P02-302

# INVESTIGATION OF MENTAL RETARDATION ETIOLOGY IN ROMANIAN CHILDREN USING CLINICAL, CYTOGENETIC AND ARRAY-CGH DIAGNOSTIC TECHNIQUES

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Introduction: Mental retardation (MR) is the most common developmental disability, affecting 2-3% of the general population. A major challenge in both clinical practice and research in the field of MR is to identify the underlying causes: genetic, chromosomal and environmental factors that have an influence on a person's development and behavior.
Objective: We present the results of our study regarding genetic abnormalities associated with mental retardation in children.
Methods: A total of 180 children were studied using a diagnostic protocol based on dysmorphologic and clinical assessment. A disease, familial and personal history were noted. All patients were evaluated by clinical and paraclinical exams (including dysmorphological features, psychological tests, neurological features, neuroimagistic studies). Genetic investigations included a karyotype with GTG banding, FISH and array-CGH.
Results: A specific causes for the mental handicap was identified in 80 children (44%). These included a chromosomal abnormality in 32 cases (17%), microdeletion syndromes in 25 children(14%), recognizable syndromes in 23 (13%). Array CGH identified a 22q11 deletion in a girl with unusual phenotype for DiGeorge syndrome, a Xp21 duplication in a girl with severe phenotype (including sever mental retardation, epilepsy, dysmorphic features, genital anomalies, glaucoma, dental anomalies), and a 4p14 deletion in a girl with moderate mental retardation, dysmorphic features, diparesis, congenital heard malformation.
Conclusions: While clinical diagnosis and conventional techniques form the mainstay of investigation of children with mental retardation, array CGH proved important diagnostic tool.
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