

Fetal brain anomalies detection during the first trimester: expanding the scope of antenatal sonography

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Objective

The purpose of this study was to describe fetal brain anomalies identified during nuchal translucency (NT) examination and their clinical management.

Methods

In this retrospective study, we evaluated charts of pregnant women performed the first trimester scan (FTS) between 1.1.2011 and 31.12.14 in a tertiary referral center. Study population consisted of 952 patients scheduled for routine NT scan for aneuploidy screening between 11.0 and 13.6 gestational weeks, and 32 referred patients due to suspicious CNS finding during previous NT scan. Targeted brain assessment was performed according to clinical judgment during routine scans and in all referred cases. Patients with suspicious CNS findings were referred to pregnancy termination or further evaluation including genetic consultation and second trimester anatomical scan.

Results

Thirty-one fetuses were diagnosed with variable brain anomalies. Acrania/anencephaly was the most common defect (nine cases) while the rarest findings were inter-hemispheric cyst, microcephaly, and Walker-Warburg Syndrome (one case each). Genetic testing revealed variable results. Twenty-six (83.9%) couples decided to terminate their pregnancies.

Conclusion

The current report highlights the evolving ability to detect CNS malformation during NT scan. FTS novel findings expand our embryological understanding of early fetal development. While severe anomalies can be managed by early termination, other defects necessitate comprehensive evaluation and repetitive imaging.