

First trimester ultrasound diagnosis of exencephaly

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Objective

The introduction of first trimester nuchal translucency measurement has resulted in earlier identification of fetuses with exencephaly. Exencephaly is an uncommon congenital malformation in which the flat bones of calvaria are absent leaving the brain completely exposed or protrudes through the defect in the cranial vault. Anencephaly may ensue as a result of prolonged exposure of the developing encephalon to amniotic fluid and trauma in utero.

Methods

i) The patient was a 32 year old Chinese female, gravida 2, para 0. Her first pregnancy ended in miscarriage. Medical history noted Type 1 diabetes mellitus. A viability scan at 9. 7 weeks of amenorrhoea demonstrated a viable embryo with no obvious abnormality; crown rump length (CRL) concurred with 8. 7 weeks. The First Trimester screening at 13. 7 weeks, CRL concurred with 12. 7 weeks. Fetal cranium was absent and brain appeared as an irregular mass. A diagnosis of exencephaly was made. Fetal anomaly scan at 14. 1 weeks confirmed the anomaly. (ii) Another patient was a 30 year-old Indian female, gravida 2 para 0. Medical history noted asthma and allergy to antibiotics. Dating scan at 12. 4 weeks showed CRL according with 10. 4 weeks. Cranium appeared absent. A repeat scan at 15. 3 weeks noted CRL to concur with 12. 6 weeks. Cranium was absent and fetal brain was noted as an irregular mass.

Results

Two and three dimensional ultrasound images of exencephaly.

Conclusion

Two and Three dimensional ultrasound demonstrated the absent cranium and exposed brain very well. As in anencephaly, facial structures and bony base of the calvarium were preserved.