

# Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

## Annual Meeting Report ([genome.gov/iscc](https://genome.gov/iscc))

Friday, April 5, 2024, 8:30am – 5:00pm ET

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### Executive Summary:

- Dr. Rich Haspel is stepping down as co-chair after seven years of service. NHGRI will be seeking a new external co-chair shortly.
- ISCC-PEG Scholars Program has now started Class #4. Following introductions by their ISCC-PEG Mentors, Class #3 Scholars presented their work, including:
  - Ava Willoughby (mentored by Barbara O'Brien, M.D.) on reproductive genetic counseling for high-risk individuals, and
  - Molly Felix (mentored by Vinaya Murthy, MPH, M.S., LCGC) on language access and the genetic counseling experience.
- NHGRI's Education and Community Involvement Branch is starting strategic planning activities for their Healthcare Provider Genomics Education portfolio and ISCC-PEG. Input was requested from all ISCC-PEG members and assistance with prioritizing high-impact activities for the future.
- ISCC-PEG Project Groups continue to be active in developing valuable resources for healthcare provider education on a variety of topics for different audiences.
- There was also the first meeting of a possible new Health Equity/Community Engagement Project Group, with collaboration from two existing Project Groups (LGBTQI+ Issues in Genomics; and Inclusion in the Practice of Genomic Medicine: Exploring the Impact of Implicit Biases Towards Disability) and NHGRI's Community Engagement with Genomics Working Group of NHGRI's Advisory Council (CEGWG).

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## **Session Notes:**

### **ISCC-PEG: Annual Meeting 2024 Roadmap**

**Rich Haspel, M.D.**

Vice Chair for Medical Education, Department of Pathology, Beth Israel Deaconess Medical Center  
Professor of Pathology, Harvard Medical School  
ISCC-PEG Co-Chair

This meeting marks an important milestone for ISCC-PEG, a collaborative group focused on genetics education for healthcare professionals. The group was founded around 2013 and has grown significantly over the years, now boasting 333 members from 61 organizations, 11 government institute groups, and 8 companies.

Dr. Haspel is stepping down from his role after serving since 2017. He emphasized the importance of this group in leveraging the diverse expertise and perspectives of its members to determine needs in genomics education and create useful resources. He highlighted the group's many accomplishments, including the development of 19 resources since 2020, and encouraged members to continue to be active in the various project groups.

The meeting is also an opportunity to strategize on the group's mission and future direction. Dr. Haspel discussed the importance of conducting needs assessments, considering dissemination plans for new resources, and leveraging the group's diverse membership to maximize the impact of their work. He encouraged members to get involved in presenting their work, applying for grants, and seeking publication opportunities.

### **NHGRI Welcome**

**Ben Solomon, M.D.**

Clinical Director, National Human Genome Research Institute

Dr. Solomon discussed the exciting developments happening in the NHGRI's intramural research program, which conducts a wide range of clinical and translational genomics studies.

Some key highlights include new gene therapy and targeted molecular treatments being developed for rare genetic disorders, efforts to expand genomic sequencing capabilities to tackle "unsolvable" cases, and the application of artificial intelligence and machine learning to improve disease diagnosis and understanding. Dr. Solomon emphasized that genomics is now permeating all of healthcare, not just rare genetic conditions. He also notes the challenges around ensuring equitable access and representation in genomics research and AI algorithms.

Finally, Dr. Solomon shares his enthusiasm for the cutting-edge work happening at NHGRI, from high-profile breakthroughs like the rapid genome sequencing of a critically ill child, to more incremental

advances in areas like microbiome research and neuroimaging of ADHD. Overall, the presentation showcases the diverse and impactful genomics research being conducted at the NIH.

## **Keynote: Future Directions in Genomic Medicine**

**Teri Manolio, M.D.**

Division of Genomic Medicine Director, National Human Genome Research Institute

Dr. Manolio discussed the past and future directions of the NIH's Genomic Medicine Meetings, which have led to the development of important initiatives like ISCC-PEG, the ClinGen Consortium, the IGNITE program, and the International 100,000 Cohorts Consortium. These meetings have brought together various stakeholders to address challenges and advance the field of genomic medicine.

A major focus of Dr. Manolio's talk was on implementation research to improve the uptake and use of genomic innovations in clinical practice. She highlighted new programs, such as the development of a network of learning health systems to test and share strategies for implementing genomic medicine, as well as a peer-to-peer e-consult service to improve access to genomic expertise. Additionally, she discussed plans to pilot studies on population screening for certain genetic conditions.

Dr. Manolio also provided an overview of the NIH's training programs in genomic medicine, ranging from pre-doctoral to faculty-level opportunities. She emphasized the importance of these programs in building a workforce capable of advancing the field of genomic medicine.

## **ISCC-PEG Scholars Class 3 Presentations**

### **Provider Practices and Perceived Barriers towards Counseling on Reproductive Options for High-Risk Individuals**

**Ava Willoughby, MS, CGC**

Research Genetic Counselor, Nationwide Children's Hospital

Ms. Willoughby's study aims to investigate the reproductive options counseling practices of high-risk genetic providers, such as maternal-fetal medicine providers and prenatal genetic counselors, in both Ohio and Massachusetts. Willoughby and colleagues hope to compare practices between genetic and non-genetic providers, as well as identify barriers to comprehensive counseling. The study design involves a survey presented to approximately 20 genetic counselors and 60 maternal-fetal medicine providers, asking about their practices, attitudes, and any barriers they face when counseling patients on reproductive options for future or current high-risk pregnancies. They plan to analyze the survey data using descriptive and inferential statistics to answer questions about which options are most/least often discussed, any differences in counseling between provider types or scenarios, and the barriers identified. The goal is to use the findings to inform the development of interventions to improve reproductive options counseling and ultimately patient care.

The discussion touched on several key points:

- Considering the characteristics and perspectives of patients/families that may influence provider decisions, and potentially including an additional survey item on this.
- Rationale for selecting Ohio and Massachusetts as the study sites was accessibility.
- Consideration can be given to leveraging ISCC-PEG connections to expand recruitment.
- Importance of piloting the survey with a small, representative group to ensure the scenarios and questions are clear and eliciting the intended responses.
- Expanding the study to include more diverse provider types and practice settings, such as rural/community hospitals and assisted reproductive technology clinics, to capture a broader view of counseling practices.

## **Genetic Counselor's Adaptations and Perceptions in Language Discordant Patient Encounters**

**Molly Felix, BS**

MS Candidate in Genetic Counseling, Virginia Commonwealth University School of Medicine

Ms. Felix's research focused on understanding how language barriers impact patients receiving clinical genetic services. Through a survey of genetic counselors, Molly aimed to explore the specific strategies they use to navigate language discordance in their initial intake appointments, such as the use of interpreters, language adaptations, and visual aids.

The key findings from the survey include:

- Genetic counselors reported frequently using various interpreters, with telehealth interpreters being the most common. They expressed a strong preference for in-person interpreters over using family/friends.
- While most genetic counselors reported positive experiences with interpreters, many also experienced frequent frustration.
- The most consistently used adaptation was increasing the duration of the appointment, while the use of visual aids was relatively less common.
- A concerning finding was that 25% of genetic counselors felt their patients did not have a good understanding of their genetic testing options by the end of the appointment, despite the counselors' efforts.

The discussion highlighted the need for more training and resources to help both genetic counselors and interpreters better communicate complex genetic information. Suggestions included developing curricula or "cheat sheets" for genetic counseling programs, collaborating with interpreter organizations, and potentially offering training sessions at the annual genetic counseling conference. The presenter acknowledged the limitations of the quantitative survey and expressed interest in conducting more qualitative research to better understand the barriers and challenges faced by both parties in these language-discordant encounters. Potential intersections with other ISCC-PEG Project Groups were encouraged.

## Strategic Planning

NHGRI's Education and Community Involvement Branch, under the direction of a new Branch Chief, Beth Tuck, is undertaking a new strategic planning effort to align branch activities with the institutional strategic vision and the action agenda for a diverse genomics workforce. The brainstorming session gathered ideas for the future direction of the ISCC-PEG group and the broader work of the NIH/NHGRI/ECIB team, with a special focus on opportunities to have a "multiplier effect" - developing programs and initiatives that can reach beyond just the immediate community. In-person participants discussed, while staff took real-time notes; and virtual participants helped populate a digital whiteboard.

Suggestions included:

- Develop case studies and educational materials focused on specific medical specialties to make genomics more accessible.
- Address common misconceptions about genetic testing.
- Improve collaboration and avoiding duplication of efforts across different groups.
- Incorporate more patient and community perspectives to understand their needs and priorities.
- Leverage the expertise of different healthcare disciplines like molecular pathologists.
- Reevaluate and update the "Common Core Principles of Genomics" - the foundational teaching resource. Ensure materials are available on the website.
- Explore the feasibility of developing a certification framework or board exam for genomics education. Engage with professional societies to gauge interest and support.
- Facilitate stronger interdisciplinary connections between healthcare providers around genomics education and implementation. Leverage the diverse membership of ISCC-PEG to make these connections.
- Establish a more formalized system to notify members about meetings, conferences, and opportunities to present or participate.
- Conduct needs assessments to better understand the priorities and pain points of different stakeholder groups (e.g. clinicians, patients, educators). Use this to inform future initiatives.
- Experiment with new program models, such as one-day retreats, to find the most impactful and feasible approaches.
- Develop best practices and strategies for ISCC-PEG members to effectively reach out and collaborate with external organizations.
- Continuously focus on cross-pollination and involvement of diverse audiences within ISCC-PEG projects and initiatives.
- Leverage the ISCC-PEG listserv as an important communication tool.
- Identify specific, actionable projects that can move the group's work forward in a measurable way.
- Language-accessible materials
- Train-the-trainer models for clinical educators
- Considering geographic accessibility, materials for rural communities
- Using AI intelligently, point-of-care clinical decision support
- Short Course with evaluation/certificate of completion
- Clarify/organize ISCC-PEG around a central goal
- More small group discussions with targeted questions relevant for all – e.g. needs assessment within your organization, how different groups can work together
- Regular updates to GenomeEd

- Bring in communication specialists, implementation scientists, payers/insurance, end-users of the products
- Patient advocacy, equity in access to genetic testing services
- Missing Specialties: Occupational therapy, Social Work, Physical Therapists, Dietitians, Speech/Language Therapist, Psychologists, more public health
- Content needs to be short - “snippets” of information to engage
- Conference attendance (travel support)

#### Wish List Items:

- Use of the NHGRI YouTube Channel, social media platforms
- Contribute to website content development
- Maintenance of Certification Channels?
- Publish on Up-to-Date, MedScape
- Video series – interviews about using genomics in practice
- CME/Education workshops using participants own PGx data
- ECHO Model – regional hubs, partnerships and career pipelines
- MOOC

The team will synthesize the ideas from this discussion and work to prioritize and implement new strategic initiatives going forward.

### **Project Group Breakout Sessions & Summaries**

Prior to the breakout sessions, ISCC-PEG Co-Chair Rich Haspel provided some guidance to help frame Project Group discussions. Members were reminded to consider a needs assessment, how the final product(s) will be disseminated, what organizations could be involved to support it, and potential funding sources like grants. When developing educational content, members should also plan for evaluation and publication opportunities, as this can help get the work recognized. Finally, Dr. Haspel reminded participants of the diverse membership of ISCC-PEG that can and should be leveraged to help with things like reaching out to relevant specialty groups or organizations that could be involved. Project groups were reminded to keep conflict of interest considerations in mind, especially if the content is intended for continuing medical education (CME), where industry representatives cannot be permanent members of the committee creating the curriculum.

#### **Project Group Breakout Sessions:**

- Direct-to-Consumer Genetic Testing - <https://bit.ly/ISCC-PEG-DTC-GT>
  - Create packaged cases for specialty-related society for CME
  - Cross-pollinate with other Project Groups
  - Needs buy-in from specialty societies
  - Need ISCC-PEG “central” organization to oversee specialty-specific cases and which project group contributes content to which case (e.g. create a case “database” or spreadsheet)
- LGBTQI+ - <https://bit.ly/ISCC-PEG-LGBTQI>
  - Working on Sexual Orientation and Gender Identity (SOGI) Collection Recommendations

- o Goal to publish educational resources on genome.gov
- Nursing Genomics - <https://bit.ly/ISCC-PEG-NursingGenomics>
  - o Manuscripts in progress
  - o Considering dissemination efforts for FAQ, Expert Commentary, Q&A
- Pharmacogenomics - <https://bit.ly/ISCC-PEG-PGx>
  - o Dissemination efforts for PGx Learning Modules
- Physician Assistants - <https://bit.ly/ISCC-PEG-PA>
  - o New Group! Get PAs to participate in other Project Groups, others (non-PAs) are encouraged to join this group
  - o AAPA Conference, Public Health Genetics Week, PA Week
- OB/GYN - <https://bit.ly/ISCC-PEG-OB-GYN>
  - o Working on Curriculum Modules for OB/GYN residents
  - o Refine cases, pilot test and validate
- Rare Diseases - <https://bit.ly/ISCC-PEG-Rare>
  - o Many workshops completed over the past year, including “Train-the-Trainer” approaches; with several ideas for future meetings to consider for presentations
  - o The Rare Disease Genomics Education Curriculum developed by this Project Group can be translated for different audiences and specialties
  - o Several new members in the Rare Disease Group, adding to the multidisciplinary nature of the group
  - o Needs assessments are planned as future activities to cater the Group’s curriculum further with different specialties
- Health Equity/Community Engagement (Beth) - <https://bit.ly/ISCC-PEG-HealthEquity>
  - o Enlist ISCC-PEG members to share community engagement resources and initiatives from professional societies – help identify high priority unmet needs
  - o Dedicate time at upcoming plenary call to discuss, curate resources
  - o Survey to listserv, ISCC-PEG members to share with colleagues
  - o Anyone tech savvy enough to help make a web crawler that could scan for resources?
  - o Opportunity to collaborate with NHGRI’s Community Engagement with Genomics Working Group (CEGWG), goal to complete paper by 2025
  - o Possible opportunities for integration with other Project Groups/Scholar projects