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AB078. Application of array comparative genomic hybridization in clinical diagnostics of intellectual disability/developmental delay in children

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Background: Array-based comparative genomic hybridization (array-CGH) has been shown to be a new successful tool in identifying genetic defects underlying intellectual disability (ID) and/or developmental delay (DD). This study was designed to analyze and evaluate the potential pathogenic genomic imbalance in children with unexplained ID/DD and its association with phenotypes, and to investigate the value of array-CGH.

Methods: A total of 72 Children with ID/DD were evaluated by the array-CGH for detection of genomic copy number variations (CNVs).

Results: In this study G-band karyotyping of peripheral blood cells showed no abnormalities in all the children. The results of the array-CGH revealed that 10 (14%) of the 72 patients had pathogenic CNVs, in that 6 cases were identified with pathogenic CNV in a single chromosome, 2 cases had multiple microdeletions and 2 cases had combined microdeletion and microduplication, 2 cases had pathogenic CNVs in chr 1p36 and Xq28 region. One case had variation of unknown significance in 15q11.2. Large bands of copy neutral loss of heterozygosity were detected in 2 cases, comprising more than 10% of genome.

Conclusions: Array-CGH allows for the etiological diagnosis in some of the children with unexplained ID/DD. As a high-throughput and rapid tool, it has a great clinical significance in the etiological diagnosis of ID/DD.

Keywords: Array-based comparative genomic hybridization (array-CGH); intellectual disability (ID); developmental delay (DD)

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