

B-23. Autism Spectrum Disorder in Boys with Fragile X: Serial Evaluation

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We have reported that the diagnosis of autism spectrum disorder (ASD) in boys with Fragile X (FraX) is associated with distinctive abnormalities in adaptive socialization and aberrant social behavior (AJMG 2004;126A:9). We also demonstrated that impairment in adaptive socialization is a better predictor of Autism Diagnostic Interview-Revised (ADI-R) scores and ASD diagnosis than communication parameters. This study examined the stability of ASD diagnosis, and associated factors, over a 1-year period in FraX boys. We evaluated 39 subjects with standardized measures of cognition, language, and adaptive, aberrant, and autistic (ADI-R) behaviors. Diagnosis was stable, across a spectrum of ASD severity (none, PDD-NOS, autism), for 72% of subjects. Of the approximately same proportion of subjects with more (13%) or less (15%) severe discrepant diagnosis, only two (5% sample) had changed from none to autism or vice versa. Adaptive socialization was still the greatest predictor of ADI-R scores and diagnoses, and other socialization-related measures were once more associated with ASD diagnosis. These data further support the presence of a distinctive ASD subphenotype in FraX, which represents a wide-ranging disturbance in social behavior. Supported by MH67092.