

P01-182 - GENETIC SYNDROMES ASSOCIATED WITH AUTISM

C.G. Cozaru, C.-A. Papari, A. Papari

Faculty of Psychosociology, "Andrei Şaguna" University, Constanta, Romania

Autism is a heterogeneous neurodevelopment disorder that can reveal impairments in social interactions, communication, and behavior. This paper aims to review the current understanding of the etiologies and the multiple pathogenetic pathways that are likely to lead to the autistic phenotype. Multiple studies evidence that most cases of autism likely have an underlying genetic cause or predisposition. Just as mental retardation, autism is likely to be caused by many different genetic mechanisms and genes rather than a single, or few, major genes or environmental effects. Applying routine genetic testing in clinical (CLIA-certified) diagnostic laboratories it's possible to identify the specific etiology and recurrence risk in 10% to 15% of autism cases and is clinically indicated for any child with autism. In this study we report 5 cases with different genetic syndromes in which autism has been described as one of the possible manifestations: Down syndrome, 22q13 deletion, „cri du chat” syndrome - 5p15 deletion, Cornelia de Lange syndrome, Sotos syndrome.

Conclusions: The etiologies of autism are complex and presence of a wide variety cytogenetic abnormalities are providing us with extremely valuable information about the role played by genetics in autism and collaboration with psychiatrists. Such recognition and understanding will help clinicians implement syndrome-specific treatments of patients identified with a genetic cause of autism spectrum disorder. Early identification of this genetic disorder is critical not only to the individual patient but for the entire family.

Keywords: Autism, chromosomal abnormalities, genes.