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A case of absent ductus venosus with associated cardiomegaly and hepatomegaly

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Objective

Agenesis of the ductus venosus (aDV) with extra-hepatic umbilical venous drainage may lead to cardiomegaly and hydrops in the third trimester of pregnancy. Trisomy 21-related transient abnormal myelopoiesis (TAM) may present at late gestation with isolated, hypoechoic hepatomegaly due to extra-medullary haemopoiesis. Prenatal diagnosis of TAM is thought to be associated with worse prognosis.

Methods

We report on the perinatal clinical course of an infant diagnosed in the third trimester of pregnancy with cardiomegaly and hepatomegaly thought to be due to aDV with abnormal drainage of the umbilical vein (UV) into the inferior vena cava (IVC).

Results

The mother was a 42-year old gravida-3 para-2 with two previous healthy children born from a non-consanguineous marriage following uncomplicated pregnancies. In the index pregnancy she booked at 26 weeks' gestation and declined serum screening for Trisomy 21 (T21). An ultrasound examination carried out in Fetal Medicine Unit at 30 weeks' gestation due to incomplete anomaly survey revealed femur length below 5th centile, cardiomegaly, hepatomegaly, moderate bowel echogenicity and aDV with extra-hepatic umbilical vein drainage to dilated IVC. A specialist fetal cardiac scan confirmed the findings. In view of negative infection screening and absence of ultrasound features of fetal anaemia, both cardiomegaly and hepatomegaly were deemed to be secondary to aDV. The parents were counseled by a multi-disciplinary team including fetal medicine specialist, fetal cardiologist and neonatologist about the increased risk of underlying chromosomal defect and uncertainty of short and long term outcome due to the severity of the clinical picture. The couple declined karyotyping in view of their commitment to the pregnancy. Indication for late preterm induction of labour was made at 36 weeks and 6 days' gestation due to onset of severe pre-eclampsia and resulted in spontaneous vaginal delivery. A male infant was born weighing 2510g and admitted to Intensive Care Neonatal Unit for respiratory distress and incipient hydrops. Clinical and cytogenetic diagnosis of Trisomy 21 was made. Haematological investigations including blood film revealed TAM. Invasive cardiac monitoring showed severe pulmonary hypertension possible linked to T21-related pulmonary hypoplasia. TAM was successfully treated with 1 week of cytarabine. However, at 6 months of age the baby remains admitted with congenital anaemia and thrombocytopaenia, conjugated hyperbilirubinaemia and recurrent infections still being ongoing causes of significant morbidity.

Conclusion

Cardiomegaly and hydrops have been frequently described in fetuses with aDV with extra- hepatic umbilical venous drainage both in cases where the UV drains directly to the right atrium or the IVC. Hepatic changes including sinusoidal congestion and focal hyperplasia have also been reported in association with this condition. However no obvious hepatomegaly, albeit theoretically possible as a result of cardiac failure, has been reported to date in cases of isolated aDV, irrespective of the karyotype. Fetal hypoechoic hepatomegaly is significantly associated with TAM and trisomy 21. Differential diagnosis includes severe isoimmunization disorder, intrauterine infection, fetal congestive heart failure, and metabolic disorder. The prognosis of TAM developing during fetal period is sometimes poor with the size and progression of hepatomegaly being suggested prognostic markers. In our case, it is likely that cardiomegaly resulted from aDV with abnormal extra-hepatic drainage of umbilical vein. It is not clear whether this significantly contributed to the hepatomegaly,

which is in fact typical of prenatal TAM particularly in T21 fetuses. Our case suggests that in fetuses with confirmed or suspected T21, late-onset abnormalities such as cardiomegaly and hepatomegaly should be regarded as predictors of poor outcome, irrespective of the etiology. Prenatal diagnosis is paramount to counsel prospective parents regarding their options and to plan delivery in an appropriate setting and with availability of intensive care neonatal unit.