

An unusual case of severe IUGR ? Genetic cause

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Objective

Intrauterine growth restriction (IUGR) refers to a condition in which a fetus is unable to achieve its genetically determined growth potential. There are multiple underlying causes for severe IUGR which can be categorised as maternal, fetal or placental/cord problems. Of all fetuses at or below the 10th centile, 40% have IUGR, 40% will be constitutionally small and the remaining 20% are intrinsically small secondary to a chromosomal or environmental problem.

Methods

A 38 year old Chinese, gravida 4, para 2 with 2 living children both born at term, booked early in pregnancy. She was fit and well, with a normal BMI and a non-smoker. Nuchal thickness was 1.6 with free Beta HCG levels were markedly elevated at 3.64 MOM and PAPP-A was 0.10 MOM. The combined screen result was positive with a risk of 1 in 2. A CVS was performed and revealed a normal karyotype. At the 14 week scan no anomaly was noted but AC and BPD were plotted on the 5th centile. The 20 week anomaly scan found all parameters for growth were well below the 3rd centile and placenta was noted to be very bulky and thick with cystic spaces. Suspicion of a partial molar pregnancy was raised in association with severe IUGR and reduced liquor. A poor prognosis with possible early onset of pre-eclampsia was discussed and parents wanted to continue with the pregnancy. Gestational diabetes was diagnosed at 28 weeks requiring insulin therapy. Pregnancy continued till 32 weeks with normal dopplers and although growth continued all measurements remained well below the 3rd centile with reduced liquor. The antenatal plan after the 32 week scan was to give steroids, monitor closely and possible in-utero transfer, aiming to achieve a gestation of 34 weeks.

Results

Results She had an emergency caesarean at 32+2; a live male infant was delivered weighing 803g. Appgars were 3, 9 and 9 and 1, 5 and 10 minutes respectively. The baby was intubated and initially fed with TPN. The infant is currently 8 weeks old weighs 1.8kg, is feeding normally and is soon to be discharged home. Parental chromosomal analysis has been advised. Despite the normal CVS, post-natal analysis of the infant's chromosomes has shown duplication of long arm of chromosome 14 (the duplication region is from bands q13.1 to q13.2) The Placental histology did not detect any anomaly, however unfortunately no clinical details were sent to pathologist to aid the assessment.

Conclusion

Conclusion: Severe IUGR is not always associated with poor prognosis as the above case demonstrates; a guarded prognosis should be discussed openly with parents. Initially the cause for severe IUGR was thought to be placental however it appears that this might be associated with genetic cause as anomaly in chromosome 14 was detected. Extra genetic material is likely to result in some problems. The specific problem depends on the chromosome segment that is duplicated, the genes it contains and in some cases the parental origin. There is currently limited knowledge regarding the impact of duplication of q13.1 – q13.2 and no known association with severe IUGR has been reported.

