Envisioning a Future in Which People Everywhere Realize the Benefits of Human Genetics and Genomics Research

2022-2023

ANNUAL REPORT

CONTENT

ABOUT ASHG

The American Society of Human Genetics (ASHG) is the leading professional scientific membership organization for human genetics and genomics researchers in the world. The Society's members include researchers, clinicians, genetic counselors, and others who have a special interest in the field of human genetics and genomics research. In addition to serving and representing its members, ASHG also works to increase the availability of reliable information about genetics and genomics to the public.

MISSION

Advance human genetics and genomics in science, health, and society through excellence in research, education, and advocacy.

VISION

People everywhere realize the benefits of human genetics and genomics research.

CORE VALUES

- **Integrity**
- **Rigor**
- **Inclusion**
- **Innovation**
- **Collaboration**
- **Social responsibility**

MESSAGE FROM THE PRESIDENT

Dear ASHG Members,

In September 1948, a group of leading researchers and clinicians of their day joined together to develop a scientific organization that sought "the furtherance of sound research" in human genetics. This event marked the founding of the American Society of Human Genetics (ASHG), the first and largest professional organization dedicated to leading research, education, and service in human genetics. Seventy-five years into ASHG's growth and evolution, we are pleased to commemorate this notable anniversary and use it to look forward to a future that holds great possibility for the continued advancement of human genetics knowledge and its growing application to serve health and society.

This ASHG 2023 Annual Report highlights ASHG's progress during the past year to support our mission "to realize the benefits of human genetics and genomics research for people everywhere." It also reflects ASHG's "One Humanity, Many Genomes" anniversary theme, which seeks to convey that human genetics tells us so much about all that we share in common through 99.9% of our genomes, as well as what makes each of us unique, and how we can use that knowledge to improve health and well-being.

Throughout 2023, ASHG advanced its mission through its own programs and its role as a scientific partner, advocate, and convenor, and it continued its successful emergence from the challenging COVID-19 pandemic. ASHG organized its first in-person meeting in three years in Los Angeles, welcoming nearly 7,000 attendees to reunite as a community, discuss the latest research, unveil new technologies and tools, and spark collaborations that advance scientific discoveries. The Society's Digital Learning Center also continued to serve the needs of members wherever they live and work, with 13 webinars, six workshops and one podcast series. In addition, ASHG published two community guidance statements addressing responsible community engagement that limits harm and maximizes benefits to the participants and the challenges of using polygenic risk scores to predict a trait or disease outcome. ASHG's journals, *The American Journal of Human Genetics (AJHG)* and *HGG Advances (HGGA)* remained field-leading venues for scientific exchange, and *HGGA* earned its first Impact Factor of 4.4, one signal of scientific quality and engagement.

It is also our responsibility to convene critical conversations about these areas of study and to ensure broad, meaningful participation in the dialogue of how and why human genetics and genomics research serves our human community. To that end, ASHG's Board of Directors recognizes that realizing a brighter future for all must include reflecting on our history. Earlier this year, the Society's "Facing Our History – Building an Equitable Future" initiative released a significant report along with a Board statement acknowledging and apologizing for past harms and outlining near- and longer-term steps it would take to address findings. The statement also encourages the larger human genetics community to engage in similar individual and institutional reflection and action. Together with the broader community, we are committed to realizing the benefits of this research for people everywhere and consistently integrating these lessons and topics throughout Society work.

ASHG is remarkable because of the dedication of countless volunteer leaders, ASHG staff, and donors. In my year as president, I have had the great honor to help guide our organization's efforts and I am enormously grateful to work with so many amazing scientists who are giving back to the community and our outstanding staff. It took the collective contributions of many individuals over 75 years to bring our Society to this point, and it will take even more collaborative efforts to remain current and significant in the years ahead. It is a bright moment in time for our field and for ASHG. I hope you join us and contribute to building our profoundly important future.

Sincerely,

Brendan Lee, MD, PhD
ASHG President

MEMBERSHIP

ASHG works to build a vibrant and collaborative community of human genetics and genomics professionals in research, medicine, and education from across the world. Members include researchers, clinicians, genetic counselors, students, and others working to advance discovery within and application of human genetics and genomics in society. ASHG offers professional development, networking, volunteer opportunities, and much more to support their work.

Member Benefits

- **Free abstract submission to the ASHG Annual Meeting**
- **Reduced rates and early housing reservation access for the ASHG Annual Meeting**
- **Free online subscription and no publishing fees for *AJHG***
- **20% off publishing in *Human Genetics and Genomics Advances***
- **Access to key professional education, career, and networking resources including:**
  - Webinars, journal clubs, workshops, and podcasts
  - Online career center and annual career fair
  - Members-only online community and shared interest groups
- **Service on the Board and committees along with voting rights**
- **Policy resources and representation in advocacy efforts**
- **Public engagement opportunities through the Genetics Engagement & Education Network**

Member Newsletters

The Society publishes several e-newsletters that deliver vital information and resources to ASHG members on upcoming events, deadlines, and exciting new advancements in the field. These include the weekly *SNP* and quarterly *Messenger*, quarterly trainee newsletter *The Nascent Transcript*, and specialty newsletters focused on public advocacy and engagement called *Advocate* and *Engage*.

"ASHG membership shapes the future for the field and society with access to breakthrough science, career resources, partnership, and connectivity through local and global events."

— Tesfaye B. Mersha, PhD
Member since 2010

"ASHG membership gives access to cutting-edge training and events designed to prepare today's scientists for tomorrow."

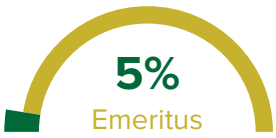
— Dwi U. Kemaladewi, PhD
Member since 2015

"ASHG membership provides access to news, webinars, podcasts, data, and publications to keep you plugged into the latest facts and analysis."

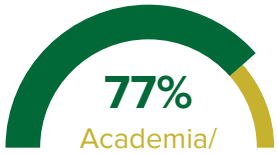
— Ninad Oak, PhD
Member since 2017

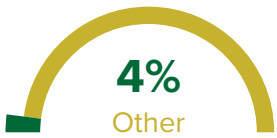
MEMBERSHIP HIGHLIGHTS

Member Types

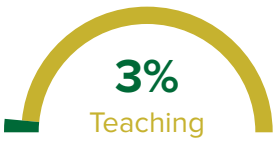


Work Settings

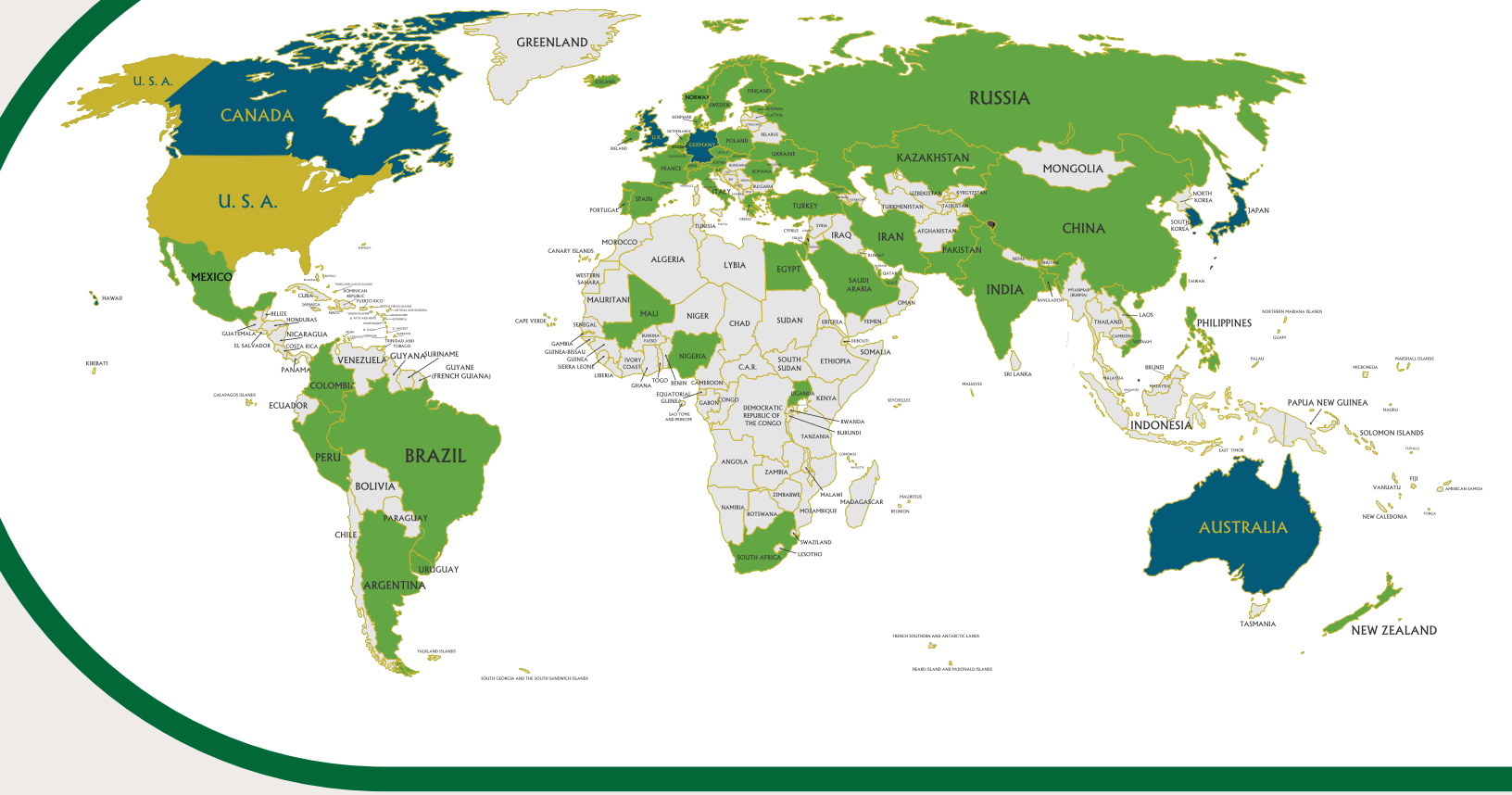




Types of Work



2022 MEMBERS 6,926



United States: 71%

1 to 5%

0.1 to 0.9%

<0.1%

ANNUAL MEETING

ASHG's 2022 Annual Meeting, held in Los Angeles, CA, for the first time in person in three years, attracted thousands of human genetics and genomics professionals to discuss the latest research, unveil new technologies and tools, and spark collaborations that advance research and medical discoveries.

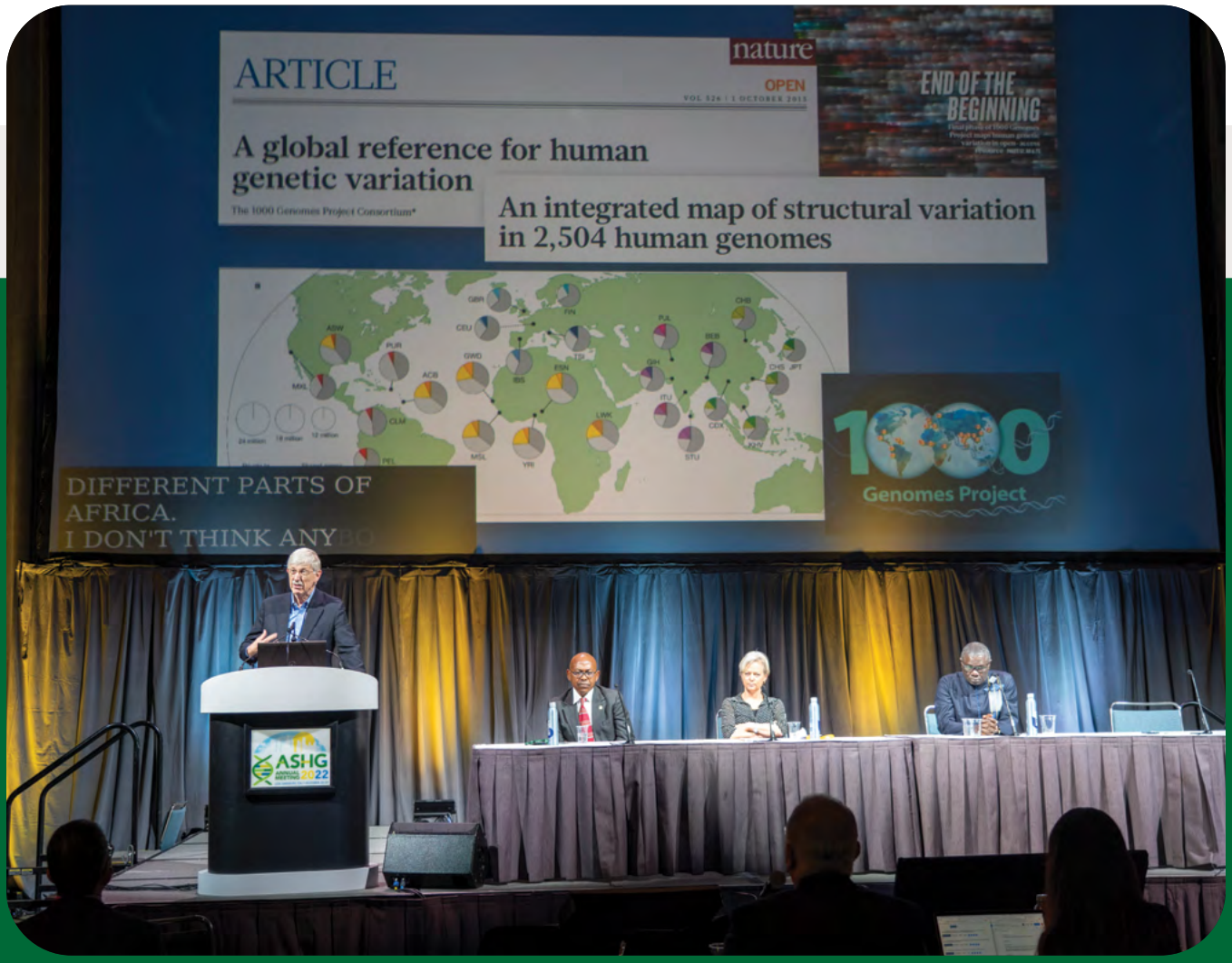
Highlights

- **6,922 Attendees** (5,443 Scientific)
- **Programs**
  - 11 Invited Sessions
  - 4 Invited Workshops
  - 15 Plenary Talks
  - 334 Platform Talks
  - 2,778 Posters
- **3,157 Abstracts Submitted + 86 Late Breaking**
- **211 Exhibitors**
- **65+ Industry-Led Sessions**

Presidential Symposium on African Genomics

The 2022 Presidential Symposium highlighted genetics and genomics in Africa, a profoundly dynamic and diverse continent, including discussing major advances, new directions and goals, emerging scientific leadership, exciting investment in technology infrastructure, and more. 2022 ASHG President, Charles Rotimi, PhD, and former NIH Director, Francis Collins, MD, PhD, moderated the session and invited distinguished speakers from Africa who included:

- **Christian Happi**, PhD, *African Centre of Excellence for Genomics of Infectious Disease, Ede, Nigeria*
- **Julie Makani**, PhD, *Muhimbili University of Health and Allied Sciences (MUHAS); Sickle Pan Africa Research Consortium (SPARCO), Dar es Salaam, Tanzania*
- **Mayowa Owolabi**, MD, *University of Ibadan, Ibadan, Nigeria*
- **Nicola Mulder**, PhD, *University of Cape Town; H3ABioNet, Cape Town, South Africa*



PUBLICATIONS

Through the Society's journals and other publications, ASHG shares the latest scientific breakthroughs and perspectives on important issues with the research community.

The American Journal of Human Genetics

Known for its scientific excellence and rigor, *The American Journal of Human Genetics (AJHG)* is one of the leading journals in the field of genetics and genomics. Since 1948, *AJHG* has published thousands of research and review articles on human heredity and the application of genetic principles in medicine and public policy.

AJHG Editorial Board

Most Downloaded Articles

The individual and global impact of copy-number variants on complex human traits

A multi-layer functional genomic analysis to understand noncoding genetic variation in lipids

Systematic comparison of family history and polygenic risk across 24 common diseases

ASHG75 American Society of Human Genetics 75 YEARS

Highlights

Impact Factors:
AJHG: **9.8** and *HGG Advances*: **4.4**

Articles Downloaded:
Over **3.3 million**

Articles Published:
200

Human Genetics and Genomics Advances

ASHG's fully open-access journal *Human Genetics and Genomics (HGG) Advances* allows for the rapid dissemination of cutting-edge, high-impact research across the breadth of human genetics that is immediately accessible to researchers and the science-interested public.

HGG Advances Editorial Board

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Deputy Editor
Jessica Chong, PhD

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Sara Mostafavi, PhD
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Charmaine Royal, PhD
Avni Santani, PhD
Fasil Tekola-Ayele, PhD
Frank Tüttelmann, MD
Jennifer Wagner, JD, PhD
Nicole Weaver, MD
Heather Wheeler, PhD
Joon-Ho Yu, MPH, PhD

Most Downloaded Articles

Improving polygenic prediction with genetically inferred ancestry

From pharmacogenetics to pharmaco-omics: Milestones and future directions

HLA-A*03:01 is associated with increased risk of fever, chills, and stronger side effects from Pfizer-BioNTech COVID-19 vaccination

ASHG75 American Society of Human Genetics 75 YEARS

PUBLICATIONS CONTINUED

Awards for Outstanding Publications

Each year, *AJHG* and *HGG Advances* recognize trainees and early-career professionals who authored articles published in the journals within the previous year and whose published work the editors feel best represent outstanding scientific contributions to the field of human genetics and genomics.

2022 *AJHG* C.W. Cotterman Award

(Now known as the *AJHG* Award for Outstanding Trainee Publication)

Shawn Fayer, MS, CGC
Closing the gap: Systematic integration of multiplexed functional data resolves variants of uncertain significance in BRCA1, TP53, and PTEN

Sam Smith, PhD
Enrichment analyses identify shared associations for 25 quantitative traits in over 600,000 individuals from seven diverse ancestries

2022 *HGG Advances* Early-Career Investigator Award

(Now known as the *HGG Advances* Award for Outstanding Early Career Publication)

Lindsay Fernández-Rhodes, PhD
Ancestral diversity improves discovery and fine-mapping of genetic loci for anthropometric traits—The Hispanic/Latino Anthropometry Consortium

Heather Wheeler, PhD
Transcriptome prediction performance across machine learning models and diverse ancestries

AWARDS

From prestigious awards for outstanding achievements in the field to supporting the next generation of geneticists through merit-based travel grants to the annual meeting, ASHG honors excellence in and promotes member work across the span of their careers.

Updates to Award Names: In June 2023, ASHG announced the renaming of the Society's awards to descriptive titles that can more quickly and clearly communicate the values and attributes of the specific awards. The new names will begin with the 2023 awards.

2022 Professional Awards

William Allan Award

Recognizes substantial and far-reaching scientific contributions to human genetics. *Now known as the ASHG Lifetime Achievement Award.*

Sir Peter Donnelly, FRS, FMedSci
Genomics plc

Victor A. McKusick Leadership Award

Recognizes individuals whose professional achievements have fostered and enriched the development of human genetics as well as its assimilation into the broader context of science, medicine, and health. *Now known as the ASHG Leadership Award.*

David L. Nelson, PhD
Baylor College of Medicine

Curt Stern Award

Recognizes genetics and genomics researchers who have made significant scientific contributions during the past decade. *Now known as the ASHG Scientific Achievement Award.*

Heidi Rehm, PhD, FACMG
Massachusetts General Hospital/Broad Institute

AWARDS CONTINUED

Early Career Award

Recognizes the contributions of genetics and genomics scientists in the first 10 years of their careers as independent investigators.

Eimear Kenny, PhD
Icahn School of Medicine at Mount Sinai

Advocacy Award

Recognizes individuals who have exhibited excellence and achievement in application of human genetics for the common good, in areas such as facilitating public awareness of genetics issues, promoting funding for biomedical research, and integrating genetics into health systems.

Meow-Keong Thong, MD
University of Malaya Medical Center

Mentorship Award

Recognizes individuals with significant accomplishment as mentors for graduate students, postdoctoral fellows, residents, or clinical fellows.

Cinnamon Bloss, PhD
University of California, San Diego

Arno Motulsky-Barton Childs Award for Excellence in Human Genetics Education

Recognizes individuals for exceptional contributions to human genetics education. *Now known as the ASHG Education Award.*

Maurice Godfrey, PhD
University of Nebraska Medical Center

ASHG/NHGRI Genomics and Public Service Fellowships

In June 2023, ASHG and NHGRI announced the expansion and renaming of our joint fellowship program through a five-year, nearly $7 million contract. The newly named **ASHG/NHGRI Genomics and Public Service Fellowships** program will continue and enhance the prestigious post-graduate Genetics & Public Policy Fellowship and Genetics Education & Engagement Fellowship and launch two new fellowships: a post-graduate Genomics Communication Fellowship and Post-Baccalaureate Genomics Analyst Fellowship. Applications open for all programs in fall 2023 with the first cohort starting rotations in summer 2024.

Genetics & Public Policy Fellowship

Designed for those interested in genetics-related health and research policies at a national level. Fellows participate in policy analysis at NHGRI, ASHG, and the U.S. Congress.

Albert Hinman, PhD: 2022-2024 Genetics & Public Policy Fellow

Genetics Education & Engagement Fellowship

Designed for those interested in genetics-related public engagement and/or professional development for students to professionals. Fellows participate in projects at NHGRI, ASHG, and a third organization.

Nancy Sey, PhD: 2022-2024 Genetics Education & Engagement Fellow

Charles Epstein Trainee Awards for Excellence in Human Genetics Research

The Charles Epstein Trainee Awards, now known as the ASHG Trainee Research Excellence Awards, honor excellence in research conducted by predoctoral and postdoctoral trainees, recognizing highly competitive abstracts submitted and presented at the ASHG Annual Meeting. The 2022 winners included the following:

Predoctoral

Layla Siraj, Harvard University

Frederik Filip Vinggaard Stæger, University of Copenhagen

Marianne Lemée, University of Strasbourg and IGBMC

Postdoctoral

Hai Nguyen, University of California San Francisco

Nathan Nakatsuka, New York Genome Center

Yosuke Tanigawa, MIT

LEARNING & CAREER DEVELOPMENT

ASHG offers year-round online access to professional education and career development programs for genetics and genomics professionals across all career stages and interests.

ASHG Learning Center

The ASHG Learning Center provides access to all ASHG digital programs including live and on-demand webinars, interactive workshops, podcasts, and Annual Meeting content. An ongoing grant from the Illumina Corporate Foundation since 2020 has enabled and supported the growth of ASHG online education programming.

2,818 Registrants

13 Webinars

6 Workshops

Genetics and Genomics Digital Forum

After relaunching an in-person Annual Meeting following two years of being virtual due to the pandemic, ASHG convened a Digital Forum immediately following the Annual Meeting for those unable to attend the in-person meeting or who wanted additional content and networking following the in-person meeting. Programming included live and on-demand talks, workshops, and networking sessions.

6 Plenary Talks

12 Poster Talks

4 Workshops

3 Industry Education Sessions

6 Epstein Award Winner Talks

2 Networking Sessions with 5 Breakout Rooms

ASHG Career Center

The ASHG Career Center features year-round resources and opportunities for genetics and genomics professionals, no matter their career stage. The ASHG Job Board has become one of the biggest resources for our members since its launch in 2019. Over 250 employers posted over 2,000 open positions on the site this past year.

Career and Professional Development Events

ASHG also held multiple events focused on career and professional development including:

- **ASHG Annual Meeting events**
  - 2nd ASHG Career Fair, which boasted 12 booths, career coaches offering free resume review, and professional headshots
  - Self-Discovery Workshop: Finding Balance & Joy in Your Work
  - Trainee Social Reception

- **Webinars focused on**
  - Caregiving for children, elders, and personal relations *(part of ASHG's Work-Life Balance Webinar Series)*
  - Perspectives from ASHG Members Working in Industry

TOP MILESTONES IN

HUMAN GENETICS AND GENOMICS

Every advance paved the way for those that followed, culminating in the sequencing of the human genome, the complete set of genetic information contained in each person's DNA, and progress made in understanding the human genome. These accomplishments have led to major progress in elucidating the role that DNA variants play in health and disease, how to use this information for personalized care and precision health, and the ability to go beyond coding DNA to other parts of the genome.

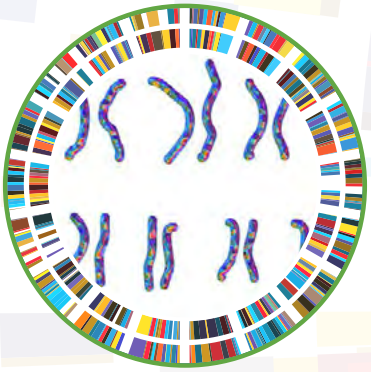
DNA and the Human Genome

The successful completion of the Human Genome Project would not have been possible without the many discoveries that preceded it. From understanding the double helix structure of DNA to learning how to sequence the nucleotides that make up DNA to developing the tools needed for research, progress in genetics has been incremental, a step-by-step process that continues to evolve.

Every Person and Genome Is Unique: Variation in the Human Genome

State-of-the-art tools perfected during the 2000s led to the discovery of the tremendous variation in the human genome. Knowledge gained from this work included learning that the genomes of two human individuals are 99.5% identical at the DNA level, yet every person has variants that make them unique. Also, it was understood that sets of genetic variants could be passed along from one generation to the next and great progress was made in identifying variants that could result in disease. Moving forward, the goal is to sequence the genomes of as many people worldwide as possible—the most effective way to uncover the whole catalogue of variants of humanity.

Progress in human genetics and genomics research spans decades and is made possible by emerging tools and technologies. Below are major highlights. We encourage you to visit ASHG's website https://www.ashg.org/discover-genetics/timeline/ to see the many advances over decades that made these milestones possible.



The Genetic Basis of Disease

Beginning in the 1950s, researchers first used a molecular technique called karyotyping to visualize human chromosomes and their differences linked to conditions such as Down syndrome. Much progress has been made since then. We have identified many other causes of genetic anomalies, including single nucleotide variants and larger structural changes that alter protein-encoding genes. We are now starting to understand the role of variation in regions that control the expression of genes and how that impacts human diseases. We are just now starting to understand the role of variation in non-protein coding parts of the genome and their role in human diseases.

How Genomics and Technology Are Changing the Health Care Landscape

Today, when genetic diseases are detected, physicians can do more than just treat the symptoms. In some cases, they can actually replace defective genes with healthy ones or repair a misspelling in the DNA. These technologies are the tip of the iceberg, a preview of what will be possible in the coming years.

Genome Function Beyond DNA

With many questions about DNA now answered, scientists are now looking at genes in a broader context. For example, how do they interact with the environment? What can we learn from gene expression profiles of different tissues in the body? Information like this can be used widely in a range of research projects and to better understand disease and improve people's lives.

ENVISIONED ADVANCES IN

HUMAN GENETICS AND GENOMICS

Equitable Expansion of Precision Medicine

- Every patient with a suspected genetic disorder can be whole-genome sequenced to obtain an accurate diagnosis of their disease, eliminate the diagnostic odyssey, and potentially guide management and treatment based on the specific molecular defect in the patient
- Full integration of our understanding of inherited characteristics and individual genomic information, along with environmental and lifestyle factors, into the day-to-day medical care to better assess and predict risk of diseases (e.g., monogenic and polygenic health conditions, including common infectious and noncommunicable diseases such as diabetes, heart disease and cancer)

Increase Awareness of Genetics

- Increased education and awareness of genetics concepts and topics among the general public and health professionals to enable informed discussions and decision making about individuals' health based on their genomic information

The field of human genetics and genomics is relatively young and advancing at great speed, but it continues to tackle scientific challenges and raise societal implications. ASHG asked several field leaders who are also ASHG members to identify some areas of scientific and community interest for years and decades to come. Read more detailed information about these possibilities at https://www.ashg.org/discover-genetics/forward-looking-advances-for-human-genetics-and-genomics/.

Increase Diversity of Genomics Research

- Increasing diversity and worldwide representation of participants in genomics research in an equitable and respectful manner
- Creation and whole-genome sequencing of longitudinal biobanks from populations around the world

Explore New Avenues for Research and Clinical Impact

- Using patient-derived models such as organoids to test efficacy of medications and therapeutic avenues on patients prior to clinical implementation
- Full understanding and functional characterization of all protein coding and noncoding genes in the human genome, their normal biological functions, and associations with diseases or traits

REALIZING THE FULL POTENTIAL OF

"ONE HUMANITY, MANY GENOMES"

As part of ASHG's 75th anniversary celebration, ASHG engaged geneticists from different parts of the world and pursuing different aspects of the field. Some are in earlier career stages, while others have experienced firsthand the enormous progress in genetics and genomics over decades. Some are pursuing fundamental research while others work primarily with patients, and many apply their time engaging patient communities and integrating ethical and societal implications and other vital facets of genomics dialogue. In so doing, they all help pursue ASHG's mission to advance human genetics and genomics research, education, and advocacy to benefit science, health, and society.

Gail Jarvik, MD, PhD, the Arno G. Motulsky Endowed Chair in Medicine, Joint Professor of Medicine and Genome Sciences, Head of the Division of Medical Genetics, and Adjunct Professor of Epidemiology at the University of Washington Medical Center and Affiliate Member of the Fred Hutchinson Cancer Research Center. 2021 ASHG President.

Dr. Jarvik celebrates the past decades' major milestones in identifying the Huntington's disease gene and the sequencing of the full human genome. Yet she knows genetic databases are missing genomic sequences from underrepresented people around the world. That's why she has taken a leadership role in the National Institutes of Health *All of Us* project. In 2022, the project announced completion of the first 100,000 genome sequences, with 50% of those from individuals from groups that have been historically underrepresented in research.

"The project has also shown that by engaging with the community to gain trust, more people are willing to enroll and become part of this scientific endeavor," says Dr. Jarvik. ***"With greater diversity in the database, the resource will become indispensable for health-related research."***

Another project in which she is involved, the eMERGE consortium, is using genome-wide association studies (GWAS) to analyze data from a database that contains records of people with a range of diseases and conditions. Researchers are looking at the impact of small genetic and genomic variations to learn about the causes underlying disease.

Dr. Jarvik hopes to see more genetic knowledge translated into treatments for common diseases, such as heart disease and cancer. ***"As we gain a better understanding of how genes interact with the environment and with each other, it will become possible to improve prevention measures and treatment,"*** she adds.

FIELD LEADERS REFLECT ON PROGRESS, POTENTIAL FOR HUMAN GENETICS AND GENOMICS RESEARCH

Benjamin Neale, PhD, Co-director of the Program in Medical and Population Genetics at the Broad Institute and Director of Genetics at the Institute's Stanley Center for Psychiatric Research.

Dr. Neale came of age with technology, and he sees analytical and computational tools as the most effective way to analyze large datasets. He thinks biobanks ***"will slowly but surely transform how we think about human health, what environmental exposures matter, and what treatments may be possible."***

The focus of Dr. Neale's research is severe mental illnesses, particularly schizophrenia, bipolar disorder, and attention deficit hyperactivity disorder. ***"These diseases are complicated and challenging to study from a genetics perspective, but this work may shed light on what variants increase an individual's risk of developing one of these disorders,"*** he says.

Dr. Neale and his team conducted an analysis for the Schizophrenia Exome Sequencing Meta-Analysis consortium. They found variants in 10 genes that have a significant impact on a carrier's risk of developing schizophrenia. He also has analyzed data from the first ADHD GWAS study. He believes sharing data from large datasets is the wave of the future — the best way to build scientific knowledge and to address such pressing issues as the relationship between genetic and phenotype variation and how to measure genetic differences.

"It is my passion to figure out the underlying causes of schizophrenia and bipolar disorder and to help develop better treatments," Dr. Neale says.

"We share much of our DNA, but many small differences have big consequences, causing people a great deal of suffering. Genetics presents a new way to look at the world and an opportunity to improve the quality of life for individuals with complex disorders."

— Benjamin Neale

Claudia Gonzaga-Jauregui, PhD, Principal Investigator, International Laboratory for Human Genome Research, LIIGH, UNAM. Founder Red Mexicana de Enfermedades Raras

Dr. Gonzaga-Jauregui was in high school when the Human Genome Project was completed. As a PhD student at Baylor College of Medicine, she was part of the team that conducted one of the first whole genome analyses, opening the doors to understanding the link between genetic variations and disease. She returned to Mexico determined to bring her experience and knowledge to Latin America. ***"Our goal is to bridge the gap between a rare disease and its genetic cause,"*** Dr. Gonzaga-Jauregui explains. ***"We're focusing on Mendelian disorders, those caused by mutations in single genes, and using genomic and exome sequencing to provide a diagnosis and characterize genetic variation in Latin America."***

To date, Dr. Gonzaga-Jauregui and her team at the University of Mexico have sequenced the genomes of 15 families and are planning to sequence another 15 sets. The hope is to better understand the molecular architecture of rare diseases that run in families. Over the long term, Dr. Gonzaga-Jauregui would like to see biotechnology companies bring sequencing technologies to other parts of the world to achieve equity in the makeup of genomic databases. Furthermore, much work still needs to be done to educate people about why this work is important.

Stephen B. Montgomery, PhD, Associate Professor of Pathology, of Genetics, of Biomedical Data Science and, by courtesy, of Computer Science

Scientists have long known that before genetic variations manifest themselves as diseases, they are already active at the molecular level. Dr. Montgomery is investigating why and the impact of variants on gene function.

With others, he is analyzing information in the Genotype-Tissue Expression (GTEx) Dataset, which includes tissues from 800 people who have died. Dr. Montgomery's lab investigated expression levels of different variants throughout the body. For example, when looking at the variant associated with obesity, Dr. Montgomery's team questioned whether the variant was impacting expression of adipose cells in the body or the hypothalamus in the brain. They also investigated whether the variant affected the pancreas and how insulin is secreted.

"As we uncovered where in the genome the variation was, we developed a map of these variant effects," Dr. Montgomery explains. ***"Other researchers can now use this resource to dig deeper into the relationship between variants, gene functioning, and the onset of disease, [including] changes during different developmental stages."***

Recently, Dr. Montgomery is looking at rare, undiagnosed diseases. He is making use of a large database called GREGoR, or Genomics Research to Elucidate the Genetics of Rare Diseases, developing computational approaches to help clinicians identify and diagnose disease. ***"We're looking for the right tools and technologies to facilitate that diagnosis."***

Michele Ramsay, PhD, Professor, Division of Human Genetics, National Health Laboratory, University of the Witwatersand, Johannesburg, South Africa

Dr. Ramsay has devoted her life to studying diversity at the genetic level and pushing for more sequencing across the African continent.

"I'm particularly interested in understanding susceptibility to diseases such as diabetes, as well as obesity and stroke," she says. *"To realize this goal, it's also important to understand the gene environment in the broadest sense, which includes climate, diet, and exposure to alcohol and smoking."* Her work has also shown that the link between genetic variants and health outcomes isn't always straightforward. *"These links aren't always black and white, making it important to be careful how data is interpreted."*

Dr. Ramsay believes that it's crucial to add more understudied populations from diverse groups to worldwide databases. Yet with larger, more diverse databases, the ethics of how to describe people needs to be considered thoughtfully. *"Respecting the privacy of individuals and protecting the data from bad actors while sharing data must be done simultaneously,"* Dr. Ramsay says. *"It's possible to put in place systems that promote fair data sharing."*

Neil Hanchard, MBBS, DPhil, Senior Investigator, Center for Precision Health Research, National Human Genome Research Institute, National Institutes of Health

Dr. Hanchard describes his entrée into genetics research as "pure serendipity." He found his passion while studying genetics at the University of Oxford in the United Kingdom.

"I found genetics fascinating," Dr. Hanchard recalls. *"I focused on rare disorders in pediatrics, and I wanted to figure out why some children developed these diseases. By looking at diseases through a genetic lens, we've gained insight into what causes them. I'm particularly interested in diseases that have been around a long time but whose physiology still eludes us."*

Sickle cell disease is a case in point. Physicians and scientists had long noticed that some patients were at increased risk of producing antibodies following lifesaving transfusions. Dr. Hanchard's team discovered mutations that appear to cause this problem.

> **"I think of humans as having a shared experience and a shared ancestry. While most of the genome is the same, the portions that are different drive the richness of humanity."**
>
> **— Neil Hanchard**

DIVERSITY, EQUITY, & INCLUSION

As both a strategic and scientific imperative, ASHG is committed to advancing diversity, equity, and inclusion within the genetics and genomics workforce and meaningful engagement of underrepresented groups as participants in genetics and genomics research.

Report on Facing Our History – Building an Equitable Future Initiative

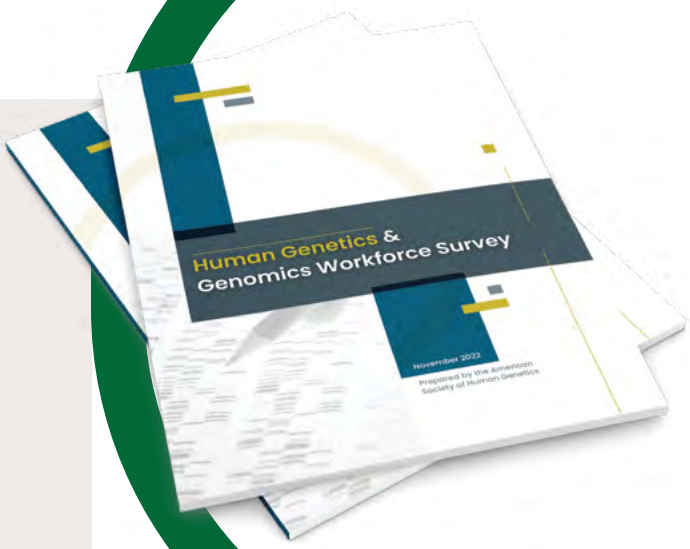
In Winter 2023, ASHG released a report on the *Facing Our History – Building an Equitable Future Initiative* which involved a year-long effort to acknowledge and reckon with a history of past injustice as well as progress toward justice in the field and Society. An expert panel composed of leading human geneticists, historians, clinician-scientists, social scientists, and equity scholars led the effort with support from ASHG staff and an outside research firm. The report process included an intensive research and environmental scan, four Expert Panel meetings, and community dialogue, leading to the identification of four major themes and several immediate actions that ASHG commits to take.

Major Themes

1. ASHG and the American Eugenics Movement
2. ASHG Was Silent When Genetics Was Misused to Justify Social Harms
3. ASHG's Evolving Role to Advance Ethical and Legal Protections
4. ASHG Strives for a More Equitable and Just Future

Major Actions

1. Publish the full report and statement publicly online and in *AJHG*.
2. Increase integration of equity into scientific and training initiatives at ASHG.
3. Sustain advocacy for research DEI through policy and communications agendas.
4. Continue to build the diversity and inclusivity of ASHG's leadership.
5. Assess the names of ASHG professional awards for the future.
6. Update and prioritize DEI objectives as part of refreshing ASHG's strategic plan.



Human Genetics & Genomics Workforce Survey Report

In fall 2022, ASHG released a comprehensive report examining diversity in the human genetics and genomics workforce. ASHG, in partnership with NHGRI, the American College of Medical Genetics and Genomics (ACMG), and the National Society of Genetic Counselors (NSGC), conducted the survey to understand the demographics of the genetics and genomics workforce, as well as the climate and culture in training programs and the workplace. Drawing from 4,367 responses across the spectrum of the workforce (i.e., researchers, clinicians, and genetic counselors), the report's findings confirm that progress continues to be made and provide insights about what challenges the field and organizations within need to address to build a more diverse workforce.

Respondent Demographics

- **Primary Area of Work**
  - Genetic Counseling (45.7%)
  - Research (30.4%)
  - Academic (23.4%)
- **Gender**
  - 74.7% Women
  - 23.3% Men
  - 0.5% Nonbinary or Transgender
- **Race, Ethnicity, or Ancestry**
  - 67.0% White.
  - 7.4% Asian
  - 1.5% Black, African American, or African
  - 2.0% Hispanic, Latino, or Spanish
  - 1.1% Middle Eastern or North African
  - <1% American Indian or Alaska Native
  - <1% Native Hawaiian or Other Pacific Islander (<1%)
  - 4.8% Multiracial

"Successful and pioneering research relies on the talent of the research workforce and the goal of this project is to assess the demographic landscape of the human genomics workforce and the factors affecting diversity, equity, and inclusion in the field. Our field is only as strong as the individuals that contribute to shaping frontiers and maximizing the benefits of this science."

— Charles Rotimi, PhD, 2022 ASHG President

DIVERSITY, EQUITY, & INCLUSION CONTINUED

Human Genetics Scholars Initiative

Made possible with continued foundational support from NHGRI and Biogen along with annual support from GSK, Merck & Co., and Roche, the Human Genetics Scholars Initiative supports the professional success of diverse early-career genetics and genomics researchers by providing them with year-round education, networking, and mentoring while also working to develop a community of researchers and leaders committed to workforce diversity, equity, and inclusion at their institutions.

2022-2024 Human Genetics Scholars

Kimberly Diaz Perez, BS
PhD Candidate
Emory University

Paige Haffener, BS
PhD Candidate
University of Utah

Gabrielle Hampton, MS
PhD Candidate
Vanderbilt University

Eric Sosa, BS, MS
MD/PhD Candidate
Albert Einstein College of Medicine

Lizeth Tamayo, BS, MPH
PhD Candidate
University of Chicago

Adelaide Tovar, PhD
Postdoctoral Fellow
University of Michigan

Travis Tu'ifua, BS
PhD Candidate
University of Utah

Matanel Yheskel, BS
MSTP Student
Albert Einstein College of Medicine

Impact Partnerships for Equity in Genetics and Genomics

Knowing that collective effort is required to ensure all people benefit from genetics and genomics research, ASHG launched the Impact Partnerships for Equity in Genetics and Genomics Research in January 2022. Illumina and Invitae joined as founding partners demonstrating their shared commitment to advancing DEI in human genetics and genomics. Through their financial contributions, Impact Partners support the Society's diversity, equity, and inclusion efforts. ASHG and Impact Partners also meet periodically throughout the year to share knowledge and experiences and discuss opportunities for future efforts aimed at advancing DEI in the field.

ADVOCACY & POLICY

ASHG, through its leadership and member advocates, speaks out on behalf of the human genetics and genomics community by advocating in Congress for robust and sustained investment in research; writing perspectives on the implications of genetics and genomics discoveries; advocating for greater diversity, equity, and inclusion in genomics research; and promoting the value and impact of science for personal and public health and society at large.

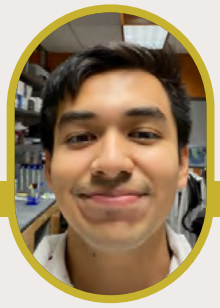
Advocacy Certificate for Human Genetics and Genomics Trainees

The Society provides training, opportunities, and resources to support members in engaging with policymakers including the Advocacy Certificate for Human Genetics and Genomics Trainees (ACGT). Now in its second year, ACGT helps trainees build their professional skills within science policy and advocacy. This past year, ACGT members joined ASHG at the Rally for Medical Research and participated in ASHG's virtual Capitol Hill Day, during which they spoke with congressional offices and leaders about the impact and value of NIH research support and other policy priorities.

2023 Advocate Program Trainees


Anna Capria


Hubert Chen


Richard Coca


Ricardo Harripaul


Christy LaFlamme


Jasmine Lewis


Nandana Rao


Grace Tietz


Leonard Wang

ADVOCACY & POLICY CONTINUED

ASHG Guidance on Community Engagement and Polygenic Risk Scores

ASHG recognizes that researchers do not always have formal training on the ethical or social implications of their research. To address this need, ASHG develops and disseminates through *AJHG* timely publications called "Guidance" to help members consider and address responsible application of their research. In fall 2022, ASHG published two guidance statements on the topics of:

1 responsible community engagement that limits harm and maximizes benefits to the participants, and

2 the challenges of using polygenic risk scores to predict a trait or disease outcome.

SUCCESS STORY

Advocating for an NIH Community Engagement Program for Genomics Research

ASHG advocated successfully to Congress for inclusion of specific language in the Consolidated Appropriations Act of 2023, passed in December 2022, that establishes a community engagement program at the National Human Genome Research Institute, modeled after the NIH Community Engagement Alliance (CEAL) Against COVID-19 Disparities program. ASHG encouraged language that includes fostering diverse public participation in research and supporting efforts to increase the participation of individuals historically underrepresented and hesitant to participate in human genetics and genomics research.

PUBLIC ENGAGEMENT

ASHG actively engages the public in learning more about their genomes, gaining a new appreciation of humanity's diversity and our shared genetic heritage as a single species, and exploring the benefits and implications of genetics and genomics for society as a whole.

One Humanity, Many Genomes: Celebrating 75 Years of ASHG

Established in 1948, ASHG marks our 75th anniversary in 2023 through the lens of "One Humanity, Many Genomes," which emphasizes how humans are more similar than we are different but that tiny genomic differences can markedly affect our health and disease risk, especially combined with differing environments.

DNA Day Essay Contest

ASHG 2023 DNA DAY ESSAY CONTEST

Each year, ASHG works to spark excitement and learning among the next generation of genetics professionals and foster greater public awareness through our annual DNA Day Essay Contest. In 2023, the Society received over 950 submissions from nearly 30 U.S. states and 40 countries outside of the U.S. The question for the 2023 annual contest asked students to explain what "One Humanity, Many Genomes" means to them in relation to how advances in understanding our genomes impact our lives. For the first time, ASHG invited the first-place winner to record a video with their teacher about their essay and the writing process.

2023 DNA Day Essay Contest Winners

Additional DNA Day celebrations included a discussion panel on the Ask Science Reddit page, a social media campaign recognizing pioneering leaders in the field, and a webinar and fact sheet focused on career paths in human genetics targeted to middle and high school students.

Jennifer Zhong

Teacher:
Ms. Maria Zeitlin

School:
Smithtown High School East

Location:
Saint James, New York

Bolin Miao

Teacher:
Ms. Mary Frances Hanover

School:
Dana Hall School

Location:
Wellesley, Massachusetts

Olivia Park

Teacher:
Ms. Cindy Law

School:
William Lyon Mackenzie C.I.

Location:
Toronto, Canada

Read the winning essays at ashg.org/dnaday

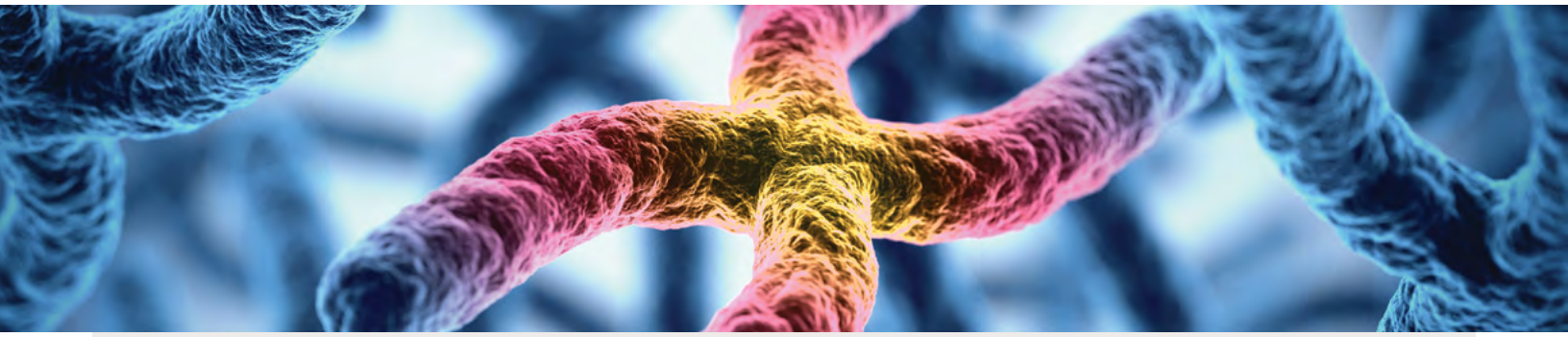
ASHG75 American Society of Human Genetics 75 YEARS

PUBLIC ENGAGEMENT CONTINUED

Discover Genetics

ASHG's public-facing website, Discover Genetics, added new resources this past year, including:

- Two fact sheets focused on analyzing DNA and career paths in genetics.
- Weekly news updates that highlight exciting stories and developments within genetics and genomics.
- A timeline of top milestones in the field over the last 75 years.
- Perspectives of six members about the year's theme and how their work impacts the field and society.



Social Media

ASHG social media channels expand our reach to the greater public to garner added interest from broader audiences into genetics and genomics and Society programming, resources, and membership. This past year, ASHG hosted a Twitter Chat with the National Hispanic Medical Association (NHMA) and current chair of the Public Engagement and Awareness Committee (PEAC), Kenneth Ramos, MD, PhD, to discuss how ASHG's diversity messaging project relates to NHMA programs.

50,910 followers with **337** new per month

22,513 followers with **43** new per month

7,761 followers with **278** new per month

4,440 followers with **66** new per month

FINANCES

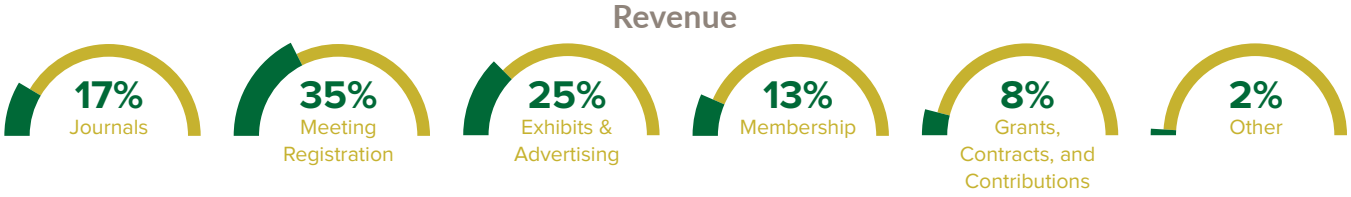
ASHG's finances remained sound in fiscal year 2023 despite the continued impact of the COVID-19 pandemic and inflationary pressures. Preliminary financial results are presented here with audited financial statements being available online later this year.

Due to the COVID-19 pandemic's impact on operations from FY20-23, ASHG experienced substantial variability in operational revenue and expense over the four years but emerged with only a modest net loss thanks to sustained attention to quality programming and member services, careful operations management, and prudent insurance and other risk-mitigation practices.

Operations: FY23 operating revenue totaled $7.6 million while operating expenses totaled $8.7 million, producing an operating loss of $1.1 million. Operating expenses exceeded revenue in large part due to 2022 Annual Meeting performance, which was the first in-person meeting since 2019 and occurred just as public events were beginning to reopen more fully as the bivalent vaccine became available. While scientifically successful, attendance and exhibit revenue was lower than expected and expenses were high due to acute broader inflation. The Society also gratefully accepted valued member and company donations and secured several large grants, contracts, and contributions to support digital learning, DEI initiatives, and fellowship programs.

Reserve Portfolio: The Society's investments remain prudently invested for both long-term performance and protection of the Society. The Society's investment reserve portfolio weathered the market fluctuations this past year, reporting a reserve totaling $19.2 million at fiscal year-end.

Strategic Investment Funds: The strength of ASHG's reserve portfolio provides the opportunity to reinvest further in Society programs and services. As such, the Board authorized use of up to 4% of the reserve value in FY23 to support strategic initiatives. These investments totaled $500K and included DEI programs such as the history initiative, advocacy training for early-career professionals, COVID-19 safety measures at the annual meeting, investment in journal marketing, strategic planning, and more.



PARTNERS

ASHG partners with numerous organizations and government agencies to fund and collaborate on programs and initiatives advancing our collective vision that people everywhere realize the benefits of human genetics and genomics research.

Grants & Sponsorships

Thank you to those organizations who joined in support of programs and initiatives that benefit the human genetics and genomics research community through their generous financial and volunteer contributions.

National Human Genome Research Institute

ASHG/NHGRI Fellowships
Human Genetics Scholars Initiative
Genomic Workforce Diversity Initiative

illumina®

Impact Partnership (2022-2023)
ASHG Learning Center

INVITAE

Impact Partnership (2022)

Biogen

Human Genetics Scholars Initiative

gsk do more feel better live longer

Human Genetics Scholars Initiative

Variant Bio

Resource-Limited Country Awards

BIOMARIN®

Charles Epstein Trainee Awards

Roche

Human Genetics Scholars Initiative

MERCK

Human Genetics Scholars Initiative

Collaborations

The Society collaborates with multiple other nonprofit and governmental organizations to pursue policies and programs that advance scientific research.

- Ad Hoc Group for Medical Research
- American Association for the Advancement of Science
- American Board of Medical Genetics and Genomics
- American College of Medical Genetics and Genomics
- Annual Biomedical Research Conference for Minority Students
- Association of Professors of Human and Medical Genetics
- European Society of Human Genetics
- Federation of American Societies of Experimental Biology
- International Federation of Human Genetics Societies
- Minority Genetic Professionals Network
- National Human Genome Research Institute
- National Science Teachers Association
- National Society of Genetic Counselors
- Research!America
- Society for Advancement of Chicanos/Hispanics and Native Americans in Science
- U.S. National Academies of Science, Engineering, and Medicine

DONORS

Our donors make a huge impact on the genetics and genomics community and society at large. Thank you for all you do for the Society and the community. The individuals and organizations listed below made charitable gifts between July 1, 2022, and June 30, 2023.

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