An atypical case of Turner’s Syndrome

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Introduction

Turner’s syndrome is a genetic disease with a karyotype of 45, X or 46, XX/45, X (mosaicism). 1/2500 female live births are affected, although 15% of spontaneous miscarriages show an X monosomy and only 1% of fetuses will survive to term. Typical prenatal findings are nuchal cystic hygroma, nonimmune hydrops, and renal and cardiac abnormalities. However, neither sonography nor maternal serum screening can be considered diagnostic of Turner syndrome, and karyotype confirmation is mandatory.

Case

38 yrs old III Gravida II Para presented for first trimester screening in 12+4 gestational weeks. The first child was born with biliaar atresia and needed living liver donation at the age of 3 weeks. The 2nd child is healthy. This fetus presented with a nuchal fold of 3.3 mm and generalized edema at a CRL of 55 mm. The nasal bone was hypoplastic. The fetus showed hypertelorism, low set ears and a strawberry shaped head. The IT was not visible and a sacral spina bifida was detected. Besides an omphalocele, there was an congenital heart defect suspicious for TOF with pericardial effusion, cardiomegaly, VSD, overriding aorta and suspected pulmonary stenosis. CVS was performed but contaminated by maternal cells. In the framework of scientific research postnatal molecular karyotyping could finally confirm 45/X0 twice.

Conclusion

Turner syndrome can present with a broad heterogeneity of signs and symptoms. The wide range of prenatal features indicates that a number of different X-located genes are responsible for the complete phenotype. Nevertheless affected fetuses show a substantial number of early-onset signs that can be detected in the first trimester, i.e. cystic hygroma, fetal hydrops, cardiac defects and increased nuchal translucency. The difference in the appearance of nuchal fold in each chromosomal defect may reflect the heterogeneity of causes, hence invasive diagnostics is mandatory.