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Molecular analysis of patients with autistic behaviour

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Background

Autism and Pervasive Developmental Disorders (P.D.D.) belong to the group of neurodevelopmental disorders with a prevalence of 5-10/10,000 and a male to female ratio of 3-4:1. During the last decade there has been significant progress internationally in the identification of genes predisposing individuals to autism [1]. This study aimed to analyse the Neuroligin 3 gene (NLGN3, Xq13) in patients with Autism Spectrum Disorders (A.S.D.) [2,3].

Materials and methods

The population studied includes 367 individuals (169 children, 154 mothers, 44 first-degree relatives). All patients had been characterised as autistic by neurologists and psychiatrists according to the DSM-IV criteria. In addition, all patients were examined by clinical geneticists and chromosomal aberrations as well as Fragile X syndrome were excluded. The mutations Y74Y and R451C of the NLGN3 gene were screened in our sample.

Results

The total of 367 samples were examined with ARMS PCR but none of them was positive for the mutation Y74Y. The dHPLC screen for the R451C mutation has so far been performed in 200 out of 367 samples. These samples didn't prove positive for the existence of this mutation.

Conclusions

This is the first molecular study of individuals with A.S.D. in Greece. In our sample the research performed didn't reveal the Y74Y or R451C mutations in the NLGN3 gene. The study will be continued for the completion of the R451C mutation screen and will extend to screen the NLGN4 gene, which is also a candidate for A.S.D. [3,4]. The final results will allow for a genotype-phenotype correlation in the Greek population.

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