2019 Presidential Address: An Unexceptional President[1]

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This address encapsulates two distinct, but related themes. First, that it is the core objective of our Society to provide opportunities for a wide range of individuals—exceptional and not—to succeed in our field and second, that genetic exceptionalism must be extinguished for human genetics to succeed.

The first theme of exceptionalism is that this geneticist considers himself fundamentally unexceptional in our field. This raises a host of questions, not the least of which is, "How could such an unremarkable individual have the opportunity to serve as the president of this remarkable Society?" The answer to this is the Society itself—how ASHG fostered aspirations and opportunities in this unexceptional geneticist has much to do with what an extraordinary association the ASHG has been and what it should strive to be in the future.

My career began with puzzling patients who presented in the nursery and infant wards with poorly understood presentations of congenital anomaly syndromes. This was a yawning need in clinical medicine: to determine what was wrong with these children and what could be done to help them. Fortuitously, being married to a newly trained genetic counselor, I had the opportunity to attend the 1982 meeting of the ASHG as a fourth-year medical student guest attendee. The clear picture in my mind was that of us ascending the escalators to the center of the Renaissance Center in Detroit. As a fourth-year medical student, I was overwhelmed by the goings on of this meeting; nearly all of it was over my head and unintelligible. Yet, colleagues of my wife were generous, welcoming, and nearly instantly set about encouraging me to enter this field. That September, 27 years ago, set my course. In my genetics training at Michigan, the human genetics faculty were unrelenting supporters of ASHG as a venue to learn and grow as a geneticist on the national level. My engagement with the machinery of the Society began in the early 1990s with an opportunity to serve as a member of the ”rapid action task force on informed consent” that addressed the challenging issues surrounding consent for stored specimens. As the junior person on the working group, I was warmly welcomed into the committee and encouraged and supported to advance my views. This openness and support by the Society was just one of a string of opportunities I had to serve the Society, which included multiple years of abstract reviewing for the Program Committee, being a member of the Program Committee, chairing the Program Committee, serving on the *AJHG* Editorial Board, and serving as the chair of the Future of the Annual Meeting Working Group, as a member of the Board of Directors, and finally as President.

The key conclusion to take away from these opportunities to serve is that they were offers to do work to advance the mission of the Society; they were not honors granted for accomplishments. This is the key take home from my history with ASHG: the Society saw in me an able and willing worker to develop and execute the activities of the Society, which advanced the interests of all members. That I was an unexceptional geneticist was irrelevant. I belabor this point because I want to inspire all members to step up and offer their efforts to realize our shared goals. All of you can participate in this valuable and rewarding work.

Upon assuming the presidency and reflecting on how ASHG had stoked this unexceptional clinician's enthusiasm for genetics, I concluded that the highest priority of the Society should be to promote an optimistic and positive view of genetics and genomic science. This was

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[1] This article is based on the address given by the author at the meeting of the American Society of Human Genetics (ASHG) on October 15, 2019, in Houston, Texas. The audio of the original address can be found at the ASHG website.

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robustly discussed and enthusiastically endorsed by the Strategic Planning Group and the Board of Directors. The result of this was the new vision for ASHG: "People everywhere realize the benefits of human genetics and genomics research." Our goal in this vision statement is that every member can engage with this positive view of our field and that we can attract the best and the brightest trainees to enter and expand our field. As your advocate, the Society must first and foremost support you in your efforts to realize the benefits but never deny the real downsides and risks of genomics. This led to my views on genetic exceptionalism, which are discussed below.

Another lesson from my experience with the Society was that participation in the work of the Society is not only apparently beneficial to the Society, but also provides a vibrant and diverse network of colleagues and exposure to many facets of our field. To give all of our members as many of these opportunities as possible, we have developed a strategic goal of developing year-round programming, expanded committee membership, and developed a more transparent and democratic process for committee membership and chair appointments. Our strategic goal of year-round programming is responsive to member calls for increased opportunities to learn about cutting-edge scientific opportunities and to practice science with the highest standards of ethics and social benefit.

Equally important to year-round programming, we have a strategic goal to increase diversity in genetics and genomics research—we are firmly committed to increasing the representation of individuals of previously under-represented groups in our field and supporting their participation in Society activities through our code of conduct policy (see Web Resources) and targeted programming. We are equally committed to increasing diversity among our research participants—to generate data on genomic benefits for people everywhere. Your board has been inspired to make these changes and improve these opportunities so that every member of the Society (and potential future members) can grow and benefit from their membership in ASHG and that the public can benefit from our work.

All of these efforts were part of a very thoughtful, thorough, and broad-based effort at strategic planning initiated by 2018 President David Nelson and completed during my term. Ideally, every member will pull up the strategic plan, review it carefully, and build components of that plan into your practice of genetic and genomic medicine and science.

The second theme of this address is genetic exceptionalism. The historic origins of genetic exceptionalism were addressed in the symposium at the 2019 meeting, titled "Genetic Exceptionalism—From Its Beginning to Its End?" Without detracting from the forthcoming publication on the content of that symposium, "exceptionalism" is defined as "… a state of being special, exceptional, or unique" or "the theory or belief that something … does not conform to a pattern or norm." Exceptionalism thus sets a thing apart from all other things.

In our field, this manifests in a number of ways that we must expunge in order for us to succeed. Overall, it is an assertion that genetic science and medicine are exceptional to all other forms of science and medicine. Among the broader field of biology, there is a stereotype that geneticists are just taxonomists, obsessed with arcane knowledge of little utility, endlessly refining what was already established, which is that species and diseases can be organized into a taxonomic dendrogram and the central dogma of genetics. In medicine, this is perhaps worse, being associated with a number of false and destructive beliefs, one form of which is a kind of therapeutic nihilism, based on the stereotype that because genetic disorders are untreatable and diagnosing and studying them is pointless. Other forms of exceptionalism are that genetic risk prediction is uniquely imprecise and that the risks of genetic practice and testing are greater because genetic diagnosis is more emotionally fraught. Finally, there is an elitist exceptionalism that genetics is too challenging for non-geneticists, especially primary care providers, and thus should only be practiced by geneticists. These and other exceptionalist views unnecessarily limit the growth and expansion of the science and practice of genetics and genomics.

The adverse consequences of exceptionalism have not gone unnoticed by previous ASHG presidents, even though the consequences they identified were not directly coupled to the concept of exceptionalism. At the 1986 meeting, Dr. Charles Scriver concluded his address with this rhetorical question, likening the field of genetics to an emerging species: "Do we continue to be a somewhat reductive and [isolated] phenomenon, or do we emerge as a major organism on behalf of human genetics and its associated issues in the world of today?"[1] Scriver argued strongly for the generalizability of a genetic view of health and disease across biology and medicine, which he felt was lacking. A similar thread was present in the address by Barton Childs, the ASHG President of 1976: "But genetics is a discipline whose usefulness in medicine is directly proportional to the number of physicians who embrace it."[2]

Indeed, if genetics is limited to geneticists, we have failed. If genetics is taken up by many others, we have succeeded. In discussing my views with colleagues, I am often rejoined by something along the lines of "If genetics is taken up by others, what will there be for us to do?" Broadly expanded implementation of genetics and genomics across science and health care will provide more, not fewer, opportunities for geneticists. There will always be cutting-edge technologies and complex cases and problems that demand the attention of the geneticist. We can leave the routine to the non-specialist. Encourage the evolutionary biologist and nephrologist to run sequencing instruments; they will discover insights that are useful to their disciplines and will contribute to the larger tapestry of genetic and genomic knowledge that will be woven by the geneticists. They will make observations that are not explainable by current genetic models and that the geneticists will untangle.

We practice our science in an environment of rapidly expanding knowledge and rapidly changing scientific and technologic tools—the excitement and opportunity for geneticists lies there—not by constraining and limiting the practice of genetic science and genetic health care. Childs' address focused more on the remedies rather than the causes of the isolation (what I would call exceptionalism), but among the causes, he emphasized the need to change the external forces that were creating barriers to human genetics. In addition to Childs' plea, I would add that it is just as important to change the forces within our field. We must work to provide a balanced, but positive view of genetics and genomics and encourage all of our colleagues to do so as well. We must help our colleagues to not go down the rabbit hole of the innumerable hypothetical risks and harms. It is as if some geneticists are afraid of their own shadow—more fearful than optimistic about our field. To be successful, we must expunge exceptionalism and embrace a positive view of our field: that people everywhere realize the benefits of human genetics and genomics research.

Finally, I close by acknowledging a giant of genetics, James Neel, who provided in his presidential address the disclaimer that should be part of every such address: "Little that I have to say is new, but this will scarcely come as a disappointment to any of you, since new ideas in science are, after all, extremely rare and practically never encountered in presidential addresses." The address of this president is certainly not exceptional in that regard.

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Web Resources

ASHG Code of Conduct, https://www.ashg.org/meetings/2020meeting/attendees/policies/

ASHG Strategic Plan, https://www.ashg.org/about/mission-strategic-plan/

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