Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC) Meeting Minutes

February 25, 2020

National Human Genome Research Institute (NHGRI)
National Institutes of Health (NIH)
Natcher Conference Center
Bethesda, Maryland

Attendees: 78 registrants (47 external; 16 from NHGRI; 15 from other NIH institutes)

Welcome and Introductions (slides): ISCC-PEG Co-Chairs Richard Haspel, M.D., Ph.D. (Beth Israel Deaconess Medical Center) and Carla Easter, Ph.D. (National Human Genome Research Institute

Dr. Rich Haspel highlighted the goals and progress for ISCC-PEG, including putting the "work back in teamwork" and continuing to create resources and support project groups. He also relayed some logistics, including introducing the new dissemination and review policy, the need for a new logo, and summarized the new google drive available to ISCC-PEG. He concluded with future plans, expanding membership, spreading the word about ISCC-PEG, continuing current working groups and creating new ones, and expanding strategic planning and funding.

Keynote: Tools for Public Education about Research Ethics, Jeffrey Botkin, MD, MPH, Professor of Pediatrics and Adjunct Professor of Human Genetics and Internal Medicine at the University of Utah (slides)

Dr. Jeffrey Botkin, MD, MPH, delivered the keynote address summarizing barriers to population screening and biobanking. His presentation also included a listing of available tools for public education in genomics. After addressing the many reasons that population screening is important, he highlighted a case study of biobanking that illustrated issues such as ethical questions surrounding informed consent and bloodspot collection in different states with different laws. Citing the importance of assessing the public attitude to developing acceptable public policies, Dr. Botkin summarized a research study using a video on newborn screening and residual bloodspot retention followed by a focus group with expert moderators. Some of the significant findings to be considered were that simple messages and brevity worked best, with an appropriate reading level for low literacy and a balanced set of information useful to the public. Dr. Botkin finished by asking "the important question": will more information make people more or less supportive of newborn screening? Several studies were presented to answer this question, with the basic conclusions being that video and interactive tools work best, people want to be informed and want to have a choice, and enhanced information usually increases support for the programs.

Comments and Questions

What do parents willing to consent their samples for "important research" define as "important"? What is stigmatized?
There was not that level of detail in the video nor from the focus groups. There was, however, a specific concern against uses in industry. At the federal level, selected senators have paid attention to this issue and debated the need for "human subject" consideration for de-identified samples.

What are the different needs and interests by region? Is it possible to create one video addressing a topic, or does it need to be targeted to specific issues of interest of a region?
They are working on this. Right now, they are trying to keep it general.

The importance of diversity in the videos and focus groups was also discussed (language, ethnicity; the importance of local stories).

Extramural Training Opportunities/News: Heather A. Colley, M.S., Program Director, Division of Genomic Medicine, NHGRI (slides)

Heather Colley highlighted the many different extramural training and funding opportunities available to researchers in basic and clinical genomics research and genomic research education.

- Institutional Training Grants (T32)
  - Pre and postdocs
  - Basic and clinical research but not clinical care of patients
  - Essential ELSI component
  - PA-18-403 and NOT-HG-20-003
- R25: NHGRI diversity action plan
  - Expose under-represented minorities students to genomics
  - Undergrads, post-bacs, grad students
  - Career development
  - PAR-19-380
- R25: Genomic research education courses
  - Supports short term/long term development programs
  - Provides innovative, SOTA, evidence-based education
  - Doctoral plus
  - Up to 5 years of support
  - PAR-19-185
- PAR-19-185Protected Research Time
  - K08 clinical investigator development award
    - For genomic medicine
  - K01 research scientist development award
    - For genome science and ELSI
  - K99/R00 pathway to independence award
  - K43 emerging global leaders award
    - Supports research scientists from LMIC countries
- Fellowships
  - F30 MP/PhD or dual degree
  - F31 predoc
  - F32 postdoc
  - F33 senior fellows
- Promoting workforce diversity
  - Administrative supplements: 2 years support
  - NOT-HG-19-010

Heather Colley also addressed the findings of the NHGRI Training task force that was charged to evaluate current extramural training efforts. Their recommendations for improvement included increasing budget, data science, diversity, and modular training:
https://www.youtube.com/watch?v=Ix_-7PWXQZE
Report of the NHGRI Training and Education Task Force, October 22, 2019:
https://www.genome.gov/sites/default/files/media/files/2020-02/NHGRITrainingTaskForceReport_October22_2019.pdf

Comments and Questions
Do R25s apply to people like genetic counselors or others without a PhD or MD?
May be able to use R25s to help in this category.

Has the R25 funded courses yet?
Yes, but they aren't published yet.

Ethical, Legal and Social Implications Research Program: Dave Kaufman, Ph.D., Program Director, Division of Genomics and Society, NHGRI (slides)

Dave Kaufman, Ph.D., began by giving an introduction to the NHGRI ELSI program, which was established in 1990 and receives 5% of NHGRI's extramural budget, 50% of which is distributed between research priority areas of genetic and genomic research, genetic and genomic medicine, and legal policy and societal issues. Past issues addressed have been informed consent, incidental findings, understanding of results, research/clinical barriers, while emerging issues include gene editing, personal utility, direct-to-consumer genetic testing therapy, alternatives to in-person counseling, obligations and practices, equity and diversity, and clinical genomics. Dr. Kaufman went on to encourage researchers from many disciplines to get involved in ELSI research, citing ELSI applicants have represented disciplines such as ethics/bioethics, law, philosophy/humanities, public policy, medicine, biomedical science, public health, and social sciences. He concluded by highlighting ELSI funding mechanisms such as the R01 (3-5 years, <$500K direct costs annually), R21 (2 years, $275K total), and R03 (2 years, $50k direct costs per year). Funding announcements and all other information can be found at http://www.genome.gov/elsi.

Comments and Questions
Does ELSI fund education projects?
Straight development of curricula and dissemination are not fundable. It has to be research and assessment. It would fund education research with a rigorous scientific assessment framed around a research question

Organization Members and ISCC Collaboration: Robert Ostrander, MD, SUNY Upstate Medical University, representative from American Academy of Family Physicians (AAFP) (slides)

Dr. Robert Ostrander, MD, discussed his experience with project groups working hard and generating ideas that never got past that phase, cycling and recycling as membership turns over and new variations are introduced. He continued by summarizing possible approaches to breaking the cycle and moving forward. He also provided a brief overview of his personal trajectory, how he came to view genomic education of physicians to be extremely important, taking advantage of opportunities to have an impact. He discussed broadly the stimuli and barriers to improvement. Finally, as an example, he highlighted a project where the ISCC-PEG Pharmacogenomics Project Group, with him as the initial intermediary, is collaborating with AAFP to develop a pharmacogenomics educational module around the use of clopidogrel, highlighting how that collaboration came to be. It is near completion and going to the implementation, dissemination and assessment phase. The vision is to create a series of similar modules across the genetic/genomics domains with the audience being primary care physicians.

Comments and Questions

Can you list some opportunities you would like to see developed in a genomics educational module?
ELSI piece, newborn screening, prenatal genetic testing, cancer genomics in terms of risk an enhanced screening, cancer subtypes with treatment choice, chronic condition.

Could you provide context or perspective regarding the competition for attention of healthcare provider education? There are a lot of modules, how do you develop a competitive tool that people will use?
Consider the audience/reach and correlate that to impact. Make it clear that this is important, and sell that to people who are behind this effort.

How do we get materials on genome.gov? What should be considered regarding branding and endorsement?
It’s an ongoing discussion.

Any additional comments?
Finding technical expertise was difficult for Project Inclusive Genetics and should be something to consider for other groups.

ISCC Business: Discussion of ISCC-PEG Description and Draft “Policy on Review of Materials and Dissemination” document: Richard Haspel, M.D., Ph.D. and Carla Easter, Ph.D., ISCC-PEG Co-Chairs

Anything mentioning ISCC-PEG should be given to the ISCC-PEG Co-chairs to review. Co-chairs can also suggest outside review. Any submissions should have to include the prewritten disclaimer, and if it is developed by ISCC-PEG it needs to be made available for free.

Project Group Meetings

Direct-to-Consumer Genetics Testing (DTC-GT)

This group began by going the over the main project: FAQ website for healthcare providers. They went over content, worked on defining the target audience, and worked on where it would be housed. There was also discussion on how to address regulatory challenges such as avoiding giving medical advice. The group also discussed the development of a point of care flow chart tool.

Inclusive Genetics

Time was given to complete the online module at Bucme.org/activity/8001 . Next, the group went over the preliminary data on the existence of explicit and implicit bias in providers and whether it varies by disability or provider. They also expressed their need to get data from other provider demographics. Finally, they reviewed whether the module simply assesses bias, or if it actually helps reduce bias in the providers upon completing the module.

Pharmacogenomics (PGx) (slides)

The group discussed various topics including the introduction of a new focus group on psychiatry, genomics and pharmacogenomics, creation of a universal framework from idea to action for developing educational materials, and how to handle endorsement of materials.

There was also discussion of new projects for PGx to add to the group of modules for the collaborative effort with AAFP. Topics include newborn screening, cancer genetics, cancer treatment, chronic condition genetics, approaches to patients with undiagnosed diseases, and how to deal with direct-to-consumer genetic testing.

Rare Diseases (slides)

This group discussed project goals and focused on developing dissemination methods, including targeting specific professional demographics, raising awareness about available resources for rare genetic diseases, and working broadly to cover rare disease topics and strategies as opposed to focusing on individual diseases. The group also discussed utilizing the TRIG Universal Modules (http://www.pathologylearning.org/trig/resources) and trying to find an annual meeting to implement or involve residency training programs.

The group also discussed the preliminary work being done on a creative matrix project focused on making a diagnosis, treatment and management, referrals and specialty centers, and patient support.

Project Group Meeting Updates and “Asks” of ISCC Membership

G2C2 and G3C overview

The Genetics/Genomics Competency Center (G2C2) (https://genomicseducation.net/) is a searchable repository of genomics education resources primarily targeted to healthcare providers and educators. Previously, all resources were reviewed and vetted by the G2C2 Editorial Board and resources were mapped to published genomic competencies. Genomics educational resources submitted by ISCC-PEG societies and assembled in the Compendium will be recognized as having been reviewed by the submitting societies and will not undergo review by an editorial board. Selections from the Compendium will be added to the G2C2 repository using this format:

> Compendium: Society Name: Resource Title
> Format / CME/CE (Yes or No) / Free or Paid

This format will allow G2C2 users to differentiate between resources submitted through the ISCC-PEG Compendium and those submitted through review by the G2C2 Editorial Board.

The Global Genetics and Genomics Community (G3C) (https://www.genomicscases.net/en) contains educational resources contributed to by ISCC-PEG members. Interactive video-based cases provide information about basic genomic concepts used in a variety of genomic healthcare applications.

Project Group Chairs have the following "Asks" of ISCC-PEG Membership:

- DTC-GT asks for ISCC collaboration on appropriate channels for dissemination of resources. How have people had dissemination/upkeep success? How to make this information available to all?
- Inclusive Genetics asks for advice and help in expanding the reach of the module to more professionals in various medical professions and positions.
- Pharmacogenomics did not have any specific asks.
- Rare Diseases asks for advice on ensuring program success, longevity, and dissemination.

General discussions and suggestions for ISCC

ISCC-PEG Scholars

ISCC-PEG would like to add a student group. The membership expressed great enthusiasm for this idea and mentioned that genetic counseling and physician's assistant students would be especially interested. A potential name of this group could be "ISCC Scholars", with possible NHGRI funding. The idea would be to have the students within a project group and have a faculty sponsor associated with each student. This student would be tied to a particular effort that the project group would be working towards. For consideration: Should there be an application for the "ISCC Scholars" or should new members be paired with a mentor before being able to join? The group suggested admitting a few students each year who apply and give an example of a project to work on, and then potentially fund them to come to meeting.