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Copy number imbalances detected with a BAC-based array comparative genomic hybridization platform in congenital diaphragmatic hernia fetuses

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ABSTRACT. Congenital diaphragmatic hernia (CDH) is a phenotypically and genetically heterogeneous disorder, with a complex inheritance pattern. Structural abnormalities of almost all chromosomes have been described in association with CDH. We made a molecular analysis through array comparative genomic hybridization (array CGH) of a group of fetuses with prenatal ultrasound diagnosis of CDH and normal G-banded karyotypes. A whole genome BAC-array CGH, composed of approximately 5000 BAC clones, was carried out on blood samples from fetuses with prenatal ultrasound diagnosis of CDH and a normal karyotype (500-band level). All potential cytogenetic alterations detected on the arrays were reported. The array CGH analysis showed copy number gains and losses in 10 of 12 cases. Eighty-five clones showed genomic imbalances, and 29 clones displayed described copy number variations. We

identified a recurrent gain in 17q12 in two of 12 cases, which has not been previously described. Our results may contribute to determining the effectiveness and applicability of array CGH for prenatal diagnosis purposes, and also to elucidate the submicroscopic genomic instability of CDH fetuses.

Key words: Congenital diaphragmatic hernia; Genetic pathways; Array comparative genomic hybridization; Prenatal diagnosis

