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Subtelomeric region of chromosome 2 in patients with autism spectrum disorders

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ABSTRACT. Autism spectrum disorders are severe psychiatric diseases commonly identified in the population. They are diagnosed during childhood and the etiology has been much debated due to their variations and complexity. Onset is early and characterized as communication and social interaction disorders and as repetitive and stereotyped behavior. Austistic disorders may occur together with various genetic and chromosomal diseases. Several chromosomal regions and genes are implicated in the predisposition for these diseases, in particular those with products expressed in the central nervous system. There are reports of autistic and mentally handicapped patients with submicroscopic subtelomeric alterations at the distal end of the long arm of chromosome 2. Additionally, there is evidence that alterations at 2q37 cause brain malformations that result in the autistic phenotype. These alterations are very small and not identified by routine cytogenetics to which patients are normally submitted, which may result in an underestimation of the diagnosis. This study aimed at evaluating the 2q37 region in patients with autistic disorders. Twenty patients were studied utilizing the

fluorescence *in situ* hybridization technique with a specific probe for 2q37. All of them were also studied by the GTC banding technique to identify possible chromosomal diseases. No alterations were observed in the 2q37 region of the individuals studied, and no patient presented chromosomal diseases. This result may be due to the small sample size analyzed. The introduction of routine analysis of the 2q37 region for patients with autistic disorders depends on further studies.

Key words: Autism spectrum disorders; Chromosome 2; Fluorescence *in situ* hybridization; FISH technique

