Dos Decision Regret Correlate with Results Received? A Study of Parents of Children who Have Undergone Whole Exome Sequencing

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ABSTRACT

Whole exome sequencing (WES) is pushing its way to the forefront of genetic testing strategies amongst both adults and children with complex disease. For affected individuals that have phenotypes that do not point to a specific causative gene, or have had previous inconclusive test results, WES is proving itself to be extremely useful in a clinical setting. WES is the most comprehensive genetic testing currently available, leading to a conclusive test result for approximately 25-30% of individuals, many of whom have had prior, inconclusive evaluations. However, since this test is new in the clinical realm, little research exists examining the psychological implications of WES, specifically decision regret. This is important to understand as the clinical use of this testing becomes more common. We surveyed parents of children who have undergone WES to determine whether decision regret correlated with their test result. Results demonstrated no statistically significant difference in decision regret amongst those with a positive, negative, or uncertain result. This study indicates that type of results may be a poor predictor of decision regret for those undergoing whole exome sequencing, and highlights the need for future research to identify potential factors that may influence decision regret amongst these patients.

METHODS

Study participants were recruited from a larger retrospective cohort study at New York-Presbyterian/Columbia University Medical Center (NYP/CUMC). Participants consisted of parents of children who had undergone genetic counseling for and testing via WES at NYP/CUMC and who completed a child primary survey. After eliminating incomplete responses, 205 surveys remained for data analysis of decision regret. Ages of respondents ranged from 20 to 67 with a mean age of 39.52 (SD=8.19). The majority of respondents reported themselves as female, married or living as married with a partner, white non-Hispanic, college or greater level of education, and employed full time. Indications for testing were varied and included developmental delay, cardiomyopathy, birth defects, hearing loss, cancer, dysmorphic features, autism, and seizures.

RESULTS

Two hundred and eighteen responses were recorded for individuals that had completed a child primary survey. After eliminating incomplete responses, 205 surveys remained for data analysis of decision regret. Ages of respondents ranged from 20 to 67 with a mean age of 39.52 (SD=8.19). The majority of respondents reported themselves as female, married or living as married with a partner, white non-Hispanic, college or greater level of education, and employed full time. Indications for testing were varied and included developmental delay, cardiomyopathy, birth defects, hearing loss, cancer, dysmorphic features, autism, and seizures.

Figure 1. Distribution of decision regret amongst those with a positive, negative and uncertain result. Positive cohort, n=82. Negative cohort, n=68. Uncertain cohort, n=55. Overall, there was not a statistically significant difference in decision regret amongst those with positive, negative, or uncertain results, (F(2, 202) = 0.324, p = 0.724). Medians for such can be viewed in Figure 1.

Figure 2. Distribution of decision regret scores between those of different education levels. Levels other than college degree, n=68. College degree or greater, n=145.

Figure 3. Mean regret score of those of different education levels.

CONCLUSIONS

• Results demonstrated no statistically significant difference in decision regret scores among those with a positive, negative, or uncertain result from WES
• There was a statistically significant correlation between education level and decision regret demonstrating that those with higher education levels (college degree or greater) experienced less decision regret
• The majority of individuals (54.9%) had a regret level of zero indicating that the majority of parents whose children underwent WES did not experience regret regardless of type of result received
• Overall, these results indicated families undergoing WES experienced low levels of decision regret

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