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Abstract

Title: Prenatal Exome Sequencing: Issues of Clinical Utility and Beyond

Prenatal exome sequencing (PES) is now being conducted in limited clinical settings. The purpose of this study was to assess genetic counselors’ perspectives toward previously identified issues with PES and to provide insight for future professional guidelines for this technology.

A survey was developed based on qualitative data, literature review, and expert opinion, which was distributed through eblast to the entire NSGC membership. Questions assessed attitudes toward issues surrounding PES, including clinical utility (CU), return of results, and others.

182 participants completed at least some portion of the survey. 82% agreed that there is CU using PES to diagnose fetuses with ultrasound abnormalities and 63% to prepare for a treatable childhood-onset condition. Participants showed less agreement regarding CU of other potential indications for PES and mostly disagreed that there would be CU for preparing for adult-onset conditions (treatment: 75%; no treatment: 85%). 80% agreed that PES differs from the use of ES in postnatal settings, mostly due to the option for termination (80%). There was disagreement as to what types of results to return prenatally and the relevance of current ACMG guidelines for return of secondary

findings. Participants prioritized returning secondary findings indicating childhood-onset conditions over adult-onset, with less priority given to treatment availability. There was agreement that there could be potential barriers to access PES, such as the costs involved (overall: 93%; out-of-pocket: 96%). Further research into the clinical utility of PES (50%) was ranked as the most important area of future study regarding this technology.

Participants are generally supportive of the current applications of PES technology. However, they are not as supportive of other potential uses of PES, and discrepancies exist in their opinions as to what types of information should be returned. These discrepancies, along with the identified potential for harms that PES brings, illustrate a need for professional guidance for this technology.