Outros

(21520) - 8P INVERTED DUPLICATION/DELETION SYNDROME DETECTED IN TWO PRENATAL CASES

Joana Trindade[1]; Cláudia Alves[1]; Fernando Santos[2]; Isabel Cerveira[2]; Ana Azeredo[1]; Fernanda Baltar[1]; Mafalda Lopes[1]; Maria João Oliveira[1]; Paula Lindo[1]; Cecília Correia[1]; Margarida Reis-Lima[1]

1 - Unidade de Citogenética, SYNLABHEALTH Genética Médica, Porto; 2 - Unidade de Medicina Fetal - DPN, Hospital S. Teotónio, Viseu

Introdução

The 8p inverted duplication/deletion syndrome or invdupdel(8p) [ORPHA 96092] is a very rare structural rearrangement. The clinical manifestations include CNS anomalies, intellectual deficit, severe development delay, facial dysmorphisms and skeletal, orthopedic and heart defects. Only few reports described invdupdel(8p) in prenatal diagnosis.

Ultrasound is a fundamental tool for early fetal abnormalities screening, leading to early genetic diagnosis and medical intervention.

Objectivos

Cytogenetic studies in two prenatal cases with the same rare invdupdel(8p) rearrangement.

Metodologia

Case 1: amniocentesis at 21 weeks of gestation, due to fetal agenesis of corpus callosum. Array-CGH and karyotype analysis were performed.

Case 2: CVS at 12 weeks after high risk serum screening for trisomy 21. Chorionic villus karyotype followed by amniotic fluid karyotype and CGH microarray were performed.

Resultados e Conclusões

Case 1: Multiplex-PCR results showed a normal female fetus and the karyotype revealed a structural abnormality of chromosome 8 short arm [46,XX,der(8)del(8)(p23.1)dup(p23.1p11.2)]. Array-CGH analysis revealed a 8Mb deletion involving 8p23.3p23.1 regions (162 genes), and a 31Mb duplication of the 8p23.1p11.1 segment, encompassing several genes.

Case 2: Multiplex-PCR results showed a normal female fetus. Chorionic villus karyotype revealed an abnormal result confirmed by amniotic fluid karyotype [46,XX,der(8)?del(8)(p23.1)dup(8)(p11.2p23.1)]. Array-CGH analysis revealed a 6,8Mb deletion involving chromosome 8p23.3p23.1 regions (81 genes), and a 24,7Mb duplication of the 8p23.1p11.23 segment (325 genes).

In both cases parental karyotypes were normal, allowing us to classify the abnormalities as de novo. Pregnancies were terminated.

In the literature only few reports describe this anomaly detected prenatally, the majority arising de novo. In our cases, the array-CGH allowed a more accurate characterization of this rearrangement. Array-CGH and karyotype analysis provided a better understanding of the chromosome imbalance, therefore stressing that in many circumstances it is very important to rely on them as complementary techniques.

Genotype-phenotype correlations are crucial for genetic counselling and the orientation of medical decisions.

Palavras-chave : invdupdel(8p)