Identification of gene copy number variations in patients with mental retardation using array-CGH: Novel syndromes in a large French series

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Résumé en anglais	Array-CGH has revealed a large number of copy number variations (CNVs) in patients with multiple congenital anomalies and/or mental retardation (MCA/MR). According to criteria recently listed, pathogenicity was clearly suspected for some CNVs but benign CNVs, considered as polymorphisms, have complicated the interpretation of the results. In this study, genomic DNAs from 132 French patients with unexplained mental retardation were analysed by genome wide high-resolution Agilent 44K oligonucleotide arrays. The results were in accordance with those observed in previous studies: the detection rate of pathogenic CNVs was 14.4%. A non-random involvement of several chromosomal regions was observed. Some of the microimbalances recurrently involved regions (1q21.1, 2q23.1, 2q32q33, 7p13, 17p13.3, 17p11.2, 17q21.31) corresponding to known or novel syndromes. For all the pathogenic CNVs, further cases are needed to allow more accurate genotype-phenotype correlations underscoring the importance of databases to group patients with similar molecular data.

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