Semi-Annual Colorado Genetic Counselors Symposium

Date: October 11, 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 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.

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8:00-8:45 The Evolution of Newborn Screening in Colorado
Erica Wright, MS, CGC

Emerging therapies are improving the outcome of numerous genetic diseases and propelling the expansion of newborn screening. The Colorado Newborn Screening program continues to add additional disorders and refine its current screening panel including adding Pompe Disease and MPSI in 2022 and X-linked Adrenoleukodystrophy in 2023. In the coming year, Colorado will add MPSII and GAMT as well as targeted congenital CMV. While beneficial to the newborns of Colorado, this rapid expansion of the disorders can be disruptive and come with struggles. Hence, this presentation will highlight both the successes and barriers to expanding the Colorado NBS as well as the feasibility and sustainability of a newborn screening in the public health setting.

Learning Objectives

- Provide update on the Colorado newborn screening program including addition of disorders and continued quality improvement projects.
- Describe impact of expansion on both laboratory and follow-up/clinical side of the NBS system
- Examine the feasibility and sustainability of a newborn screening in the public health setting.

8:45-9:30 Newborn Screening and Downstream Considerations
Leighann Sremba, MS, CGC and Janell Kierstein, MS, CGC

In this talk, we will review the newly added conditions (MPSI, Pompe, X-ALD) to Colorado's state newborn screening program and to the federal Recommended Uniform Screening Panel (RUSP) (MPS II). The addition of these conditions has created new dimensions of newborn screening follow up in the outpatient clinic by identifying those who are at risk for later onset disease rather than immediately affected newborns. There is also an expansion of family member testing due to the addition of an X-linked semidominant condition to NBS. We will discuss this new paradigm shift and our one year follow up experience with patient cases.

Learning Objectives

- Review the natural history of MPS I, MPS II, Pompe and XALD
- Describe the complexities that go along with early diagnosis of later onset disorders
- Discuss the downstream considerations for family member testing, monitoring and management in X-ALD

9:30-10:15 Varying Journeys of Different ALD NBS Parents
Miranda McAuliffe

In this talk, Miranda will share her experience as the mother of a son identified to have X-linked adrenoleukodystrophy (X-ALD) by newborn screening and discuss the varying journeys of X-linked X-ALD families, parent perspectives, and the impact of newborn screening. Miranda will also review her role in advocacy as part of the ALD Alliance.

Learning Objectives

- Describe X-ALD family member experiences as they relate to diagnosis, newborn screening, and psychosocial impacts
- Discuss the benefits of ALD Alliance for families with X-ALD

10:15-10:30 Break

10:30-11:15 Post-Pregnancy and Pre-Conception Reproductive Genetics Care: Bridging the Gap
Manesha Putra, MD

In this talk we will discuss the role of reproductive genetics service in bridging the care for families who have had prior pregnancy with genetic/metabolic diagnoses. Post-pregnancy period is an ideal time to summarize prior genetic/metabolic testing that had been done and to perform additional testing as necessary. On the other hand, pre-conception period is an ideal time to discuss plan of care prior to and during the subsequent pregnancy. We will discuss common issues we typically encounter in these settings.

Learning Objectives:

- Describe the utility and importance of post-pregnancy and pre-conception reproductive genetics care
- Review common reproductive genetics issues in post-pregnancy period
- Discuss common reproductive genetics issues in pre-conception period

11:15 - 12:15 PEARL Trial: PrEnAtal enzyme Replacement for Lysosomal disorders
Billie Lianoglou, MS

This session will provide updates for diagnosing fetuses with LSDs including benefits/limits to expanded carrier screening, sonographic findings for specific conditions as well as challenges related to fetal sequencing. In addition, an overview of the ongoing phase 1 clinical trial of in utero ERT for patients with LSDs, including interim results and opportunities for other potential prenatal therapies.

Learning Objectives

- Recognize prenatal presentations of LSDs.
- Describe the rationale for the current on-going phase 1 clinical trial of in utero enzyme replacement therapy as well as the potential benefits and risks.

12:15 – 12:45 LUNCH BREAK SPONSORED BY: Ambry Genetics®

12:45-1:45 RNA Testing Methodologies and Clinical Uses
Rebecca Carr, MS, CGC and Victoria Suslovitch, MS, CGC

This presentation will review current RNA testing methodologies and the implication RNA analysis has on diagnostic genetic testing including increasing diagnostic yield and reducing uncertainty. Presenters will also review a variety of case examples utilizing RNA across multiple clinical indications and specialties.

Learning Objectives

- Discuss RNA testing methodologies.
- Reflect on implications of RNA analysis in diagnostic genetic testing.
- Review case examples spanning multiple specialties.

1:45-2:30 Recruitment and Retention of Genetic Counselors from Diverse Educational Backgrounds
Kami Schneider, MS, CGC

This presentation will review results of a quantitative survey of GCs evaluating how GCs started their higher educational journeys prior to GC graduate training and comparing resiliencies and demographics between those who attended community college and those who did not. Plans for ongoing qualitative research to explore lived experiences, resiliencies and barriers of GCs who started higher education experiences at a community college will be discussed. The hope is for this research to provide guidance on how the profession of genetic counseling can better recruit, train, and retain individuals from diverse educational backgrounds.

Learning Objectives:

- Understand the importance of recruitment and retention considerations for GCs who attend(ed) community college.
- Recognize aspects of higher education experiences of GCs prior to genetic counseling training regarding pathways, perspectives, resiliencies, and demographic factors
- Identify future research opportunities to help with diversity recruitment and retention in the GC profession.

2:30 - 2:45 Break

2:45-3:30 Next Generation Advocacy in Rare Disease Communities
Karmen Trzupek, MS, CGC

“Next generation advocacy” sees patients and families playing an active and crucial role in driving research and development for rare disease therapies. The shared mission and vision of Global Genes and RARE-X is to provide expanded tools and resources to support such advocacy efforts. This presentation highlights real-world successes of next generation advocacy across different rare disease communities and explores how genetic counselors may empower patients and families through their advocacy journeys.

Learning Objectives

- Describe the history and mission of Global Genes and the RARE-X program.
- .Illustrate how different advocacy activities help drive research and inspire hope in rare disease organizations, even in the absence of clinical trials or approved therapies.
- Examine aspects of genetic counseling that can support rare disease community engagement, advocacy, and research.

3:30 – 4:30 Genetic Counselors in Advocacy and Public Policy
Amy Gaviglio and Marianna Raia

This presentation will highlight genetic counselor evolving roles in advocacy including founding and supporting organizations, working closely with families and stakeholders, and informing on public policy. Two genetic counselors will describe their experiences highlighting their roles in Expecting Health and Rare Disease Advisory Councils (RDACs). Expecting Health works collectively with organizations, healthcare professionals, people, and communities to provide the right information at the right time - from planning a pregnancy to early infant care. Rare Disease Advisory Council (RDAC) acts as an advisory body that gives the rare disease community a stronger voice in state government.

Learning Objectives

- Describe Expecting Health and goals of engaging families as partners in driving health care system change.
- Discuss NORDs efforts to establish RDACs and the roles RDACs play across the United States informing on public policy
- Illustrate experiences and provide resources that empower families to engage, advocate, and lead in health care and public policy.

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