



Introduction

It is often pointed out that Nuchal Translucency (NT) thickening can be related to not only chromosomal anomalies but also structural abnormalities such as congenital heart disease, congenital diaphragmatic hernia, skeletal dysplasia and so on. Though the relation between first trimester increased NT and chromosomal abnormalities is well-defined, little has been known about the importance of NT thickening in fetuses with normal karyotype. We experienced a case of increased NT with normal karyotyping fetus from mother with abnormal karyotype and report with a literature review.

Case Report

A 35-year-old primigravid woman with balanced translocation between 6 and 21 undergone laparoscopic myomectomy 9 months ago made her first visit for first pregnancy trimester. Estimated date of conception was fixed from crown rump length at 10 weeks of gestation, with NT of 3.3 mm. She was referred to higher medical institution for non-invasive prenatal genetic testing (NIPT) and genetic counseling, and normal karyotype of the fetus was revealed. The fetus got the highest NT of 7 mm at 13 weeks of gestation, but increased NT had been diminished and gradually vanished before 24 weeks. She has been taken regular prenatal checkup and no other anomalies has been detected.

Discussion

- (1) Increased NT despite normal karyotyping can be associated with adverse pregnancy outcome.
Niroomanesh S, et al. Pregnancy outcomes of normal karyotype fetuses with increased nuchal translucency. Caspian J Intern Med. 2023 Fall; 14(4):732-736.
- (2) With an increase in NT, even if the karyotype is normal, the incidence of structural defects, genetic syndromes, abortion, and perinatal death increases.
Mula R, et al. Increased nuchal translucency and normal karyotype: perinatal and pediatric outcomes at 2 years of age. Iran J Obstet Gynecol Infertil. 2012;7:32-36.
- (3) Complications are directly related to the amount of NT in the first-trimester examination and it seems the higher NT level has a greater risk.
Souka AP, et al. Increased nuchal translucency with normal karyotype. Am J Obstet Gynecol. 2005;192:1005-1021.

Conclusion

We experienced a case of increased NT with normal karyotyping fetus from mother with abnormal karyotype. Tight observation of both mothers and fetuses should be done in normal karyotype fetuses with increased NT, and providing appropriate information through ultrasonography can be extremely useful.

Declaration of interest

The authors have no conflicts of interest.



Figure 1. Transvaginal ultrasonography finding at 10 weeks of gestation. Fetus with NT of 3 mm was revealed.



Figure 2. Transabdominal ultrasonography finding at 13 weeks of gestation. Thick NT of 6 mm was detected.