NRXN1 EXONIC DELETIONS IN BRAZILIAN PATIENTS WITH IDIOPATHIC AUTISTIC SPECTRUM DISORDERS

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Introduction. Autism spectrum disorders (ASD) represent a group of neurodevelopmental disorders characterized by a core set of social-communicative and behavioral impairments. The neurexin-1 (NRXN1) gene mapped on chromosome 2p16.3 encodes a cell adhesion molecule in the nervous system. Inherited or de novo deletions suggested neurexin as a candidate gene for the ethiopathogenesis of ASD. **Objective.** To investigate Copy Number Variations (CNVs) in NRXN1 gene in patients with idiopathic ASD. Methods. A CNVs screening in NRXN1 of 400 idiopathic ASD and 200 healthy controls Brazilians population was made using Multiplex Ligation dependent-Probe Amplification (MLPA) and custom array – CGH. The clinical data of the positive patients were compared with those of previously described. **Results.** We found two patients with exonic deletions in NRXN1, a 0.5% frequency of this alteration in ASD population. All controls were normal. The clinical features were similar those previously described. **Conclusions.** The data of screening for CNVs on NRXN1 gene in ASD Brazilian individuals suggests that NRXN1 exonic deletions can be not so rare in this population. Dysmorphisms and behavioral phenotype can be related to the locations of exonic deletions in NRXN1. Based on gene function NRXN1 in central nervous system, can be suggested the involvement of CNVs observed in the pathogenesis of psychiatric illness of our patients, which can direct the process of genetic counseling of the families involved.

Keywords: Neurexin-1; ASD; dysmorphic; MLPA; array-CGH

Finantial: support: BAP/FAMERP