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FRANCESCA SPINOSI, MS, CGC, **MEMBER-AT-LARGE**

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January Meeting Schedule

Meeting Date: Tuesday, January 14, 2025
Meeting Registration: [Registration Link](#)

- 10:00 – 10:30 AM **Executive Committee Meeting**
CLOSED MEETING - Executive committee only
- 10:30 – 11:30 AM **Concurrent Committee Meetings**
Everyone is encouraged to join a committee.
Education Committee
Professional Issues Committee
Public Relations Committee
Community Outreach Committee
- 11:45 AM – 12:45 PM **New Advances in Inborn Errors of Metabolism**
Rebecca Ahrens-Nicklas, MD, PhD
Children's Hospital of Philadelphia
- 12:45 – 1:30 PM **General Membership Meeting**
President's Report
Secretary's Report
Treasurer's Report
Introduction of Members
Committee Reports
New Business
- 1:30 – 3:00 PM **Where Biology Ends and Bias Begins***
Shoumita Dasgupta, PhD
Boston University
- 3:00 – 4:00 PM **CRISPR for Variant Classification: An Introduction to MAVEs and Their Impact on Genetic Testing**
Catherine Mayo, MS, CGC
Ambry Genetics
- 4:00 PM **Adjournment**

* *The presentation is a discussion of medical ethics and fulfills the requirement for genetic counseling licensure in New Jersey.*

This event has been submitted to the National Society of Genetic Counselors (NSGC) for approval of 0.35 Category 1 CEUs. The American Board of Genetic Counseling (ABGC) accepts CEUs approved by NSGC for purposes of recertification. Approval for the requested CEUs and Contact Hours is currently pending.



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Summary of Educational Presentations

New Advances in Inborn Errors of Metabolism

Speaker: Rebecca Ahrens-Nicklas, MD, PhD

11:45 AM – 12:45 PM (60 minutes, 1 contact hour)

Objectives:

1. Advance the understanding of how novel gene therapies may be developed for rare disease patients.
2. Examine challenges to widespread, equitable implementation of novel therapies.

Summary:

Novel approaches to gene therapy for patients with inborn errors of metabolism will be discussed.

Where Biology Ends and Bias Begins*

Speaker: Shoumita Dasgupta, PhD

1:30 – 3:00 PM (90 minutes, 1.5 contact hours)

* *The presentation is a discussion of medical ethics and fulfills the requirement for genetic counseling licensure in New Jersey.*

Objectives:

1. Recognize that questionable eugenic principles were used to justify forced sterilizations in recent American history and that similar ideas could be applied to modern genetic technologies.
2. Define disparities in health and health care at the population level and recognize the multiple roots of those inequities including the impact of racism and biases as well as potential solutions.
3. Articulate personal spheres in which anti-oppressive actions can be implemented.

Summary:

This presentation will aim to foster dialogue on issues like racial justice, reproductive rights, LGBTQ+ equity, and disability inclusion—integrating the foundational science that dispels harmful misconceptions. Methods to do this by disentangling social and biological identities and considering ways in which to apply this framework to our professional activities will be explored.

CRISPR for Variant Classification: An Introduction to MAVEs and Their Impact on Genetic Testing

Speaker: Catherine Mayo, MS, CGC

3:00 – 4:00 PM (60 minutes, 1.0 contact hours)

Objectives:

1. Summarize the role of CRISPR in MAVE design and describe the clinical impact of MAVE studies on variant interpretation
2. Differentiate a Multiplex Assay of Variant Effect (MAVE) from a traditional/low-throughput functional assay.

Summary:

This presentation will review the concept and application of multiplex assays of variant effect (MAVEs), including the role of CRISPR in the study design, in the context of variant interpretation and clinical genetic testing. The talk will also explain the difference between MAVEs and traditional, low-throughput functional assays.

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*All talks/modules above are presented via PowerPoint presentations and sound. The presenters have visual aids and key points listed on their PowerPoint presentations.*

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