

# **Semi-Annual Colorado Genetic Counselors Symposium**

**Date: October 10, 2025**

**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 is approved by the National Society of Genetic Counselors (NSGC) for approval of 7.5 hours (0.75) Category 1 CEUs. The American Board of Genetic Counseling (ABGC) accepts CEUs approved by NSGC for purposes of recertification.

## **EVENT SPONSORS**



Special Thank You to Natera as a Featured Speaking and Lunch Sponsor

## **8:00-9:00 Understanding Genetic Aortic Disease**

**Leslie McCallen, MS, CGC (In person)**

What is the aorta and what does it do? Why should a non-cardiac genetic counselor care? This presentation will define some basic concepts for those of us who do not work in a cardiovascular clinic so you can better understand these issues if they come up with your patients, and differentiate what to be concerned about and why.

### Learning Objectives:

- Identify high-risk clinical presentations which should be considered 'red flags' for cardiac genetics referral.
- Compare evaluation options including imaging, genetic testing, and family member screening.
- Contrast management strategies including medication, activity restrictions, and surgical interventions.

## **9:00-10:00 Anti-Fat Bias in Healthcare and Genetic Counseling**

**Mindy Simonson, MS, CGC (In person)**

This talk explores the pervasive issue of anti-fat bias in healthcare, with a particular focus on its presence and impact within the field of genetic counseling. It begins by defining the various types of anti-fat bias—explicit, implicit, and structural—and how they manifest in clinical settings. The presentation then reviews current evidence highlighting the prevalence and consequences of such bias in healthcare and genetic counseling practices. Finally, it outlines intersectional strategies to recognize and mitigate anti-fat bias in genetic counseling, promoting more equitable, patient-centered care.

### Learning Objectives

- Define the types of anti-fat bias.
- Summarize the current state of anti-fat bias in healthcare and genetic counseling.
- Identify intersectional strategies for mitigating anti-fat bias in genetic counseling.

## ***10:00-10:15 Break***

## **10:15-11:15 Genetic Counseling in Sickle Cell Disease: Navigating a New Era of Treatment and Hope**

**Barabara Harrison, CGC from Howard University (virtual)**

This talk explores the evolving role of genetic counseling in sickle cell disease (SCD) as advances in treatment—especially curative options like gene therapy, bone marrow transplantation, and CRISPR-based approaches—transform the clinical landscape. While SCD remains a significant public health concern with complex psychosocial and systemic impacts, recent breakthroughs demand updated counseling strategies that address not only inheritance and carrier screening but also patient education, decision-making, and long-term care planning. The presentation will dive into current and emerging therapies, including the implications of curative treatments on identity, access, reproductive planning, and survivorship. Culturally sensitive approaches and health disparities in SCD care will be highlighted throughout. Real-world scenarios will illustrate how genetic counselors can support patients and families in understanding complex treatment options, manage uncertainty, and advocate for equitable access to care.

### Learning Objectives

- Learning Objectives
- Describe the inheritance pattern and pathophysiology of sickle cell disease and its implications for family-based risk assessment.

- Summarize current and emerging treatment options for SCD, including gene therapy, hematopoietic stem cell transplant, and pharmacologic advances
- Examine genetic counseling approaches for individuals and families affected by SCD, including those considering curative therapies.
- Illustrate the psychosocial, ethical, and cultural considerations in counseling patients about treatment decisions, fertility, and long-term survivorship.

**11:15-12:15 +cfDNA to Confirmed Diagnosis: Understanding the Lived Experience of Parents Raising a Child with a Sex Chromosome Trisomy**  
**Kayla Molison, BS (in person)**

This presentation explores the real-world experiences of parents who receive a noninvasive prenatal screening (cfDNA) result suggesting a sex chromosome trisomy (SCT)—such as 47,XXY (Klinefelter syndrome), 47,XXX (Triple X syndrome), or 47,XYY—and go on to receive a confirmed diagnosis postnatally or prenatally. While cfDNA has transformed prenatal screening, the unique nature of SCTs—often variable in phenotype, under-recognized, and inconsistently understood—often results in a complex emotional and informational journey for families. Data collected from the eXtraordinary Babies Study, a natural history study of children with prenatally identified SCT, will illustrate patterns and themes in how parents raising a child with SCT navigate uncertainty, diagnosis, stigma, and long-term care planning. It examines how genetic counseling plays a critical role at each stage—from screening and diagnostic decision-making, to postnatal support and developmental monitoring. Emphasis will be placed on improving counseling strategies, enhancing psychosocial support, and addressing gaps in healthcare provider knowledge around SCTs.

Learning Objectives:

- Describe the typical diagnostic pathway from cfDNA screening to confirmed diagnosis of a sex chromosome trisomy.
- Examine the lived experiences, challenges, and emotional trajectories of parents raising a child with prenatally identified SCT.
- Identify areas of improvement for counseling for SCTs.

***12:15-12:45 Lunch Break & Announcements***

**12:45-1:45 Lunch Sponsored Talk:**  
**Beyond the Fetus: Maternal Health Insights from NIPT and Carrier Screening**  
**Katie Wiens, MS, CGC From Natera, Inc (in person)**

This presentation will examine unexpected maternal health findings that may arise through non-invasive prenatal testing (NIPT) and carrier screening, with a focus on their implications for genetic counseling practice. Topics will include incidental findings from NIPT such as maternal X chromosome mosaicism and atypical findings suspicious for maternal malignancy, along with current recommendations for clinical follow-up. We will also explore exceptions to the commonly communicated message that "carriers are typically asymptomatic," and the implications these findings can have on obstetric care and beyond.

Learning Objectives:

- Summarize incidental maternal findings that may be detected through NIPT, including X chromosome mosaicism and atypical findings suspicious for maternal malignancy.
- Identify conditions included on carrier screening panels that may have maternal manifestations during pregnancy and beyond.

## **1:45-2:00 Break & Announcements**

### **2:00 – 3:00 Long-Read Sequencing in Genomic Medicine Austin Larson, MD (in-Person)**

This talk introduces long-read sequencing technologies (such as PacBio and Oxford Nanopore) and their role in clinical and research genomics. Compared with short-read sequencing, long-read methods can span complex and repetitive regions of the genome, improving detection of structural variants, phasing, methylation abnormalities, and characterization of areas such as tandem repeats and GC-rich sequences. The presentation will review how they work, their strengths and limitations, and their current and emerging clinical uses in rare disease. Case examples will highlight how long-read sequencing has identified diagnoses where short-read sequencing tests were nondiagnostic.

#### Learning Objectives:

1. Explain how short-read and long-read sequencing differ
2. Recognize situations where long-read sequencing is potentially clinically useful
3. Discuss current clinical and research applications, with examples from rare disease

### **3:00-4:00 Counseling For Skeletal Dysplasia: Variables and Challenges Janet Bell, MS, CGC**

This presentation will provide an overview of skeletal dysplasia, which encompasses an ever-growing number of genetic conditions that affect the development of bones, joints and cartilage. This session will highlight the diagnostic process, the genetic role, patient education, risk assessment, psychosocial support and community integration, and the evolution of a multidisciplinary model of care. Emphasis will include insight into clinical variability, diagnostic tools, emerging therapies, as well as patient and community perspectives. Case examples will illustrate challenges faced in counseling, including family dynamics, social pressures and perspectives, and quality of life considerations. This presentation will also address the ethical and emotional facets of genetic counseling in skeletal dysplasia, including decision making and uncertainty given variability.

#### Learning Objectives

- Illustrate the clinical features and genetic basis of skeletal dysplasias commonly seen in MDC clinic
- Describe genetic testing strategies used in the diagnosis of skeletal dysplasia
- Explore the psychosocial and ethical challenges in counseling families

## **4:00-4:30 Interesting Cases and Dilemmas (In Person)**

**Moderator: Melissa Gibbons, MS, CGC**

The session will be an interactive session which will allow genetics professionals to present cases of rare knowns and unknowns. These will include cases that are rare knowns of diagnosed malformations, genetic syndromes, or potential genetic syndromes. The rare knowns presentation may be of assistance to others in practice. The session will also provide an opportunity for individuals to present cases of unknowns for assistance with management suggestions or potential diagnoses. It will also be a time to illustrate the need for a multidisciplinary approach and communication for optimal care of patients and families. Additionally, this provides a forum to discuss cases of genetic disorders and management options.

### Learning Objectives

- Reproduce a case presentation from chief complaint to diagnosis
- Design a differential diagnosis
- Examine diagnostic workup suggested by experts
- Illustrate clinical features of rare cases

**For information about our conference please visit:**

**<https://sites.google.com/view/cogc/home>**