

Exome-Backbone Panels: Genetic Professionals' Perspectives in the Next Generation of Panel Testing

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Exome-backbone panels are an emerging genetic testing technology that blend traditional panel testing and genomic sequencing methods. There are no known studies assessing how genetics professionals understand this technology, how they perceive the benefits and limitations, or how these new panels are being marketed to these individuals. It is important to know the perspectives and needs of ordering providers regarding these tests in order to implement better communication with testing laboratories and improve quality of care for patients. This study recruited both genetic counselors and medical geneticists who had offered clinical genetic testing within the last 3 years. All participants were invited to complete an anonymous survey, which consisted of both multiple-choice questions and free text responses. In total, 195 eligible participants completed the survey. Of those, 188 were genetic counselors and 7 were medical geneticists. About 47% of participants had experience with ordering an exome-backbone panel. Reported benefits of the testing were the ability to reflex to other genes or exome data and the ability to customize panels. Participants felt drawbacks of the testing were concerns over incidental findings, inadequate gene coverage, or read depth, and a higher rate of variants of uncertain significance. The clinics in which exome-backbone panels were thought to have the most utility were pediatric and adult genetics, neurology, and cardiology, and those with the least suspected utility were prenatal and cancer genetics. This study shows that while exome-backbone testing is a newer approach to genetic testing, adoption of these tests is relatively high. However, perspectives of the testing are significantly different among those with experience with the test and those without experience. Additionally, differences in ordering practices in terms of consenting and results return suggest that ordering providers could benefit from professional guidelines for the implementation of these tests to provide better quality of care.