P-764 - (MICRO)DELETION/(MICRO)DUPLICATION SYNDROMES IN CHILDREN WITH MENTAL RETARDATION

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Objective: (Micro)deletion/(Micro)duplication syndromes in one of the most common cause of intellectual disability in children, often in association with a sever phenotype. Introducing of new genetic techniques of molecular diagnosis, like array-CGH, allowed identification of new microdeletion/microduplication syndromes. This paper presents our experience regarding diagnosis and management of children with (micro)deletion/(micro)duplication syndromes.
Material and methods: 250 children with mental retardation, selected using a diagnostic protocol based on personal and familial history, general and neurological examination, dysmorphologic and psychological assessment, specific paraclinical tests, were included in our study. In 130 children genetic investigations, including karyotype with GTG banding, FISH and array-CGH, were performed.
Results: 44 (micro)deletion/(micro)duplication syndromes were diagnosed: 16 cases with Williams syndrome, 10 cases with Angelman syndrome, 3 cases with Prader-Willi syndrome, 2 cases with Wolf-Hirschorn syndrome, 3 cases with cri-du-chat syndrome, and one from the following syndromes: DiGeorge, 1q deletion, 3p deletion, 3q duplication, 4p deletion, 8p deletion, 9p deletion, 12p duplication, Xp duplication, Xq duplication. The management of these children included physical therapy, speech therapy, behavioral therapy, the therapy of associated conditions (epilepsy, malformation etc.).
Conclusions: An early diagnosis of (micro)deletion/(micro)duplication syndromes is very important for a proper management of these conditions. New molecular genetics tests are useful for identification of some new or very rare anomalies.
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