

Cardiovascular anomalies detected at very early anatomical scan in fetuses with increased nuchal translucency

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Introduction

Increased nuchal translucency (NT) is associated with congenital heart disease and is a common indication for early fetal cardiac evaluation. Advances in high-resolution ultrasound (US) enable assessment of fetal cardiac anatomy in the late first trimester, although experience with systematic evaluation at this stage remains limited. We aimed to describe cardiovascular anomalies detected at a very early anatomical scan in fetuses with increased NT.

Methods

This retrospective study included pregnancies referred to a tertiary center between 2018–2025 following detection of increased NT (≥ 3.0 mm). A detailed scan was performed at 12–13 weeks using transabdominal and transvaginal approaches, including systematic assessment of fetal cardiac and extracardiac anatomy. US findings, genetic testing results, follow-up imaging, and pregnancy outcomes were recorded.

Results

A total of 144 fetuses were included. Cardiovascular anomalies were detected in 67 cases (46.5%). The spectrum of cardiovascular anomalies ranged from major congenital heart defects, such as atrioventricular septal defect, to subtler findings, including chamber asymmetry and great vessel disproportion. Among cases with suspected cardiovascular anomalies, 61 pregnancies were terminated, often associated with extracardiac malformations (n=49) and genetic abnormalities (n=30). In 3 of the 14 cases with an isolated cardiovascular anomaly (21.4%), parents chose termination of pregnancy (TOP) based on the early diagnosis of a major cardiac anomaly without genetic testing. Postmortem evaluation was not available following TOP. Among the six cases that continued, follow-up US confirmed the anomaly in three cases, while in one case the anomaly was ruled out, and in two cases the great vessels asymmetry resolved.

Conclusion: Very early anatomy scan in fetuses with increased NT can identify a broad spectrum of cardiovascular anomalies. Early recognition may facilitate timely genetic evaluation, assessment for extracardiac anomalies, and informed counselling regarding pregnancy management.