

Recurrent case of fetal cystic hygroma in a subsequent pregnancy: a case study

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Background

Fetal cystic hygroma (CH) is a developmental malformation of the lymphatic system resulting in fluid filled sacs, most commonly at the neck. It may also occur at the axilla, mediastinum or abdomen¹. The incidence in first trimester is 1:285 although some will resolve in-utero². About 50% of cases of CH are associated with chromosomal abnormalities, including Turner syndrome and Trisomies 13, 18 and 21². Recurrence in subsequent pregnancies is rare.

Case

A 27-year-old, G4P1 presented in her 4th pregnancy at K12 following a first trimester ultrasound showing an increased nuchal translucency (6.5mm), and diffuse subcutaneous oedema consistent with bilateral CH and hydrops.

She had further ultrasound evaluation and proceeded with a medical termination of pregnancy at K14 due to likelihood of poor fetal outcome. DNA extraction revealed a normal fetal karyotype. Her second pregnancy had a similar outcome, with the diagnosis of bilateral CH on first trimester screen. She underwent amniocentesis which showed a 46XY karyotype. Following progression of the CH, she was induced at 22 weeks gestation and delivered a stillborn fetus. The patient's first pregnancy was uncomplicated and resulted in the term delivery of a live female infant. She suffered a first trimester miscarriage in her third pregnancy. Both the patient and her partner had no major medical problems and were non-consanguineous. She was rhesus negative and had had appropriate Anti-D throughout her pregnancies.

References

Discussion

The prognosis of CH is variable but there are poorer outcomes among fetuses with large, multi-loculated or septated hygromas, with a chromosomal abnormality or with other major structural abnormalities³. To have subsequent pregnancies affected by CH is rare. In those with a normal karyotype, it can be inherited as an autosomal recessive trait⁴. In this case, a 427kb deletion at 17p12 was present in the mother and first affected fetus, however not in the second affected fetus. The significance of this deletion is unknown. Genetic counselling is recommended for parents who have had pregnancies affected by CH.

Fig 1: Fetal septated cystic hygroma³



1. Malone FD, First-trimester septated cystic hygroma: prevalence, natural history, and pediatric outcome. *Obstet Gynecol* 2005 2. Scholl J, First-Trimester Cystic Hygroma: Relationship of Nuchal Translucency Thickness and Outcomes. *Obstet Gynaecol* 2012 3. Shanal, C. Prenatal management, pregnancy and pediatric outcomes in fetuses with septated cystic hygroma. *Braz J Med Biol Res* 2014 4. Baxi L, Recurrent cystic hygroma with hydrops. *Fetal Diagn Ther* 2009