ASHG ANNUAL MEETING 2022

LOS ANGELES, CA • OCTOBER 25-29

The ASHG ANNUAL MEETING

Program at a Glance

THE ASHG 2022 ANNUAL MEETING
PROGRAM-AT-A-GLANCE

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Program-at-Glance as of September 2022. Check the online planner and mobile app for the most up-to-date program including abstracts, poster listings and more! This program is being provided as a convenience for attendees who prefer to have a printed copy of the program and has not been edited for style consistency.

Tuesday, October 25, 2022

11:15 AM - 12:45 PM

Baylor College of Medicine: New Sequencing Technologies for Clinical and Translational Disease Diagnosis (Ancillary Event)
Conv Ctr/Room 402/South Building

11:30 AM - 12:30 PM

NIH/NLM/NCBI: A Conversation with NCBI about the NIH Comparative Genomics Resource (CGR) (Ancillary Event)
Conv Ctr/Room 407/South Building

1:00 PM - 1:30 PM

S01. Presidential Welcome and Address: One Human Race: Billions of Genomes
Conv Ctr/West Hall A/West Building
Charles Rotimi, PhD, ASHG 2022 President

1:30 PM - 2:15 PM

S02. Awards Recognition I
Conv Ctr/West Hall A/West Building

President Charles Rotimi will present the 2022 William Allan Award, Victor A. McKusick Award, and Arno Motulsky-Barton Childs Award for Excellence in Human Genetics Education. He will also recognize the 2020 and 2021 recipients of the awards. *AJHG* Editor-in-Chief Bruce Korf will recognize the 2022 *AJHG* Cotterman Awardees and *HGG Advances* Editor-in-Chief Michael Bamshad will recognize the inaugural *HGGA* Early-Career Investigator Awardees.

2:15 PM - 4:00 PM

S03. Featured Plenary Abstract Session I
Conv Ctr/West Hall A/West Building
Moderator: Davis, E.
Moderator: Matthijs, G.

14:15   014. Deep learning to understand the genetic architecture of the human skeletal form. Kun, E.
14:35   015. Leveraging sequencing data in the UK Biobank to detect parent-of-origin effects in 80,821 individuals.Hofmeister, R.
14:55   016. Characterizing 54 type-2 diabetes candidate genes using CRISPR/Cas9, *in vivo* imaging and deep learning in zebrafish larvae. Mujica, E.
15:15   017. Integrated multiomic analyses reveal LSD genes as a rich source of FXTAS modifiers. Shelly, K.
15:35   018. Tissue-specific dynamic eQTLs in response to a high cholesterol, high fat diet in baboons. Lin, W.

4:30 PM - 6:00 PM

S04. COVID-19 in the post-pandemic era: Long COVID, vaccine response, and beyond (Invited)
Conv Ctr/Room 502/West Building

Moderator: Schulte, E.
Moderator: Verma, S.

16:31   021. Multi-omics analysis to identify molecular determinants of virus-host interaction in SARS-CoV-2 infection. Girault, V.
16:46   022. Clinical implications of COVID-19 genetics. How can we use genetic risk to improve patient care?   Nakanishi, T.
17:01   023. Genetics of vaccine side-effects. Cirulli, E.
17:16   024. Lessons learned from the Long COVID Working Group of the COVID-19 Host Genetics Initiative–Understanding genetic risk factors for Long COVID. Ollila, H.
17:31   Questions and Answers

S05. Demographic history, natural selection, and disease risk in diverse global biobanks (Invited)
Conv Ctr/Room 515/West Building
Moderator: Biddanda, A.
Moderator: Martin, A.

16:31   027. Inferring population size dynamics of Niger-Congo speakers using deep whole genome sequences. Choudhury, A.
16:51   028. Pan-Arab genome wide genotyping array and its associated reference panel: empowering the region with the necessary tools for increasing diversity in genetic studies. Badji, R.
17:11   029. Nationwide biobank in Mexico unravels demographic history and complex trait architecture from 6,000 genomes. Sohail, M.
17:31   030. Leveraging Fine-Scale Population Structure in Health Systems. Belbin, G.
17:51   Questions and Answers

S06. Genome mapping technologies – Enabling next-generation cytogenetics (Invited)
Conv Ctr/Petree D/West Building
Moderator: Iqbal, M.
Moderator: Sahajpal, N.

16:32   033. Principles of Genome Mapping and Long-read Sequencing Technologies for Structural Variant Detection. Mantere, T.
16:47   034. Optical Genome Mapping allows for Comprehensive and Efficient Detection of Constitutional Chromosomal Aberrations. El-Khattabi, L.
17:07   035. Integrated Genomic Evaluation Using Optical Genome Mapping Combined with Targeted NGS in Hematological Malignancies. Kanagal-Shamanna, R.
17:22   036. How Genome Mapping Technologies Will Change Clinical Cytogenetics Practice. Smith, A.
17:37   Questions and Answers/Panel Discussion

S07. Genomic perspectives on biological sex (Invited)
Conv Ctr/Concourse Hall F/West Building
Moderator: Raznahan, A.
Moderator: Disteche, C.

16:31   039. The impact of sex on gene expression and its genetic regulation across human tissues. Stranger, B.
16:46   040. Linking X-Y gametologue co-expression patterns to sex differences in disease. Raznahan, A.
17:01   041. X chromosome regulation and gene expression in stem cell derivatives. Disteche, C.
17:16   042. The influence of common X-linked genetic variation on neuroanatomical variation in humans. Mallard, T.
17:31   Questions and Answers

S08. Mosaicism, vascular anomalies, and emerging therapeutics: Models for using the rare to understand the common (Invited)
Conv Ctr/Petree C/West Building
Moderator: Sheppard, S.
Moderator: Bennett, J.

16:31   045. A novel genetic cause for central conducting lymphatic anomaly. Vikkula, M.
16:46   046. Towards a quantitative understanding of genetic PI3Kalpha activation. Madsen, R.
17:01   047. A MEKINIST-ic approach to target abnormal lymphatics in Gorham-Stout disease. Dellinger, M.
17:16   048. MEK inhibition for the treatment of RAS-pathway related vascular malformations. Dayneka, J.
17:31   Questions and Answers

S09. New advances in computational genome interpretation: From prediction to the clinic (Invited)
Conv Ctr/Concourse Hall E/West Building
Moderator: Bakolitsa, C.
Moderator: Radivojac, P.

16:35   051. Presentation title: Evidence-based calibration of computational tools for missense variant pathogenicity classification. Pejaver, V.
16:55   052. Performance of diagnostic methods in identifying disease-causing variants: assessment of the Rare Genomes Project CAGI challenge. Stenton, S.
17:15   053. Progress in complex phenotype prediction: the CAGI6 PRS challenge. Chun, S.
17:35   054. Ethical challenges in the era of genomic medicine: lessons from CAGI. Fullerton, S.
17:55   Panel Discussion - Future of computational genome interpretation

6:00 PM - 8:00 PM

Washington University School of Medicine, St. Louis: Faculty, Alumni and Friends Reception (Ancillary Event)
Conv Ctr/Room 407/South Building

6:00 PM - 9:00 PM

Bionano Genomics Cocktail Reception: Eat, Drink & Transform with us at the Bionano Social (Registration Required) (Ancillary Event)
JW Marriott Hotel/The Mixer Room/Lobby Level

NIH's Gabriella Miller Kids First and INCLUDE Programs: Accelerating Pediatric Genomics Research Through Collaboration: Networking & Poster Session (Ancillary Event)
JW Marriott Hotel/Platinum A/3rd floor

6:30 PM - 8:00 PM

Excellence in Genetics Awards Recognition Reception (by invitation only)
Conv Ctr/Room 408/South Building

Trainee Social Reception (Admission by advance ticket purchase only)
Conv Ctr/Petree Plaza/West Building

7:00 PM - 8:15 PM – Concurrent Poster Talks
Poster Talks give attendees a sneak peek at some of the top-scoring posters across a variety of topics through rapid-fire presentations. The presenters still present at Poster Sessions.

S11. Poster Talks II
Conv Ctr/Petree D/West Building

19:00   626 / PB3257. Web-CSEA: Web-based Cell-type-Specific Enrichment Analysis of Genes. Dai, Y.

19:03   627 / PB2358. Updated clinical practice guidelines for managing children and adults with 22q11.2 deletion syndrome. McDonald-McGinn, D.

19:06   628 / PB2318. Investigating the frequency of parental gonosomal mosaicism in Neurodevelopmental Disorder cohorts to improve the genetic diagnostic rate. Sandran, N.

19:09   629 / PB2329. Multiple molecular diagnoses identified by clinical whole genome sequencing in an international cohort of more than 1,200 individuals. Thorpe, E.

19:12   630 / PB2591. Functional characterization of a heat shock regulated enhancer at coronary artery disease associated locus that controls CALCRL expression in endothelial cells. Selvarajan, I.

19:15   631 / PB2615. Hematopoietic Loss of Y Chromosome Leads to Cardiac Fibrosis and Heart Failure Mortality. Forsberg, L.

19:18   632 / PB2547. Consequences of loss of function burden effects on the human plasma proteome in 54,306 UK Biobank participants. Whelan, C.

19:21   633 / PB2585. Fine-mapping causal tissues and genes at disease-associated loci. Strober, B.

19:24   634 / PB2704. The role of introgressed archaic DNA in the human genome. Young, R.

19:27   635 / PB2891. A spatial map of neurodevelopmental disorder risk in the developing human cortex. Segato Dezem, F.

19:30   636 / PB2937. Clinical use of the amniotic fluid cells transcriptome in deciphering Mendelian disease. Lee, M.

19:33   637 / PB2938. Closing the gap: Solving complex medically relevant regions of the human Genome. Sedlazeck, F.

19:36   638 / PB3083. Multiset correlation and factor analysis enables exploration of multi-omic data. Brown, B.

19:39   639 / PB3141. Single-cell allele-specific expression analysis reveals dynamic and cell-type-specific regulatory effects, Qi, G.

19:42   640 / PB2742. Novel genetic signals identified for angiotensin-converting enzyme inhibitor-induced cough. Coley, K.

19:45   641 / PB2086. Genetic deconvolution of fetal and maternal cell-free DNA in maternal plasma enables next generation non-invasive prenatal screening. Zhang, J.

19:48   642 / PB2108. Phenome and genomewide recessive scan of coding variants reveals novel associations for female infertility. Ruotsalainen, S.

19:51   643 / PB3356. Deep-PheWAS.R: an innovative R package for phenome wide association of novel phenotypes. Packer, R.

19:54   644 / PB3387. Evaluating strategies to infer continental ancestry using single-cell RNA-seq datasets: an application to the Human Cell Atlas. Yao, J.

19:57   645 / PB3401. Functional Annotations-Informed Whole Genome Sequence Analysis Identifies Novel Rare Variants for AD in the Alzheimer's Disease Sequencing Project. Lee, S.

20:00   646 / PB3540. Performance and accuracy of imputation panels for genetic association studies in sub-Saharan African populations. Sengupta, D.

20:03   647 / PB3630. Winner's curse in rare variants association analysis: bias depends on effect direction and the pooled methods used. Soave, D.

20:06   648 / PB3421. Genome-wide study on 72,298 Korean individuals in Korean biobank data for 76 traits identifies hundreds of novel loci. Nam, K.

20:09   649 / PB3511. MENDEL-modified segregation analysis of 168 *TP53*-positive families estimates age-specific risks for cancer types beyond the established spectrum for Li-Fraumeni syndrome. Fortuno, C.

20:12   650 / PB3601. The impact of 22q11.2 copy number variants on human traits in the general population. Zamariolli, M.

S10. Poster Talks I
Conv Ctr/Petree C/West Building

19:00   601 / PB1223. Uncovering gene fusions with 3D genomics: from clinical validation to actionable insights for undiagnosable solid tumors. Schmitt, A.

19:03   602 / PB1229. Utilizing Electronic Health Records (EHR) and Tumor Panel Sequencing to Demystify Prognosis of Cancer of Unknown Primary (CUP) patients. Moon, I.

19:06   603 / PB1047. Clinicogenomic Neural Networks with Real World Evidence: Predicting Outcomes and Targeting Interventions at a Comprehensive Cancer Center. Flagg, M.

19:09   604 / PB1136. Identifying molecular markers of early stage ovarian cancer pathogenesis through whole genome CRISPR screening of mutant *BRCA1* fallopian tube cells. Dhungana, S.

19:12   605 / PB1039. Chromatin accessibility of primary human cancers ties regional mutational processes and signatures with tissues of origin. Reimand, J.

19:15   606 / PB1508. Investigating the influence of genetic ancestry on gene-environment interactions on polygenic risk score and acculturation: Results from the Hispanic Community Health Study/Study of Latinos. Sharma, J.
19:18   607 / PB1354. Estimating the causal influence of body mass index on gut microbiome variation. Hughes, D.
19:21   608 / PB1500. Integrative analysis of large-scale multi-ancestry genome-wide association study and single-cell omics data provides high-resolution insight of cell-types in the pathogenesis of type 2 diabetes. Suzuki, K.
19:24   609 / PB1649. Somatic mutations in chronic lung disease are associated with reduced lung function. Yun, J.
19:27   610 / PB1542. Multi-ancestry HLA allele calling using whole-exome sequencing in the UK Biobank reveals 353 novel genome-wide significant HLA alleles associated with 11 auto-immune phenotypes. Butler-Laporte, G.
19:30   611 / PB1512. Key discoveries of the genetic basis of stuttering. Pruett, D.
19:33   612 / PB1596. Prioritizing genes and gene programs for disease by integrating genetic and perturbation data. Dey, K.
19:36   613 / PB2408. Epigenetic Age Acceleration is Suggestively Associated with Stroke Precursor Phenotypes. Dueker, N.
19:39   614 / PB2396. Dissecting the molecular machinery and sequence of histone to protamine transition during spermiogenesis. Rabbani, M.
19:42   615 / PB2388. Cross-tissue patterns of DNA hypomethylation reveal genetically distinct histories of cell development. Scott, T.
19:45   616 / PB2861. Understanding natural selection in the European Holocene using Ancient DNA. Pandey, D.
19:48   617 / PB2237. Using variant databases to estimate disease prevalence for rare recessive disease. O'Donnell-Luria, A.
19:51   618 / PB2052. Long-term efficacy and safety of elamipretide in patients with Barth syndrome is presented through the 192-week open-label extension results of TAZPOWER. Campbell, J.
19:54   619 / PB1863. GWAS of Down Syndrome Associated Atrioventricular Septal Defect identifies three novel loci. Feldman, E.
19:57   620 / PB1874. Human brains with Tay Sachs disease exhibit altered transcriptomes during fetal development. Han, S.
20:00   621 / PB1909. Loss of C-terminal Mediator Complex subunit-11 impairs fetal brain development and cause severely progressive neurodegeneration. Cali, E.
20:03   622 / PB1990. The impact of NGS on rare and complex epilepsies: identification of novel causative variants for an efficient diagnostic pathway and personalized treatments. Gellera, C.
20:06   623 / PB1764. Bi-allelic LETM1 variants perturb mitochondrial ion homeostasis leading to a clinical spectrum with predominant nervous system involvement. Kaiyrzhanov, R.
20:09   624 / PB1892. Investigating tissue specific defects in mitochondrial bioenergetics and quality control: new implications for cellular pathogenesis and therapeutic targeting in Barth syndrome. Sniezek, O.
20:12   625 / PB1919. Molecular insights into the pathogenesis of Chediak-Higashi syndrome and the biology of Lysosomal Trafficking Regulator. Morimoto, M.

7:30 PM - 9:30 PM

Baylor College of Medicine Genetics Reception (Ancillary Event)
JW Marriott Hotel/Platinum D/3rd floor

Wednesday, October 26, 2022

7:00 AM - 8:00 AM

Industry Education Session Presented by Agilent Technologies: New technology adoption for implementing germline whole exome sequencing at a rural academic medical center (Industry Solutions)
Conv Ctr/Room 301/South Building

Industry Education Session Presented by Amazon Web Services (Industry Solutions)
Conv Ctr/Room 402/South Building

Industry Education Session Presented by Parse Biosciences: Introduction to Single Cell Technologies and Combinatorial Barcoding (Industry Solutions)
Conv Ctr/Room 306/South Building

Industry Education Session Presented by Resolve Biosciences: How do you answer biological questions through spatial genomics? (Industry Solutions)
Conv Ctr/Room 304/South Building

Industry Education Session Presented by UK Biobank: How UK Biobank is enabling scientific discoveries in population health (Industry Solutions)
Conv Ctr/Room 303/South Building

7:00 AM - 8:30 AM

ClinGen Polygenic Risk Score Translation Needs Assessment (Ancillary Event)
JW Marriott Hotel/Georgia I/3rd floor

NHGRI/NIH: Applying for NIH Grants - Strategies for Success (Ancillary Event)
Conv Ctr/Room 404/South Building

University of Helsinki: FinnGen Consortium Breakfast Meeting (Ancillary Event)
Conv Ctr/Room 407/South Building

S12. Genomic research in the All of Us Researcher Workbench: Advancing precision medicine for all (Admission by advance ticket purchase only)
Conv Ctr/Room 403/South Building

S13. Investigating public variant datasets with the UCSC Genome Browser (Admission by advance ticket purchase only)
Conv Ctr/Room 408/South Building

8:30 AM - 10:00 AM

S14. A collection of cardiovascular and metabolic disease conundrums (Platform)
Conv Ctr/Petree D/West Building
Moderator: Tcheandjieu, C.
Moderator: Horne, B.

08:30　064. A large-scale multi-ethnic mitochondrial-wide association analysis for cardio-metabolic traits. Zhou, J.
08:45　065. Genome-wide association study of coronary microvascular disease assessed by stress cardiac perfusion positron emission tomography. Verma, S.
09:00　066. Genome-wide analysis reveals novel mechanisms underlying atrial fibrillation and the clinical utility of a polygenic predictor for cardioembolic risk. Miyazawa, K.
09:15　067. Pathway and cell state specific polygenic risk scores shed light into the genetic basis of coronary artery disease. Kaikkonen, M.
09:30　068. Discovery of novel pharmacogenomic biomarkers of clopidogrel response in African Americans using multi omics. Yang, G.
09:45　069. Identification of *BRAF* as a novel gene for autosomal dominant dilated cardiomyopathy. Lam, C.

S15. Applying Mendelian randomization to complex traits (Platform)
Conv Ctr/West Hall A/West Building
Moderator: Walters, R.
Moderator: Seyerle, A.

08:30　072. Integration of single-nucleus RNA-sequencing into Mendelian randomization reveals an adipose cell-type-origin effect of abdominal obesity on non-alcoholic fatty liver disease. Lee, S.
08:45　073. Mendelian randomization and colocalization characterize the impact of the plasma proteome on human complex diseases. Baird, D.
09:00　074. Proteome-wide Mendelian randomization implicates nephronectin as an actionable mediator of the effect of obesity on COVID-19 severity. Yoshiji, S.
09:15　075. Pleiotropy testing identifies novel loci associated with lipid traits in multiple ethnicities. Lorincz-Comi, N.
09:30　076. Genetically proxied PCSK9 inhibition provides indications of lower prostate cancer risk: A Mendelian randomization study. Fang, S.
09:45　077. Mendelian randomization (MR) identifies possible causal gene expression in multiple sclerosis (MS). Harley, J.

S16. Considering ancestry in study populations (Platform)
Conv Ctr/Room 515/West Building
Moderator: Tamayo, L.
Moderator: Jin, J.

08:30　080. Genetic and phenotypic associations of whole blood gene expression in the admixed population of Greenland. Hanghoej, K.
08:45　081. Multi-ancestry genome-wide association study in >2.5 million individuals reveals distinct biological pathways driving type 2 diabetes susceptibility with heterogeneous effects across diverse population groups. Hatzikotoulas, K.
09:00　082. Leveraging global multi-ancestry meta-analysis in the study of idiopathic pulmonary fibrosis genetics. Partanen, J.
09:15　083. Genetic architecture of the inflammatory bowel diseases across East Asian and European ancestries. Liu, R.
09:30　084. The extent of allelic heterogeneity in a trans-ancestry GWAS meta-analysis of alcohol and tobacco addiction in 3.4 million individuals. Saunders, G.
09:45　085. Epigenetic variation impacts ancestry-associated differences in the transcriptional response to influenza infection. Aracena, K.

S17. Genetic and functional advances in inherited neuromuscular disease (Platform)
Conv Ctr/Room 502/West Building
Moderator: Scott, A.
Moderator: Meyer-Schuman, R.

08:30　088. A whole exon targeted PCR/Nanopore sequencing assay that reveals SNVs, indels and CNVs across *SMN1* and *SMN2* with implications for SMA carriers and disease severity. Rao, P.
08:45　089. *SPTSSA* variants alter sphingolipid synthesis and cause a complex form of hereditary spastic paraplegia. Pan, X.

09:00   090. A reverse genetics and genomics approach to gene paralog function and disease: Myokymia and the juxtaparanode. Marafi, D.
09:15   091. Predicting gene regulatory networks related to the development and maintenance of Schwann cells in the peripheral nervous system. Khullar, S.
09:30   092. A *C. elegans* model of neuropathy-associated *GARS1* mutations. Pierluissi, J.
09:45   093. Development of mini-gene therapy for Charcot-Marie-Tooth disease type 4B3 using patient-derived iPSCs. Jacobs, E.

S18. Genetic impacts on the epigenome and beyond (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator: Ramaker, M.
Moderator: Lu, T.

08:30   096. Enhancer cluster swapping reveals latent context sensitivity at the Igf2/H19 locus. Maurano, M.
08:45   097. Genetic regulation of methylation across East Asian and European populations. Xia, Y.
09:00   098. Genetic determinants of DNA methylation in African Americans: An meQTL mapping study in GENOA. Shang, L.
09:15   099. Identifying molecular mechanisms across common and complex neurological diseases by integrating single-cell epigenomics. Marderstein, A.
09:30   100. Targeted activation of the imprinted PWS locus via CRISPR/Cas9-based epigenome editing. Rohm, D.
09:45   101. Single-sample, whole-genome parent-of-origin inference from imprinting control regions using ultra-long and Pore-C nanopore reads. McKenzie, S.

S19. Genetics of human immunity, inflammation and infection (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator: Li, D.
Moderator: Ramos, P.

08:30   104. Dissecting IBD GWAS loci using single-cell and bulk multi-omics analysis. Xu, Z.
08:45   105. Methylation patterns associated with inflammation traits in racially and ethnically diverse populations. Lundin, J.
09:00   106. Single-cell models reveal more pervasive T cell state-dependent regulatory effects than in bulk analyses. Nathan, A.
09:15   107. Genetic and epigenetic regulation of immune cell subpopulations across health and ages. Fave, M.
09:30   108. Influenza GWAS of >1 million individuals identifies and experimentally validates two loci in *ST6GAL1* and *B3GALT5* and shows that risk factors for COVID-19 are largely distinct from flu. Kosmicki, J.
09:45   109. Novel genetic associations underlying symptom severity and infection susceptibility to SARS-CoV-2. Andrews, S.

S20. High-throughput characterization of coding variants, from benchtop to desktop (Platform)
Conv Ctr/Petree C/West Building
Moderator: Yang, J.
Moderator: Adams, D.

08:30   112. Using Saturation Mutagenesis-Reinforced Functional Assays (SMuRF) to accelerate the pathogenicity identification of variants of uncertain significance. Ma, K.
08:45   113. Machine learning and genome editing to resolve variants of uncertain significance in *TSC2*. Biar, C.
09:00   114. Functional classification of all possible SNVs in *VHL* using saturation genome editing. Findlay, G.
09:15   115. A novel method to profile missense variation in extracellular proteins at scale reveals biochemical features important for expression and secretion of coagulation factor IX. Popp, N.
09:30   116. The landscape of regional missense mutational intolerance quantified from 125,748 exomes Wang, L.
09:45   117. Prediction of recessive inheritance for missense variants in human disease. Petrazzini, B.

10:00 AM - 10:30 AM

CoLab Session Presented by 10X Genomics: Integrated spatial transcriptomics and genomics in high-grade gliomas (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Bionano Genomics: Multi-site clinical study of optical genome mapping and comprehensive genome analysis of SNVs and SVs utilizing a single software solution (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by Genomenon: Curating the Genome to Drive Precision Diagnosis in Clinical Care (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

10:45 AM - 12:15 PM

S21. Averting Alzheimer's as soon as possible (Platform)
Conv Ctr/Petree D/West Building
Moderator: Celis, K.
Moderator: Cornejo-Olivas, M.

10:45   120. Single-cell transcriptomic and epigenomic dissection of Alzheimer's disease pinpoints causal GWAS variants and reveals epigenome erosion. James, B.
11:00   121. Genome-wide circRNA dysregulation contributes to Alzheimer's disease pathogenesis. Li, Y.
11:15   122. Spatial transcriptomics based gene interactions analysis in AD brain. Wang, S.
11:30   123. Large-scale multi-omic analyses in CSF identified multiple causal and druggable targets for Alzheimer's disease. Cruchaga, C.
11:45   124. Rare-variant analysis of whole-genome sequence data obtained from multi-ancestry families identifies new genes associated with late-onset Alzheimer's disease. Leal, S.
12:00   125. Multi-ancestry genome-wide association analysis of late-onset Alzheimer's disease (LOAD) in 60,941 individuals identifies novel cross-ancestry loci. Naj, A.

S22. Epigenomic associations with disease (Platform)
Conv Ctr/Room 502/West Building
Moderator: Kitzman, J.
Moderator: Kundu, S.

10:45   128. Method for lung cancer detection using BAL fluid based on differential methylation pattern analysis by MRE-seq. Min, N.
11:00   129. Multi-level epigenome profiling reveals a distinct role for DNA methylation in driving cellular differentiation. Hodges, E.
11:15   130. Profiling pre-diagnosis plasma cell-free DNA methylomes up to seven years prior to clinical detection reveals early signatures of cancers. Cheng, N.
11:30   131. MeOW: genome-wide identification of differentially methylated regions from Oxford Nanopore long-read sequencing data. Galey, M.
11:45   132. Whole blood-based epigenome-wide association study of renal cell carcinoma reveals hypomethylation of a human-specific sequence in the second intron of *PCBD2*. Shimizu, A.
12:00   133. An epigenome-wide association study (EWAS) of allergic sensitization in children of diverse ancestry using a custom allergy & asthma array reveals an enrichment for differentially methylated high-value CpGs compared to the EPIC array. Thompson, E.

S23. Let's talk about sex and its contributions to disease risk (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator: Hodonsky, C.
Moderator: Chikowore, T.

10:45   136. Testing for interaction using over 1 million individuals and 20 diseases shows considerable differences in polygenic risk score estimates across age groups and sexes. Jermy, B.
11:00   137. GWAS meta-analysis of WHRadjBMI identifies differences across sexes and populations. Wilson, E.
11:15   138. Phenome-wide sex differences in environmental effects in the National Health and Nutrition Examination Survey (NHANES) 1999-2006. Gonzalez Zarzar, T.
11:30   139. Regulatory network approaches reveal sex differences in cancer gene regulation. Lopes-Ramos, C.
11:45   140. *ADGRG6* is involved in gender-specific fat distribution. Nguyen, H.
12:00   141. A proteome-wide investigation of sex differences in genetic control of brain protein abundance in 1100 human brain samples. Wingo, T.

S24. Massively parallel variant characterization (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator: O'Donnell-Luria, A.
Moderator: Azaiez, H.

10:45   144. Deciphering sequence effects of splicing in health and disease with massively parallel reporter assays. Yu, A.
11:00   145. Improving estimation of variant functional effects from high-throughput assays. Yu, T.
11:15   146. Massively parallel functional assessment of missense variants in cardiac arrhythmia genes *KCNQ1* and *KCNE1* elaborates structure-function relationships. Muhammad, A.
11:30   147. Saturation genome editing reveals 10% of missense SNV alleles in functional domains of PALB2 as functionally abnormal. Dawood, M.
11:45   148. Massively parallel screen for rare 3' UTR variants regulating mRNA abundance. Fu, T.
12:00   149. Quantifying the functional effects of 234,448 likely causal regulatory variants underlying complex human traits. Siraj, L.

S25. Novel statistical genetics methods for complex traits (Platform)
Conv Ctr/Room 515/West Building
Moderator: Ge, T.
Moderator: Patel, R.

10:45   152. A unified method for estimating direct genetic effects and performing genome-wide association studies. Guan, J.
11:00   153. Inference of causal networks using bi-directional Mendelian randomization and network deconvolution with GWAS summary data. Lin, Z.
11:15   154. A variational Bayesian approach to characterize pleiotropic components across thousands of human diseases and complex traits using GWAS summary statistics. Zhang, Z.
11:30   155. Fast multiple-trait genome-wide association analysis for correlated longitudinal measurements. Abdel-Azim, G.
11:45   156. Flexibly encoded GWAS identifies novel nonadditive SNPs in individuals of African and European ancestry. Zhou, J.
12:00   157. Tree-based phasing and imputation increases speed and accuracy of SNP-array imputation and opens the door to population scale inference. Palmer, D.

S26. Somatic mosaicism in human health and disease (Platform)
Conv Ctr/Petree C/West Building
Moderator: Conlin, L.
Moderator: Aldinger, K.

10:45   160. Delineating the rates and determinants of human somatic variation among tissues and individuals. Xu, H.
11:00   161. Detection, causes and consequences of Y chromosome mosaicism. Zhao, Y.

11:15   162. Genetic investigation of mosaic loss of the X chromosome in peripheral leukocytes of 918,085 women identifies germline predisposition and strong signals of haplotype selection. Liu, A.
11:30   163. Meiotic and mitotic aneuploidies drive preimplantation mortality of in vitro fertilized human embryos. McCoy, R.
11:45   164. Comprehensive multiomic profiling of somatic mutations in malformations of cortical development. Chung, C.
12:00   165. Profiling *PIK3CA* variants in disorders of somatic mosaicism. Mojarad, B.

S27. Therapeutic development for Mendelian disorders (Platform)
Conv Ctr/West Hall A/West Building
Moderator: Morini, E.
Moderator: Kemaladewi, D.

10:45   168. Development of a novel oral treatment that rescues gait ataxia and retinal degeneration in a phenotypic mouse model of familial dysautonomia. Morini, E.
11:00   169. High-throughput transcriptome analyses from ASPIRO, a phase 1/2/3 study of gene replacement therapy for XLMTM. Andreoletti, G.
11:15   170. Therapeutic outcomes of selumetinib treatment in pediatric and adult Korean patients with neurofibromatosis type 1 and inoperable plexiform neurofibromas. Lee, B.
11:30   171. Reversible epigenome editing of *PCSK9* as a therapeutic strategy. Whittaker, M.
11:45   172. Dosage dependent suppression and replacement gene therapy for *KCNH2*-mediated arrhythmias. Bains, S.
12:00   173. Base editing as a treatment approach for correction of c.2988+1G to A, the most common CF-causing variant in individuals of African descent. Kavanagh, E.

12:15 PM - 1:45 PM

Lunch Break: Explore the Exhibit Hall, View Posters, and Join Industry Sessions
Conv Ctr/Exhibit/Poster Hall/South Building

Diversity, Equity, and Inclusion Luncheon (Admission by advance ticket purchase only)
Conv Ctr/Room 408/South Building

Self-Discovery Workshop: Finding Balance & Joy in Your Work (Admission by advance ticket purchase only)
Conv Ctr/Room 403/South Building

Broad Institute: A2FKP (Ancillary Event)
Conv Ctr/Room 409/South Building

Genes MDPI: Editorial Board Member Gathering (Ancillary Event)
Conv Ctr/Room 505/West Building

Springer Nature: Meet the genetics editors from Nature Portfolio journals (Ancillary Event)
Conv Ctr/Room 407/South Building

12:30 PM - 1:00 PM

CoLab Session Presented by Golden Helix: Maximizing Profitability in your NGS Testing Lab: develop repeatable cancer and germline interpretation workflows scaling from panels to whole genomes (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Roche Sequencing and Life Science: It is time to take a leap forward in digital PCR technology with Roche's Digital LightCycler® System (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Thermo Fisher Scientific (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

12:30 PM - 1:30 PM

Industry Education Session Presented by Advanced Cell Diagnostics: Visualize diverse gene expression signatures at single-cell resolution using RNAscope™ (Industry Solutions)
Conv Ctr/Room 301/South Building

Industry Education Session Presented by Covaris (Industry Solutions)
Conv Ctr/Room 411/South Building

Industry Education Session Presented by DNAnexus (Industry Solutions)
Conv Ctr/Room 306/South Building

Industry Education Session Presented by Illumina: Illumina technology improvements driving genomic insights (Industry Solutions)
Conv Ctr/Room 304/South Building

Industry Education Session Presented by PacBio: The long and short of it – learn about game changing capabilities of PacBio sequencing (Industry Solutions)
Conv Ctr/Room 402/South Building

Industry Education Session Presented by SomaLogic: Cerebrospinal fluid proteogenomic analyses: identification of novel proteins and therapeutic targets for Alzheimer's and Parkinson's disease (Industry Solutions)
Conv Ctr/Room 303/South Building

1:15 PM - 1:45 PM

CoLab Session Presented by BGI / MGI: More accurate, reliable, and affordable sequencing platforms through MGI DNBSEQ technology! (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Metabolon (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by PerkinElmer Diagnostics: Expanding the Functionality of scRNAseq (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

1:45 PM - 2:45 PM

S28. Ancestry and admixture in diverse populations (Platform)
Conv Ctr/West Hall A/West Building
Moderator: Lin, M.
Moderator: Mancuso, N.

13:45   176. An ancestry-specific allele frequency browser at 142 million variants and an imputation reference panel derived from exome and whole genome sequencing of the Mexico City Prospective Study. Ziyatdinov, A.
14:00   177. IBD-Mix: An accurate IBD segment-based local ancestry inference method. Zhi, D.
14:15   178. Using topological ancestry groups to describe population structure in large multi-ethnic biobanks. Diaz-Papkovich, A.
14:30   179. On the number of genealogical ancestors tracing to the source groups of an admixed population. Mooney, J.

S29. Emerging topics in biobank-scale association analysis (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator: Wu, Y.
Moderator: Border, R.

13:45   182. Genetic analysis of breast density phenotypes derived using deep learning applied to the UK Biobank MRI dataset. Geraghty, B.
14:00   183. Age-dependent topic modelling of comorbidities in UK Biobank identifies disease subtypes with differential genetic risk. Jiang, X.
14:15   184. Refined patient stratification leveraging biomarker and other quantitative traits from UK Biobank to enhance PheWAS analyses. Vitsios, D.
14:30   185. Participation bias correction reveals substantial impact on genetic association and downstream analyses in the UK Biobank. Schoeler, T.

S30. Genotypes and phenotypes of Mendelian disorders (Platform)
Conv Ctr/Petree D/West Building
Moderator: Thiffault, I.
Moderator: Manickam, K.

13:45   188. The inner junction protein CFAP20 functions in motile and non-motile cilia and is critical for vision. Au, P.
14:00   189. Defining the clinical and molecular spectrum of a *KDM6B*-related neurodevelopmental disorder through large cohort analysis, 3D-protein structure analysis, and *Drosophila* functional assays. Jakub, T.
14:15   190. *De novo* variants in *MAST4* cause a neurodevelopmental disorder with variable brain malformations and epilepsy. Houghtaling, S.
14:30   191. Standardized phenotypic similarity analysis in 10,705 exome sequencing trios with 619,109 clinical annotations reveals hidden patterns in neurodevelopmental disorders. Ganesan, S.

S31. How do we express ourselves? Examining promoter and enhancer biology (Platform)
Conv Ctr/Room 515/West Building
Moderator: Burton, E.
Moderator: Luca, F.

13:45   194. Allelic specific transcription factor binding and chromatin accessibility shape promoter kinetics in human cell lines. Jin, B.
14:00   195. Massively parallel reporter assay reveals promoter position-dependent and tissue-specific effects in islet TSSs. Bose, M.
14:15   196. Adaptive sequence divergence forged new neurodevelopmental enhancers in humans. Mangan, R.

14:30   197. A global high-density chromatin interaction network reveals functional long-range and trans-chromosomal relationships. Iohia, R.

S32. Influence of germline variants on somatic genomic features in cancer risk and progression (Platform)
Conv Ctr/Petree C/West Building
Moderator: Wang, Z.
Moderator: Hennessey, R.

13:45   200. Germline cancer gene expression quantitative trait loci influence local and global tumor mutations. Liu, Y.
14:00   201. A comprehensive analysis of clinical and polygenic germline influences on somatic mutational burden with implications for survival. Taraszka, K.
14:15   202. The impact of common germline risk on somatic alterations and clinical features across cancers. Namba, S.
14:30   203. Genomic analysis of skin cancers from xeroderma pigmentosum subgroups revealed new mechanisms of UV mutagenesis. Nikolaev, S.

S33. Investigating complex genomic regions (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator: Rodriguez, O.
Moderator: Turner, T.

13:45   206. Repeat polymorphisms underlie top genetic risk loci for glaucoma and colorectal cancer. Mukamel, R.
14:00   207. Extensive mosaicism by somatic L1 retrotransposition in normal human cells. Ju, Y.
14:15   208. Evaluation of long reads across challenging medically relevant genes and their implications for *All of Us*. Mahmoud, M.
14:30   209. A global reference for human genetic variation at tandem repeats. Ziaei Jam, H.

S34. New methods for revealing repeats (Platform)
Conv Ctr/Room 502/West Building
Moderator: Seifert, B.
Moderator: Miller, D.

13:45   212. RExPRT: a machine learning tool to rank repeat expansions by pathogenicity. Fazal, S.
14:00   213. Precise and ultrafast tandem repeat variant detection in massively parallel sequencing reads. Wang, X.
14:15   214. Genome-wide analysis of mutations and epigenetic changes of tandem repeat regions in rare genetic disease cases. Dolzhenko, E.
14:30   215. Non-linear effects of short tandem repeats on gene expression. Lamkin, M.

3:00 PM - 4:45 PM – Poster Session I
Poster authors will be at their boards on Wednesday or Thursday from 3:00 to 4:45 pm. See pages xx-xx for Wednesday listings.

3:00 PM - 3:30 PM

CoLab Session Presented by Beckman Coulter Life Sciences: Optimizing your NGS lab with a game-changing, easy-to-use automation solution (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by Integrated DNA Technologies: ctDNA for minimal residual disease research by hybridization capture in resected cancer samples (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by PacBio: Unlock the next wave of discoveries in Human Genetics with PacBio technologies (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

3:45 PM - 4:15 PM

CoLab Session Presented by DNAnexus: Detection of Subtle Genomic Changes and Patient Subtyping (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Parse Biosciences: Democratizing Access to Single Cell RNA Sequencing with Evercode Whole Transcriptome (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Sony Biotechnology (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

4:30 PM - 5:00 PM

CoLab Session Presented by Amazon Web Services (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by Oxford Nanopore Technologies: The sequencing platform for multiomics – where will Oxford Nanopore's PromethION take you? (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Quantabio (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

5:00 PM - 5:15 PM

S35. Awards Recognition II
Conv Ctr/West Hall A/West Building

President Charles Rotimi will present the 2022 Curt Stern Award and Early-Career Award. He will also recognize the 2020 and 2021 recipients of the awards.

5:15 PM - 7:00 PM

S36. Featured Plenary Abstract Session II
Conv Ctr/West Hall A/West Building
Moderator: Assimes, T.
Moderator: Reddy, T.

17:15   221. Multi-omic profiling of 204,494 cells and nuclei from human islet donors under basal and stimulatory conditions nominates causal cell types, genes, and regulatory element contexts at T1D GWAS loci. Robertson, C.
17:35   222. Polygenic score performance varies across the continuum of genetic ancestry in all human populations. Ding, Y.
17:55   223. Rapid and scalable preclinical evaluation of personalized antisense oligonucleotides using organoids derived from rare disease patients. Younger, S.

18:15   224. A genome-wide atlas of recurrent repeat expansions in human cancer. Erwin, G.

18:35   225. An oligogenic inheritance test discovers novel risk genes and interactions in congenital heart defects. Pittman, M.

7:00 PM - 8:30 PM

Broad Institute: GIANT Consortium Face-to-Face Meeting (Ancillary Event)
Conv Ctr/Room 407/South Building

7:00 PM - 9:00 PM

CIHR-SickKids Canadian Mixer (Ancillary Event)
JW Marriott Hotel/Platinum A/3rd floor

Duke University Alumni Reception (Ancillary Event)
JW Marriott Hotel/Platinum G/3rd floor

GIM Editorial Board Meeting (Ancillary Event)
JW Marriott Hotel/Georgia I/3rd floor

Illumina / ClinGen & Medical Genome Initiative Reception (Ancillary Event)
Conv Ctr/Room 406/South Building

Resolve Biosciences Customer Dinner (Ancillary Event)
JW Marriott Hotel/Plaza I/3rd floor

UCLA Department of Human Genetics and UCLA Intercampus Medical Genetics Training Program Reception (Ancillary Event)
Conv Ctr/Room 409/South Building

7:30 PM - 9:00 PM

Diversity, Equity, and Inclusion Reception (ASHG Reception)
JW Marriott Hotel/Platinum D/3rd floor

Thursday, October 27, 2022

7:00 AM - 8:00 AM

Industry Education Session Presented by 10X Genomics: Access the full richness of biological complexity with single cell and spatial multiomics (Industry Solutions)
Conv Ctr/Room 304/South Building

Industry Education Session Presented by Amazon Web Services (Industry Solutions)
Conv Ctr/Room 411/South Building

Industry Education Session Presented by Azenta Life Sciences: Do More with Less: Harnessing the Power of Multiomics from a Single Sample (Industry Solutions)
Conv Ctr/Room 402/South Building

Industry Education Session Presented by Element Biosciences: High accuracy, high-resolution long and short read sequencing for transcriptomes, metagenomes, targeted sequencing and more (Industry Solutions)
Conv Ctr/Room 306/South Building

Industry Education Session Presented by Parse Biosciences: Educational Seminar 2: Evercode WT v2 and Beyond (Industry Solutions)
Conv Ctr/Room 301/South Building

Industry Education Session Presented by Standard BioTools: Achieve a fast, high throughput PGx workflow that detects actionable SNPs and CNVs while supporting customization (Industry Solutions)
Conv Ctr/Room 303/South Building

7:00 AM - 8:30 AM

Global Lipids Genetics Consortium Meeting (Ancillary Event)
JW Marriott Hotel/Georgia I/3rd floor

8:30 AM - 10:00 AM

S37. Presidential Symposium: African Genomics (Plenary)
Conv Ctr/West Hall A/West Building

From participating in the development of the International HapMap and 1000 Genomes projects to the establishment of the African Society of Human Genetics and the Human Heredity and Health in Africa Initiative (H3Africa), the African genomics ecosystem has experienced tremendous growth. The increasing participation of African scientists and communities is transforming human genetics and genomics and is generating profound new insights and capacity to ask novel questions and advance human health. The 2022 Presidential Symposium will highlight this profoundly dynamic and diverse continent's major advances, new directions and goals, emerging scientific leadership, exciting investment in technology infrastructure, and more. How can and will genomics in Africa 'spread its wings' and what areas are most exciting? Join a global community for an exciting dialogue about what African genomics is - and is becoming. The session moderated by Charles Rotimi and Francis Collins includes the following speaker line-up followed by a panel discussion with questions from the audience.

Speaker Happi, C.
Speaker Makani, J.

Speaker Owolabi, M.
Speaker Mulder, N.
Moderated Panel/Q&A

10:00 AM - 10:30 AM

CoLab Session Presented by Bio-Rad Laboratories: Overcoming Limitations of RNA-Seq Library Construction from FFPE samples using a Novel Workflow (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by Illumina: Illumina Complete Long-Read technology – Benefits of Illumina's novel long-read technology (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by PerkinElmer Diagnostics: Improving NGS Library Preparation and Cell Free DNA Analysis (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

10:45 AM - 12:15 PM

S38. Disorders of bone pathology X-rayed using common and rare disease approaches (Platform)
Conv Ctr/Petree D/West Building
Moderator: Mendoza-Londono, R.
Moderator: Ramadesikan, S.

10:45   234. EFEMP1 regulates bone structure and effects fracture risk: A discovery from HRPQCT bone microarchitecture GWAS screening confirmed by CRISPR knockout zebrafish and mice: The Bone Microarchitecture International Consortium (BoMIC). Hsu, Y.
11:00   235. Converging evidence from rare and common variants implicates target genes for osteoporosis. Zhou, S.
11:15   236. COL11A2 as a candidate gene for vertebral malformations and scoliosis. Sobreira, N.
11:30   237. PTBP1 cytoplasmic retention is associated with osteochondrodysplasia and variable neurodevelopmental anomalies. Masson, A.
11:45   238. Single-cell RNAseq uncovers novel hidden mechanisms in a model of osteogenesis imperfecta. Zieba, J.
12:00   239. Low-dose infigratinib, an oral selective fibroblast growth factor receptor tyrosine kinase inhibitor, demonstrates activity in a preclinical model of hypochondroplasia. Dambkowski, C.

S39. Navigating the complex genetic landscape of neurodevelopmental disorders (Platform)
Conv Ctr/Room 502/West Building
Moderator: Smail, C.
Moderator: Carvill, G.

10:45   242. Functional genomics provide key insights to improve the diagnostic yield of hereditary ataxia. Chen, Z.
11:00   243. Contribution of copy number variants to schizophrenia in East Asian populations. Feng, Q.
11:15   244. *In vivo* functional screening in the mouse brain uncovers autism-relevant enhancers. Lambert, J.
11:30   245. Single-cell transcriptomic benchmarks of human iPSC-derived neuronal cultures and their implications for modeling neurological disorders. Gao, D.
11:45   246. Dosage imbalance of the chromatin remodeler *CHD1L* contributes to the 1q21.1 CNV-associated mirrored neurodevelopmental phenotypes. Lemée, M.
12:00   247. Bi-allelic pathogenic variants in *TMEM147* cause moderate to profound intellectual disability with dysmorphic facial features. Thomas, Q.

S40. Polygenic prediction of complex disease (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator: Zhang, H.
Moderator: Eissman, J.

10:45   250. Genome-wide meta-analysis identifies novel loci and improves disease prediction of Age-related Macular Degeneration. He, W.
11:00   251. Incorporating genetic information into clinical prediction models: Assessment of technical considerations and value in Crohn's disease. Morley, T.
11:15   252. Impacts of individual uncertainty in polygenic risk score estimation on lung cancer risk stratification and prediction. Wang, X.
11:30   253. Evaluation of type 2 diabetes polygenic risk scores in prospective longitudinal cohort study in Korean population. Kim, N.
11:45   254. Integrating large scale genetic and clinical information to predict cases of heart failure. Wu, K.
12:00   255. Investigating genetic drivers of cardiovascular disease variability in admixed individuals. Tietz, G.

S41. Populations evolving: Modeling genetic variation to understand evolutionary processes (Platform)
Conv Ctr/West Hall A/West Building
Moderator: Ochoa, A.
Moderator: Gao, Z.

10:45   258. Inference of coalescence times and allele ages using deep neural networks identifies signatures of natural selection. Tsangalidou, Z.
11:00   259. Genetic evidence for ancient population shifts and migrations in Central and Southern California. Nakatsuka, N.
11:15   260. 1,000 ancient genomes uncover 10,000 years of natural selection in Europe. Le, M.
11:30   261. Causal effects on complex traits are similar across local ancestries within admixed individuals. Hou, K.
11:45   262. The cumulative effects of natural selection on protein truncating variants is associated with fecundity, mortality, morbidity, and life expectancy in humans. Vy, H.
12:00   263. A mutation rate model at the basepair resolution identifies new mutagenic effects and improves population genetics inference. Lee, D.

S42. The methylome and transcriptome of complex traits (Platform)
Conv Ctr/Room 515/West Building
Moderator: Harripaul, R.
Moderator: Trajanoska, K.

10:45   266. Single-cell transcriptional hallmarks of Alzheimer's disease across 427 individuals. Tanigawa, Y.
11:00   267. A cell-type-specific enhancer-gene map built from multimodal assay of RNA and ATAC-seq in 160,000 single cells pinpoints causal variants and genes in human diseases. Sakaue, S.
11:15   268. Combining comparative DNA methylation profiling and patient EHR data to characterize enhancer-gene networks in T2D genetic risk. Agan, V.
11:30   269. Universal DNA methylation age across mammalian tissues. Lu, A.
11:45   270. Fine mapping of Alzheimer's disease susceptibility loci prioritizes functional variants within myeloid cell enhancers. Ravi, A.
12:00   271. Molecular mechanisms of healthy blood aging at single-cell resolution. Bader, E.

S43. Using omics to dissect GWAS signals (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator: Manning, A.
Moderator: Justice, A.

10:45   274. Proteogenomic analysis in UK Biobank identifies potential proteomic consequences of genetic susceptibility to Parkinson's disease. Robins, C.
11:00   275. Identifying potential causal genes by integrating molecular intermediate phenotypes and GWAS signals in psychiatric disorders. Gedik, H.
11:15   276. Deconvolution of bulk RNA-seq reveals cell-type specificity mechanism in Alzheimer's disease. Wen, J.
11:30   277. Large-scale transcriptome-wide association study identifies novel candidate genes underlying prostate-specific antigen levels. Chen, D.
11:45   278. Applications of Individual-level Imputed Transcriptomes in the UK Biobank. Wang, X.
12:00   279. A multi-ancestry genome-wide meta-analysis, fine-mapping, and target gene prioritization to characterize the genetic architecture of adiponectin. Sarsani, V.

S44. Voices from the community (Platform)
Conv Ctr/Petree C/West Building
Moderator: Applegate, C.
Moderator: Sanghavi, K.

10:45   282. The Silent Genomes 'Precision Diagnosis Study': A strategy to improve access to translational genomics research for Indigenous Peoples in Canada. Jacob, K.
11:00   283. Assessing Vietnamese American patient views toward incorporating genomics in primary care: A community engaged research approach. Lemke, A.
11:15   284. The perspective of community gatekeepers on genomic risk information in the context of orofacial cleft in an African population. Oladayo, A.
11:30   285. Participants as partners: how research participants guide the approach for returning genetic health-related information in the All of Us Research Program. Neben, C.
11:45   286. Effectiveness of the *Family Heart Talk* communication tool in improving family member screening for dilated cardiomyopathy: Results of a randomized trial. Kinnamon, D.
12:00   287. Creation of a Justice, Equity, Diversity and Inclusion Action Plan to inform the National Society of Genetic Counselors' organizational goals. Zierhut, H.

12:15 PM - 1:45 PM

Lunch Break: Explore the Exhibit Hall, View Posters, and Join Industry Sessions
Conv Ctr/Exhibit/Poster Hall/South Building

Behind-the-Scenes: ASHG Publications Workshop (Admission by advance ticket purchase only)
Conv Ctr/Room 408/South Building

ClinGen Resource Luncheon (Ancillary Event)
Conv Ctr/Room 406/South Building

NHGRI: Building a Diverse Workforce – Listening to the Voices of Trainees and Early-Stage Scientists (Ancillary Event)
Conv Ctr/Room 404/South Building

Simons Foundation: SPARK Medical Genetics Committee Meeting (Ancillary Event)
Conv Ctr/Room 407/South Building

12:30 PM - 1:00 PM

CoLab Session Presented by Bio-Rad Laboratories: Advanced Multiplexing with the New QX600 Droplet Digital PCR System (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by Jumpcode Genomics: CRISPR depletion in single cell analysis: Improvements in discovery and sequencing efficiency (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Scale Biosciences: Combinatorial indexing methods for high-throughput, cost-effective single-cell sequencing library preparation (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

12:30 PM - 1:30 PM

Industry Education Session Presented by BGI Genomics: Cancer Multiomics & Alzheimer's Disease Spatial Transcriptomics (Industry Solutions)
Conv Ctr/Room 304/South Building

Industry Education Session Presented by Children's Hospital of Philadelphia (Industry Solutions)
Conv Ctr/Room 402/South Building

Industry Education Session Presented by Olink Proteomics (Industry Solutions)
Conv Ctr/Room 306/South Building

Industry Education Session Presented by Oxford Nanopore Technologies: Unravelling complex human genomes – one sequencing platform for all your biology (Industry Solutions)
Conv Ctr/Room 411/South Building

Industry Education Session Presented by Singular Genomics (Industry Solutions)
Conv Ctr/Room 301/South Building

Industry Education Session Presented by Thermo Fisher Scientific (Industry Solutions)
Conv Ctr/Room 303/South Building

1:15 PM - 1:45 PM

CoLab Session Presented by Beckman Coulter Life Sciences & University of Michigan: Improving high-throughput genomic protocols with statistical design of experiments (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Cellecta: Approaches for the Discovery of Drug Targets, Resistance Mechanisms and Biomarkers (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Resolve Biosciences: Functional biology occurs in 3 dimensions, so should transcriptomic analysis. Molecular Cartography at nanometer resolution throughout the entire cell (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

1:45 PM - 2:45 PM

S45. All of Us for All of You (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator: Tsosie, K.
Moderator: Goleva, S.

13:45   290. Genomic data in the *All of Us* Research Program: Advancing precision medicine for all. Bick, A.
14:00   291. Considerations for meaningful collaboration with Tribal Nations and self-identified American Indians and Alaska Natives in the *All of Us* Research Program. Hahn, M.
14:15   292. Frequency of known pathogenic variants across ancestries in the All of Us Research Program cohort. Venner, E.
14:30   293. Pharmacogenomic investigation of SSRI or SNRI-induced SIADH in *All of Us*. Mo, H.

S46. Genetics of substance use disorders (Platform)
Conv Ctr/Room 502/West Building
Moderator: Zhou, H.
Moderator: Duncan, L.

13:45   296. Cross-ancestry meta-analysis of tobacco use disorders based on electronic health record data uncovers novel loci and reveals associations with numerous health outcomes. Jennings, M.
14:00   297. Exome-wide association study in 750,000 individuals identifies CHRNB2 as a potential drug target for smoking. Rajagopal, V.
14:15   298. Multi-omics insights into the biological mechanisms underlying gene-by-smoking and gene-by-alcohol consumption interactions. Bentley, A.
14:30   299. Polygenic scores for tobacco use provide insights into disease associations in a diverse EHR-linked biobank. Venkateswaran, V.

S47. Novel methods for rare variants (Platform)
Conv Ctr/Room 515/West Building
Moderator: Pottinger, T.
Moderator: Daley, D.

13:45   302. Heritability and genetic architecture of rare coding variation across 394,000 exomes. Nadig, A.
14:00   303. Rare variant polygenic risk scores. McRae, J.
14:15   304. PheWAS of ultra-rare deleterious variant burden in the UK Biobank: Insights into hundreds of complex traits with high locus heterogeneity. Carss, K.
14:30   305. Exome sequencing analysis of 14 diseases studied by the GBMI identifies likely effector genes of GWAS signals. Gurski, L.

S48. Reading between the reads: Getting the most out of sequencing results (Platform)
Conv Ctr/West Hall A/West Building
Moderator: Reish, O.
Moderator: Gilissen, C.

13:45   308. Seeing beyond the target: leveraging off-target reads in targeted clinical tumor sequencing to identify prognostic biomarkers and survival. Patel, Y.

14:00   309. Exome versus panel germline sequencing for pediatric cancer predisposition syndromes: Is more always better?. Desrosiers, L.
14:15   310. Integrated DNA and RNA analysis of hereditary cancer associated genes: More answers for more patients. Nykamp, K.
14:30   311. Whole genome sequencing in rare disease diagnosis: Update from the completion of the UK 100,000 Genomes Project. Rendon, A.

S49. RNA based mechanisms in neuropsychiatric disorders (Platform)
Conv Ctr/Petree D/West Building
Moderator: Schaffer, A.
Moderator: Yang, C.

13:45   314. Isoform-level transcriptome-wide association studies uncover novel mechanisms underlying genetic associations with neuropsychiatric disorders. Bhattacharya, A.
14:00   315. Spliceosome malfunction causes neurodevelopmental disorders with autistic features. Li, D.
14:15   316. The dynamic FMRP-RNA interactome across the human lifespan and brain regions. Lee, A.
14:30   317. Novel genes associated with the severe autism subphenotype disproportionate megalencephaly. Nishizaki, S.

S50. The current environment for gene-environment interactions (Platform)
Conv Ctr/Petree C/West Building
Moderator: Tovar, A.
Moderator: North, K.

13:45   320. Using polygenic scores to detect gene-environment interactions associated with human complex traits. Poyraz, L.
14:00   321. Gene-environment interaction between *TRIP4 l*ocus and air pollution exposure influences risk of coronary artery disease. Hartiala, J.
14:15   322. Genome-wide gene-environment interaction scans reveal novel putative drug targets: an Alzheimer's disease study. Zhang, Y.
14:30   323. Evidence of gene x environment interactions acting on body mass index using genotyping and whole exome sequencing data. Dawes, A.

S51. Using EHRs to withdraw new insights from biobanks (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator: Fritsche, L.
Moderator: Shoaib, M.

13:45   326. Variability in lifetime risk of 20 complex diseases across European countries and polygenic score strata in over 1 million individuals. Wolford, B.
14:00   327. Neuroimaging-guided PheWAS using electronic health records from UK Biobank and eMERGE cohorts yield novel associations between brain imaging and disease outcomes. Veturi, Y.
14:15   328. Leveraging genomic diversity for discovery in an EHR-linked biobank: The UCLA ATLAS Community Health Initiative. Johnson, R.
14:30   329. Phenome-wide association study of polygenic risk for central adiposity. Josyula, N.

3:00 PM - 4:45 PM – Poster Session II

Poster authors will be at their boards on Thursday from 3:00 to 4:45 pm. See pages <mark>xx-xx</mark> for Thursday listings.

3:00 PM - 3:30 PM

CoLab Session Presented by BioSkryb Genomics: Uniting WGA and WTA from a Single Cell: ResolveOME™ Chemistry Plus BaseJumper™ Software Unlock Unprecedented Discoveries in a Simple Application (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by CENTOGENE: Ready to Revolutionize a Patient's Life With MOx, Our Multiomic Solution? (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Olink Proteomics (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

3:45 PM - 4:15 PM

CoLab Session Presented by Thermo Fisher Scientific (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by Vizgen: In situ single cell transcriptomic imaging in FFPE tissues with MERSCOPE (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Watchmaker Genomics: Sensitivity Meets Speed: Improved gene detection from low input and FFPE RNA utilizing a rapid NGS library prep workflow (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

4:30 PM - 5:00 PM

CoLab Session Presented by Agilent Technologies: Maximizing the Power of Exome Sequencing for Clinical Research (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Arima Genomics: A New Frontier for Discovery – How 3D genomics reveals novel disease mechanisms and therapeutic targets missed by other technologies (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

CoLab Session Presented by PacBio: The best keeps getting better – product portfolio updates from PacBio (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

5:00 PM - 5:15 PM

S52. Awards Recognition III
Conv Ctr/West Hall A/West Building

President Charles Rotimi will present the 2022 Advocacy Award and Mentorship Award. He will also recognize the 2020 and 2021 recipients of the awards. ASHG Awards Committee Chair France Gagnon will announce the 2022 winners of the Charles J. Epstein Awards for Excellence in Human Genetics Research

5:15 PM - 7:00 PM

S53. Featured Plenary Abstract Session III
Conv Ctr/West Hall A/West Building
Moderator. Civelek, M.
Moderator. Campbell, C.

17:15   335. Systematic analysis of nonsense-mediated decay escaping protein-truncating variants in 97,728 clinical exomes identifies new Mendelian disease genes. Torene, R.
17:35   336. The correlation between CpG methylation and gene expression is driven by sequence variants. Sigurpalsdottir, B.
17:55   337. Australia's National Centre for Indigenous Genomics enabling the inclusion of First Nations peoples in genomics. Patel, H.
18:15   338. Novel therapeutic target discovery using circulating proteins in up to 42,000 UK Biobank participants through systematic Mendelian randomization and genetic colocalization. Chen, L.
18:35   339. A deep catalog of protein coding variation from one million individuals. Balasubramanian, S.

7:00 PM - 8:00 PM

RELAGH: The Latin-American and Caribbean Networking Initiative for Developing Human Genetics and Genomics Meeting (Ancillary Event)
Conv Ctr/Room 505/West Building

7:00 PM - 9:00 PM

Association of Chinese Geneticists in America (ACGA) Annual Meeting (Ancillary Event)
Conv Ctr/Room 406/South Building

CWRU, University Hospitals and Cleveland Clinic Reception (Ancillary Event)
JW Marriott Hotel/Platinum I/3rd floor

Emory University Human Genetics Reception (Ancillary Event)
JW Marriott Hotel/Platinum A/3rd floor

Ohio State's Scarlet and Gray Reception (Ancillary Event)
JW Marriott Hotel/Platinum J/3rd floor

Rare Bone Disease Alliance: Treatment of Rare Bone Diseases from Bench to Bedside (Ancillary Event)
Conv Ctr/Room 404/South Building

Regeneron Genetics Center Reception (Ancillary Event)
JW Marriott Hotel/Gold 3/3rd floor

UCSF Alumni & Friends 2022 Reception (Ancillary Event)
JW Marriott Hotel/Platinum I/3rd floor

University of Chicago Dessert Reception (Ancillary Event)
JW Marriott Hotel/Platinum C/3rd floor

Friday, October 28, 2022

7:00 AM - 8:00 AM

Industry Education Session Presented by MGI / BGI: MGI DNBSEQ technology redefines the sequencing possibilities and expands the horizon of your sequencing capacities (Industry Solutions)
Conv Ctr/Room 304/South Building

8:30 AM - 10:00 AM

S54. ASHG/ESHG Building Bridges: Complete Genomes Require New Directions to Democratize Data (Invited)
Conv Ctr/Room 515/West Building
Moderator. Davis, E.
Moderator. Reymond, A.

08:35   342. Expanding studies of global genomic diversity with complete, telomere-to-telomere (T2T) assembly of diploid genomes. Miga, K.
09:00   343. The assembly of a human pangenome. Li, H.
09:25   344. The NIH Comparative Genomics Resource. Schneider, V.
09:50   Panel Discussion

S55. Long-read RNA-seq to illuminate splice-driven mechanisms of human genetic diseases (Invited)
Conv Ctr/Concourse Hall F/West Building
Moderator. Sheynkman, G.
Moderator. Castaldi, P.

08:40   348. Long-read RNA-seq of human microglia reveals novel isoforms and elucidates Alzheimer's-associated genetic regulation of splicing. Humphrey, J.
08:55   349. Interpreting the effects of GWAS loci on splicing using long-read RNA-seq. Abood, A.
09:10   350. Evidence for high levels of mis-annotation of protein coding genes known to have an associated pseudogene revealed by long-read RNA-sequencing. Ryten, M.
09:25   351. A more comprehensive landscape of RNA alterations in cancer with long-read sequencing. Brooks, A.
09:40   Questions and Answers   , .

S56. Novel preclinical models of Down syndrome elucidate the complex genetic etiology of neurodevelopmental phenotypes (Invited)
Conv Ctr/Petree D/West Building
Moderator. Thyme, S.
Moderator. Moyer, A.

08:31   354. Leveraging C. elegans for an unbiased screen of human 21st chromosome gene overexpression. Sanchez, S.
08:46   355. Elevated levels of Down syndrome cell adhesion molecule cause presynaptic overgrowth in Drosophila and mouse models of Down syndrome. Ye, B.
09:01   356. New genetic models of Down syndrome in mouse and rat. Reeves, R.
09:16   357. Down syndrome induced senescence disrupts the nuclear architecture and function of neural progenitors. Meharena, H.
09:31   Questions and Answers

S57. The impact of ascertainment, phenotyping, and population structure on human genetic research (Invited)
Conv Ctr/Petree C/West Building

Moderator. Polimanti, R.
Moderator. Yengo, L.

08:31   360. Human genetics provides insights into participation to research studies and nonresponse to questionnaires. Ganna, A.
08:46   361. The impact of socioeconomic status in the polygenic risk of psychiatric traits and disorders: evidence of assortative mating in UK Biobank. Cabrera Mendoza, B.
09:01   362. Year of birth bias in the association of polygenic score within the PsycheMERGE network. Niarchou, M.
09:16   363. Using polygenic scores distribution to detect ascertainment in observational studies. Campos, A.
09:31   Questions and Answers

S58. To report or not to report: The quandary of variants of uncertain significance (VUSs) (Invited)
Conv Ctr/Concourse Hall E/West Building
Moderator. Rehm, H.
Moderator. Callier, S.

08:31   366. Challenges of communicating VUSs from genetic testing results. Hanson-Kahn, A.
08:46   367. The landscape of VUSs in a large clinical cohort undergoing molecular testing for hereditary disease. McKnight, D.
09:01   368. Not all VUSs are created equal. Rehm, H.
09:16   369. Reporting and return of results involving VUSs: Opinions from genetics providers. Saitta, S.
09:31   Questions and Answers

S59. Upset the set up: Moving from community engagement to community empowerment (Invited)
Conv Ctr/Room 502/West Building
Moderator. Martschenko, D.
Moderator. Smith, M.

08:31   372. Decolonizing DNA Through Storytelling. Jeff, J.
08:46   373. Bezos to Bottlenecks: The Chasm between Scientific Altruism & Extraction from the Amerindigenous. Yracheta, J.
09:01   374. LGBTQIA+ Community Engagement with Genomic Studies on Same-sex Sexual Behavior: Lessons. Learned   Wedow, R.
09:16   375. From Objects to Subjects: Anti-Ableism and Community Engagement in Precision Medicine Research. Sabatello, M.
09:31   Questions and Answers

10:00 AM - 10:30 AM

CoLab Session Presented by DISPENDIX (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by PacBio: Solving the unsolvable and sequencing the unsequenceable – translational impact of PacBio technologies (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by TALG: Clinical Trial Patient Privacy Considerations for Sponsors and CRO(s) selection (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

10:30 AM - 12:00 PM

S60. Computational approaches for understanding noncoding variants (Platform)
Conv Ctr/Room 502/West Building

Moderator. Quillen, E.
Moderator. Neale, B.

10:30   378. A genome-wide mutational constraint map quantified from variation in 76,156 human genomes improves the functional interpretation of non-coding regions. Chen, S.
10:45   379. Trans-PCO: a powerful approach to detecting *trans*-QTLs associated with regulatory networks. Wang, L.
11:00   380. Performing TWAS on proteomic data to understand *cis* and *trans* gene regulatory mechanisms underlying complex traits. Wittich, H.
11:15   381. Modeling tissue and gene co-regulation reveals causal tissues for disease. Amariuta, T.
11:30   382. Genetically predicted metabolite levels identify candidate pathways for common diseases. Nyasimi, F.
11:45   383. Multi-biofluid metabolome-wide association study illuminates the molecular basis in complex trait genetics. Huang, K.

S61. Extended applications of polygenic risk scores (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator. Harris, J.
Moderator. Adeyemo, A.

10:30   386. The UK Biobank polygenic risk score release. Plagnol, V.
10:45   387. Can genetic associations for disease onset be used to predict disease prognosis? The glass is half empty. Mars, N.
11:00   388. An atlas of associations between plasma protein biomarkers and polygenic risk scores for complex human diseases. Chasioti, D.
11:15   389. Selection, optimization, and validation of 10 polygenic risk scores for clinical implementation in diverse populations. Kenny, E.
11:30   390. The contributions of rare inherited and polygenic risk to autism spectrum disorder in multiplex families. Cirnigliaro, M.
11:45   391. Germline polygenic risk scores predict cancer patient response and treatment failure. Kurant, D.

S62. Genetics and integrated-omics of diabetes and associated metabolic disorders in diverse populations (Platform)
Conv Ctr/Room 515/West Building
Moderator. Baier, L.
Moderator. Voight, B.

10:30   394. Sequencing of 448 Greenlandic Inuit reveals a unique genetic architecture exemplified by a common high-impact *HNF1A* variant affecting type 2 diabetes. Stæger, F.
10:45   395. Trans-ancestry GWAS meta-analysis of random glucose provides insights into diabetes pathophysiology, complications, and treatment stratification. Kaakinen, M.
11:00   396. Integrating transcriptomics, metabolomics, and GWAS helps reveal molecular mechanisms for metabolite levels and disease risk. Yin, X.
11:15   397. Integrated ca/eQTL analyses uncover diabetes GWAS variants modulating human islet proinflammatory cytokine responses. Bhuiyan, R.
11:30   398. New common genetic loci for gestational diabetes mellitus reveal a distinct genetic architecture from type 2 diabetes mellitus. Elliott, A.
11:45   399. Identification of paradoxical genetic metabolic effects in Europeans, South Asians and Africans using 450,000 exomes and 150,000 whole genomes. Hawkes, G.

S63. Hearing and seeing the advancements in auditory and vision genetics (Platform)
Conv Ctr/Petree C/West Building
Moderator. Morton, C.
Moderator. De Baere, E.

10:30   402. *Chd7* and *Sox2* cooperate to regulate development of the inner ear. Martin, D.
10:45   403. Syntaxin 4 is essential for hearing in human and zebrafish. Bharadwaj Mathilakath, T.

11:00   404. Whole genome sequencing for rare variants and imaging analyses in model organisms identify *SLC16A8* as a significant contributor for age-related macular degeneration risk. Nouri, N.
11:15   405. Implication of a non-coding variation of *FOXE3* in an individual displaying a complex microphthalmia. Fares Taie, L.
11:30   406. Beyond Mendelian Inheritance: An approach to digenic inheritance in cases of retinitis pigmentosa, deafness and Alport syndrome in a cohort of Colombian patients. Galvez, M.
11:45   407. *FOXP4*: A novel candidate gene for angle closure glaucoma. Presley, W.

S64. New molecular and analytical tools to study Mendelian disorders (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator. Girirajan, S.
Moderator. Grochowski, C.

10:30   410. PhenoScore: AI-based phenomics to classify genetic variants. Dingemans, A.
10:45   411. A new locus for craniosynostosis caused by copy number variants flanking *FOXD3.* Wilkie, A.
11:00   412. MARRVEL-AI: Knowledge based artificial intelligence for variant pathogenicity prediction for Mendelian disorders. Mao, D.
11:15   413. Determining the parent of origin of homologous chromosomes without parental data. Hanlon, V.
11:30   414. The use of optical genome mapping in genetically unsolved neurodevelopmental disorders. Schrauwen, I.
11:45   415. Reducing uncertainty and improving diagnostic yield in the MSSNG cohort using DNA methylation signatures. Choufani, S.

S65. Population screening: From patient identification to return of results (Platform)
Conv Ctr/West Hall A/West Building
Moderator. Riggs, E.
Moderator. Madden, E.

10:30   418. Assessing a legal pathway to implement a cascade traceback screening program for ovarian cancer. Wagner, J.
10:45   419. The Precision Population Health Initiative, scaling genomic medicine into population health. Green, R.
11:00   420. Geno4ME: Establishment of an equitable whole-genome sequencing-based platform for clinical screening in a large healthcare system. Wagner, J.
11:15   421. Genome-first evaluation with exome and clinical data uncovers underdiagnosis of genetic disorders in a large healthcare system. Forrest, I.
11:30   422. Tests and procedure rates following return of medically actionable monogenic variants within the eMERGE cohort. Linder, J.
11:45   423. Addition of chatbot to the return of genetic results process for biobank participants. Larson, N.

S66. Towards the defeat of neurodegenerative disease (Platform)
Conv Ctr/Petree D/West Building
Moderator. Feliciano, P.
Moderator. Chahrour, M.

10:30   426. Polygenic architecture of a novel MRI endophenotype: "The hemochromatosis brain". Loughnan, R.
10:45   427. Immune system and blood-brain barrier-wide biomarker analyses provide causal evidence for autoimmunity in dementia causing diseases. Lindbohm, J.
11:00   428. Genetic analysis of multiple sclerosis severity identifies a novel locus and implicates CNS resilience as a major determinant of outcome. Harroud, A.
11:15   429. Single-cell multi-region dissection of Alzheimer's disease progression reveals selectively vulnerable neurons. Boix, C.
11:30   430. Regional genetic correlations highlight relationships between neurodegenerative diseases and the immune system. Lona Durazo, F.
11:45   431. Cell-type transcriptome-wide association studies and fine-mapping via deconvolution using single-cell RNA-seq. Shi, H.

12:15 PM - 1:45 PM

Lunch Break and Poster Viewing: Explore the Exhibit Hall, Posters and Network
Conv Ctr/Exhibit/Poster Hall/South Building

Johns Hopkins University: Epic Genomic Indicators Braintrust (Ancillary Event)
Conv Ctr/Room 407/South Building

12:15 PM - 12:45 PM

CoLab Session Presented by Fluent Biosciences (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Gencove (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Oxford Nanopore Technologies: It takes two – Haplotype specific identification of genetic and epigenetic variation using Nanopore sequencing (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

12:30 PM - 1:45 PM

How to Get Started Engaging Under-Represented Communities in Genetics & Genomics Research (Admission by advance ticket purchase only)
Conv Ctr/Room 403/South Building

12:30 PM - 1:30 PM

Industry Education Session Presented by GSK: Genomic Sciences at GSK – Driving Drug Discovery and Development (Industry Solutions)
Conv Ctr/Room 304/South Building

Industry Education Session Presented by Luna Genetics (Industry Solutions)
Conv Ctr/Room 303/South Building

Industry Education Session Presented by PhenX Toolkit: Using the PhenX Toolkit to design studies optimized for data sharing (Industry Solutions)
Conv Ctr/Room 306/South Building

Industry Education Session Presented by SOPHiA Genetics: Fully integrated workflows to accurately interpret genomic variants associated with rare and inherited diseases (Industry Solutions)
Conv Ctr/Room 301/South Building

1:00 PM - 1:30 PM

CoLab Session Presented by ASHG Impact Partners: Broadening access to and participation in genetic testing and research: a discussion about barriers and potential solutions (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 2

CoLab Session Presented by Genomenon & Inozyme Pharma: Curating the Genome to Drive Precision Diagnosis in Clinical Care for ENPP1 Deficiency (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 1

CoLab Session Presented by Spectrum Solutions (Industry Solutions)
Conv Ctr/Exhibit/Poster Hall/CoLab 3

2:00 PM - 3:30 PM

S67. Context matters! Tissue, cell type, and condition-specificity of epigenetics and expression (Platform)
Conv Ctr/Petree D/West Building
Moderator. Battle, A.
Moderator. Ahituv, N.

14:00   434. Application of RNA sequencing on transdifferentiated patient fibroblasts for genetic diagnosis of neurological disorders. Li, S.
14:15   435. Cell-type specific DNA methylome signatures reveal epigenetic mechanisms for neuronal diversity and neurodevelopment disorder. Jin, Y.
14:30   436. Single-nucleus multiomics reveal enrichment of disease heritability in cell-state-dependent regulatory elements. Gupta, A.
14:45   437. Leveraging human genetic variation and single cell RNA-seq to reveal the catalog of influenza-induced splicing QTLs. Wang, L.
15:00   438. Cell type specific and disease associated eQTLs in the human lung. Natri, H.
15:15   439. Brain cell-type-specific protein interactomes of rare variant schizophrenia risk genes highlight shared biology with neurodevelopmental disorders. Hsu, Y.

S68. Leveraging population genetics to inform diverse cohorts and biobanks (Platform)
Conv Ctr/West Hall A/West Building
Moderator. Gopalan, S.
Moderator. Atkinson, E.

14:00   442. Advancing fine-scale population health monitoring systems in a Los Angeles biobank. Caggiano, C.
14:15   443. A comprehensive genetic profile of 140,000 adults from the Mexico City Prospective Study. Torres, J.
14:30   444. Analysis of a large cohort of admixed Greenlandic siblings shows that genetic load of metabolic phenotypes differs between Inuit and Europeans. Lin, L.
14:45   445. Genomic analysis of 15,154 individuals from India: A largest study to-date to explore understudied Indian sub-populations. Kapoor, M.
15:00   446. Integrative functional genomic analyses identify novel genetic variants influencing skin pigmentation in Africans. Feng, Y.
15:15   447. Multiadaptive shrinkage improves cross-population transcriptome prediction for transcriptome-wide association studies in underrepresented populations. Araújo, D.

S69. Methods and databases: Open, benchmarked and FAIR (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator. Eulalio, T.
Moderator. Wenz, B.

14:00   450. Comparing open-source tools to detect alternative splicing and gene expression outliers in RNA-seq to improve diagnostic yield. Jaramillo Oquendo, C.
14:15   451. GestaltMatcher Database: A FAIR database for medical imaging data of rare diseases. Lesmann, H.
14:30   452. Polygenic risk score prediction accuracy: A retrospective analysis. Aschard, H.

14:45   453. The ENCODE 4 long-read RNA-seq resource reveals distinct classes of isoform diversity. Reese, F.
15:00   454. Evaluating gene prioritization methods and their ability to identify successful drug targets. Sadler, M.
15:15   455. The Association to Function Knowledge Portal: An open-access resource for translating variant associations to biological knowledge. Brandes, M.

S70. Molecular investigations into disease mechanisms (Platform)

Conv Ctr/Room 515/West Building
Moderator. Li, Y.
Moderator. Antonellis, A.

14:00   458. Long-read sequencing and profiling of RNA-binding proteins reveals the pathogenic mechanism of aberrant splicing of an *SCN1A* poison exon in individuals with epilepsy. Happ, H.
14:15   459. 5,510 bp deletion upstream of NCK2 is associated with Alzheimer's disease and deletes 6 CREs in microglia PU.1 super-enhancer. Farrell, J.
14:30   460. Genomic discovery and functional validation of MRP1 as a novel therapeutic target for sickle cell disease. Lessard, S.
14:45   461. DNA sequence is the primary determinant of R-loop formation across genomes. Walker, L.
15:00   462. Chromatin accessibility is a potential factor contributing to the lower risk effect of the *APOE* ε4 allele in individuals of African ancestry. Young, J.
15:15   463. Bi-allelic variants in SPOUT1, an RNA methyltransferase functioning in spindle organization, cause a novel neurodevelopment disorder. Dharmadhikari, A.

S71. Preconception carrier screening and rapid WGS of newborns (Platform)

Conv Ctr/Room 502/West Building
Moderator. Chaudhari, B.
Moderator. Actkins, K.

14:00   466. Outcomes of reproductive genetic carrier screening in 9,111 couples screened for 1,281 autosomal and X-linked genes: results from the Australian Reproductive Genetic Carrier Screening Project ("Mackenzie's Mission"). Kirk, E.
14:15   467. Carrier rates for prenatal-lethal genomic variants: A case for expanded genetic screening. Aminbeidokhti, M.
14:30   468. The PREGCARE study: Personalized recurrence risk assessment following the birth of a child with a pathogenic *de novo* mutation. Goriely, A.
14:45   469. A genome sequencing system for universal newborn screening, diagnosis, and precision medicine for severe genetic diseases. Mowrey, W.
15:00   470. Experiences of rapid whole genome sequencing (rWGS) at Baylor Genetics: a powerful and comprehensive first-tier genetic test. Dai, H.
15:15   471. SeqFirst: Improving equitable access for precise genetic diagnosis in critically ill infants. Wenger, T.

S72. Sequencing vs panel testing: Is more better? (Platform)

Conv Ctr/Petree C/West Building
Moderator. Strande, N.
Moderator. Byrne, A.

14:00   474. Whole genome sequencing is a powerful "one-stop shop" screening assay for uncovering undiagnosed conditions in apparently healthy pediatric cohort. Balciuniene, J.
14:15   475. An integrated multiomic panel as an excellent tool for the genetic diagnosis of metabolic diseases: Our first 3,720 patients. Pinto Basto, J.
14:30   476. Clinical application of next-generation sequencing for the diagnosis of Mendelian disorders in a highly consanguineous population: results from more than 1400 Iranian families. Abolhassani, A.
14:45   477. Microarray analysis using a comprehensive array with enhanced probe coverage for disease genes and SNP probes. Bi, W.
15:00   478. Whole Genome Sequencing Analyses of 45,090 Individuals Reveal Rare Coding and Noncoding Variants Associated with Kidney Function. Li, Z.

15:15   479. Diagnostic yield of pediatric and prenatal exome sequencing in a diverse population. Slavotinek, A.

S73. Structural variation in population and disease (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator. Yuan, B.
Moderator. Chong, Z.

14:00   482. Resolving the exact breakpoints and sequence rearrangements of large neuropsychiatric copy number variations (CNVs) at single base-pair resolution using CRISPR-targeted ultra-long read sequencing (CTLR-Seq). Zhou, B.
14:15   483. Structural variation analysis on 16,905 whole-genome sequencing data from Alzheimer's Disease Sequencing Project (ADSP). Wang, H.
14:30   484. Detection of complex structural genome variants using ARC-SV and their enrichment inside genes of neurodevelopmental pathways. Hughes, C.
14:45   485. Advances in long-read sequencing and telomere-to-telomere assembly enable discovery of cryptic ring chromosome breakpoints and highlight complex rearrangements of acrocentric p-arms. Mostovoy, Y.
15:00   486. Pangenome graphs for the analysis of rare genetic diseases. Groza, C.
15:15   487. Human Y chromosome - *de novo* assembly and comprehensive analysis of genetic variation across 45 diverse haplotypes. Hallast, P.

4:00 PM - 5:00 PM

S74. Challenges in everyday clinical genetics (Platform)
Conv Ctr/Petree C/West Building
Moderator. Jose, M.
Moderator. Singh, K.

16:00   490. Costs and diagnostic yield of whole genome sequencing in neurodevelopmental disorders. Johansson Soller, M.
16:15   491. Insurance denials and diagnostic rates in a pediatric genomic research cohort. Zion, T.
16:30   492. Whole genome germline sequencing: Its role in general health screening in family practice, the first study in the UK. Jones, A.
16:45   493. The ClinGen General Inborn Errors of Metabolism Gene Curation Expert Panel: Assessing the clinical validity of genes implicated in metabolic disorders. Goldstein, J.

S75. Gee, What A Session! (GWAS) (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator. Seielstad, M.
Moderator. Loh, P.

16:00   496. Multi-ethnic GWAS meta-analysis identifies 17 loci associated with nonalcoholic fatty liver disease that define new disease subtypes, mechanisms, and predict advanced liver disease. Du, X.
16:15   497. Genome-wide meta-analysis identifies novel risk variants and enables polygenic prediction of preeclampsia and gestational hypertension. Honigberg, M.
16:30   498. Genome-wide association study of chronic kidney disease progression in the Million Veteran Program and BioVU. Robinson-Cohen, C.
16:45   499. Trans-ancestry genome wide association of uterine fibroids in All of Us Research Program. Williams, A.

S76. Genetic architecture of adiposity (Platform)
Conv Ctr/Room 515/West Building
Moderator. Grant, S.

Moderator. Buchner, D.

16:00   502. Characterising the circulating proteome of adiposity through use of weight loss interventions and Mendelian randomisation. Goudswaard, L.

16:15   503. Identification of rare functional variants in known genes for monogenic obesity among American Indians with severe obesity. Koroglu Altok, C.

16:30   504. Discovery of novel loci that uncouple adiposity from its cardiometabolic comorbidities in the UK Biobank: towards precision medicine of obesity. Chami, N.

16:45   505. Rare loss of function variants in the hepatokine gene *INHBE* protect from abdominal obesity. Deaton, A.

S77. Genetic variants and cancer risk in diverse populations (Platform)
Conv Ctr/Concourse Hall E/West Building
Moderator. Middha, P.
Moderator. Amos, C.

16:00   508. Risk allele associated with childhood acute lymphoblastic leukemia at the IKZF1 locus is associated with Indigenous American ancestry and absent in European ancestry populations. Jeon, S.

16:15   509. A multi-population African GWAS for prostate cancer reveals novel disease associations and within-continent heterogeneity of cancer risk. Janivara, R.

16:30   510. MeSuSie: a novel method for discovering shared and unique putative causal variants by fine-mapping across diverse ancestries. Gao, B.

16:45   511. Genotype-first breast cancer prevention - experience with transferring monogenic findings from a population biobank to clinical setting. Nõukas, M.

S78. Let me sleep on it (Platform)
Conv Ctr/Room 502/West Building
Moderator. Miller, A.
Moderator. Klimentidis, Y.

16:00   514. Whole-exome sequencing study identified novel genes for self-reported, diagnosis, and accelerometer-based sleep and circadian rhythm traits and disorders. Chen, C.

16:15   515. Enabling PheWAS for psychiatric disorders through deep phenotyping of behavior from smartphones sensors. Balliu, B.

16:30   516. Identification of a putatively adaptive promoter variant in *PRKAA1* in Andean highlanders and associations with hypoxia adaptation. Fassardi, S.

16:45   517. An integrated multi-omics analysis of sleep disordered breathing traits across multiple blood cell types. Sofer, T.

S79. Splicing together the story (Platform)
Conv Ctr/West Hall A/West Building
Moderator. Iyengar, A.
Moderator. Reddy, T.

16:00   520. Single-cell differential splicing of Alzheimer's disease across 1.9 million cells and 416 individuals. Hwa, C.

16:15   521. Introme and SpliceVarDB: Predicting and validating splice-altering variation to improve diagnostic yield. Sullivan, P.

16:30   522. High-throughput assessment of isoform-specific function by targeting exon-exon junctions in a CRISPR/Cas13 forward screen. Isaev, K.

16:45   523. Investigating X-Linked Dystonia-Parkinsonism signatures in post-mortem brain samples and designing ASOs against XDP signatures in neuronal cell models. Yadav, R.

S80. Therapeutic insights leveraged from preclinical models of disease (Platform)
Conv Ctr/Petree D/West Building
Moderator. Al-Hertani, W.
Moderator. Rossignol, F.

16:00   526. CRISPR base editing to treat phenylketonuria. Wang, X.
16:15   527. 4-Phenylbutyric acid obviates ER stress-induced neurodegeneration in the spinal cord of a mouse model of GM2 gangliosidosis. Weaver, F.
16:30   528. Potential therapies for Kir7.1 channelopathy. Kabra, M.
16:45   529. Probucol ameliorates the autoimmune, lipodystrophic and neurodegenerative phenotypes observed in Clec16a KO mice. Pandey, R.

5:30 PM - 6:30 PM

S81. Late Breaking Plenary Abstract Session
Conv Ctr/West Hall A/West Building

Check the online planner and mobile app for listings.

7:00 PM - 9:00 PM

ASHG Friday Socials (ASHG Reception)
Convention Center
Check the online planner for locations and details.

North Carolina Macular Dystrophy: Non-Coding Mutations (Ancillary Event)
Conv Ctr/Room 407/South Building

Saturday, October 29, 2022

7:30 AM - 9:00 AM

S82. How to Use The Human Pangenome Reference in AnVIL (Admission by advance ticket purchase only)
Conv Ctr/Room 403/South Building

S83. Using the Ensembl Variant Effect Predictor (VEP) to interrogate coding and non-coding variants (Admission by advance ticket purchase only)
Conv Ctr/Room 408/South Building

9:00 AM - 10:00 AM

S84. A mix of murine models for mechanistic mapping (Platform)
Conv Ctr/Room 502/West Building
Moderator. Yang, X.
Moderator. Monroe, T.

09:00   540. The ENCODE mouse postnatal developmental time course identifies regulatory signatures of cell type maturation. Rebboah, E.
09:15   541. Muscle-specific knock-out of *Sucla2* exhibits differential cellular and *ex vivo* phenotypes as well as striking functional perturbations *in vivo*, providing a novel model of mitochondrial myopathy. Anderson, M.
09:30   542. *Psmb5* is essential for mouse early embryo development and possibly for zygote formation. Gu, Y.
09:45   543. Profiling three-dimensional nuclear telomeric architecture as a biomarker of myelodysplastic syndromes and acute myeloid leukemia in a mouse model. Gadji, M.

S85. Non-coding variation and cancer (Platform)
Conv Ctr/Concourse Hall F/West Building
Moderator. Cooper, S.
Moderator. Jones, M.

09:00   546. Non-coding aberrations in mismatch repair genes underlie a substantial part of the missing heritability in Lynch syndrome. te Paske, I.
09:15   547. Single cell chromatin accessibility based deep learning models prioritize functional non-coding genetic variants in colorectal cancer GWAS loci affecting distinct cell types. Kotler, E.
09:30   548. Identifying non-coding somatic driver mutations in regulatory elements for pancreatic cancer. Zhong, J.
09:45   549. Oncogenic non-coding RNAs activated by distal enhancers through somatic genome rearrangements in cancer. Yang, L.

S86. Novel approaches to increase the diagnostic yield of genetic disorders (Platform)
Conv Ctr/Room 515/West Building
Moderator. Murray, S.
Moderator. Arboleda, V.

09:00   552. Using a gene pathogenicity tool 'GenePy' identifies missed biallelic diagnoses in the 100,000 Genomes Project. Seaby, E.
09:15   553. Genome reference choice impacts RNA-seq interpretation and rare disease diagnosis. Goddard, P.
09:30   554. Variant effect prediction based on custom long-read transcriptomes improves clinical variant annotation. Salz, R.
09:45   555. Automatically constructed pedigrees accurately identify patients at risk for monogenic and polygenic conditions. Hebbring, S.

S87. The many ways to make polygenic risk scores (Platform)

Conv Ctr/Concourse Hall E/West Building
Moderator. Landry, L.
Moderator. Mathias, R.

09:00   558. The genetic architecture of complex traits and its relevance to polygenic score performance and divergence. Lachance, J.
09:15   559. Increased prediction accuracy for complex traits by combining phenotypic information from known and inferred relatives with polygenic scores. Moore, M.
09:30   560. Addressing overfitting bias due to sample overlap in polygenic risk scoring. Jeong, S.
09:45   561. Potential clinical utility of polygenic risk scores in disease prognosis. The glass is half full. Wanner, J.

S88. Unwrapping the role of chromatin in neurodevelopmental disorders (Platform)

Conv Ctr/Petree C/West Building
Moderator. Greenwald, E.
Moderator. Raj, T.

09:00   564. A framework for summarizing chromatin state annotations within and identifying differential annotations across groups of samples. Vu, H.
09:15   565. *De novo* mutations in replication-independent histone genes and an unexplored class of rare pediatric Mendelian syndromes. Lubin, E.
09:30   566. Epigenetic mechanisms of growth retardation in Kabuki syndrome 2 and correction of disrupted H3K27 methylation as an approach to treatment. Gao, C.
09:45   567. Uncovering the role of EZH1 in neural development and neurodevelopmental disorders. Gracia Díaz, C.

10:30 AM - 11:30 AM

S89. Detection and effect of CNVs in general populations (Platform)

Conv Ctr/Concourse Hall F/West Building
Moderator. Li, Y.
Moderator. Dennis, M.

10:30   570. A high-resolution map of copy number and structural variation in Qatari genomes and their contribution to quantitative traits and disease. Aliyev, E.
10:45   571. Copy number variants differ in prevalence across ancestral populations. Schultz, L.
11:00   572. The impact of rare coding CNVs in 197,306 UK Biobank exomes. Fu, J.
11:15   573. Analysis of gene dosage shows that the majority of the coding genome impacts. Huguet, G.

S90. Expanding roles of repeats (Platform)

Conv Ctr/Concourse Hall E/West Building
Moderator. Stergachis, A.
Moderator. Savatt, J.

10:30   576. Polymorphic short tandem repeats make widespread contributions to blood and serum traits. Margoliash, J.
10:45   577. Phenotypic effects of common coding copy-number variation. Hujoel, M.
11:00   578. Characterization of full-length CNBP expanded alleles in myotonic dystrophy type 2 patients by Cas9-mediated enrichment and nanopore sequencing. Delledonne, M.
11:15   579. Long-read sequencing reveal s*de novo* mutations in repetitive regions of the genome. Noyes, M.

S91. Genetic and functional underpinnings of epilepsy (Platform)

Conv Ctr/Petree C/West Building

Moderator. Russell, B.
Moderator. Breuss, M.

10:30   582. Computational and functional analysis identifies AP3D1 as a potential candidate gene associated with epilepsy. Nagai, T.
10:45   583. Haploinsufficiency of *CHASERR*, a human long non-coding RNA, causes a severe neurodevelopmental disease, implicating dosage sensitivity of *CHD2* in brain development. Ganesh, V.
11:00   584. Missense variants in *RPH3A* cause defects in synaptic function and are associated with a neurodevelopmental disorder characterized by epilepsy. Pavinato, L.
11:15   585. *De novo* and biallelic variants in *R3HDM1*, encoding an Encore-like RNA binding protein hosting the microRNA MiR-128-1, disrupt cortical development and lead to neurodevelopmental phenotypes and epilepsy. Scala, M.

S92. The past matters for cancer treatment and risk (Platform)
Conv Ctr/Room 502/West Building
Moderator. Huang, Y.
Moderator. Chapman Hannah, L.

10:30   588. Examining genetic susceptibility to anthracycline-related cardiomyopathy in cancer survivors using a gene-level approach. Sharafeldin, N.
10:45   589. Cancer predisposition variants and subsequent-malignancy-related late-mortality among long-term survivors of childhood cancer. Chen, C.
11:00   590. Genomic characterization of lymph node metastases in papillary thyroid carcinoma following the Chernobyl accident reveals an expression profile specific to the metastatic process. Lee, O.
11:15   591. Selection acting on somatic structural variation in blood impacts molecular function and cancer risk among humans. Skead, K.

S93. Understanding GWAS signals: From variants to function (Platform)
Conv Ctr/Room 515/West Building
Moderator. Asgari, S.
Moderator. Landstrom, A.

10:30   594. Cross-cohort eQTL fine-mapping utilizing TOPMed whole genome sequencing identifies tens of thousands of independent eQTLs signals and thousands of eQTLs colocalizing with complex trait-associated variants. Orchard, P.
10:45   595. Integrative analysis of metabolite GWAS illuminates the molecular basis of pleiotropy and genetic correlation. Smith, C.
11:00   596. Massively parallel reporter assays of QT interval GWAS enhancer variants. Kapoor, A.
11:15   597. From variants to functions for coronary artery disease: Systematic Perturb-seq links GWAS loci to disease programs in endothelial cells. Engreitz, J.

12:00 PM - 1:00 PM

S94. Thematic Roundtable Discussions Moderated by PC Members
Convention Center, Petree C, Petree D, Room 502, Room 515

Stay through Saturday for the final session of the meeting to discuss the most exciting discoveries presented during the week. Leave ASHG 2022 prepared to tell your colleagues what you learned and how you want to use your this in your work setting. Scientific leaders from the ASHG community will share important breakthroughs and practical takeaways in this energetic, interactive session rounding out the meeting. Look for questions posted on social media to take part in shaping the content for this session. There will be four rooms focused on the following discussion areas:

- Room 1: Basic Science Research: Complex traits
- Room 2: Basic Science Research: Mendelian phenotypes and cancer
- Room 3: Clinical Research: Complex traits

- Room 4: Clinical Research: Mendelian phenotypes and cancer

POSTER LISTINGS
Conv Ctr/Exhibit/Poster Hall/South Building

Poster authors will be at their boards on Wednesday or Thursday from 3:00 to 4:45 pm based on the schedule below.

3:00 PM - 4:45 PM – Wednesday Posters

Cancer Posters - Wednesday (Poster)
PB1001. 3D genomics with Arima Hi-C sequencing enables detection of clinically relevant gene fusions in pediatric cancer samples. Sikkink, K.

PB1003. A B-ALL pediatric patient with IGH rearrangement. Zhao, A.

PB1005. A cross-ancestry meta-analysis identifies 451 susceptibility loci for prostate cancer and yields a genetic risk score effective in multiple populations. Wang, A.

PB1007. A genome-wide gene expression signature of Breast cancer in the Moroccan women. Ayoubi, S.

PB1009. A polygenic risk score for prostate cancer risk prediction. Shi, M.

PB1012. AmpliconClassifier detects the mechanisms of focal genome amplifications in cancer. Luebeck, J.

PB1014. An active learning framework improves tumor variant interpretation. Capra, J.

PB1016. An integrative genomic transcriptomic and proteomic investigation to characterize differences between isogenic radioresistant prostate cancer cell lines. Haas, R.

PB1018. Analysis of the spatial transcriptomic profiles in tumor and tumor microenvironment of colorectal cancer in Alaska Native patients. Thomas, M.

PB1019. ArCCH: Improving the Performance of Clonal Hematopoiesis Variant Calling and Interpretation using a Consensus Based Approach. Chan, I.

PB1021. Assessing the relationship between coding variation and cancer risk in known cancer GWAS susceptibility regions. Hammermeister Suger, A.

PB1023. Association of the rs12587 variant in the KRAS gene in patients with breast cancer and control group of the Mexican population. Garibaldi Rios, A.

PB1025. B cell repertoire analysis in prostate cancer identifies gene expression pattern differences in FC gamma receptors. Reddy, R.

PB1026. Breast cancer genetics in non-Ashkenazi breast cancer patients - carrier rates and variant classification in a genetically diverse population. Levy-Lahad, E.

PB1027. Breast cancer polygenic risk score as a tool to risk stratify participants in the Healthy Nevada Project. Hajek, C.

PB1031. Cancer-driving mutations are enriched in genic regions intolerant to germline variation. Dhindsa, R.

PB1033. Case-control likelihood ratio calculation for clinical classification of *BRCA1* and *BRCA2* variants of uncertain significance. Zanti, M.

PB1035. Cell-intrinsic metabolism and cofactor balancing define the metabolomic impact of NAD+supplementation in pancreatic cancer cell metabolism. Elsea, S.

PB1037. Characterization of nucleosome positions and chromatin interactions of regulatory elements comparing Hi-C, Micro-C, and promoter capture Micro-C. Rhie, S.

PB1039. Chromatin accessibility of primary human cancers ties regional mutational processes and signatures with tissues of origin. Reimand, J.

PB1041. Chromosome Arm Copy Number Profiling using a Clinical, Targeted NGS Panel. Gascoyne, J.

PB1043. *Cis*- and *trans*-eQTL TWAS of breast and ovarian cancer identify more than 100 risk genes in the BCAC and OCAC consortia. Head, T.

PB1045. Clinical and genetic features of patients with multiple endocrine neoplasia type 1 in Korea. Kim, B.

PB1047. Clinicogenomic Neural Networks with Real World Evidence: Predicting Outcomes and Targeting Interventions at a Comprehensive Cancer Center. Flagg, M.

PB1049. -Co-delineating genomic and transcriptomic modes of resistance to MEK inhibition in individual triple negative breast cancer cells. Arvapalli, D.

PB1051. Comprehensive Characterization of Functional Cancer Susceptibility at the 5p15.33 TERT/CLPTM1L Pan-Cancer Risk Locus. O'Brien, A.

PB1053. Comprehensive study of gene expression outliers and their regulation mechanisms in pan-cancer. Han, J.

PB1055. Concurrent Core-binding factor beta subunit (CBFB) gene rearrangement/inv(16)(p13.1q22) in p210 BCR-ABL1 positive chronic myelogenous leukemia. Reid, J.

PB1057. Congenital uterine abnormalities and the risk of gynecological cancers. Abdelmoula, N.

PB1059. Cost-utility of universal screening for common BRCA1 and BRCA2 variants among Ashkenazi Jewish women: a real-life analysis. Michaelson-Cohen, R.

PB1061. Cross disorder genetic analysis identifies autoimmune disease loci inversely associated with diverse cancer types. Chen, J.

PB1064. Deep learning identifies gene expression and DNA methylation as highly predictive of lung cancer histologic subtype. Betti, M.

PB1066. Detecting pleiotropic breast cancer susceptibility variants from genome-wide association studies. Li, X.

PB1068. Detection of APC and MUTYH germline mutations in two south Indian families with hereditary colorectal cancer. Akula, S.

PB1070. Detection of *EGFR* exon skipping variants using clinical sequencing and its application. Kim, S.

PB1072. Determining the mechanisms of radio resistance in breast cancer. McBean, B.

PB1074. Development and Clinical Validation of a Bioinformatics Pipeline for Cancer Transcriptome Sequencing. Cao, K.

PB1076. Development of a breast cancer risk prediction model with carrier status, a polygenic risk score, and epidemiologic risk score. Kraft, P.

PB1078. Diagnostic gene fusion detection in cancer by adaptation of short-read CHOP Fusion Panel to Oxford Nanopore long-read sequencing. Rybacki, K.

PB1080. Differentially expressed serum RNAs as potential early markers for primary sclerosing cholangitis-associated cholangiocarcinoma. Hemmrich-Stanisak, G.

PB1081. Discovery of novel predisposing coding and noncoding variants in familial Hodgkin lymphoma. Myers, J.

PB1083. Dissecting admixture effects on a 313-variant polygenic risk score model for breast cancer in a census-based cohort of Brazilians. Almeida, T.

PB1085. Dosing-specific modes of resistance to quizartinib in individual acute myeloid leukemia cells exposed by ResolveOME combined genomics and transcriptomics chemistry. Velivela, S.

PB1087. EagleC: A deep-learning framework for detecting a full range of structural variations from bulk and single-cell contact maps. Luan, Y.

PB1089. EG VEGF in ovarian cancer and preeclampsia as an early diagnostic marker: toward a targeted therapy in the treatment of tumors, especially those resistant to chemotherapy. Benfateh, M.

PB1091. Elongin C (*ELOC/TCEB1*) gene: a novel cause of von Hippel-Lindau disease. Andreou, A.

PB1094. Evaluating biomarker potential of germline genomic factors for predicting clinical outcomes in prostate cancer. Zeltser, N.

PB1096. Evaluation of non-coding regulatory variants in susceptibility of Chordoma. Yepes Torres, S.

PB1098. Exome-wide discovery of the contribution of coding variants identifies novel genes associated with breast cancer risk. Wilcox, N.

PB1100. Exploring the non-invasive potential of miRNA 145 and miRNA 363 in Prostate Cancer. Manoj, A.

PB1102. Filling gaps in whole genome analysis in hematology: A chance for optical mapping and long-read NGS. Savara, J.

PB1104. Frequently mutated TOP2B binding sites reveal candidate structural and regulatory cancer drivers of hepatocellular carcinoma. Uuskula-Reimand, L.

PB1106. Functional screens implicate candidate breast cancer risk genes in T cell-mediated immune-surveillance. Beesley, J.

PB1108. Gene fusion detection and characterization in long-read cancer transcriptomes with FusionSeeker. Chen, Y.

PB1110. Genetic ancestry correlates with somatic mutations in endometrial cancer. Bremseth-Vining, R.

PB1112. Genetic Modifiers of *KRAS*-Mutant Colorectal Cancer. Pollock, N.

PB1114. Genome and transcriptome landscape of familial pancreatic cancer and implications for altered glucose metabolism in moderate-penetrance cancer susceptibility. Dixon, K.

PB1116. Genome-wide association study identifies 4 novel risk loci including a missense variant in LGR5 for small intestinal neuroendocrine tumors. Kumar, A.

PB1118. Genome-wide profiling of DNA N6methylation from a breast cancer and a matched normal cell lines. Xiao, C.

PB1120. Germline functional non-coding variants associated with ovarian cancer predisposition. Ezquina, S.

PB1122. Germline sequencing in Brazilian pancreatic carcinoma patients. Munhoz Rodrigues, L.

PB1124. Germline variants in the Ah Receptor in Colombian smoker patients with squamous cell carcinoma of the head and neck. Trujillo, N.

PB1126. Going beyond the atlas: Mapping the tumor landscape with complete genomic signatures. Kennedy, K.

PB1128. Hemophagocytosis related to acute promyelocytic leukemia is also positive for the *PML::RARA* gene fusion. García Romero, A.

PB1130. Heterozygous loss of *AURKB* in the germline associates with protection from metastasis and death from cancer. Ward, L.

PB1132. High-throughput functional screening of candidate causal variants from lung cancer susceptibility loci. Patel, H.

PB1134. Identification of target genes and pathways involved in triple-negative breast cancer proliferation and stemness using genome-wide CRISPR screens. Li, T.

PB1136. Identifying molecular markers of early stage ovarian cancer pathogenesis through whole genome CRISPR screening of mutant *BRCA1* fallopian tube cells. Dhungana, S.

PB1138. Impact of polygenic risk, pathogenic variants, and family history on detection of breast cancers within and outside the national mammography screening programme on 117,252 women over 1992-2019. Mars, N.

PB1140. Implementation, utilization, and diagnostic outcomes of a technology-enabled telehealth-based program for individuals to access multi-cancer early detection testing. Weissman, S.

PB1142. Integrating GWAS and 3D chromatin interactome data to identify multi-cancer risk genes in hormone-related cancers. Rivera, S.

PB1145. Isolated multinodular goiter in a family with DICER1 heterozygous germline variant. Dalal, P.

PB1147. Leveraging network architecture and transfer learning for precision oncology. Yi, S.

PB1149. Liquid biopsy for the detection of actionable human variants in the blood of cancer-diagnosed dogs: Opening the door to comparative oncology and therapeutic development. Chorny, I.

PB1151. Low CD2 expression in sentinel lymph nodes of early breast cancer patients is associated with postoperative tumor recurrence or metastasis. Kang, S.

PB1153. Mechanistic insight into HIF1α-Notch1 axis in chronic myeloid leukemia patients and its diagnostic implications. Singh, V.

PB1155. Metagenomics and metabolomics of persistent high-risk Human Papillomavirus (hrHPV) infections and cervical cancer. Adebamowo, C.

PB1157. Mitonuclear genotype remodels the metabolic and microenvironmental landscape of Hürthle cell carcinoma. Liu, E.

PB1161. Mosaic chromosomal alterations detected in HIV infected men and its relationship to non-Hodgkin lymphoma. Khan, S.

PB1163. Multi-modal characterization of ultra-rare germline genetic variants driving breast cancer risk in the indigenous Arab population. Chu, H.

PB1165. Multi-omic single-cell profiling of endogenous and engineered T cells in patients undergoing CAR T cell therapy for high-grade glioma. Banovich, N.

PB1167. Multi-population GWAS analysis identified functional variants in ever- and never-smoking lung cancer. Li, Y.

PB1169. Next Generation SP DNA Sample Prep and DLS Labeling Readies Optical Genome Mapping Workflows for Adoption at Scale. Sadowski, H.

PB1171. Novel Germline Pathogenic Variants of *APC* and *BMPR1A* genes in Algerian patients with Hereditary Polyposis Syndromes. Cherbal, F.

PB1173. OM2BFB: Detecting and elucidating Breakage Fusion Bridge structures in cancer genomes using Optical Mapping data. Raeisi Dehkordi, S.

PB1175. Online implementation of combined polygenetic protein prediction algorithm for oncology (C3PO): Tumor mutation contributions to cancer hallmarks based on proteomics. Caryotakis, G.

PB1178. Pan-Cancer Analysis introduces HLF as a tumor suppressor gene and a promising biomarker for prognosis and targeted therapy in human tumors. Ahmadi, M.

PB1180. Parent-of-origin-aware genomic analysis in the management of patients and families with genetic disease. Schrader, K.

PB1182. Pathogenic Variants in Adult-Onset Cancer Predisposition Genes Identified by Pediatric Tumor Genomic Testing: Prevalence, Tumor Features, and Germline Testing Outcomes. Oak, N.

PB1184. Potential misrepresentation of inherited breast cancer risk by common germline alleles. Letsou, W.

PB1188. Prevalence of suspected pathogenic or likely pathogenic germline *TP53* variants in population sequencing databases: A genotype-first analysis. De Andrade, K.

PB1190. Prognostic Value of Bile Acid Transporter SLC10A1 Expression in Hepatocellular Carcinoma. Chen, H.

PB1192. Rare germline variants in Deubiquitylating enzymes (DUBs) in young women with breast cancer. Grimaldo, L.

PB1195. Resolving clone-and haplotype-specific copy number variation and DNA methylation in heterogeneous tumors with nanopore sequencing. Aganezov, S.

PB1197. Rewired phospho-signaling networks by genetic variants in Clear Cell Renal Cell Carcinoma. Mao, S.

PB1199. Serum biomarkers are altered in UK Biobank participants with mosaic chromosomal alterations. Hubbard, A.

PB1201. Shared predisposition to cancer and metabolic disorders using large-scale genomic data. Pascat, V.

PB1203. Single cell transcriptomics of Pituitary Neuroendocrine Tumors (PitNETs). Brunner, M.

PB1205. Single-cell identity and state coupled with genome-wide SNV and CNV in primary breast cancer with ResolveOME profiling. Zawistowski, J.

PB1207. Single-cell multi-omic characterization of MPAL phenotypes reveals patient heterogeneity associated with poor treatment outcomes. Hayford, C.

PB1209. *SMARCA4* Schwannomatosis: a known cancer susceptibility gene, a new phenotype. Chan-Pak-Choon, F.

PB1211. t(6;9)(p22;q34) translocation with DEK-NUP214 fusion identified in one patient with chronic myeloid leukemia. Zhang, Y.

PB1215. The effect of weight-loss on the colorectal transcriptome and its relation to colorectal cancer risk. Hazelwood, E.

PB1217. The role of senescent tumor cells in cancer progression. Park, T.

PB1219. The use of *TNF-α* polymorphic forms as a possible risk assessment tool for the management of breast cancer among Nigerian women. Alamukii, N.

PB1221. Transcriptome-wide association study identifies new breast cancer susceptibility genes in Latinas. Middha, P.

PB1223. Uncovering gene fusions with 3D genomics: from clinical validation to actionable insights for undiagnosable solid tumors. Schmitt, A.

PB1225. Unraveling the mechanisms behind the malignant behavior of mast cell tumors using laser capture microdissection mediated RNA sequencing on canine models. Vander Plaetsen, A.

PB1227. Using human genetics to evaluate the causal role of circulating inflammatory markers in risk of adult cancer. Yarmolinsky, J.

PB1229. Utilizing Electronic Health Records (EHR) and Tumor Panel Sequencing to Demystify Prognosis of Cancer of Unknown Primary (CUP) patients. Moon, I.

PB1231. Whole genome CpG-resolution DNA methylation profiling of HNSCC reveals previously ignored heterogeneity among HPV(+)tumors and clinical implications. Qin, T.

Evolutionary and Population Genetics Posters - Wednesday (Poster)

PB2754. A Bayesian framework to capture the probability of polymorphism with sequence context across the human genome. Adams, C.

PB2756. A robust kinship inference approach based on machine learning method for missing person identification. Huang, M.

PB2758. Analysis of evolutionary pressure on gene expression identifies patterns of selection in tissues and disease. Sartori, A.

PB2760. Analysis of Latvian genome diversity combining the whole-genome sequencing with genome-wide genotyping data. Klovins, J.

PB2762. Ancestral haplotype prediction using neural networks. Belsare, S.

PB2764. Ancient virome analyses of ancient individuals who lived in the Japanese archipelago. Nishimura, L.

PB2765. Between-population genetic differences for human complex traits. Hivert, V.

PB2767. CAFE: Ancestry and genotype calling uncertainty-adjusted ancestry-specific allele frequency estimation from admixed samples. Wang, J.

PB2769. Challenging continental groupings of humans using a data-driven approach. Palma Martínez, M.

PB2771. *Cis*-regulatory genetic variants influence distinct expression profiles of adipose and muscle tissue transcripts between African and European ancestry individuals. Langefeld, C.

PB2773. Comparing human centromeres with TandemAligner. Bzikadze, A.

PB2775. Conservation and function of the *NXF2* palindrome in male fertility. Lawson, A.

PB2777. Contrasting patterns of positive selection at *Plasmodium falciparum* and *Plasmodium vivax* associated loci in Oman. Haffener, P.

PB2779. Detection of Genomic Regions undergoing Selection in post-Neolithic Transition. Bhattacharyya, C.

PB2781. Discovering determinants of short tandem repeat stability in local sequence context using gradient-based attribution scores. DeVito, R.

PB2783. Evaluation of imputation performance of different reference panels in a Pakistani population. Xu, J.

PB2784. Evidence of potential natural selection in African American individuals post admixture. Jaworski, J.

PB2786. Exploring population health disparities through identity by descent genetic communities in The Biobank at The Colorado Center for Personalized Medicine. Shortt, J.

PB2788. Fine-scale ancestry mapping revealed population-specific associations with complex disease. Isshiki, M.

PB2790. Genetic and phenotypic high-altitude adaptation at *EPAS1/HIF2A* in Andean highlanders. Gu, W.

PB2792. Genetic variation related to the regulation and expression of hemoglobin isoforms among global populations. Hall, J.

PB2794. Genomic deserts: A global survey of underrepresented populations in genomics research. Mangul, S.

PB2796. Germline genetic determinants of T-cell fraction in 110,000 diverse individuals. Poisner, H.

PB2798. Haplotype-based analyses of phylogeny and regional genome diversity among laboratory rats using the latest rat reference genome. Pan, Y.

PB2800. Human populations and demographics in Qatar from the Neolithic to the Late Iron Age. D'Aurelio, A.

PB2802. Imputation around the world: Assessing imputation quality across diverse global populations. Cahoon, J.

PB2805. Inference of the distribution of fitness effects using local genealogical trees. Ortega-Del Vecchyo, D.

PB2807. Inferring between-population genetic variation using admixed populations: challenges and limitations. Huang, J.

PB2809. Insights into non-equilibrium population genetics under strong natural selection from a new theoretical formalism. Balick, D.

PB2811. Introgressed sequences and positive selection profiles of the Japanese population revealed by the analysis of 3,256 Japanese whole-genome sequencing (WGS) data. Tomizuka, K.

PB2813. Investigating selection and the genetics of height in Central African Rainforest Hunter-gatherers. Ju, D.

PB2815. Large-scale analysis of 6,000 whole genomes from Qatar uncovers genetic structure of Arab and Middle Eastern populations and establishes a valuable resource for understanding personalized disease risk and causality   . Naeem, H.

PB2817. Leveraging Base Pair Mammalian Constraint to Understand Genetic Variation and Human Disease. Gazal, S.

PB2819. Lower allele frequency among males is a marker for pathogenicity among non-pseudoautosomal X-chromosome variants. Ciesielski, T.

PB2822. Mosaic chromosomal alterations and longevity. Leshchyk, A.

PB2824. Newfoundland and Labrador: A mosaic founder population of an Irish and British diaspora from 300 years ago. Gilbert, E.

PB2827. Patterns of selection on human gene regulatory variation. Colbran, L.

PB2829. Population-specific Recombination Maps Reduce False Positive Findings in Genomic Scans of Adaptations. Dinh, B.

PB2831. Primate comparative genomics and infectious diseases; COVID-19, Monkey Pox and what comes next. Roodgar, M.

PB2833. Recovering signatures of ghost admixture using ancestral recombination graphs. Biddanda, A.

PB2835. Relating the evolutionary fitness costs of loss-of-function mutations to their pathogenic consequences in humans. Agarwal, I.

PB2837. Resolution of structural variation in diverse mouse genomes reveals dynamic chromatin remodeling due to transposable elements. Ferraj, A.

PB2839. Risk of venous thromboembolism in individuals with supernumerary sex chromosome aneuploidies in two large population-based cohorts. Berry, A.

PB2841. Sequence variants affect the genome-wide rate of germline microsatellite mutations. Kristmundsdóttir, S.

PB2843. Signatures of Mutational Processes in Human DNA Evolution. Ebrahimi, D.

PB2845. Simulating effects from genetic and experimental perturbations to gene regulatory networks. Aguirre, M.

PB2847. South Asian and Greater Middle Eastern Enrichment of *NOTCH3* p.Arg1231Cys Variant and Associated Stroke Risk. Rodriguez-Flores, J.

PB2849. The differential identification of cross-species lung scRNA expression via integrative analysis. Lin, W.

PB2850. The genetic diversity of Japanese populations inferred from the whole genome sequencing data. Kawai, Y.

PB2852. The genomic history of human populations in the North American Central and Southern Plains. Sykora, L.

PB2855. The landscape of tolerated genetic variation in humans and primates. Gao, H.

PB2857. The NHGRI Sample Repository for Human Genetic Research: biospecimens and a new genomic data search tool. Liautaud, A.

PB2859. Tracing the genetic link of Mizo people of India: Genetic & archaeological evidence. Bankura, B.

PB2861. Understanding natural selection in the European Holocene using Ancient DNA. Pandey, D.

PB2863. Unsupervised Learning of Ancestry Informative Markers and Genetic Admixture Coefficients From Biobank Data. Ko, S.

PB2865. Variable representation of duplicated sequence in recent canine genome assemblies. Nguyen, A.

Genetic Therapies Posters - Wednesday (Poster)

PB2017. A conditional knock-in preclinical mouse model for the *TRPV4* skeletal disorders. Nevarez, L.

PB2019. A drug repurposing screen to identify therapies for the rare disease DPAGT1-CDG. Dalton, H.

PB2020. A Phase 1, Open-Label, Dose-Escalation Study to Evaluate the Safety and Efficacy of HMI-103, a One-Time Phenylalanine Hydroxylase Gene-Editing Vector in Adult Participants with Classical PKU Due to PAH Deficiency. Sandhu, A.

PB2022. AAV delivery of *ELP1* exon-specific U1 snRNA rescues retinal degeneration in a mouse model of familial dysautonomia. Kirchner, E.

PB2025. An allele specific strategy for targeting mutant senataxin in patients with amyotrophic lateral sclerosis type 4. Grunseich, C.

PB2027. An *in vivo* screen identifies small molecule modulators of retinitis pigmentosa and the ER stress response. Hope, K.

PB2030. Comparison of AAVmyo and MyoAAV as promising vectors to deliver gene therapy in the VCP ^{R155H} KI mouse model. Weiss, L.

PB2032. Development of an AAV gene therapy for GNE myopathy: AAV9-CK8e-SV40-*hGNE1*-V5 shows robust GNE expression in mouse muscle tissue. Koczwara, K.

PB2034. Down-regulation of *SCN8A* as treatment for developmental and epileptic encephalopathy. Yu, W.

PB2036. Dynamin-2 interacts with SPEG and its reduction rescues the skeletal myopathy of SPEG-deficient mouse model. Li, Q.

PB2038. Effect of respiratory resistance exercise done remotely on respiratory function in individuals with familial myopathy. Halseth, M.

PB2040. Evaluation of Ataluren efficacy in fibroblasts from Neurofibromatosis Type 1 patients with nonsense mutation. Kim, S.

PB2042. Genetic subgroup learnings is presented from the MMPOWER-3 trial: elamipretide improves six-minute walk test in individuals with mtDNA replisome disorders. Sullivan, A.

PB2044. Genotyping analysis in patients with retinitis pigmentosa due to mutations in the *RHO*, *PDE6A*, or *PDE6B* gene: the PHENOROD2 study. Chung, D.

PB2046. Harnessing the power of natural variations of the human genome to support the discovery of new therapeutics: A systematic review. Trajanoska, K.

PB2048. Interrogation of the role of androgens and their antagonists in the pathogenesis and treatment of Vascular Ehlers-Danlos Syndrome. Juzwiak, E.

PB2050. Leveraging Machine Learning to Improve Guide RNA Design for Efficient and Selective ADAR-Mediated RNA Editing. Jiang, Y.

PB2052. Long-term efficacy and safety of elamipretide in patients with Barth syndrome is presented through the 192-week open-label extension results of TAZPOWER. Campbell, J.

PB2054. Novel ASO based treatment shows promise in treating CMT2S, preclinical data. Przychodzen, B.

PB2056. Personalized RNA interference approach as a nanotherapy for Crouzon syndrome: design of allele-specific siRNAs targeting FGFR2 mutant allele delivered by recombinant human nanoferritin. Tiberio, F.

PB2058. RNA editing of founder nonsense mutations causing inherited retinal diseases using site-directed endogenous adenosine deaminase acting on RNA. Sharon, D.

PB2060. Testing gene therapy for achromatopsia-like visual deficits in nonhuman primates affected by naturally occurring missense mutations in *PDE6C*. Rogers, J.

PB2062. Two years of venglustat combined with imiglucerase shows continued positive effects on neurological features and brain connectivity in adults with Gaucher disease type 3. Zheng, R.

Complex Traits Posters - Wednesday (Poster)

PB1233. A comparative PRS-PheWA of 14 autoimmune disorders, and their genetic relationship. Topaloudi, A.

PB1235. A comprehensive genetic map of cytokine responses to pathogens in 1063 Lyme patients reveals novel regulatory mechanisms underlying diseases. Botey-Bataller, J.

PB1237. A genome wide association study of chronic spontaneous urticaria risk and heterogeneity. Chang, D.

PB1239. A genome-wide association study of lifetime estrogen exposure in Korean postmenopausal women. Yuk, M.

PB1241. A Haptoglobin (HP) Exon Deletion Polymorphism Alters the Effect of *APOE* Alleles on Alzheimer's Disease in Individuals of European Ancestry with *APOEε4*. Bai, H.

PB1243. A Mendelian randomization study of genetic liability to Post-traumatic stress disorder and risk of Ischemic Stroke. Soremekun, O.

PB1245. A multi-ethnic genome-wide association study in type 1 diabetes. Michalek, D.

PB1246. A nationwide approach to understand the role of health, socioeconomic and genetic information in predicting COVID-19 vaccination uptake. Ganna, A.

PB1248. A pan cancer immunogenomic atlas and its applications for immune checkpoint blockade immunotherapy. Yang, J.

PB1250. A second wave SARS-CoV-2 variant in Quebec is associated with persistent infection. Fournelle, D.

PB1252. A Systematic Multi-omics Approach Identifies miRNA and Proteomic Dysregulation in Human Post-traumatic Stress Disorder. Wang, J.

PB1255. Activity-dependent transcriptional program in glutamatergic neurons enriched for genetic risk for schizophrenia. Ma, Y.

PB1257. Age differences in preadipocyte proportions and expression profiles link to obesity and insulin resistance. Kar, A.

PB1259. Alzheimer Disease plasma biomarker pTau-181 in individuals of diverse admixed ancestral backgrounds. Griswold, A.

PB1261. Alzheimer's disease pathology drives distinct homeostatic and active microglia phenotypes revealing clues to early pathogenic transcriptional switches. Jayadev, S.

PB1263. An integrated genomic and phenotypic analysis of sleep traits in autism spectrum disorder. Weissenkampen, J.

PB1265. Analysis of 3,273 undiagnosed neurodevelopmental disorder trios reveals over-transmission of polygenic risk and a female protective effect. Wigdor, E.

PB1267. Analysis of both shared and specific associations among various autoimmune disorders greatly helps the identification of the causal gene(s) and the functional mechanisms. Yang, W.

PB1269. Analysis of nascent RNA reveals distinct gene signatures for acute inflammation in cytokine stimulated keratinocytes. Patrick, M.

PB1271. Analysis of Relationship between Metabolic Syndrome and Genome-Wide Association Study (GWAS) of Vitamin D in Koreans: Community-Based Cohort. Kim, H.

PB1273. Assessing T-Cell Receptor Diversity and Genetic Variation in Complex Phenotypes. Ercelen, D.

PB1275. Association analysis of structural variants in whole-genome sequencing of 150K UK Biobank participants. Liu, J.

PB1277. Association between Blood Pressure-related Polygenic Risk Scores and Hypertension among White and Black Individuals Across the Life Course. Sun, X.

PB1279. Association between polymorphisms in *CD32, CD36, CD40, CD54* and Malaria phenotypes among under-fives in Ibadan South-west Nigeria. Olajide, T.

PB1281. Association of African Ancestry with risk for fibroproliferative diseases is consistent with selection for a Th2 favored genome in African derived populations. Hampton, G.

PB1283. Association of Genetically-Predicted Placental Gene Expression and Diseases of the Cardiovascular System. Pigg, A.

PB1285. Association of Structural Variants with Coronary Artery Disease. Iyer, K.

PB1287. Associations between polygenic risk scores, alcohol use, and differences in brain measures in the UK Biobank. Thornton, V.

PB1289. Autism Spectrum Disorder (ASD) in Qatar: Whole genome sequencing of 100 affected families highlights Dominant and Recessive risk genes of ASD. Abdi, M.

PB1291. Burden of functional variants in epilepsy patients using a deep learning approach. Girard, A.

PB1293. Cell-type-specific activity-dependent multiomic profiling in single hiPSC-derived neurons for neuropsychiatric disorders . Zhang, S.

PB1295. Characterization of a diverse Frontotemporal Dementia cohort, enriched for Caribbean Hispanic patients. Nuytemans, K.

PB1297. Characterization of sequencing-based HLA alleles in a Quebec COVID-19 biobank. McClelland, P.

PB1299. Cholesterol and LDL-C levels are elevated prior to symptom onset in Alzheimer's disease independent of APOE-4. Chase, B.

PB1301. Classical HLA Alleles are Associated with Fasting Glucose and Type 2 Diabetes in Multiple Populations. Chen, G.

PB1303. Clinical utility of polygenic scores for cardiometabolic disease in Arabs. Shim, I.

PB1305. Collagen gene cluster expressions role on liver fibrosis in biliary atresia patients. Gunadi, F.

PB1307. Combining genetic risk with machine learning derived biological age improves prediction of type 2 diabetes. Leiby, J.

PB1309. Common and rare variant associations with spinal stenosis in 29,488 cases. Otto, J.

PB1311. Concordance of directional effects between sexes suggests pervasive epistasis and omnigenic regulation of complex metabolic traits in mice. Miller, A.

PB1313. Construction of clinically significant mutation map and their spatio-temporal single cell transcriptomics trajectories in congenital heart disease. Tambi, R.

PB1315. Copy number variants and mental health in a community pediatric sample. Engchuan, W.

PB1318. Curation burden mitigation: strategies for empowering high throughput gene panel analysis using whole genome sequencing. Strom, S.

PB1320. Data-driven Clustering of Human Cardiovascular Disease (CVD) Related Variables. Mandal, M.

PB1322. *De novo* CNVs impacting constraint overlapping genes in neurodevelopmental disorders and congenital anomalies. Safizadeh Shabestari, S.

PB1324. *De novo* variant analysis of childhood-onset obsessive-compulsive disorder in the French-Canadian population. Bornais, K.

PB1326. Deep embedded clustering by phenotype and genome-wide association study in autism from SPARK individuals. Ueno, F.

PB1328. Deep venous thrombosis: genotyping and whole-exome sequencing data to improve genetic risk prediction. Lo Faro, V.

PB1330. Differential splicing occurs in individuals with disseminated coccidioidomycosis infections. Jensen, S.

PB1332. Direct participation of individuals scales the development of cardiometabolic polygenic risk score models. Lopez Pineda, A.

PB1334. Discovering the early molecular markers of lipid dysregulation using deep phenotyping and adipose tissue gene expression. Tomlinson, M.

PB1335. Discovery of rare variants in 219 genes associated with adult human height via burden analysis in exomes of 785,210 individuals. Locke, A.

PB1337. Discrepancy between genetically-predicted and actual body mass index is a significant predictor of incident cardiovascular disease. Rhee, T.

PB1339. Dissecting single cell splicing isoforms in human hearts using single cell nanopore sequencing. Lu, L.

PB1341. Dissecting the role of pleiotropy in the genetic adaptation of Tibetans to high altitudes. Thornburg, A.

PB1343. Drug repurposing for osteoporosis in men by integrating multi-omics and pharmacogenomics. Greenbaum, J.

PB1345. Effect of cortisol on cortical organoids: Building a "stress in a dish" model system. Purmann, C.

PB1347. Effects of the Tau Haplotype on Cognition in Childhood. Dokuru, D.

PB1350. Enrichment of Patients with Ehlers Danlos Syndrome in Idiopathic Gastroparesis - A Gene Set Enrichment Analysis. Carlin, J.

PB1352. ERAP1, ERAP2, and two copies of HLA-Aw19 alleles increase the risk for Birdshot Chorioretinopathy in HLA-A29 carriers. Gelfman, S.

PB1354. Estimating the causal influence of body mass index on gut microbiome variation. Hughes, D.

PB1356. Evaluating the causal effect of tobacco smoking on white matter brain aging: a Mendelian randomization analysis in UK Biobank. Mo, C.

PB1358. Evaluation of genetic support for targeting the IL-23 pathway: current and future therapeutic opportunities. Hart, A.

PB1360. Evaluation of polygenic risk scores to differentiate between type 1 and type 2 diabetes. Shoaib, M.

PB1362. Examination of PTSD symptoms in older veterans as a function of AD risk genes and combat implicates *CLU* as a stress-response gene. Logue, M.

PB1364. Exome sequencing of six singlet Qatar families identified nine neurodevelopmental candidate genes. Gupta, V.

PB1366. Exploring disease-course trajectories using genomic scores in the Finnish SUPER study - a cohort of 10,407 psychotic individuals. Kämpe, A.

PB1368. Exploring the complexity of scleroderma etiology by trio whole genome sequencing. Dai, H.

PB1370. Expression profiling of small bowel tissue from Crohn's disease patients reveals alterations of immune functions and dampening of epithelial response. Lee, Y.

PB1372. Findings at both known and novel loci in genome wide linkage and association study of a depression endophenotype, major depressive disorder with chronic pain. Nolan, D.

PB1374. Fine-mapping 56 lipidomics loci identified by univariate and multivariate genome-wide association analyses. Ottensmann, L.

PB1376. Fine-mapping and signal co-localization of asthma and white blood cell traits within the chr17q12-21 locus across diverse ancestries. Ben-Eghan, C.

PB1378. Fine-mapping to find differences in effect sizes across ancestries for skeletal phenotypes. Bartell, E.

PB1380. Functional validation of rs7132908 as the causal variant at the childhood obesity locus on chr12q13. Littleton, S.

PB1381. Gene-based association studies of rare variants identify novel genes for asthma-associated quantitative traits in African-American children. Clay, S.

PB1383. Gene-by-stress interaction in Alzheimer's disease risks. Singh, A.

PB1385. Gene-level GWAS fine-mapping combined with single cell RNA-seq yields insight into neurodegenerative disease biology. van de Geijn, B.

PB1387. Genes causing significant effects on the dynamics of peroxisomes. Roy, N.

PB1388. Genetic analysis of bruxism and its associations to sleep, psychiatric and behavioural traits. Strausz, T.

PB1390. Genetic and environmental regulation of caudate nucleus transcriptome in schizophrenia. Benjamin, K.

PB1392. Genetic architecture of insulin-stimulated glucose uptake reveals novel regulators of post-prandial glucose metabolism. Williamson, A.

PB1394. Genetic characterization of central serous chorioretinopathy and pleiotropic effects with age-related macular degeneration. Rämö, J.

PB1396. Genetic epidemiology of carotid intima-media thickness in Sub-Saharan African populations contributes to atherosclerosis biology. Boua, P.

PB1398. Genetic Interactions Implicate Disruption in Ciliogenesis in the Etiology of Non-syndromic Orofacial Clefts. Alade, A.

PB1400. Genetic Overlap among Alzheimer's Disease and Five Psychiatric Disorders: Genetic Convergence and Specificity. Enduru, N.

PB1402. Genetic pleiotropy effects on kidney function and soluble receptor for advanced glycation end-products: The Long Life Family Study (LLFS). Feitosa, M.

PB1404. Genetic Predictors of Memory Performance in Older Adults. Archer, D.

PB1406. Genetic Regulation of Gene Expression in HIV+ T Cells Associated With Control of HIV spVL in African Populations. Tough, R.

PB1408. Genetic susceptibility for autoimmune diseases and white blood cell count variability. Vaitinadin, N.

PB1410. Genetic variants in Olfactory Response Genes associated with Osteoarthritis progression to Hip or Knee Replacement among African and Hispanic American Veterans. Nair, A.

PB1412. Genetically Informed Prediction of Short Term Parkinson's Disease Progression. Javedani Sadaei, H.

PB1414. Genetics determinants of Renin-Angiotensin-Aldosterone System (RAAS) in the Cooperative Health Research In South Tyrol study. Foco, L.

PB1415. Genetics of Gulf War illness, a genome wide association study. Vahey, J.

PB1416. *Genetics of ventricular septal defects: A novel genetic interaction between Sox7 and Wnt4 is associated with abnormal endocardial cushion morphogenesis.* Hernandez-Garcia, A.

PB1418. Genome sequencing in brain samples from the ROSMAP cohort to examine the relationship between telomere length, mitochondrial copy number, and β-amyloid. Lynch, M.

PB1420. Genome-wide analysis in over 1 million individuals reveals over 2,000 independent genetic signals for blood pressure. Keaton, J.

PB1422. Genome-wide association analysis of composite sleep scores in 413,904 individuals. Wang, H.

PB1424. Genome-wide association meta-analysis reveals 99 risk loci for pain susceptibility and pleiotropic relationships with psychiatric, metabolic, and immunological traits. Mocci, E.

PB1426. Genome-wide association of NASH and Mendelian randomization with plasma protein levels identifies putative protein changes resulting from disease. Smith, E.

PB1428. Genome-wide association studies of human and ratbody mass converge on a conserved molecular network. Leger, B.

PB1430. Genome-wide association study for 233 circulating metabolic traits in 136,000 participants reveals extensive pleiotropy and novel associations. Karjalainen, M.

PB1432. Genome-wide association study in mesial temporal lobe epilepsy with hippocampal sclerosis. Bruxel, E.

PB1434. Genome-wide association study of longitudinal changes in motor symptomology in Parkinson's Disease. Kim, W.

PB1436. Genome-wide association study of opioid use disorder for diverse populations in All of Us research program. Gui, H.

PB1438. Genome-wide association study of venous thromboembolism in women using combined oral contraceptives. Trégouët, D.

PB1440. Genome-wide association study to identify genetic variants for obesity in Korean population. Yang, S.

PB1442. Genome-wide meta-analysis identifies a potential therapeutic targets for new loci and functional pathways influencing Alzheimer's disease risk. Kim, J.

PB1444. Genome-wide metabolite quantitative trait loci analysis in red blood cells from routine blood donors. Moore, A.

PB1446. Genome-Wide SNP Interaction Tests with Polygenic Risk Score for Ischemic Stroke Identifies Associations with Alzheimer Disease at *VCPKMT* and *KNDC1* by Age-Stratified Analysis. Chung, J.

PB1448. Genomic discovery with functional annotation of *CNTNAP5* in the phenotypic extremes of primary angle closure glaucoma. Chakraborty, S.

PB1449. Genomic structural equation modeling to unravel kidney-specific variants from multiple biomarkers. Fujii, R.

PB1451. Genotypic and Phenotypic Heterogeneity in Prader-Willi Syndrome: A Bangladeshi cohort study. Mim, R.

PB1453. Global Long COVID Host Genetics Initiative identifies *FOXP4* locus as the first genetic risk factor associated to Long COVID. Lammi, V.

PB1455. GWAS of the age-of-onset of type 1 diabetes reveals HTATIP2 as a novel T cell regulator. Cardinale, C.

PB1457. Heterozygous *RYR2, ABCG8, PIK3C2G*, and *RASSF9* variants as the cause of premature coronary artery disease in five Iranian families. Najmabadi, H.

PB1459. HLA in autoimmune encephalitis. de Araujo, T.

PB1461. HLA typing and COVID-19 disease outcome associations: GENCOV Study Canada. Frangione, E.

PB1463. *HLA-DRB1*15:01* modifies sphingolipid levels in multiple sclerosis. Briggs, F.

PB1465. How are *APOE4*, changes in weight/BMI, and longevity related? Insights from a Causal Mediation Analysis. Holmes, R.

PB1467. Hypertension Prediction From Metabolomics For Two Cardiovascular Disease Cohorts and Transfer Learning of Resultant Models. Gong, H.

PB1469. Identification of genetic loci associated with peanut specific IgG4 in high risk children from the LEAP study. Kanchan, K.

PB1471. Identification of novel genetic variants for association with rheumatoid arthritis susceptibility in Pakistani population. Ali, S.

PB1473. Identification of shared genes and pathways between psychiatric GWAS and monogenic neurodevelopmental disorders. Jansen, P.

PB1475. Identifying Developmental Stuttering comorbidities using Electronic Health Records. Scartozzi, A.

PB1477. Identifying Metabolomic Signatures of Genetic Liability to Type 2 Diabetes in East Asian Adolescents. Chen, S.

PB1479. Identifying phenotypic signatures of circulatory microRNAs. Mustafa, R.

PB1481. Identifying shared genetic architecture across eye diseases using electronic health records. Scalici, A.

PB1484. Improving Common Disease Risk Prediction with Clinical Risk Factors using Polygenic Scores in a Primary Care Physician Network of 35,000 Patients Followed for 16 years. Mandla, R.

PB1486. Improving the selection of variants included in polygenic risk scoring models. Arehart, C.

PB1487. In silico analysis to examine COPD genetic associations using scRNA-seq-based CRISPRi approach in iPSC derived alveolar type 2 (iPSC-AT2) cells. Malik, V.

PB1491. Individuals with GCK-MODY are not at increased risk for common complications associated with T2D as measured by the Diabetes Complications Severity Index (DCSI). McEwen, L.

PB1493. Insights into pediatric inflammatory bowel disease subtypes using single-cell RNA-sequencing. Keever-Keigher, M.

PB1495. Integrating Genomic Risk into Absolute Risk Estimates for Coronary Heart Disease. Hamed, M.

PB1497. Integration network-based analyses and Mendelian Randomization to screen drug repurposing candidates for osteoporosis. Liu, D.

PB1499. Integrative analyses to identify plausible target genes on Alzheimer's Disease genes with genetic evidence. Leung, Y.

PB1501. Integrative genomic and transcriptomic analyses revealed new driver variants associated with progression of the non-alcoholic fatty liver disease. Kim, D.

PB1502. Interactions between Valley Fever and genetic ancestry: does genetic ancestry affect risk of developing disseminated coccidioidomycosis?. Spendlove, S.

PB1504. Interplay of age and Alzheimer's disease diagnosis in estimating heritability. Leonenko, G.

PB1506. Intragenic loci within *TOMM40* modulate *APOE* expression in human microglia. M Ramirez, A.

PB1509. Irina St. Louis. St. Louis, I.

PB1511. Isoform usage differences in schizophrenia. Giusti-Rodriguez, P.

PB1513. Known Alzheimer disease candidate variants influence the severity of neuropathologic lesions underlying Alzheimer disease and related dementias. Godrich, D.

PB1515. Large-scale chromatin accessibility fine-mapping of blood lipids GWAS loci in human liver cell types. Wenz, B.

PB1517. Leveraging aggregation of juvenile idiopathic arthritis in large pedigrees to investigate susceptibility genes. Avery, C.

PB1519. Leveraging spatial patterns in the distribution of dental caries to resolve genetic heterogeneity. Haworth, S.

PB1521. Limited effect of Y chromosome variation on coronary artery disease and mortality in UK Biobank. Wilson, J.

PB1522. Longitudinal transcriptomic profiling of IBD patients reveals therapeutic decision-making determinants of drug response. Cervera Seco, L.

PB1524. Loss-of-function variants association study highlights potential new causal genes implicated in blood pressure regulation. Lecluze, E.

PB1526. Machine learning in COVID-19 phenotype prediction. Novelli, G.

PB1528. Mapping Autism Spectrum Disorder Endophenotypes to Genomic Regions in Thousands of Families. Stockham, N.

PB1529. Metabolomic signatures of body mass index in the Hispanic Community Health Study/ Study of Latinos (HCHS/SOL). Buchanan, V.

PB1531. Metagenomic analysis of the gut resistome in dyslipidemia and healthy individuals. Raza, S.

PB1533. Mitochondrial*TXNRD2*and*ME3*genetic risk scores are associated with specific primary open-angle glaucoma phenotypes. Aboobakar, I.

PB1536. Modification of coronary artery disease clinical risk factors by coronary artery disease polygenic risk score. Truong, B.

PB1540. Molecular states during acute COVID-19 reveal distinct etiologies of long-term sequelae. Thompson, R.

PB1543. Multi-ancestry meta-analysis of asthma improves polygenic risk prediction across populations. Tsuo, K.

PB1546. Multi-tissue splicing transcriptome-wide association study identifies 25 new candidate susceptibility genes and a new risk region for Alzheimer's disease. Gao, G.

PB1548. Multitrait analysis genome-wide association study of atherosclerosis phenotypes. Hasbani, N.

PB1549. Multi-trait rare variant analysis of cardiometabolic traits. Bone, W.

PB1551. Natural language processing and modelling of clinical brain disease trajectories. Holtman, I.

PB1555. NGS testing and risk stratification of malignant cardiac arrhythmias. Kokalj Vokac, N.

PB1557. Non-Mendelian inheritance patterns and extreme deviation rates of CGG repeats in autism. Kooy, F.

PB1559. Novel Insights into Pediatric Scoliosis Revealed by Genome-wide Association Study and Whole Exome Sequencing. Qu, H.

PB1561. Opportunities in national SARS-CoV-2 genomic surveillance programs for accelerating study on human genetic determinants of differential COVID-19 severity. Szemes, T.

PB1563. Overexpression screen of chromosome 21 genes reveals modulators of Sonic hedgehog signaling relevant to Down syndrome. Moyer, A.

PB1565. Partitioned polygenetic risk scores of adipocyte marker genes and sex-specific GWAS variants explain sex-specific differences in abdominal obesity. Huang, H.

PB1568. Perceived benefits and barriers to implementing polygenic risk scores in primary care: results of a national physician survey. Vassy, J.

PB1570. Phenome-wide analyses with nonsynonymous variants in SOS2 demonstrate remarkable pleiotropic associations in the UK Biobank. Bruse, S.

PB1572. Phenome-Wide Association Study (PheWAS) investigates the role of long-read discovered structural variants in cardiovascular disease risk in the UK Biobank using an imputation framework. Basile, A.

PB1576. Polycystic Ovarian Syndrome Physiologic Pathways Implicated Through Clustering of Genetic Loci. Stamou, M.

PB1578. Polygenic risk and complex trait prediction for East Asians using the Taiwan Precision Medicine Initiative and Taiwan Biobank datasets (N = 500k). Chang, C.

PB1580. Polygenic risk for body mass index is associated with atypical antipsychotic induced weight gain. Justice, A.

PB1582. Polygenic risk scores affected by different strategies of selection of relevant genomic positions and equations used for calculations. Radvanszky, J.

PB1584. Polygenic risk scores identify heterogeneity in disease diagnosis and treatment in asthma and COPD. Moll, M.

PB1586. Polygenic score prediction of atrial fibrillation following cardiac surgery: a retrospective hospital biobank study. Jeuken, A.

PB1587. Population level genetic screening for coronary artery disease with polygenic risk scores and rare variants in a diverse electronic health record linked biobank. Boulier, K.

PB1589. Portability of a multiethnic polygenic risk score for low-density lipoprotein cholesterol in a Samoan population. Carlson, J.

PB1591. Predicted loss-of-function variants for blood lipids in over a million individuals. Koyama, S.

PB1593. Prediction of Alzheimer's disease conversion using polygenic risk scores grouped by transcriptome profiles from blood and brain. Goldstein, D.

PB1595. Prioritization of causal genes at Parkinson's disease associated loci using machine learning. Yu, E.

PB1599. PRS-PheWAS of psychiatric/ behavioral traits to disentangle clinical heterogeneity in Substance Use Disorders. Vilar-Ribo, L.

PB1600. Quantification of race, ethnicity, and genetic ancestry disparities in anti-hypertensive drug efficacy in the All of Us Research Program. Goleva, S.

PB1601. Quantifying differential multiple sclerosis risk via polygenic risk scores in African American and Hispanic cohorts. Hernandez, A.

PB1605. Rare copy number variation in the GR@ACE/DEGESCO dementia dataset of spanish population. de Rojas, I.

PB1607. Rare protein-truncating DNA variants in APOB or PCSK9, low-density lipoprotein cholesterol, and risk of coronary artery disease. Dron, J.

PB1609. Rare variant genetic architecture of mitochondrial DNA copy number from 415,422 exomes. Pillalamarri, V.

PB1611. Rare variation in ADHD and related diagnoses in the Mass General Brigham Biobank. Satterstrom, F.

PB1613. Relationship between the gut microbiome and depression in individuals with post traumatic stress disorder. Finnicum, C.

PB1615. *RGS3* and *IL1RAPL1* missense variants implicate defective neurotransmission in early-onset inherited schizophrenias. Kanwal, A.

PB1617. Risk factors that affect performance of polygenic risk scores across diverse cohorts. Hui, D.

PB1619. RNASeq in nasal epithelium from African ancestry subjects in the CAAPA consortium reveal *IL4* and *TGFB1* as upstream regulators of differentially expressed networks for asthma. Szczesny, B.

PB1621. SARS-CoV-2 sequencing: A comparison of high-throughput methods. Olaso, R.

PB1623. Secretoglobin family 1D member 2 (SCGB1D2) protein inhibits growth of Borrelia burgdorferi and affects susceptibility to Lyme disease. Strausz, S.

PB1625. Sequencing-based genome-wide association study of triglycerides in East Africans. Meeks, K.

PB1627. Serum proteomic signatures predicting unintentional weight loss in patients with chronic obstructive pulmonary disease. Chiles, J.

PB1629. Sex-specific analysis of rare variant associations with quantitative traits in the UK Biobank. Hoffing, R.

PB1631. Sex-Specific associations of gene expression in brain with Alzheimer's disease pathology and cognitive performance. Clifton, M.

PB1633. Sex-specific genetic predictors of brain amyloidosis in Alzheimer's disease. Wang, T.

PB1637. Sex-stratified meta-analysis of age-related cognitive decline across neurocognitive domains. Acharya, V.

PB1639. Shared genetic basis informs the roles of polyunsaturated fatty acids in brain disorders. Xu, H.

PB1641. Shriners Children's international program on the genetics of rare pediatric disorders. Shazand, K.

PB1643. Single cell patch-seq in understanding the regenerative ability of cortical spinal motor neurons. Kim, H.

PB1645. Single-Cell RNA-Seq Reveals the connections of amyloid-beta and ferroptosis tendency. Jin, Y.

PB1647. SNX8 as a novel predisposing factor by an integrative study for the risk of childhood atopic dermatitis in COCOA. Choi, E.

PB1649. Somatic mutations in chronic lung disease are associated with reduced lung function. Yun, J.

PB1651. Stratification of a PMS population based on their response to Human Growth Hormone and Insulin-like Growth Factor 1. Moffitt, B.

PB1653. Study of gene-environment interactions suggests a personalised medicine approach for ALS. Cooper-Knock, J.

PB1655. Telomere length associated rare variant candidate gene study in idiopathic pulmonary fibrosis. Radder, J.

PB1657. THE ASSOCIATION OF *FSHR* POLYMORPHISMS WITH PCOS IN PUNJABI POPULATION. Kaur, A.

PB1659. The causal effect of blood lipid metabolism related traits with depression phenotypes: the evidence from mendelian randomization study. Tao, S.

PB1661. The first GWAS on intrahepatic cholestasis of pregnancy unveils novel bile acid metabolism related associations. Tyrmi, J.

PB1663. The genetic drivers of young- and early-onset Parkinson's Disease in India. Andrews, S.

PB1665. The heritable component of human longevity is highly polygenic and pleiotropic. Jabal-Ameli, R.

PB1667. The impact of exercise on gene regulation in association with complex trait genetics. Vetr, N.

PB1670. The kynurenine pathway in Alzheimer's disease: A systematic review and meta-analysis. Inam, M.

PB1672. The Pan-UK Biobank project improves locus discovery, facilitates genetic architecture comparisons, and increases the resolution and generalizability across diverse populations. Kanai, M.

PB1674. The prevalence, penetrance, and expressivity of mitochondrial disorders in a population-based cohort. Hall, T.

PB1676. The role of host genetics on weight gain following ART initiation in people living with HIV. Jia, T.

PB1678. The variant in *FLNA* identified in a patient with progressive supranuclear palsy. Kume, K.

PB1680. Tissue-associated examination of risk factors uncovers genetic risk profiles that vary across patients of inflammatory bowel disease. Gaite, A.

PB1682. Trans-ancestry genome-wide analysis of kidney disease in individuals with type 2 diabetes. Richard, E.

PB1684. Transcriptome analysis of familial dysautonomia reveals tissue-specific gene expression disruption in the peripheral nervous system. Harripaul, R.

PB1686. Transcriptome-wide and Proteome-wide association study of Tourette's Syndrome. Shekhar, S.

PB1688. Transcriptome-wide association study identifies novel candidate susceptibility genes for migraine. Meyers, T.

PB1690. Transcriptomic analysis of whole blood in ancestrally diverse Alzheimer Disease cohorts implicates convergent immune and lipid processing molecular pathways. Gu, T.

PB1692. Trans-ethnic meta-analysis in a multi-ethnic population refines multiple sclerosis susceptibility loci and identifies novel locus. Beecham, A.

PB1694. Understanding the effect of non-coding de novo mutations within craniofacial enhancers in trios with orofacial clefts. Curtis, S.

PB1696. Understanding the molecular basis of SARS-CoV-2 pathogenesis at a gene expression level using Applied Biosystems™ TaqMan™ Flexible Array Panels. Gupta, A.

PB1698. Universal clinical Parkinson's disease axes identify a major influence of neuroinflammation. Universal clinical Parkinson's disease axes identify a major influence of neuroinflammation. Sandor, C.

PB1700. Unsupervised learning revealed metabolic syndrome sub-groups with differing phenotypic and genotypic traits. Lim, A.

PB1702. Using genetics to uncouple highly correlated metabolic phenotypes and test their separate role in disease. Martin, S.

PB1704. Using Stimulation of B cells to uncover novel disease-associated QTLs. Murphy, D.

PB1706. USP53 variant associated with psychosis in a consanguineous pedigree. V Pardo, J.

PB1708. Variant analysis of disease causing genes and risk factor genes in patients with Parkinson's Disease. Andriamboavonjy, L.

PB1710. Variant-to-gene mapping in human microglial cell models with clonal CRISPR validation implicates *RTFDC1* and *CASS4* at the Alzheimer's disease '*CASS4*' locus. Burton, E.

PB1712. Whole exome sequencing identifies two heterozygous novel variants in CCL21 and ITGA2 as a cause of Hypertrophic cardiomyopathy. Carlus, F.

PB1714. Whole exome sequencing suggests a role for rare genetic variation in the *NLRX1* gene in HIV disease progression in pediatric African populations of Botswana and Uganda. Amujal, M.

PB1716. Whole Genome Sequencing Analyses of 87,652 Individuals Reveal Rare Variants in Promoter of *HMGA1* Associated with Height. Li, X.
PB1718. Whole genome sequencing of a diverse Hispanic IBD population in the United States reveals differences in previously identified risk alleles. Haritunians, T.
PB1720. Whole-exome sequencing in five families with specific language impairment (SLI) suggests novel SLI genes and confirms previous findings. Raza, M.
PB1722. X Chromosome Wide Association Study on Latin American cohort reveals new potential loci associated with Parkinson's Disease. Peixoto Leal, T.

Genetic Counseling, ELSI, Education, and Health Services Research Posters - Wednesday (Poster)

PB2133. "Grown up" minors: Recontact after withdrawal or attrition. Patrinos, D.
PB2135. "I just wanted more": Hereditary cancer syndromes patients' perspectives on the utility of circulating tumour DNA testing for cancer screening. Clausen, M.
PB2137. A Sociotechnical Analysis of Returning Genomic Informed Risk Assessments in Primary Care Pediatrics. Terek, S.
PB2140. Advancing Health Equity in Genomics: Reflections and Recommendations for Future Research Directions from an NHGRI Workshop. Hindorff, L.
PB2142. Assessing the performance of the Clinician-reported Genetic testing Utility InDEx (C-GUIDE): Further evidence of inter-rater reliability. Hayeems, R.
PB2144. Assessment of reading comprehension levels of interpretation sections of genetic testing reports. Gillentine, M.
PB2146. Barriers to genetic testing when indicated at diagnosis for autism spectrum disorder. Wang, A.
PB2148. Cancer patients' experience of receiving variant of uncertain significance results: An Asian perspective. Ishak, N.
PB2150. Clinical cancer and direct-to-consumer genetic test result sharing behavior among US adults: Findings from HINTS 2020. Shete, S.
PB2152. Communicating genetic and socioeconomic factors underlying racial health disparities in medical education. Orozco Scott, P.
PB2154. Community Engagement in genetic and genomic research in a low literate setting: Challenges and perspectives. Diallo, K.
PB2156. Considerations for embedding genomic discovery research in clinical settings: Perspectives from medical geneticists. Muenzen, K.
PB2160. Designing a Comprehensive Education Program for Participants, Providers, and Research Teams: The eMERGE Consortium Experience. Connolly, J.
PB2162. Development and utility of a clinical research informatics application for participant recruitment and management for a return of results pilot trial in the Million Veteran Program. Yi, T.
PB2164. Do digital tools support a diversity of patient values? A qualitative study using the Genetics Adviser.   Krishnapillai, S.
PB2166. Electronic health record-based recruitment for genomic research studies. Miller, A.
PB2168. Engaging a local community & participant advisory board in a national precision medicine study: Experiences from the New York City Consortium *All of Us* Research Program. Trousdale, R.
PB2170. Ensuring participant understanding of informed consent through an automated consent chatbot for a national genome sequencing study. Andrew, E.
PB2172. Evaluation of out-of-province and self-pay for genetic testing in two publicly funded provincial programs in British Columbia, Canada. Elliott, A.
PB2174. Expanding ethics education for trainees: creating and evaluating a course on genetics, ethics, and society. Patel, R.
PB2176. Exploration of a Novel Cancer Genetics Learning Format: A Pilot Self-Directed Course in Genetic Cancer Risk Assessment. Tanna, D.
PB2178. Exploring the ethical implications of genomic data sharing in cloud-based ecosystems: Views of developers and users. Nelson, S.
PB2180. Family communication choices about neurodevelopmental/psychiatric genetic results: A social-network assessment of adult participants in a population-based genomic screening program. Wain, K.
PB2182. Fragile X syndrome: Experiences with receiving a genetic diagnosis and communicating genetic risk in a Cameroonian family. Kengne Kamga, K.
PB2184. Gender medicine, Gender-Omics and Sex-Omics: What we are dealing with? Karra, A.
PB2186. Genetic Counselors' Attitudes Towards Religiously-Based Policies and Their Impact on Professional and Ethical Obligations in the Prenatal Setting. Freedkin, L.

PB2188. Genome sequencing and preferences for secondary findings in ostensibly healthy COVID-19 positive individuals: GENCOV study Canada.   Casalino, S.

PB2190. Healthcare provider confidence in understanding unsolicited genomic results: Insights from the eMERGE III experience. Wiesner, G.

PB2192. Homozygous *APC* p.I1307K Mutations in an Ashkenazi Jewish Female: Implications for Genetic Counseling and Clinical Management. DeMarco, T.

PB2193. Identifying ELSI needs in newborn screening research. Lumpkins, C.

PB2195. Implementing human genetics training and research at a university hospital in Nairobi, Kenya. Ilovi, S.

PB2197. Improving Community Based Genomics Research in Black Populations: A Review of Lessons Learned and Ideas for Need Based Solutions. Whitted, C.

PB2199. Individual attitudes towards identity, privacy, and health insights from personal genomics: Evidence from a full archival search on Twitter. Zhao, B.

PB2200. Integrating Predictive Genetic Testing into clinical care in the Preventive Genomics Clinic. Srisukajorn, C.

PB2202. Is it time for a paradigm shift? Design and formative evaluation of a randomized clinical trial of the sequence of genetic counseling and testing to optimize efficiency, patient empowerment and engagement, and medical adherence for cardiovascular genetic testing indications *(RESEQUENCE-GC).*   Pendleton, C.

PB2204. Low interest and high drop off in population genetic screening of adults: Results from a "real world" pilot study. Rao, N.

PB2206. Medical genetics and genomics services *across the lifespan*: Patient and referral trends over 17 years at the Maritime Medical Genetics Service. Mackley, M.

PB2208. Non-inferiority of letter versus telephone genetic counseling for negative exome-based cancer genetic test results disclosure in a historically underserved population.   Gilmore, M.

PB2210. Partnering with patients to explore the psychosocial and socioeconomic impacts of hereditary cancer syndromes. Etchegary, H.

PB2212. Patterns and barriers to clinical trial participation for patients with primary mitochondrial disease: A survey of patients/caregivers, physicians, and pharmaceutical industry professionals. DiMatteo, M.

PB2214. Population DNA screening for medically actionable conditions in young adults: The DNA Screen national pilot study. Lacaze, P.

PB2216. Preferences of parents from diverse backgrounds on genomic screening in newborns. Omorodion, J.

PB2218. Prioritizing Research in Newborn Screening: Tools from the Newborn Screening Translational Research Network. Taylor, J.

PB2220. Q-A sequences in prenatal genetic counseling in Japan; conversation analytic study of NIPT and NT consultations. Kawashima, M.

PB2222. Real-world implementation of an efficient germline Point-of-Care Testing model in a multidisciplinary cancer center. Schaffer, K.

PB2224. Return of secondary genomic findings: Experiences of sickle cell disease research participants. Floyd, J.

PB2226. Shared in Rare: Engaging stakeholders to develop a shared ELSI research agenda across rare diseases. Berrios, C.

PB2227. The future of diversity, equity, and inclusion: Achieving the vision of genetic counseling for the All of Us Research Program. Onyeneho, K.

PB2229. The Quebec Participatory cohort, a citizen science projet for direct-to-consumer genetic testing participants. Girard, S.

PB2231. The SeDeN-p2 study : Perceptions of French health professionnals on the extension of newborn screening with or without genetics as a first-line test.   Level, C.

PB2233. Transforming genetics service delivery: Genetics providers' perspective on integrating digital tools into clinical practice.   Luca, S.

PB2234. Tribal community perspectives on genomics research and data sharing: a mixed-methods study. Tsosie, K.

PB2235. Understanding views towards gene editing in Switzerland. Ormond, K.

PB2237. Using variant databases to estimate disease prevalence for rare recessive disease. Baxter, S.

PB2239. Validation of automated electronic health record (EHR)data capture of hereditary breast and ovarian cancer and Lynch syndrome phenotypes. Savatt, J.

PB2241. Whole-exome sequencing in Puerto Rican patients: approach to diagnosis in complex traits. Albino, E.

Genetic, Genomic, and Epigenomic Annotations, Databases and Resources Posters - Wednesday (Poster)

PB3188. A comprehensive analysis of the reusability of public omics data across 2.8 million research publications. Vahed, M.

PB3190. A comprehensive Japanese genetic variation database TogoVar. Katayama, T.

PB3192. A novel reference architecture for multi-party federation: enabling joint analysis of large-scale clinical-genomic data across distributed Trusted Research Environments. Davison, C.

PB3194. A reference panel composed of Alzheimer's Disease enriched single-nucleotide and structure variants for structure variants discovery on SNP array data. Cheng, P.

PB3196. ALS/FTD Compute: an open, centralized repository of genomic data for ALS/FTD research. Traynor, B.

PB3198. Assessing the completeness of immunogenetics databases across diverse populations. Huang, Y.

PB3200. Blockchain and Artificial Intelligence-Enabled Stratified Trial System (BESTS) - A patient driven data sharing platform that leverages genomic and health data to accelerate clinical trial recruitment for precision therapies. Brady, L.

PB3202. Brotman Baty Institute Clinical Variant Database (BBI-CVD). Folta, A.

PB3204. Clinical Variants for Brain Gene Curation: A Powerful and Under-Utilized Resource. Chopra, M.

PB3206. CombVar: A pipeline to combine variant impact from prioritizers, integrators, and annotations. Nato, A.

PB3208. Distance-based panel generation optimizes gene selection for targeted gene panel design. Isakov, O.

PB3210. Early development of a locus specific database for *GUSB*, the gene associated with Mucopolysaccharidosis type VII: hints of a higher predicted prevalence. Daugherty, S.

PB3212. Efficient reinterpretation of rare disease cases using Exomiser. Vestito, L.

PB3214. Global landscape of primary omics data generation and its secondary analysis across 193 countries and territories. Peng, Q.

PB3216. HLA-SPREAD: a natural language processing based resource for curating HLA association from PubMed abstracts. Dholakia, D.

PB3218. Identification and characterization of genomic variations from population scale whole genome sequencing in south India. Kahali, B.

PB3220. Indiana Biobank: a resource of linked electronic health records, omics data, genetic data, and biospecimens for the research community. Lai, D.

PB3222. Lifebit delivers a federated Trusted Research Environment to allow researchers seamless access to 35,000 whole genomes in a novel COVID-19 cohort. Raine, N.

PB3223. MANE Select and beyond: expanding the joint NCBI and EMBL-EBI transcript set. Loveland, J.

PB3225. MetaMatchMaker: An artificial Intelligence tool for data discovery and meta-data harmonization. Page, G.

PB3227. National Center Biobank Network (NCBN) and genomic information for NCBN bioresources. Tokunaga, K.

PB3230. North Carolina Macular Dystrophy (NCMD/MCDR1): Analysis of our entire database, a model disease of non-coding mutations. Small, K.

PB3232. Partly Cloudy: analysis of blood lipids across All of Us and UK Biobank demonstrates the promise and limitations of cross-biobank analyses in the cloud. Selvaraj, M.

PB3234. PhenX presents Bone and Joint updates and expansion of Social Determinants of Health. Krzyzanowski, M.

PB3237. Rare Disease Phenotyping: What is Next? Adams, D.

PB3239. Role of Python APIs, Big data and Cloud computing in understanding the Genetic basis of a Disease. Murthy, S.

PB3241. Study design using the PhenX COVID-19 Collection and Variable Compare Tool. Williams, D.

PB3243. The BaseJumper™research platform for single cell multiomic analysis and interactive visualization. Weigman, V.

PB3245. The ClinGen Allele Registry, Linked Data Hub, and Evidence Repository Support Curation Efforts Through Registration of Variants, Aggregation of Supporting Evidence, and Publication of Pathogenicity Classifications. Riehle, K.

PB3247. The International Mouse Phenotyping Consortium: A unique resource underpinning the study of human disease. Groza, T.

PB3249. The One-Sided Matchmaking Platform (OSMP) - Accelerating genomic matchmaking and gene discovery through robust data sharing. Osmond, M.

PB3251. TT-Mars: structural variants assessment based on haplotype-resolved assemblies. Yang, J.

PB3253. Variant reclassification based on the allele frequency information from exome sequencing data of 20,455 patients enriched with under-represented populations. Kwon, K.

PB3255. VuTR: Visualising the consequences of high-impact 5' UTR variants. Dsouza, E.

PB3257. Web-CSEA: Web-based Cell-type-Specific Enrichment Analysis of Genes. Dai, Y.

Mendelian Phenotypes Posters - Wednesday (Poster)

PB1723. 4-years of Face2Gene in a General Genetics Clinic: Insights from Retrospective Analysis of Diagnosed Cases. Muriello, M.

PB1725. A case of atypical inheritance in late-onset Pompe disease. Mroczek, M.

PB1727. A case of fatal cardiac arrest in a neonate diagnosed with VLCADD postmortem. Singh, P.

PB1729. A case with mosaicism pigmentary and Prader-Willi Syndrome: importance of molecular diagnosis in cases of mosaicism. Gasparini, Y.

PB1731. A dysmorphology physical examination entry system facilitates structured genetic phenotype capture and natural language processing. Campbell, I.

PB1733. A homozygous variant in a novel gene "*AP2A2*" causes an early-onset hereditary spastic paraplegia in a Malian family. Diarra, S.

PB1735. A new case of YARS1 associated multisystemic disorder with compound heterozygous in a patient with Klinefelter syndrome. Kuan, J.

PB1737. A novel homozygous missense mutation in *ARSK* causes a new subtype of MPS. Sun, M.

PB1739. A Novel Homozygous Variant in Homologous Recombination Repair Gene *ZSWIM7* Causes Azoospermia in Males and Primary Ovarian Insufficiency in Females. Nawaz, S.

PB1741. A novel *LZTR1* variant in a Brazilian familial case with Noonan syndrome: comparative modeling and structural analysis. Chaves Rabelo, N.

PB1743. A novel splice-site variant causes *MYH2*-associated myopathy in a large family. Cassini, T.

PB1745. A *Plod2* mutant mouse model of Bruck syndrome. Kot, A.

PB1747. A recessive variant in *TFAM* causes mtDNA depletion associated with primary ovarian insufficiency, seizures, intellectual disability and hearing loss. Ullah, F.

PB1749. A single-center genetic analysis of oculopharyngeal myopathy. Shimizu, T.

PB1751. A Zebrafish Model of Congenital Muscular Dystrophy Caused by POMT1 Loss of Function. Manzini, M.

PB1753. Alteration of mitochondrial proteostasis in Costello syndrome. Lacombe, D.

PB1755. Angelman syndrome with mosaic paternal uniparental disomy caused by mitotic nondisjunction. Fujimoto, M.

PB1757. *ARHGAP32*, encoding the Rho GTPase Activating Protein 32, is a novel candidate gene involving in autosomal dominant neurodevelopmental disorder spectrum. Cao, Y.

PB1759. Atypical 260 kb deletion on distal 22q11.22 involving *TOP3B* shows the significance of this gene in autism spectrum disorder. Evans, D.

PB1762. Biallelic *DMD* variants in a mildly-affected female without a Duchenne or Becker phenotype. Yang, X.

PB1764. Bi-allelic LETM1 variants perturb mitochondrial ion homeostasis leading to a clinical spectrum with predominant nervous system involvement. Kaiyrzhanov, R.

PB1766. Bi-allelic loss of function variant in the *NRCAM* gene is associated with hereditary polyneuropathy phenotype. Elahi, Z.

PB1768. Bi-allelic loss-of-function variants in *PPFIBP1* cause a severe neurodevelopmental disorder with microcephaly, epilepsy and periventricular calcifications. Platzer, K.

PB1770. Biallelic pathogenic variants in the endosomal transport regulator *SNAPIN* cause a rare prenatal neuroanatomical syndrome. Yousaf, H.

PB1772. Biallelic variants in *CSPG4* cause a novel neurodevelopmental disorder with intellectual disability, global developmental delay, and facial anomalies. Da'as, S.

PB1774. Bibliometric analysis of Alport Syndrome - the impact of next generation sequencing on diagnosis. Salia, E.

PB1776. Biotin-thiamine responsive basal ganglia disease: A retrospective review of the clinical, radiological and molecular findings of cases in Kuwait with novel variants. Aburezq, M.

PB1781. Characterization of *CFTR* Variants Across Ancestries. Ideozu, J.

PB1783. Characterizing human disease-causing mutations: A zebrafish-based approach. Burgess, J.

PB1785. *CHP2* is a genetic modifier of risk of chronic *Pseudomonas aeruginosa* airway infection in cystic fibrosis.   Buckingham, K.

PB1787. Clinical and molecular characteristics of idiopathic midaortic syndrome in pediatric patients - preliminary results. Pelc, M.

PB1790. Combined brain abnormalities in patients with beta-tubulin mutations. Bae, H.

PB1792. Compound heterozygosity for variants in *DCAF1* in fraternal twins with neurodevelopmental disorder. Pappas, J.

PB1794. Compound heterozygous variants in *UFM1* gene in a case of hypomyelinating leukodystrophy. Kang, B.

PB1796. Congenital Anomalies Cluster: Linking clinicians and model system experts. Twigg, S.

PB1798. C-X3-C motif chemokine receptor 1 (*CX3CR1*) gene variants associated with lesion burden in Cerebral Cavernous Malformation. Weinsheimer, S.

PB1800. *De novo* missense variants in *RPS5* cause a newly characterized ribosomopathy. Ladha, F.

PB1802. De novo non-synonymous *CTR9* variants are associated with motor delay and macrocephaly. Suzuki, H.

PB1804. Deciphering a Diagnostic Odyssey of Atypical Free Sialic Acid Storage Disorder Associated with Tissue Specific Mosaic Variant in *SLC17A5*. Shinawi, M.

PB1807. Defining the clinical heterogeneity of *GARS1*-related Charcot-Marie-Tooth disease. Marte, S.

PB1809. Delayed developmental milestones in *MBD5*-associated neurodevelopmental disorder (MAND) associated with 2q23.1 deletions highlight need for early diagnosis. Zhan, L.

PB1811. Deletions of 14q32.2 result in severe neurodevelopmental outcomes and multiple congenital anomalies: three affected males and review of the literature. Black, J.

PB1813. Developmental Delay at the Single Cell Level in Prader-Willi Syndrome. Reiter, L.

PB1815. Diagnostic rate and clinical utility of whole genome sequencing in adults with intellectual disability. Sabo, A.

PB1817. Dilated cardiomyopathy in a 3-month-old female: The answer in genetics. Patel, M.

PB1819. Effect of FOXO3 and Air Pollution on Cognitive Function: A Longitudinal Cohort Study of Older Adults in China. Ji, J.

PB1821. Ellis-Van Creveld Syndrome: Clinical and Molecular Analysis of 50 Individuals. Aubert-Mucca, M.

PB1823. Enzymatic testing for MPSI in Kuwait: A Pilot Study toward newborn screening. Alsharhan, H.

PB1827. Exome sequencing leads to the identification of a rare case of an autosomal dominant non-syndromic hearing disorder in a German family. Birkenhager, R.

PB1830. Expanding the phenotypic spectrum of *TAB2* related disorders. Gupta, P.

PB1832. Exploration of Ubiquitin-Proteasome System involvement in neurodevelopmental diseases using cellular models. Deb, W.

PB1834. Exploring the distinct phenotypes of nine Romanian patients harboring a homozygous Arg355* variant in *BBS12*. Focsa, I.

PB1836. First patient reported with a *TPP1* missense mutation pathogenic variant predicted insilico causing *CLN2*. Moreno Giraldo, L.

PB1838. Fracture prevalence in children diagnosed with Ehlers-Danlos Syndrome and Generalized Joint Hypermobility. Mendoza, R.

PB1840. Frameshift *PPP1R12A* pathogenic variant in a mexican patient with differences of sex development (DSD), middle line defects and hemangiomata. Contreras-Capetillo, S.

PB1842. Functional analysis of *EZH1-A678G* in *Drosophila melanogaster*. Jangam, S.

PB1844. Gaps in the phenotype descriptions of ultra-rare genetic conditions: Systematic review and recommendations. Almail, A.

PB1846. Genealogy as a predictor of disease progression in patients with myotonic dystrophy type 1: A demonstration of the power of intersectorial research. Bouchard, J.

PB1848. Genetic analysis reveals that GNE Myopathy remains an underdiagnosed neuromuscular disorder. Rossignol, F.

PB1850. Genetic and phenotypic heterogeneity of childhood-onset hearing loss and implications for the success of cochlear implants. Carlson, R.

PB1852. Genetic Mapping Identifies A Founder Mutation of *GHRHR* Gene in Pakistani Family with Isolated Growth Hormone Deficiency Type 4. Khan, M.

PB1854. Genetic modifiers of nutritional status in CF are both unique to CF and shared with the general population. Ling, H.

PB1855. Genetic underpinnings of moderate to severe hearing loss in singleton individuals born to consanguineous couples in Pakistan. Naz, S.

PB1857. Genome sequencing identifies coding and non-coding variants in hereditary deafness missed by exome sequencing. Ramzan, M.

PB1859. Genomic ascertainment and reverse phenotyping diabetogenic variants in a phenotypically unselected cohort. Wilczewski, C.

PB1861. GGC repeat expansion within *NOTCH2NLC* causes behavioral deficits and neurodegeneration in a mouse model of Neuronal Intranuclear Inclusion. Kang, Y.

PB1863. GWAS of Down Syndrome Associated Atrioventricular Septal Defect identifies three novel loci. Feldman, E.

PB1865. Haplotype in interleukin 6 is associated with a reduction of blood transfusion events in sickle cell anemia patients. Fong, C.

PB1867. Here, There and Everywhere: Characterizing Variability in Approaches to Ascertaining Individuals with Rare Mendelian Disorders from EHRs. Veatch, O.

PB1869. High Molecular Diagnostic Yields and New Phenotypic Expansions Involving Anorectal Malformations. Belanger Deloge, R.

PB1871. Highly recurrent histone H4 mutations cause a neurodevelopmental disorder. Bicknell, L.

PB1873. How many diseases can one gene cause: Why mechanism matters for gene curation, variant classification, and clinical management. Radtke, K.

PB1875. Human mutations in *Slitrk3* implicated in GABAergic synapse development in mice. Efthymiou, S.

PB1877. Identification and Functional Evaluation of Autosomal Recessive Non-Syndromic Hearing Impairment Genes in Rwanda. Uwibambe, E.

PB1879. Identification of a novel pathogenic variant in *HIST1H1E* encoding the H1 histone linker in a patient with a complex phenotype including progeroid feature, intellectual disability and lipodystrophy. Contribution of additional variants to the complexi. Mellone, S.

PB1881. Identification of a pathogenic gene mutation in Med12. Serrano Rodriguez, G.

PB1883. Identification of molecular causes of recessively inherited ataxic and neuropathic disorders in consanguineous Pakistani families. Aslam, F.

PB1885. Identifying phenotypic expansions for congenital diaphragmatic hernia plus (CDH+) using DECIPHER data. Hardcastle, A.

PB1887. Incontinentia pigmenti female with the *IKBKG/NEMO*del4-10 deletion: A mosaic form. Ursini, M.

PB1889. Intraflagellar transport protein RABL5/IFT22 is required for normal visual function and retinal photoreceptor survival. Yang, Y.

PB1891. Investigating the role of seryl-tRNA synthetase in mitochondrial biology and human recessive disease. Del Greco, C.

PB1893. Investigating variants of uncertain significance in orofacial cleft trios. Diaz Perez, K.

PB1895. *ITGB8* is a candidate disease gene for autosomal dominant and recessive trait forms of muscular dystrophy and neurological disease. Barish, S.

PB1897. *KIF1A* novel variant p.(Ser887Profs*64) exhibits clinical heterogeneity in a Pakistani family individuals with HSANIIC. Azam, M.

PB1899. Large-scale zebrafish-based functional analysis of genes associated with neurodevelopmental disorders. Thyme, S.

PB1901. LIM Homeobox 1 gene variants contribute to Mayer-Rokitansky-Küster-Hauser syndrome. Kira, D.

PB1903. Lissencephaly spectrum disorders: Clinical, radiological molecular results of 70 Egyptian families. Zaki, M.

PB1905. Long Read Sequencing and Expression Studies of *AHDC1* Deletions in Xia-Gibbs Syndrome Reveal a Novel Genetic Regulatory Mechanism. Chander, V.

PB1907. Long-term clinical course of Heyn-Sproul-Jackson syndrome. Futagawa, H.

PB1909. Loss of C-terminal Mediator Complex subunit-11 impairs fetal brain development and cause severely progressive neurodegeneration. Cali, E.

PB1911. *LYRM7* mutations causing mitochondrial complex 3 deficiency nuclear type 8 (MC3DN8)in a Five-year-old boy without cavitating leukoencephalopathy. Alsharhan, H.

PB1913. Meta transcriptomics detects emerging multidrug resistant Candida auris in a family with a mild TP63 associated ectodermal dysplasia. Fortina, P.

PB1915. Modeling dystroglycanopathy in zebrafish through CRISPR-Cas9-mediated knockout of *pomgnt2*. Flannery, K.

PB1917. Molecular and clinical review of 95 Polish pediatric patients with the clinical suspicion of Alport syndrome. Halat-Wolska, P.

PB1919. Molecular insights into the pathogenesis of Chediak-Higashi syndrome and the biology of Lysosomal Trafficking Regulator. Morimoto, M.

PB1921. Monogenic Familial Hypercholesterolemia in the eMERGE Network: Penetrance and Associated Coronary Heart Disease Risk. Dikilitas, O.

PB1923. Mutational analysis of a Romanian Bardet-Biedl syndrome cohort revealed an overabundance of causal *BBS12* variants. Khan, S.

PB1925. Myelomeningocele: targeted search for novel variants in Mexican families. Medina, Q.

PB1927. Novel clinical entity: Autosomal recessive missense GPC6-related skeletal dysplasia (ARM-GSD). Crenshaw, M.

PB1929. Novel homozygous variant in *TFAP2B*in a colombian family with Char syndrome and distinctive neurobehavioral features. Bernal, I.

PB1931. Novel locus identification for primary congenital glaucoma in a Pakistani family. Khan, H.

PB1933. Novel variants in the *PKD1* and *PKD2* genes in German patients with autosomal dominant polycystic kidney disease. Zarbock, R.

PB1935. Osteogenically differentiated human fibroblasts - an alternative model to study bone diseases. Pekkinen, M.

PB1937. Patients with primary familial brain calcification due to variants in SLC20A2 are not commonly diagnosed with Parkinson's disease. Chang, J.

PB1939. Phenotypic Characterization of 127 Individuals with *MECP2* Duplication Syndrome. Fatih, J.

PB1941. PhenylEc: Novel genotypes and nutritional management in phenylketonuria patients from Ecuador. Romero, V.

PB1942. Polygenic risk scores and protein language models predict phenotype severity in individuals with rare clinical pathogenic variants. Wei, A.

PB1943. *PPP2R5D*-related Neurodevelopmental Disorder can be inherited from a mildly affected parent. Jougheh Doust, S.

PB1945. Pre-clinical studies in induced Pluripotent Stem Cell (iPSC) lines with *SORD* mutations linked to a recessive neuropathy. Yanick, C.

PB1949. Pulmonary function and structure in young adults with types III, IV and VI osteogenesis imperfecta. Gochuico, B.

PB1951. Rare *MECP2* variant associated with the development of Rett syndrome: a case report. Crescenti, S.

PB1953. Rate of Deleterious Copy Number Variants Similar in Early Onset Psychosis and Autism Spectrum Disorders: Implications for Clinical Practice. Brownstein, C.

PB1955. Reanalysis of whole exome sequencing data of 50 Iranian families with hereditary hearing loss. Shokouhian, E.

PB1957. Recessive variants in the C-terminal domain of *UFSP2* are associated with severe spondyloepimetaphyseal dysplasia. Weisz-Hubshman, M.

PB1959. Report of a novel *LAMB3* variant and an overview of *LAMB3* variants associated with junctional epidermolysis bullosa. Tan, E.

PB1961. Rewiring our understanding of the enteric nervous system: Identification of diverse developmental lineages of enteric neurons and cell-state specific roles in maintenance across aging. Slosberg, J.

PB1963. RNF220, RING finger E3 ubiquitin ligase, is required for the generation of p2-derived V2 interneurons and its loss-of-function causes an interneuronopathy with a broad spectrum of neurodevelopmental phenotypes. Kim, H.

PB1965. Screening for Genetic Modifiers of *MED12*/*kto* Using Naturally Occurring Variation in *Drosophila melanogaster*. Bussey, K.

PB1967. Sequencing analysis of nonsyndromic cleft lip / palate in Brazilian patients reveals CTNNA1, ESRP1 and PRICKLE1 as novel candidate genes. Brito, L.

PB1969. Shared and divergent transcriptional dysregulation is present across brain regions and cell types in adult *Chd8* haploinsufficient mice. Nord, A.

PB1971. Six patients with genetic disorders with atypical symptoms: how to perceive them. Uehara, T.

PB1973. Splicing noise is variable across human introns, tissues and age and modelling its characteristics can improve our understanding of age-related diseases of the human brain. Garcia-Ruiz, S.

PB1975. Subtle genomic duplication on Chromosome 6: a hypothesis for a causative mechanism. John, A.

PB1977. Systematic bioinformatic analysis promotes a missense variant in *XK* as potential cause of X-linked intellectual disability. Litster, T.

PB1979. Taking the *CPLANE INTU* the light: further defining the orofaciodigital syndrome phenotype association with biallelic *INTU* alterations. Hunter, J.

PB1981. *TCEAL1* loss-of-function results in an X-linked dominant neurological syndrome and drives the neurological disease trait in Xq22.2 deletion. Hijazi, H.

PB1983. TGFβ signaling is important for spermatogenesis. Kumar, U.

PB1985. The ClinGen Kidney Disease Clinical Domain Working Group: Defining solutions to the challenge of nephropathy ontology and nomenclature. Byrne, A.

PB1986. The genetic and phenotypic landscape of *STUB1*/*TBP*-associated disorders: Two genes and three patterns of inheritance sharing a continuous neurodegenerative phenotypic spectrum. Magri, S.

PB1988. The genetic landscape of *ATP7B* provides insights into Wilson Disease variant penetrance and prevalence. del Angel, G.

PB1990. The impact of NGS on rare and complex epilepsies: identification of novel causative variants for an efficient diagnostic pathway and personalized treatments. Gellera, C.

PB1993. Three cases of (likely) pathogenic *FGF10* variants: Examples of phenotypic variability in Lacrimoauriculodentodigital syndrome. Drost, M.

PB1995. TMEM161B mediates radial glial scaffolding in neocortical development. Wang, L.

PB1997. Transcriptional response of Fundulus heteroclitus embryos exposed to carbaryl. Torano, O.

PB1999. Truncating variants in *RFC1* cause cerebellar ataxia, neuropathy vestibular areflexia syndrome (CANVAS). Das, S.

PB2001. Two novel compound heterozygous variants identified in a new case of classic Donohue syndrome and literature review. Wang, H.

PB2002. Understanding the role of *TTC5* in the developing mouse brain: A novel null TTC5 mouse model. Rushforth, R.

PB2005. Using remote methods to assess communication in Phelan-McDermid Syndrome. Quadri Valverde, S.

PB2006. Variable expressivity in a four-generation ACDMPV family with a noncoding hypermorphic SNV in *trans* to the frameshifting *FOXF1* variant. Stankiewicz, P.

PB2008. Vici syndrome in Israel: Clinical and molecular insights. Pode-Shakked, B.

PB2010. Whole exome sequencing in intellectual disability patients identifies de novo mutations in KCNB1, PPP1R3F, SHANK2, and SYNGAP1 genes. Alkhateeb, A.

PB2012. Whole exome sequencing will miss Congenital Myotonic Dystrophy in the child on an asymptomatic mother. Appiah, F.

PB2014. Whole-Exome Sequencing of a French Canadian Cohort Reveals New Candidate Founder Mutations. Ashton-Beaucage, D.

PB2016. Whole-genome sequencing for comprehensive genetic modifier identification in CMT1A. Xu, I.

Molecular and Cytogenetic Diagnostics Posters - Wednesday (Poster)

PB2243. A calibrated automated patch clamp assay can interpret any VUS in *KCNH2.* Vandenberg, J.

PB2245. A combination of exome sequencing and optical genome mapping unveils a dual molecular diagnosis in a case with an unknown neurodevelopmental disorder. Acharya, A.

PB2247. A heterozygous de novo VUS (p.M130I) in candidate gene PSMC5 in a male with autism, mild developmental delay, speech deficiency, dysmorphic features, hypospadias and chordee. Hajianpour, M.

PB2249. A multiple congenital anomalies newborn with coexistence of extra der(22) chromosome marker and balanced t(11;22) maternally inherited. Albalwi, M.

PB2251. A new neurodevelopmental disorder with microcephaly and neural tube defect Loss of NARS1 leads to microcephaly and neural tube defects. Temel, S.

PB2253. A rare association of congenital hypohydrotic ectodermal dysplasia, secondary hypogonadism, and severe iron deficiency anemia. Selvarajan, D.

PB2254. A recurrent *AFF3* pathogenic variant associated to KINSSHIP syndrome in a Mexican patient. Flores Gallegos, L.

PB2256. A variant interpretation framework based on probabilistic graphical modeling that provides continuous high-resolution estimates of variant pathogenicity with improved interpretation accuracy over current discrete five-category frameworks. Nussbaum, R.

PB2258. Advancing Biobanking in the Genomics Era: Development of Molecular Omics Approaches for Interrogating the Diversity of Population Genetics for Early and Late-Stage Human Disease Prevention. Sheldon, M.

PB2260. Anthropometric parameters in males with chromosome 9p microdeletion syndrome in the first years of life: preliminary evidence. Pajouhanfar, S.

PB2261. B-allele frequency based approach to detecting absence of heterozygosity using optical genome mapping. Raksi, A.

PB2264. Child with Rhabdomyosarcoma and unusual features: suspected Rasopathy. Charaya, S.

PB2266. Chromosome conformation capture to detect structural variation in medical diagnostics. Holwerda, S.

PB2268. Clinical description of patients with CNV in the 15q11.2 region: What do we know? Chaparro Solano, H.

PB2270. Clinical utility of parental testing for the reclassification of likely pathogenic and uncertain copy number and sequence variants in a pediatric neurodevelopmental disorders cohort. Bilancia, C.

PB2272. Clinical whole-genome sequencing of oral samples and microbial contamination. Ameziane, N.

PB2273. Comparing the analytical performance of exome sequencing and traditional panel testing in a cancer population. Reble, E.

PB2275. Comparison of diagnostic yield between exon-targeted microarray and standard microarray for patients with congenital heart defects (CHD). Sulpizio, S.

PB2278. CREBBP related Rubinstein Taybi syndrome: Report of 3 patients. Panigrahi, I.

PB2280. Deep exome sequencing to detect mosaic variants in craniofacial microsomia. Parmalee, N.

PB2282. Development and clinical implementation of genome-based germline exome testing. Dumas, K.

PB2284. Diagnosis of genetic causes of intellectual disability and multiple congenital abnormalities in center of excellence for human genetics in Egypt. Mohamed, A.

PB2286. Diagnostic Utility and Lessons Learned from Deep Sequencing Vascular Malformations. Myers, C.

PB2288. Diagnostic yield of trio and individual Whole Exome Sequencing in a group of colombian patients with a suspected monogenic disease. Rodriguez-Alvarino, E.

PB2290. Does a larger gene panel size change the mutation detection rate in hypertrophic or dilated cardiomyopathy ? Fokstuen, S.

PB2292. Evaluate using artificial intelligence and machine learning in to predict deletion detection possibility in a cohort of chromosome 22q11.2 deletion syndrome cases. Alabdulkareem, I.

PB2294. Evidence review of the use of first-line genome sequencing to diagnose rare germline disorders. Wigby, K.

PB2296. Gabriella Miller Kids First Data Resource Center (DRC): Collaborative Platforms for Accelerating Pediatric Research. Higgins, D.

PB2298. Genetic Testing Interpretation Consult (GTIC): A Novel Electronic Health Record (EHR) Support Tool to Further Characterize Variants of Interpretation for the Nongenetic Provider to Support Their Clinical Practice. Hamid, R.

PB2300. Genetic testing of the ATP7B gene for patients with suspected Wilson disease. Kyriss, M.

PB2302. Genome Sequencing Enables Precision Clinical Care in Genetic Leukoencephalopathies. Muirhead, K.

PB2306. High diagnostic yield from clinical genome sequencing supports genome sequencing as first-tier genetic test: Evidence-based from 2002 index cases. Guo, F.

PB2308. Identification of Balanced and Unbalanced Complex Chromosomal Rearrangement Involving Chromosomes 2, 4,5,18 and 21 in a family. Venkateshwari, A.

PB2309. Identification of somatic Neurofibromatosis 1 mosaicism using targeted next-generation sequencing of café-au-lait-macules. Korpershoek, E.

PB2312. Inadequate parental sequencing depth burdens African ancestry patients with inaccurate de novo inheritance calls. Choi, S.

PB2314. Increased Diagnostic Yield With Long Read Sequencing In Patients With Undiagnosed Neurodevelopmental Disorder. Jamuar, S.

PB2316. Infinium Global Screening Array for CNVs detection. Vieira, L.

PB2318. Investigating the frequency of parental gonosomal mosaicism in Neurodevelopmental Disorder cohorts to improve the genetic diagnostic rate. Sandran, N.

PB2320. *KANSL1*, mosaic or not? Weerts, M.

PB2322. Long-read HiFi genome sequencing reveals a 2.7 kilobase intronic insertion in *NR5A1*as a cause of 46,XY disorder of sexual development. Del Gobbo, G.

PB2326. Mitochondrial variants and short tandem repeat expansions detected by clinical whole genome sequencing. Schulze, K.

PB2328. Monozygous twins with distal 5p duplication: a case report. Lebel, R.

PB2330. Mutation Spectrum of Polycystic Kidney Disease-Toronto Genetic Epidemiology Study. Haghighi, A.

PB2332. Novel potential biomarkers for *DICER1* syndrome. Wu, M.

PB2334. Optical genome mapping identified a likely pathogenic POLR3B variant in an undiagnosed male with ataxia, hypotonia, and cerebellar atrophy. Ortega, A.

PB2336. Optical genome mapping improves clinical interpretation of constitutional copy number gains. Raca, G.

PB2338. Pathogenic variants detected by RNA sequencing in Cornelia de Lange syndrome. Seyama, R.

PB2340. Potentially noteworthy intronic variants in the *COL5A1* and *COL5A2* genes in patients with classical Ehlers-Danlos syndrome: The necessity for RNA study and genetic diagnostic approaches. Heo, W.

PB2342. Recombinant chromosome with proximal 14q duplication in two sibs with mental retardation and facial dysmorphism due to maternal translocation t(14q21;21p11). Tayel, S.

PB2344. Review of Cytogenetic Findings of patients with Turner Syndrome and its variants among Filipinos from 1991 to 2020. Maceda, E.

PB2346. Statistical method for detection of uniparental disomy using SNP microarray or NGS technologies. Roytman, M.

PB2348. The perplexing 22q11.2 duplication syndrome: an important cause of congenital anomalies, medical conditions, cognitive deficits, and behavioral phenotypes - or nothing at all. McGinn, D.

PB2350. The utility of whole exome sequencing in atypical cases of heterogeneous neurological disorders. Ali, Z.

PB2352. Transposable element insertions in 1000 Swedish individuals. Eisfeldt, J.

PB2354. Ultra-low DNA input with long-read sequencing identified complex chromosomal rearrangements involving *NIPBL* in a Cornelia de Lange syndrome patient. Jiang, N.

PB2356. Unmasking of a chromothripsis event using the integrated approach of Chromosomal Microarray Analysis (CMA) and Optical Genome Mapping (OGM). Loddo, S.

PB2358. Updated clinical practice guidelines for managing children and adults with 22q11.2 deletion syndrome. McDonald-McGinn, D.

PB2360. Value of DNA testing in the diagnosis of Sickle Cell Anemia in Childhood in an environment with a high prevalence of other causes of anemia. Mbayabo Gloire, G.

Molecular Effects of Genetic Variation Posters - Wednesday (Poster)

PB2493. A gain-of-function recurrent missense variant leads to a GABAergic/glutamatergic imbalance in a forebrain organoid model of PACS1 syndrome. Rylaarsdam, L.

PB2495. A Homozygous IER3IP1 Mutation Causes Secretory Protein Trafficking Defects in Neural Progenitor Cells. Ahn, L.

PB2497. A lupus-associated variant in IRF7 amplifies IFN-α production. Virolainen, S.

PB2499. A rare genetic variant in the cleavage site of *prepro-orexin* is associated with idiopathic hypersomnia. Miyagawa, T.

PB2501. A single-nucleus transcriptome-wide association study implicates novel genes in depression pathogenesis. Zeng, L.

PB2503. A structural variant of the C-terminal prion-like domain of TDP-43 causes vacuolar muscle degeneration. Ervilha Pereira, P.

PB2505. *A2ml1*-Knockout Mouse as a Potential Model of Chronic Otitis Media. Elling, C.

PB2507. Abnormal cell fate allocation in embryogenesis caused by *Zic3* loss-of-function. Haaning, A.

PB2511. Allele-specific expression in blood cells during aging. Harwood, M.

PB2513. An African-specific Alzheimer disease-associated *ABCA7*frameshiftdeletion results in altered microglia functionality. Dykxhoorn, D.

PB2515. An integrated machine learning and functional analysis approach for resolution of variants of uncertain significance (VUS) in *PCDH19.* Calhoun, J.

PB2519. Assessing the contribution of variants with functional effects on post-transcriptional regulation to genetics of psychiatric disease. Gu, J.

PB2521. Assessment of genotype-phenotype correlation of human *TEX11* variants associated with non-obstructive azoospermia in mouse. Hardy, J.

PB2523. Association of MTHFR C677T polymorphism with obesity in women: A case-control study and meta-analysis. Irfan, M.

PB2525. Body mass index as an environmental context in functional genomic annotations. Signer, R.

PB2526. Calibration set for high throughput functional assay for *RYR1* Variants. Johnston, J.

PB2528. Cellular consequences of Hirschsprung disease-associated RET variants studied utilizing CRISPR-mediated genome engineering. Fries, L.

PB2530. Characterization of haplotype diversity in the human immunoglobulin heavy chain constant region using long-read sequencing. Jana, U.

PB2532. Characterizing a novel missense variant in the methionine salvage gene *MRI1* that conveys protection for type 2 diabetes. Bandesh, K.

PB2534. Chromatin conformation during CD4+ T cell activation implicates putative autoimmune disease candidate genes. Pahl, M.

PB2536. Clustering of missense mutations follows a non-random distribution across the human genome and correlates with specific classes of genes. Rivolta, C.

PB2538. Colocalization of blood cell traits GWAS associations and variation in *PU.1* genomic occupancy prioritizes causal noncoding regulatory variants. Jeong, R.

PB2540. Comparative titin gene frequencies and distributions across four population cohorts. Lai, P.

PB2541. Comparison of pedigree-based graph workflow to more traditional workflows for rare candidate variant analysis. Pusey, B.

PB2543. Comprehensive signal identification highlights the contribution of protein QTLs to complex trait genetics. Chiou, J.

PB2545. Congenital Myasthenic Syndrome Due to *DOCK7* Gene Mutation Case Study Report. Lebron Ilarraza, A.

PB2547. Consequences of loss of function burden effects on the human plasma proteome in 54,306 UK Biobank participants. Whelan, C.

PB2549. CRISPR/Cas9-mediated knockout and cDNA rescue using sgRNAs that target exon-intron junctions in *Drosophila*enable functional analysis of pathogenic variants in a tissue-specific manner. Yoon, W.

PB2551. CYP2C19 Polymorphism in Ischemic Heart Disease Patients Taking Clopidogrel after Percutaneous Coronary Intervention in Egypt. El-Zawahri, M.

PB2553. Deletion mapping of regulatory elements for *GATA3*reveals a distal T helper 2 cell enhancer involved in allergic diseases.. McVicker, G.

PB2555. Determining the rate of unintended on-target and off-target effects induced by CRISPR-Cas gene editing in iPSC clones. Shum, C.

PB2557. Development of a yQTL Discovery Pipeline Applicable for Both Unrelated and Related Individuals. Li, M.

PB2559. Disease-specific analysis improves prioritization of non-coding genetic variants. Liang, Q.

PB2561. *Drosophila* assays for variant interpretation in the polycomb repressive complex 2 related syndromes. Cyrus, S.

PB2563. Ehlers-Danlos Syndrome: A Likely Pathogenic Variant Found in *COL5A1* gene. Rodriguez Rodriguez, C.

PB2565. Elucidating mechanisms of genetic interactions and oligogenic inheritance in Autism Spectrum Disorder. Sertie, A.

PB2567. Epigenomic and transcriptomic analyses define core cell types, genes and targetable mechanisms for kidney disease. Liu, H.

PB2569. Escape from Nonsense-Mediated Decay: Annotation of Transcripts with Protein Truncating Variants. Klonowski, J.

PB2571. Establishment of *in vitro* assay system for evaluating of *RERE* variants. Kim, B.

PB2573. Evaluation of regulatory potential of European ancestry *APOEε3 TOMM40*-523' repeat haplotypes with differential risk effects. Lipkin Vasquez, M.

PB2575. Evidence from a massively parallel reporter assay and genome engineering for non-coding variants at 1q32-*IRF6* that directly influence risk for orofacial cleft. Kumari, P.

PB2577. Examination of predicted loss of function (pLoF) variants in the general population reveals high rates of rescue and artifacts. Gudmundsson, S.

PB2579. Exome-wide association analyses of 1,463 proteins in 31k participants of the UK Biobank reveal the spectrum of coding variant effects on protein levels. Kim, H.

PB2581. Expansion of the histidine-rich domain of *DYRK1A* in a patient with developmental delay, short stature, and microcephaly: A case report. Fox, R.

PB2583. Exploiting a human stem cell-based modeling approach to functionally characterize loss-of-function of lncRNA RMST as a cause of Kallmann Syndrome. Stanton, L.

PB2585. Fine-mapping causal tissues and genes at disease-associated loci. Strober, B.

PB2587. Fragile X related genes and Dlg4 gene are involved in process formation in neuronal cells. Shimizu, H.

PB2589. Functional analysis of genetic variations found in clinical exomes of ASD patients. Bhattacharya, A.

PB2591. Functional characterization of a heat shock regulated enhancer at coronary artery disease associated locus that controls CALCRL expression in endothelial cells. Selvarajan, I.

PB2593. Functional Characterization of Novel UBE3A Variants Associated with Angelman Syndrome. Cousin, M.

PB2596. Gene discovery and pathogenic mechanisms of disease in worms, flies, and fish: The Model Organisms Screening Center for the Undiagnosed Diseases Network. Wangler, M.

PB2598. Gene prioritization for rare diseases integrating genotype, RNA-seq and phenotype - lessons from a CAGI 6 challenger team. Yepez, V.

PB2600. Genetic determinants of blood transcript splicing and impact on molecular phenotypes in 4732 healthy individuals. Tokolyi, A.

PB2602. Genetic effects on chromatin accessibility and nucleosome positioning using snATAC data across 284 skeletal muscle biopsies. Wang, X.

PB2604. Genetic regulation of OAS1 nonsense-mediated decay underlies association with COVID-19 hospitalization in patients of European and African ancestries. Florez-Vargas, O.

PB2606. Genetically Decreased CPS1 Activity Attenuates Atherosclerosis in Humans and Mice Through Sexually Dimorphic Patterns. Hilser, J.

PB2608. Genome-wide association study of serum metabolites and lipids in Multiple Sclerosis. Yang, C.

PB2610. Genome-wide microRNA expression quantitative trait loci from 596 adult brains. Vattathil, S.

PB2612. Genome-wide transcriptome correlation analysis identifies shared pathways between coronary heart disease and hematologic traits. Abe, H.

PB2614. Glucocorticoids unmask silent non-coding genetic risk variants for common diseases. Nguyen, T.

PB2616. Human genetic variation reveals regulators of *Yersinia pestis* cellular Infection. Keener, R.

PB2618. Hypotonia due to homozygous mutation of *NALCN*. Ope, O.

PB2620. Identification of polyadenylation signals relevant to Mendelian disease variant interpretation. Shiferaw, H.

PB2622. Identification of susceptibility loci for frailty through a genome-wide scans and genetic association study of aging-related variants in Korean population. Kwak, Y.

PB2624. Identifying functional variants related to heart development in African populations. Xie, N.

PB2626. Improving generalizability of gene expression prediction models from African-ancestry populations. Esquinca, E.

PB2627. Inhibiting aurora kinase A in the HMC3 immortalized human microglia cell line increases phagocytosis of amyloid beta *in-vitro.* Cook, K.

PB2630. Investigating the impact of non-coding structural variants flanking epilepsy-associated gene, *FOXG1* on chromatin architecture and gene regulation. Ramamurthy, A.

PB2632. Investigating the potential roles of Importin-7 and Importin-βin LINE-1 retrotransposition. Mortimer, S.

PB2634. KMT5B is required for early motor development. Feser Stessman, H.

PB2636. Leveraging Mosaic Epileptogenic Human Brain Tissue to Study Cell-type Specific Transcriptional Changes Associated with a Pathogenic*PIK3CA*Variant. Gade, M.

PB2638. Long-chain fatty acid oxidation disorder gene variants identified through a gene panel sponsored program: a diverse gene variant landscape. Rangel Miller, V.

PB2639. Mapping the effects of genomic deletions and duplications on cognitive ability across cortex. Kazem, S.

PB2641. Massively parallel saturation genome editing of an essential mitochondrial targeting sequence. Forrest, M.

PB2643. Mechanisms of adaptation in high-altitude pregnancy: association of genotype with oxygen delivery and placental metabolism. O'Brien, K.

PB2645. Metabolomics to clarify the mechanism of *ALDH2* polymorphism-induced alcoholic liver injury. Harada, S.

PB2647. Misexpression of inactive genes is associated with nearby rare structural variants. Vanderstichele, T.

PB2650. Modeling single-cell activation states enhances power to identify ex vivo stimulation response eQTLs. Valencia, C.

PB2652. Mosaicism of a *de novo TP53* mutation in a patient with breast cancer: value of two germ layers study. Aguilar, D.

PB2654. Mutant *Wac* mice exhibit phenotypes relevant to DeSanto-Shinawi Syndrome and provide initial insights into molecular and developmental mechanisms. Seban, N.

PB2656. NASP contributes to autism via epigenetic dysregulation of neural and immune signaling pathways. Li, J.

PB2658. Novel actionable targets discovered by proteomic genome-wide association of 1715 unstudied proteins. Kuliesius, S.

PB2661. Novel plasma and brain proteins that are implicated in multiple sclerosis risk. Lin, X.

PB2663. Nuclear abnormalities in novel LMNA variant segregating with LMNA-associated cardio cutaneous progeria syndrome. Vernet Machado Bressan Wilke, M.

PB2665. Parallel functional screening of human duplicated genes in neurodevelopment at single-cell resolution using zebrafish. Uribe-Salazar, J.

PB2667. --Pathogenic variants in *PLEKHO2* predispose to heritable thoracic aortic disease. Duan, X.

PB2670. PIEZO2 as a molecular candidate of respiratory phenotype induced by Congenital Central Hypoventilation Syndrome mutations in *PHOX2B*. Hedgecock, T.

PB2672. Population genetic study for 16 autosomal STR loci < and > for > in unrelated 15,546 Korean individuals. Park, H.

PB2674. Population-specific RNA-seq data empowers the functional follow-up of disease-associations from FinnGen. Benoit-Pilven, C.

PB2676. Predicted nasal epithelial transcriptome-wide association study in African-ancestry populations. Johnson, R.

PB2678. Profiling Local Ancestry Effects on Gene Regulation in An Admixed Population. Chen, H.

PB2680. Rare protein-coding variants in the insulin receptor gene (*INSR*) identified in women with polycystic ovary syndrome (PCOS). Bauer, R.

PB2682. Regulatory rare variant trait associations in 127,724 UK Biobank genomes. Ribeiro, D.

PB2683. Role of SNPs of Leptin gene in Etiology of Venous Thromboembolism among Females contraceptive users in Pakistan Population. Agha, Z.

PB2685. Seamless integrative pipeline for QTL datasets enhance the discovery of putative causal variants for Alzheimer's Disease. Cifello, J.

PB2687. Sex differences in gene expression affect host response to influenza A infection. Jones, A.

PB2688. Short chain fatty acid abnormalities: Genotype-Phenotype correlation in thirteen Puerto Rican patients. R. Pascual, J.

PB2689. Single-cell caQTL reveals cell-state-dependent regulation in immune cells in rheumatoid arthritis. Mu, Z.

PB2691. SMN modulates adverse effects associated with GEMIN5-mediated neurodevelopmental syndrome. Pandey, U.

PB2695. Targeted-NGS across the β-globin gene cluster identifies multiple linkage disequilibrium patterns in north Indian non-transfusion dependent β-thalassemia patients. Singh, N.

PB2697. The contribution of mitochondrial DNA variation to disease risk in a diverse cohort. Zaidi, A.

PB2699. The effect of rare protein-coding sequence variants on the human plasma proteome. Prins, B.

PB2701. The human myocardial transcriptomic response to the interaction effect of genetic variants and ischemic stimulus identified by a novel approach. Yazdani, A.

PB2703. The Multiple *de novo* Copy Number Variant (M*dn*CNV) phenomenon: peri-zygotic DNA mutational signatures and multilocus pathogenic variations. Du, H.

PB2705. The Unmasking Of 'Mitochondrial Adam' And Structural Variants Larger Than Point Mutations As Stronger Candidates For Traits, Disease Phenotype And Sex Determination. Singh, A.

PB2707. Two types of variants pointing out a crucial role for RNF216 in healthy and diseased brain. Versluys, L.

PB2709. Types of cis- and trans-gene regulation of expression quantitative trait loci across human tissues. Fu, A.

PB2711. UK Biobank whole genome sequencing and large-scale proteomics identifies novel protein quantitative trait loci (pQTLs) contributing to human disease. Hou, L.

PB2713. Uncovering sources of human gene expression variation in a globally diverse cohort. Taylor, D.

PB2714. Unsupervised Density Estimation for Noncoding Variant Effect Prediction. Rastogi, R.

PB2716. Using massively parallel reporter assays to dissect context-specific regulatory grammars in type 2 diabetes. Tovar, A.

PB2718. Variant-to-gene mapping at the insomnia *WDR90* locus and subsequent luciferase assay analyses implicates *PIGQ* as an effector gene in sleep dysregulation. Sonti, S.

PB2720. x-QTL: A mixture-model for identification and characterization of trans-eQTLs. Wu, C.

Omics Technologies Posters - Wednesday (Poster)

PB2866. A bioinformatics pipeline to aid in the design of allele-specific antisense oligonucleotides for patients with rare genetic conditions. Zhang, Q.

PB2868. A Comparative Bioinformatics Analysis to Propose a COVID_19 Candidate Vaccine Epitope at the ACE-Bionformatics in Mali (West Africa). Kouyate, M.

PB2870. A computational approach to design a COVID-19 vaccine against a predicted SARS-CoV-2 variant: high immunogenicity, efficacy and safety of DELLERA vaccine.   Paone, R.

PB2872. A Deep-learning based RNA-seq Germline Variant Caller. Cook, D.

PB2874. A framework for evaluation of new or modified sequencing technologies for use in human genomics. Gatzen, M.

PB2876. A Hybrid Machine Learning and Regression Method for Cell Type Deconvolution of Spatial Barcoding-based Transcriptomic Data. Yan, X.

PB2878. A massive proteogenomics screen identifies thousands of novel human coding sequences. Xing, J.

PB2882. A new framework for efficient Perturb-seq enables cheap large-scale dissection of the innate immune response and provides insight into regulatory eQTL relationships. Yao, D.

PB2884. A novel auto-normalizing pooled library construction method that streamlines sample preparation prior to targeted hybrid capture. Costello, M.

PB2886. A novel enzymatic fragmentation library preparation workflow that prevents sequencing artifacts for production-scale DNA-seq. Haimes, J.

PB2888. A rigorous benchmarking of methods for SARS-CoV-2 lineage detection in wastewater. Knyazev, S.

PB2890. A software platform for real-time and automated simultaneous analysis and detection of genetic diseases. Garrido Navas, M.

PB2892. A submodular fairness metric for sequence-to-function models in precision genomics. Robson, E.

PB2894. A two-channel deep learning framework for accurate prediction of the change in protein folding free energy upon mutations. Liu, Q.

PB2896. A validated clinical whole genome sequencing system for the detection of germline variants. Glidewell-Kenney, C.

PB2898. Algorithmic and Assay-based Simplification of Multiallelic Variants for Genotyping. Main, B.

PB2900. An algorithmic framework for isoform-specific functional analysis. Robinson, P.

PB2902. An evaluation of the Ultima Genomics sequencing platform: Scalable, high-throughput sequencing for low-cost whole genome sequencing. Coole, M.

PB2904. Analysis of pathogenic tandem repeat variation in congenital disorders. Altman, G.

PB2906. Application of DRAGEN Graph read alignment to challenging medically relevant genes and other difficult regions in GRCh38 and T2T-CHM13 genomes. Roddey, C.

PB2908. Assessing the impact of bioinformatics tools on genomic reproducibility: opportunities and pitfalls. Liu, F.

PB2910. Assessment of Whole Genome Sequencing Quality Metrics from a Large Cohort of Saliva-derived DNA Samples. Smith, G.

PB2913. Automated Walk away NGS Sample Preparation Using a Flexible Library Prep System with Enhanced Error Correction. Bigdeli, S.

PB2915. AUTOSurv: Interpretable autoencoder aided deep learning framework for breast cancer survival analysis incorporating multi-omics data. Jiang, L.

PB2917. Benchmarking algorithms for joint integration of unpaired and paired single-cell RNA-seq and ATAC-seq data. Lee, M.

PB2919. Benchmarking of computational methods for ancestry prediction from RNA-seq data. Yadav, A.

PB2921. Beyond the "one gene, one disease" paradigm with DIVAs, an Explainable AI tool for phenotype-driven digenic variants interpretation. Rizzo, E.

PB2923. Blended Genome Exome (BGE) Sequencing as an Alternative to SNP Arrays for Cost Effective Imputation of Variants in Globally Diverse Cohorts. DeFelice, M.

PB2925. Breast Cancer Specific Enhancer-Interactome Analysis Identifies Candidate Target Genes Enriched in Common Variant Risk Regions. Davis, B.

PB2927. bulk and single-cell gene co-expression network analysis of AD using PyWGCNA. Rezaie, N.

PB2929. Cas9 targeted enrichment and adaptive sampling for characterizing repetitive elements. Mumm, C.

PB2931. CCC: An efficient not-only-linear correlation coefficient based on machine learning. Pividori, M.

PB2933. Characterizing the genetic etiology of patients with neuromuscular disease through the integration of omics data. Triassi, V.

PB2935. Circulating proteomic signatures of pulmonary function. Lee, M.

PB2937. Clinical use of the amniotic fluid cells transcriptome in deciphering Mendelian disease. Lee, M.

PB2939. Combination of annotation enrichment approaches for characterization of blood-based RNA sequencing biomarkers. Listopad, S.

PB2941. Combined multiple structural variant algorithms achieved the highest recall rate in GIAB large deletion callsets. Chen, P.

PB2943. Comparison of exome performance in an inherited cancer context. Johnson, C.

PB2945. Competing sampling biases balance concordance of single-cell and nucleus RNA-seq. Chamberlin, J.

PB2947. Comprehensive detection of trait-associated structural variations using short read sequencing data. Terao, C.

PB2949. Comprehensive Structural Variant Detection: From Population to Mosaic level. Paulin, L.

PB2951. Cryptic splice variants contribute to the genetic etiology of congenital heart disease. Lesurf, R.

PB2953. D-CoMEx: Differential co-expressed module extraction to identify phenotype-specific miRNA biomarkers related to diseases. Kakati, T.

PB2956. Deep immune repertoire analysis of SARS-CoV-2 antibodies from a vaccinated human patient via mass-spectrometry and synthetic long reads. Castellana, N.

PB2958. Deep plasma protein profiling of Alzheimer's subjects with a novel unbiased and scalable proteogenomics approach. Siddiqui, A.

PB2960. DeepConsensus v0.3: Gap-aware sequence transformers for sequence correction. Belyaeva, A.

PB2962. DeepMod2: A deep learning framework for DNA and RNA modification detection using Oxford Nanopore sequencing. Ahsan, M.

PB2965. Detecting and assembling non-reference LINE-1 insertions using clustered long reads. Blacksmith, M.

PB2967. Detection of somatic variation in human neurons using long-read sequencing approaches. Zhou, W.

PB2969. Development of computational methods for analyzing single-cell spatial transcriptomic data with applications to murine spermatogenesis. Vargo, A.

PB2971. Diagnostic metabolomic profiling of hypo myelinating leukodystrophy caused by DEGS1 deficiency. Gijavanekar, C.

PB2975. Efficient DNA sample contamination metric estimation using a novel variant representation and algorithm. Lu, W.

PB2977. Enabling flexible low throughput sample preparation for multiple sequencing platforms using the Miro Canvas. Day, A.

PB2979. Ensure sample identity in sequencing workflows with the Twist Sample ID Kit. Oh, S.

PB2981. Establishing best practice for structural variant discovery with long read sequencing in the Gabriella Miller Kids First Pediatric Research Program using Sentieon's haplotype-resolved variant evaluation tool. Li, Z.

PB2983. Evaluating deep learning for predicting epigenomic profiles. Tang, Z.

PB2985. Evaluation of single nuclei RNA sequencing in brain tissue using Pre-templated Instant Partitions (PIPseq). Meltzer, R.

PB2987. Exome sequencing reanalysis complemented with combined multi-omics approach reached to 60% diagnostic yield in previously undiagnosed rare disease cohort. Ounap, K.

PB2989. Extensive differential cell type-specific gene expression and regulation by sex in human skeletal muscle. Hanks, S.

PB2991. faiGP: an evolutionary approach to discover governing equations in high-dimensional genomic data. Razavi, S.

PB2993. FastRNA: an efficient exact solution for PCA of single-cell RNA sequencing data based on a batch-accounting count model. Lee, H.

PB2995. Fixing falsely duplicated and collapsed regions of the GRCh38 reference genome. Behera, S.

PB2997. Gene regulatory network inference using single-cell multiome ATAC-seq and RNA-seq data. Wang, Y.

PB2999. Generative Adversarial Networks Create High-Quality Synthetic Prostate Cancer Magnetic Resonance Images. Ledesma, B.

PB3001. Genetics guided approach to infer unobserved covariates in functional genomics data. Yamamoto, R.

PB3003. Genome-wide prediction of chromatin profiles from gene expression. Fu, J.

PB3005. Genomic atlas of the plasma metabolome prioritizes metabolites implicated in human diseases: an analysis of the Canadian Longitudinal Study on Aging (CLSA). Chen, Y.

PB3007. GestaltMatcher supports classification of ultra-rare disorders and delineation of novel syndromes by facial phenotype descriptors. Hsieh, T.

PB3009. HD-Seq: A novel method for high accuracy sequencing. Shore, S.

PB3011. High throughput single-cell epigenomic profiling through split-pool combinatorial barcoding. Sayar, Z.

PB3013. High-throughput CRISPR Inhibition Screen to Improve Interpretation of Noncoding Variants in Developmental Epileptic Encephalopathies. Almanza Fuerte, E.

PB3015. High-throughput RNA isoform sequencing using programmable cDNA concatenation. AlKhafaji, A.

PB3017. High-Throughput Sample Processing workflow for rapid Vaginal microbiota profiling of women's health samples. Kelvekar, J.

PB3018. High-throughput targeted enrichment for long read third-generation sequencing: A flexible, scalable, and cost-efficient method achieving similar throughput as for short read NGS. Steiert, T.

PB3020. Human cytomegalovirus infection extensively re-organizes the human genome at human disease risk loci. Weirauch, M.

PB3022. Identification of FZD3 and SEMA3A as potentially secreted proteins involved in proliferative sickle cell retinopathy. Lima Camargo, A.

PB3024. Identifying high-confidence de novo mutations in somatic and germline cells through duplex sequencing of diverse tissue types. Lulla, S.

PB3026. Improved data quality for renal cell carcinoma samples with HIVE scRNAseq integrated storage. Sergison, E.

PB3029. Improving gene detection sensitivity from clinically relevant low input and FFPE RNA samples utilizing a rapid whole transcriptome library prep workflow. Sanders, T.

PB3031. Imputing functionally impactful *CYP2A6* structural variants from SNP array data. Langlois, A.

PB3033. Increased mitochondrial expression is associated with increased *APOEe4* expression and affected by ancestry and sex. Muniz Moreno, M.

PB3035. In-depth analysis and comparison of different whole genome methylation sequencing library preparations with low DNA input. Sun, Z.

PB3037. Induced pluripotent stem cells (iPSCs) for functional genetic screen applications. Isachenko, N.

PB3039. Inference of menopausal status from female reproductive tissue gene expression signatures. Theusch, E.

PB3041. Insights into Molecular Pathways Associated with Juvenile Myositis through Single-Cell Transcriptome Profiling. Kao, C.

PB3043. Integrated Proteotranscriptomics Analysis of Breast Cancer using Variational Autoencoders to Prioritize Pathogenic Genes. Jhee, J.

PB3045. Integrating Knowledge in Breast Cancer Subtyping Deep Learning Classifiers. Davidson, J.

PB3047. Integration of single nucleus and bulk adipose RNA-seq reveals distinct adipocyte subtypes for obesity and insulin resistance. Alvarez, M.

PB3049. Integrative single-nucleus multi-omics across 287 skeletal muscle biopsies reveals context-specific e/caQTL and extensive caQTL-GWAS colocalization. Parker, S.

PB3053. ioSearch: A tool for searching disease-associated interacting omics. Das, S.

PB3055. IsoMiGA: Combining long and short read RNA-seq in human microglia reveals novel isoforms and splicing events and insight into Alzheimer's disease. Brophy, E.

PB3057. Learning nonlinear causal relations in complex biological systems using a deep-learning approach and knockoff statistics. Park, H.

PB3059. Linking GWAS risk variants to disease genes by epigenomic mapping and prediction of functional enhancer-promoter interactions. Guo, Y.

PB3061. Long-amplicon variant-robust genome capture of SARS-CoV-2 using molecular inversion probes. Boyden, E.

PB3063. Long-read isoform sequencing reveals aberrant splicing of *PSEN2*, but not *PSEN1*, in individuals with sporadic Alzheimer's disease. Valdmanis, P.

PB3065. LongReadSum: A fast and flexible quality control tool for long-read sequencing data. Perdomo, J.

PB3067. Low-pass whole genome sequencing as a cost-effective and improved alternative to genotyping arrays. Zhang, P.

PB3069. Major cell-types in imbalanced multiomic single-nuclei datasets impact statistical modeling of links between regulatory sequences & genes. Leblanc, F.

PB3072. Max Read™: a novel high-throughput sequencing format for patterned flow cells. van Wietmarschen, N.

PB3074. Metagenomic analyses coupled with metabolic and deep immune profiling reveal coordinate effects on host-microbe interactions in chronic kidney disease   . Su, S.

PB3076. MHConstruct creates personalized full-length MHC locus haplotype sequences by finding optimal paths through a population variation graph.   Mumphrey, M.

PB3078. Mitigating challenges of large-scale single cell data management, querying, and analysis with REVEAL SingleCell. Sharma, K.

PB3079. Mitochondrial Genomic ratio (mitoscore) is an efficient biomarker to predict the implantation efficacy of human embryos. Chettiar, S.

PB3081. Multi-Omics Integration for Osteoporosis Prediction through Mixture-of-Experts Framework. Gong, Y.

PB3084. Neuroimmune insights through single-cell transcriptomics of paired brain and blood from living human subjects. Liu, D.

PB3086. New genotyping and sequencing methods for high throughput molecular blood group typing. Franke, A.

PB3088. Novel sequencing platform using an open fluidics architecture and mostly natural chemistry for cost-efficient whole genome sequencing. Lipson, D.

PB3090. ODER: Optimising the Definition of Expressed Regions, a publicly available R package to improve annotation of RNAseq data and investigate novel transcription using short-read RNA-sequencing data. Brenton, J.

PB3092. Optimizing microbiome extraction methods for human intestinal resections.. Ryu, E.

PB3094. Outcomes from the Care4Rare Canada unsolved research pipeline for 950 patients with non-diagnostic exome sequencing data. Boycott, K.

PB3096. PacBio HiFi sequencing provides highly accurate CpG methylation calls without bisulfite treatment. Saunders, C.

PB3098. Papillary renal cell carcinoma (pRCC) functional heterogeneity unraveled by single nucleus sequencing (Sn-Seq) technologies. Xin, R.

PB3100. Perturb-seq analysis reveals key mediators of TNFα-induced transcriptional response. Tedesco, D.

PB3102. PhaseDancer: Targeted assembly of the complex syntenic regions in non-human primates of the HSA2 fusion site. Poszewiecka, B.

PB3104. PIPseq, a novel and highly scalable technology based upon Pre-templated Instant Partitions (PIPs), is powering single-cell RNA sequencing into the million cell era. Fontanez, K.

PB3106. Population Scale Genetic Interpretation Software for Reporting Pathogenic and Likely Pathogenic Variants Impacting the ACMG59 Genes. Olson, R.

PB3108. Prokaryotic and viral genomes recovered from 787 Japanese gut metagenomes revealed microbial features associated with diets, populations, and diseases. Tomofuji, Y.

PB3113. PubCaseFinder: Implementation of the new function designing Mondo-based virtual gene panels to increase diagnostic efficiency for rare genetic diseases. Fujiwara, T.

PB3115. pyTCR: a comprehensive and scalable platform for TCR-Seq data analysis to facilitate reproducibility and rigor of immunogenomics research. Peng, K.

PB3117. Rapid whole genome and whole exome variant detection using a novel fluorescently labeled reversible terminated nucleotide sequencing system and GPU-based accelerated analysis. Looney, T.

PB3119. Regulome-wide association study identifies chromatin accessibility associated with pancreatic cancer risk. Liu, S.

PB3121. Rigorous benchmarking of HLA callers for RNA sequencing data. Ayyala, R.

PB3123. RNA monsters generated by human mitochondrial RNA polymerase. Greenwald, E.

PB3125. SARS-CoV-2 genomic surveillance in Rwanda: Introductions and local transmission of the B.1.617.2/Delta variant of concern. Butera, Y.

PB3127. Searching for Somatic Variation in Rapid-onset Obesity with Hypothalamic dysfunction, Hypoventilation and Autonomic Dysregulation (ROHHAD). Barclay, S.

PB3129. Sentieon DNAscope LongRead: a highly accurate, fast, and efficient pipeline for germline variant calling from PacBio HiFi reads. Freed, D.

PB3131. Sequence-to-expression models of compact promoters for cell-type-specific promoter design. Reddy, A.

PB3133. Sequencing By Binding (SBB) enables a lower limit of detection for Tuberculosis resistance genes *gyrA* and *katG.* Wike, C.

PB3135. Simplified RNA-Seq Library Prep: Improved RNA sequencing results for FFPE and blood samples. Putnam, E.

PB3137. Simultaneous dimensionality reduction and cell-type annotation of single-cell RNA-seq data using marker enriched uniform manifold approximation and projection. Khan, A.

PB3140. Single neuron whole genome sequencing for somatic CNVs using three different genome amplification methods. Proukakis, C.

PB3142. Single-cell dissection of ALS and frontotemporal dementia in the human motor and prefrontal cortices.   Pineda, S.

PB3144. Single-cell multi-omics reveals gene regulation dynamics during mouse secondary palate development. Yan, F.

PB3146. Single-molecule, modified base sequencing to identify frequency and cause of rAAV vector breakpoints. Thompson, J.

PB3148. Single-stranded library preparation for cfDNA identifies unique fragment length signature in myelodysplastic syndrome with multi-lineage dysplasia and ring sideroblasts. Schwartz, C.

PB3151. sparQ mRNA-Seq: Consistent and high-quality mRNA library preparation from both abundant and limiting samples. Conley, K.

PB3153. STRT-N: A newly optimised single-cell RNA sequencing method. Boskovic, N.

PB3155. Structuring information via an immune-focused ontology enables the construction of a high-quality knowledge graph for the study of autoimmune diseases. Truong, V.

PB3157. Systematic evaluation of transcriptome sequencing applied to real-world rare disease cohorts: Insights and limitations. Silverstein, S.

PB3159. Targeted sequencing of a 1 Mb carrier screening panel using molecular inversion probes. Amaral, J.

PB3161. Targeted transcriptome sequencing enables exponential scaling of combinatorial barcoding. Tran, V.

PB3163. Temporal motif discovery in biological interaction networks. Jazayeri, A.

PB3165. The impact of insert length on variant calling quality in whole genome sequencing. Lajoie, B.

PB3167. Transcriptome analyses of congenital heart disease tissue from participants with Trisomy 21. Morton, S.

PB3169. Transcriptome signature of sodium intake in and links to cardiovascular traits. Gaye, A.

PB3171. Transcriptomic analysis reveals long-term effect of SARS-CoV-2 infection on host genetic regulation. Zhu, W.

PB3172. Ultrasensitive Duplex Sequencing for Diverse Applications on the Singular Genomics G4 Platform. Fitzgerald, D.
PB3174. Undergraduate research assistant. Williams, M.
PB3176. Using an NGS readout for high-throughput proteome-wide analysis in large population health studies. Lawley, C.
PB3178. Using single cell CRISPR/dCas9-based regulatory element screening to dissect complex genetic loci. Bounds, L.
PB3180. Utilization of Agena massARRAY to improve quality assessment of saliva samples for clinical genome sequencing. Ho, C.
PB3182. Utilizing paraformaldehyde fixation to expand opportunities for single-cell RNA-sequencing. Meta, B.
PB3184. Visualize RNA biomarkers to spatially interrogate complex tissues using the RNAscope™ HiPlex v2 in situ hybridization assay. Dikshit, A.
PB3186. Whole genome sequencing of low input tagmentation-based libraries results in high quality somatic variant calling comparable to ligation-based PCR-free libraries. Rice, E.

Pharmacogenomics Posters - Wednesday (Poster)
PB2721. A Pharmacogenetic Integration Solution for Clinical Decision Support Software. Katzov-Eckert, H.
PB2723. Accurate CYP2D6 star (*) allele diplotyping for long-read PacBio HiFi sequencing. Harting, J.
PB2725. An NGS approach to detecting novel LOF variants in *DPYD* across >69,000 individuals. Kim, G.
PB2727. Convergence of bipolar disorder treatments and gene knockdown on the transcriptome. Vuokila, V.
PB2729. DEEPCT(Deep-learning-based Clinical Trial) simulates a drug clinical trial by exploiting germline variants as random assignment of drug. Kim, J.
PB2731. Elucidating the multi-omic drivers of hepatic drug metabolism in African Americans. Clark, C.
PB2733. Evaluating the frequency and the impact of pharmacogenetic variants in an ancestrally diverse Biobank population. Keat, K.
PB2735. Genetic associations of iron absorption among individuals of East Asian and European descent. Barad, A.
PB2736. Genomic analysis of hydroxychloroquine-associated retinal toxicity in a large cohort. Ullah, E.
PB2738. Implementation of clinical pharmacogenomics: How NCBI's Medical Genetics Summaries and MedGen can help educate and support clinicians. Malheiro, A.
PB2740. LOF *CYP2C19* allele evaluation identifies significant number of Indian CAD patients are low responders to Clopidogrel. Rastogi, G.
PB2742. Novel genetic signals identified for angiotensin-converting enzyme inhibitor-induced cough. Coley, K.
PB2744. Pharmacogenomic analysis of drug metabolizing enzymes associated with plasma clozapine/N-desmethylclozapine ratio. Park, S.
PB2746. Predicted expression of enzymes in the thiopurine metabolic pathway and risk of side effects in patients with inflammatory conditions treated with azathioprine Daniel, L.
PB2749. The Alabama Genomic Health Initiative Integrating Genomics into Primary Care. Limdi, N.
PB2750. Vaccinations against pneumonia and the flu may protect carriers of particular genotypes against Alzheimer's disease. Ukraintseva, S.
PB2752. Variation in *CYP2A6*, a nicotine metabolism gene, associates with lung diseases: A Phenome-wide Association and Mendelian Randomization study. Giratallah, H.

Prenatal, Perinatal, and Developmental Genetics Posters - Wednesday (Poster)
PB2065. Assessing the accuracy of variant detection and coverage bias in mitochondrial DNA (mtDNA) after whole genome amplification of single fibroblast cells with known mtDNA heteroplasmic mutations to effectively optimize preimplantation genetic testing of mtDNA related disorders. Novoselska, A.
PB2067. Assisted reproductive technology and DNA methylation at imprinted genes. Lyle, R.
PB2069. Cell free fetal (cff) DNA assessment using Y- chromosome specific sequences and amplicon NGS of targeted SNPs for possible implementation in non-invazive prenatal testing of common chromosomal aneuploidies. Vodicka, R.
PB2071. Characterization of the prenatal presentation of novel truncating variants in *ANKRD11.* Van Ziffle, J.
PB2074. Contribution of BBS9 isoforms to the etiopathogenesis of non-syndromic craniosynostosis. Sibilia, D.
PB2076. Developmental programming of vitamin b12 deficiency model in zebrafish. Verma, A.
PB2078. Discordances among NIPT, QF-PCR and cytogenetic results in prenatal cases. Thuriot, F.
PB2080. Down Syndrome therapeutics: The integrated stress response and restoration of protein synthesis. Korenberg, J.
PB2081. Explore SOX9 haploinsufficiency in neural stem cells. Chan, S.
PB2083. Fetal genetic autopsy: A five-year retrospective audit monocentric audit. Martinovic, J.
PB2085. Genetic Characterization of VACTERL Association-Like Congenital Malformations. Pang, H.

PB2087. Genetic determinants of gestational length in Indian women: A two-stage genome-wide association study. Bhattacharjee, E.

PB2089. Heteroplasmy dynamics and segregation in pre-implantation embryos: Rare insights from complete mitochondrial genome sequencing of clinical trophectoderm biopsy. Aggarwal, A.

PB2091. Host genomics and human milk exposure are associated with sex-specific changes in the gut microbiota of infants with asthma in the CHILD Cohort Study. Stickley, S.

PB2093. Identifying genes associated with preeclampsia using whole exome sequencing data from participants in the Penn Medicine BioBank. Xiao, B.

PB2095. Investigating uptake and impact of genetic and genomic evaluation following a perinatal demise. D'Orazio, E.

PB2097. Large-scale genome-wide association study meta-analysis of Hyperemesis Gravidarum confirms the nausea and vomiting hormone gene *GDF15* is the greatest genetic risk factor and identifies additional risk loci. Fejzo, M.

PB2099. Leveraging duplex DNA sequencing to investigate germline mutation rates in the context of sperm selection and paternal age. Kunisaki, J.

PB2102. Molecular Epidemiological Status of Group B Streptococcus in Ile Ife South Western Nigeria. Omololu-Aso, J.

PB2104. *Nlrp2* is a Maternal Effect Gene Required for both Placental and Embryonic Development. Sharif, M.

PB2106. Ovarian subfertility genetic variants in females with unexplained infertility. Roman, R.

PB2108. Phenome and genomewide recessive scan of coding variants reveals novel associations for female infertility. Ruotsalainen, S.

PB2110. Placenta fraction of maternal blood cell-free RNA associates with pre-eclampsia during pregnancy. Yan, Q.

PB2112. Prenatal and postnatal changes in chromatin accessibility and gene expression in mouse gut development for understanding Hirschsprung disease (HSCR). Chubaryov, H.

PB2114. Prenatal diagnosis of recurrent severe form of infantile galactosialidosis exemplifying a pitfall in parental carrier screening. Araújo Castro, M.

PB2116. Prenatal onset of Spondyloepimetaphyseal Dysplasia SIK3 Type (SEMDK). Wachtell, D.

PB2118. Reconsidering the "advanced" maternal age for invasive prenatal testing. Maya, I.

PB2120. Rescue of limb hemorrhage in a cohesinopathy mouse model with severe limb reduction. Strasser, A.

PB2122. Single-nucleus chromatin accessibility and RNA sequencing reveal impaired brain development in prenatally e-cigarette exposed neonate's brain. Chen, Z.

PB2124. Successful implementation of a short turn-around time prenatal exome sequencing process in a perinatal centre. Barnett, C.

PB2126. The abnormal expression of has-miR-196b-5p, FAS, and FAS-AS1 genes in Iranian patients with recurrent pregnancy loss. Ghaderian, S.

PB2128. Understanding the burden of clonal and non-clonal sperm mosaicism by bulk and single-cell genome sequencing. Yang, X.

PB2130. Variant reclassification in expanded carrier screening brings challenges for clinical management: experience of a Hong Kong prenatal diagnosis centre. Zheng, Y.

PB2132. Whole-genome sequencing analysis in families with pregnancy loss. Workalemahu, T.

Statistical Genetics and Genetic Epidemiology Posters - Wednesday (Poster)

PB3258. 8 Billion Polygenic Risk Score distributions: building unique reference populations based on genetic ancestry to increase the performance of PRS. Busby, G.

PB3260. A fast Bayesian screen to identify pleiotropic loci and describe pleiotropic profiles. Huo, S.

PB3262. A framework to improve the interpretation and prediction of variant effect sizes using non-linear functional models. Cheng, S.

PB3264. A generalized framework to decorrelate family or population structure that can easily incorporate epistasis, GxE interaction or multiple-variant analysis in GWAS and NGS studies. Li, D.

PB3266. A genome-wide search for parent-of-origin effects in 4,505 unrelated individuals identifies the *8q21.3* locus as a novel genetic regulator of Factor V plasma levels. Gendre, B.

PB3268. A machine-learning approach for disease prediction improves GWAS and polygenic scores. Eick, L.

PB3270. A network based approach for fine-scale population structure inference in genetic datasets. Shemirani, R.

PB3271. A novel approach identified eight gene x alcohol or gene x smoking interactions that contribute to serum lipids. Zhu, X.

PB3273. A Novel Gene-Based Test for Sequencing Studies Based on a Bayesian Variable Selection of Rare Variants. Xu, J.

PB3275. A novel method for annotation-agnostic differential transcript usage analysis. Eldjarn Hjoerleifssoon, K.

PB3277. A pipeline for predicting pathogenicity of monogenic cardiovascular disease genetic variants. Ramaker, M.

PB3279. A polygenic score method including recessive model for improving odds ratio. Ota, R.

PB3281. A Rare Missense Variant of Large Effect is Associated with Cataract in Puerto Ricans. Shi, J.

PB3283. A scalable framework for robust linear mixed model association testing. Kalantzis, G.

PB3285. A scalable statistical framework for genome-wide interaction testing harnessing cross-trait correlations with an application to Alzheimer's Disease. Bian, S.

PB3287. A statistical framework for integrative genetic association analysis. Okamoto, J.

PB3289. A transcriptomic signature of late-life depression. Matan-Lithwick, S.

PB3292. Accurate and cost-effective imputation of genotypes at whole genome level with Sparse Denoising Autoencoders. Loguercio, S.

PB3294. Accurate *in silico* confirmation of rare copy number variant calls from exome sequencing data using transfer learning. Tan, R.

PB3297. Admixture mapping for phenome-wide discovery in a diverse health system biobank. Cullina, S.

PB3299. Allelic imbalance of chromatin accessibility in cancer identifies candidate causal risk variants and their mechanisms. Grishin, D.

PB3301. AlphaCluster: Coevolutionary driven residue-residue interaction models enable quantifiable clustering analysis of *de novo* variants to enhance predictions of pathogenicity. Obiajulu, J.

PB3303. An accurate and efficient causal gene network inference method that handles many confounding variables. Kvamme, J.

PB3305. An alternative approach to the estimation of biological age. Fischer, K.

PB3307. An Exomiser-based R pipeline for gene burden testing aids novel gene discovery in rare Mendelian diseases. Cipriani, V.

PB3309. Analysis of follow up data in large biobank cohorts: a review of methodology. Kolde, A.

PB3311. Ancestry-specific eQTL associations in HIS/NHW Alzheimer's disease cohorts. Wheeler, N.

PB3313. Application of the colorectal cancer polygenic risk score to the VA CSP #380 "Prospective Evaluation of Risk Factors for Large (≥1cm) Colonic Adenomas in Asymptomatic Subjects" longitudinal colonoscopy screening cohort. Qin, X.

PB3315. Assessing the safety of lipid-modifying medications among East Asian adolescents using genetics: Evidence from Hong Kong's "Children of 1997" birth cohort. Luo, S.

PB3317. Assessment of correlated multi-SNP Mendelian Randomization method using brain eQTLs to discover schizophrenia genes as a case study. Baird, D.

PB3318. Association Between Brain Structure and Alcohol Use Behaviors in Adults: A Mendelian Randomization and Multiomics Study. Mavromatis, L.

PB3320. Association testing in admixed and multiethnic data sets nominate genetic modifiers of age at onset of Alzheimer's disease. Blue, E.

PB3322. Auxiliary classifier generative adversarial network for phenotypic conditional genomic data synthesis. Lim, E.

PB3324. Bi-ancestral phenome-wide association of complement component 4 haplotypes in 550,000 individuals. Venkatesh, S.

PB3327. Broad- and narrow-sense heritability-contributing regions are shared across age, genetic sex and geographical cohorts. Udwin, D.

PB3329. Building ancestry-informed tools for more inclusive genomics and improved clinical interpretation in admixed populations. Atkinson, E.

PB3331. Calcium signaling in Alzheimer's disease: Insights from *PTK2B* and *miR146a* interaction analyses. Yashin, A.

PB3333. CARMA: Novel Bayesian model for fine-mapping with high-dimensional functional data. Yang, Z.

PB3335. Cell composition inference and identification of layer-specific transcriptional profiles with POLARIS. Chen, J.

PB3337. Characterizing features affecting Local Ancestry Inference in Latin American populations. Mauer, J.

PB3339. Colorectal cancer polygenic risk score is inversely associated with eye phenotypes. Rosenthal, E.

PB3341. Common and Rare Variants Genetic Association Analysis of Late-Onset Alzheimer's Disease in 500,000 Exomes from UK Biobank. Hwang, Y.

PB3343. Comparing methods to adjust for fine-scale population structure in rare variant analyses. Marker, K.

PB3345. Comparison of trans-ancestry meta- and mega-analyses of cis-eQTLs in whole blood and LCLs. Nandakumar, P.

PB3347. Controlled Discovery and Localization of Causal Variants via Bayesian Linear Programming. Spector, A.

PB3349. Credible set determination for multi-ancestry fine-mapping. Shen, J.

PB3351. *De novo* mutation hotspots in homologous protein domains identify function-altering mutations in neurodevelopmental disorders. Hampstead, J.

PB3352. Deep Learning-based Phenotype Imputation on Biobank-scale Data Increases Genetic Discoveries. An, U.

PB3354. Deep sequencing of DNA from urine of kidney allograft recipients to estimate donor/recipient-specific DNA fractions. Belkadi, A.

PB3356. Deep-PheWAS.R: an innovative R package for phenome wide association of novel phenotypes. Packer, R.

PB3358. Deprivation index as a predictor in PheWAS recapitulates associations between socio-economic status and phenotypes captured by ICD9 codes. March, M.

PB3360. Detecting shared and sex-specific causal genetic effects on complex traits. Zhang, W.

PB3362. Detecting variable-number tandem repeat (VNTR) variation using Oxford Nanopore sequencing data at population scale. Beyter, D.

PB3364. Developing an African American population-based transcriptome prediction model from the GENE-FORCAST cohort. Goodney, G.

PB3366. Differential gene expression analysis based on linear mixed model corrects false positive inflation for studying quantitative trait. Tang, S.

PB3368. Differentiate association and causal effects of cardiovascular disease risk factors on Alzheimer's disease and its age-at-onset. La, J.

PB3370. Discovery of rare variants associated with alcohol problems improved by leveraging machine learning phenotype prediction and empirical functional variant weighting. Ahangari, M.

PB3372. Disentangling the genetic overlap and causal relationship between primary open-angle glaucoma, brain morphology, and major neurodegenerative disorders. Gharahkhani, P.

PB3374. Diversity is key for POAG: the proportion of variance for primary open-angle glaucoma explained by covariates and genetic risk scores varies by ancestral group in the Million Veteran Program. Waksmunski, A.

PB3376. DosaCNV: A deep multiple instance learning framework for jointly predicting copy number variation pathogenicity and gene dosage sensitivity. Liu, Z.

PB3380. Efficient tests of marginal epistasis reveal the impact of interactions on complex traits. Pazokitoroudi, A.

PB3382. Estimating heritability explained by local ancestry and stratification bias in admixture mapping from summary statistics. Chan, T.

PB3384. EVai's "Suggested Diagnosis" feature: a new AI-based method to increase diagnostic yield in Rare Disease Patients. Limongelli, I.

PB3386. Evaluating human genetic evidence for existing drug targets in the UK Biobank. McInnes, G.

PB3388. Evaluating the accuracy genotype imputation in selected African populations. Nanjala, R.

PB3390. Evaluation of GENESIS, SAIGE and REGENIE for genome-wide association study of binary traits in correlated data. Gurinovich, A.

PB3392. Exome sequencing of 628,388 individuals identifies common and rare variant associations with clonal hematopoiesis phenotypes. Kessler, M.

PB3394. Exploring germline genetics of *in situ* and invasive cutaneous melanoma. Ingold, N.

PB3396. Expression and Splice QTLs found in COVID-19 patients and controls show differential colocalization and heterogeneity between infectious and non-infectious states.   Farjoun, Y.

PB3398. Extremely sparse models of linkage disequilibrium in diverse GWAS. Wohns, A.

PB3400. fSuSiE: new approach for fine mapping functional phenotypes. William, D.

PB3403. Gene-by-gene and gene-by-sex interactions identified in the UK Biobank using Explainable Boosting Machines. Nagafuchi, Y.

PB3404. Gene-sodium intake interactions reveal new blood pressure loci in a multi-ancestry genome wide association study in UK Biobank and CHARGE. Kho, M.

PB3406. Genetic determinants of a metabolic model for age. Xiao, S.

PB3408. Genetic evidence of assortative mating in the Japanese population. Yamamoto, K.

PB3410. Genetically predicted counts of white blood cells are associated with lower risk of obesity: a Mendelian randomization study. Sun, Y.

PB3412. Genome wide association study of asthma in 1587 French Canadian subjects. Eslami, A.

PB3414. Genome-wide analysis of dental caries variability identifies genotype-by-environment interactions. Zou, T.

PB3416. Genome-wide association studies of metabolic traits in Samoans. Wehr, J.

PB3418. Genome-wide association study of the human metabolome in diverse ancestries identifies an association of the *OPLAH* locus with 5-oxoproline (pyroglutamic acid) in individuals with African ancestry. Krishnan, M.

PB3420. Genome-wide imputed differential expression enrichment (GIDEE) analysis identifies trait-relevant tissues. Nyholt, D.

PB3423. GrafPop: A tool to quickly infer subject ancestry from multiple large genotype data sets. Jin, Y.

PB3425. GWAS of breast cancer family history reveals evidence for indirect genetic effects on maternal cancer risk. Sun, Z.

PB3427. GWAS of serum ALT and AST using whole genome sequencing data for 119,009 UK Biobank participants yields novel associations with rare variants. Holleman, A.

PB3429. Haplotype-based fine-mapping of variant associations for complex traits. Massarat, A.

PB3431. HLA Analysis Tool Kit v2.0. Choi, W.

PB3433. HybridGWAIS-Web: A fast and secure epistasis detection web service. Wienbrandt, L.

PB3435. Identification of key biomarkers associated with obesity using multi-omics data integration. Liu, A.

PB3438. Identifying common genetic susceptibility underlying comorbid phenotypes using Binomial Regression. Ghosh, S.

PB3440. Identifying functionally-related SNPs and genes from phenome-wide genetic effect correlations using GWAS summary statistics. Chen, L.

PB3442. Identifying the conserved versus divergent structural sequences of the human pangenome. Lee, H.

PB3444. Imaging derived phenotypes of the knee reveal novel genetic and clinical risk factors associated with knee osteoarthritis severity. Flynn, B.

PB3446. Implementing Mendelian Randomization to Assess Foetal Risk from Intrauterine Prescriptive Drugs for the Treatment of Diabetes, Hypertension and Thyroidism in Pregnancy. Barry, C.

PB3448. Improved genotyping of rare variants from AppliedBiosystems™ Axiom™ microarrays, using Support Vector Machine (SVM) prediction models. Webster, T.

PB3449. Improved Prediction Performance of Polygenic Risk Score with Gene Expression Data. Xu, H.

PB3451. Improvements in performance of polygenic risk scores for Alzheimer's disease in the Midwestern Amish using founder population-specific weights. Osterman, M.

PB3453. Improving fine-mapping by modeling infinitesimal effects. Cui, R.

PB3455. Impseer: Machine learning prediction of imputation quality. Saad, M.

PB3457. In silico GWAS: the rapid and accurate (pre)computation of genetic disease associations using only population-level data. Foley, C.

PB3459. Incorporating functional annotations of rare variants in interpretable neural networks identify novel Alzheimer's disease risk loci. Das, A.

PB3461. Inference for set-based effects in genome-wide association studies with multiple interval-censored outcomes. Choi, J.

PB3463. Integrating external controls by regression calibration for genome-wide association study. Zhu, L.

PB3465. Integration of gene expression data in Bayesian association analysis of rare variants. Zhong, G.

PB3467. Integrative approaches to unravel sex- and age-associated gene signatures and networks. Johnson, K.

PB3469. Inverted genomic regions between reference genome builds in humans impact imputation accuracy and decrease the power of association testing. Chiang, C.

PB3471. Investigating the effect of body size between menarche and first birth on breast cancer risk in later life: A lifecourse Mendelian randomization study. Power, G.

PB3473. Investigating the potential impact of PCSK9-inhibitors on mood disorders using eQTL-based Mendelian randomization. Aman, A.

PB3475. Investigation of Genetic Variants and Causal Biomarkers Associated with Brain Aging. Lee, J.

PB3477. Is the portability of polygenic scores a matter of genetic ancestry alone? Wang, J.

PB3479. Joint analysis of GWAS and multi-omics QTL summary statistics reveals a large fraction of GWAS signals shared with molecular phenotypes. Wu, Y.

PB3481. Leveraging cohorts from diverse ancestries to uncover generalizable genotype driven fetal- and adult-specific brain transcriptomic mechanisms in psychiatric disorders. Jajoo, A.

PB3483. Leveraging functional genomic annotations and genome coverage to improve polygenic prediction of complex traits within and between ancestries. Zeng, J.

PB3485. Leveraging multiple reference panels by stacked regression TWAS identifies 6 novel independent risk genes for Parkinson's disease. Parrish, R.

PB3487. Leveraging polygenic risk scores of biomarkers to identify obesity endotypes. Bagheri, M.

PB3489. Leveraging tissue-wide functional regulatory information improves portability of trans-ancestral polygenic risk score applications. Crone, B.

PB3491. Local ancestry at the MHC region is associated with disease heterogeneity in a multi-ethnic lupus cohort. Solomon, O.

PB3493. Longitudinal, multi-modal EHR prediction of incident CKD and ESRD for risk stratification and genetic studies. Banerjee, N.

PB3495. Low-coverage sequencing induces uncertainty in polygenic scoring. Petter, E.

PB3497. LTPI: A generalized liability threshold model combining dichotomous and continuous phenotypes in EHR increases the association power. Lee, C.

PB3499. Machine Learning Approaches to Genome-wide association studies. Enoma, D.

PB3501. Machine learning guided transcriptomic analyses reveal a novel 9-gene module associated with schizophrenia risk. Guan, F.

PB3503. MagicalRsq: Machine learning based genotype imputation quality calibration. Sun, Q.

PB3504. MANOCCA : An innovative powerful test to detect predictors of the gut microbiome variability. Boetto, C.

PB3506. MATS: A novel multi-ancestry transcriptome-wide association study to account for heterogeneity in the effects of cis-regulated gene expression on complex traits. Pan, W.

PB3508. Mendelian randomization and colocalization of amino acids in the UK Biobank NMR metabolomic profiles. Willis, C.

PB3510. Mendelian randomization indicates sex-specific causal effects of estradiol levels on kidney function. Nasr, M.

PB3512. Metabolite prediction models in UK Biobank: A metabolome-wide association study (MWAS). Huang, L.

PB3514. Methods and software for empirical Bayes multivariate multiple testing and effect size estimation. Yang, Y.

PB3516. Metric projection—AUCs, correlations, and heritability—for biobank scale data: using sparse prediction in the UK Biobank to plug diversity gaps in future biobanks. Raben, T.

PB3520. Multi trait GWAS for diverse ancestry : Mapping the knowledge gap. Julienne, H.

PB3522. Multi-ancestry polygenic risk scores for venous thromboembolism. Jee, Y.

PB3525. Network-based cross-phenotype risk scoring models for non-linear compositing multiple disease risks using biobank-scaled PheWAS data. Nam, Y.

PB3527. New perspectives in the genetics of persistent opioid use: reexamining candidate gene studies and presenting new results from the Michigan Genomics Initiative. Annis, A.

PB3529. NGS Variant Detection as a Sequence-to-Sequence Modeling Problem. O'Fallon, B.

PB3531. Novel Approach to Age Estimation of Rare Variants Enables Prioritization of Functional Variants. Liao, K.

PB3533. Omnibus proteome-wide association study (PWAS) method identified 28 novel risk genes for Alzheimer's disease. Hu, T.

PB3535. One-sample and two-sample mendelian randomization studies consistently showed that an increase in adiposity raises serum uric acid level in the East Asian populations. Lee, S.

PB3537. PathWAS: Combining transcriptomics with proteomics to predict pathway functionality and associations with complex traits. May-Wilson, S.

PB3539. Pathway analysis using expression factors reveals biological processes underlying genetics of complex traits. Sun, X.

PB3541. Personality traits are consistently associated with blood mitochondrial DNA copy number estimated from genome sequences in two genetic cohort studies. Oppong, R.

PB3543. Phenotype integration improves power and preserves specificity in biobank-based genetic studies of MDD. Dahl, A.

PB3545. Phenotypic subtyping via contrastive learning reveals heterogeneity in the genetic architecture of traits. Gorla, A.

PB3547. PLEIOVAR - Assessing Pleiotropic Effects of SNPs on Traits in Genome-wide Association Studies. Meirelles, O.

PB3549. Polygenic risk scores for coronary heart disease in diverse populations using population-specific optimization. Smith, J.

PB3551. Polygenic scores for insulin resistance are associated with brain volumes in the NHLBI Trans-Omics for Precision Medicine (TOPMed) Program. Sarnowski, C.

PB3553. PopMLvis: A Tool for Analysis and Visualization of Population Structure in Genome-Wide Association Studies. Elshrif, M.

PB3556. Practical screening for differences in predictive value of polygenic scores across the phenotypic range. Mefford, J.

PB3557. Predicting cancer risk from germline next-generation sequencing data using a novel context-based variant aggregation approach. Guan, Z.

PB3559. Predicting how our genetic variants regulate splicing and impact human phenotypes. Jacobs, H.

PB3561. Prediction of congenital heart disease subgroups associated with copy number variants using a multinomial machine learning-based classifier. Penaloza, J.

PB3563. Prospective analysis of disease incidence and progression with genetic, clinical and lifestyle risk factors. Wang, W.

PB3565. Proteome-Wide Association Analysis in the Women's Health Initiative Study. Chen, B.

PB3567. PUMICE-S: Prediction Using Models Informed by Chromatin conformations, Epigenomics and Summary Statistics. Wang, L.

PB3569. Quantifying the causal impact of modifiable risk factors and biomarkers on total healthcare burden via Mendelian randomization. Lee, J.

PB3571. Rapid identification of clusters of multi-level colocalized traits within hundreds of candidate traits and in the presence of multiple causal variants. Kuncheva, Z.

PB3572. Rare variation analyses of over 6,900 ALS cases in a multi-ethnic population. Pottinger, T.

PB3573. Rare-variant aggregation analysis reveals new genes involved with IgG glycosylation. Klaric, L.

PB3575. Reaching 95.0 years of age: The genetic association in the elderly Croatian population. Celinšcak, Ž.

PB3577. Reliability of genotyping arrays for detection of rare variants: design & performance of a bespoke array focusing on obesity & diabetes. Almansoori, S.

PB3579. SDPRX: A statistical method for cross-population prediction of complex traits. Zhou, G.

PB3583. Shared genetic etiology and causality between COVID-19 and venous thromboembolism: evidence from genome-wide cross trait analysis and bi-directional Mendelian randomization study. Liu, Z.

PB3585. Shared Genetics Drive Mate Selection. Burghardt, K.

PB3587. Simple phenotype transformations help elucidate genetic architecture in the tails of biomarker distributions. Baya, N.

PB3589. Single-cell transcriptomic landscape in Alzheimer's disease. Swarup, V.

PB3591. Specific HLA risk alleles associated with the highly variable region of T cell receptors (TCRs) in Leukemia, Melanoma and COVID-19 patients. Han, J.

PB3594. Supervised modeling on CRISPR gene regulatory screening and cell-type-specific gene regulatory landscapes to prioritize GWAS Targeted Genes. Tsai, M.

PB3596. Systematic Integration of Multi-omics Data for the Study of Coronary Artery Disease and Subclinical Atherosclerosis. Yang, C.

PB3598. The effect of mental diseases on chronic kidney disease: a bidirectional Mendelian randomization study. Yu, S.

PB3600. The hidden factor: the importance of accounting for covariate effects in power and sample size computation when analyzing a binary trait. Zhang, Z.

PB3602. The impact of genetic variation on hearing acuity variability. Duran, J.

PB3604. The largest deep-coverage whole genome seq meta-analysis on osteoporosis identified novel GWAS loci. Yu, F.

PB3606. The relationship between kinship and heritability estimation accuracy. Chen, D.

PB3608. The unusual suspects: genome-wide significant sex-difference in minor allele frequency in gnomAD v3.1.2 whole genome sequence via a novel population-aware retrospective regression. Sun, L.

PB3610. Tissue-specific impacts of aging and genetics on gene expression patterns in humans. Chung, R.

PB3614. Transcript-aware gene burden analyses in the UK Biobank reveal isoform-specific compositions and functions. Jakubosky, D.

PB3616. Transcriptomics-based drug-repurposing screen discovers potential novel therapies for primary open-angle glaucoma. Duchinski, K.

PB3617. Transferability of polygenic scores for anthropometric traits to the Native Hawaiian population. Lo, Y.

PB3620. Uncovering ancestry-specific differences in genetically driven transcriptomic dysregulation in schizophrenia and bipolar disorder. Mathur, D.

PB3622. Unravelling the genetic contributions to gestational duration in Autism Spectrum Disorder individuals: results from the SPARK sample. Chatzigeorgiou, C.

PB3624. Using external reference panel and meta-analysis summary statistics for rare-variant aggregation tests. Ryan, B.

PB3628. Variants in *CRY2* and *MTNR1B* influence the diurnal variability of blood glucose levels. Jones, S.

PB3630. Winner's curse in rare variants association analysis: bias depends on effect direction and the pooled methods used. Soave, D.

Epigenetics Posters - Wednesday (Poster)

PB2363. A long short-term memory autoencoder approach for detecting CRISPR genomic edits. Hinson, P.

PB2365. A simple workflow for methyl DNA analysis in FFPE-preserved Alzheimer and other neurodegenerative brain samples. Jackson, S.

PB2367. A single-cell atlas of 3D genomic structure and DNA methylation in human subcutaneous adipose tissue. Chen, Z.

PB2369. Additive effects of stress and alcohol exposure on accelerated epigenetic aging in Alcohol Use Disorder. Jung, J.

PB2371. Alterations of regulatory regions in T-cell Prolymphocytic Leukemia. Yan, H.

PB2373. Analysis of methylation QTLs in breast cancer characterises the influence of germline variation on the abnormal cancer methylome. Hannah, R.

PB2375. Atypical Prader-Willi and Angelman syndrome deletion: Importance of parent of origin detection. Al-Sweel, N.

PB2377. Catalysis of histone H3K27me3 by Polycomb-independent mechanisms. Mercado-Hernandez, I.

PB2378. Characterization of nucleosome positioning and DNA methylation signatures using prostate cancer cells from diverse ethnic groups using NOMe-EM-seq. Gonzalez-Smith, L.

PB2382. Clinical Epigenomic Testing in Canada: Discovery and Clinical Assessment of Episignatures. McConkey, H.

PB2384. Cord blood DNA methylation alterations potentially mediate associations between adverse pregnancy outcomes and childhood blood pressure. Hu, J.

PB2386. Critical epigenomics association between methylation and clinical phenotype. Kulikowski, L.

PB2388. Cross-tissue patterns of DNA hypomethylation reveal genetically distinct histories of cell development. Scott, T.

PB2390. Defining the genetic background of immunoglobulin G galactosylation in humans. Frkatovic, A.

PB2392. Differentially methylated probes and regions associated with the Healthy Eating Index. Yuan, K.

PB2394. Disparities in epigenetic modifications in *NRF1* and *FTO* genes in children.. Patel, P.

PB2396. Dissecting the molecular machinery and sequence of histone to protamine transition during spermiogenesis. Rabbani, M.

PB2398. Divergent age-related methylation patterns in long and short-lived mammals. Haghani, A.

PB2400. DNA methylation changes associate with measured glomerular filtration rate in an American Indian cohort with type 2 diabetes. Day, S.

PB2402. DNA Methylation Episignature of Valproate Embryopathy. Relator, R.

PB2404. DNA methylation profile in multiple system atrophy and progressive supranuclear palsy. Reho, P.

PB2406. Dysregulation of serum and exosomal miR-7-1-5p and miR-223-3p in Parkinson's disease. Citterio, L.

PB2408. Epigenetic Age Acceleration is Suggestively Associated with Stroke Precursor Phenotypes. Dueker, N.

PB2409. Epigenetic profiling of isolated blood cells reveals highly cell-type specific smoking signatures and links to disease risk. Wang, X.

PB2411. Epigenetic signatures of asthma in nasal epithelium from African ancestry populations from the CAAPA consortium. Boorgula, M.

PB2413. Epigenome-wide association of DNA methylation markers for dilated cardiomyopathy in left ventricular heart tissues. Tan, K.

PB2415. Epigenome-wide association study reveals CpG sites associated with thyroid function and regulatory effects on *KLF9*. Teumer, A.

PB2416. Epstein-Barr Nuclear Antigen 2 (EBNA2) types 1 and 2 have shared and distinct host DNA binding partners. Viel, K.

PB2418. Exploring novel regulatory elements in X-chromosome inactivation. Beseiso, D.

PB2420. Finding the Right Match: the Role of ZCWPW1 in Mammalian Meiosis. Xie, W.

PB2422. Functional Analysis of CHD2 and CHD8 *De Novo* Mutations and Related Molecular Events Involved in Neurodevelopmental Disorders. Muhammad, T.

PB2424. Genetic variation in correlated regulatory region of immunity. Avalos, D.

PB2426. Genome-wide dysregulation of R-loops in Ataxia Telangiectasia neurological pathogenesis. Westover, K.

PB2428. Genome-wide mapping implicates 5-hydroxymethylcytosines in diabetes and Alzheimer's disease. Zhang, Z.

PB2430. Genome-wide neonatal epigenetic changes associated with maternal exposure to the COVID-19 pandemic. Kocher, K.

PB2432. Global identification of cortical circRNAs and circRNA clusters associated with Alzheimer's disease. Wang, F.

PB2434. High-dimension to high-dimension screening for detecting genome-wise epigenetic regulators of gene expression. Ke, H.

PB2436. Human epi- and transcriptomic trajectories in the postprandial state. Costeira, R.

PB2438. Identification of diagnostic DNA methylation episignatures associated with large structural chromosomal variations in patients with neurodevelopmental disorders. Rooney, K.

PB2440. Immune cell-type level epigenome-wide association analysis on chronic HIV infection highlights hallmark genes for HIV pathogenesis and cancer. Zhang, X.

PB2442. Investigation of the effects of alcohol exposure on the chromatin binding and localization of cohesin and CTCF. Cummings, C.

PB2444. KDM6A/B inhibition rescues an osteoblast phenotype and altered gene expression in a novel mouse model of Weaver Syndrome with skeletal overgrowth. Fahrner, J.

PB2446. Learning the cis and trans sequence features of gene regulatory divergence between human and rhesus macaque. Fong, S.

PB2448. Loss of DNMT3A or TET2 activity in stimulated macrophages alters transcription factor binding and enhances inflammatory gene expression. Rodrigues, K.

PB2450. Methylation-based machine learning classification of osteoarthritis in the Multicenter Osteoarthritis Study. Okoro, P.

PB2452. Multiscale phase separation by explosive percolation with the single chromatin loop resolution. Plewczynski, D.

PB2454. Nascent RNA reveals new links between genetic variation and disease at multiple levels of regulation. Buen Abad Najar, C.

PB2457. Peripheral immune roles in bipolar disorder: genomic, epigenomic, and phenotypic integration. Hou, L.

PB2459. Reduced-Representation Methylation Sequencing (RRMS): Oxford Nanopore's alternative to RRBS enables accurate and easy to use methylation calling in CpG islands, shores, shelves, and promotor regions using a MinION flowcell. Rescheneder, P.

PB2461. Role of TET1-mediated epigenetic modulation in Alzheimer's disease. Armstrong, M.

PB2463. Serum miR-451a is up-regulated in sarcopenic patients compared to healthy controls. Agostini, S.

PB2465. Single-cell sequencing links reduced glucose metabolism to cocaine addiction-like behavior in rats. Zhou, J.

PB2467. SnapHiC-G: identifying long-range enhancer-promoter interactions from single cell Hi-C data via a global background model. Liu, W.

PB2469. Stability of genetic regulation of gene expression over time. Raza, Y.

PB2471. TDP-43 depletion results in aberrant gene expression and TE activation via dysregulation of R-loop homeostasis. Hou, Y.

PB2473. The interplay of epigenetic, genetic, and traditional risk factors on blood pressure: Findings from the Health and Retirement Study. Zhang, X.

PB2475. Tobacco smoke-derived metabolites in plasma are associated with cardiometabolic traits, are genetically regulated, and may modulate adipose tissue DNA-methylation in African Americans. Das, S.

PB2477. Transfer learning of a methylation based COVID19 severity model for DNA viral infection by eczema herpeticum. Zhou, W.

PB2479. Trinity of chromatin architects - The coordination of CTCF, RNAPOL2 and Cohesin in shaping the genomic landscape. Agarwal, A.

PB2481. Using allele-specific expression to assess intra- and inter- individual heterogeneity in X-chromosome inactivation within human tissues. Markus, H.

PB2483. Using machine learning to predict 3D genome organization across thousands of diverse individuals reveals conservation and population differentiation. Gilbertson, E.

PB2485. Variable*cis*-regulatory landscapes associate with gene function and expression patterns. Benton, M.

PB2487. *Variation in miRNAs may contribute to orient future therapeutic strategies in hidradenitis suppurativa; An epigenome-wide* screening approach. Aaren, V.

PB2489. Whole genome bisulfite sequencing identifies methylation markers in cord blood linked to maternal hyperglycaemia and childhood metabolic abnormalities. Li, Y.

PB2491. Widespread age-associated changes in the chromatin architecture of skeletal muscle cell type populations. Moo, K.

3:00 PM - 4:45 PM – Thursday Posters

Poster authors will be at their boards on Wednesday or Thursday from 3:00 to 4:45 pm. See pages <mark>xx-xx</mark> for Wednesday listings.

Cancer Posters - Thursday (Poster)

PB1002. 4C guided cis- and trans-interaction networks associated with the RCCD1 gene at the 15p26.1 breast and ovarian cancer risk locus. Plummer, J.

PB1004. A Case of A Hyperdiploid Complex Karyotype in a Patient with a Myelodysplastic Syndrome. Robinson, H.

PB1006. A genome-wide association study to identify predictive markers for the risk of nivolumab-induced immune-related adverse events. Zembutsu, H.

PB1008. A novel immunohistochemical marker helps in the diagnosis of Lynch syndrome: detection of deficient DNA mismatch repair pathway proteins crypts within non-tumor colonic mucosa. Coulet, F.

PB1011. Accelerated Tumor Only Variant Analysis Utilizing a GPU Framework. Vats, P.

PB1013. Amplification of *RUNX1* in a patient with AML. Hurtado, R.

PB1015. An AML patient showing an abnormal hyperdiploid karyotype. Hamid, B.

PB1017. Analysis of second-hit events in Tuberous Sclerosis Complex normal tissues and lesions. Klonowska, K.

PB1020. Artemis disease-associated variants and their effect on VDJ recombination and genome instability. Gavilan, M.

PB1022. Assessment of genomic instability in aggressive forms of breast cancer in Mali. Ongoiba, C.

PB1024. Associations of genetic ancestry to the somatic mutational landscape from tumor profiling data of 100,000 cancer patients. De La Vega, F.

PB1028. Can impairment of kidney function cause kidney cancer? A bidirectional Mendelian randomization study. Yang, Y.

PB1030. Cancer subclone detection based on DNA copy number in single cell and spatial omic sequencing data. Wu, C.

PB1032. Case report: Olaparib use in metastatic lung adenocarcinoma with *BRCA2* pathogenic variant Soon, J.

PB1034. Cell-free DNA fragmentation patterns predict the response to immunotherapy in head and neck squamous cell carcinomas. Zheng, H.

PB1036. Characterization of molecular differences between normal weight and overweight/obese cancer patients. Huang, F.

PB1038. Characterization of the *2q22* renal cancer susceptibility locus. Souza, A.

PB1040. Chromosomal alteration profiling of lymph node metastases in non-small cell lung cancer. Valdebenito, J.

PB1042. Circulating miRNA signature for diagnostic prediction in breast cancer. Yerukala Sathipati, S.

PB1044. Classification of Central Nervous System Tumors with DNA methylation - The Brazilian Experience. Wolff, B.

PB1046. Clinical utility of genomic sequencing for hereditary cancer syndromes: A retrospective chart review. Kodida, R.

PB1048. CML with a typical BCR-ABL1 fusion in a 15-year-old male patient presenting with marked leukocytosis, thrombocytosis, left-shifted granulocyte maturation in the peripheral blood. Solanki, M.

PB1050. Combined FISH and cytogenetic testing increases the accuracy of classification and risk stratification of multiple myeloma samples.Smith, J.

PB1052. Comprehensive CRISPR-based dissection of a complex common and familial melanoma risk locus on chromosome band 9p21. Hennessey, R.

PB1054. Computational method for the detection of microsatellite instability in tumor tissue samples. Budiš, J.

PB1056. Confirmation of pathogenic germline variants identified by tumour testing in British Columbia. Cheng, P.

PB1058. Copy number variation at *ATF7IP* is associated with increased risk for testicular germ cell tumors. Fan, M.

PB1060. *CRLF2* Rearrangements in a Patient with B-ALL. Lin, Y.

PB1062. Deciphering a prognostic DNA methylation signature in tubo-ovarian high-grade serous cancer. Jorgensen, B.

PB1063. Deep and error corrected sequencing via the low-cost Ultima Genomics platform enables ultra-sensitive circulating tumor DNA cancer monitoring. Cheng, A.

PB1065. Deep learning morphology profiling identifies and enriches carcinoma cells from effusion samples in real-time for cytological and molecular analysis. Mavropoulos, A.

PB1067. Detection of androgen receptor splice variants from clinical sequencing. Hwangbo, S.

PB1069. Detection of circulating tumor DNA in resectable pancreatic ductal adenocarcinoma. Lee, J.

PB1071. Detection of somatic copy number alterations using paired DNA and RNA sequencing. Lastrapes, M.

PB1073. Developing a high-quality, sample-to-result Hereditary Breast and Ovarian Cancer Panel Assay Pipeline for using a novel sequencing platform. Bhattacharjee, A.

PB1075. Development and Clinical Validation of a Liquid Biopsy Assay for Neurofibromatosis 1. Xu, F.

PB1077. DHGAN, Digital Histology Generative Adversarial Networks Used in Creating Synthetic Genitourinary Tissue Slides. Arora, H.

PB1079. Dietary factors and endometrial cancer risk: A Mendelian randomization study. O'Mara, T.

PB1082. Discovery of transcriptomic predictors of colorectal cancer mortality by race and ethnicity. Yin, H.

PB1084. Does non-coding variation in known breast cancer susceptibility genes contribute to breast cancer risk? Akamandisa, M.

PB1086. Dynamic play between human N a-acetyltransferase D and H4-mutant histones: Molecular dynamic study. Srivastava, K.

PB1088. Efficacy of universal testing for hereditary cancer syndromes: A community based cancer center (CBCC) experience. Hamblett, A.

PB1090. Eligibility, uptake and response to germline genetic testing in women with DCIS. Ellsworth, R.

PB1092. Enhanced expression of mutant alleles of cancer driving genes in pan-adenocarcinoma. Lo, C.

PB1093. Estimating total tumor-specific microRNA content in human tissues using computational deconvolution. Montierth, M.

PB1095. Evaluation of miR-21 and miR-938 expression in the progression of gastric cancer in the Magellanic Chilean cohort - MAGIC.. Zapata-Contreras, D.

PB1097. Evidences for polygenic inheritance in familial cancer. Lista, M.

PB1099. Expanding the clinical and molecular spectrum of hereditary *TINF2* cancer predisposition syndrome reporting a family with Hodgkin Lymphoma. Agolini, E.

PB1101. Family History-Based Risk of BRCA Mutations in the Malian Populations. Baba, B.

PB1103. Finding Therapeutic Targets in Cancer Cells Using Essential Genes. Tran, T.

PB1105. Functional characterization of the 1p36.33 pancreatic cancer GWAS locus. Connelly, K.

PB1107. Functionalizing methylation-derived gene panels in the UK Biobank, TCGA, and HCA datasets. Mudgal, U.

PB1109. Gene-environment interaction of antioxidant genes and a healthy lifestyle index (HeLiX) on breast cancer risk reduction. Gómez Flores Ramos, L.

PB1111. Genetic ancestry differences in tumor mutation between early and average onset colorectal cancer. Rhead, B.

PB1113. Genetically Predicted Telomere Length and Risk of Multiple Primary Cancers in the Prostate, Lung, Colorectal and Ovarian Cancer Screening Trial. He, S.

PB1115. Genome-wide Analysis of Rare Haplotypes Associated with Breast Cancer Risk: Discovery, Replication, and Generalizability Evaluation. Wang, F.

PB1117. Genome-wide association study of intracranial germ cell tumors: a common deletion at *BAK1* attenuates the enhancer activity and confers risk for the rare disease. Sonehara, K.

PB1119. Genomic landscape of malignant peripheral nerve sheath tumor (MPNST) reveals novel pathways of tumor evolution that correlate with tumor behavior. Miller, D.

PB1121. Germline genetics of venous thromboembolism (VTE): Non-cancer polygenic risk scores improve risk stratification in cancer patients. Groha, S.

PB1123. Germline sequencing of DNA damage repair genes in two hereditary prostate cancer cohorts reveals rare risk-associated variants. Foley, G.

PB1125. GLUT10 is a biomarker for poor prognosis and a predictor of vitamin C combination therapy in breast cancer. Lee, Y.

PB1127. HDM2 differentially modulates the molecular phenotype of lung tumor cells of varying p53 genotypes. Wang, Y.

PB1129. Hereditary Cancer Variants & Homologous Recombination Repair Deficiency in Biliary Tract Cancer. Nakagawa, H.

PB1131. High tumor mutational burden by whole-genome sequencing in resected non-small cell lung cancer in never smokers is associated with worse prognosis. Ruel, L.

PB1133. Identification of key molecular mechanisms in IDH mutant brain tumors to enable precise risk stratification. Bhattacharya, S.

PB1135. Identification of *USP9X* as a leukemia susceptibility gene. Sisoudiya, S.

PB1137. Identifying strong instrumental variables for pleiotropic assessment in Mendelian randomization: CALGB/SWOG 80405 (Alliance). Yazdani, A.

PB1139. Implementation of new ClinGen/CGC/VICC recommendations for classification of oncogenicity of somatic variants using AI. Einhorn, Y.

PB1141. Inference on the genetic architecture of breast cancer risk. Yasui, Y.

PB1143. Integrative data analysis to uncover genes and pathways underlying the aggressiveness of lung cancer brain metastasis and the overall prognosis. Asakereh, R.

PB1144. Investigating the link between cholesterol-lowering genes and risk of skin cancers. Ong, J.

PB1146. Large-scale whole genome sequencing reveals that APOBEC mutagenesis is a common process in normal human small intestine. Wang, Y.

PB1148. Leveraging somatic cancer mutation data to predict the pathogenicity of germline missense variants. Haque, B.

PB1150. Loss of function of *neurofibromin* affects *afadin* -6 in neurofibromatosis type-1. Sulaiman, M.

PB1152. Massively parallel sequencing-based panel strategy for microsatellite instability testing. Styk, J.

PB1154. Metabolic Profiling of Primary and Metastatic Breast Cancer Cells and New Treatment Approaches. Boccuto, L.

PB1156. Methylation-sensitive restriction enzyme sequencing (MRE-seq)-based early detection of cancer using deep neural network. Kwon, H.

PB1158. Modulating the immune system not to reject an allograft. McDaniel, D.

PB1160. Molecular signature predicts immunotherapy resistance in gynecologic cancer patients treated with immune checkpoint inhibition. Ni, Y.

PB1162. Multigene deregulation 12q12 renal cancer-susceptibility locus. Andrade Costa, L.

PB1164. Multiomic sequencing analysis with whole genome association study at chromosome 16 of wilms tumor patients. Chang, C.

PB1166. Multiplex ligation dependent probe amplification versus fluorescent in situ hybridization for screening RB1 copy number variations in Egyptian patients with retinoblastoma. Eid, o.

PB1168. Multi-stage germline exome sequencing study of 17,546 men with aggressive or non-aggressive prostate cancer identifies genes that may inform clinical gene panel testing. Darst, B.

PB1170. No evidence of association between clonal hematopoiesis and risk of prostate cancer in large samples of European ancestry men. Xu, Y.

PB1172. Nucleotide resolution of large tandem duplications in cancer genomes. Audano, P.

PB1174. Oncological biomarkers identified by NGS: characterization of the colombian population tumors from 2019 to 2022. Parada Niño, L.

PB1176. Ovarian sex cord stromal tumor is a likely additional phenotype of *BAP1*-Tumor Predisposition Syndrome. Abdel-Rahman, M.

PB1177. *PALB2*-mutated human mammary cells display a broad spectrum of morphological and functional abnormalities beyond defective DNA damage response. Winqvist, R.
PB1179. Pan-cancer polygenic risk distinctly contributes to prediction of heterogeneous subsequent malignancies across treatment profiles in survivors of childhood cancer. Im, C.
PB1181. Pathogenic genetic variants from highly connected cancer susceptibility genes confer the loss of structural stability and further link with diabetes. Hossain, M.
PB1183. Phylogeny-aware detection of single-nucleotide variants and mode of evolution from cancer single-cell DNA sequencing data. Edrisi, M.
PB1185. Potential subtype-informative genetic risk variants and genes identified from a case-case analysis for breast cancer. Sun, X.
PB1187. Prevalence of mutations in hereditary cancers of Czech patients. Koudová, M.
PB1189. Primary myelofibrosis and progression to acute myeloid leukemia in a young patient with variant in *CARL* Tamayo. Palacio, A.
PB1191. Proteome studies of breast, prostate, ovarian, and endometrial cancers implicate plasma protein regulation in cancer susceptibility. Gregga, I.
PB1193. Rare variant association study in lymphoid cancers. Ralli, S.
PB1194. Relative involvement of human papillomavirus constitutional genomic instability: Cellular sequela syndromes. McGhee, E.
PB1196. Revisiting tumor evolution in multifocal hepatocellular carcinoma with different clonal origins. Tang, X.
PB1198. Risk of cancer in persons with a single Fanconi anemia gene variant: A population-based study of MyCode Cohort. Altintas, B.
PB1200. Sex differences in the Lung Adenocarcinoma transcriptome: Analysis of TCGA LUAD and GTEx lung tissue. Printzis, C.
PB1202. Single cell RNA sequencing reveals global cellular landscape of appendiceal cancer. Bui, L.
PB1204. Single Sample Gene Set Enrichment Analysis (ssGSEA) to Identify Dysregulated Pathways in a Biomarker Matched Pilot Study of Mantle Cell Lymphoma. Hill, H.
PB1206. Single-cell methylation and 3C sequencing in prostate tumors shows substantial epigenome reprogramming. Li, T.
PB1208. Single-cell sequencing of genomic DNA: Assessing clonal populations of tumor cells in endometrial cancer. Velazquez Villarreal, E.
PB1210. Splicing analysis of endometrial cancer GWAS risk loci: splicing-associated variants reveal *NF1* and *SKAP1* as candidate susceptibility genes. Glubb, D.
PB1212. Targeting mutant HLA-bound cancer peptide demonstrated in vivo efficacy in delaying tumour growth in a low mutational burden aggressive ovarian cancer. Nguyen-Dumont, T.
PB1213. Telomeric nuclear organization of urinary circulating cells in bladder cancer (BC): A new method uncovers unreported markers. Doumbia, O.
PB1218. The Role of Tumor Microenvironment and Tumor-Active Pathways in the Progression of Glioma. Bayram, F.
PB1220. Transcriptome Wide Association Study identifies novel candidate risk genes for testicular germ cell tumors. Ugalde Morales, E.
PB1222. Transcriptome-wide association study identifies novel susceptibility genes for childhood acute lymphoblastic leukemia in Latinos. Kachuri, L.
PB1224. UNISON: a software toolkit for streamlined detection of clonal hematopoiesis of indeterminate potential in large-scale sequencing studies. Tian, S.
PB1226. Unravelling RecQ helicase function in genome stability using Strand-seq. Hamadeh, Z.
PB1228. Utility of hereditary cancer panel testing in individuals with a known familial variant. Weltmer, E.
PB1230. Variants in *FANCM* confer risk to estrogen receptor-negative breast cancer among Latinas. Nierenberg, J.

Epigenetics Posters - Thursday (Poster)
PB2361. A Bayesian interaction model to estimate cell-type-specific Methylation Quantitative Trait Loci (meQTLs) incorporating priors from flow-sorted sequencing data. Cheng, Y.
PB2364. A methylation-based COVID-19 classification model to predict severe disease in vaccinated individuals. Harrison, G.
PB2366. A single cell chromatin accessibility and transcriptome atlas of the human heart improves the identification of risk variants and genes of Atrial Fibrillation. Selewa, A.
PB2368. Activation of *Xist* by an evolutionarily conserved function of KDM5C demethylase. Samanta, M.
PB2370. Alcohol use-disorder associated DNA methylation differences in human nucleus accumbens and dorsolateral pre-frontal cortex. White, J.

PB2372. Altered and allele-specific open chromatin landscape reveals epigenetic and genetic regulators of innate immunity in COVID-19. Zhang, Z.

PB2374. *ASXL3* links chromatin biology to neurodevelopment disorders. Peirent, E.

PB2376. Cardiac development in mammalian models lacking the PR-DUB component ASXL3. Regan, S.

PB2379. Characterization Of Pten Expression Cis-Regulatory Elements Involved In Cowden Disease. Matis, T.

PB2381. ChromGene: Gene-Based Modeling of Epigenomic Data. Jaroszewicz, A.

PB2383. Conserved GATA4/5/6-mediated gene regulation in cardiac development and disease. Wilson, M.

PB2385. Could the difference in DNA methylation status explain phenotypic variability in patients with 5p- syndrome? Almeida, V.

PB2387. Cross-species and tissue imputation of species-level DNA methylation samples. Maciejewski, E.

PB2389. Defining the gene regulatory roles of non-coding variants in the pathogenesis of autism. Sosa, E.

PB2391. Differential newborn DNA methylation among individuals with complex congenital heart defects and pediatric lymphoma. Richard, M.

PB2393. Digitally untangling the 3D genome. Fudenberg, G.

PB2395. Dissecting mechanisms underlying expression divergence of human duplicated genes. Shew, C.

PB2397. Distinct signatures in malignant PEComa and leiomyosarcoma identified by integrative RNA-seq and H3K27ac ChIP-seq analysis. Giannikou, K.

PB2399. DNA methylation changes among Ethiopian women diagnosed for cervical cancer. Kumbi Jufara, B.

PB2401. DNA methylation epi-signature and biological age in attention deficit hyperactivity disorder patients. Carvalho, G.

PB2403. DNA methylation in twins discordant for van der Woude syndrome. Petrin, A.

PB2405. DNA methylation signatures in Arab and Middle Eastern families with neurodevelopmental disorders provide insight to disease etiology. Nour, A.

PB2407. Early Embryos Employ a Unique Mechanism of CENP-A Equalization during the First Cell Cycle. Tower, C.

PB2410. Epigenetic regulation of Wnt signaling pathway associated with age-related mobility loss. Quillen, E.

PB2412. Epigenome-wide association meta-analysis of DNA methylation with lifetime cannabis use. Fang, F.

PB2414. Epigenome-wide association study in multi-ethnic Asian populations identifies novel markers for incident type 2 diabetes. Loh, M.

PB2417. Expanded studies of a methylation-based COVID-19 classification model to predict severity of disease and its ability to differentiate from other respiratory viruses. Peterson, B.

PB2419. Extracellular vesicles microRNAs in traumatic brain injury patients with and without intracranial abnormality. Vorn, R.

PB2421. First trimester human placenta DNA methylation correlates to maternal estradiol levels. Gonzalez, T.

PB2423. Gene regulatory network synchronizes genetic and epigenetic signals, prioritizes GWAS SNPs, and identifies repurposable drug candidates for multiple sclerosis. Manuel, A.

PB2425. Genome-wide DNA methylation profiling in subcortical regions reveals epigenetic signatures associated with PTSD and MDD. Li, H.

PB2427. Genome-wide evaluation of the effect short tandem repeat variation on local DNA methylation. Martin Trujillo, A.

PB2431. Genome-wide screening of epigenetic variations in the EpiSign Knowledge Database. Relator, R.

PB2433. High dimensional co-expression network analysis unravels transcriptomic drivers of diverse biological systems. Morabito, S.

PB2435. High-resolution analysis identifies ancestry-specific chromatin 3D interactions. Xu, W.

PB2437. Human gene regulatory evolution is driven by divergence in both *cis* and *trans.* Hansen, T.

PB2439. Identifying functional interactors of conserved meiotic histone reader ZCWPW1. Bazzano, D.

PB2441. Investigating aberrant DNA methylation in pediatric epilepsies. LaFlamme, C.

PB2443. Investigation of Tissue Level of Molecular Pathways Effective in Vascular Calcification at Chronic Renal Insufficiency Patients. Arslan, E.

PB2445. Learning a generalized regulatory model from paired chromatin accessibility and transcriptome. Xiong, L.

PB2447. Linking genetic variants associated with gluteofemoral fat storage to determine their role in adipogenesis. Belanich, J.

PB2449. Mapping chromatin accessibility QTL in 138 liver tissue samples identifies coordinated regulation, links regulatory elements to genes, and predicts mechanisms at GWAS loci. Currin, K.

PB2451. MIMOSA: A method for improved methylome imputation increases power to identify CpG site-phenotype associations. Melton, H.

PB2453. NANOME: A Nextflow pipeline for haplotype-aware allele-specific consensus DNA methylation detection by nanopore long-read sequencing. Liu, Y.

PB2455. Network analysis across cellular reprogramming states. Thangavelu, C.

PB2456. Nucleosome spike-in controls identify best-in-class antibodies and enable reliable next-generation epigenomic mapping approaches. Husby, N.

PB2458. Rare inherited Copy Number Variants as genetic modifiers of Developmental Disorders. Atzori, M.

PB2460. RFX transcription factors regulate genes involved with primary cilium in glioma. Chua, S.

PB2462. Scoring individual cells in snATACseq dataset based on GWAS fine-mapping identifies disease-relevant cell-types in neurodegenerative diseases. Mohamed, A.

PB2464. Simultaneous sequencing of genetics and epigenetics provides opportunities for new biological insights.   Lumby, C.

PB2466. Single-molecule architecture and heterogeneity of human telomeric DNA and chromatin. Stergachis, A.

PB2468. SpliceVI: a visualization tool for predicting protein consequences based on SpliceAI data. Cho, Y.

PB2470. Subsetting systemic lupus erythematosus patients based on clustering of DNA methylation at the time of disease flare. Horton, M.

PB2472. The association of cigarette smoking with DNA methylation and gene expression in human tissues. Tamayo, L.

PB2474. The role of genetic and epigenetic mechanisms in the definition of the circular RNA landscape in multiple sclerosis. Paraboschi, E.

PB2476. Transcriptome and chromatin accessibility dynamics across 25 brain regions identify novel susceptibility gene sets for neuropsychiatric disorders. Dong, P.

PB2478. Transposable elements are associated with the variable response to influenza infection. Chen, X.

PB2480. Uncovering Novel Functions of Histone Demethylase KDM5 Through a Genome-wide Approach. Yheskel, M.

PB2482. Using iPSC-derived microglia and oligodendrocytes to study ancestry-specific gene expression in the context of Alzheimer's Disease (AD). Moura, S.

PB2484. Validation and Methylome analysis of the CRISPR-edited DUX4 locus in immortalized myoblast cell lines modeling FSHD using long-read sequencing. Sakr, J.

PB2486. Variation in DNA methylation is associated with cognitive function in post-surgery breast cancer patients prior to adjuvant therapy. Liu, S.

PB2488. Vitamin c contributes to epigenetic regulation of genes related to diabetic retinopathy in retinal endothelial cells. Sant, D.

PB2490. Whole-methylomics reveals differentially methylated genes in blood associated with Late-onset Alzheimer's disease. Breen, C.

Complex Traits Posters - Thursday (Poster)

PB1232. A catalog of genetic predictors impacting lifelong medication use patterns in hyperlipidemia, hypertension and type 2 diabetes. Kiiskinen, T.

PB1234. A comprehensive coronary artery disease risk prediction framework incorporating genetic and nongenetic risk factors. Chen, S.

PB1236. A *Drosophila* platform to validate and assess pathogenicity of LMNA variants identified from dilated cardiomyopathy patients. Han, Z.

PB1238. A genome-wide association study in Peruvians suggests new risk loci for Alzheimer disease. Rajabli, F.

PB1240. A genome-wide association study of mammographic density phenotypes among pre-menopausal women of European ancestry. Harrison, T.

PB1242. A large-scale genome-wide association meta-analysis of polycystic ovary syndrome in over 540,000 women provides insights into biological mechanisms. Actkins, K.

PB1244. A multi-ancestry genome-wide association study identifies two novel loci associated with increased risk of diabetic macular edema. Stockwell, A.

PB1247. A novel missense variant in melanopsin associated with delayed sleep phenotype: A whole genome sequencing study. Brzezynski, J.

PB1249. A polygenic score-based approach improves discrimination between familial, idiopathic, and pathologic short stature in a cohort of pediatric patients presenting for evaluation. Shelley, J.

PB1251. A single nucleus transcriptomics study of alcohol use disorder in postmortem brain tissue. Clark, S.

PB1253. A trans ancestry genomics based approach to study the interplay between the immune system, infectious type, and HLA type, ancestry and sepsis outcome. Chhugani, K.

PB1254. Accurate identification of causal variants of intracellular LDL uptake via Bayesian modeling of base editor reporter screens. Ryu, J.

PB1256. Advancing rare disease research through web-based recruitment: the 23andMe Systemic Sclerosis Research Study. Kukar, K.

PB1258. Altered expression levels of *TAS1R1, TAS1R3* genes among SARS-CoV-2 variants. Ergoren, M.

PB1260. Alzheimer's disease genetic risk and plasma biomarker profiles in the Midwestern Amish. Dorfsman, D.

PB1262. An emirati cohort of autism spectrum disorders. Nassir, N.

PB1264. An unprecedented level of complexity in the schizophrenia-associated 3q29 region of the human genome with unique segments that increase the risk for non-allelic homologous recombination. Yilmaz, F.

PB1266. Analysis of adipose and liver expression profiles from dual-tissue transcriptomic cohort discovers 10 serum biomarker candidates for NAFLD.   Darci-Maher, N.

PB1268. Analysis of MRI-derived spleen iron in the UK Biobank identifies genetic variation linked to iron homeostasis and hemolysis. Sorokin, E.

PB1270. Analysis of poison exons caused by splice donor variants in genes associated with developmental brain disorders. Hare-Harris, A.

PB1272. *APOE* genotype and rare variants of APP processing complex in autopsy-confirmed rapidly progressive Alzheimer disease. Wang, P.

PB1274. Association analysis of *LHCGR* gene polymorphisms with Polycystic Ovary Syndrome in Punjabi population. Singh, S.

PB1276. Association between a type 2 diabetes polygenic risk score and type 2 diabetes-related risk factors and complications. Guo, B.

PB1278. Association between genomic runs of homozygosity and complex traits in the Taiwan Biobank. Feng, Y.

PB1280. Association of a genetic risk score with incident coronary heart disease in the Hispanic Community Health Study/Study of Latinos (HCHS/SOL) cohort. Hutten, C.

PB1282. Association of genetic predisposition and physical activity with risk of gestational diabetes mellitus in nulliparous women. Radivojac, P.

PB1286. Associations between polygenic risk scores and gene expression identify core genes for bipolar disorder. Lapinska, S.

PB1288. Autism and Cognitive Ability CNV- Genome Wide Association study (CNV-GWAS). Poulain, C.

PB1290. Automated identification, GWAS, and ExWAS of enlarged perivascular space burden in human brain MRI in the UK Biobank. Parikshak, N.

PB1292. Causal effects of gut-related metabolites on human psychiatric disorders. Ihejirika, S.

PB1294. Cellular context-specific gene regulation of neuropsychiatric disorders in single human neurons. Wood, W.

PB1296. Characterization of gene expression profile and molecular pathways involved in type 2 diabetes among self-identified Hispanic American individuals. Yaser, M.

PB1298. Characterizing 31 cardiometabolic candidate genes for a role in early vascular inflammation and foam cell formation using CRISPR-Cas9, *in vivo* imaging and deep learning. den Hoed, M.

PB1300. Choroidal thickness as a biomarker for age-related macular degeneration progression: Genome-wide Association Study in Amish. Cooke Bailey, J.

PB1302. CLEC16A regulates the adipogenic and lipolytic capacity of adipocytes. Bakay, M.

PB1304. Clonal behavior across multiple timepoints in clonal hematopoiesis. Mack, T.

PB1306. Colocalization and variant-to-gene mapping nominates pleiotropic bone mineral density effector genes. Conery, M.

PB1308. Combining polygenic and monogenic causes for improved osteoporotic fracture risk prediction. Lu, T.

PB1310. Comparison of clinical characteristics among survivor and non-survivor of covid-19 infected patients: Scoping review and meta-analysis.   Adebiyi, M.

PB1312. Considerations in developing a framework for defining clinical actionability of polygenic risk scores. Hunter, J.

PB1314. Construction of mutational landscape and characterization of associated genes in a Bangladeshi cohort of neurodevelopmental disorders. Akter, H.

PB1316. Coronary artery disease risk assessment in Asian Indian Punjabis using a genome wide polygenic risk score. Rout, M.

PB1317. Cross-tissue single-nucleus RNA-sequencing reveals distinct tissue-specific expression profiles of adipocytes, depending on their residency in human adipose tissue or heart. Das, S.

PB1319. Cytokine treatment increases *APOEε4* expression in reactive astrocytes in European local ancestry but not in African local ancestry. Oron, O.

PB1321. Data-driven genome-wide association study of longitudinal pain and mobility trajectories among cases of Parkinson's disease from the Fox Insight Data Exploration Network. Liu, S.

PB1323. *De novo* germline and somatic variants as modifiers of cleft lip/palate severity in mice. Rao, S.

PB1325. Deciphering the causal relationship between blood pressure and white matter integrity: a Mendelian Randomization study in the UK Biobank. Ye, Z.

PB1327. Deep learning models of gene regulation in Alzheimer's disease improve functional fine-mapping and thereby cross-ancestry PRS portability. Lin, T.

PB1329. Differential gene expression analysis of alcohol use disorder across postmortem human brain tissues. Willis, C.

PB1331. Differentiating Mesenchymal Stem Cells from iPSCs for Analysis of Type 2 Diabetes and Related Trait GWAS Loci. Ventresca, C.

PB1333. Discovering disease context eQTLs in a patient cohort with active psoriatic arthritis. Guan, M.

PB1336. Discovery of Type 2 Diabetes genes using an accessible tissue. Davtian, D.

PB1338. Dissecting autism heterogeneity by genotype phenotype analysis among autism risk gene carriers in SPARK . Shu, C.

PB1340. Dissecting the relationship between recurrent pregnancy loss and type 1 and type 2 diabetes in Western Ukrainian population. Sharhorodska, Y.

PB1342. Dose-responsive mRNA biomarkers of alcohol consumption and placebo response: A transcriptome-wide gene expression analysis. Shetty, A.

PB1344. Effect of artificial light on metabolic disorders and circadian gene expression. Fatima, N.

PB1346. Effects of epigenetic age acceleration on kidney function: a mendelian randomization study. Pan, Y.

PB1348. Elevated levels of CXCL16 in severe COVID-19 patients: Effects on mortality. Johnson, C.

PB1349. Enriching genetic hypotheses of schizophrenia through neuroimaging transcriptomics. Bledsoe, X.

PB1351. Epstein-Barr virus- and genotype- dependent transcriptional regulation in B cells from patients with multiple sclerosis. Granitto, M.

PB1353. Essential hypertension genomic regions from linkage analysis studies. Magalhaes Borges, V.

PB1355. Ethnicity- and sex-specific genome-wide association study on Parkinson's disease. Chung, S.

PB1357. Evaluating the frequency and impact of structural variation in amyotrophic lateral sclerosis. Dilliott, A.

PB1359. Evaluation of mitochondrial DNA variation in autism and neurodevelopmental disorders. Wang, Y.

PB1361. Evidence of allelic series with fine-mapped protein quantitative trait loci across 1,470 protein abundance measurements and 30 biochemistry measurements in UK Biobank participants. Cortes, A.

PB1363. Exome analysis reveals plausible glaucoma causing mutations in novel genes in dominant JOAG families of Pakistani origin. Ayub, H.

PB1365. Exome-wide association study of DSM-5 antisocial personality disorder in a nationally representative sample. Zhang, H.

PB1367. Exploring genome-wide association results for neuroticism as a proxy for mental health disorders in individuals of African ancestry. Kaka, M.

PB1369. Expression of adipokine genes associated with gestational diabetes mellitus. Issa, R.

PB1371. Extensive Transcriptional Complexity of Autism Associated *SHANK* Family Genes by Capture Based and Long Read sequencing Method. Lu, X.

PB1373. Fine-mapping 110 migraine risk loci using 98,375 migraine cases. Hautakangas, H.

PB1375. Fine-mapping and genomic analyses identify causal variants and genes for hypertension. Bell, C.

PB1377. Fine-mapping the association of *CYP2A6* with nicotine metabolism in African ancestry smokers. Pouget, J.

PB1379. Functional analysis of rare copy number variations across psychiatric disorders. Engchuan, W.

PB1382. Gene-based tests for early and late onset Alzheimer Disease: common and non-overlapping factors. Lucio da Fonseca, E.

PB1384. Gene-environment interaction of coffee with body mass index in multiple populations. Shivakumar, M.

PB1386. Genes at asthma-associated GWAS loci are induced in activated CD4 tissue resident memory T cell. Schoettler, N.

PB1389. Genetic analysis using large biobank controls validates known genes and discover novel genes in Hirschsprung disease. Fu, M.

PB1391. Genetic architecture of asthma in African American Patients. Chang, X.

PB1393. Genetic associations of rare variants in alcohol and tobacco use in up-to 526,400 individuals. Jang, S.

PB1395. Genetic colocalization between Covid-19 and diseases of the immune system. Zechner, M.

PB1397. Genetic Exploration of Military Sudden Cardiac Arrest (GEMini): Genome Sequencing Results from the First 24 Participants. Hellwig, L.

PB1399. Genetic Mechanisms of Supraventricular Tachycardias. Weng, L.

PB1401. Genetic overlap of idiopathic pulmonary fibrosis and hypertension. Parcesepe, G.

PB1403. Genetic prediction of weight and waist circumference regain after weight loss with Intensive Lifestyle Intervention: A secondary analysis of the Look AHEAD trial. Revsbech Christiansen, M.

PB1405. Genetic profiling and improved predictive capability with protein-based risk scores for IBD and disease progression using UK Biobank data. Zeng, X.

PB1407. Genetic regulation of gene expression in the frontal cortex across the human lifespan. Young, H.

PB1409. Genetic Variability of *CYP2B6* Polymorphisms (785A>G, 64>CT and 516G>T) in Southwest Nigerian Population: Implications for Malaria Treatment. Ademola, S.

PB1411. Genetic variation in fatty acid amide hydrolase (*FAAH*) influences early drinking and smoking behaviors. Alsaafin, A.

PB1413. Genetically-associated body-mass index, vitamin D levels, and age at puberty are not associated with Parkinson's Disease related phenotypes. Misicka, E.

PB1417. Genome scans of early childhood caries implicate bitter taste receptors. Orlova, E.

PB1419. Genome wide association study of clinically predicted suicide liability. Jespersen, A.

PB1421. Genome-wide association analyses of sepsis and septic shock in a large practice-based biobank. Jiang, L.

PB1423. Genome-wide association meta-analysis and burden of rare coding variants in African American patients with Acne. Saeidian, A.

PB1425. Genome-wide association of Copy Number Variation for Cleft Lip with or without Cleft Palate in families from a multi-ethnic study sample. Mukhopadhyay, N.

PB1427. Genome-wide association studies identify genetic variants associated with comorbidities in atopic dermatitis. Hartley, A.

PB1429. Genome-wide association study analysis of glucose response in oral glucose tolerance test (OGTT). Thaker, V.

PB1431. Genome-Wide Association Study in a Rat Model of Temperament Identifies Multiple Loci for Exploratory Locomotion and Anxiety-Like Traits. Chitre, A.

PB1433. Genome-wide association study of diabetic retinopathy in the Million Veteran Program identifies four significant loci. Breeyear, J.

PB1435. Genome-wide association study of nephrotic syndrome replicates *APOL1* and MHC loci, while predicted gene expression analysis identifies *C4A*. Hellwege, J.

PB1437. Genome-wide association study of plasma amyloid β levels in older adults. Aslam, M.

PB1439. Genome-wide association study on skin tags. Liu, M.

PB1441. Genome-wide Interaction Study with Smoking Identifies *FHIT* and *SLC22A23* Associated with Alzheimer's Disease. Han, X.

PB1443. Genome-wide meta-analysis identifies novel susceptibility loci for acne vulgaris. Kals, M.

PB1445. Genome-wide Polygenic Risk Scores in Diabetes Risk Prediction: Results from the Asian Indian Diabetic Heart Study/Sikh Diabetes Study. Sanghera, D.

PB1447. Genomic architecture of Autism Spectrum Disorder from comprehensive whole-genome sequence annotation. Trost, B.

PB1450. GenoPipeR: An Automated Pipeline for Predictive Modelling of Complex Phenotypes using Genome-wide Data. Elsheikh, S.

PB1452. Global analysis of RNA editing in Alzheimer's disease across multiple brain regions. Huang, E.

PB1454. GWAS and Polygenic prediction of anthropometric traits in sub-Saharan African populations. Adebamowo, S.

PB1456. Heterozygous loss-of-function mutation in *SORL1* causes neuronal dysregulation of endosomal trafficking and processing of APP. DeRosa, B.

PB1458. HLA Haplotype Analysis and Genome Wise Association Study of Nephrotic Syndrome in the Million Veteran Program. Hung, A.

PB1460. HLA types are associated with hundreds of complex traits and diseases in an ancestry-dependent manner. D'Antonio, M.

PB1462. *HLA* associations with autoantibody-defined subgroups in idiopathic inflammatory myopathies. Diaz-Gallo, L.

PB1464. Host Genetics in Resistance to COVID-19: Hints from recovered Brazilian super elderly. de Castro, M.

PB1466. Hypermethylation of *PM20D1* promoter is associated with atherosclerosis in Dominican Families. Wang, L.

PB1468. Identification of a locus associated with major depressive disorder in a Colombian population. Lattig, C.

PB1470. Identification of genetic loci with divergent effects between Crohn's disease and ulcerative colitis. Kim, Y.

PB1472. Identification of platelet aggregation genetic determinants: Results from a cross-ancestry GWAS meta-analysis. Thibord, F.

PB1474. Identifying compounds to treat opiate use disorder by leveraging multi-omic data integration and multiple drug repurposing databases. Stratford, J.

PB1476. Identifying genetic variations that connect iron homeostasis to metabolic disease. Praggastis, S.

PB1478. Identifying novel gene dysregulation associated with opioid overdose death: a meta-analysis of differential gene expression in human prefrontal cortex. Carter, J.

PB1480. Identifying Rare Variants Associated with Type 2 Diabetes Mellitus in GENNID: a Multiplex, Multiethnic, Family Study. Simon, L.

PB1482. Identifying the genetically dysregulated risk and preventative pathways in Alzheimer's disease. Li, X.

PB1483. Impact of Interleukin-4, Interleukin-13 Gene Polymorphisms and HLA-DQ Alleles on Genetic Susceptibility of Type-1 Diabetes Mellitus (T1DM) in Kuwaiti Children. Haider, M.

PB1485. Improving model predictivity and explainability by combining genotypes and immunophenotypes in SARS-CoV-2 infected people. Renieri, A.

PB1488. Increased burden of pathogenic variants in known aortopathy genes in individuals with early onset thoracic aortic dissection. Guo, D.

PB1490. Increased frequency of predicted loss of function variants in *PKP2* in African ancestry individuals. Winters, A.

PB1492. Insights from HostSeq :Canada's National Whole Genome Sequencing Cohort of 10,000 Canadians Infected with SARS-CoV-2. Garg, E.

PB1494. Integrating common and rare variants into a genetic risk score for Alzheimer's disease risk prediction. Suh, E.

PB1496. Integration Analysis of Multi-Omics Data for Osteoporosis Biomarker Discovery Identification. Qiu, C.

PB1498. Integration of proteomics quantitative trait loci into genetic association analysis of stroke in the African American population. Cai, Y.

PB1500. Integrative analysis of large-scale multi-ancestry genome-wide association study and single-cell omics data provides high-resolution insight of cell-types in the pathogenesis of type 2 diabetes. Suzuki, K.

PB1505. Interrogation of shared genetic architecture of infectious disease traits and Alzheimer's disease. Sale, R.

PB1507. Investigating the cellular and molecular response to hyperglycemiain iPSC-derived cardiomyocytes. Johnson, O.

PB1508. Investigating the influence of genetic ancestry on gene-environment interactions on polygenic risk score and acculturation: Results from the Hispanic Community Health Study/Study of Latinos. Sharma, J.

PB1510. Is Whole-Blood Cell Mitochondrial Copy Number Lower in Chronic Obstructive Pulmonary Disease in the NHLBI trans-omics for precision medicine (TOPMED) Program? Rocco, A.

PB1512. Key discoveries of the genetic basis of stuttering. Pruett, D.

PB1514. Large scale genome-wide association study highlights the causality of vasospastic angina. Hikino, K.

PB1516. Large-scale exome sequencing identifies common and rare coding variants contributing to tinnitus. Ayer, A.

PB1518. Leveraging genetic variation to evaluate risk factors and therapeutic opportunities for aortic valve stenosis: a mendelian randomization analysis. Urquijo, H.

PB1520. Leveraging the nutritional geometry framework to dissect the impact of macronutrient composition on metabolic function and gene regulation in fat tissue. Farris, K.

PB1523. Loss of chromosome Y in leukocytes as a risk factor for critical COVID-19 in men. Bruhn-Olszewska, B.

PB1525. Lung-specific alternative splicing is a shared etiological risk for COVID-19 severity and chronic respiratory diseases. Nakanishi, T.

PB1527. Machine learning-based classification of Age-related Macular Degeneration using gene expression profiles. Ratnapriya, R.

PB1530. Metabolomics profiling upranks the role of natural steroids and phenylalanine metabolism over oxidative stress in insulin resistance amongst lean subjects. Diboun, I.

PB1532. Mitochondrial and Nuclear genetic variants demonstrate mitochondrial function determines severity but not risk of amyotrophic lateral sclerosis. Harvey, C.

PB1534. Modeling cardiac cell developmental trajectories at high temporal resolution. McIntire, E.

PB1535. Modeling gene by environment interactions in post-traumatic stress disorder using hiPSC-derived neurons. Seah, C.

PB1537. Molecular and clinical characterisation of Polish Temple syndrome patients with 14q32 alterations. Jurkiewicz, D.

PB1539. Molecular epidemiologic and family history evidence suggests that myalgic encephalomyelitis (ME)/chronic fatigue syndrome (CFS) may be an autoimmune disorder. Moslehi, R.

PB1542. Multi-ancestry HLA allele calling using whole-exome sequencing in the UK Biobank reveals 353 novel genome-wide significant HLA alleles associated with 11 auto-immune phenotypes. Butler-Laporte, G.

PB1544. Multi-ancestry meta-analysis of X chromosome-wide associations for height. Vedantam, S.

PB1545. Multiple HLA haplotypes and a variant altering immunogenicity of minor histocompatibility antigen epitopes encoded by *CTSH* are associated with age at type 1 diabetes diagnosis. Roshandel, D.

PB1547. Multi-tissue transcriptome-wide association study (TWAS) uncovers 10 novel genes associated with multiple ageing outcomes. Navoly, G.

PB1550. Multivariate GWAS of 3D cranial vault shape in multi-ethnic and admixed children. Goovaerts, s.

PB1552. Neurobiological pathways underlying opioid addiction identified by systems biology multi-omic gene integration.   Sullivan, K.

PB1553. Neuroimmunogenic architecture in brains of Alzheimer's disease at single-cell resolution. Chen, Y.

PB1554. Neuronal Assay for Investigating Circadian Rhythm Defects in Prader-Willi and Schaaf-Yang Syndrome. Victor, A.

PB1556. Non-desmosomal genes in Arrhythmogenic Cardiomyopathy: genetic variants rating. Bueno Marinas, M.

PB1558. Not one and done: A rare finding of two deleterious variants contributing to the onset of rhabdomyolysis. Kunovac, A.

PB1560. Obesity genomic loci are heterogeneously associated with lipid profiles in ancestrally diverse *Population Architecture using Genomics and Epidemiology (PAGE)* study. Kim, D.

PB1562. Osteoarthritis has high genetic heritability in 488,421 multi-ancestry participants from MVP and the UK Biobank. Wilson, A.

PB1564. Parsing Transcriptomic and Variant Signatures in Tetrapartite Brain Regions Uncover a Collection of Novel Genes in the Neuropeptide-Neurotransmitter Axis Conferring Addiction Risk. Veerappa, A.

PB1566. Patterns of human germline hypermutability identified with whole-genome sequencing. Dong, S.

PB1567. Pediatric Manifestations of Genetic Risk Factors for Polycystic Ovary Syndrome. Zhu, J.

PB1569. Performance of externally developed polygenic risk scores in the All of Us Research Program Database. Hull, L.

PB1571. Phenome-wide association studies of deleterious variants in the Han Taiwanese people. Ko, W.

PB1573. Phenome-wide perspective into the role of Finnish Y chromosome variation in complex diseases. Preussner, A.

PB1574. Phenome-wide PGS portability in the Colorado Center for Personalized Medicine biobank suggests overlooked challenges in diverse populations. Lin, M.

PB1575. PLA2G6 associated late onset Parkinson's disease in a Sudanese family: A case report. Bakhit, Y.

PB1577. Polygenic burden of major depressive disorder and suicidality in children of diverse ancestries: a US-population-based study. Lee, P.

PB1579. Polygenic risk burden is associated with early psychosis and endophenotypes of psychotic disorders. Warren, T.

PB1581. Polygenic Risk score based phenome wide association study for Tourette Syndrome. Jain, P.

PB1583. Polygenic risk scores for quantitative estimation of complex vitamin intake needs to lower homocysteine in the adult population: from population based to personalized dietary intake recommendations. Hager, J.

PB1585. Polygenic score for height influences adolescent growth spurt in Southwestern Indigenous Americans. Ramirez Luzuriaga, M.

PB1588. Population-scale proteo-genomic analysis reveals novel loci associated with Parkinson's disease. Doostparast Torshizi, A.

PB1590. Potential effects of SORBS1 variants on serum immunoglobulin E levels specific to milk intake in relation to prevalence of atopic dermatitis in Korean children. Lee, H.

PB1592. Predicted polygenic transcriptional risk score supports the inference of canalization of polygenic risk of common diseases and traits in the UKBiobank. Nagpal, S.

PB1594. Pre-infection antiviral innate immunity attenuate SARS-CoV-2 infection and viral load in iPSC derived alveolar epithelial cells type-2. Kumar, S.

PB1596. Prioritizing genes and gene programs for disease by integrating genetic and perturbation data. Dey, K.

PB1598. Profiling the neurobiology underlying brain structure in living human subjects. Lund, A.

PB1602. Quantifying mitochondrial dysfunction at biobank scale using insights from rare disease. Gupta, R.

PB1604. Rare copy number variants in *CACNA1H* implicated in essential tremor. Medeiros, M.

PB1606. Rare *Plasmodium falciparum* coronin gene mutations following ACT treatment of malaria in South Western Nigeria. Ajibaye, O.

PB1608. Rare protein-truncating variants in *ZNF518A* are associated with shorter female reproductive lifespan. Shekari, S.

PB1610. Rare variants in ADAMTS13 lead to COVID-19 autosomal dominant disorder, conditioned by SARS CoV-2 infection, sex, and age. Fallerini, C.

PB1612. Rare-Inherited variations in Sensory genes increased burden of Autism Spectrum Disorders: A Whole Exome Sequencing study of Indian families. Siddappa Niranjana Murthy, A.

PB1614. Results from largest GWAS in Gastroparesis point to macrophage polarization etiology. Smieszek, S.

PB1616. Risk factors involved in Congenital Diaphragmatic Hernia, a case-control study in Bogotá and Cali, Colombia. Acevedo Castaño, C.

PB1618. RNA editing regulates interferon induction and host immune response in SARS-CoV-2 infection. Huang, M.

PB1620. Role of amino acids in COVID-19 virulence and pathogenicity: An observational study. Singh, H.

PB1622. Screening of variant rs11190870 nearby *LBX1* gene for association with Adolescent Idiopathic Scoliosis in the population of North India. Singh, H.

PB1624. Self-reported and device-based physical activity and risk of Alzheimer's disease and related dementias: a Mendelian randomization study. Arani, G.

PB1626. Serum osteopontin levels in Mexican patients with systemic lupus erythematosus with lupus nephritis and molecular interaction review. Rivera-Cameras, A.

PB1628. Sex dependent transcriptional changes in response to stress in patients with myalgic encephalomyelitis/chronic fatigue syndrome. Gamer, J.

PB1630. Sex-specific and multiomic integration to enhance accuracy of peripheral blood biomarkers of major depressive disorder. Delahaye-Duriez, A.

PB1632. Sex-specific genetic loci linked to early and late onset type 2 diabetes. Berumen, J.

PB1634. Sex-specific genetic predictors of memory performance in older adults. Eissman, J.

PB1636. Sex-specific risk scores for coronary artery disease using machine learning models. Ye, Q.

PB1638. *SHANK3* domain specific functional characterization of a spectrum of neurodevelopmental disorders phenotypes. Kosaji, N.

PB1642. Significant heritability enrichment for Schizophrenia located in single neuron electrophysiology genes. Li, X.

PB1644. Single nucleotide variants genotyping of IL-6, IL-1β, TNF-α genes promoter region and its protein levels associated to inflammatory response in obstructive sleep apnea and periodontal disease. Sir-Mendoza, F.

PB1646. SnRNA-seq probing of the differential vulnerability of motor neurons in ALS. Alipour, P.

PB1648. Socioeconomic Status and Risk of Stroke: A Mendelian Randomization Study. Myserlis, E.

PB1650. Spectrum of causal genetic variants in inherited cardiomyopathies and arrhythmias in a primarily French-Canadian Population: A 14-year retrospective study from a specialized Cardiovascular Genetic Center. Grondin, S.

PB1652. Stratified LD score regression with 3D genomic features in metabolically relevant cell types implicates a role for pancreatic alpha cells in the pathogenesis of childhood obesity. Trang, K.

PB1656. Telomere length, and Parkinson's disease risk, age at onset, and presentation: Mendelian randomization analyses suggest no causal relationships. Iyengar, A.

PB1658. The bidirectional causal effects of brain morphology across the life course and risk of Alzheimer's disease: A cross-cohort comparison and Mendelian randomization meta-analysis. Korologou-Linden, R.

PB1660. The effect of population structure and ancestry-related assortative mating on the genetic architecture of autism spectrum disorder. Zhang, J.

PB1662. The genetic architecture of the corpus callosum and its subregions. Campbell, M.

PB1664. The genetic risk score affects the lifetime blood pressure and obesity modify the effect of genetic risk. Park, H.

PB1666. *The impact of copy number variants in the genetic testing yield of LQTS patients.* Celeghin, R.

PB1668. The impact of HIV-1 Vpr on host transcription in macrophages. Virgilio, M.

PB1669. The interacting effects of environmental factors and polygenic risk scores on BMI: application to the multiethnic GENNID family study. Wan, J.

PB1671. The Largest Asian GWAS for Systemic Sclerosis Identified a Novel High Risk Candidate Causal SNP in the Fcγ-Receptor Gene Region. Ishikawa, Y.

PB1673. The *PPARG1* gene regulates the aging retina by the modulation of mitochondrial functions, autophagy and senescence in a mouse model of age-related macular degeneration. Blasiak, J.

PB1675. The relationship of Alzheimer disease, genetic risk, and education in individuals of African Ancestry. Akgun, B.

PB1677. The role of *POLG* in the genetic etiology of Amyotrophic Lateral Sclerosis. Russell, N.

PB1679. The whole blood transcriptional regulation landscape in 465 COVID-19 infected samples from Japan COVID-19 Task Force. Wang, Q.

PB1681. To annotate the neuroprotective role of Camel α Lactalbumin and Oleic acid (CLOA) complex against Parkinson's induced model. Ubaid, S.

PB1683. Trans-ancestry PRSs of lipid traits: characterization and evaluation of clinical utility in the diverse PAGE study. Wang, Z.

PB1685. Transcriptome-wide analysis in Cameron County Hispanic Cohort (CCHC) provides novel insights into gene expression signatures of blood pressure. Zhang, X.

PB1687. Transcriptome-wide association analyses in CD4 T-cells identify genes dysregulated in frequent cocaine users and associated with HIV latent reservoir. Quach, B.

PB1689. Transcriptomic analysis of severe obesity identifies novel genes in Hispanic/Latino populations with a high burden of disease. Sharma, P.

PB1691. Transcriptomic and Epigenomic Consequences of Heterozygous Loss-of-function Mutations in *AKAP11*, a Bipolar Disorder Risk Gene, and their Effects on Responsiveness to Lithium. Farhangdoost, N.

PB1693. *TTN* truncating variants in cardiac-expressed exons show high penetrance for cardiomyopathy in carriers with atrial fibrillation. Barrett, K.

PB1695. Understanding the etiology of Alzheimer's disease using genetically predicted gene expression and brain feature models. Wu, E.

PB1697. Undiagnosed complex cases with neurodevelopmental disorders (NDD) after exome sequencing-what's next? Trajkova, S.

PB1699. Unraveling the genetic basis of autoimmune hypothyroidism. Bujnis, M.

PB1701. Untargeted metabolomics on nutrient density and their genomic interaction among Mexican Americans. Chung, S.

PB1703. Using Independent Component Analysis to disentangle obesity heterogeneity discovers genetic drivers of clinical disease signatures and their variation with BMI status. Shuey, M.

PB1705. Using whole genome sequencing to understand the genetic architecture of cerebral palsy in a Canadian cohort.   Wintle, R.

PB1707. Validation of a rapid TTR genotyping assay as a point-of-care tool for cardiac amyloidosis diagnosis in low-income settings. Anyika, C.

PB1709. Variants in *GBA*and *MAPT* influence parkinson disease risk and age at onset in a colombian patient. Satizabal Soto, J.

PB1711. Variation and impact of polygenic hematological traitsin monogenic sickle cell disease. Lettre, G.

PB1713. Whole Exome Sequencing revealed spectrum of mutations associated with different myopathy in clinically suspected DMD patients of Bangladesh. Eshaque, T.

PB1715. Whole genome sequence analysis of long non-coding RNAs for plasma lipid traits. Wang, Y.

PB1717. Whole genome sequencing enables identification of a genetic susceptibility locus for idiopathic pulmonary fibrosis in the 16p subtelomere. Donoghue, L.

PB1719. Whole genome sequencing study in multiple sclerosis identifies novel CTBP2 association with change in brain lesion burden. Bhangale, T.

PB1721. Whole-genome sequencing analysis of fructosamine and glycated albumin in Black and White participants in the Atherosclerosis Risk in Communities (ARIC) Study. Venkataraghavan, S.

Evolutionary and Population Genetics Posters - Thursday (Poster)

PB2755. A likelihood-based framework for demographic inference from genealogical trees. Fan, C.

PB2757. Addressing population stratification in GWAS with variational autoencoders. Yang, D.

PB2759. Analysis of human disease variants from ancestrally diverse Asian genomes. Chan, S.

PB2761. Ancestral genetic tracing defies socio-cultural structure in the north western human populations of India. Singh, A.

PB2763. Ancient trans-species polymorphism at the Major Histocompatibility Complex in primates. Fortier, A.

PB2766. Biological functions and spatial distribution of rapidly evolving brain-expressed genes in the human lineage. Ajumobi, T.

PB2768. Cardiovascular associations and signals of selection at the *Adrenergic Receptor Alpha-1A* gene (*ADRA1A*) in high-altitude Andeans. Moya, E.

PB2770. Characterizing the origin of African ancestral tracts in admixed individuals of the UK Biobank. Shah, N.

PB2772. Coding variants associated with healthy aging: Findings from the Long Life Family Study (LLFS). Gunasekaran, T.

PB2774. ConsensuSV - HPC-ready, ML-enhanced automated pipeline for Illumina-based variant detection. Chilinski, M.

PB2776. Continuous measures of genetic diversity in biomedical research. Dixit, A.

PB2778. *De novo* assembled and phased human genomes from Persian Arab trios show divergent and novel sequence versus CHM13 and GRCh38, providing valuable population specific reference genomes for middle eastern region. Ghorbani, M.

PB2780. Did Neanderthals have different 3D genome folding from modern humans? McArthur, E.

PB2782. Disease-associated STR loci in diverse populations. Steely, C.

PB2785. Examining polygenic adaptation in time-stratified genome samples. Cheng, X.

PB2787. Extensive sex-biased gene flow in Native Americans across the Andes. Borda, V.

PB2789. Generation of synthetic genotypes with neural networks. Geleta, M.

PB2791. Genetic landscape and demographic history of Britain and Ireland. Gilbert, E.

PB2793. Genome-wide patterns of tandem repeat abundance across species are primarily driven by retrotransposons. Gymrek, M.

PB2795. Genomic variation and chromatin structure across the human-specific NOTCH2NL duplications. Real, T.

PB2797. Guaranteeing unbiasedness in selection tests based on polygenic scores. Blanc, J.

PB2799. High coverage whole genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios. Byrska-Bishop, M.

PB2801. Improved detection of evolutionary selection highlights potential bias from different sequencing strategies in complex genomic-regions. Hayeck, T.

PB2803. Imputation of ancient genomes. Mota, B.

PB2804. Inference of natural selection on epigenetic marks: implications for the evolution of gene regulation and germline mutation rates. Boukas, L.

PB2806. Inferences about archaic introgression and positive selection from Lithuanian whole-genome sequences. Urnikyte, A.
PB2808. Inferring germline CpG methylation signature accumulated along the human history from genetic variation catalogs. Si, Y.
PB2810. Integrative approaches linking divergent selection to disease association studies. Shaw, G.
PB2812. Investigating human migrations and genetic diversity in Northeast India over the last   4,000 years using ancient genomics. Bandyopadhyay, E.
PB2814. Kidd Lab 55 AISNP panel shows a better performance than the SNP*for*ID 34-plex for genetic ancestry estimates in a Brazilian population sample. Mendes-Junior, C.
PB2816. Late Night Eating or Circadian disruption could be more dangerous than our think Late Night Eating or Circadian disruption could be more dangerous than our think. Naz, Q.
PB2818. Long-range regulatory effects of Neandertal DNA in modern humans. Yermakovich, D.
PB2820. *MaLAdapt* reveals novel targets of adaptive introgression from Neanderthals and Denisovans in worldwide human populations. Zhang, X.
PB2821. Modeling recurrent mutations predicts allele frequencies and enables precise inference in large samples.   Koch, E.
PB2823. Neanderthal introgression modifies anti-inflammatory response in modern humans. Boye, C.
PB2825. No reduction in diagnostic yield of exome sequencing in prenatal and pediatric patients with non-European ancestries. Mavura, Y.
PB2826. On the Genes, Genealogies, and Geographies of Quebec Anderson-Trocme, L.
PB2828. Phased, long read assemblies from Central African participants represent a more comprehensive, inclusive future for human pangenomics. LoTempio, J.
PB2830. Presenilin-1 G206A mutation in Puerto Ricans with Alzheimer Disease reveals it is as a founder event on the African haplotype. Celis, K.
PB2834. Relatedness inference up to 3rd degree from low coverage ancient genomes in presence of contamination, long runs of homozygosity and ascertainment bias. Popli, D.
PB2836. Repeats R/T ratio is associated with Human Y chromosome Haplogroups. Puurand, T.
PB2838. Resurrecting the alternative splicing landscape of archaic hominins using machine learning. Brand, C.
PB2840. SALAI-Net: Species-agnostic local ancestry inference network. Oriol Sabat, B.
PB2842. Sequencing of 230 primate species identifies conserved enhancers underlying complex human disease. Rashid, S.
PB2844. Simple scaling laws control the genetic architectures of complex traits. Simons, Y.
PB2846. Simulating Genome-phenome Dataset of 1,000,000 Individuals for the European 1+ Million Genomes Initiative. Hiekkalinna, T.
PB2848. Structure-function analysis of a tRNA-derived SINE. Koch, E.
PB2851. The genetic making of Bangladesh. Singh, P.
PB2853. The impact of background selection on deleterious alleles and its consequences on trait evolution and DFE inference. Li, X.
PB2854. The influence of demographic history and genetic architecture on complex phenotypes via runs of homozygosity. Szpiech, Z.
PB2856. The lingering effects of Neanderthal introgression on human complex traits. Wei, X.
PB2858. The spatial distribution of rare deleterious alleles: implications for study design in human genetics. Steiner, M.
PB2860. Understanding ancestry-specific disease allelic effect sizes by leveraging multi-ancestry-matched single-cell RNA-seq and GWAS datasets. Wang, J.
PB2862. Understanding the structure of regional populations with founder effects. Gagnon, L.
PB2864. Using a population-specific reference panel improves genotype imputation accuracy in individuals of African ancestry. Mayanja, R.

Genetic Counseling, ELSI, Education, and Health Services Research Posters - Thursday (Poster)

PB2134. "Frankly, I thought this was one and done": Patient and provider perspectives on return of reclassified variants in clinical oncology. Makhnoon, S.
PB2136. A New System for Triaging Patients in a Medical Genetics Clinic Saves Time and Improves Quality. Randolph, L.
PB2138. A systematic review of health policy guiding the identification, analysis, and management of genomic secondary findings. Majeed, S.
PB2139. Access to Clinical Genetics Service and Testing for Patients with Retinal Dystrophies: a Quality Improvement Study. Shao, Z.
PB2141. Anxiety is a prominent neurobehavioral feature in Kabuki syndrome that significantly affects quality of life. Kalinousky, A.
PB2143. Assessment of cancer patients using a distress thermometer to evaluate perceived and calculated stress in order to identify individuals requiring additional psychological support. Bhende, T.

PB2145. Awareness about rare diseases through Indian cinema. Nanda, A.

PB2147. Benefits, harms and costs of newborn genetic screening for long QT syndrome: estimates from the PreEMPT Model. Christensen, K.

PB2149. Challenges in incorporating polygenic risk scores in the electronic health record. Bangash, H.

PB2151. Clinical utility of returning all clinically relevant secondary findings from genomic sequencing: A systematic review. Mighton, C.

PB2153. Community data-driven approach for generating cross-ethnic population carrier screening panel. Einhorn, M.

PB2155. Considerations for assessing and optimizing remote participant recruitment for a pragmatic trial of polygenic risk score testing. Brunette, C.

PB2157. Cultivating a data-driven computational culture within biomedical institutions by empowering graduate students with code-based data science skills. Ronkowski, C.

PB2159. Delivery of an unfavorable genetic diagnosis: Developing a methodology framework. Chorin, O.

PB2161. Developing Recommendations for Direct-to-Consumer Genetic Testing. Ayoubieh, H.

PB2163. Do Automated Family Histories Significantly Improve Risk Prediction in an EHR? Huang, X.

PB2165. Early Mentoring for Medical Students in Research of Medical Genetics. Samassekou, O.

PB2167. Engagement and Feedback on Genetic Ancestry and Trait Results from >100,000 All of Us Participants. Hoban, H.

PB2169. Engaging Families and Clinicians to Advance Care Coordination and Improve Health Outcomes in Genetic Disease: The Long-Term Follow-Up Cares and Check Initiative. Chan, K.

PB2171. Ethical considerations for genomic newborn hearing screening from the SEQaBOO (SEQuencing a Baby for an Optimal Outcome) study. Morton, C.

PB2173. Evidence-Based Assertions of Clinical Actionability of Secondary Findings: Update from the ClinGen Actionability Working Group. Pak, C.

PB2175. Expanding genomic testing to underserved pediatric populations. Cohen, A.

PB2177. Exploring stigma and discrimination among adults after receipt of neurodevelopmental/psychiatric genetic results in a population-based genomic screening program. Matshabane, O.

PB2179. Factors contributing to preimplantation genetic testing for polygenic disorders (PGT-P) screening and recommendations by reproductive medical professionals. Shah, S.

PB2181. *FLCN* variant of unknown significance complicating a kidney donation process in a family with Birt-Hogg-Dubé. Geurts-Giele, W.

PB2183. Fragmented systems of care: An overview of Canadian health system care models for hereditary cancer syndromes. Sam, J.

PB2185. Genetic counselors and legal recognition: a made-for-Canada approach. Zawati, M.

PB2187. Genetics education in neurodevelopmental disorders for non-genetic health professionals: A scoping review. Peltekova, I.

PB2189. Genomic knowledge, orientation, and empowerment in a demographically diverse population: Preliminary findings from the Texome project. Murali, C.

PB2191. Healthcare providers' experiences counseling patients with results from consumer genomic testing. Trottier, M.

PB2194. Implementation and evaluation of digital patient-reported oncology questionnaire at the East Genomic Medicine Service Alliance. Buczkowicz, P.

PB2196. Implementing public health genomics: The familial hypercholesterolemia model. Manace, L.

PB2198. Incidental findings in study participants: What is the researcher's obligation? Schaare, D.

PB2201. International Best Practices for Genomic Data Sharing in the Health Technology Industries. Knoppers, B.

PB2203. Lessons learned from population-based biobank participants' responses to individual genomic results. Metspalu, A.

PB2205. Masking for whole-exome sequencing data: An analysis of the impact of masking choices in rare coding variants association tests. Nguyen, T.

PB2207. Newborn Screening: a multidisciplinary approach based on landscape analysis, genetic screening, whole genome sequencing, and artificial intelligence to achieve an early diagnosis of rare diseases. Ferlini, A.

PB2209. Parents' attitudes towards return of results from genome sequencing research for their healthy children: A qualitative study. Di Carlo, C.

PB2211. Patient reported utility of cancer results from genomic sequencing. Shickh, S.

PB2213. Personal utility in genomic medicine: what is it and can we measure it? Turbitt, E.

PB2215. Predictors and Trends in Cancer Genetics Clinic Attendance Rate After the Adaptation of Telemedicine During the COVID-19 Pandemic. Smullin, A.

PB2217. Priorities to promote participant engagement in cancer genomics research: A report from the Participant Engagement and Cancer Genome Sequencing (PE-CGS) Network. Schuster, A.

PB2219. Psychosocial impacts in patients/parents with secondary findings reporting from exome sequencing: 1st French multicenter qualitative and quantitative study (FIND Study). Viora-Dupont, E.

PB2221. Raising Genetic Disease Awareness: NBSTRN Efforts to Leverage Social Media to Realize the Promise of Genomics. Unnikumaran, Y.

PB2223. Rethinking general consent for stem cell-based embryo model research. Mittleman, B.

PB2225. Secondary findings in a large Pakistani cohort tested with whole genome sequencing. Skrahin, A.

PB2228. The Million Veteran Program - Return Of Actionable Results (MVP-ROAR) Study: Participant Baseline Characteristics. Danowski, M.

PB2230. The role of oncology nurses in the genetic counseling workflow: A literature scan. Hasser, E.

PB2232. The Texome Project: Initial enrollment in a rare and undiagnosed disease program for underserved communities in Texas. German, R.

PB2236. Users' evaluation of artificial intelligence-based diagnostic program in pediatric rare diseases. Choi, I.

PB2238. Utilizing chatbots for family communication: Uptake and engagement among familial hypercholesterolemia probands from a prospective, pragmatic trial. Walters, N.

PB2240. Voices from the online right: A qualitative study of race and genetics discourse. Sommers, O.

PB2242. Your genes & you: patient motivations regarding secondary findings from genomic studies. Brock, P.

Genetic Therapies Posters - Thursday (Poster)

Conv Ctr/Exhibit/Poster Hall/South Building

PB2018. A cross-species approach using an *in vivo* evaluation platform in mice demonstrates that sequence variation in the human *RABEP2* gene modulates ischemic stroke outcomes. Lee, H.

PB2021. A Structural Screen Approach and Molecular Simulation Identifies Potential Ligands Against the K700E Hot Spot Variant and Functional Pockets of SF3B1 to modulate splicing in Myelodysplastic Syndrome. Garcia, R.

PB2023. Adjunct treatment with glycogen synthase (GYS1) antisense oligonucleotides and Enzyme replacement therapy (ERT) reduced glycogen in the Pompe disease mouse model. Kimonis, V.

PB2024. An allele- discriminative approach for efficient and specific CRISPR/Cas9 based gene therapy of Late Onset of Alzheimer's disease. Kantor, B.

PB2026. An alternate translational start site allows 5' nonsense variants to escape NMD so that readthrough compound ELX-02 and modulators can restore CFTR function. Bowling, A.

PB2028. Antisense oligonucleotide-based exon skipping therapeutic strategy for Cohen syndrome. Ansar, M.

PB2029. Characterization of novel mouse model with 35 bp deletion in *Champ1* gene and exploration of AAV gene therapy to correct syndromes arising from mutations in the *CHAMP1*. Carneiro, T.

PB2031. Deep phenotyping APOC3 Knockouts in a population with high consanguinity. Khalid, S.

PB2033. Dkk1 inhibition normalizes limb phenotypes in a mouse model of *Fzd2* associated omodysplasia Robinow syndromes. Stottmann, R.

PB2035. Drug repositioning network in rare and intractable diseases based on drug target gene analyses. Sakate, R.

PB2037. ECLIPSE, an automated CRISPR platform for the large-scale generation of cell models for the iPSC Neurodegenerative Disease Initiative (iNDI). Deng, P.

PB2039. Efficacy and safety of elamipretide in subjects with primary mitochondrial disease resulting from pathogenic nuclear DNA mutations (nPMD) is investigated through this phase 3 study design. Abbruscato, A.

PB2041. Generation of eight human induced pluripotent stem cell lines from immortalized lymphoblastoid cells. Sertori Finoti, L.

PB2043. Genome-wide CRISPR/Cas9 Screening Reveals ATR as a Therapeutic Target That Overcomes Osteosarcoma Chemoresistance. Tang, S.

PB2045. Harmonization of phenotype ontologies and identification of novel gene/drug-disease associations with Natural Language Processing. Melas, I.

PB2047. Implementing a Genetics-First Strategy in Clinical Development for Rare Brain Diseases. MItchell, A.

PB2049. Learnings from a randomized clinical trial with ARID1B patients. van der Sluijs, P.

PB2051. Localization of KK13 peptide and their potential against ALS. Bhuiyan, A.

PB2053. New class of anaplerotic compounds ameliorates substantial loss of lysine succinylation in propionyl-CoA carboxylase deficient cells. Mohsen, A.

PB2055. Novel siRNA-based therapeutic approach for Megacystis Microcolon Intestinal Hypoperistalsis Syndrome (MMIHS). Baldini, I.

PB2057. Pre-clinical antisense oligonucleotide treatment of CMT2E in a human induced pluripotent stem cell (iPSC)-derived motor neuron model. Medina, J.

PB2059. Sustained efficacy and safety up to 3.5 years in adults with glycogen storage disease type Ia (GSDIa): results from a phase 1/2 clinical trial of DTX401, an AAV8-mediated, liver-directed gene therapy. Mitchell, J.

PB2061. The Jackson Laboratory Center for Precision Genetics: Preclinical research for rare diseases. Lutz, C.

PB2063. Venglustat, a novel, investigational, brain-penetrant glucosylceramide synthase inhibitor, for Gaucher disease type 3: phase 3 LEAP2MONO trial design. Heine, W.

Genetic, Genomic, and Epigenomic Annotations, Databases and Resources Posters - Thursday (Poster)

PB3189. A comprehensive database of Long-chain fatty acid oxidation disorder gene variants. Richbourg, H.

PB3191. A globally representative panel of human genomes: Resource and tutorials to support analyses of diverse populations. Yohannes, M.

PB3193. A Private, universal registry for patient engagement in biomedical research - Xia-Gibbs Syndrome as a model. Hansen, A.

PB3195. A systematic assessment of the completeness of TCR databases across *Mus musculus* strains. He, Y.

PB3197. An improved platform for sharing genomic information of underrepresented populations: BIPMed 2.0. Lopes-Cendes, I.

PB3201. Breast cancer related genes: a genome-first approach of the Geisinger and UK Biobank cohorts. Kim, S.

PB3203. Burden of Mendelian disorders in a large middle eastern biobank. Aamer, W.

PB3205. Colorado Biobank Portal is a cloud platform for the interactive exploration of genome-wide association studies results. Pozdeyev, N.

PB3207. Concordance in variant calls from short-read sequencing data of admixed individuals using different reference genomes. de Oliveira, T.

PB3209. Dynamically querying thousands of genomes to identify genetically matched cohorts with Intel® Optane™ Persistent Memory. Schneider, K.

PB3211. Efficient querying of genomic reference databases with *gget.* Luebbert, L.

PB3213. Genomic database query tracking to understand the prevalence of rare diseases: *ASAH1* in the VarSome database. Confer, N.

PB3215. Growing a research ecosystem in the cloud: Early insights from the All of Us Research Program. Lunt, C.

PB3217. How UK Biobank is democratising access to large-scale genomic and phenotypic data for discovery science. Lacey, B.

PB3219. Identifying Variants of Interest in Samples with Whole Genome Sequencing Data using Kids First Variant WorkBench. Guo, Y.

PB3221. JBrowse 2: a modular genome browser for visualizing synteny and structural variants. Holmes, I.

PB3224. Metadata retrieval from sequence databases with *ffq.* Galvez Merchan, A.

PB3226. MGeND: Integrated database of clinical and genomic information to encourage precision medicine in Japan. Kamada, M.

PB3228. NCBI dbGaP FHIR API Provides Access to Thousands of Studies to Improve Data Integration and Interrogability for Biomedical Research. Phan, L.

PB3229. NIA Genetics of Alzheimer's Disease Data Storage Site (NIAGADS): 2022 Update. Issen, H.

PB3231. Participant-provided case-level data from GenomeConnect can impact variant classification. Morgan, A.

PB3233. Path to independence: Overview of challenges and opportunities of computational data-driven research in biology. Ramesh, T.

PB3236. PreSiBO: Database system for genome guided and data driven drug repurposing. Priyadarshi, D.

PB3238. Regeneron Genetics Center (RGC) Cohort Identification Application. Sharma, D.

PB3240. STR Truth Set for Evaluating Genome-wide STR Calls. Weisburd, B.

PB3242. The Alzheimer's Disease Sequencing Project - Follow Up Study ADSP-FUS: APOE genotype status and demographic characteristics across datasets. Mena, P.

PB3244. The cellxgene suite is an online analytical platform and the largest repository of standardized single-cell data. Aevermann, B.

PB3246. The importance of assessment of gene-disease association in the clinical practice. Bauer, P.

PB3248. The NINDS Human Genetics Resource Center: 20 Years of Impact. Jian, X.

PB3250. The Osteogenesis Imperfecta Variant Database: current state. Stolk, S.

PB3252. Using RGD to analyze genes associated with obesity. Vedi, M.

PB3254. VariantMatcher a tool to enable connections amongst individuals with interest in a specific variant. Paulo Martin, R.

PB3256. Web Tools and Resources for Generating and Accessing ACMG Criteria Specifications for Variant Classification by ClinGen Variant Curation Expert Panels. Shah, N.

Mendelian Phenotypes Posters - Thursday (Poster)

PB1724. A A recurrent de novo mutation in *ZMYND11.* Carrion, T.

PB1726. A case of Fanconi anemia diagnosed due to *BRCA2* gene mutation triggered by cancer genome profiling test. Fukuda, K.

PB1728. A case report of an Egyptian patient with a severe neurodevelopmental disorder and a novel biallelic loss-of-function variant in *GOLGA2*. Khang, R.

PB1730. A Difficult Dual Diagnosis: Dilemmas in the Era of Next Generation Sequencing. Rekab, A.

PB1732. A Highly Polymorphic VNTR in the *DRD4-DEAF1* Intergenic Region. Vandenbergh, D.

PB1734. A multi-branch family with mutation in *SLC26A4* gene from a village in the southeastern region of Iran. Mohseni, M.

PB1736. A novel de novo nonsense variant in TBX2 cause osteochondrodysplasia. Umair, M.

PB1738. A novel homozygous variant in *HECW2* gene is associated with intellectual disability and epilepsy in a Malian family. Sangare, M.

PB1740. A novel homozygous variant in *IBA57* causing multiple mitochondrial dysfunction syndrome type 3. Lang, S.

PB1742. A novel *PUM1* heterozygous missense variant causing severe developmental delay and epilepsy from infancy with dysmorphic features. Sato, E.

PB1744. A novel XL gene for autism with psychiatric illness in adults. Alakeel, H.

PB1746. A prospective, longitudinal observational natural history study of patients with NGLY1 Deficiency. Tong, S.

PB1748. A recurrent missense variant in *ITPR3* causes demyelinating Charcot-Marie-Tooth neuropathy. Beijer, D.

PB1750. A substantial proportion of high myopia is caused by mutations in *CACNA1F* (XL CSNB). Hoefsloot, L.

PB1752. Allele-specific inactivation of epidermolysis bullosa simplex mutations using CRISPR-Cas9 and spraying of the corrected cell suspension onto the wounds. Bchetnia, M.

PB1754. An initiative for the diagnosis and study of rare and undiagnosed diseases in Mexico. Gonzaga-Jauregui, C.

PB1756. *ARG1* variants in Arginase 1 Deficiency: genetic characterization of participants in the pegzilarginase clinical trials. McNutt, M.

PB1758. ATP2C1</i>as a candidate for Interstitial Cystitis/Bladder Pain Syndrome. Estrella, E.

PB1760. Atypical molecular findings in patients with capillary malformations. Montano, C.

PB1761. Bi-allelic ACBD6 variants lead to a neurodevelopmental syndrome with progressive complex movement disorders. Maroofian, R.

PB1763. Biallelic founder mutation in PDE2Acausesparoxysmal dyskinesia with Intellectual disability in Pakistani families. Fatima, A.

PB1765. Biallelic Loss of Function Mutations in PYGM Cause Hereditary Macular Dystrophy. Hussein, R.

PB1767. Biallelic loss-of-function variants in mitochondrial phospholipase *PNPLA8* decrease in the number of basal radial glial cells and lead to microcephaly with simplified gyral pattern. Nakamura, Y.

PB1769. Biallelic nonsynonymous variants in LTV1 are associated with LIPHAK syndrome, a novel poikiloderma-like disorder. Han, J.

PB1771. Biallelic *RAD51C* loss-of-function variants drive perizygotic SNV/indel hypermutator phenotype in a subject with Fanconi anemia complementation group O. Zemet Lazar, R.

PB1773. Bialletic variants in *FICD* leading to inactivation of BiP cause motor neuron disease. Rebelo, A.

PB1775. Billalelic *ATOH1* Variant in Siblings With Pontocerebellar Hypoplasia, Developmental Delay, and Hearing Loss. Writzl, K.

PB1777. Brachyolmia with amylogenesis imperfecta: Identification of a Novel LTBP3 gene Mutation in several Druze Arab patients from the north of Israel. Hadid, Y.

PB1780. Characterization of associated non-classical phenotypes in patients with deletion in WAGR region identified by chromosomal microarray: new insights and literature review. Mendes-Júnior, C.

PB1782. Characterization of transthyretin *TTR* missense variants for associations with hereditary transthyretin-mediated (hATTR) amyloidosis and hallmark symptoms in 470,000 UK Biobank whole exome sequences. Plekan, M.

PB1784. Chd8 mutation in mouse impacts cerebellar function across anatomical, genomic, electrophysiological and behavioral axes. Canales, C.

PB1788. Clinical diversity and molecular mechanism of VPS35L-associated Ritscher-Schinzel syndrome. Otsuji, S.

PB1789. Clinical usefulness of a custom Next Generation Sequencing gene panel in providing molecular diagnosis to patients with a broad spectrum of Neurodevelopmental Disorders. Giordano, M.

PB1791. Comparing phenotypes across five developmental and epileptic encephalopathies (DEEs) through evaluation of 2490 patient data years. Mallory, E.

PB1793. *Compound heterozygous IFT81 variants causing brachydactyly with cone-shaped epiphyses and generalized metaphyseal dysplasia.* Carter, E.

PB1795. Confirmation of association of TGFBI p.Ser591Phe mutation with variant lattice corneal dystrophy. Chung, D.

PB1797. Co-occurring anomalies in cases with achondroplasia. Stoll, C.

PB1799. De novo (deep-)intronic splicing mutations in patients with neurodevelopmental disorders. van der Sanden, B.

PB1801. *De novo* missense variants in *SEPHS1* Cause a Novel Neurodevelopmental Disorder with Developmental Delay, Hypotonia, Muscle Weakness, Speech Delay, and Growth delay. Mullegama, S.

PB1803. *De novo* truncating *ADNP* variants as a recurrent cause of neurodevelopmental disorders. Gusic, M.

PB1805. Deciphering the phenotypic variability of the most common deafness-causative *GJB2* p.V37I variant in Asia. Chiang, Y.

PB1806. Deep resequencing of the 1q22 locus in non-lobar intracerebral hemorrhage. Parodi, L.

PB1808. Defining the nuclear genetic architecture of a maternally-inherited mitochondrial disorder. Boggan, R.

PB1810. Deleterious *SNAPC4* Variants are Associated with a Neurodevelopmental Disorder. Frost, F.

PB1812. Delineation of gain-of-function MYCN-induced novel megalencephaly syndrome and possible out-of-brain complications implicated by a mouse model. Nishio, Y.

PB1814. Diagnosis, treatment, and follow-up care for patients with Barth syndrome is presented through the results from 1:1 interviews of pediatric cardiologists. Kelly, G.

PB1816. Diagnostic utility of comprehensive genomic and transcriptomic profiling of anorectal malformations. Ramadesikan, S.

PB1818. *Drosophila* model of de novo *MRTF-B* variants highlights the critical role of actin regulation. Andrews, J.

PB1820. Effective Genetic Diagnosis for Unilateral and Asymmetric Pediatric Hearing Loss via Exome Sequencing. Redfield, S.

PB1822. Enrichment of *RAI1* genetic aberrations associated with sleep disturbances in Smith-Magenis syndrome and autism spectrum disorder. Kaden, A.

PB1824. ERF-Related craniosynostosis: further delineation of the new craniosynostosis syndrome. Santillán Martínez, R.

PB1826. Exome Sequencing in Understanding the Etiologies of Ataxia in Children. Phadke, S.

PB1828. Exome sequencing uncovers novel variants in two Malian families with Epileptic encephalopathies. Maiga, A.

PB1829. Expanding the Phenotypic spectrum of *SMARCA5*-related neurodevelopmental disorder. Zambrano, R.

PB1831. Expert perspectives on sequencing newborns for treatable genetic conditions. Gold, N.

PB1833. Exploring cancer cachexia on the spatial plane. Park, Y.

PB1835. Family with case of HMG-CoA lyase deficiency with novel pathogenic variant in *HMGCL.* Froukh, T.

PB1837. First report of severe hemolysis associated with *SCARB1.* Tanpaiboon, P.

PB1839. Fragile X Syndrome in a female patient: A case of strong family history and developmental delay. Marbaker, B.

PB1841. Frequency of Y chromosome microdeletion and its correlation with spermatogenesis defect in north Indian infertile males. Sharma, H.

PB1843. Functional characterization of recurrent truncating variant in *UBAP1* associated with hereditary spastic paraplegia. Gu, S.

PB1845. *GATA3*-related pedigrees showing characteristic audiologic profile and inheritance pattern. Kim, B.

PB1849. Genetic and clinical landscape of childhood cerebellar hypoplasia and atrophy. Sakamoto, M.

PB1851. Genetic insights from consanguineous cardiomyopathy families. Jamshidi, Y.

PB1853. Genetic modifiers in Niemann-Pick type C1 disease. Sitarska, D.

PB1856. Genetics of intellectual disability in pakistani consanguineous families. Rasheed, M.

PB1858. Genomic and transcriptomic characterization of a novel *RYR1* variant in the French-Canadian population. Labrecque, M.

PB1860. Genomic region identified on chromosome 22 contributing to lymphedema in Phelan-McDermid syndrome. Sarasua, S.

PB1862. GWAS of cleft palate trios reveals novel associations and subtype-specific effects. Robinson, K.

PB1864. Haploinsufficiency and loss-of-function of LEF-1 cause a novel Mendelian disorder by dysregulating WNT signaling. Asif, M.

PB1866. Hemizygous CNV in chromosome region Xq23 is associated with a complex psychiatric and bone mineral density disorder. Kapalanga, J.

PB1868. Heterogeneity of Inherited Cone Dysfunction Disorders with Normal Fundus Appearances. Joo, K.

PB1870. High prevalence of frontotemporal dementia in females of five Hispanic families with R159H VCP multisystem proteinopathy. Shmara, A.

PB1872. Homozygous *EMC1* variant in four families from the same Kuwaiti tribe with cerebellar atrophy, visual impairment, and psychomotor retardation. Alenzi, M.

PB1874. Human brains with Tay Sachs disease exhibit altered transcriptomes during fetal development. Han, S.

PB1876. Hypophosphatemia gene panel sponsored program: a comparison of findings from 2019 through 2022 describing *PHEX* variant landscape and molecular diagnosis of XLH. Miller, N.

PB1878. Identification of a novel compound heterozygous mutation in *RyR1* gene in an Indian family affected with congenital myopathy. Kirola, L.

PB1880. Identification of a Novel Therapeutic Target Underlying Atypical Manifestation of Gaucher Disease. Do, H.

PB1882. Identification of *INHB*-related neurodevelopmental disorder. Pierson, T.

PB1884. Identifying modifier genes in a PIGA-CDG pedigree with reduced penetrance. Thorpe, H.

PB1886. *In Silico* genomic interrogation reveals 16 autosomal dominant neurodevelopmental candidate genes at 1p13.3. Ben Mahmoud, A.

PB1888. Integrating Genomic and Phenotypic Analyses of Autonomic Nervous System Dysfunction in a Rare Neurological Disease Cohort. Rivera-Munoz, E.

PB1890. Investigating SLC6A1 targeted therapeutics in *Drosophila.* Jay, K.

PB1892. Investigating tissue specific defects in mitochondrial bioenergetics and quality control: new implications for cellular pathogenesis and therapeutic targeting in Barth syndrome. Sniezek, O.

PB1894. Isolated acanthosis nigricans in a Mexican girl with a nonsense variant in *FGFR3* gene. Abreu González, M.

PB1896. Joint analysis of de novo mutations in multiple traits and gene-expression data improves statistical power for the prioritization of genes associated with disease. Nguyen, T.

PB1898. Large-scale genomic analyses of 150 consanguineous kindreds from the Middle East and North Africa identify novel neurodevelopmental disease mechanisms. Duan, R.

PB1900. Leveraging orthogonal sequencing and optical mapping technologies for the precision diagnosis of neurodevelopmental disorders in a Middle Eastern family based cohort. Siddig, Z.

PB1902. Lisch epithelial corneal dystrophy is caused by heterozygous loss-of-function variants in *MCOLN1.* Patterson, K.

PB1904. Local versus systemic control of bone and skeletal muscle mass by targeting components of the transforming growth factor-β signaling pathway using pharmacological and transcriptional approaches. Chandok, H.

PB1906. Long term prognosis in two adult patients with malonyl CoA decarboxylase deficiency. Yano, S.

PB1908. Loss of adipocyte phospholipase gene*PLAAT3*causes lipodystrophy with neurological features due to inactivated arachidonic acid-mediated PPARℽ;;;; signaling. Schuermans, N.

PB1910. Loss of FOCAD, operating in the SKI mRNA surveillance pathway, causes a pediatric syndrome with liver cirrhosis. Moreno Traspas, R.

PB1912. *MED13L* knockout in cerebral organoids leads to a shifted developmental program through abnormal cis-regulatory element activation. Ghoumid, J.

PB1914. Metatranscriptomics reveals association of α-, β-, and γ-HPVs with typical epidermodysplasiaverruciformis in a large cohort of patients with CIB1, TMC6, or TMC8 mutations. Youssefian, L.

PB1916. Molecular and behavioral characterization of a novel mouse model of Snyder-Robinson Syndrome: A path towards therapeutic development. Kemaladewi, D.

PB1918. Molecular Genetic Defects of Hereditary Angioedema in Saudi Arabia. Alajlan, H.

PB1920. Monoallelic variation in the DExH-box helicase DHX9, a product of the *DHX9* gene paralog, perturbs neurodevelopment & causes peripheral nerve axon degeneration. Calame, D.

PB1922. Multi-omic approach identifies a novel non-coding deletion at Xq28 in a patient with X-linked primary immunodeficiency. Bonner, D.

PB1924. Mutation-specific pathophysiological mechanisms of *AFF3.* Bassani, S.

PB1926. New *trans*-modifier genes in the most common monogenic eye disorder, ABCA4/Stargardt disease. Zernant, J.

PB1928. Novel homozygous nonsense mutation of *MLIP* and compensatory alternative splicing in a late-onset distal myopathy. Mezreani, J.

PB1932. Novel variants expand understanding of *PPP1R13L* - related cardio-cutaneous syndrome. Close, C.

PB1934. One family, two disorders, three affected: a diagnostic odyssey highlighting the importance of WES as first-tier testing in patients with a specific phenotype but a consanguineous background. Alsaleh, N.

PB1936. Patients with biallelic GGC repeat expansions in *NOTCH2NLC* exhibiting a typical neuronal intranuclear inclusion disease phenotype. Kameyama, S.

PB1938. PHACE syndrome: New strategies to identify the disease mechanism for this unsolved recognizable condition. Cuillerier, A.

PB1940. Phenotypic variation of outcomes for tail-domain *KIF5A* related variants. Jose, M.

PB1944. Precision diagnostics for GLUT1 disorder using deep mutational scanning. Ricardo, B.

PB1946. Predictive modeling to define the locus heterogeneity of tRNA synthetase-related peripheral neuropathy. Cale, A.

PB1947. Probing variants associated with neurodevelopmental disorders by studying social behaviors in *C. elegans.* Pierce, J.

PB1948. Proposal of intervention of the IL-10 rs1800872 (-592 C>A) as a modulator in the cardiovascular variant of Fabry disease. Ruiz Ramirez, A.

PB1950. Rapid Turnover Skeletal Disease from In-frame Tandem Duplication of Exons 4-9 in *TNFRSF11A* Encoding RANK. Mumm, S.

PB1952. Rare variants in syndromic ciliopathy genes as novel causes of isolated renal disease in adults. Sentell, Z.

PB1954. RBM12 Variants Lead to a Novel Neurodevelopmental Syndrome With Atypical Genotype-Phenotype Correlations. Bhoj, E.

PB1956. Recessive variants in *SLC9A1* cause a syndrome of cerebellar ataxia, amelogenesis imperfecta and variable sensorineural hearing loss. Pellerin, D.

PB1958. Reconsidering SIX5 as a monogenic cause for Congenital Anomalies of the Kidney and Urinary Tract (CAKUT) and Branchio-oto-renal syndrome (BOR). Chang, J.

PB1960. Report of *PLP1* Gene Mutation in Pelizaeus-Merzbacher Disease. Seyedhassani, S.

PB1962. *RFC1* expansions in Italian population: Molecular analysis, structure/size determination and clinical features of patients with CANVAS/spectrum phenotypes. Di Bella, D.

PB1964. Rothmund Thomson syndrome with congenital cataracts and severe growth restriction is associated with a not previously described gene in an autosomal recessive inheritance pattern in seven families. Yamamoto, G.

PB1966. Seizures in a patient with Autosomal recessive Intellectual disability type 5. Aliu, E.

PB1968. Severe ABCA4-retinal dystrophy caused by homozygosity for a novel double mutated allele. Aslaksen, S.

PB1970. Siblings with hypophosphatemic rickets and homozygous *CYP27B1* variants. Wang, L.

PB1972. Slc9a6 mutation causes Purkinje cell loss and ataxia in the *shaker* rat. Figueroa, K.

PB1974. *SRSF1*haploinsufficiency is responsible for a syndromic developmental disorder with intellectual disability and variable marfanoid habitus. Garde, A.

PB1976. Sunken symptoms of *SRCAP* mutation: A case report of atypical phenotype of Floating Harbor Syndrome. Nesheiwat, N.

PB1978. Systematic identification of disease-causing near-coding variants in 29,759 individuals with rare disease. Whiffin, N.

PB1980. Targeted deep sequencing analyses of long QT syndrome in a Japanese population. Nagata, Y.

PB1982. Temtamy Preaxial Brachydactyly Syndrome: penetrance in heterozygotes for a *CHSY1* missense variant. Sabeh, P.

PB1984. The benefits and yields of whole exome sequencing data reanalysis in Iranian undiagnosed patients with intellectual disability. Fattahi, Z.

PB1987. The genetic background in a group of 155 Polish pediatric patients with unexplained epilepsy. Chalupczynska, B.

PB1989. The genetic landscape of neurodevelopmental disorders in a large cohort of multiplex consanguineous families from Turkey. Reis, A.

PB1991. The mechanism and current concepts and future research direction of rare hereditary disease White Sponge Nevus. Cai, W.

PB1992. The Spectrum of PLEC Sequence Variants and Related Plectinopathies Including Novel Association with Epidermolysis Bullosa Pruriginosa. Uitto, J.

PB1994. Three new founder Bukharian Jewish mutations in *COL4A4* are causative of autosomal dominant and recessive Alport syndrome. Levy, M.

PB1996. Toward precision medicine in Neurodevelopmental disability: identifying phenotypic differences in individuals with global developmental delay. Cuppens, T.

PB1998. Translated mutant *DSPP* mRNA expression level impacts the severity of dentin defects. Kim, J.

PB2000. Tumor burden among members of the DiscovEHR cohort harboring deleterious constitutive variants in*NF1*. Pemov, A.

PB2003. Using artificial intelligence driven NGS re-analyses followed by full genome profiling (HiFi-GS) for unsolved pediatric white matter disorders. Thiffault, I.

PB2004. Using genetic relatedness within biobanks to understand genetic architecture of Retinitis Pigmentosa. Baker, J.

PB2007. Variants of *LRP2*, encoding a multifunctional cell surface endocytic receptor, are associated with hearing loss and retinal dystrophy. Faridi, R.

PB2009. Whole exome sequencing identifies novel variants in familial myoclonic epilepsy in Mali. Bamba, S.

PB2011. Whole exome sequencing reveals known and candidate genes in non-syndromic hearing impairment in Mali. Yalcouyé, A.

PB2013. Whole genome sequencing identifies biallelic *SCLT1* splice-region variants in a child with a suspected ciliopathy: Delineating the *SCLT1*related ciliopathy spectrum. Gillesse, E.

PB2015. Whole-exome sequencing of Pakistani consanguineous families identified variants in novel and known genes of severe intellectual disability. Abdullah, A.

Molecular and Cytogenetic Diagnostics Posters - Thursday (Poster)

PB2244. A Case of Concurrent 3p26.3-p25.3 Deletion and 9q34.2-q34.3 Duplication in Monozygotic Twin Males: A Case Report. Maini, T.

PB2246. A framework designed to improve the evaluation and reporting of incidental findings: Implementation and experience of a clinical laboratory in the first 15 months. Brown, C.

PB2248. A multiomics approach to resolving small supernumerary marker chromosomes. Grochowski, C.

PB2250. A national initiative working toward equitable translational genomic research for Canadian Indigenous families: Early findings indicate the need to reduce uncertainty in variant interpretation. Jacob, K.

PB2252. A novel FAME1 repeat configuration in a European family identified using a combined genomics approach. Maroilley, T.

PB2255. A robust NGS detection workflow for the *IKBKG* gene. Yen, H.

PB2257. Accurate Total and Allele-Specific Copy Number Quantification of the Cardiovascular Disease-Associated Lipoprotein (a) Kringle-IV Type-2 Tandem Repeat Using Illumina Sequencing. Belyeu, J.

PB2259. Analysis of mRNA and protein expression levels of MECP2 in missense mutant iPSC and lymphoblast lines against isogenic controls. Thiruvallur Madanagopal, T.

PB2262. Beyond NGS: RNA-based functional evaluation improves ACMG classification of VUS with potential Loss of function. Blouin, J.

PB2263. Challenges of characterisation of medically relevant tandem repeats in whole genome sequencing data. Lojova, I.

PB2265. Chromosomal and molecular investigations of Senegalese children bearing disorders of sex development. Younoussa, H.

PB2267. Clarifying genetic disease with long range sequencing: Tipping the balance for structural variant detection in SMAD3. Safgren, S.

PB2269. Clinical long-read genome sequencing: Analytical performance of germline small variant detection using HiFi genome sequencing. Hammond, N.

PB2271. Clinical validation of end-to-end whole genome sequencing and interpretation applied to both panel-based and diagnostic genome testing. Salman, A.

PB2274. Comparison of chromosomal inversions in three different datasets. Bozkurt, T.

PB2276. Comparison of positive findings between CMA/exome combo and Whole Genome Sequencing: Aclinical lab experience. Zhao, X.

PB2277. Copy Number Variations (CNVs) detected in 14% of patients presenting with peripheral neuropathies in a French cohort. Lia, A.

PB2279. Deciphering the genetic etiology of idiopathic male infertility by low-pass mate-pair genome sequence analysis. Qian, J.

PB2281. Development and analytical validation of an innovative 397 gene genome-based inherited kidney disease panel. Weymouth, K.

PB2283. Development of a comprehensive whole genome sequencing test for cardiovascular disease patients. Amendola, L.

PB2285. Diagnostic approach of transthyretin amyloidosis in a Colombian patient. Estela-Zape, J.

PB2287. Diagnostic utility of the targeted next generation sequencing panel test for suspicious genetic glomerular diseases. Kim, J.

PB2291. *DST* variants are responsible for neurogenic arthrogryposis multiplex congenita confirming the large clinical spectrum of type VI hereditary sensory autonomic neuropathy. Capri, Y.

PB2293. Evaluating the potential for using internal data in assigning benign classification to variants of uncertain significance. Wang, J.

PB2295. Examining the factors impacting molecular diagnosis during clinical exome sequencing re-analysis. Chan, A.

PB2297. Generic genome sequencing: one lab flow for all. Schobers, G.

PB2299. Genetic testing of *STRC* for hearing loss: a clinical lab's approach to pseudogenes. Luo, M.

PB2301. Genome sequencing enables parent of origin analysis of the X chromosome in probands with Klinefelter syndrome. Seifert, B.

PB2303. Genome-wide investigation of potentially pathogenic copy number variants & mechanisms fomenting their origins. Dardas, Z.

PB2304. Genomic sequencing generates less uncertainty than panel-based testing: Results of over 1.5 million tests. Rehm, H.

PB2305. GMCK-RD a sucess story: Implementation of genomic medicine in Stockholm healthcare region, update on first 10,000 samples. Lindstrand, A.

PB2307. Identification of a small duplication in the upstream noncoding enhancer of *SOX9* in a family with 46,XX disorder of sexual development by clinical whole genome sequencing. Sajan, S.

PB2311. Importance of the reclassification of the clinical significance of new genetic variants in non-classical mucopolysaccharidosis type IV A. Gomez, R.

PB2313. Increased diagnostic yield from negative whole genome-slice panels using automated reanalysis. Berger, S.

PB2315. Increasing global access to clinical whole genome sequencing for under-resourced undiagnosed individuals. Terry, S.

PB2317. Integrating population-scale data into structural variant visualization. Chowdhury, M.

PB2319. Is 22q11.2 deletion syndrome truly less common in African American patients? McDonald-McGinn, D.

PB2321. Leveraging genomic advances to ascertain the cause of congenital limb defects in Indian subcontinent. Langeh, N.

PB2323. Low-level paternal mosaicism of novel pathogenic *DNM1L* variant confirms EMPF1 diagnosis in proband, highlighting importance of deep sequencing when parental carrier status is ambiguous. Akler, G.

PB2325. Minimizing false negatives in NGS-based diagnostic testing by optimizing variant filtering strategy. Guan, B.

PB2327. Molecular characterization of a rare case of chromosome 20p duplication syndrome in an Egyptian male patient with multiple congenital anomalies. Abd Allah, S.

PB2329. Multiple molecular diagnoses identified by clinical whole genome sequencing in an international cohort of more than 1,200 individuals. Thorpe, E.

PB2331. Non-invasive detection of *TEK* mutations in a cohort of children with isolated venous malformations using cell-free DNA. Au, N.

PB2333. Optical genome mapping and whole genome sequencing in a case of multiple chromosomal rearrangements. Levy, J.

PB2335. Optical genome mapping identifies double parental paracentric inversions as risk factor for atypical monocentric recombinant chromosomes in offspring. Kuentz, P.

PB2337. Pallister-Killian syndrome: Characterization of the isochromosome 12p by fluorescent in situ hybridization: A Case Report. Lavu, S.

PB2339. Positive Findings in Clinical Mitochondrial Genome Testing by NGS. Yang, J.

PB2341. Rapid whole genome sequencing in the neonatal intensive care unit of Brazilian hospitals.. Migliavacca, M.

PB2343. Return of incidental genetic findings to pediatric patients: considerations and opportunities from experiences in genome sequencing. Bowling, K.

PB2345. Somatic Mutational Landscape of Extracranial Arteriovenous Malformations and Phenotypic Correlations. Eyries, M.

PB2347. Strategies to Reduce Inconclusive Genetic Testing Results for Inherited Cardiomyopathies: Perspectives from a Clinical Diagnostic Laboratory. Bronicki, L.

PB2349. The power of whole genome sequencing: Identification of an undocumented homozygous partial gene deletion in a patient with a neurological phenotype. Fischer, G.

PB2351. Towards dried blood spot-based RNAseq in routine diagnosis of genetic disease. Beetz, C.

PB2353. Two new Tunisian cases of achondroplasia/hypochondroplasia : phenotype-genotype correlation. Abdelmoula, B.

PB2355. Unexpected CNV anomalies using exome or genome-wide approach for complex phenotypes: report of a Regional Genetic Center experience. Plaiasu, V.

PB2357. Unusual X-chromosome composition in mother and daughter with different karyotype constitutions. Melaragno, M.

PB2359. Utilization of the Age-based Semi Quantitative Metric (ASQM) to evaluate gene-condition pairs for inclusion in Early Check's newborn sequencing panel. Cope, H.

Molecular Effects of Genetic Variation Posters - Thursday (Poster)

PB2492. A Catalogue of Transcriptomes and Associated Genetic Effects on 2,000 Qataris Uncovering the Functional Impact of Middle Eastern Genetic Variation and Identifying Novel Pathways Underlying Human Traits and Diseases. Mokrab, Y.

PB2494. A genome-first approach characterizing the frequency and phenotype of potentially pathogenic germline *DGCR8* variants in two large, unselected cohorts. Kim, J.

PB2496. A humanized yeast model to identify disease-associated, dominant-negative effects on protein function. Antonellis, A.

PB2498. A Novel De novo Likely Pathogenic Variant of *WFS-1* gene in a Pakistani Child. Hanif, M.

PB2500. A risk variant for Barrett's esophagus and esophageal adenocarcinoma at chr8p23.1 affects enhancer activity and implicates multiple gene targets. Buas, M.

PB2502. A splicing variant found in the human myostatin gene encodes an isoform that inhibits myostatin. Maeta, K.

PB2504. A uniquely prevalent deleterious variant within *POMC* gene identified in Estonian Biobank cohort. Abner, E.

PB2506. Aberrant splicing prediction across human tissues. Wagner, N.

PB2508. Abundance of immuneprotein CD99 affected by loss of chromosome Y. Mattisson, J.

PB2510. All-but-one conditional analysis of eQTL isolation in peripheral blood sheds light onto causal variant prioritization. Brown, M.

PB2512. Amplicon sequencing-based noninvasive fetal genotyping for RHD-Positive D antigen-negative alleles. Hori, A.

PB2514. An Alzheimer's disease risk variant in *TTC3*modifies the growth and transcriptional profile of iPSC-derived forebrain neurons. Cukier, H.

PB2516. An updated genetic atlas of the plasma proteome: Findings from the UK Biobank Pharma Proteomics Project (UKB-PPP). Sun, B.

PB2517. Antibody repertoire heavy chain gene usage is explained by common genetic variants in the immunoglobulin heavy chain locus. Rodriguez, O.

PB2518. *APOE* Genotype Contributes to Distinct Profiles in Human iPSC Astrocytes and Neurons. Clayton, S.

PB2520. Assessing the genetic control of whole blood RNA editing and its role in modulating risk of common diseases. Stacey, D.

PB2522. Association of GWAS and candidate gene loci of dopaminergic system with major depression, schizophrenia and bipolar disorder in the Pakistani population. Hashmi, A.

PB2524. Bayesian fine-mapping of sex-specific gene regulation during neurodevelopment shows link to neurodevelopmental disorders. Lalli, J.

PB2527. CD4+ T cell gene expression influences COVID-19 severity and susceptibility: results from single cell sequencing and Mendelian randomization. Willett, J.

PB2529. Characterization of a leaky splice mutation in *AIRE*, associated with milder phenotype and late debut of autoimmune polyendocrine syndrome type 1. Berger, A.

PB2531. Characterization of mutants in SOS1 in an endothelial cell model of lymphatic anomaly. Matsuoka, L.

PB2533. Characterizing the effects of a novel *RDH12* donor splice site variant using *in vivo*, *in vitro*, and *in silico* methods. Chung, M.

PB2535. Clonal hematopoiesis of indeterminate potential and kidney function decline in the general population. Vlasschaert, C.

PB2537. COL9A1 and FLNB: Variants corresponding to complex clinical phenotype. Vázquez, J.

PB2539. Combining human genetics and functional genomics identifies regulators of the lysosomal lipid BMP. Leto, D.

PB2542. Comprehensive detection of trans-regulatory signal from scRNA-seq and perturbation datasets. Babushkin, N.

PB2544. Conditionally distinct adipose eQTL signals in 2,256 individuals identify hundreds of colocalized genes for cardiometabolic traits. Brotman, S.

PB2546. Connecting rare variation to extremes of plasma protein levels. Xie, X.

PB2548. CRISPR perturbation screens identify predicted regulatory sequences and genes within GWAS loci associated with erythrocyte density traits. Brosseau, N.

PB2550. Cryptic AS-NMD elements harbor relevant variants in probands with early onset genetic disease. Felker, S.

PB2552. Defining the effects of noncoding genetic variation on human regulatory element activity. Strouse, K.

PB2554. Detection of rare Synaptogyrin 3 (*SYNGR3*) gene missense mutations in patients with schizophrenia. Cheng, M.

PB2556. Development of a multiplexed assay of variant effects for the DNA repair gene. MUTYH   Hemker, S.

PB2558. Discovery and finemapping of eGFR loci in metaanalysis GWAS of 80K African ancestry individuals. Kintu, C.

PB2560. Dissecting the downstream effects of IL-6 signaling on atheroprogression: a proteome-wide Mendelian randomization study. Georgakis, M.

PB2562. Dual inhibition of mTORC1 and mTORC2 activation effectively rescues hyperproliferative lymphatic sprouting in a cell-based model system of complex lymphatic anomalies. Battig, M.

PB2564. Elucidating genomic disease associations using transcriptomic, proteomic and metabolomic data in 4,732 individuals. Persyn, E.

PB2566. Elucidating the mechanism of a neurological syndrome caused by germline mutations in *H3F3A* and *H3F3B.* Sangree, A.

PB2568. Epitranscriptomic m6A alterations in C9orf72 ALS cerebellum. Ross, J.

PB2570. Establishing a deep mutational scan for functional classification of MSH6 missense variants. Scott, A.

PB2572. Estimating Diagnostic Noise in Panel-Based Genomic Analusis. Beaumont, R.

PB2574. Evaluation of the ACMG PM5 variant pathogenicity evidence guideline. Maston, G.

PB2576. Evolutionary study and variant analysis in the mcph1 gene in patients diagnosed with non-syndromic hearing impairment. Oluwole, O.

PB2578. Exome analysis for bronchial asthma in the Mennonite population reveals an association with *NOTCH4* polymorphisms. Bucco, I.

PB2580. Expanding the rare *DHCR24*-related sterol biosynthesis disorder: genotype -phenotype correlation. Cocciadiferro, D.

PB2582. Explicable prioritization of genetic variants by integration of rule-based and deep learning algorithms for diagnosis of rare Mendelian disorders. Lee, K.

PB2584. Fast Forward - is Multiomics a resurgence of old? Hegde, M.

PB2586. FOXI3 pathogenic variants cause one form of craniofacial microsomia. Antonarakis, S.

PB2588. Full penetrance of a hypomorphic TYR gene variation in oculocutaneous albinism. Velez, C.

PB2590. Functional and *in silico* analyses of variants found in mitochondrial trifunctional protein deficiency patients. Vieira Neto, E.

PB2592. Functional characterization of new non-coding pathogenic variants creating upstream Open Reading Frames in the 5UTR of the Endoglin gene and causing Hereditary Hemorrhagic Telangiectasia. Soukarieh, O.

PB2594. Functional characterization of polycystic ovary syndrome-associated risk loci identifies genetic regulatory regions. Sankaranarayanan, L.

PB2595. Functional impact of rare variants and sex across the x chromosome and autosomes. Ungar, R.

PB2597. Gene expression profiling with direct long-read RNA sequencing uncovers functional variation affecting transcripts production. Real, A.

PB2599. Genetic control of mRNA splicing as a potential driver for incomplete penetrance of coding variants. Einson, J.

PB2601. Genetic determination of fetal hemoglobin levels in patients with sickle cell disease. Ojewunmi, O.

PB2603. Genetic inactivation of zinc transporter SLC39A5 improves liver function and hyperglycemia in obesogenic settings. Chim, S.

PB2605. Genetic regulation of splicing in macular and peripheral retina reveals novel molecular mechanisms for age-related macular degeneration. Mehta, P.

PB2607. Genome-First Approach to Explore Prevalence and Cancer Risk of Adults with Pathogenic and Likely Pathogenic Variants in RASopathy Genes. Astiazaran Symonds, E.

PB2609. Genome-wide characterization of selective constraint on variation within CTCF Binding Sites. Tubbs, C.

PB2611. Genome-wide pleiotropy study identifies association of *PDGFB* with Age-Related Macular Degeneration and COVID-19 infection outcomes. Sun, X.

PB2613. Genomic characterization of the immunoglobulin light chain lambda locus from individuals of European, Asian and African origin. Gibson, W.

PB2615. Hematopoietic Loss of Y Chromosome Leads to Cardiac Fibrosis and Heart Failure Mortality. Forsberg, L.

PB2617. Human pancreatic islet microRNAs implicated in diabetes and related traits by large-scale genetic analysis. Taylor, H.

PB2619. Identification of bidirectional regulatory regions for *FADS1* and *FADS2.* Yang, S.

PB2623. Identifying context-specific genetically regulated expression changes in monocytes associated with resilience to Alzheimer's Disease. Mustafa, Y.

PB2625. Impact of rare heterozygous mutations of *PCSK1* on obesity: implication for treatment with MC4R agonists. Baron, M.

PB2628. Integrated analysis of retinal eQTLs, meQTLs and eQTMs identifies target genes and epigenetic mechanisms contributing to age-related macular degeneration. Advani, J.

PB2629. Integrative 3D structural analysis of *de novo* missense variants and their associated protein structures inferred by AlphaFold2 identifies the importance of β-strand/sheet features for congenital heart disease. Xie, Y.

PB2631. Investigating the molecular and cellular effects of pathogenic variants of *GNAI1* in developmental and epileptic encephalopathy. Fathi, E.

PB2633. KDM5A, a chromatin remodeler and newly identified autism gene, has a cell type-specific function in the hippocampus. El Hayek, L.

PB2635. Large-scale integrative analysis of 1.5 million single cells across diverse human tissues and immune cell types reveals dynamic regulatory effects on *HLA* gene expression. Kang, J.

PB2637. Limited overlap of eQTLs and GWAS hits due to systematic differences in discovery. Mostafavi, H.

PB2640. Massively parallel reporter assays with multi-layer annotations identified cell-type-specific functional variants and genes associated with melanoma. Long, E.

PB2642. Maternally Inherited novel *SUMO2* variant associated with a complex phenotype. Otohinoyi, D.

PB2644. Mendelian randomization reveals the causal interaction of *MICA* transcription levels with the risk of Graves' disease. Sutoh, Y.

PB2646. Mimicking NDD phenotypes and functional characterization of *PPP1R9A*$^{+/-}$ mutation using iPSCs- derived neurons and single-cell transcriptomics. Uddin, M.

PB2648. Mitochondrial Complex III (CIII) deficiency case study: Analysis of a likely pathogenic variant in *UQCRC2* gene as the etiology of intellectual disabilities and obesity. Arroyo Figueroa, G.

PB2649. Mitotic recombination is a common mechanism of cellular mosaicism in Fanconi anemia. Donovan, F.

PB2651. Modeling the effects of *KCNQ1* SNPs on type 2 diabetes risk in iPSC-derived pancreatic islets identifies an early role of methylation that alters islet composition and beta-cell mass. Nair, A.

PB2653. Multiomics identifies dysregulation of Wnt-signaling through epigenetic gene regulation across tissues in Bohring-Opitz Syndrome. Lin, I.

PB2655. Mutational scanning to determine pathogenicity of Variants of Uncertain Significance in genes in the Sonic Hedgehog Pathway. Baldridge, D.

PB2657. Natural variants in *SEL1L* modify lethality, ERAD, and proteasome function in a model of NGLY1 deficiency. Tu'ifua, T.

PB2660. Novel non-coding variant in NMNAT1 reduces the transcription of NMNAT1, which leads to Leber Congenital amaurosis. Han, J.

PB2662. Novel splice variant in PRUNE1 leads to aberrant neurodevelopment and neurodegeneration. Srivastava, A.

PB2664. Organ-specific prioritization of non-coding regulatory variants with stacking generalization. Zhao, N.

PB2666. Pathogenic mechanisms associated with a recurrent *CUX2* missense variant in epilepsy. Cheng, J.

PB2668. Patient-derived iPSC-CMs from TANGO2-deficient disorder revealed multi-channel defects, functional association between ERK1/2 and TANGO2, and folate as a potential therapeutic agent to mitigate arrhythmias in patients. Xu, W.

PB2669. Phenotypic and functional genomic impact of complex structural variation at the 17q21.31 locus. Zhang, P.

PB2671. Pooled RNA-IP approach to investigate variant effects on RBP binding and splicing. Schertzer, M.

PB2673. Population-specific non-coding and coding putative causal variants shape quantitative traits. Liu, X.

PB2675. Possible complex Di-Genetic inheritance in Primary Congenital Glaucoma. Bercovich, D.

PB2677. Predicting functional and fitness effect of missense variants. Zhao, Y.

PB2679. Quantifying negative selection in human 3'UTRs uncovers deleterious non-coding genetic variation. Findlay, S.

PB2681. Regulation of stress granules by VCP in health and genetic degenerative diseases. Helton, N.

PB2684. Scalable mQTL analysis of biochemical pathways with the UK Biobank using REVEAL:Biobank. Pitluk, Z.

PB2686. SEPT-GD: a decision tree to prioritise potential RNA splice variants in cardiomyopathy genes for functional splicing assays in diagnostics. Alimohamed, M.

PB2690. Single-nucleus transcriptome analysis of 424 aging brains identifies cell type and subtype specific *cis*-eQTL and their relation to neurodegenerative disease susceptibility. Fujita, M.

PB2692. Spectrum of pathogenic ATM variants in Indian ataxia telangiectasia patients. Shukla, R.

PB2693. Structural and functional characterization of missense variants in*NPR2*augments genetic associations, phenotypic prediction, and mechanistic understanding of skeletal development. Covarrubias, S.

PB2694. Systematic analysis of the impact of short tandem repeats on gene expression. Benton, S.

PB2696. Testing the association of Grantham score with functional impact for clinical variant interpretation. Owens, K.

PB2698. The contribution of short tandem repeats to splicing variation in humans. Li, Y.

PB2700. The genetic architectures of gene expression in individuals of African and European ancestry: results and consequences of eQTL studies. Fletez-Brant, K.

PB2702. The importance of the use of genomics in early diagnosis of tuberous sclerosis: case report. Lambraño, A.

PB2704. The role of introgressed archaic DNA in the human genome. Young, R.

PB2706. *TP53* codon 72 polymorphism modulates macrophage polarization through altered PI3K/Akt signaling pathways. Silwal, A.

PB2708. TxEWAS identifies genetic modifiers of drug response and side effects through retrospective gene-environment association studies. Sadowski, M.

PB2710. UBA5-related Epilepsy: From Cellular Models to Novel Therapies. Chen, H.

PB2712. Unbiased linkage of SNV to expression in single cells for regulatory variant discovery influencing cancer drug resistance. Salas-Gonzalez, I.

PB2715. Use of reference population data for resolving variants of uncertain significance in hematopoietic genes. Avramovic, V.

PB2719. Variation of 5'untranslated regions by dosage sensitivity reveals differences in post-transcriptional regulation important for interpreting variant effect. Wieder, N.

Omics Technologies Posters - Thursday (Poster)

Conv Ctr/Exhibit/Poster Hall/South Building

PB2867. A Cloud-scalable Software Suite for Large-Scale Proteogenomics Data Analysis and Visualization. Gajadhar, A.

PB2869. A comparison of the sensitivity and diagnostic rate of long-read vs. short-read whole genome sequencing in critically ill infants in the SeqFirst Project. Miller, D.

PB2871. A deep learning framework for structural variant discovery and genotyping. Popic, V.

PB2873. A demonstrated workflow to unleash the multiomic potential of a single blood draw. Nui, B.

PB2875. A High Throughput Dual Nucleic Acid Extraction Method Enables Whole Genome Sequencing from PAXGene Blood RNA Tubes. Alba, C.

PB2877. A Machine Learning-based approach to extract the gene-disease association discovery information from OMIM. Rahit, K.

PB2879. A method for ABO genotyping by Sanger DNA sequencing. Schreiber, E.

PB2881. A new approach to assess the allele frequency of small insertions and deletions. Milo Rasouly, H.

PB2883. A new method for detecting genes differentiated in expression variance using single-cell RNA sequencing data. Chen, M.

PB2885. A novel Bayesian factor analysis method improves detection of genes and biological processes affected by perturbations in single-cell CRISPR screening. Zhou, Y.

PB2887. A platform for high-resolution morphology analysis reveals tumor heterogeneity and enables label-free enrichment of target cell subpopulations. Jovic, A.

PB2891. A spatial map of neurodevelopmental disorder risk in the developing human cortex. Segato Dezem, F.

PB2893. A systems biology framework to evaluate the contribution of cellular crosstalk in Alzheimer's disease genetic risk and other psychiatric traits. D'Oliveira Albanus, R.

PB2895. A unified computing environment for genomics data storage, management, and analysis: NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-Space (AnVIL). Mosher, S.

PB2897. Age-dependent accumulation of somatic mutation and cell fusion in human cardiomyocytes revealed by single-cell whole-genome sequencing. Huang, Y.

PB2899. An Algorithm for Sequence Location Approximation using Nuclear Families (ASLAN) Validates Regions of the Telomere-to-Telomere Assembly and Identifies New Hotspots for Genetic Diversity. Chrisman, B.

PB2901. An empirical comparison of calling pipelines for whole genome sequencing demonstrates the importance of mapping and alignment. Ziegler, A.

PB2903. Analysis of alternative polyadenylation from long-read or short-read RNA-seq with LAPA. Celik, M.

PB2905. Application of a novel scRNA-seq method based on Pre-templated Instant Partitions (PIPseq) to evaluate therapeutic induced intestinal epithelium regeneration. Hettige, P.

PB2907. ASCoT - Application for Sanger COnfirmatory Testing - A graphical software interface for management of confirmatory sequencing in genomic medicine. Smith, G.

PB2909. Assessing the utility of genomic deep learning models for disease-relevant variant effect prediction. Kathail, P.

PB2911. Associations between genetically predicted levels of blood metabolites and pancreatic cancer risk, Zhong, H.

PB2912. Autoencoder-based Model for Genotype Imputation. Song, M.

PB2914. Automation of single-cell NGS library preparation for Tapestri platform to accelerate oncology discoveries. Barner, A.

PB2916. Bayesian Causal Inference Method applied to CRISPR perturbations Estimates the Causal Gene Regulatory Network of CD4+ T cells. Weinstock, J.

PB2918. Benchmarking NGS Integration Site Analysis Methods in Support of Long-Term Safety Monitoring of Cell and Gene Therapy Products. Otto, J.

PB2920. Benchmarking splice variant predictive algorithms with saturation screens. Smith, C.

PB2922. Biallelic CORO1A Mutation in a Patient with Late-onset Cutaneous Tuberculosis, Epidermodysplasia Verruciformis, and StaphylococcusAureus Infection. Vahidnezhad, H.

PB2924. Brain transcriptomic profiling of Parkinson´s disease patients reveals disease stage specific gene expression patterns. Cappelletti, C.

PB2926. Building a transcriptomic atlas of adult human ovarian tissue to gain insight into folliculogenesis. Hannum, D.

PB2928. Calling structural variants in extended rat pedigree using PacBio HiFi sequencing. Chen, D.

PB2930. Causal relationship analysis of DNA openness and RNA expression at the single cell level using scRNA-seq and scATAC-seq data. Park, J.

PB2932. Cell manufacturing genome integrity analysis by optical genome mapping. Pang, A.

PB2934. Characterizing the immune dysfunction in X-CGD using single-cell transcriptomics. Muzumdar, S.

PB2936. Clinical genome and RNA sequencing with a novel, cheaper sequencing-by-synthesis technology. Liu, P.

PB2938. Closing the gap: Solving complex medically relevant regions of the human Genome. Sedlazeck, F.

PB2940. Combination of low-coverage whole genome and deep-coverage exome sequencing is the cost-effective way to drive large-scale genetics studies forward. Yu, M.

PB2942. Community detection analysis in multilayer COVID-19 patient similarity networks. Sliwa, P.

PB2944. Comparison of genotyping arrays and low-pass sequencing for predictive genomics research. Gollub, J.

PB2946. Comprehensive characterization of structural variants in human brain genomes using single flow cell nanopore sequencing protocol. Kolmogorov, M.

PB2948. Comprehensive discovery of CRISPR-targeted sequences in the human gut metagenome. Sugimoto, R.

PB2950. CRISPR Streamline: Creating a Pipeline That Evaluates Phenotype Information From Off-Target Sequence Identification. Anderson, P.

PB2952. Custom morphology markers allow better tissue stratification to study tumor heterogeneity using NanoString® GeoMx® Cancer Transcriptome Atlas. Runyon, J.

PB2954. *De novo* variant detection with HiFi reads. Lake, J.

PB2957. Deep learning approaches for predicting virus integration sites in the host genomes. Zhao, Z.

PB2959. DeepCell, A novel cell type specific marker detection machine learning tool for single cell transcriptome. Ali, A.

PB2961. *DeepLoop* robustly maps chromatin interactions from sparse allele-resolved or single-cell Hi-C data at kilobase resolution . Zhang, S.

PB2963. Delivering Comprehensive, Single-molecule Proteomics Using Protein Identification by Short-epitope Mapping (PrISM). Kapp, G.

PB2964. Depth normalization for single-cell genomics count data. Booeshaghi, S.

PB2966. Detection of 5-hydroxymethylcytosine using a modified EM-seq protocol. Evanich, D.

PB2968. Developing local compute capacity - a Ugandan experience Integrated Biorepository of H3Africa Uganda, Makerere University College of Health sciences. Kezimbira, D.

PB2970. Diagnosis of neurodevelopmental disorders by RNA-Seq : From urine derived stem cells to induced neural stem cells, a promising alternative tissue. Riquin, K.

PB2972. Direct haplotype-resolved *5-base* HiFi genome sequencing allows for linking rare disease variants to non-coding function. Pastinen, T.

PB2973. Magnified Convolutional Enrichment Representation Model. Chen, G.

PB2974. Efficiency of mutagenesis promoted by CRISPR-Cas9 in *THRAB* gene in zebrafish. Rodrigues, L.

PB2976. Empowering Discovery in Childhood Cancer: Genomic Harmonization at the Kids First Data Resource Center. Miller, D.

PB2980. Establishing a baseline transcriptomic profile of human eccrine sweat glands via single-cell RNA-sequencing with validation by 3D imaging. Eastman, A.

PB2982. Estimating and testing cell-type-specific gene co-expression from single-cell data: A robust and efficient method that mitigates technical confounding. Su, C.

PB2984. Evaluation of cell type annotation for peripheral blood mononuclear cells using single-cell RNA versus a deconvolution approach with bulk RNA sequencing. Liu, X.

PB2986. Evercode™ Whole Transcriptome Single-Cell RNA-Seq with Focal Barcoding Enables Single-Cell Transcriptomes Coupled with Gene or Vector-Enriched Readouts. Pangallo, J.

PB2988. Exome technology innovations advancing personalized medicine. Walker, K.

PB2990. Extracellular protein monitoring in the ResolveOME genomic and transcriptomic dual workflow to uncover cancer pathology mechanisms in single cells. Morozova, T.

PB2992. Fast and gentle microfluidic cell sorting upstream of single cell transcriptomics. Ciarlo, M.

PB2994. FedExosome: mitochondrial DNA & microRNA profiling of neuronal-derived packages in Alzheimer's disease. Hall, C.

PB2996. Functional characterisation of the amyotrophic lateral sclerosis risk locus *GPX3/TNIP1.* Garton, F.

PB2998. Generating long-range sequencing information without long-read sequencing. Chen, Z.

PB3002. *Genetics of Cardiovascular diseases: Mathematical Modeling & Development of a Predicting Simulator Prediction of the heart attack, the treatment to prescribe and the risk of secondary complications.* Hassani Idrissi, H.

PB3004. *Genome-wide prediction of pathogenic gain- and loss-of-function variants from ensemble learning of diverse feature set.* Stein, D.

PB3006. Genotyping known disease-associated short tandem repeats in 7,000 Project MinE genomes reveals their relation to ALS and assists in disease-association analysis of unknown short tandem repeats. van Vugt, J.

PB3008. GTAGMe-seq reveals the genome-wide long-range coordinated cis-regulatory elements at the single-molecule level. Liu, Y.

PB3010. High accuracy Sequencing By Binding (SBB) improves human germline variant calling. Nasko, D.

PB3012. High throughput workflow for human whole genome sequencing using PacBio HiFi. Rocha, J.

PB3014. High-throughput metabolomics enabling discovery of tens of thousands of small molecule biomarkers that link genetics and disease. Long, T.

PB3016. High-throughput RNA sequencing directly from cell lysates enables reproducible phenotypic profiling for CRISPR treatment phenotyping and cell response screening applications. O'Hara, A.

PB3019. Horizon - an ultrarapid platform for NICU/PICU whole genome clinical pathogenicity prediction. Salih, G.

PB3021. Human microglia transcriptional changes associated with progression and development of Alzheimer's Disease. Kosoy, R.

PB3023. Identification of novel long non-coding RNA with distinct expression patterns in different multiple myeloma subtypes. Bauer, M.

PB3025. Illuminating Dark Proteins using Reactome Pathways. Matthews, L.

PB3027. Improved detection of functionally relevant aberrant splicing using the intronic Jaccard index. Mertes, C.

PB3028. Improved sequence mapping using a complete reference genome and lift-over. Chen, N.

PB3030. Improving sensitivity of combinatorial barcoding-based single cell sequencing. Schroeder, S.

PB3032. Imputing the whole metagenomic shotgun sequencing data using the 16s amplicon sequencing data. Jang, S.

PB3034. Increasing throughput and decreasing cost for single-cell profiling using combinatorial indexing. Nakamoto, M.

PB3036. Individualized cellular ancestry: efficient reconstruction of cell lineage trees. Jang, Y.

PB3038. Infection with SARS-CoV-2 leads to variant-specific changes in gene expression in airway cell lines and primary cell cultures. Rustagi, A.

PB3040. Insights from HLA transcriptome analysis of a cohort from the Qatar Genome Program. Fadda, A.

PB3042. Integrated genetic analysis of transcriptome sequencing data in congenital diaphragmatic hernia. Qiao, L.

PB3044. Integrated Spatial Transcriptomic and Proteomic Analysis of Fresh Frozen Tissue Based on Stereo-seq. Jiang, Y.

PB3046. Integrating multiple human toxicogenomics datasets for drug repositioning. Leclercq, M.

PB3048. Integrative computational analysis of long-read transcriptomes identifies alternatively spliced poison exons in iPSC-derived brain organoids. Broad, M.

PB3050. Integrative single-nucleus multi-omics analysis identified candidate regulatory elements and variants and their target genes in Alzheimer's disease brains. Chiba-Falek, O.

PB3052. Investigating somatic mutation rates and signatures across the length of the colon with duplex sequencing. Hiatt, L.

PB3054. Is single nucleus ATAC-seq accessibility a qualitative or quantitative trait?. Miao, Z.

PB3056. Large-scale single-cell RNA-sequencing in Japanese identified biological and host genetic involvement of innate immune cells in COVID-19 severity. Edahiro, R.

PB3058. Leveraging a multi-'omics Parkinson's Disease dataset to understand the role of mitochondrial transcriptional control in the aetiology of Parkinson's Disease. Fairbrother, A.

PB3060. Long read transcriptomes identify features not found with very deep short read sequencing. Salas-Morris, T.

PB3062. Long-read capture with Twist target enrichment system. Han, T.

PB3064. Long-read RNASeq in human brains aligned to T2T CHM13 complete human genome reveals new gene bodies and new transcripts, exons, & exon junctions in known genes. Aguzzoli Heberle, B.

PB3066. Low cost, noninvasive RNA-sequencing to enable massive scaling of transcriptome studies. Martorella, M.

PB3068. Machine learning approach embedding an alternative splicing as a strategy for a multi omics integration. Lee, Y.

PB3070. Making population-scale toxicogenomic analysis performant and cost-effective using REVEAL Biobank. Sarangi, S.

PB3071. Massively parallel reporter perturbation assays uncover temporal regulatory architecture during neural differentiation. Kreimer, A.

PB3073. Metabolomic analysis of vitreous humor from the eyes with different uveal melanoma prognostic subtypes defined by tumor gene expression profiling. Demirci, F.

PB3075. Methods for screening candidate causal regulatory variants in primary immune cells by CRISPR ribonucleoprotein. Lorenzini, M.

PB3077. MiRNA regulates potential genetic pathways involved in diabetic retinopathy: Implication for an early disease risk prediction Vishwakarma, S.

PB3080. More bang for your buck! Universal Sequence Detection: A novel full spectrum variant genotyper for NGS data. Van Den Akker, J.

PB3082. Multi-omics integration via similarity network fusion to detect subtypes of aging   . Yang, M.

PB3083. Multiset correlation and factor analysis enables exploration of multi-omic data. Brown, B.

PB3085. New generative deep models to discover novel disease-gene associations in large-scale genomic cohorts. Orenbuch, R.

PB3087. Novel Principal Component Analysis reveals rich gene expression contexts from snRNA-seq within and across cell-types. Carver, S.

PB3089. NTSM: Fast sample swapping detection and ancestry estimation on unprocessed heterogeneous raw sequencing data. Chu, J.

PB3091. On statistical modeling of single-cell RNA-seq data accounting for technological and biological dropouts. Mukhopadhyay, I.

PB3093. OrphaID: a new platform for rare intellectual disabilities in Orphanet in partnership with ERN-ITHACA. Amin, M.

PB3095. Overcoming FFPE hurdles to enable high quality hybrid capture libraries and somatic mutation detection in matched tumor-normal patient samples. Chavadi, S.

PB3097. Paired whole genome and whole transcriptome sequencing of a large cohort of undiagnosed pediatric trios with neurological phenotypes. Wang, R.

PB3099. Peripheral leukocyte transcriptome dynamics following ischemic stroke of large vessel and cardioembolic etiologies. Carmona-Mora, P.

PB3101. Peruvian Hospital&ltUrban Antibiotic Resistance found in Local Wastewater. Jaramillo Valverde, L.

PB3103. Phenome-wide gene prioritisation leveraging Knowledge Graphs, Graph Convolutional Networks and UK Biobank PheWAS. Vitsios, D.

PB3105. Population diversity and selection of recent gene duplications detected using a complete human genome sequence. Soto, D.

PB3107. Predicting the transcriptional activity and mechanism of action of small molecules using deep learning. Barnhill, C.

PB3109. Proteoform inference using proteogenomic approach in non-small cell lung cancer and healthy control plasma proteomes reveals disease-associated protein isoforms. Donovan, M.

PB3110. Proteogenomic approaches improve genetic discovery across diverse human diseases. Koprulu, M.

PB3112. Pseudogenes limit the identification of common, functionally important transcripts generated by their parent genes. Gustavsson, E.

PB3114. Pyro-Velocity: Probabilistic and scalable RNA velocity inference from single-cell data. Pinello, L.

PB3116. Quantifying regional DNA methylation improves detection of biologically relevant associations in Alzheimer's Disease. Eulalio, T.

PB3118. RedSpa: a method for Reference-based domain detection in multiple-sample Spatial transcriptomics studies. Ma, Y.

PB3120. Revealing the sequence of neutralizing antibodies produced during infection using VyCAP B cell screening equipment coupled to an optimized single cell RNA sequencing protocol. Rubben, K.

PB3122. rMATS-turbo: An efficient and flexible computational tool for alternative splicing analysis of large-scale RNA-seq data. Adams, J.

PB3124. Robust mapping of cell states from multiomics data using implicit feature selection. Hu, H.

PB3126. scNanoGPS enables high throughput single cell nanopore sequencing of same cell mega-omics in human tumors Shiau, C.

PB3128. SeeNV: a pipeline ready tool for the integration and visualization of CNVs and their callers. Bradshaw, M.

PB3130. Sentieon DNAscope: high accuracy small variant calling using machine learning. Gallagher, B.

PB3132. Sequencing By Binding (SBB) demonstrates superior performance in low-pass whole-human-sequencing applications. Chen, K.

PB3134. *SigAlign* (Similarity-guided Align): A new, high-performance generalized pairwise alignment algorithm using simplified and intuitive regularization criteria. Bahk, K.

PB3136. Simulating pathogenic and likely pathogenic variants for bioinformatics pipeline testing. Zeng, Q.

PB3138. Single cell dissection of Alzheimer's disease and related dementias. Berg, A.

PB3139. Single cell RNA-seq analysis of Bone Marrow-derived Osteoblasts. Dillard, L.

PB3141. Single-cell allele-specific expression analysis reveals dynamic and cell-type-specific regulatory effects. Qi, G.

PB3143. Single-cell genome-wide association reveals a nonsynonymous variant in *ERAP1* confers increased susceptibility to influenza virus. Schott, B.

PB3145. Single-cell RNA sequencing (scRNA-seq) of fresh, neuronal tissue using Pre-templated Instant Partitions (PIPseq) and comparison to alternative scRNAseq methods. Kugler, K.

PB3147. Single-nucleus profiling of the cerebellar cortex in essential tremor reveals Bergmann gliosis and oligodendrocyte myelin abnormalities as hallmarks of disease. Castonguay, C.

PB3149. SnapFISH: a computational pipeline to identify chromatin loops from DNA FISH data. Lee, L.

PB3150. Somatic genomic alterations in single neurons from brains with chronic traumatic encephalopathy (CTE). Ma, C.

PB3152. STOC: A Simple Tool Of Compression. Klein, R.

PB3154. Structural and copy number variant detection, filtering, annotation, and classification by optical genome mapping in constitutional disorders. Clifford, B.

PB3156. SVPred: An integrated framework for Structural Variant Discovery. Sarwal, V.

PB3158. Tagmented, Indexed, and Pooled followed by ChIP Sequencing (TIP-ChIP) to generate high-throughput multi-target ChIP-Seq Results. Ngoc Tran, S.

PB3160. Targeted sequencing of native telomeres reveals patterns of telomere length and subtelomere methylation at single chromosome resolution in human cells. Tyer, C.

PB3162. Tempo: an unsupervised Bayesian algorithm for circadian phase inference in single-cell transcriptomics. Auerbach, B.

PB3164. The impact of ageing on the transcriptome profile of human germline and somatic cells. Pham, M.

PB3166. Tissue specific transcript and protein isoforms in the human neural retina. Riepe, T.

PB3168. Transcriptome sequencing of RNA isolated from small volumes of blood stabilized in Tempus solution: a technical assessment of different extraction methods and DNase treatment. Tomei, S.

PB3170. Transcriptomic analysis of circulating endothelial colony-forming cells in patients with sickle cell anemia and ischemic stroke. Nicoliello Pereira de Castro, J.

PB3173. Ultra-sensitive TCR/BCR clonotyping and immunophenotyping. Chenchik, A.

PB3175. Untargeted metabolomics profiling in patients with and without epilepsy. Oja, K.

PB3177. Using combinatorial barcoding to simultaneously profile the transcriptome and immune repertoire of 1 million T cells. Papalexi, E.

PB3179. Using single nuclei multiomics and spatial transcriptomics to identify drivers of early pulmonary fibrosis. Vannan, A.

PB3181. Utilization of multi-omic, multiplexed cell atlases and benchmark analysis demonstrates purity of product in the manufacture of induced pluripotent stem cell derived natural killer cells. Denholtz, M.

PB3183. Utilizing REVEAL SingleCell for single cell spatial transcriptomics data storage, analysis, and visualization. Bragdon, C.

PB3185. WES, with its limitations, continues to provide an attractive mode for genomic data collection. Rockowitz, S.

PB3187. Why long-read sequencing is poised to become key to clinical genetics: an episodic ataxia study. Audet, S.

Pharmacogenomics Posters - Thursday (Poster)

PB2722. A systematic review and comparison of statistical methods for testing genotype-treatment interaction effects in trans-ethnic pharmacogenomics GWAS. Zhong, W.

PB2724. Allelic diversity of the pharmacogene *CYP2D6* in New Zealand Māori and Pacific peoples. Kennedy, M.

PB2726. Clinical genetic testing and reporting of homoplasmy or low-level heteroplasmy for *MT-RNR1* m.1555A>G conferring an increased risk for aminoglycoside induced ototoxicity and irreversible hearing loss. Cody, N.

PB2728. CRISPR enrichment of complex *CYP2D6-D7-D8* loci allows for accurate detection of *CYP2D6* structural variation and phased haplotyping. Turner, A.

PB2730. Dynamics in blood transcriptomic networks responding to TNF inhibitors in patients with rheumatoid arthritis. Yu, C.

PB2732. Estimating UK Biobank population-specific PGx allele and phenotype frequencies using PharmCAT. Li, B.

PB2734. Exploring polygenic risk scores as a biomarker of treatment response in systemic lupus erythematous. Esparza Gordillo, J.

PB2737. Impact of global and local ancestry on pharmacogenetics in African American persons with multiple sclerosis. Davis, M.

PB2739. Investigating pharmacogenetics of dupilumab in the treatment of atopic dermatitis in an Asian clinical cohort Yew, Y.

PB2741. Meta-analyses of genome wide association studies of toxicity to oxaliplatin and fluoropyrimidine chemotherapy in 1800 patients with advanced colorectal cancer. Watts, K.

PB2743. Pharmacogenetics of tenofovir clearance among Southern Africans living with HIV. Cindi, Z.

PB2745. Pharmacogenomics for All of Us: Approach to testing and return of results in 1M participants. Empey, P.

PB2747. *PTPN2* and Discontinuation of Azathioprine Attributed to Myelotoxicity. Dickson, A.

PB2748. Testing for common pharmacogenomic predictors of vigabatrin-associated visual field loss, under univariate & polygenic models. Boothman, I.

PB2751. Variant Calling and Orthogonal Verification of 20 Pharmacogenomics (PGx) Clinical Pharmacogenetics Implementation Consortium (CPIC) Level A Genes including 4 Challenging Genes Using Illumina DRAGEN Bio-IT Platform. Catreux, S.

PB2753. Whole Exome Sequencing adds power to prioritize drug targets when combined with other genetic data types. Ranganathan, S.

PB3634. Local ancestry stratified GWAS identifies pharmacogenomic variants associated with metformin glycemic response in African American individuals with type-2 diabetes. Xiao, S.

Prenatal, Perinatal, and Developmental Genetics Posters - Thursday (Poster)

Conv Ctr/Exhibit/Poster Hall/South Building

PB2064. A novel human mitochondrial donating cell type promotes maturation of GV oocytes. Lee, T.

PB2066. Assessing the magnitude of clinical utility of preconception expanded carrier screening for prevention of neurodevelopmental disorders. Boonsawat, P.

PB2068. Capacity Building: Exploring the Use of Newborn Screening Data to Understand Neurodevelopmental Outcomes. Talebizadeh, Z.

PB2070. Cell-informed atlas of gene, isoform, and splicing regulation in the developing human brain identifies candidate causal variants for neuropsychiatric GWAS. Wen, C.

PB2073. Comprehensive preimplantation genetic testing for monogenetic disorders (PGT-M) and aneuploidy (PGT-A) using whole genome sequencing and haplotyping. Chen, S.

PB2075. *De novo* mutations disturb early brain development more frequently than common variants in schizophrenia/ Itai, T.

PB2077. Developmental trajectory analysis of differentiating mouse sensory interneurons to further improve stem-cell differentiation protocols. Heinrichs, E.

PB2079. DNA damage and antioxidant capacity as prenatal markers of placental epigenetic aging clock. Tekola-Ayele, F.

PB2082. Exploring roles of distal enhancers in *SOX9* haploinsufficiency in sex determination. Ou, F.

PB2084. Gaucher disease: systematic review. Gómez, G.

PB2086. Genetic deconvolution of fetal and maternal cell-free DNA in maternal plasma enables next generation non-invasive prenatal screening. Zhang, J.

PB2088. Genome-wide analysis of preeclampsia identifies novel loci and loci previously associated with blood pressure traits. Greene, C.

PB2090. High-risk *APOL1* genotypes and infant growth restriction associations in Black women affected by preeclampsia. Durodoye, R.

PB2092. Human engineered heart tissues uncover*MYH7*alleles associated with a frail sarcomere and ventricular noncompaction. Monroe, T.

PB2094. Improving rare conditions diagnostic rates by standardizing practice and offering preclinical testing. Delot, E.

PB2096. Knowledge and attitude of pregnant women in the Kingdom of Saudi Arabia toward Noninvasive prenatal testing: A single center study. Akiel, M.

PB2098. Lethal genes and Mendelian disorders. Cacheiro, P.

PB2100. Metabolomic analysis reveals altered omega-6 fatty acid processing as a marker of *FMR1* premutation carriers with FXPOI compared to premutation carriers without ovarian insufficiency. Allen, E.

PB2101. Molecular dysregulation in *Sox2*-expressing pituitary stem cells lacking *Prop1.* Masser, B.

PB2103. Next-Generation Sequencing based pre-implantation genetic Testing in Nepal. Khanal, S.

PB2105. Non-invasive prenatal testing (NIPT): A reliable accurate prenatal non-invasive diagnosis setting in Nepal. Thapa, S.

PB2109. Phenomic and genomic analyses of endometriosis in the All of Us Research Program. Schlueter, D.

PB2111. Preeclampsia risk is influenced by apolipoprotein L1 genotype and micronutrient deficiencies. Bruner, W.

PB2113. Prenatal cytogenomic microarray analysis (CMA) reference model. Ghochani, M.

PB2115. Prenatal diagnostic testing in cases of cystic hygroma or increased nuchal translucency: A single center experience. Owen, N.

PB2117. Rapid testing of congenital diaphragmatic hernia novel variants using CRISPR/Cas9 mouse embryo editing. Bogenschutz, E.

PB2119. Recurrent constitutionnal chromosome 5 inversion. Doco-Fenzy, M.

PB2121. Sex-specific variation in the commonly shared mothers' milk and infant gut microbiota are associated with sex disparity in childhood asthma risk. Fang, Z.

PB2123. Sperm mosaicism predicts transmission of *de novo* mutations to human blastocysts. Breuss, M.

PB2125. Tensor decomposition on electronic health records identifies latent subtypes of preterm birth that associate with polygenic risk scores. Cruz Gonzalez, S.

PB2127. Understanding caudal developmental abnormalities using single-nucleus multi-omics data from wild type and Danforth's short tail mouse E9.5 tailbuds. Zajac, C.

PB2129. Utility of preconception carrier screening in couples with unexplained infertility. Yatsenko, S.

PB2131. Whole genome sequencing and analysis of 4,053 individuals in trios and mother-infant duos from the Born in Guangzhou Cohort Study. Huang, S.

Statistical Genetics and Genetic Epidemiology Posters - Thursday (Poster)

PB3259. A Cox proportional hazards whole genome sequencing study of ischemic stroke risk in a Brazilian sickle cell population. Earley, E.

PB3261. A fast variational inference approach for powerful genome-wide association of quantitative traits. Loya, H.

PB3263. A gene-based association test of interactions for maternal-fetal genotypes identifies genes associated with non-syndromic congenital heart defects. Huang, M.

PB3265. A genetic correlation disease-disease network (gcDDN) for the improved identification of novel phenotype relationships Sriram, V.

PB3267. A locus near *ICOS* modifies risk for *Mycobacterium avium* complex airway infection in people with cystic fibrosis. Gordon, W.

PB3269. A multi-omics risk score approach to identify individual risk for incident type 2 diabetes. Ng, H.

PB3272. A novel computational deconvolution method to estimate sample-level cell-type-specific gene expression profile. Kang, K.

PB3274. A novel haplotype based approach allows for conducting ancestry agnostic genome wide association studies. Perry, D.

PB3276. A novel method for multiple phenotype association studies based on genotype and phenotype network. Cao, X.

PB3278. A polygenic framework nominating causal SNPs, their target genes, and cell-types of action. Kim, A.

PB3280. A precise, combinatorial approach for the estimation of *de novo* mutation rate from short-read WGS data. Kohailan, M.

PB3282. A robust whole-genome Mendelian randomization approach for improved estimation and inference of causal effects. Zhang, H.

PB3284. A scalable pipeline for robust relatedness inference with application to a study of 1,144,542 individuals. Gaynor, S.

PB3286. A simple distance-based model predicts effector genes at GWAS loci. Seaton, D.

PB3288. A statistical framework for local genetic correlation across populations using GWAS summary data. Zhang, C.

PB3290. A two-stage mixed effects model to analyze longitudinal zero-inflated microbiome count data. Wang, J.

PB3291. A virtual transcriptome approach identifies drug repurposing candidates for Alzheimer's disease. Grabowska, M.

PB3293. Accurate and efficient estimation of local heritability using summary statistics and linkage disequilibrium. Li, H.

PB3295. Accurate variant calling of the camouflaged LPA KIV-2 VNTR in whole exome sequencing data. Schoenherr, S.

PB3296. Adjusting for genetic confounders leads to reliable detection of causal genes from transcriptome-wide association studies. Crouse, W.

PB3298. Allele frequency differences of causal variants explain the bulk of low portability of cross-ancestry PRS. Liu, X.

PB3300. Alpha-2A adrenergic receptor (*ADRA2A*) modulates susceptibility to Raynaud's disease. Tervi, A.

PB3302. An accelerated, massively scalable variant calling and filtering workflow for the detection of de novo mutations in whole genome sequencing data. Dawson, E.

PB3304. An allelic series rare variant association test for candidate gene discovery. McCaw, Z.

PB3306. An evaluation of ancestry specific polygenic risk scores (PRS) for uterine fibroids across populations. Piekos, J.

PB3308. An experimentally-based functional gene embedding. Brechtmann, F.

PB3310. Analysis of vitamin D receptor-binding variants in susceptibility to pediatric-onset multiple sclerosis. Yilmaz, D.

PB3312. Ancestry-Specific Polygenic Risk Scores for Telomere Length in TOPMed. Jin, J.

PB3314. Applying human phenomics to electronic health records provides a framework for understanding skin-aging related phenotypes. Abbasi, A.

PB3316. Assessing the value of non-coding variants in 500,000 whole-genome sequences in the discovery of gene-phenotype associations. Liu, J.

PB3319. Association of genetically predicted iron homeostasis biomarkers in type 2 diabetes and glycemic traits: A two-sample Mendelian Randomization study. Liang, Y.

PB3321. Asymmetric non-portability across populations of different ancestry. Ballard, J.

PB3323. Being overweight in late life can be a causal factor contributing to longevity: a Mendelian Randomization study. Duan, H.

PB3325. Boosting SNP discoverability: How genetic architecture impacts the power of multi-trait GWAS. Suzuki, Y.

PB3328. BTS: Bayesian Tissue Significance scores for functional evaluation of GWAS results. Kuksa, P.

PB3330. Burden testing of imputed rare variants to inform therapeutic hypotheses. Fuller, Z.

PB3332. Calibrated and adaptive prediction interval improves polygenic score (PGS) performance in diverse populations. Xu, Z.

PB3334. Causal inference for the relationship between transcription and DNA methylation in breast cancer. Karki, B.

PB3336. Characterizing copy number variations and their association with plasma proteins using short and long-read sequencing. Schmitz, D.

PB3340. Combining family history with a polygenic risk score enables highly effective risk stratification for glaucoma. Macgregor, S.

PB3342. COMPADRE: a robust, cloud-optimized genetic relatedness platform. Evans, G.

PB3344. Comparing the relationships of genetically proxied PCSK9 inhibition with mood disorders, cognition, and dementia between men and women: a drug-target Mendelian randomization study. Bell, A.

PB3346. Constructing and benchmarking omnibus polygenic risk scores using GWAS summary statistics. Zhao, Z.

PB3348. Copy-number variants as modulators of common disease susceptibility. Auwerx, C.

PB3350. Cross-trait meta-analysis reveals shared genetic architecture between PCOS and chronic inflammation markers. Brixi, G.

PB3353. Deep Representational Clustering for Genetic Endotype Discovery from Clinical Data. Averitt, A.

PB3355. DeepPerVar: a multimodal deep learning framework for functional interpretation of genetic variants in personal genome. Wang, Y.

PB3357. Defining longitudinal disease trajectories in 146,000 individuals with hypertension from Penn Medicine Electronic Health Records. Singhal, P.

PB3359. Detecting haplotype association using synthetic genetic variants. Guan, Y.

PB3361. Detecting Somatic Mosaicism at Tandem Repeats. Sehgal, A.

PB3363. *Detection of underdiagnosis of complex diseases due to underrecognition of high polygenic risk.* Marquez-Luna, C.

PB3365. Development and validation of an RNA-seq-based transcriptomic risk score for asthma. Mersha, T.

PB3367. Differentially Private Release of Genomic Summary Statistics Using Factorization Mechanisms</i>. Neel, S.

PB3369. Discovery of clinically relevant gene-phenotype associations from 454,712 whole exomes in the UK Biobank. Fiziev, P.

PB3371. Disentangling the common genetic architecture and causality of rheumatoid arthritis and systemic lupus erythematosus with COVID-19 outcomes: genome-wide cross trait analysis and bi-directional Mendelian randomization study. Zhao, J.

PB3373. Distinct explanations underlie gene-environment interactions in the UK Biobank. Durvasula, A.

PB3375. DNARecords: An extensible sparse format for petabyte scale genomics analysis. Mañas Mañas, A.

PB3377. DrFARM: Identification and inference for master regulator variants in multi-trait GWAS. Chan, L.

PB3378. Effect of type 2 diabetes and its genetic susceptibility on severity and mortality of COVID-19 in UK Biobank. Chung, W.

PB3379. Efficient multivariable Mendelian randomization for confounder adjustment using public GWAS databases. Morrison, J.

PB3381. Elucidate trans-ancestry cross trait genetic architecture for tobacco and alcohol use phenotypes using 3 million Individuals. Wang, X.

PB3383. Estimating indirect maternal genetic effects on children's autism spectrum disorder risk. Wu, Y.

PB3385. Evaluating differential expression of imputed miRNA expression in the subgenual anterior cingulate cortex. Drake, J.

PB3387. Evaluating strategies to infer continental ancestry using single-cell RNA-seq datasets: an application to the Human Cell Atlas. Yao, J.

PB3389. Evaluating the impact of parallel mutations on the accuracy of statistical phasing. Beck, A.

PB3391. Evaluation of germline variants in pediatric low grade glioma using non-negative matrix factorization. Chapman Hannah, L.

PB3393. Exploiting the mediating role of the metabolome to unravel transcript-to-phenotype associations. Porcu, E.

PB3395. Exploring the genetic, socio-economic and environmental determinants of drug adherence. Cordioli, M.

PB3397. Extending Genome-Wide Association Studies to admixed cohorts with high degrees of relatedness. Tan, T.

PB3399. Fine-mapping one chromosome at a time: Software and an initial application to UK Biobank. Yuan, K.

PB3401. Functional Annotations-Informed Whole Genome Sequence Analysis Identifies Novel Rare Variants for AD in the Alzheimer's Disease Sequencing Project. Lee, S.

PB3402. Genealogy-wide association improves detection of rare and low-frequency variants in under-sequenced populations. Gunnarsson, A.

PB3405. Genetic association models are robust to common population kinship estimation biases. Hou, Z.

PB3407. Genetic Determinants of Circulating Glycine and Cardiovascular Disease: Causal Relationship or Red Herring? Biswas, S.

PB3409. Genetic variants associated with serum IGF-1 levels in the Long Life Family Study. Minster, R.

PB3411. Genetically-guided phenotype imputation of partial or missing traits in biobank data increases power for genetic discovery. Eijsbouts, C.

PB3413. Genomes, exomes, and imputation: Comparing technologies for variant association and discovery in large-scale genetic studies. Joseph, T.

PB3415. Genome-wide association studies of brain imaging endophenotypes derived from unsupervised learning identifies genes relevant to brain structure. Xie, Z.

PB3417. Genome-wide association study of obstructive sleep apnea in the Million Veteran Program uncovers heterogeneity by sex and genetic ancestry. Kurniansyah, N.

PB3419. Genome-wide classification of epigenetic signal reveals regions of enriched heritability in complex immune traits. Stricker, M.

PB3421. Genome-wide study on 72,298 Korean individuals in Korean biobank data for 76 traits identifies hundreds of novel loci. Nam, K.

PB3424. GWAS meta-analysis of DXA-derived bone mineral density identifies 42 novel loci and uncovers biological pathways not previously identified in larger studies of ultrasound-derived bone mineral density. Frysz, M.

PB3426. GWAS of pericarditis derived from a natural language processing model on self-reported free text data identifies a genome-wide significant association on chromosome *2q14.1*. German, C.

PB3428. Hand grip strength and its underlying genetics is associated with widespread protein changes revealed by population-based plasma proteomics. Huang, Y.

PB3430. Hierarchical scanning strategy reveals signal regions in 260 genes implicated in regulation of gene expression on chromosome 19q. Zhang, X.

PB3432. Hybrid autoencoder for robust ancestry inference in the presence of data artifacts and relatedness. Yuan, M.

PB3434. Identification of age-related genetic and molecular determinants and their link to disease. Michalettou, T.

PB3436. Identification of rare genetic coding variants associated with Alzheimer's disease. Khurshid, Z.

PB3437. Identifying circulating proteins as biomarkers for age at menarche and age at natural menopause: insights from a Mendelian randomization study. Yazdanpanah, N.

PB3439. Identifying composite biomarkers in multi-omics datasets of osteoporosis for drug discovery. Alam, M.

PB3441. Identifying non-common variant sources of clinical comorbidities through integration of electronic health data and genetics for schizophrenia. Vessels, T.

PB3443. Identity-by-descent strategies uncover cryptic relatedness, deliver novel pedigrees, and facilitate gene discovery in amyotrophic lateral sclerosis. Williams, K.

PB3445. Impact of heterogeneity-by-ancestry on GWAS in admixed populations. Mester, R.

PB3447. Improved breast cancer risk stratification by integration of a cross-ancestry polygenic model with clinical risk factors. Tshiaba, P.

PB3450. Improved risk prediction using functionally calibrated polygenic risk scores in admixed populations. Wang, X.

PB3452. Improving Cross-Population Polygenic Risk Scores with a Tree-Guided Deep Learning Method. Layne, E.

PB3454. Improving the accuracy and interpretation of polygenic risk score through modeling the pathways of disease and multiple risk factors. Yang, Y.

PB3456. Imputation of heritable gene expression reveals transcriptomic associations with neuroimaging phenotypes. Hoang, N.

PB3458. Incorporating functional annotation with bilevel continuous shrinkage for polygenic risk prediction. Zhuang, Y.

PB3460. Incorporating related individuals in genome-wide association studies to reduce biases from familial effects and population stratification in Mendelian randomization. Jiang, W.

PB3462. Insights into the comorbidity between type 2 diabetes and osteoarthritis: a genetics view. Arruda, A.

PB3464. Integrating rare and common variant association data to improve therapeutic target discovery. LoGerfo, P.

PB3466. Integration of International Mouse Phenotype Consortium (IMPC) and UK Biobank data to identify genes associated with blood molecular phenotypes. Haseli Mashhadi, H.

PB3468. Interrogating causality of epidemiological risk factors on adolescent idiopathic scoliosis: a two-sample Mendelian randomization study. Manousaki, D.

PB3470. Investigating miRNA role in the neuropathology of Major Depression in a large postmortem brain sample. Taylor, Z.

PB3472. Investigating the impact of the rare pathogenic variants linked to Intellectual Disability genes on the cognitive ability in adults: exome analysis of the UK Biobank cohort. Kumar, A.

PB3474. Investigating the relationship between immune disease genetic risk and Alzheimer's Disease incidence. Clark, K.

PB3476. Investigations of the potential causal effects of the immune response to varicella-zoster virus on multiple health conditions: a Mendelian randomization phenome-wide association study. Yu, X.

PB3478. Isoform level transcriptome wide association study of prostate cancer risk reveals large number of transcript specific associations. Barman, P.

PB3480. Latent factor analysis reveals genetic components across GWAS traits. Omdahl, A.

PB3484. Leveraging Molecular-Diagnosis Workflow to Identify Modifier Genes in Diseases. Schmitz-Abe, K.

PB3486. Leveraging multiple traits to detect shared non-additive genetic variation in genome-wide association studies. Bass, A.

PB3488. Leveraging the use of meta-analysis summary statistics to improve gene expression prediction models. Mews, M.

PB3490. Linking the joint genetic structure of neuroanatomical phenotypes with psychiatric disorders. Auvergne, A.

PB3492. Loci for Cognitive Preservation in the Midwestern Amish. Main, L.

PB3494. Low-coverage sequencing imputation with 150,119 UK Biobank reference samples. Rubinacci, S.

PB3496. LT-Free: A novel method for leveraging family history in genetic association studies of arbitrarily complex diseases. Matthews, J.

PB3498. Machine learning approach for identifying diabetes nephropathy and non-diabetic chronic kidney disease cases and controls using urinary high-throughput transcriptomic data. Traglia, M.

PB3500. Machine learning based methods for predicting guide RNA effects on cell fitness and gene expression in CRISPR epigenomic experiments. Luo, T.

PB3502. Machine learning model discriminates false from true CNVs with a 98% accuracy. Renne, T.

PB3505. MaSk-LMM: a matrix sketching-based fast and scalable linear mixed model for association studies in large biobanks. Burch, M.

PB3507. mBAT-combo: a gene-based association test to decipher masking effects. Li, A.

PB3509. Mendelian randomization implicates GDF15 blood levels as a causal factor in inflammatory disease. Timmers, P.

PB3511. MENDEL-modified segregation analysis of 168 *TP53*-positive families estimates age-specific risks for cancer types beyond the established spectrum for Li-Fraumeni syndrome. Fortuno, C.

PB3513. Meta-imputation combining the East Asian and multi-ethnic reference panels enhanced the imputation performance of rare variants for East Asian population. Kim, Y.

PB3515. MethylPipeR: Multi-omic Prediction of Complex Traits. Cheng, Y.

PB3517. miRNA Metabolome - wide association study: A multi-omics integrative approach to Asthma. Sharma, R.

PB3519. MTClass: Identification and annotation of multi-tissue cis-eQTLs using machine learning. Li, R.

PB3521. Multi-ancestry and sex-specific phenome-wide association meta-analysis of genetically-predicted height. Marouli, E.

PB3523. MultiSuSiE: Multi-population fine-mapping under the sum of single effects model. Rossen, J.

PB3524. Nearest Neighbor Simulated Annealing Cohort Selection for Improved PCA Genome Matching. Laboulaye, R.

PB3526. New paradigm for uncovering the clinical consequences of genetic variation. Xiong, M.

PB3528. Next generation phenotyping of DEGCAGS syndrome. Freeman, R.

PB3530. Non-additive outlier effects of gene expression on anthropometric traits. Brown, A.

PB3532. Omics profiling in overweight and obese children in the California Bay Area during childhood development and the effects of a multi-modal health intervention program tailored to a majority Latinx community. Li-Pook-Than, J.

PB3534. On the polygenic trait model in population-based human genetics studies: What is random and what is fixed. Tang, Y.

PB3536. OTTERS: A powerful TWAS framework leveraging summary-level reference data. Dai, Q.

PB3538. Pathway analysis identifies novel non-synonymous variants contributing to extreme vascular outcomes in Williams-Beuren syndrome. Liu, D.

PB3540. Performance and accuracy of imputation panels for genetic association studies in sub-Saharan African populations. Sengupta, D.

PB3542. Phenome-wide Mendelian randomization study of plasma triglycerides and 2,600 disease traits. Park, J.

PB3544. Phenotypic and genetic relationships between physical activity, sleep, and brain health. Guo, Y.

PB3546. PheWAS-based clustering of Mendelian Randomization instruments reveals distinct mechanism-specific causal effects between obesity and educational attainment. Darrous, L.

PB3548. Polygenic prediction across populations is influenced by ancestry, genetic architecture, and methodology. Wang, Y.

PB3550. Polygenic scores enable discovery of widespread genetic interactions associated with quantitative traits in the UK Biobank. Ferreira, L.

PB3552. Polygenic transcriptome risk scores can translate genetic results between species. Sanchez-Roige, S.

PB3554. Population diversity in global reference genome cohorts. Skrahina, V.

PB3555. Power Window: a power-based sliding window method to identify the rare and novel variants underlying gene-based association signals. Bolze, A.

PB3558. Predicting ExWAS results from GWAS data: A shorter path to causal genes. Liang, K.

PB3560. Prediction of atrial fibrillation and stroke using machine learning models. Papadopoulou, A.

PB3562. Prioritization of causal genes from genome-wide association studies by Bayesian data integration across loci. Mousavi, Z.

PB3564. Protecting privacy in polygenic risk score calculation by homomorphic encryption. Kim, H.

PB3566. Publicly Available Privacy-preserving Benchmarks for Polygenic Prediction. Witteveen, M.

PB3568. Quantifying portable genetic effects and improving cross-ancestry genetic prediction with GWAS summary statistics. Miao, J.

PB3570. Quantifying the effects of high-dimensional cross-trait assortative mating on complex trait genetic architectures. Border, R.

PB3574. Rare-variant association studies: When are aggregation tests more powerful than single-variant tests? Bose, D.

PB3576. REGENIE v3: more efficient analysis of rare genetic variation with an extended set of gene-based tests. Mbatchou, J.

PB3578. sCLC: a Novel Statistical Method for Association Studies of Multiple Phenotypes and Genetic Variants based on GWAS Summary Statistics. Wang, M.

PB3580. Sensitive detection of within-species contamination from low-pass whole genome sequencing data. Liu, A.

PB3582. Sex-stratified vs. sex-combined analysis in the presence of genetic effect heterogeneity. Lin, B.

PB3584. Shared genetic etiology between Alzheimer's disease and stroke: A large scale genome-wide cross-trait analysis. Wang, Z.

PB3586. Short tandem repeat expansions are present in up to 25% of sporadic amyotrophic lateral sclerosis and frontotemporal dementia patients. Henden, L.

PB3588. Single cell RNA-seq data analysis of the Huntington's disease cerebellum using the interactive webserver, ICARUS. Jiang, A.

PB3590. Smoking-informed methylation QTLs in human nucleus accumbens. Carnes, M.

PB3592. ssCTPR: summary statistic based cross-trait penalized regression. Weidmann, M.

PB3593. Subcontinental Admixture in Individuals with European Ancestry and Implications for Genetic Epidemiology Studies. Gouveia, M.

PB3595. Supervised multiset sparse partial least squares discriminant analysis in multi-omics data integration. Su, K.

PB3597. The effect of CA and D vitamins on the course of COVID 19 from the plasma osmolality perspective in a Turkish Cohort. Ulgen, A.

PB3599. The effects of demographic-based selection bias on GWAS results in the UK Biobank. Galama, T.

PB3601. The impact of 22q11.2 copy number variants on human traits in the general population. Zamariolli, M.

PB3603. The impact of rare regulatory variation from epigenetics to protein. Li, T.

PB3605. The nature of nurture is not in the parents' genes: estimating indirect genetic effects from parents using imputed grandparental genotypes. Young, A.

PB3607. The role of sleep in human brain and heart health: an investigation using 40,000 brain and cardiac magnetic resonance images from the UK Biobank. Fan, Z.

PB3609. The use of population-based common controls in GWAS of infectious diseases can result in biased association signals. Duchen, D.

PB3611. Tools for efficient manipulation and meta-analysis of summary statistics from multi-exposure, genome-wide, gene-environment interaction studies. Westerman, K.

PB3613. Tradeoff between prediction accuracy and transferability in the design of polygenic risk scores. Dominguez, A.

PB3615. Transcriptome and GWAS identified disease-specific eosinophilia-pathways and their upstream regulators in primary biliary cholangitis. Ueno, K.

PB3618. Transformer generative adversarial networks and variational autoencoder for causal analysis of genetic variation in the presence of unobserved confounding. Zhao, J.

PB3619. TWAS of the X Chromosome in Neurodegeneration. Evans, P.

PB3621. Unpicking the Gordian knot: Mendelian randomisation identifies priority groups for prophylactic EBV vaccination. Muckian, M.

PB3623. Unsupervised representation learning significantly improves genomic discovery for lung function and respiratory disease prediction. Yun, T.

PB3625. Using genetic associations from shared clustered genetic predictors to infer causation. Batool, F.

PB3627. Validation of large-scale Hispanic S-PrediXcan findings by independent Hispanic RNA-Seq data analysis confirms novel lipid-associated genes. Petty, L.

PB3629. Whole-exome sequencing in 545,835 individuals implicates 27 genes in eosinophil biology. Sidore, C.

PB3631. Machine learning-based phenotyping significantly improves power for genetic discovery in NAFLD and NASH. Somineni, H.

PB3633. A unifying statistical framework to discover disease genes from GWAS. McManus, J.

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10X Genomics	1222
23andMe	1248
23andMe Careers	1249
3billion	913
Active Motif	807
Advanced Cell Diagnostics (ACD) / Bio-Techne	803
Agilent Technologies	1103
AiLife Diagnostics	1823
Allelica	1155
American Board of Medical Genetics and Genomics (ABMGG)	945
American College of Medical Genetics and Genomics (ACMG)	943
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Anchor Molecular Inc	507
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Variant Bio	519
VarSome	1719
VectorBuilder, Inc.	603
Vertex Pharmaceuticals, Inc. Careers	1334
Vizgen	1018
Washington University School of Medicine-St. Louis	937

Watchmaker Genomics	825
Zebrafish Genetics and Disease Models Core	855