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Reversed ductus venosus A-wave during first-trimester screening revealing trisomy 21: report of two cases

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Introduction

First-trimester screening for chromosomal abnormalities combines nuchal translucency measurement with Doppler assessment of the ductus venosus. An absent or reversed A-wave in the ductus venosus has been associated with an increased risk of aneuploidy, particularly trisomy 21.

Case

We report two cases identified during routine first-trimester screening at 12 weeks of gestation.

In both cases, ultrasound examination showed normal early fetal anatomy, present nasal bones and nuchal translucency within normal limits. However, Doppler evaluation of the tricuspid valve showed no regurgitation (**Figure 1**) but the Doppler of the ductus venosus demonstrated a reversed A-wave (**Figure 2**), raising suspicion of chromosomal abnormality despite otherwise reassuring findings.

Following genetic counseling, invasive diagnostic testing was performed and trisomy 21 was confirmed in both fetuses. After multidisciplinary discussion regarding prognosis and management options, the patients opted for termination of pregnancy.

Fetopathological examination confirmed phenotypic features consistent with trisomy 21.

Conclusion

These cases highlight the importance of systematic Doppler assessment of the ductus venosus during first-trimester screening. Even in the presence of normal nuchal translucency and apparently normal early anatomy, a reversed A-wave may represent an early marker of chromosomal abnormalities and should prompt further genetic investigation.



Figure 1. Doppler of the ductus venosus



Figure 2. Doppler of the tricuspid valve