

A case of ambiguous genitalia

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Objective

A case report to aid diagnosis of genital ambiguity and management options to exclude chromosomal anomaly, pseudohermaphroditism and genitourinary anomaly.

Methods

A case report of a 38 year old primigravida spontaneous conception after one year of infertility. No evidence of serological analytical importance, who was taking folic acid. She presented at 8 weeks with vaginal bleeding and diagnosed with a threatened miscarriage. She was prescribed Utrogestan and advised complete rest for three days. At 11 week she had vomiting that resolved spontaneously.

Results

Aneuploidy screening indicated a high T21 risk of 1: 110. Amnioscentesis showed a normal karyotype. Routine anomaly screening at 20 weeks showed a normal fetal morphology and female genitalia. At 24 weeks gestation a diagnosis of gestational diabetes were made, which was treated with diet and exercise. An ultrasound examination performed at 33 weeks revealed ambiguous genitalia. No other genitourinary abnormalities were observed.

Conclusion

The diagnosis of sex for ultrasound is not always easy. When faced with genital ambiguity, a through structural examination examination of the fetus should be conducted to rule out genitourinary anomalies. A karyotype should also be performed to exclude chromosomal anomalies, as in this case.

