**Semi-Annual Colorado Genetic Counselors Symposium**

**Date: Friday May 12th, 2023**

**Time: 8:00am – 4:30pm  
Hybrid (virtual & in-person) event**

**Virtual (Zoom):** [**https://ucdenver.zoom.us/j/97320967155**](https://ucdenver.zoom.us/j/97320967155)

**In Person: Mt. Yale, Children’s Hospital Conference Center**

**Prenatal & Preconception focus**

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

**8:00- 8:45 Michael Zaretsky, MD**

*In Utero Surgery for Congenital Diaphragmatic Hernia – The FETO procedure*

Fetoscopic endoluminal tracheal occlusion, or FETO, is a surgical procedure to treat the most severe cases of congenital diaphragmatic hernia (CDH) that are diagnosed in utero. Today’s presentation will review the procedure details and highlight the recent case done here at the Colorado Fetal Care Center.

Learning objectives

1. Identify criteria for fetal surgery (FETO Procedure) in the presence of a congenital diaphragmatic hernia.
2. Evaluate challenges associated with this procedure and outcomes

**8:45 – 9:30 Emily Forbes, MD & Marrisa Lafreniere, MS, CGC**

*FMR1 Premutation: Updates on the Adult Phenotype and Management*

This presentation will review how the genetics of the FMR1 repeat expansion differentially affects adults as compared with children. We will review the risks and phenotypic spectrum of the FMR1 premutation, as well as discuss management options, referrals, and resources.

Learning objectives

1. Compare and contrast the genetic causes of the different FMR1 disorders
2. Summarize the different premutation phenotypes in adults
3. Identify proper referrals for people with the FMR1 premutation.

**9:30 - 9:45 Break**

**9:45 - 10:30 Mikayla Stoecker, MS, CGC & Chandra Perez-Gill, MS, CGC**

*Genetic Testing in Pregnancy Loss*

Cytogenetic practices such as FISH and chromosome analysis are routinely ordered in cases of unexplained fetal demise or stillbirth. However, such evaluations have a significant risk of failure due to the compromised nature of the sample. We will discuss the use of SNP microarray in the evaluation of pregnancy loss at various stages of development and review published literature on the detection rate of anomalies in this population.

Learning objectives

1. Identify the limitations and causes of test failures using common cytogenetic techniques on POC samples
2. Summarize the utility of SNP microarray in the setting of fetal demise

**10:30- 11:15 Shannon Mulligan, MS, CGC**

*NIPT for Fetal Sex: A Cautionary Case*

This case report will discuss a patient referred for pretest counseling with the indication of advanced maternal age. It will chronicle the cascade of events that can follow an abnormal result for a sex chromosome aneuploidy and the unique addition of unexpected ultrasound findings, concluding with an update on postnatal outcome and evaluation with pediatric genetics.

Learning objectives

1. Examine the potential complexities subsequent to NIPT identifying a fetus at risk for a sex chromosome aneuploidy.
2. Illustrate the patient experience navigating an evolving diagnosis during pregnancy following “routine” testing.

**11:15- 12:00 Jessica Giordano, MS, CGC**

*Moving Towards Prenatal Genome as a One-Stop Test*

In the last decade, we have seen tremendous advances in prenatal screening and diagnosis through the use of CMA, NIPT, carrier screening, and next-generation sequencing. The use of exome sequencing has become standard practice for fetal anomalies at many centers, allowing for improved counseling on prognosis, management, and recurrence risk. Integration of genome sequencing into prenatal diagnosis will allow for a rapid, single, one-stop test.

Learning objectives

1. Summarize the current literature on the clinical utility of exome sequencing in fetal anomalies and stillbirth
2. Examine the value of genome sequencing in prenatal diagnosis as a single-tier, comprehensive test

**12:00-12:30 LUNCH**

**12:30-1:15 Aranza Gonzalez, MS, CGC & Samantha Montgomery, MS, CGC**

*Impact of Abortion Bans on Genetic Counseling in Texas and Beyond*

This presentation will briefly review the history of abortion access in the State of Texas through the fall of Roe and after. Additionally, we will discuss how abortion bans in Texas impact prenatal genetic counseling services and patients living in Texas and nationwide. The discussion will include a Q&A portion to address attendants' questions and concerns regarding abortion bans.

Learning objectives

1. Summarize history and abortion legislation in Texas up to fall of Roe
2. Examine the impact abortion restrictions have on genetic counseling practice & patients

**1:15- 2:00 Carly Peterson, MS, CGC & Rachel Swihart, MS, CGC**

*PGT-yay or PGT-nay: Indications for appropriate referrals*

While many genetic counselors are familiar with the option of PGT (preimplantation genetic testing) for a variety of genetic conditions, we plan to provide a genetic counselor-based view of complex PGT cases in which PGT-M (PGT for monogenic disease)/PGT-SR (PGT for structural rearrangement) has been utilized to reduce reproductive risks for CCRM patients. We will walk you through the process as we explore testing avenues and options that come up when designing such specialized testing. With so many requirements necessary for testing, there are indications in which PGT is not possible or is not the most appropriate risk reduction strategy.

Learning objectives

1. Illustrate the utility of PGT-M/PGT-SR for various indications
2. Examine the stages of the PGT process for complex scenarios from referral to pregnancy
3. Define cases in which PGT is an acceptable intervention for reducing reproductive risk

**2:00- 2:15 Break**

**2:15- 3:00 Jennifer Hoskovec, MS, CGC**

*Single Gene NIPT: A deep dive into clinical performance for autosomal recessive conditions and fetal antigen status.*

This session will discuss the technology and workflow of clinically available single gene NIPT assays for common recessive disorders and fetal antigen status. A review of validation data and clinical case examples will also be presented.

Learning objectives

1. Define carrier screening with reflex with single gene NIPT and fetal antigen NIPT
2. Identify appropriate single gene NIPT and fetal antigen NIPT applications through case presentations
3. Compare the performance of traditional carrier screening to carrier screening with reflex to single gene NIPT

**3:00 – 3:45 Manesha Putra, MD, Hannah Elfman, MS, CGC & Kestutis Micke, MS, CGC**

*Debate: Should secondary findings be offered on prenatal whole exome?*

Review pros and cons of offering secondary findings on prenatal whole exome sequencing. The speakers will each take a side and argue their reasoning, with the goal for the audience to think critically about whether reporting of secondary findings should be included as standard practice.

Learning objectives

1.     Summarize current guidelines for inclusion of secondary findings in prenatal whole exome sequencing

2.     Defend the merits of including variants outside the clinical indication in prenatal whole exome sequencing

3.     Evaluate the potential consequences of secondary finding disclosure in prenatal settings

**3:45 – 4:30 Interesting Cases/Diagnostic Challenges**

*Moderator: Leslie McCallen, 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 the management options.

Learning objectives:

1. Reproduce a case presentation from chief complaint to diagnosis
2. Design a differential diagnosis
3. Examine diagnostic workup suggested by experts
4. Illustrate clinical features of rare cases