Confined Placental Mosaicism in Chorionic Villus Sampling
- Case Report -

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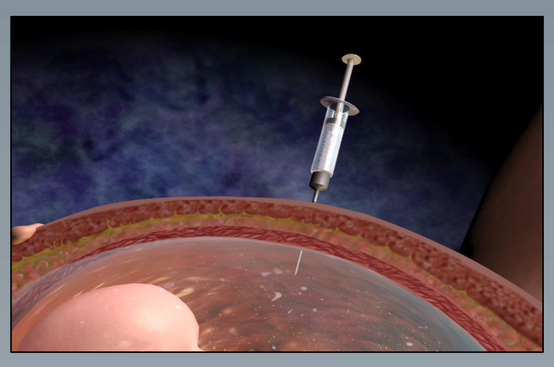
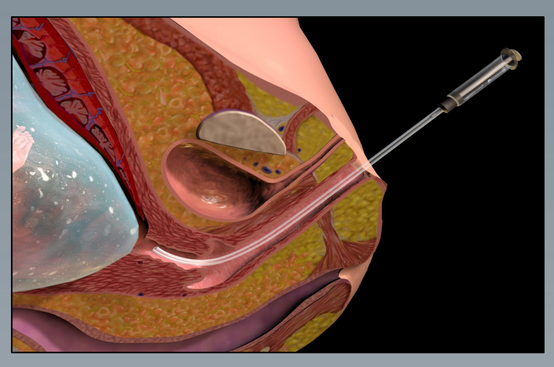
OBJECTIVES

Chorionic Villus Sampling (CVS) has several advantages over amniocentesis: it may be performed at an earlier gestational age, the results are quicker to obtain and there's a lower miscarriage risk – 1%. However, the higher prevalence of discrepant fetal and villus sampling material's karyotype findings is a disadvantage of this technique – 0.5%. This is caused, amongst other causes, by placental mosaicism which consists of two genetically different cell lines. There are three types of placental mosaicism according to the abnormal cell line location: Type I – in the cytotrophoblast; Type II – in the villus' stroma; Type III – in both the above locations.





MATERIAL AND METHODS

We present a case report about a 36-year-old pregnant woman going through our Department's 1st trimester combined screening program; a CVS was performed, which showed Confined Placental Mosaicism (CPM).

RESULTS AND CONCLUSION

Although the pregnant woman was in the low-risk group for aneuploidy, the patient wanted the cytogenetic study to be performed in order to reduce maternal anxiety. CVS was performed at the gestational age of 12 weeks + 5 days and the karyotype was 47XY+2/46XY. For the correct interpretation of this data an amniocentesis was performed at the gestational age of 15 weeks + 6 days, which showed a 46XY karyotype. We therefore conclude that the cytogenetic analysis of the CVS was the result of a CPM. A careful follow-up including fetal echocardiogram and seriated ultrasonographic monitoring was used to safely exclude malformations and fetal growth restriction. We verified no occurences throughout pregnancy, delivery and perinatal period.

CVS practice was recently implemented in our country and has many advantages over amniocentesis. Besides the fact that an earlier gestational age usually means less affective bonding to the fetus and therefore makes medical termination of pregnancy somewhat less difficult, one should consider specific situations like the one reported in which CPM may be diagnosed. This condition is associated with increased risk of fetal growth restriction, so the clinician should be aware of the need for a more careful follow-up, since perinatal complications, which should be anticipated and treated, can be expected in 16-21% of these cases.

References:
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