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Complex phenotypes of autism: a case study on AUTS2 and high intelligence

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Statement of the Problem: In neurodevelopmental disorders, Autism (ASD) susceptibility to gene 2 (AUTS2) has been associated with severe clinical manifestations, e.g. intellectual disability (ID) and congenital malformations. Hitherto, genetic investigation has focused on syndromic rather than non-syndromic ASD. Although some reports have investigated the genetic etiology in non-syndromic, high-functioning ASD, no study has described AUTS2 disruption associated with high intelligence in ASD. Methodology & Theoretical Orientation: The development of RP, a boy diagnosed with autism, high intelligence and a microduplication of AUTS2 was followed during 13 years of his life, from 10 to 23 years old. Four neuropsychological assessments were performed, evaluating intelligence, fine motor skills and executive functions. The findings will be interpreted considering the genetic and bioinformatics results, analyzing the contributions of the study case to the description of the ASD phenotypes associated with AUTS2.

Findings: Bioinformatics research displayed 87 CNVs overlapping AUTS2: a phenotype was described for six of the twelve duplications, including delayed speech and language development, seizures, ASD, mild developmental delay, global developmental delay, cleft palate, ataxia, and ADHD. RP's neuropsychological assessment showed improvement in motor fine skills over time. Two assessments using the Wechsler scales revealed an IQ=123. A discrepancy between executive IQ and verbal IQ was consistently observed. Processing speed was the highest score in both assessments followed by perceptual organization. Vocabulary was the lowest score on the first assessment and increased on the second assessment. In contrast, comprehension was average on the first assessment, and decreased later, which may reflect the patient's socio- pragmatic deficiencies. Attention and executive function difficulties observed in the first assessment persisted over time.

Conclusion & Significance: Actually, the genetic investigation is far more frequently developed in syndromic than in non-syndromic ASD. Few reports have been published investigating genetic etiology in non-syndromic, normal high intelligence persons having ASD.

Biography

Giulia Moreira Paiva is a neuropsychologist. She concluded her master study and is current a PhD student in Neuroscience at Federal University of Minas Gerais (UFMG). Giulia has extensive experience with literacy and inclusive education, as well as developing assessments and interventions aimed at children with learning disabilities, neurodevelopmental disorders and genetic syndromes. She is member of the Board Committee of the Brazilian Society of Neuropsychology (SBNp), collaborator at the Developmental Neuropsychology Laboratory (LND- UFMG) and researcher at INCEI-TREINITEC. At INCEI she develops research and innovative tools aimed at inclusive education and interventions for children and adolescents on the autism spectrum.

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