



Normal size baby with abnormal dopplers. Think aneuploidy?

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Objective

To describe a case of fetus with Down's syndrome with low risk at combined screening test, presented in the third trimester with abnormal dopplers.

Methods

This is a case report.

Results

A 23 years old patient with previous obstetric history of caesarean section was referred to the Fetal Medicine Unit (FMU) due to abnormal third trimester ultrasound findings. She had previously received a low risk for Down's syndrome during first trimester screening (risk < 1: 1800). Furthermore, at the anomaly scan no obvious fetal abnormalities were identified and her pregnancy was overall uncomplicated. At 30 weeks she was referred to the obstetric ultrasound department for a fetal wellbeing scan as she was measuring large for dates. As this ultrasound revealed significant polyhydramnios, short femur and abnormal PI of the umbilical artery (UA) doppler, the patient was referred to FMU which confirmed the above. In view of the above findings an amniocentesis was offered to the patient, which confirmed trisomy 21.

Conclusion

Abnormal umbilical artery dopplers at third trimester does not necessarily mean placental dysfunction or intrauterine growth restriction. This warrants expert fetal review to exclude other markers of fetal aneuploidy. A few previous studies have also concluded that third trimester fetuses with trisomy 21 can often present with raised PI of the UA Doppler even in the absence of placental insufficiency.