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A case of increased fetal nuchal translucency thickness and normal karyotype

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Objective

Increased nuchal translucency (NT) is associated with aneuploidies and other chromosomal abnormalities, a number of genetic syndromes and with structural congenital anomalies (mainly cardiac defects). The majority of fetuses with NT ≥ 3. 5 mm have a normal karyotype and the pregnancy outcome is highly dependent on the absence of anomalies on expert fetal ultrasound examination. The aim of this case report is to present prenatal and postnatal outcomes of fetus with increased NT and normal karyotype.

Methods

This is a case report.

Results

A 26-year old primigravida was referred at 12+0 weeks of single gestation for a routine ultrasound scan. None of the parents reported any family history of aneuploides, genetic syndromes or congenital malformations. The pregnancy was dated according to the CRL (59mm) and the NT was above the 99th percentile. (figure 1). The PRISCA 1 results showed low risk (PAPP-A=0. 92 MoM and fb-HCG=1. 13 MoM). A normal karyotype was obtained by NIPD*(A NIPD revealed a normal karyotype). We excluded infection and immunological causes (TORCH, type of blood group, Rh factor). The parents were informed in detailed and they decided to continue the pregnancy. That was very stressful for them. The ultarasound assessment was performed at 4 weeks intervals. The increased nuchal thickness was found at 19w3d (NT=7, 1mm) (figure 2.), but detailed US examination and fetal echocardiography didn't reveal any malformation (Figure 3-). The intrauterine growth was normal. In the 40th week of gestation, a female baby was delivered (4030g/52cm and APGAR score of 8/9). Nine months after delivery, the baby has normal physical and neural development without any complications.

Conclusion

After exclusion of chromosomal abnormalities, most fetuses with increased NT have an adverse pregnancy outcome. However, if the detailed ultrasound examination at around 20 weeks is normal, a favorable outcome can be expected with confidence, irrespective of initially increased NT. Parents should be informed in detail about the condition, diagnostic methods, possible consequences and probable outcomes of the pregnancy.