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Cf DNA for Monosomy X and X chromosome variants in relation to ultrasound findings

Bedei I, Gehrke T, Gloning KP, Wolter A, Schenk J, Weber A, Axt-Fliedner R
Justus-Liebig University Germany, Gießen, Germany

Objective

Evaluation of the performance of cf DNA for Monosomy X (MX), mosaics/X chromosome variants in relation to anomalies on prenatal Ultrasound. Further, investigation of the uptake of invasive testing after a high risk cf DNA result, prenatal phenotype in monosomy X and X chromosome variants and outcome of affected pregnancies.

Methods

Multicenter retrospective survey study.

Results

Cf DNA results, phenotype, pre-or postnatal karyotype and outcome was available in 55 pregnancies. 48/55 were high risk for MX. Of these, 23 were TP and 25 were FP. 32/48 with a high risk cf DNA result did not show anomalies on prenatal ultrasound. 7/32 fetuses with high risk result and no US anomaly were TP, all were mosaics or genetic variants and not MX. All 16 cases with high risk NIPT and abnormal US were TP. Fetuses with MX showed more often "typical" anomalies like cystic hygroma and fetal hydrops, while genetic variants showed other anomalies on prenatal US. 7/55 were false negative, all but one of these fetuses showed anomalies on prenatal ultrasound. 44 pregnant women with a high risk cf DNA for MX opted for prenatal invasive testing. 29 of these did not show anomalies on prenatal US. Life birth rate was significantly higher in fetuses with a genetic variant of the X chromosome (63.6%) than for fetuses with MX (16.7%), p= 0.036.

Conclusion

Cf DNA can detect both, MX and X chromosome variants. In the presence of fetal anomalies, the result is more likely to be TP. In the absence of US anomalies, it is most often a false positive result or a fetus with a mosaic/X chromosome variant. Uptake of invasive testing after a high risk cf DNA result is high, independent on anomalies on prenatal US. Fetal outcome is less favorable in case of MX.