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GENETICAL ANALYSIS IN 148 BRAZILIAN CASES WITH HOLOPROSENCEPHALY

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Holoprosencephaly (HPE) is a malformation sequence where the cerebral hemispheres fail to separate into distinct left and right halves. It can be associated with midline structural anomalies of the central nervous system and/or face. Classic HPE has a live birth prevalence of 1/16,000, but an incidence as high as 1:250 in conceptuses, and a worldwide distribution. The etiology of HPE is complex, with both environmental and genetic factors being implicated. Chromosomal abnormalities have been attributed as the main commonly identified cause. The subjects of this study were ascertained from a sample of 148 patients within the clinical spectrum of HPE belonging to the Hospital for Rehabilitation of Craniofacial Anomalies, University of São Paulo (HRAC-USP-Bauru/SP), and previously screened for mutation of candidate genes (SHH, ZIC2, TGIF, GLI2 and PTCH1) using the kit SALSA MLPA KIT P187 Holoprosencephaly Version 03 (MRC Holland, Amsterdam, Netherlands), with none mutation found. The MLPA technique revealed five aberrations out of 148 cases analyzed. These aberrations involved five deletions and one duplication in genes related to holoprosencephaly. Three non related individuals presented deletions at the SHH gene, which counts with 66% of cases. TGIF gene was deleted in one case and a duplication in ZIC2 gene was found in another case. Our results support the hypothesis that presence of chromosomal abnormalities in HPE are part of the etiology and highlights the potential of MLPA technique as an important tool in the diagnosis of HPE and genetic counseling.

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