Fetal Brain Anomalies Detection during the First Trimester: Expanding the Scope of Antenatal Sonography

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Objective
To describe fetal brain anomalies identified during nuchal translucency (NT) examination and the clinical management following these findings.

Methods
In this retrospective study we evaluated the charts of all pregnant women referred for FTS between 1.1.2011 and 31.12.14 in a single tertiary referral centre. Studypopulation consisted of 952 low-risk pregnant women scheduled for primary NT scan for aneuploidy screening between 11.0 and 13.6 gestational weeks, and 32 patients referred for secondary scan because of suspicious CNS finding during previous NT scan. Targeted brain assessment was performed according to clinical judgement among the primary scan cohort and in all secondary scans. Patients diagnosed with suspicious findings were referred either to pregnancy termination or further evaluation according to their findings including genetic consultation and second trimester anatomical scan.

Results
Thirty-one foetuses were diagnosed with variable brain anomalies. Acrania/anencephaly was the most common defect (9 cases) while the rarest findings were inter-hemispheric cyst, microcephaly, and Walker-Warburg Syndrome (1 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 the NT scan. FTS novel findings expand our embryological understanding of early foetal development. While severe anomalies can be managed by early termination, other defects necessitate comprehensive evaluation and repetitive imaging.