FCP169

MULTIPLE CONGENITAL ANOMALIES IN A FETUS WITH APPARENTLY BALANCED DE NOVO TRANSLOCATION, 46,XY,T(3;8)(Q27-29;Q21)

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Apparently balanced de novo translocations detected in prenatal analyses may be associated with phenotypic abnormalities in the fetus. We describe a de novo reciprocal translocation between the long arms of chromosome 3 and 8 in a foetus with multiple congenital anomalies.

A 26-year-old primigravida was referred for genetic counselling because of ultrasonographic findings of cleft lip, pes equinovarus, and single umbilical artery at her 23th week of gestation. Cytogenetic analysis of cordocentesis material revealed 46,XY,t(3;8)(q27-29;q21) karyotype. Karyotype of the parents were normal. The pregnancy was terminated at 24th week. Post-mortem examination of this male fetus revealed facial dysmorphism characterised by hypertelorism, proptosis, depressed nasal bridge, low-set ears, unilateral cleft lip and palate, absence of left nostril and micrognathia. Claw appearance of the hands, simian crease on the right hand, bilateral pes equinovarus, and hypoplastic right kidney with pelvic localisation were other abnormalities.

Previous prenatal studies show that approximately 6% of de novo reciprocal translocations have a risk of serious congenital anomaly. Normal karyotypes of parents and apparently balanced translocations do not exclude particular hot spots among the breakpoints, causing serious abnormalities in the offspring.

FCP170

A NEW AUTOSOMAL RECESSIVE SYNDROME WITH ABDUCTED THUMB, BRACHYDACTYLY, ROCKER BOTTOM FEET, AND JOINT DEFORMITIES: SIGNIFICANCE OF DETAILED PRENATAL ULTRASONOGRAPHY CONFIRMED BY POST-MORTEM EXAMINATION

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For a reliable genetic counseling concerning following offspring it is of primary importance to describe fetal malformations in detail during pregnancy. We report on two fetuses with hand and foot abnormality first diagnosed by prenatal ultrasonography.

Case 1. The female fetus was the product of consanguineous couples (third cousins). The mother was referred at 23th week of gestation because of ultrasonographic abnormality and anomalous fetus history of the first pregnancy. Karyotype of cultured amnioctyes was normal. The pregnancy was terminated at 24th week. Postnatal examination showed asymmetric face, high forehead, hypertelorism, flat nose, dysplastic ears, and hypoplastic mandible. The extremities were short. On the right hand there was a complete cutaneous syndactyly of the fingers 4-5 and partial webbing of fingers 2-3 and 4-5. 1st toes were bowed laterally. Fingers 2-5 were deviated laterally. Rocker-bottom feet, wide metatarsals and flexion deformities of both knees were other remarkable features. The autopsy was normal.

Case 2. Third pregnancy was terminated at 17th week of gestation due to the similar abnormalities. Postmortem examination showed mild facial dysmorphism with oedematous eye lids, hypertelorism, and depressed nose. At first sight short extremities were noted. There were flexion deformities of hands and knees. 1st toes were bowed laterally. Internal organs and placenta were normal.

The main compenents of fetuses are mild facial dismorphism, short extremities, brachydactyly, adducted thumb, rocker bottom feet, laterally placed 1th toes and a wide gap between 1st ve 2nd toes. This findings are not consistent with any previously recognised syndrome and represent a new condition with possible autosomal recessive inheritance.