

2017 NHGRI Research Training and Career Development Annual Meeting

Report from the DACC

The NHGRI Research Training and Career Development Program 2nd Annual Meeting was held on April 12-14, 2017 at the Chase Park Plaza in St. Louis, MO. This meeting continues to provide a venue for trainees supported by the National Institutes of Health (NIH) National Human Genome Research Institute (NHGRI) in Genomic Sciences, Genomic Medicine, and in Ethical, Legal and Social Implications (ELSI) fields to present their research and to form collaborations with other trainees and established investigators from training programs across the United States.

There were 244 registered to attend the meeting (see **Appendix A**). Of these, 179 were **trainees**, including those from the Diversity Action Plan programs (DAP), the T32 training programs, individual awardees of fellowship or career-development (F or K awards) or Centers of Excellence in ELSI Research (CEER) awards, and a NHGRI intramural intern.

OPENING SESSIONS

- **Welcome and Introduction of Participants:** Carolyn Hutter, Director of the Division of Genome Sciences, welcomed meeting attendees by highlighting that the Annual Meeting serves to convene the NHGRI-supported trainees, showcase genomics research and share findings, and provide opportunities for attendees to network with other trainees, mentors, and NHGRI staff. Carolyn reminded trainees that the meeting provides tools for career development and that NHGRI has always been supportive of its training programs. Tina Gatlin, NHGRI, followed the welcome with meeting announcements. Tina introduced staff from the NHGRI and the DACC as well as the advisors. Tina highlighted Agenda sessions and called attention by name to three genomic new medicine T32 programs (University of Utah, University of Alabama at Birmingham (UAB), and Vanderbilt University) as well as to a new ELSI T32 Program at Stanford University.
- **The Keynote Presentation:** The keynote presentation was given by Dr. Carol Bult, an NHGRI training program advisor and faculty at The Jackson Laboratory, gave a talk entitled, “Why it Matters to be FAIR.” Dr. Bult described the FAIR principles of data management: findable, accessible, interoperate, and reusable. To understand the genetic and genomic basis for human disease, one needs to apply these principles. With an unprecedented amount of data being produced, it is imperative that researchers maintain that their data is accessible for computational assessment. Data-driven sciences rely on data integration so that queries can be answered by many different databases at once.

FEATURED PANEL SESSIONS

- **Whole Genome Sequencing Variant Case Review Session:** Dr. Bruce Korf, chairman of the Department of Genetics at UAB, moderated this panel session made up of three teams of faculty from Utah, UAB, and Vanderbilt plus nine trainees who volunteered to participate in the session. Each team reviewed three different case summaries including DNA sequencing variants identified by whole genome sequence to determine the potential clinical impact of the variants. Each case summary included a clinical description, inheritance of the variant (when known), and information

about the variant's predicted impact. Panel participants discussed their preparation for the session, including literature reviews of each variant and team deliberations whether or not findings, even if incidental, should be returned to the patient. Teams deliberated each case's findings and formed conclusions on the variant's pathogenicity in context of one of the five ACMG categories (benign, likely benign, uncertain significance, likely pathogenic, and pathogenic). Teams reached broad consensus matching conclusions reached by teams at HudsonAlpha who originally worked on these cases.

- **Mock Grant Review:** Drs. Rudy Pozzatti (NHGRI) and Mike Boehnke (University of Michigan) gave trainees a look into the NHGRI grant review process by moderating a mock grant review. Dr. Pozzatti first briefly reviewed the grants process (e.g., Scientific Review Officers read each grant application and then recruit a chair and panel members to review and score grants for their scientific and technical merit). The mock review panel consisted of three teams each with three trainee volunteers. Each team reviewed one of three real (i.e., actually submitted to NHGRI) grant applications and discussed the grant's aims, strengths, and weaknesses by considering each grant application's significance, investigators, innovation, approach and environment. Scores were later assigned. The session also allowed trainees to ask questions about the grant review process.

FEATURED TALKS

- **Avoiding Common Pitfalls in Research Integrity: Data-Informed Tips for Genetic Researchers:** James DuBois, faculty at Washington University School of Medicine noted that many investigators find themselves not meeting compliance and ethical standards during their careers. His professionalism and integrity in research program trains investigators in avoiding these common pitfalls including failure to follow protocol, informed consent issues, oversight failures, and privacy violations. Dr. DuBois encouraged trainees to avoid these pitfalls by seeking help early, managing stress and time, holding regular team meetings, and normalizing compliance as a part of doing research.
- **WU Program Showcase:** Two faculty members affiliated with Washington University's T32 and DAP Programs, Drs. Barak Cohen and Cherilynn Shadding, respectively, highlighted some of the unique features of their programs. Specifically, Barak discussed how the T32 program focuses on "Antedisciplinary" Training, defined as training an individual to be knowledgeable in multiple (inter- and cross-disciplinary) fields, fluent in both theoretical and experimental fields. Cherilynn reviewed the gap in STEM careers among underrepresented minorities (URMs) as compared to the percentage of URMs in the general population and highlighted the results of her program showing success in retaining URM students in STEM fields during her 2nd year of grant funding. The third speaker was Ramin Lalezari of Sling Health. Ramin talked about his organization's vision to improve patient care by teaching students how to become entrepreneurs and to provide students the space (e.g., lab, office, workshop), funding, legal services (e.g. patent support), and mentorship (e.g., industry experts and clinicians) needed to develop and commercialize medtech solutions.
- **Plenary Lunch, For the Sake of All: Translating Evidence into Action for Community Health:** Jason Purnell, Faculty, at the George Warren Brown School of Social Work at Washington University in St. Louis, gave a presentation during Friday afternoon's lunch to highlight health

inequities in the St. Louis region. He discussed how his community health project, *For the Sake of All*, uses research translation and civic education to address inequities while intervening on the social determinants of health. Dr. Purnell reviewed the goals of his project, including influencing the policy agenda on health disparities, engaging community members around recommendations for improvement, and activating key private and public sector stakeholders to implement recommendations.

- **A Brief Introduction to the Precision Medicine Initiative All of Us:** Joshua Denny, faculty at Vanderbilt University, described the fundamental problem the Precision Medicine Initiative (PMI) seeks to solve: better ways of treating patients. He highlighted the basic tenets of PMI which include focuses on diversity and engagement and that at least 1 million participants be followed over time and can be re-contacted for follow-up. Dr. Denny noted that Precision Medicine is not a new idea, but the *All of Us* Initiative works to advance personalized medicine by having data accessible to researchers, providers, and industry via the standardization of the electronic medical health record.

CONCURRENT SESSIONS

- **Career Development Session:** Carol Moakley and Thi Nguyen, both from the Career Center at Washington University in St. Louis, focused on trainees' transferrable skills. These are the skills that trainees may gain in the lab but can then be translated into action verbs in an interview or on a resume, especially when trainees are considering new jobs or career fields. Transferable skills include analytical thinking, team-based problem solving, and grant writing. The speakers highlighted that trainees need to learn the invaluable skill of telling their research or career story to a lay person in thirty seconds or less. The speakers also encouraged trainees to contact their Institution's career centers and explore alumni contacts when planning the next career steps.
- **Executive Session:** The executive session hosted by Tina Gatlin, NHGRI, and Treva Rice, DACC, and was attended by advisors, PIs, training coordinators, NHGRI staff and other DACC members. This session is **summarized in Appendix B: Business Section**.

CLOSING REMARKS

- Bettie Graham, NHGRI, encouraged trainees to stay focused on their work and in their research intentions and reminded trainees that NHGRI is available as a resource for trainee's questions. She reiterated NHGRI's commitment to increasing the number of URMs participating in research, nothing the importance for trainees and mentors to not leave the recruitment and retention of URMs to their Institution's office of diversity, but to instead act now and locally within their own Programs. Bettie acknowledged that the Annual Meeting would not be possible without the participation of the trainees, speakers, the DACC, and NHGRI.

TRAINEE PARTICIPATION

Presenting Research: In addition to the structured sessions listed above, trainees presented their research either as an oral platform talk or as a poster. Of a total of 144 abstracts submitted, 140 were

actually presented at the meeting (4 trainees who submitted abstracts for presentation were later unable to attend the Annual Meeting).

Final number of Abstracts presented at the meeting	140**
Presented as Oral Platform Presentation	15
Presented as Poster	130
**65 of the 140 Abstracts presented at the meeting wished to be considered for the potential of being chosen as a Platform Presentation	

The program committee was extremely impressed with the quality of the trainees' presentations and the potential impact that their work may have on the future of research in the genomic fields. While the program committee would have liked to provide an award to every trainee, the following individuals were singled out as providing noteworthy presentations.

- **Oral Platform Talks:** A panel of expert judges (from the ranks of the DAP and T32 PI's as well as the ELSI members of NHGRI) considered 65 abstracts for oral presentations. New this year was increase the number of trainee talks by five and hold three platform talk sessions, one session for each major field of research supported by NHGRI: genomic sciences, medicine, and ELSI. The following 15 trainees were chosen to give oral platform talks. Congratulations to the following presenters:
 - Genomic Science Trainee Platform Talks (Moderator: Emily Holzinger, NHGRI)
 - **Nicole Teran:** ChAR-Seq: A New Method for Discovering Chromatin Associated RNAs
 - **Jacob Luber:** Elucidating Horizontal Gene Transfer Events in Community Metagenome Samples Using Ultra Long Reads
 - **Eugene Gardner:** Full-Length LINE-1 as a Driver of Human Genetic Diversity and Disease
 - **Amy Wilfert:** Genome-wide Significance Testing of Genetic Variation from Single Case Exomes
 - **Robin Kirkpatrick:** Programmable Tools to Rewire the 3D Spatial Organization of the Genome
 - Genomic Medicine Trainee Platform Talks (Moderator: Malachi Griffith, Washington University in St. Louis)
 - **Nicole Rockweiler:** Whole-body Maps of Somatic Point Mutations in 550 Healthy Individuals
 - **Jesse Cochran:** Rapid In Vitro Evaluation of Genes of Uncertain Disease Significance Implicated in Gene Expression and Seizures from Patients with Developmental Delay and/or Intellectual Disability
 - **Ryan Collins:** Defining the Spectrum of Large Inversions, Complex Structural Variation, and Chromothripsis in Autism
 - **Erin Young:** KRT16 Germline Mutation Associated with Familial Syndrome of Tylosis with Esophageal Cancer (TOC)
 - **Adam Brown:** Enabling Reproducibility in Computational Drug Repositioning
 - ELSI Trainee Platform Talks (Moderator: Danton Char, Stanford University)

- **Aaron Neiman:** The Politics of Approximation: An Epistemological Critique of the Hardy-Weinberg Equilibrium
 - **Christi Guerrini:** IP and Genomic Citizen Science: Who Owns Research Outcomes?
 - **Martine Lappe:** Researcher Perspectives on the Social and Ethical Implications of Epigenetics
 - **Anya Prince:** Should Life Insurers Use an Applicant's Genetic Information?: Policy Lessons from the UK, Australia, and Canada
 - **Kurt Christensen:** Short-term Costs of WGS in Cardiology and Primary Care: Findings from MEDSEQ
- **Posters:** The *poster presentations* provided an opportunity for trainees to present their research and answer questions from their peers and other scientists. This year's poster sessions were held in one large room across two sessions, morning and afternoon, paired with snacks, to promote a networking session among all meeting attendees. A panel of poster judges reviewed 130 posters and chose the top posters in each of 3 career levels based on the visual presentation of the poster and the potential for the trainee's science to make a significant impact on the field. Congratulations go to the following trainees for their noteworthy posters:
 - **Undergraduate**
 - 3rd Place: **Jessica Arozqueta**, Fezf2+ Neuronal Stem Cells Contribute Distinct Subtypes of Olfactory Bulb Interneurons
 - 2nd Place: **Emily Alden**, Single Stranded Transposon Insertion
 - 1st Place: **Yarid Mera**, Establishing Dilated Cardiomyopathy Mice Model to Study Gene Therapy Techniques
 - **Graduate**
 - 3rd Place: **Jacob Pfeil**, Explaining Outlier Gene Expression Levels in Pediatric Brain Tumors
 - 2nd Place: **Rebecca Wilbanks**, Biohackers in science and fiction: a literary pre-history of synthetic biology
 - 1st Place: **Ariel Gewirtz**, Constructing Tissue-Specific Gene Co-Expression Networks through Bayesian Biclustering
 - **Post -Graduate**
 - 3rd Place: **Amanda Pendleton**, Selective sweeps highlight pivotal role of neural crest development in early dog domestication
 - 2nd Place: **Furqan Fazal**, Mapping the spatial organization of RNA genome-wide in mammalian cells
 - 1st Place: **Danton Char**, How clinicians caring for critically ill children with congenital heart disease anticipate and perceive the impact of whole genome sequencing (WGS) on acute care choices
 - **Networking:** In addition to formal presentations (posters and oral talks), other opportunities were provided throughout the meeting to encourage trainees to network. Networking activities included both structured and informal opportunities to talk about research and to interact with other investigators (peers and program PIs, mentors and coordinators) interested in similar research questions, opening the door for developing collaborations among participants.

- **Opening Reception:** A casual 90-minute reception was held in the Zodiac Ballroom on the 11th floor. Passed appetizers, food stations, and a cash bar were provided in the early evening the day before the meeting began. Meeting attendees had the opportunity to network inside the Ballroom or outside on the balcony while enjoying the nice weather and viewing local points of interest.
- **Networking Lunch (by Scientific Discipline):** This session was held on the first day. Each of the attending program PIs, training mentors and NHGRI researchers were free to sit at tables assigned by Scientific Discipline (Genomic Medicine, ELSI, and sub disciplines of Genomic Sciences). Trainees were asked to sit and talk with other trainees and Investigators at tables whose research was of interest to the trainee.
- **Happy Hour:** An informal and casual well-attended networking happy-hour was held at the Drunken Fish at the end of the first full day of meetings.