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A case of microcephaly

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Objective

This is a case report of congenital microcephaly.

Methods

The patient was a 42 year old G2P1 of Indian origin. Medical complications included type 2 diabetes mellitus. First trimester screening was intermediate risk (NT 1.8mm, NB seen, T21 1: 329, T18 1: 1387, T13 1: 1996), cfDNA returned a low risk, fetal fraction 12.1%. Karyotype was 46XY, TORCH screen and Zika PCR were negative. Fetal anomaly scan at 21 weeks' noted head circumference on the 5th centile, decreasing to below the 5th centile at 26 weeks'. No other structural abnormalities were noted. The couple was counselled that the aetiology of the microcephaly was not apparent but fetal growth restriction could not be excluded, and that there is a risk of developmental delay associated with microcephaly. Toxoplasma IgG/IgM negative Rubella IgG + IgM negative CMV IgG + IgM negative. HSV IgM negative.

Results

A male infant with a birthweight of 2045g was delivered at 35^{+5} weeks.

Conclusion

The couple were referred to the geneticist for counselling.