Co-Occurrence of Behavioral, Psychiatric, and Medical Issues in Families Ascertained for Autism and Language Learning Impairment

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Introduction: Converging evidence suggests that in families where there is an individual with an autism spectrum disorder (ASD), relatives may also exhibit behavioral, medical, and/or psychiatric characteristics that are milder but qualitatively similar to the defining features of autism. This observation has been coined the Broad Autism Phenotype—BAP (see Losh et al., 2008). Furthermore, research suggests that these conditions occur more frequently in families where there is an individual diagnosed with autism when compared to control families. Before undertaking any family linkage and association analyses, strong phenotypic characterization of these family members is essential.

The New Jersey Language and Autism Genetics Study (NJLAGS) has studied families ascertained through (at least) one proband with autism and a second proband with a significant language-learning impairment. Each family member has also been directly assessed to identify family members who exhibit elevated scores on ratings of (1) language deficits, (2) rigid personality traits/social aloofness, and (3) medical and psychiatric inventories, yet do not meet any of the criteria for autism. The primary goal is to create behavioral biomarkers related to language and other conditions associated with autism. Once proband criteria are met, the family completes a comprehensive neuropsychological battery and donates a blood sample for DNA analysis.

Study Ascertainment & Methods: Autism probands are diagnosed using the ADI-R, the ADOS, and a comprehensive physician’s examination including the DSM-IV.

• Language-Learning Impaired probands are diagnosed by a licensed Speech/Language Pathologist and is defined as a family member with oral and/or written language skills significantly below his/her peers.
  1) reflected on comprehensive standardized oral (speaking and listening) and written (reading and spelling) language tests, or
  2) as significant weaknesses in specific language domains interventions.

Children’s Yale-Brown Obsessive Compulsive Scale

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