Prenatal sonographic diagnosis of Beckwith-Wiedemann syndrome in a fetus with omphalocele

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DESCRIPTION

A 28 years old primigravida was referred to Department of Obstetrics and Gynaecology, King George's Medical University, as 35 weeks pregnancy with polyhydramnios with suspected fetal omphalocele. The defect was diagnosed at 24 weeks at some external centre and the patient was counselled regarding continuation of pregnancy with postnatal surgical repair of malformation after delivery. The diagnosis of Beckwith-Wiedemann syndrome (BWS) was missed at that time.

Phenotypic presentation of BWS has several variations and low occurrence of multiple abnormalities simultaneously limits the sonographic detection rate. The genotypic detection is feasible only if the molecular defect is known.

We are submitting images of this fetus which shows a fetus with large abdominal wall defect covered by a wall containing only bowel loops and not the liver (*figure 1*). Along with it the fetus also shows bilateral enlarged kidneys with a suspicious cystic lesion on upper pole of right kidney (*figure 2*) and macroglossia (*figure 3*). Fetal parameters were more than 90th centile and liquor was increased (amniotic fluid index 20 cm). This classically fitted in the diagnosis of BWS¹ rather than being an isolated omphalocele.

Prenatal diagnosis of BWS helps in postnatal care of the baby who has risks of neonatal hypoglycaemia, accelerated bone maturation and embryonal tumours which may arise in 7% of cases.² BWS is sporadic but in 15% of patients it could be familial with autosomal dominant mode of inheritance and this needs to be explained to the couple so as to have a high index of suspicion for early diagnosis in subsequent pregnancy.

Figure 2 Enlarged kidney with hetroechoic lesion.

Figure 3 Enlarged tongue protruding out.

Figure 1 Anterior abdominal wall defect with sac containing loops of intestines.

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Learning points

- ▶ Beckwith-Wiedemann syndrome (BWS) is a significantly more morbid condition compared to an innocent isolated omphalocele.
- ▶ Any fetus presenting with omphalocele must be screened for BWS which makes significant difference in postnatal management.
- ▶ Early diagnosis can be made keeping a high index of suspicion.

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Contributors NK performed the sonography. SA carried out the literature search. VD and AP helped in preparing the final report.

Competing interests None declared.

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