

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

Emily Rosen, MS¹, Julia Wynn, MS, CGC², Jill Fischer, MS, CGC¹, Nancy Frye, PhD¹, Wendy Chung, MD, PhD²
¹Long Island University-Post, Brookville, NY ²New York-Presbyterian/Columbia University Medical Center, New York, NY

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 total 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.

BACKGROUND

- The diagnostic rate of whole exome sequencing is estimated to be 25-30% across several ethnic backgrounds and for multiple indications. Many of these individuals have had previously inconclusive evaluations (Farwell et al., 2015, Iglesias et al., 2014, Yang et al., 2014, Yang et al., 2013).
- WES has been used to diagnose various Mendelian conditions such as retinitis pigmentosa, Knobloch syndrome, and amelogenesis imperfecta (Haghighi et al., 2014, O'Sullivan et al., 2011, Züchner et al., 2011). However, WES is also a powerful tool for discovery of candidate genes for complex disorders.
- In addition to the powerful diagnostic capabilities of WES, it has proved to be more effective and cost-efficient than the traditional method of testing a single to a few genes for individuals with complex, unexplained phenotypes (Valencia et al., 2015).
- Anxiety, depression, distress, and several other factors are known to exist within populations undergoing genetic testing. However, due to the novelty of clinical WES, there currently is little research examining the specific psychological implications of undergoing such. Particularly, there are few studies that examine the psychological implication of decision regret.

OBJECTIVES

- Determine decision regret amongst patients who have undergone WES
- Determine how the test result influenced decision regret levels
- Determine possible correlation between levels of decision regret and feelings about the testing experience

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 received results between April 2012 and October 2015. Respondents reported type of results for their child. These were grouped into three categories: (1) positive, (2) negative, or (3) uncertain. Each respondent completed the Decision Regret Scale (Brehaut et al., 2003) and a scale score was created. Higher scale scores indicated greater decision regret. Scale scores were then averaged within each group (positive, negative or uncertain). Difference in level of regret across types of results was measured with a one-way ANOVA. Patients also reported demographic information, such as gender and education level. Lastly, patients were asked to detail the (1) positive and (2) negative aspects of WES. Gender, education level, and number of aspects positive and negative about WES were then analyzed for correlation with decision regret.

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.

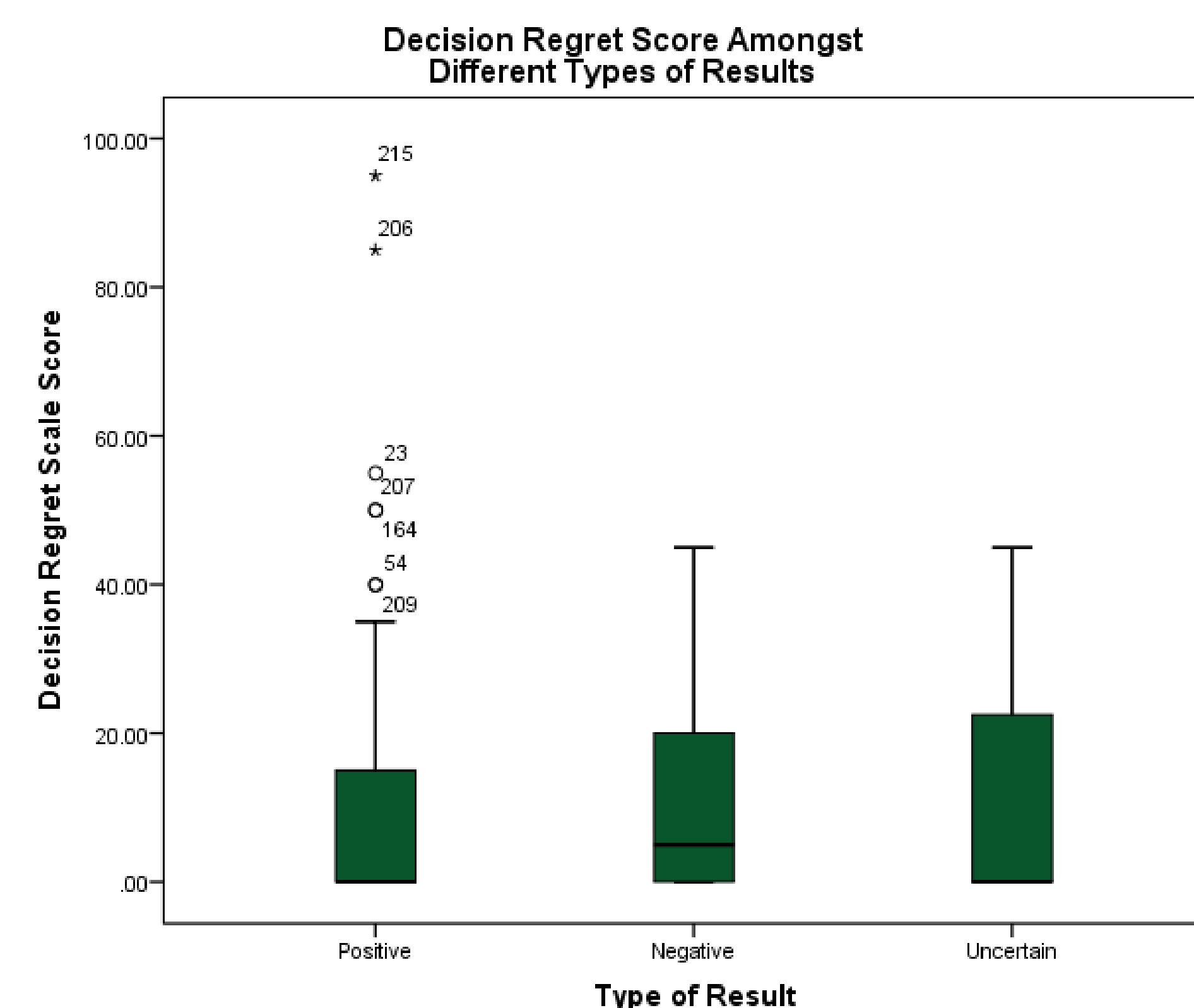


Figure 1. Distribution of decision regret amongst those with a positive, negative and uncertain result. Positive cohort, n=82. Negative cohort, n=68. Uncertain cohort, n=55.

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**.

RESULTS

The majority (54.9%) of respondents had a scale score of zero indicating that they did not experience any decision regret. Additionally, 43.8% of participants had scale scores between 5.00 and 50.00. Only 1.5% of respondents had scale scores higher than 50.00. Considering that higher regret scores indicate higher levels of regret, these results demonstrate that the majority of parents did not experience high levels of decision regret. Respondents with higher regret scores reported a greater number of aspects of the WES experience to be negative ($r(213) = 0.277, p < .05$), and fewer aspects to be positive ($r(213) = -0.348, P < .05$). There was no statistically significant association among gender, type of results received and decision regret score ($F(2,196) = .003, p = .997$). Likewise, there was no statistically significant association among education level, type of result received, and decision regret score ($F(2,198) = 0.380, p = 0.684$). There was; however, a statistically significant correlation between education level and decision regret when type of results were not taken into account ($F(1,198) = 5.079, p = < .05$) indicating that those with higher education levels experienced less decision regret.

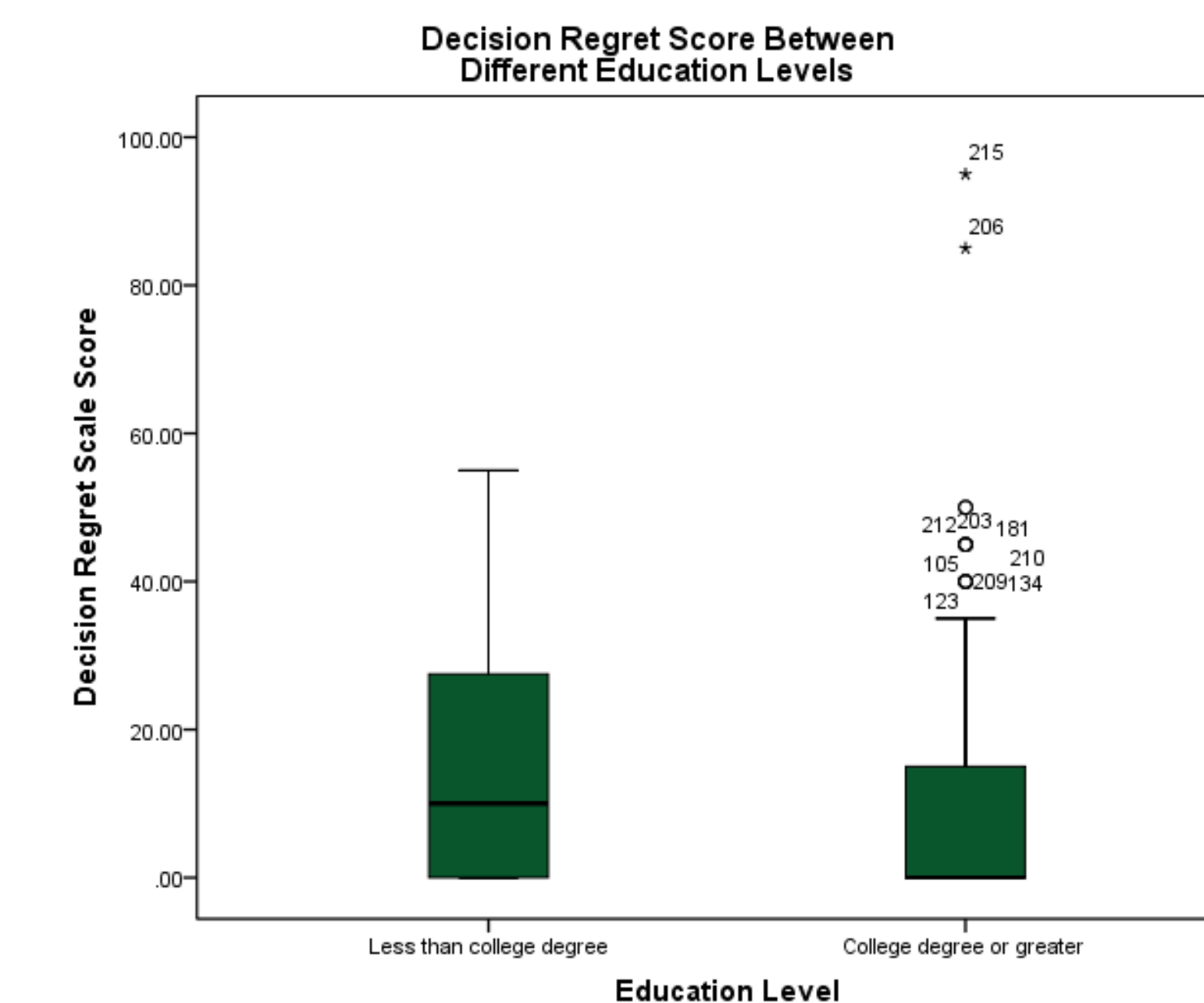


Figure 2. Distribution of decision regret scores between those of different education levels. Less than college degree, n=68. College degree or greater, n=145.

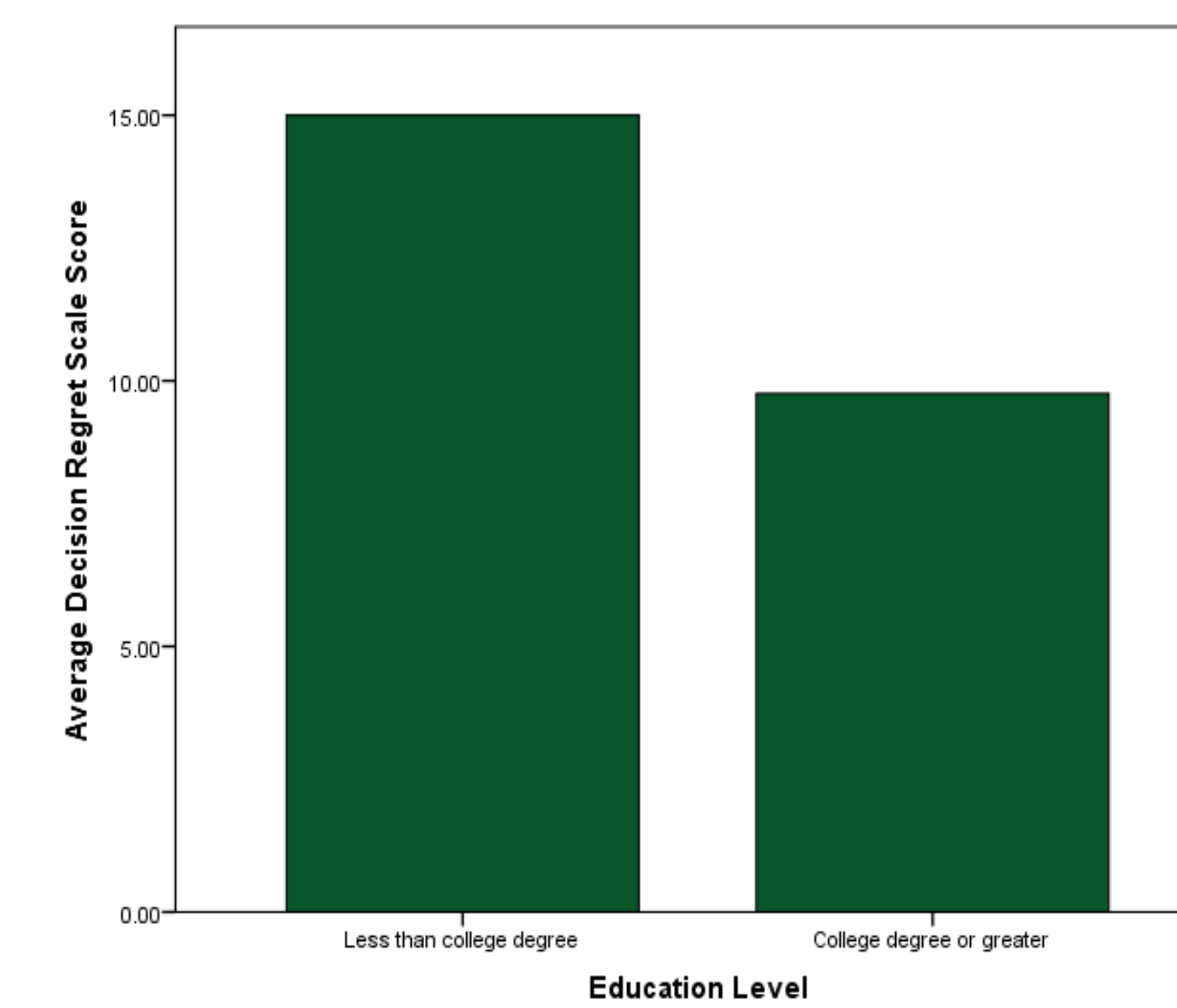


Figure 3. Mean regret score of those of different education levels.

CONCLUSIONS

- Results demonstrated no statistically significant difference in decision regret levels amongst 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

ACKNOWLEDGEMENTS

It is with immense gratitude that I acknowledge the support and help of: Long Island University-Post Genetic Counseling Graduate Program New York-Presbyterian/Columbia University Medical Center