P02-208

XQ28 DUPLICATION IN A BOY WITH MENTAL RETARDATION, HYPERKINESIA AND DYSMORPHIC FEATURES - A CASE REPORT

B. Budisteanu[1], A. Arghir[2], A. Tutulan-Cunita[2], S.M. Chirieac[2], M. Budisteanu[2,3], A. Lungeanu[2]

[1]Child and Adolescent Psychiatry, 'Prof. Dr. Alex. Obregia' Clinical Hospital of Psychiatry, [2]Laboratory of Medical Genetics, 'Victor Babes' National Institute of Pathology, [3]Pediatric Neurology, 'Prof. Dr. Alex. Obregia' Clinical Hospital of Psychiatry, Bucharest, Romania

Introduction: X-linked mental retardation (XLMR) is a common, clinically complex and genetically heterogeneous disease arising from many mutations along the X chromosome. Although research during the past decade has identified >90 XLMR genes, many more remain uncharacterized. In XY males, duplication of any part of the X chromosome leads to functional disomy of the corresponding genes.

Objective: In this paper we present the case of a boy with a syndrome of Xq28 duplication.

Methods: We present a 6 years old boy, admitted in the Department of Pediatric Psychiatry for evaluation. He presented sever mental retardation, autistic features, speech delay, hyperkinesia, and dysmorphic features (high forehead, partial palpebral ptosis, small nose, carp-shaped open mouth, micrognathia), recurrent infections. Cerebral MRI was normal. Genetic investigations, including katyotype with GTG banding and array-CGH, were performed.

Results: Array-CGH indicated a dup(X)(q28) of less than 1.5 Mb. There were 15 duplicated genes, including MECP2 gene, which is involved in autism and mental retardation.

Conclusions: Duplications at Xq28 are often associated with autistic features/non-syndromic MR; alterations in MECP2 gene (duplicated in our patient) are described in Rett syndrome or as a specific phenotype. The alteration occurring at Xq28 band is responsible for the patient´s phenotype. Clinical manifestation of this child will be compared with those of other patients with the same duplication previously described to further delineate this syndrome.

Acknowledgments: National Research Program PN II, Project 42-130, CAPACITATI 29/2007-2009 Project; CNCSIS, Project 1203.