A case of increased nuchal translucency in an euploid fetus with heart abnormality

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Objective

Increased fetal nuchal translucency (NT) thickness is associated with chromosomal abnormalities, fetal death and a wide range of fetal malformations and genetic syndromes. An association has been reported between increased NT and increased risk of the heart and the great vessels' abnormalities even in the chromosomally normal fetus. We present a case of hypoplastic left heart syndrome diagnosed in a fetus with normal karyotype and increased NT at first trimester.

Methods

A 31 year-old woman gravida 2 para 1 was referred to our unit for increased fetal nuchal translucency (NT). We measured the fetal NT as 4, 2 mm with septate at 12 weeks of gestation. Chorion villus sampling procedure was offered to parents for karyotyping. The result was 46 XX, normal fetal karyotype. Detailed structural ultrasound examination at 18 weeks' gestation revealed an appearently normal fetus. Fetal echocardiography examination was performed and hypoplastic left heart was founded.

Results

The poor prognosis for hypoplastic left heart was explained to parents. The parents decided to terminate the pregnancy. The pregnancy was terminated at 19th weeks' gestation using vaginal misoprostol.

Conclusion

Increase NT is associated with chromosomal anomalies and is a strong marker for adverse pregnancy outcome such as miscarriage, intrauterine death, congenital heart defects and numerous other structural defects and genetic syndromes. For this reason, they are offered detailed ultrasound examinations later in the pregnancy. The risk of adverse outcome is proportional to the degree of NT enlargement.