

Genetic Counselors' Current Practices, Challenges, and Needs for Support with Clinical Exome and Genome Sequencing

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In recent years, exome and whole genome sequencing (ES/WGS) have shown marked clinical utility in discovering the underlying etiology for complex or rare genetic diseases. These technologies have become increasingly incorporated into clinical practice as useful tools in the diagnostic journey for patients who have not had success with traditional genetic testing methods. At many institutions, genetic counselors play a central role in the process of ordering and disclosing results from genomic tests and are uniquely qualified to address some of the inherent challenges that have accompanied the rapid emergence of genomic technologies. A nationwide survey of genetic counselors was conducted to investigate these challenges and elucidate specific areas where support is needed. An anonymous online survey was distributed to genetic counselors through the American Board of Genetic Counseling (ABGC) and the National Society of Genetic Counselors (NSGC). 220 genetic counselors submitted surveys for analysis, of whom 77% currently utilize ES/WGS in clinical care. Results indicated that, in general, complexity of informed consent, results disclosure, and needs for support are all greater for ES/WGS than for other types of genetic testing. Genetic counselors who had been utilizing ES/WGS for less than 2.5 years were more likely to rate all three as 'significantly' more complex ($p < 0.05$). Respondents reported needing the greatest amount of support with variant interpretation and medical management related to both primary and secondary results. Those not yet involved with ES/WGS anticipated significantly higher needs for support than individuals who currently use it ($p < 0.05$), except with regard to interpretation ($p = 0.1$) and medical management ($p = 0.06$) of primary results. Finally, coding of qualitative data from open-ended survey items revealed trends related to unequal access of ES/WGS as a result of lack of insurance coverage, differences in provider thresholds for ordering testing, and lack of established eligibility criteria. These results suggest that institutional support services related to assessment of candidate patients, variant interpretation, and education of non-genetics providers would improve equality of patient access to testing and consistency in patient care. Ultimately, this would be beneficial for genetic counselors and other providers involved in ES/WGS as its scope and utilization across specialties continues to increase.