A Case of Recurrent Pregnancy and Child Loss in an Irish Traveller

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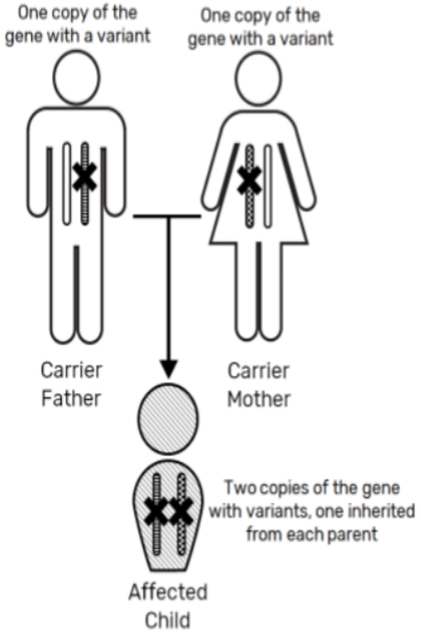
Objective: This case illustrates the challenges faced whilst providing care in a high risk pregnancy, with a structural abnormality, in the absence of a genetic diagnosis.

Case Report: A 36 year old lady (G10 P4+5), from the Irish Traveller Community, in a consanguineous marriage with her first cousin, was seen in the high risk clinic at 15+6 weeks gestation. The patient had 5 first trimester miscarriages, one child who passed away at the age of five (global developmental delay and respiratory disease) and one neonatal death at 22 hours of life (severe IUGR-900g at birth and a renal anomaly). Two of her children are alive and healthy. In their previous pregnancies, this couple declined invasive prenatal tests, post mortem examinations and genetic tests offered for their live children. In this pregnancy, foetal ventriculomegaly was noted, foetal echocardiogram revealed hypoplasia of the aortic valve and EFW consistently measured below the 5th centile. At 35 weeks gestation an emergency CS was carried out for persistently reduced foetal movements and increased resistance on dopplers. At delivery, the female infant was noted to be small with dysmorphic features (low set ears, overriding fingers, a thick nuchal fold, increased tone and wide set nipples). DNA microarray revealed a diagnosis of Beaulieu- Boycott Innes syndrome which is a rare autosomal recessive (AR) neurodevelopmental disorder linked to the THOC6 gene. The infant subsequently deteriorated and comfort care was offered.

Discussion: Poor obstetric outcomes are more common in Irish travellers compared to the general population. A number of factors play a role in this, including a higher incidence of rare genetic disorders (often AR). Advances in modern medicine, especially with DNA microarray technology, has improved outcomes, as a management plan can be triggered once a diagnosis is made. IUGR was a common finding in previous pregnancies and these children may have had the same disease with different phenotypes. If AR conditions amongst this population are suspected, rapid diagnosis should be facilitated and the patient should have access to a geneticist with experience with their particular population. An alternative approach is to offer diagnosis based on foetal phenotype or parental carrier testing.



Conclusion: This case demonstrates that a number of rare genetic disorders present with congenital anomalies and may be detected with foetal ultrasound and therefore specialised follow up, although challenging without a diagnosis, in a foetal medicine unit may be of benefit.