An Evaluation of Parents' Experience with Expanded Genetic Testing for Autism Spectrum Disorders

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The autism spectrum disorders (ASDs) are a wide-ranging spectrum of conditions characterized by behavioral and developmental deficits in social interaction, language and communication abilities, and a restricted range of interests and activities. Although the etiology remains elusive in a majority of cases, research overwhelmingly supports genetic factors as contributory. Until quite recently, genetic testing by karyotype analysis and fragile X molecular testing were standard of care for individuals with ASD. Recent findings have updated this recommendation to include whole-genome comparative microarray analysis (CMA) in standard of care practices. As this recommendation is adopted, an increasing number of families may be referred for this "expanded" genetic testing for ASD. Via anonymous survey, this study investigated the attitudes and experiences of 58 parents after having a child with ASD undergo expanded genetic testing at Children's Hospital Boston (CHB). Most parents reported a positive experience with the expanded genetic testing process (86%), felt that their child's expanded genetic testing results were adequately explained to them (66%), and indicated that, overall, the experience was positive (~77%). Sixteen percent of parents whose child received normal genetic testing results inaccurately believed that their child had never undergone any genetic testing. Parents whose child received positive genetic testing results by karyotype analysis or CMA testing consistently indicated that their child had received an abnormal/positive genetic test result, but inaccurately indicated which test result was abnormal (100%). Overall, parents whose child received Variant of Uncertain Significance (VUS) genetic testing results by CMA most accurately reported the genetic testing that their child had undergone and the results of that testing. Regardless of when genetic testing was offered in relation to when their child was diagnosed with ASD, the majority (93%) of respondents were satisfied with the timing of genetic testing. Based on these and other findings, the most valuable issues to address in a genetic counseling session in this population were determined.