Since January 2020 Elsevier has created a COVID-19 resource centre with free information in English and Mandarin on the novel coronavirus COVID-19. The COVID-19 resource centre is hosted on Elsevier Connect, the company's public news and information website.

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Genetic Counseling Abstracts (poster)

eP293

Patient satisfaction with telehealth genetic counseling across multiple subspecialties

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Introduction: Studies of telehealth genetic counseling (THGC) services, including videoconferencing and telephone counseling, have highlighted the need for further research on the patient experience. The COVID-19 pandemic resulted in a rapid shift from in-person genetic counseling to THGC. Most studies to date focused on THGC for cancer genetics, with only a handful of studies investigating patient satisfaction for other subspecialties. This study assessed patients' perspectives of the advantages and disadvantages of THGC and their satisfaction with this service delivery model for multiple subspecialties.
Methods: A patient satisfaction survey was designed to assess the experience of study participants at the time of referral, scheduling, during, and after the THGC appointment, as well as participants' overall satisfaction with the THGC process. Survey invitations were emailed to 485 patients between December 2020 and September 2021 following their last anticipated THGC appointment. Descriptive statistics were used for the analysis. One-way ANOVA was used to measure differences in reported satisfaction across subspecialties.
Results: A total of 103 patients responded (21.2%; 103/485). Almost half of participants reported that they were referred for hereditary cancer counseling (48.5%; 49/101) followed by reproductive genetics (13.9%; 14/101), neurogenetics (12.9%; 13/101), cardiogenetics (7.9%; 8/101), clinical genomics/exome sequencing (3%; 3/101), and ophthalmology (2%; 2/101). Most respondents (90.7%; 88/97) elected a telephone appointment versus videoconferencing, with the most common reason being "because it was easier" (70.8%; 63/89). Patient-perceived advantages of a THGC appointment were not needing to travel (94.5%; 86/91) and the ability to get an appointment that worked with their schedule (82.4%; 75/91). Safety concerns related to COVID-19 were cited as a benefit of THGC for 38.5% (35/91). Patient-perceived drawbacks included someone overhearing the conversation who was not invited (14.4%; 14/97) and distraction by other adults or children (7.2%; 7/97). Only 7.9% (7/89) of respondents agreed or strongly agreed with the statement "If I needed genetic counseling again, I would choose an in-person appointment." Nearly all respondents agreed or strongly agreed with the following statements: "I received the same care by telephone or video as I would expect at an in-person appointment" (95.5%; 85/89), "I would recommend a phone or video appointment for genetic counseling to family and friends" (89.9%; 80/89), and "Overall, I am satisfied with the quality of the appointment" (98.9%; 88/89). There was no statistical difference in reported satisfaction across subspecialties ($p = 0.823$).
Conclusion: Respondents for all subspecialties overwhelmingly reported that they were satisfied with the THGC experience. Some studies have suggested that providers prefer videoconferencing appointments. However, the vast majority of patients who responded to this survey elected telephone appointments, providing insight into patient preferences for THGC services. Although this survey was distributed during the pandemic, the most frequently selected benefits were related to logistical and scheduling issues as opposed to safety concerns related to COVID-19. These data support the use of THGC across multiple specialties as a patient-desired model of care, and helps to fill a gap in the literature by examining the experiences of patients seen for multiple subspecialties, beyond cancer genetics.

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eP294

Return of genome sequencing results in ostensibly healthy COVID-19 positive individuals: GENCOV Study Canada

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Introduction: Genome sequencing (GS) can identify genetic factors that may influence variability in COVID-19 symptoms and outcomes. In this context, GS can also be used to screen for secondary findings (SF) about inherited predispositions to several other diseases. Attitudes towards the use of GS as a screening tool in the general Canadian population is limited, warranting exploration of individuals' uptake of SF, knowledge of GS, as well as attitudes toward healthcare and genetics. This study aims to: 1) Determine the uptake of SF (clinically actionable conditions, rare genetic conditions, carrier status, drug reactions, and polygenic risk scores for common conditions) from GS; 2) determine baseline attitudes toward genetics and healthcare; 3) assess the impact of pre-test genetic counseling (GC) interventions on GS knowledge; and, 4) assess the impact of learning SF on patient attitudes, feelings, health outcomes, and health behaviors.
Methods: Online surveys were administered to ostensibly healthy individuals previously diagnosed with COVID-19 after consent but before pre-test GC. Surveys administered at baseline (T0) assessed: 1) sociodemographic characteristics; 2) SF preferences; 3) knowledge of GS; 4) attitudes toward healthcare, genetics and technology; and, 5) health literacy. Surveys administered after pre-test GC and return of GS results will address health outcomes and behaviors. Responses were analyzed with descriptive statistics.
Results: To date, 410/548 responses were received for at least part of survey T0. The majority were female (57%), ≥40 years of age (54%), and had a Bachelor's degree or higher (67%). Fifty-four percent indicated they were white/European. Almost all (407/410; 99%) said they wished to learn SF from GS. Eighty-six percent (352/407) selected clinically actionable findings, 74% (305/407) rare genetic diseases, 86% (353/407) common conditions, 81% (331/407) carrier status, and 83% (340/407) drug reactions. Less than 50% (n≤205/410) of respondents answered correctly to 5/11 statements about GS knowledge. Forty-eight percent (178/372) had positive attitudes toward genetics and 52% (194/372) had negative/mixed attitudes. Approximately half (175/374; 47%) agreed that the government will ensure a high-quality healthcare system, and 86% (320/374) reported it was important for them to access advanced tests and medical procedures. Most (347/370; 94%) reported that decisions about healthcare programs should be based primarily on the advice of experts, and 6% (23/370) on the general public's views. Most (337/369; 91%) reported that decisions about healthcare programs should be based on scientific evidence of the risks and benefits involved, and 9% (32/369) on moral/ethical issues. Overall, health literacy was adequate (mean score 18.5 out of 20, SD 2.3, n=371). Adequate health literacy scores fall into the highest of three categories within the BRIEF health literacy scale (scores 4-12: inadequate; 13-16: marginal; 17-20: adequate). Post-counseling and return of results survey data on attitudes, knowledge, and health outcomes/behaviors will be presented at the conference.
Conclusion: These preliminary findings suggest that most individuals wish to learn SF from GS despite low baseline knowledge. Generally positive and/or mixed attitudes toward healthcare and genetics in addition to relatively high health literacy scores suggests that the use of GS as a screening tool in healthy Canadians may be accepted. Pre-test GC to discuss SF and GS may improve participants' knowledge. Future surveys will assess the impact of GC interventions and return of SF results on knowledge, feelings, and health behaviors in the context of an otherwise healthy population.

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eP295

Barriers to uptake of genetic services in families of pediatric hypertrophic cardiomyopathy patients

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Introduction: Hypertrophic cardiomyopathy (HCM) is a common cardiovascular condition that is defined by unexplained left ventricular hypertrophy. A causative pathogenic variant can be detected via molecular testing in the majority of HCM cases. Pediatric HCM cases are typically predicted to be more severe than adult-onset cases and are more likely to be associated with a pathogenic variant. Barriers to genetic testing that have been established by prior studies include individual barriers such as unawareness and/or lack of knowledge regarding genetic services and institutional barriers including healthcare professionals' lack of awareness and knowledge regarding genetic services. The purpose of the study was to better elucidate the barriers to genetic testing in pediatric HCM patients and their families at UPMC Children's Hospital of Pittsburgh.
Methods: Data was collected via an anonymous survey utilizing Qualtrics software. The survey was distributed through a recruitment letter and several reminder emails that contained survey links.
Results: Of the 12 respondents, 7 (58.3%) had pursued genetic testing for their child. Of the 5 participants whose children had not received genetic testing, 4 (80%) expressed interest in pursuing it but had not for reasons including insurance denial, uncertainty regarding how to pursue it, and more pressing health concerns for their child. Lastly, this study identified deficits in respondents' understanding of GINA.
Conclusion: This study identified several important findings that have public health significance and can be utilized to develop a plan to address barriers to genetic testing within this patient population. To reduce the chance the genetic testing gets denied by insurance, healthcare institutions should make every effort to ensure patients receive an intake, evaluation, education, and consent by a genetic counselor. Additionally, genetic counselors can typically offer alternative finance options by working directly with the lab, to decrease the chance that cost is a barrier. Methods to address this and additional concerns regarding education and awareness within this population can be directed by the newly formed Cardiovascular Genetics Clinic at UPMC Children's Hospital of Pittsburgh.

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eP296

Familial *MECP2* variants: a report of 4 affected families highlighting the variable phenotypic spectrum and implications for genetic counseling

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Background: Pathogenic variants in MECP2 cause a spectrum of neurodevelopmental disorders, most commonly Rett syndrome (RTT; 312750). Although historically this was considered an X-linked dominant diagnosis with male lethality, it is now known that males can also be affected, with a disease spectrum ranging from severe neonatal encephalopathy to intellectual disability (ID). Other features commonly seen in RTT are inconsistently present in males. A