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A case of mosaic trisomy 13

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Objective

Prenatal diagnosis of fetal aneuploidies is routinely done by traditional cytogenetic culture. Commonly it is also performed a quantitative fluorescent polymerase chain reaction (QF-PCR), because of its faster results to identify common aneuploidies. The aim of this work is to present a case with ambiguous prenatal test results and discuss the advantages and limitations of currently available genetic tests for the diagnosis of prenatal mosaicism.

Methods

This is a case report of a prenatal mosaic trisomy 13 and review of current genetic test available for prenatal mosaic diagnosis.

Results

A 37-year old woman with a non consanguinous second pregnancy was referred at 12 weeks and 2 days for trisomy 13 positive screening. Noninvasive prenatal testing (NIPT) was positive for trisomy 13 at 10 weeks' and first trimester screening (FTS) calculated a risk of trisomy 13 of 1: 48 and a risk of trisomy 21 of 1: 167, without ultrasound abnormalities. Chorion villus sampling performed for QF-PCR was found to be normal. Considering both positive screenings for trisomy 13, amniocentesis was performed: QF-PCR was inconclusive and karyotype confirmed a mosaic trisomy 13: 47, XX, +13[23]/46, XX[6], as well as the prenatal chromosomal microarray analysis (CMA) with a nearly 60% trisomy line. Medical interruption of the pregnancy was performed at 17 weeks'.

Conclusion

Always consider the clinical findings and the possibility of false negative results, specialy if there is a mosaic possibility. QF-PCR and karyotype to exclude a trisomy suspition are unquestionable options. However, in particular cases, direct analysis of uncultured amniocytes by fluorescence in situ hybridization (FISH) or other molecular genetic tests such as CMA or next generation sequencing (NGS) might be considered, particularly due to limitations in diagnosing mosaicisms, such as the possibility of selective growth disadvantages.