Meeting Schedule

Meeting Date: Tuesday, May 7, 2024

Meeting Location: Virtual Meeting sponsored by Kean University
Registration Link

7:30 - 8:00 AM **Executive Committee Meeting**
Executive committee only – closed meeting

8:00 - 8:30 AM **Concurrent Committee Meetings**
Everyone is encouraged to participate in a committee.
- Education
- Legislative
- Public Relations
- Professional Issues

8:30 – 10:45 AM **Student Thesis Presentations Concurrent Sessions**
- See next page for schedule

11:00 AM – 12:00 PM **Bridging the Gap: Diagnostic and Clinical Implications of RNA Analysis**
Catherine Mayo, MS, CGC
Ambry Genetics

12:00 – 12:30 PM **Break**

12:30 – 1:00 PM **General Membership Meeting**
- President's Report
- Secretary's Report
- Treasurer's Report
- Introduction of Members
- Committee Reports
- New Business
- Member Announcements

1:15 – 2:45 PM **Pompe Disease: From Bench to Bedside with a Patient Story**
Nicole Luogo, MS, PA-C
Hallie Andrew, MS, CGC
Sanofi

3:00 – 4:00 PM **The Ethics of Genetic Prognostication**
Rebecca Mueller, PhD
University of Pennsylvania
Katharine Press Callahan, MD
Children's Hospital of Philadelphia

4:00 PM **Adjournment**

This event has been submitted to the National Society of Genetic Counselors (NSGC) for approval of 0.575 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.

Class of 2024 Student Presentations

8:30 - 10:45 AM Thesis Presentations - Concurrent Sessions

Class of 2024 student presentations are divided into two breakout sessions. Each breakout session contains nine presentations. Each student will present their thesis for 10 minutes, with five minutes allotted for Q&A. Contact information is listed for each student, provided there are questions or discussions that exceed the allotted time.

Breakout Room 1: Exploring JEDI Initiatives, Improving Access, and Advancing Delivery Models

8:30AM | *Language Concordance in Genetic Counseling: Counselor Training, Education, and Workplace Requirements to Provide Care to Patients with Limited English Proficiency*

Carolina Conti, Kean University | conticar@kean.edu

Individuals with limited English proficiency (LEP) face challenges communicating effectively with healthcare professionals in the United States, contributing to existing healthcare access and outcome disparities for this population. Communication entirely in the patient's preferred language without an interpreter, known as language concordance, improves patient communication and helps achieve in-session genetic counseling goals. Although research supports that language concordance should be encouraged and aided in graduate programs and continuing education opportunities in the genetic counseling field, there is a gap in research on the educational and personal experiences of multilingual genetic counselors and the support provided in this area. This mixed-method study aims to fill this gap by shedding light on the training, support, and resources available to multilingual genetic counselors practicing in the US who conduct language-concordant sessions. A total of 13 multilingual genetic counselors participated in the study, revealing that the vast majority obtained limited formal training and education specific to genetic counseling in languages other than English, with little to no support or resources in a language other than English. This study underscores the need for comprehensive training, education, and support mechanisms for multilingual genetic counselors throughout their careers in various educational and professional settings. Participants in this study endorsed the development of official practice-based guidelines and requirements for genetic counselors who wish to conduct language-concordant genetic counseling sessions.

8:45 AM | *Increasing Readability to Online Inherited Retinal Disease Information*

Stephanie DiNenno, Rutgers University | sed146@dls.rutgers.edu

Inherited retinal diseases (IRDs) affect more than five million people in the world and many of them have overlapping features. An increasing number of individuals are using the internet for medical information; however, many sources are written at a higher literacy level than the average individual in the United States. Despite this, almost seventy-five percent of Americans use online health information to influence their medical decisions. Many online webpages for IRDs are written at a higher reading level and lack information about the value of genetic testing, information on low vision resources, and possible therapies. The purpose of this project was to create a patient-friendly webpage for both the general public and healthcare professionals. A draft of the webpage was distributed and evaluated via a survey and readability scoring. Our findings show that although readability scales may indicate information is at a higher than recommended reading level, readers are able to understand the information provided. We

also found that readers are interested in having more information about genetic counseling. Our hope is that this free, public access webpage will help to make online health information of IRDs more accessible.

9:00 AM | *Cancer Genetic Counseling Toolkit for Vietnamese Community Outreach*

Amy Huynh, Rutgers University | ah1413@dls.rutgers.edu

Background. Genetic counseling and genetic testing can inform individuals of appropriate medical management for hereditary cancer predisposition syndromes, yet many underserved populations experience barriers to genetic services. The Vietnamese American community experiences disparities due to lack of physician referrals and culturally-sensitive educational materials. Culturally-tailored outreach programs can help overcome these barriers to increase awareness and uptake rates of genetic counseling for targeted populations. This project seeks to identify themes to inform the development of a successful outreach program for Vietnamese Americans and an outreach toolkit for cancer genetic counseling in the Vietnamese community.

Methods. A literature review was performed to learn how to develop an outreach toolkit for the Vietnamese community. Themes were extracted from the content of the articles pertaining to Vietnamese cultural concerns, methods for outreach, and culturally sensitive educational materials. A toolkit of five educational documents was developed based on these themes. This toolkit was sent out via an online anonymous survey to healthcare professionals for content feedback. Revisions were made based on feedback, and documents intended for a Vietnamese-literate audience were sent to a third party to be professionally translated.

Results. Seven major themes emerged from the literature review regarding considerations for outreach in the Vietnamese community, including: concerns of financial cost, limited language proficiency and health literacy, limitations with understanding the purpose of genetic services/preventive medicine, stigma associated with cancer/illness, value of community/family, strongly trusting recommendations from physicians, and the value of lay health workers. Feedback was obtained from six healthcare professionals and was overall positive with most suggestions focused on improving conciseness and cultural sensitivity of the content.

Discussion. A cancer genetic counseling outreach toolkit for the Vietnamese American population was developed that incorporated key cultural themes based on literature. More research is needed in the future regarding the toolkit's effectiveness in this community.

9:15 AM | *Exploring the Impact of Online Networks Available to BIPOC Applicants of Genetic Counseling Master's Programs*

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The genetic counseling field remains largely homogenous with 89% of practicing genetic counselors identifying as Non-Hispanic White (National Society of Genetic Counselors, 2023). Although there have been many diversity and recruitment initiatives, these efforts have not resulted in a notable change in demographics (Channaoui et al., 2020). A number of online networks have emerged with the mission of helping prospective students, including: Minority Genetic Professionals Network (MGPN), Genetics Opportunities, Learning, Development, and Empowerment Network (GOLDEN), Genetic Counseling Prospective Student Network (GCPSN), and GC Chat on Discord. Given the initiative to recruit and sustain more diversity in the field, a look into the utilization of these networks by Black, indigenous, and people of color (BIPOC) applicants is an important next step. The purpose of this study is to determine the extent to which BIPOC applicants are using online networks and the perceived impact of these

networks. We found that common reasons for joining these online networks include needing help with applications and seeking connection to other BIPOC individuals. These networks may be offering those with limited connections and resources a way to get the exposure and experience they need to have a more competitive application. Mentors and genetic counselors are found to be major sources of support in the application process and these networks offer an avenue of connection. Overall, the utilization of these networks increased feelings of connection to the genetic counseling field, with networks aimed to support BIPOC applicants seeming to deepen that connection.

9:30 AM | *"My Sweet Sibling and I"- A Children's Book for Siblings of Individuals with Congenital Hyperinsulinism*

Chloe Koeppen, Rutgers University | ck832@dls.rutgers.edu

The focus of this capstone project is to create a resource for siblings of individuals with congenital hyperinsulinism [HI], a rare genetic disorder that affects 1 in 25,000 babies annually and can result in hypoglycemia leading to seizures, brain damage, and even death. While those with HI face significant challenges, their siblings also experience unique difficulties such as emotional distress, adjustment problems, and taking on caregiver responsibilities. These challenges may be partly attributed to a lack of communication and understanding about HI within the family structure. This resource aims to help families navigate the complexities of HI, including providing support to siblings who may be struggling with their emotions. A group of 17 patient advocates were provided with the storyline and asked to complete a corresponding Qualtrics survey to obtain feedback on the content. There was a 59% response rate, in which the majority of the respondents agreed the content was satisfactory in demystifying and educating siblings on the condition. The results indicated that the book met the project's stated goals, and we believe it will be a beneficial tool for families seeking to foster healthy relationships between children with HI and their siblings.

9:45 AM | *An Assessment of the Influence of Race and Gender on Risk Perception Determined from Pictographs*

Gabriel Koslow, Kean University | gabekoslow@gmail.com

Pictographs are visual representations used to convey ideas. They are frequently employed in healthcare settings to present data in a comprehensible manner, and are particularly beneficial for individuals who may struggle with numerical or statistical comprehension when making critical healthcare decisions. This study delves into the impact of racial and gender concordance between participants and pictographs on risk perception within a hypothetical scenario. Participants, recruited from Kean University, were presented with a hypothetical risk scenario accompanied by a pictograph illustrating the associated risk. Subsequently, participants' demographic information was collected. Pictographs depicting individuals of various races and genders were randomly assigned to participants, and their perceived level of risk, influenced by these pictographs, was documented. Our analysis revealed no significant differences between concordant and discordant groups. These findings suggest that employing visual aids featuring diverse representations of races and genders is an effective strategy without engendering unwarranted or inaccurate perceptions of risk.

10:00 AM | *Education and Training Preferences of Medicolegal Death Investigators in Collecting Family History for Cases Surrounding Sudden Unexpected Death*

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Sudden unexpected death (SUD), is a circumstance that more commonly occurs in otherwise healthy-appearing individuals. Approximately five million lives are lost per year globally to SUD. With that, in up to a third of comprehensive medicolegal investigations, no identifiable cause of death can be found. When SUD occurs, the death is typically reported to local coroner or medical examiner's offices. In this case, medicolegal death investigators (MDI) are the first to be in contact with the family to interview for history. If no physical identification of death can be found at the preliminary investigation, the concern for genetic testing increases. This study was to determine the experience of medicolegal death investigators in training in regard to family history collection surrounding a sudden unexpected death case. This study also gauged the level of confidence MDIs have in obtaining family history. Learning styles in which medicolegal investigators prefer to be trained were also investigated. Results have shown that MDIs are more likely to ask questions about sudden death, cardiomyopathy, and sudden unexpected infant death, but less likely to ask about family members involved in motor vehicle accidents and drowning. Findings also showed that MDIs in their respective roles have identified barriers such as emotional status of a witness or family member and noncompliance to hinder the questioning of family history. Investigating further into motivations and future educational topics that MDIs would like to learn more about, the interest was greatest for psychosocial areas that play into family history collection and the need for investigation tools. This study looks to help close the gap between the first-line of sudden unexpected death to a referral for cardiology for suspected inherited cardiac conditions.

10:15 AM | *Evaluation of Alternative Model of Cancer Genetics Service Delivery at St. Luke's University Health Network (SLUHN)*

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In March 2021, the Cancer Risk and Genetics Program (CRGP) at St. Luke's University Health Network piloted an alternative service delivery model for patients ages 60 and younger who were recently diagnosed with breast cancer. The program was designed to expedite the turnaround time of STAT genetic testing (GT), allowing patients to receive critical genetic information sooner and facilitate their surgical decision-making process. In a collaboration between the CRGP and the Kean University Genetic Counseling Graduate Program, the principal investigator conducted a program evaluation to assess whether the STAT GT program was effectively designed and implemented to meet the needs of this patient population, focusing on the outcome of patient satisfaction. The research team assessed levels of patient satisfaction regarding interactions with the CRGP genetics team, participation in the STAT GT process, and patient appreciation of the clinical utility of genetic information. A total of 504 patients received a survey which solicited quantitative and qualitative feedback on experiences in the STAT GT program and the responses of 40 participants were included in data analysis. Statistical methods included descriptive and correlation analysis to assess relationships between reported levels of patient satisfaction and contextual data including age at the time of diagnosis, cancer staging at the time of diagnosis, time since diagnosis, and genetic test results. Participants generally reported moderate to high levels of agreement with satisfaction measures, with average reported satisfaction scores ranging from 4.05 to 4.83 out of a possible maximum score of 5.00. Elevated levels of patient satisfaction and broadly favorable feedback reported by participants indicate that the STAT GT program was successful in meeting the needs of this patient population, irrespective of patient contextual variables. The need for improved internal communication was identified as an area for improvement in program implementation, including more effective education and coordination with staff responsible for collecting patient samples at regional SLUHN laboratory sites, as well as more effective sharing of GT results with the breast surgeon and other providers within the patient's care team to ensure timely surgical decision-making. The STAT GT program has proven to be a successful model of cancer genetics service delivery in advancing patient satisfaction and may effectively be replicated for other indications.

10:30 AM | *Awareness and Perception of the Genetic Counseling Profession in a Diverse Undergraduate Population - Before and After Educational Outreach*

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The field of genetic counseling, which involves communicating complex genetic information and providing psychosocial support to individuals and families, has struggled with issues of diversity, equity, and inclusion, notably among underrepresented minority students over the past two decades. To enhance diversity in genetic counseling, programs must better understand methods for educating, engaging, and attracting students from underrepresented backgrounds. This study aims to evaluate the efficacy of such outreach initiatives and gather feedback from underrepresented undergraduate students of diverse racial/ethnic backgrounds at Rutgers University in the United States. By eliciting feedback and assessing the impact of a presentation in classes with diverse student populations, the study seeks to refine recruitment strategies and delve into factors contributing to the lack of diversity. The study involves a pre-survey, educational outreach via a PowerPoint presentation, and a post-survey distributed to 563 non-genetic major undergraduate students at Rutgers. Results revealed prevalent misconceptions among students regarding genetic counseling, including its scope and accessibility, persisting even after the presentation. Most students were unaware of the psychosocial counseling aspect of the profession beforehand. Primary barriers cited by students considering genetic counseling were the rigorous admissions process and program costs. However, feedback on the presentation was largely positive, with students finding it informative and engaging. To mitigate these barriers, additional outreach events emphasizing genetic counseling as a potential career option, along with discussions on financial aid opportunities and application resources, are recommended. In addition, highlighting the interdisciplinary nature of the field and incorporating personal narratives for speakers may further enhance outreach effectiveness.

Breakout Room 2: Genetic Counseling and the Power of Information: Public Health, Employment, and Patient Care

8:30 AM | *Burnout Among Non-Direct Patient Care Genetic Counselors: Observations of Job-Related Attitudes and Work Environment*

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Work-related burnout, defined by depersonalization, exhaustion, and reduced personal accomplishment, has a significant impact on the retention of genetic counselors in the workforce. Existing literature predominantly focuses on burnout among direct patient care genetic counselors, leaving a notable gap in understanding the experiences of those in non-direct patient care roles. This mix-methods study aims to address this gap by investigating various work-related and personal factors contributing to burnout in non-direct patient care genetic counseling roles via a 42-question survey, providing crucial insights for the evolving landscape of the profession. Descriptive statistics were generated via IBM SPSS Statistics 27 for OSX and burnout was measured using the Maslach Burnout Inventory General Survey (MBI-GS), and 13 themes were generated from the qualitative analysis. The MBI-GS is a validated measure that uses three subscales to measure burnout: cynicism, professional efficacy, and exhaustion. Results reveal burnout may be higher among non-direct patient care genetic counselors, contrary to the hypothesis. Results from qualitative responses indicate that participants perceive burnout to be largely linked to leadership and management concerns. Additionally, variable sociodemographic factors are identified as contributing factors. The sociodemographic profile and burnout scores of 27 non-direct patient care genetic counselors are summarized, exploring the interplay between professional experiences, sociodemographics, and burnout in this evolving sector of genetic counseling.

8:45 AM | *ToTell or Not to Tell: The Influence of Disclosure on the Psychological Impact of Oocyte Donation*

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Oocyte donors are individuals who give their oocytes to a recipient individual or couple for embryo creation and pregnancy. Oocyte donation may cause lasting positive or negative psychological effects for the oocyte donor; however, there is limited and conflicting data on the long-term impact of oocyte donation. The purpose of this study is to explore the experiences of oocyte donors and investigate the influence of disclosure on the psychological impact of oocyte donation. Research participants were recruited from two Facebook groups: We Are Egg Donors and Former Egg Donor Support. Nine oocyte donors participated in semi-structured interviews via the Zoom online communication platform. Interview transcripts were coded and analyzed using content analysis and thematic analysis. Thematic analysis revealed four main themes concerning the psychological effect of oocyte donation: 1) positive psychological effects, 2) negative psychological effects, 3) neutral psychological effects, 4) ethical concerns. All nine participants disclosed their donation experience to individuals in their personal lives. Four main themes concerning the participants' disclosure decisions and support emerged: 1) positive reaction from others, 2) disclosure increased strength of support system, 3) strong support increased positive emotions, 4) social media support. Participants recommended that healthcare providers treat donors with more respect and kindness. Overwhelmingly, participants did not feel that they were the real patient, and their well-being was not a priority. Four participants (44.44%) met with a genetic counselor during their donation and found it valuable to have someone explain their genetic results and check in on how they were doing. Oocyte donors should be informed of both the potential positive and negative effects associated with oocyte donation, with emphasis that strong support systems may promote more positive psychological outcomes. Results from this exploratory study can guide future research on factors that impact the psychological effects of oocyte donation.

9:00 AM | *Clinical Utility and Incremental Yield of Expanded Prenatal Molecular Genetic Screening for the Detection of Clinically Relevant Fetal/Newborn Disease in the General Population*

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Non-invasive prenatal testing (NIPT) is the process of sequencing placental DNA from maternal blood to assess the likelihood of a fetus being affected with a chromosomal condition. Per professional guidelines, NIPT is considered standardized care offered to all pregnant patients, regardless of age, to enable early risk assessment for common chromosomal conditions identified in liveborn babies. While previous studies have demonstrated the benefits of expanded NIPT platforms that assess fetal risk for additional chromosomal and genetic conditions, there is limited information on these methodologies within the general-risk population. The purpose of this study was to evaluate the positive predictive value (PPV) and application of two expanded NIPT platforms designed for genome-wide and single-gene use (sgNIPT), within the general obstetric population of a U.S. regional perinatal center. A retrospective chart review was conducted for approximately 12,250 patients who pursued expanded NIPT, with or without reported risk factors, between January 2020 and July 2023. Descriptive statistics were generated via Microsoft Excel 2007. Data analysis was performed with test-positive results and available diagnostic testing results from at-risk pregnancies. Approximately 130 pregnancies screened positive on genome-wide NIPT and 14 pregnancies screened positive on sgNIPT. PPV was calculated as the total number of true positives divided by the number of true positives combined with the number of false positives. PPV was calculated as 54.08% (genome-wide NIPT) and 61.54% (sgNIPT). While most affected pregnancies were associated with either advanced maternal age, ultrasound findings, and/or family history, several had no high-risk indications and were identified only through routine screening. The findings of this study suggest that implementation of expanded NIPT as part of standardized prenatal screening within the general obstetric

population allows for early fetal disease risk assessment of a range of chromosomal and genetic conditions, regardless of identified risk factors.

9:15 AM | *Pregnancy Termination for Fetal Anomaly: Genetic Counselor Perspectives on Patient Coping*

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Termination of pregnancy for fetal anomaly (TOPFA) is a procedural intervention that deliberately ends a pregnancy affected by anomalies affecting bodily structures or functions. Genetic counselors are professionals trained to provide information to assist people in making well-informed, autonomous decisions while helping them adjust to and cope with revealed genetic information. Though genetic counselors frequently see patients for TOPFA, there are no specific guidelines for any healthcare providers caring for women. This study aims to fill this gap and explore genetic counselors' perspectives of women coping with TOPFA to aid in the provision of optimal care for women coping with TOPFA. Participants were recruited through National Society of Genetic Counselors (NSGC) via an email. Nine prenatal genetic counselors participated in semi-structured interviews with the researcher via Zoom. Qualitative content analysis identified eight major areas of discussion regarding genetic counselors' perspectives on patient experiences with TOPFA: 1) role of the genetic counselor, 2) resources, 3) follow up, 4) patient coping, 5) impact of TOPFA, 6) barriers to care, 7) impact of the Dobbs decision, 8) advice for genetic counselors. This exploratory study highlights the inconsistencies in care that genetic counselors provide to TOPFA patients, particularly in terms of their resource provision, emotional support, and follow up, as well as variability in their understanding of such patients' experiences. Though genetic counselors in this study feel that TOPFA is highly impactful on women's lives, they face barriers to providing optimal care to their patients and feel that patient experiences have worsened since the Dobbs decision. Genetic counselors in this study suggest an increase in psychosocial support for TOPFA patients. The findings of this study emphasize the need for precise genetic counseling guidelines for the practice of provision of care for TOPFA.

9:30 AM | *Implementing Next-Generation Sequencing as Part of Newborn Screening in State Public Health Laboratories: Status and Barriers*

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Newborn screening (NBS) programs are vital public health initiatives, facilitating early diagnosis and treatment for thousands of infants annually in the United States. Originally targeting phenylketonuria (PKU) in the 1960s, NBS has expanded to encompass a broad spectrum of disorders, aided by technological advancements such as tandem mass spectrometry and next-generation sequencing (NGS). NGS, in particular, offers the potential to revolutionize NBS. While NGS presents numerous benefits, its integration into NBS faces various challenges, including technical feasibility, financial barriers, and logistical considerations. This study aimed to assess the barriers, expectations, and plans of state NBS programs regarding NGS integration, providing insights to guide advancements and inform stakeholders. It was found that most states are not currently using NGS technology though there is interest in implementing it in the future. The biggest barriers were lack of appropriate staff to support data analysis, bioinformatics, and genetic counselors as well as the cost of sequencing and volume of samples.

9:45 AM | *The Impact of the Overturn of Roe v. Wade on Genetic Counseling Students' Decision of Program Selection and Ranking: A Multi-Perspective Survey of Genetic Counseling Students and Program Leadership*

Kimberly Schlosser, Rutgers University | kimberly.schlosser00@gmail.com

On June 24, 2022, the United States Supreme Court overturned Roe v. Wade in the case of Dobbs v. Jackson Women's Health Organization, ultimately taking away the federal constitutional right to an abortion (Kaufman et al., 2022). This groundbreaking ruling granted states authority to regulate abortion policies. While studies have evaluated prenatal genetic counselors' perception of the impact of abortion legislation on genetic counseling (Jayaraman et al., 2021), none explored the impact of the overturn of Roe v. Wade on program selection and ranking for genetic counseling applicants. This study aims to assess the most influential factors when selecting, applying, and ranking genetic counseling training programs. Additionally, to evaluate the impact of the Dobb's decision on genetic counseling program by comparing the percent change in the total number of applicants across three consecutive application cycles from 2020 to 2022. Two surveys were created, one for United States-based genetic counseling program leadership, and another for current students and alumni from 2020 to 2022. Genetic counseling programs in states with fewer abortion restrictions showed varying application trends that were not consistent with a significant impact from abortion restrictions. Survey results indicated that abortion-related factors were not among the leading factors that contributed to students' decision to apply and/or rank various genetic counseling programs. Primary factors influencing application decisions included geographical location (92.0%), program's values and culture (85.9%), and interaction with program leadership and faculty (80.3%) as compared to ranking various program, the most important factors were the program's values and culture (89.7%), variety and length of clinical rotations (87.4%), and interactions with program leadership and faculty (86.6%).

This is the first study to assess the implications of the overturn of Roe v. Wade on genetic counseling programs and students' decision-making process during this critical period. While our research did not find that abortion-related concerns as primary factors influencing students' decisions to apply and/or rank genetic counseling programs, it is evident that the significance of the topic of abortion has increased, and it was a factor considered by students during the decision-making process.

10:00 AM | *Evaluating Pregnancy Termination Rates for Fetal Chromosome and Single Gene Disorders*

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The ability to detect genetic abnormalities prenatally has expanded in recent years to include a variety of diagnoses, including aneuploidy, copy number variants (CNVs), and single-gene disorders. The purpose of this study was to report on pregnancy termination rates following an abnormal prenatal diagnosis and to determine which factors may influence this decision. We conducted a retrospective chart review of 332 pregnancies diagnosed with a genetic abnormality from 2012-2023 and collected the type of prenatal diagnosis in addition to a variety of other factors. Data analysis consisted of multivariate logistic regression comparing each variable to the outcome of elective termination. In this study, the overall termination rate was 61.45%. Three variables remained strong predictors of elective termination: type of prenatal diagnosis, paternal race and/or ethnicity, and gestational age at diagnosis. This study adds to the literature on pregnancy termination rates following an abnormal prenatal diagnosis. Additionally, this expands our understanding of what other factors may influence the decision to terminate an affected pregnancy.

10:15 AM | *A Disease Conceptual Model of KCNT1-Related Epilepsy*

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Potassium sodium-activated channel subfamily T member 1 (KCNT1) related epilepsy is a rare genetic neurodevelopmental disorder characterized by two distinct phenotypes, sleep-related hypermotor epilepsy (SHE) and epilepsy of infancy with migrating focal seizures (EIMFS). The impacts of the disorder on the person with KCNT1-related epilepsy and their caregivers are not well understood for either phenotype. Disease concept models (DCMs) are formal frameworks which highlight the lived experiences of these individuals and their primary caregivers. To further understand these impacts, we conducted twelve semi-structured open-ended interviews with primary caregivers of individuals with this condition. Of the twelve total participants, nine were caregivers of patients with EIMFS and three were caregivers of patients with SHE. Three categories of domains were used as the framework for coding and data analysis: symptoms, symptom impacts on the individual, and caregiver impacts. Symptoms reported by all caregivers of both EIMFS and SHE participants included communication, neurological and sleep symptoms. Though both phenotypes present with intellectual disability, caregivers of more severely affected individuals placed emphasis on motor issues (EIMFS participants (n=9), and the less severely affected described concerns about cognitive disabilities (SHE participants (n=3)). Inconsolable crying was described in 5 of the 9 EIMFS participants which was previously underreported in the medical literature. Of note, delayed puberty was described in one male SHE participant which, to our knowledge, is a novel concept. Commonly reported impacts of KCNT1 mutation on the individual were reported in the domains of schooling (n=10), socialization (n=8), hospitalizations (n=9), self-care and daily living tasks (n=12), and surgical interventions (n=6). The most common domains of caregiver impacts include general caregiver requirements (n=12), emotional impacts (n=12), financial impact (n=11), need for formal support (n=7), overall health impacts (n=10), and social impacts (n=12). The conceptual model of KCNT1-related epilepsy herein described expands the description of the disorder known from public medical literature.

10:30 AM | *A Conceptual Disease Model for SYNGAP1-Related Neurodevelopmental Disorders*

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SYNGAP1-related disorders are rare neurodevelopmental and epileptic encephalopathies that were first identified in 2009. These disorders are known to manifest with a great range of clinical presentations and neurodevelopmental trajectories; however, the impacts of symptoms are not well understood. To understand symptoms and their impacts, we conducted twelve qualitative interviews with caregivers of individuals with SYNGAP1-related disorders to form a preliminary disease concept model. An inductive thematic analysis approach was used to identify concepts described by participants using DeDoose software. Disease concept models (DCMs) are frameworks that examine the relationship between symptoms, symptom impacts on quality of life, and symptom impacts on caregivers to provide a basis for generating outcome measures. We comprehensively mapped symptoms and their impact on caregivers and their families to generate a disease model as a foundation for clinical endpoints in future trials. The findings of this study identified concepts that are beneficial to include in a SYNGAP1-related disorders preliminary disease concept model that would be applicable for treatment.