Research Note

Array-CGH analysis in patients with intellectual disability and/or congenital malformations in Brazil

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ABSTRACT. In several patients, intellectual disability and/or congenital malformation may be attributed to chromosomal changes. In this study, we conducted an array-CGH test of 200 patients from the Northeast of Brazil with intellectual disability and/or congenital malformation. Blood samples were collected from the proband and from their parents when possible. DNA was extracted and investigated using the array-CGH test. Findings were evaluated for the pathogenicity in databases of benign and pathogenic changes (ISCA, UCSC, DGV, and DECIPHER). Forty-seven copy number variations (CNVs) were identified in 43/200 (21.5%) patients, including 25/98 (25.5%) in males and 22/102 (21.57%) in females. We considered 33 of these to be clinically significant, reaching a diagnosis rate of 16.5%. The sizes of the CNVs varied from 102 kb to 24 Mb in deletions and from 115 kb to 140 Mb in duplications. In 10/47 (21.3%) patients, the rearrangement involved a sex chromosome. Thirty-nine patients had one chromosomal aberration, while 2 concomitant abnormalities were detected in 4 patients.

Ten of 47 CNVs (21.3%) were > 5Mb in size. Fifteen patients had CNVs related to known syndromes. This research highlights the contribution of submicroscopic chromosomal changes to the etiology of intellectual disability and/or congenital malformation, particularly the implication of chromosomal abnormalities detected using an array-CGH test, with a high rate of 16.5%. Thus, our results support the use of array-CGH replacing standard karyotype as the first-tier cytogenetic diagnostic test for patients with multiple congenital anomalies and/or intellectual disability.

Key words: Array-CGH; Congenital malformation; Intellectual disability; Northeastern Brazilian patients

