

ARRAY-CGH ANALYSIS IN BRAZILIANS PATIENTS WITH HOLOPROSENCEPHALY

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Presently array CGH has been considered the main genetic test in cases of idiopathic MR/MCA and it has been largely used for prenatal diagnosis, and as preimplantation genetic screening technology. HPE is the most common defect of the central nervous system in humans with an estimated prevalence of 1:250 conceptions and lower than 1:10.000 when live and still births were included. Few papers have been reported on chromosomal imbalances through high resolution oligonucleotide microarrays in patients with HPE; and they have confirmed the high frequency of rearrangements in these patients stressing the existence of a large number of loci corresponding to new candidate genes.

MATERIAL AND METHODS: We selected 30 individuals who presented within the more severe similar phenotype. Investigation of copy number changes was performed by array-CGH using the whole genome Cytosure™, ISCA V2 array 4X180K (Oxford Gene Technology, OGT, UK) containing ~180.000 oligonucleotides. MLPC was used in this study to validate deletions / duplications identified by arrayCGH, less than 1Mb. **RESULTS:** Array CGH analysis showed duplication in 13q14, and in 2p21 in two unrelated patients and 8p23 deletion in other patient. Discussion: Because the clinical and genetic heterogeneity existing in patients with HPE and the increasing survival of these patients we concluded that these analysis are indicated in those cases where the search for mutations in main causative genes were negative providing a better tool for genetic counseling. **CONCLUSION:** The main purpose of the present report is to stress the importance of array CGH in patients with HPE, since it represents a paramount point concerning genetic counseling and management to families.

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