ATTITUDES ABOUT PREDICTIVE MEN1 GENETIC TESTING IN MINORS

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Multiple Endocrine Neoplasia type 1 (MEN1) is a hereditary cancer syndrome characterized by tumors of the endocrine system. Tumors most commonly develop in the parathyroid glands, pituitary gland, and the gastro-entero pancreatic tract. MEN1 is a highly penetrant condition and age of onset is variable. Most patients are diagnosed in early adulthood; however, rare cases of MEN1 present in early childhood. Expert consensus opinion is that predictive genetic testing should be offered at age 5 years, however there are no evidence-based studies that clearly establish that predictive genetic testing at this age would be beneficial since most symptoms do not present until later in life. This study was designed to explore attitudes about the most appropriate age for predictive genetic testing from individuals at risk of having a child with MEN1. Participants who had an MEN1 mutation were invited to complete a survey and were asked to invite their spouses to participate as well. The survey included several validated measures designed to assess participants’ attitudes about predictive testing in minors. Fifty-eight affected participants and twenty-two spouses/partners completed the survey. Most participants felt that MEN1 genetic testing was appropriate in healthy minors. Younger age and increased knowledge of MEN1 genetics and inheritance predicted genetic testing at a younger age. Additionally, participants who saw more positive than negative general outcomes from genetic testing were more likely to favor genetic testing at younger ages. Overall, participants felt genetic testing should be offered at a younger age than most adult onset conditions and most
felt the appropriate time for testing was when a child could understand and participate in the testing process. Psychological concerns seemed to be the primary focus of participants who favored later ages for genetic testing, while medical benefits were more commonly cited for younger age. This exploratory study has implications for counseling patients whose children are at risk of developing MEN1 and illustrates issues that are important to patients and their spouses when considering testing in children.