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Pregnancy outcomes of fetuses with isolated increased nuchal translucency thickness

Melekoglu R, Faydali S, Celik E
Inonu University School of Medicine Department of Obstetrics and Gynecology, Malatya, Turkey

Objective

The purpose of this study was to evaluate the pregnancy outcomes of patients diagnosed with isolated nuchal translucency (NT) thickness (NT≥3. 5mm) in the first-trimester screening.

Methods

The records of all patients who administered for the first-trimester screening between 2014 and 2017 in the Prenatal Diagnosis and Treatment Unit of Inonu University were reviewed retrospectively, and the records of patients who underwent chorion villus sampling (CVS) for isolated increased NT thickness were evaluated. During the study period, crown-rump length (CRL) and nuchal translucency (NT) measurements were performed in accordance with the criteria established by the Fetal Medicine Foundation (FMF) in 11-13+6 week of pregnancy by two clinicians that have the FMF first-trimester screening certificate.

Results

Eighty-seven patients diagnosed with NT≥3, 5mm in the first-trimester screening between 2014 and 2017 in Prenatal Diagnosis and Treatment Unit of Inonu University were enrolled in this study. All patients were offered CVS option but 58. 6% (n=51) opted for an invasive prenatal diagnosis procedure. A normal karyotype result was detected in 56. 9% (n=29) of the patients who underwent invasive prenatal diagnosis and the remaining 43. 1% (n=22) had an abnormal karyotype. There were no pathological ultrasound findings in 37. 2% (n=19) of patients with normal karyotype in the follow-up visits that underwent CVS. Abnormal ultrasound findings were detected in 19. 6% (n=10) of patients who opted for invasive prenatal diagnosis with a normal karyotype result. Trisomy 21 was the most frequent chromosomal anomaly in patients with an abnormal karyotype outcome, whereas the most common sonographic abnormality was non-immune hydrops fetalis in fetuses with normal karyotype (Figure-1).

Conclusion

First-trimester NT screening is an essential part of the pregnancy follow-up at present. For a case with isolated increased NT (≥3. 5mm) at first-trimester screening, an invasive prenatal diagnostic test should be offered in the first step, and detailed ultrasound examination and fetal echocardiography should be performed in the early second trimester and mid-trimester to diagnose additional minor and major fetal anomalies if the results of the diagnostic test reveal a normal karyotype. Although the risk of chromosomal abnormality is high in these pregnancies, it should be kept in mind that about one-third of these fetuses will be born alive and healthy.