



ANNUAL REPORT

2017

2018

DRIVING DISCOVERY

ADVANCING HEALTH

SERVING SOCIETY



American Society of Human Genetics



ANNUAL REPORT

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MESSAGE FROM THE PRESIDENT

Our community is driving new fundamental understanding of human genetics and delivering novel applications that influence every facet of life. Emerging uses range from preventive health to treating disease, tracing family ancestries, and understanding humanity's ancient origins. This knowledge is advancing society in countless ways—and also driving important questions on topics as diverse as privacy, law enforcement, equity, biomedical ethics, diversity and inclusion, and more.

As the world's largest community of human geneticists, the American Society of Human Genetics (ASHG) is dedicated to supporting this progress, addressing larger public policy issues, advancing public education and engagement, and supporting you: researchers and clinicians who are responsible for making and applying genetic discoveries and advances.

2017-2018 was eventful for the Society, with many achievements in longstanding valued programs. This Annual Report summarizes progress in each area of our mission, including ASHG's core scientific pillars to advance science and clinical applications – the Annual Meeting and *The American Journal of Human Genetics*. We'll also highlight other important activities, including successes in education and advocacy. Information about the Society's sound financial footing is also provided.

In service to this dynamic field, the Society is also in transition with new additions, some changes, and emerging possibilities. We have moved our headquarters to new office space and expanded our administrative independence; we welcomed a new *AJHG* editor in chief and a new ASHG executive director; completed the first member survey in some years; and have become more intentional in how we welcome and engage members into the life of the Society.

We've also initiated a strategic planning process. Our intent is to explore where our field is headed, the needs of members, and how ASHG may be best-positioned to help shape and support our community. Your input is essential to guide our thoughts as we prioritize programs that serve you, the field, and society most effectively. Therefore, the member survey and discussions at the Annual Meeting are vital, and we always benefit from your participation and feedback.

It has been a pleasure to be ASHG president. Thank you for this unexpected honor, which built on my wonderful experiences as a long-time ASHG member, *AJHG* editor, and volunteer. I am enthusiastic about ASHG's progress to date and its path ahead. That journey is rooted in our successes and experiences over 70 years, and thanks to our very strong present-day Board and committee leadership, I am confident that we will sustain the Society's impressive momentum.

We encourage you to join us as a member for 2019 — renewing your commitment to the field or perhaps joining us for the first time. Thank you!

Sincerely,

David L. Nelson, 2018 President

MEMBERSHIP AT A GLANCE



81
Countries



50
States



25%
Trainees

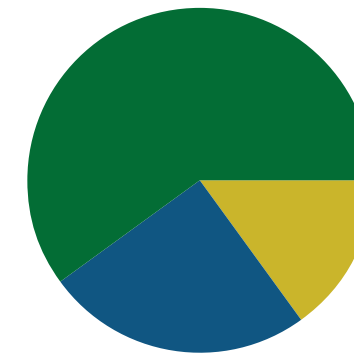


70%
Regular Members



5%
Emeritus Members

MAJOR TYPE OF WORK



RESEARCH
CLINICAL & COUNSELING
TEACHING & ADMINISTRATION

MEMBER ENGAGEMENT

91 Belong to a committee

360 Served as DNA Day essay judges

105 Reviewed 2018 meeting abstracts



2017 ANNUAL MEETING GENERATES GREAT SCIENCE & NETWORKING

The ASHG 2017 Annual Meeting in Orlando was a high point of the Society's work to advance scientific knowledge through learning, debate, and networking. In total, ASHG received more than 3,100 abstracts in 2017 and welcomed nearly 7,500 attendees and more than 220 exhibiting companies. Early indications suggest an even more vibrant meeting in San Diego in 2018.

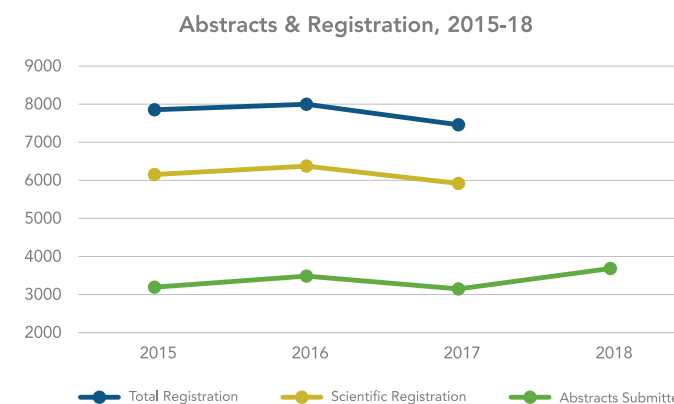
The 2017 meeting featured 12 high scoring abstracts in plenary sessions on topics such as gene expression, genetic factors associated with autism, and risk factors for breast cancer and melanoma; 360 platform sessions representing the meeting's cross-cutting science and diversity of topics; as well as 17 invited sessions organized through a competitive submission process on topics such as human brain evolution, single-cell genomics, complex traits, diversity in genomics research. Other highlights included the widely attended "Building Bridges" session with the European Society of Human Genetics, a joint symposium with the World Congress of Psychiatric Genetics on the genetics of neurodevelopmental disorders and other sessions that addressed genetics policy, communications training and education strategies. ASHG's Training and Development Committee also led a range of well received programming targeted to serve ASHG's earliest-career members.



The 2017 Presidential Symposium was especially engaging. President Nancy Cox welcomed Bill Gates, Chairman of the Bill & Melinda Gates Foundation, and NIH Director Francis Collins for a session exploring global health and its intersection with human genetics. Nancy, Bill, and Francis were joined by 2017 Program Committee Chair Peter Scacheri to discuss progress and challenges in applying growing genetic knowledge to address problems affecting millions around the globe. The event remains online for the community and public to watch.

As Nancy relayed in her opening remarks, "For decades, these two remarkable people have blazed new trails in science and technology. And through their own professional lives and personal passions, they have both been deeply committed to advancing science and serving society. Together, we'll discuss how new scientific understanding could transform how we approach infectious disease and genetic contributors to global health disparities and outcomes."

ASHG works hard each year to maintain the meeting's tremendous value to the field, as well as innovate to serve attendees even better. The meeting garnered strong satisfaction ratings from attendees, who also suggested areas for further development. While recognizing the meeting is fundamentally sound and successful, the Board tasked an ad hoc review group to explore how the Society could continue to enhance the meeting. The discussion included adding new ways to increase engagement and networking, which will be a focus in 2018 and beyond. Other new features in 2018 include an advocacy session as well as innovative "inspiration lounges," where attendees can talk informally with speakers before or after major talks, encouraging dialogue, interaction between generations, and learning opportunities.



AJHG: SHARING DISCOVERY ACROSS HUMAN GENETICS

The American Journal of Human Genetics is one of the field's leading journals. Known for its scientific excellence and rigor as well as swift time to decision, *The Journal* will enter 2019, its 70th year, with an exceptional reputation for scientific quality, breadth, and field leadership. Through its broad reach and impact, *AJHG* remains very well-positioned to chronicle—and help shape—the continued evolution of human genetics.

December 2017 was a time to say thank you and farewell to the long successful editorial leadership of David Nelson, and the ASHG Board of Directors thanked him for his service. In addition to his stewardship of the scientific content, one of David's notable passions—the rich history of the human genetics and the Society itself—was a hallmark of the *AJHG* covers during his tenure. He also highlighted that history in his final reflection as editor, in which he observed the many changes and progress points in the field over the last 65+ years and wondered what the next decades might hold.

January 2018 thus also saw the arrival of a new editor-in-chief: Bruce Korf. In an editorial, Bruce introduced himself and observed that *The Journal* possesses the unique ability “to grow along with one of the most rapidly evolving areas of scientific endeavor.” He further noted that he hoped to reinforce *The Journal* as broadly based, covering the full range of field content.” The commitment of *The Journal* to publish the best scientific research in human genetics, including population genetics, statistical genetics, molecular genetics,



BRUCE KORF, MD, PHD
EDITOR-IN-CHIEF, AJHG



THROUGH 2017, AJHG COVERS FEATURED THE PRESIDENTS OF ASHG TO HIGHLIGHT THE HISTORY OF THE FIELD AND SOCIETY.

genomics and gene discovery, and yes, clinical and translational studies, remains undiminished,” he noted. At the same time, he also made the case for papers to explore diagnostic approaches and reports on preclinical studies of potential treatments, clinical trials, cancer genetics, and even topics for policy debate and discussion.

Even with its continuing high regard as a scientific leader, *The Journal* is innovating to keep pace with changing needs in the field and forces in the scientific publishing environment. New types of content, including new feature content as well as a broad range of papers, will reinforce that innovation. In years to come, it will continue to evolve and serve the field successfully.

MOST CITED ARTICLES, 2017

ARTICLE TITLE	CITATIONS IN 2017
DNA Methylation in Newborns and Maternal Smoking in Pregnancy: Genome-wide Consortium Meta-analysis	74
The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities	62
Genotype Imputation with Millions of Reference Samples	44
Genomic Analyses Reveal Mutational Signatures and Frequently Altered Genes in Esophageal Squamous Cell Carcinoma	42
Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents	39
The Genetic History of African Americans, Latinos, and European Americans across the United States	37

EDUCATION: ENGAGING STUDENTS & THE PUBLIC, SUPPORTING HUMAN GENETICS PROFESSIONALS

Educating the diverse human genetics community, supporting trainees, and engaging students and the public are at the heart of ASHG's mission. In 2017 and 2018, the Society led many successful programs serving high school students, undergraduate students, and the larger public. It also began to explore how to best leverage its strengths to increase professional education for members.

ASHG's longest-standing, widest-reaching public education program is the DNA Day Essay Contest, which garnered extraordinary global engagement in 2018. Open to students in grades 9-12 worldwide, participants write a 750-word essay through which they examine, question, and reflect on important genetics concepts. Teachers play a central role in essay submission, and in many cases incorporate the contest into their curriculum. The 2018 contest prompted more than 1,000 submissions from students in 43 U.S. states and 23 countries. Following review by more than 300 dedicated ASHG members, outstanding essays from writers in New York, Los Angeles, and Kyiv, Ukraine were chosen. Additionally, ASHG is taking steps to increase its participation in the "DNA Day Campaign," under the leadership of the National Human Genome Research Institute, and led the successful annual workshop focused on enhancing teaching strategies for undergraduate educators.

ASHG 2018 DNA DAY ESSAY CONTEST APRIL 25, 2018

<p>1ST PLACE Diane Zhang</p> <p>Teacher: Ms. Stephanie Peborde School: Fox Lane High School Location: Bedford, NY</p>	<p>2ND PLACE Ilan Bocian</p> <p>Teacher: Mrs. Vickie Bellomo School: YULA-Boys Location: Los Angeles, CA</p>	<p>3RD PLACE Nadia O'Hara</p> <p>Teacher: Mrs. Audra Santos School: Pechersk School International Location: Kyiv, Ukraine</p>
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Read the winning essays at ashg.org/dnaday

THE 2018 DNA DAY ESSAY CONTEST RECEIVED OVER 1,000 ENTRIES FROM HIGH SCHOOL STUDENTS WORLDWIDE.

The Genomic Medicine Education Consortium, a multi-year effort of the Society, wrapped up in 2018 as an effective endeavor to explore strategies for educating non-genetics health professionals. With support from valued partners (see donors and sponsors section), the program made meaningful inroads to understand the complex challenge of improving genetic knowledge among non-genetics specialists, with content addressing pediatricians, oncologists, speech-language professionals, and others. While the Board elected to end the consortium in order to explore other emerging educational priorities, the initiative's resources will remain available on the website and through several project partners.

In the Society's effort to serve members effectively, ASHG's programs for trainees and the broader field gained traction this year. In partnership with The Jackson Laboratory, ASHG is in its second year of organizing "Conference to Career," a year-long program for as many as 130 trainees. Through a combination of in-person and online sessions, the program helps them make the most of the ASHG Annual Meeting to spark career directions. The Society also enabled nine trainees from underrepresented backgrounds to attend the ASHG Annual Meeting and hosted the trainee diversity breakfast. These activities reinforce the importance of diversity and inclusion to advance human genetics and help trainees network with one another and within the broader Society.

ASHG was also pleased to expand professional education, including a widely attended webinar with AJHG on the Matchmaker Exchange and new planned events with the National Society of Genetic Counselors to support professional development for counselors in research.



POLICY AND ADVOCACY: ADVANCING HUMAN GENETICS IN THE PUBLIC ARENA

ASHG is committed to building greater public support for human genetics and to advocating for policies that advance and protect genetic research. Over the last two years, the Society has been building its policy and advocacy impact in Washington and will be enhancing its voice on issues that affect our field and larger society. In summer 2018, ASHG launched its advocate's pledge to empower members to get involved in advocacy activities.

In the past year, ASHG has taken the lead celebrating an important policy milestone: 10 years of U.S. federal protections against genetic discrimination, a top priority for the Society. Specifically, the month of May marked 10 years since the signing into law of "GINA"—the Genetic Information Nondiscrimination Act. The anniversary sparked many ASHG activities, including a new video to explain the law's value for the public. The Society also spoke to the importance of diversity in genetic research, as illustrated in a February 2018 *Orlando Sentinel* op-ed by Past President Nancy Cox. ASHG led an international group of 11 organizations that issued a major statement on germline genome editing that garnered broad visibility. Other top policy priorities include funding for genetics research, the return of results to research participants, issues related to genetic testing, the interface of genomics research and genomic medicine, protecting genetic privacy, the teaching of evolution, and the genetics of race and ancestry.

ASHG POSITION STATEMENT

Human Germline Genome Editing

Kelly E. Ormond,^{1,19,*} Douglas P. Mortlock,^{2,19} Derek T. Scholes,^{3,19} Yvonne Bombard,⁴ Lawrence C. Brody,⁵ W. Andrew Faucett,^{6,7} Naniibaa' A. Garrison,^{8,9} Laura Hercher,^{7,10} Rosario Isasi,¹¹ Anna Middleton,^{12,13} Kiran Musunuru,¹⁴ Daniel Shriner,^{15,16} Alice Virani,^{17,18} and Caroline E. Young³

With CRISPR/Cas9 and other genome-editing technologies, successful somatic and germline genome editing are becoming feasible. To respond, an American Society of Human Genetics (ASHG) workgroup developed this position statement, which was approved by the ASHG Board in March 2017. The workgroup included representatives from the UK Association of Genetic Nurses and Counsellors, Canadian Association of Genetic Counsellors, International Genetic Epidemiology Society, and US National Society of Genetic Counsellors. These groups, as well as the American Society for Reproductive Medicine, Asia Pacific Society of Human Genetics, British Society for Genetic Medicine, Human Genetics Society of Australasia, Professional Society of Genetic Counsellors in Asia, and Southern African Society for Human Genetics, endorsed the final statement. The statement includes the following positions: (1) At this time, given the nature and number of unanswered scientific, ethical, and policy questions, it is inappropriate to perform germline gene editing that culminates in human pregnancy; (2) Currently, there is no reason to prohibit in vitro germline genome editing on human embryos and gametes, with appropriate oversight and consent from donors, to facilitate research on the possible future clinical applications of gene editing. There should be no prohibition on making public funds available to support this research. (3) Future clinical application of human germline genome editing should not proceed unless, at a minimum, there is (a) a compelling medical rationale, (b) an evidence base that supports its clinical use, (c) an ethical justification, and (d) a transparent public process to solicit and incorporate stakeholder input.

Introduction
The American Society of Human Genetics (ASHG) Workgroup on Human Germline Genome Editing developed the following position statement and explanatory paper between August 2015 and January 2017. This group, composed of a combination of basic and clinical scientists, bioethicists, health services researchers, lawyers, and genetic counselors, worked together to integrate the scientific status of and socio-ethical views toward human germline genome editing (defined as using genome-editing techniques in a human germ cell or embryo) into this statement. The group met regularly through a series of weekly conference calls and email discussions, proposed a draft statement to the ASHG Board of Directors in April 2016, presented the draft policy statement to ASHG and European Society of Human Genetics (ESHG) members at the ASHG-ESHG Building Bridges session in May 2016, and requested comments from ASHG members in June 2016. A total of 27 comments were received, 4 of which were in opposition to the statement. All comments and recommended modifications were reviewed by the explanatory paper, which was reviewed and approved by the ASHG Board of Directors in March 2017. The workgroup included representation from the following professional organizations (in alphabetical order), which then also approved the position statement and paper: the Association of Genetic Nurses and Counsellors, Canadian Association of Genetic Counsellors, International Genetic Epidemiology Society, and National Society of Genetic Counsellors. This resulting policy statement was then reviewed and endorsed by the following professional organizations (also listed in alphabetical order) before submission for publication: the American Society for Reproductive Medicine, Asia Pacific Society of Human Genetics (APSHG), British Society for Genetic Medicine, Human Genetics Society of Australasia, Professional Society of Genetic Counsellors in Asia, and Southern African Society for Human Genetics. (The APSHG would like to add a comment that we also express a concern that in some countries with inadequate ethics committee oversight or strong institutional review boards [IRBs], the potential for abuse exists. Hence, there is a strong need to

THE SOCIETY'S STATEMENT ON GERMLINE GENOME EDITING GARNERED BROAD VISIBILITY.

ASHG is also committed to supporting the scientific enterprise directly, as well as by joining forces with others to protect the vital role of science in society.

On the positive side, bipartisan Congressional support for medical research remains very strong, and ASHG is actively engaged with the broader advocacy community to push for sustained, robust increases in federal research funding. The final 2018 budget included a remarkable \$3 billion increase for NIH and 2019 negotiations featured sustained support for strong increases. This success reflects the impact



THE ASHG/NHGRI GENETICS & PUBLIC POLICY FELLOWSHIP PROVIDES GENETICISTS AN OPPORTUNITY TO CONTRIBUTE TO THE POLICYMAKING PROCESS.

of the united advocacy community in Washington that has worked over a decade to make the case that research improves health, advances knowledge, and strengthens economies.

Conversely, ASHG has been deeply concerned and active when it comes to actions that run counter to the Society's fundamental values. Following the announcement of the U.S. administration's travel ban, the Society announced its opposition strongly and clearly. We continue to monitor other issues of concern including challenges to the teaching of evolution, the misuse of genetics to

advance racial supremacy, and challenges to reproductive freedom.

Throughout, ASHG is increasingly engaged with the broader research advocacy community. This includes new membership in Research!America, a premier medical research advocacy organization, continued membership in the Federation of American Societies for Experimental Biology, and closer communication with the American Association for the Advancement of Science.

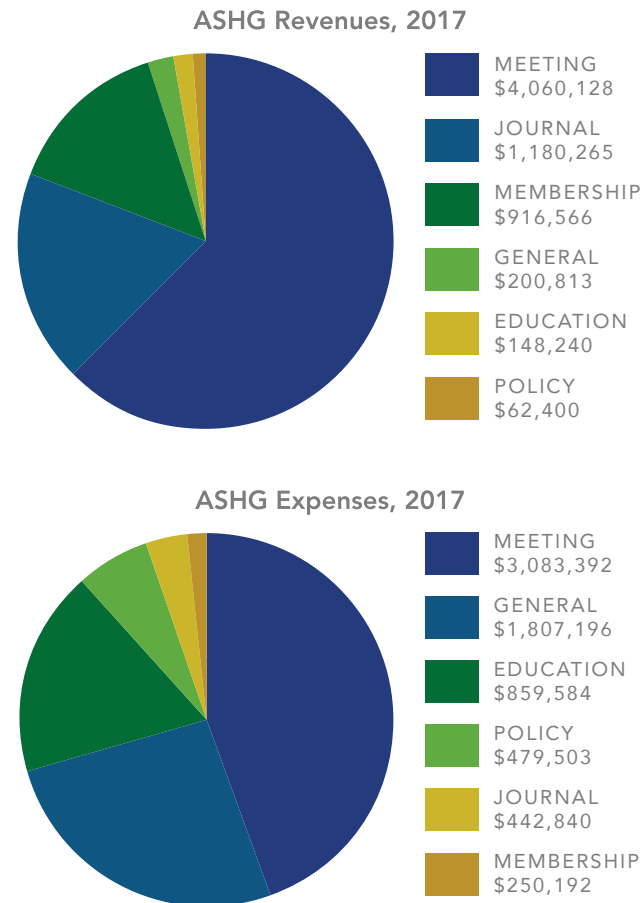
FINANCIAL HIGHLIGHTS

ASHG's financial health remains sound, with prudent management strategies that protect the Society and advance its goals. Moreover, thanks to long-time careful financial stewardship, ASHG has the capacity to look forward and begin planning for future investments that serve members, the Society, and the field.

CY2017 PERFORMANCE

Although ASHG has maintained net revenue surpluses over many years, 2017 presented a set of one-year challenges that resulted in a net operating loss of approximately \$354,000. Reduced revenue resulted primarily from lower-than-budgeted exhibit and registration revenue for the 2017 Annual Meeting, while expenses were higher than budgeted, due primarily to technology investments and the transition of the Society's executive staff position. Thanks to dedicated Board stewardship of the reserve over many years, the Society was able to absorb this one-year shortfall and the FY2019 budget anticipates a return to positive financial performance. ASHG's reserve fund was able to capitalize on strong financial markets, ending the June 30, 2018 period with a value totaling \$17,839,428.

CY2017 revenue and expenses were as follows. ASHG's CY2017 Audited Financial Statements are available online.



PRUDENT MANAGEMENT

The Society has also updated its financial management and systems to help it most effectively support Society operations and deploy resources, including changing its fiscal year from a calendar year to a July-June period. The transition will produce a single "short-year FY2018" (January-June 2018) that reflects only six months of revenue and expense. Beginning July 1, 2018, future fiscal years will again reflect full 12-month periods on the new cycle. Additionally, after many years of flat pricing for the Annual Meeting and membership, ASHG is modestly increasing rates to address inflation. In making these choices, the Society committed to reinforce the benefits of membership, including a decision to maintain the substantial member discount for Annual Meeting registration, which more than covers the cost of membership.

LOOKING FORWARD

Finally, the Society enabled limited, responsible deployment of reserves for highest-priority investments that advance the Society and the field. The Board authorized a limited use in January 2018, including sustained support for technology and infrastructure upgrades. That investment will help the Society grow, increase vital data security and integrity, and better serve members' growing technology expectations. Funds were also invested in a move to new offices in May 2018, following the end of ASHG's long-time lease on the campus of the Federation of American Societies for Experimental Biology.

Finally, the Board is also making additional investments in the future, including support for strategic planning. This began by making resources available to better understand the Society's membership composition and needs through the first member survey in some years and will continue with input from a range of stakeholders. Combined, this will help the Board prepare for a thoughtful strategic planning dialogue early in 2019. The goal will be to help chart the Society's next five years of growth, building on the solid foundation created by members and leaders over ASHG's 70 years of distinction and achievement.

2018 SOCIETY LEADERSHIP, EDITORS, AND STAFF

The American Society of Human Genetics gratefully recognizes the dedication of its volunteers, editors, and staff. The Society could not serve the field so effectively without your engagement and expertise. Thank you!

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As this report spans the years 2017-2018, a number of the activities began under the leadership of former Executive Director Joseph McInerney, who retired in July 2017. The Society recognizes and thanks Joe for his extraordinary service to the Society and his career-long dedication to advancing the field of human genetics.

2017-18 PARTNERS AND DONORS

Progress in human genetics will advance only through the active contributions and cooperation of many actors – researchers and clinicians, institutions, individuals private sector companies, federal agencies, advocacy partners, and fellow international organizations. We acknowledge and thank our program partners and donors for your support and active collaboration in 2018.



ASHG is grateful for the generosity of the individual donors listed below.

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| Delozier, Dawn | Hisama, Fuki | McGillivray, Barbara | Seguin-Spillman, Lana | Zang, Klaus |
| Demarchi, Mario | Hong, Yeon | Mendelsohn, Nancy | Sen, Askin | Zito, Antonino |
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