

Semi-Annual Colorado Genetic Counselors Symposium

Date: May 10, 2024

Time: 8am - 4:30pm

Location: Children's Hospital Colorado Conference Center

Zoom Link to be provided upon registration.

This Colorado Genetic Counselors Symposium aims to provide an educational review and update regarding genetic counseling practice and current educational issues. Presentations are given by practicing genetic counselors and professionals actively involved in the local and national genetic counseling community.

This event has been submitted to the National Society of Genetic Counselors (NSGC) for approval of 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.

PLATINUM SPONSOR

The logo for GeneDx, featuring the word "GeneDx" in a blue, sans-serif font. The "D" is stylized with a blue outline and a white fill.

GOLD SPONSOR



Ambry Genetics®

SILVER SPONSOR

The logo for COMBINED, featuring the word "COMBINED" in a blue, sans-serif font. The letter "O" is replaced by a stylized orange sun or cell icon with a central dot and radiating lines. An orange arrow starts from the top of the "O" and curves upwards and to the right.

8:00-8:45 An Overview of the Undiagnosed Disease Network- The Duke Experience

Kelly Schoch, MS, CGC & Rebecca Spillman MS, CGC

We will provide a brief history of the Undiagnosed Diseases Network, and more specifically the experience of the Duke Clinical Site. We will describe the diagnostic approach and application of new genomic technologies for patients and explore feedback from participants on the experience of living with an undiagnosed illness.

Learning Objectives

Describe the diagnostic approach and application of new genomic technologies for patients in the Undiagnosed Diseases Network.

Explore feedback from participants on the experience of living with an undiagnosed illness.

8:45-9:30 Genetic Counseling Roles in Industry

Breanna Roscow MS, CGC

Genetic counselors often practice outside of the clinical sphere, integrated into commercial and industry settings, facing unique professional challenges. In reflection of training competencies, often designed primarily for clinicians, further discussion of aspects applicable to both worlds would more adequately prepare genetic counselors for industry roles and employment opportunities.

Learning Objectives

Illustrate the roles held by genetic counselors in a variety of commercial and industry settings. Examine the effectiveness and availability of genetic counseling training in relation to industry roles.

Review the key competencies of genetic counseling and how they are applied in industry-based roles.

9:30-10:15 One Genetic Counselor's Experience Working at a Rare Disease Foundation

Casey McKenna, MS, CGC

After 10 years of clinical practice, Casey decided to move out of direct patient care to pursue alternative roles that continue to support the genetic disease community. With Casey's career path as a backdrop, this presentation will explore the history of genetic counselors working in the non-profit sector and the many professional roles suited by the genetic counseling skill set. Work within rare disease foundations will be highlighted, including a personal narrative of the challenges and successes faced in the transition.

Learning Objectives

Summarize the history of Genetic Counselors working in the non-profit sector using the Professional Status Survey.

Examine Genetic Counselor competencies and their overlap with disease advocacy work.

10:15-10:30 Break

10:30-11:15 Partnerships with Patient Advocacy Groups Katie Angione, MS, CGC

Patient advocacy groups (PAGs) provide education, advocacy, and support to patients and families with a specific disorder or groups of disorders, raise awareness and funding, and promote research to better understand, prevent, or treat that disorder. Many rare-disease PAGs are started and run by parents, after receiving a diagnosis for their child and connecting with other families from across the country or across the world. Genetic counselors can partner with PAGs in clinical, research, and educational settings in order to further their shared goals of awareness, advocacy, and specialized, comprehensive care. Katie Angione is a genetic counselor who sees patients in neurogenetic multidisciplinary clinics supported by PAGs, partners with these groups in rare-disease research, and regularly attends and speaks at disorder-specific conferences and family meetings. Katie will speak to her experience working with PAGs and how it has shaped her role as a genetic counselor in the rare-disease space.

Learning Objectives:

Identify areas in which genetic counselors can partner with patient advocacy groups.
Discuss how roles outside the clinic can shape one's career path.

11:15 - 12:15 EpiSign: Methylation Understood Robin Fletcher, MS, CGC and Matt Tedder, PhD

The clinical validation and implementation of DNA methylation episinature testing has provided a complementary approach to traditional DNA-based genetic testing. This methodology utilizes a genome-wide methylation array to identify robust and reproducible methylation patterns that have been identified in over 90 genetic disorders. Additionally, this assay can detect imprinting abnormalities and Fragile-X syndrome. This lecture will provide an overview of DNA methylation episinature testing, the strengths and limitations of the technology, and the clinical indications for testing. Clinical case examples will be discussed to demonstrate the utility of this approach and highlight the points discussed.

Learning Objectives

Examine epigenetics and methylation as a disease mechanism
Illustrate translation from research to clinical usage
Highlight the utility of Genome-Wide Methylation Analysis through case examples

12:15 – 12:45 LUNCH BREAK
SPONSORED BY:



12:45-1:45 Exome-Based Testing for Patients with Seizures: Advantages over Panel-Based Testing

Kirsten Kelly, MS, CGC

Epilepsy genetics is a rapidly developing field, in which novel disease-associated genes, novel mechanisms associated with epilepsy, and precision medicine approaches are continuously being identified. Currently, there are numerous genetic testing options available for individuals with unexplained epilepsy. Recently, the American Epilepsy Society (AES) endorsed the 2022 National Society of Genetic Counselors (NSGC) practice guideline which recommends exome/genome sequencing and/or a multi-gene panel (>25 genes) as first-tier testing for individuals with unexplained epilepsy. At GeneDx, we conducted a systematic review of our internal data to answer the questions: What is the diagnostic yield of exome sequencing for epilepsy in a clinical diagnostic laboratory? How do exome sequencing results compare to expected results for commercial epilepsy panels? Overall, this session seeks to empower genetic counselors with the latest advancements in genetic testing for unexplained epilepsy, enabling them to optimize patient care, navigate regional considerations, and contribute to advancements in the field.

Learning Objectives

Describe the utility of exome sequencing (ES) as a first-line diagnostic tool for patients with unexplained epilepsy.

Compare and contrast the diagnostic advantages of ES over epilepsy panels.

Detail the wider availability and broader insurance coverage for ES and understand the implications for clinical practice

1:45-2:45 Current and Future Directions in Diagnosis of Rare Genetic Neurodevelopmental Disorders

Elizabeth Roundree, MBA

Parents of children with genetic neurodevelopmental disorders face a long diagnostic odyssey. Less than a third of patients are diagnosed and on average, it takes families 30 months to get a diagnosis from the time of symptom onset. However, early diagnosis is critical to achieving optimal patient outcomes. In this presentation, we will explore the early symptoms of rare genetic neurodevelopmental disorders, the current literature on diagnostic yield in symptomatic infants and children, and future directions for reducing the time to diagnosis. We will also review infant case studies, assess which infants should receive genetic testing, and learn the outcomes of genetic testing in these patients.

Learning Objectives:

Recognize the early symptoms of rare genetic neurodevelopmental disorders in infants.

Assess which infants should receive genetic testing (3) Decide the appropriate type of genetic testing for symptomatic infants

2:45-3:00 Break

3:00-3:45 A Look at Advocacy: The National Fragile X Foundation's Advocacy Day on The Hill
Susan Howell, MS, CGC

Advocacy work on behalf of rare genetic disorders matters. Advocacy impacts not only policies that affect families and patients living with rare conditions, but also funding for research that allows professionals to devote their careers to helping these patients and their families. This presentation will walk the audience through the experience of Advocacy Day on The Hill for the National Fragile X Foundation in February 2024, highlighting the facilitators, stakeholders, process, and outcomes.

Learning Objectives

1. Describe how preparation for and day-of Advocacy Day on The Hill for NFXF happens
2. Explain the “Asks” for Advocacy Day 2024 and why an organized effort matters
3. Examine how the Advocacy efforts over history has impacted families and patients with Fragile X and related conditions and professionals dedicated to these conditions

3:45 – 4:30 The Other Hats Genetic Counselors Wear
Kelsey Zegar, MS, CGC

Genetic counselors master a variety of skills in their work in genetic healthcare settings. Many of these skills are unique to the field yet are highly valuable in other positions that may not carry a title of “genetic counselor”. This interactive session will explore real-world examples of genetic counselors leveraging their experience and core competencies to serve in different capacities beyond the walls of a clinic or laboratory.

Learning Objectives:

Examine published literature and case examples from the Colorado community to understand roles that genetic counselors fulfill beyond “traditional” clinical work.
Illustrate practice-based competencies for genetic counselors that serve as transferable skills to other roles.

For information about our conference please visit:
<https://sites.google.com/view/cogc/home>