Case report

Chromosomal mosaicism on amniotic interphase nuclei detected by multiprobe FISH

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Abstract. So far classical prenatal detection of chromosome aberrations has been limited to the evaluation of metaphase by means of time-consuming cytogenetic techniques. The MultiVision PGT test enables a simultaneous detection of aneuploidies of chromosomes 13, 18, 21, X, and Y, even 24 h after amniocentesis. In the presented case, this test detected prenatally a chromosomal mosaicism 69,XYY[35]/46,XY[65]. This result was not confirmed after birth, by the same test on blood smear. The discrepancy is difficult to explain.

Key words: mosaicism, multicolour-FISH, polyploidy, uncultured amniocytes.

We present a case of a pregnant woman from a group of high-risk pregnancy. Ultrasound examination, performed in the 17th week, revealed increased echogenicity of the foetal intestine, together with the presence of a “sandal gap” and shortening of the auricle. In the 24th week a two-week discrepancy between the foetal age evaluated by ultrasound and foetal age calculated from the last menses was observed. The biochemical results of CNS malformations markers were normal. However, in the MV PG-T test, 69,XYY triploidy was detected in 35% of cells, which could be evidence for 69,XYY/46,XY mosaicism (Figure 1). Despite this the foetal karyotype analyzed 10 days later was that of a normal male and did not confirm the preliminary diagnosis. Additionally, the MultiVision PGT test was performed following a standard culture of amniocytes. The test revealed an increased percentage of interphase nuclei with a triploidy – 69,XYY – at the level of 42% in comparison to uncultured amniotic cells. The pregnancy was
continued until the 33rd week, when it was terminated by a caesarean section because of dramatically increasing symptoms of gestosis. Newborn hypotrophy was found (birthweight 1710 g, Apgar score 6/7). Clinical and radiological diagnosis of congenital pneumonia was established. The MultiVision PGT test performed on blood smear after birth, did not confirm the mosaicism detected prenatally. Interpretation of the above facts posed serious medical problems. We presume that the presence of a triploid line in the early stages of embryonic development blocked cellular divisions in the transformed cells, the only clinical consequence of which was foetal hypotrophy (QUIROZ et al. 1985, TOPALOGLU et al. 1998). The mother had a history of viral infection (influenza) in the first trimester of gestation, with high body temperature. It cannot be excluded that the observed effect may have resulted from that infection, but the mechanism by which it could have been possible, is not clear.

The presented case may prompt further studies on the correlation between the detection of a cellular line (69,XY Y), where the cells were unable to proceed in the life cycle in vivo or in vitro to metaphase due to a metabolic block, and foetal hypotrophy detected both pre- and postnatally. At the age of 3, the boy demonstrates normal psychological development, but during the initial 12 months of his life, he required motor rehabilitation. Physical development below the lower limit (body weight: > 11 kg, below the 3rd percentile, body height: 95 cm, between the 3rd and 10th percentiles). The results of the MultiVision PGT
test enabled including the pregnant woman in the group of high-risk pregnancies and intense monitoring of the course of pregnancy, and in effect a delivery which was safe for both the mother and the child.

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REFERENCES


