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4D scanning: a diagnostic tool of choice for detection of fetal limb reduction defects

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DESCRIPTION

A 32-year-old primigravida, with low background risk at booking, referred to our fetal medicine department after a missing left lower limb in the fetus was diagnosed on scan at 26 weeks gestation. Hypoplastic left thigh and limb reduction defect were confirmed (figures 1 and 2) using 4D scan, and parents counselled. They remained committed to the pregnancy. Subsequent scans showed good growth and polyhydramnios. She underwent an emergency caesarean section at 41 weeks for fetal distress, and baby weighed 4175 g. On examination, baby had absent left lower limb and ectopic left hemiscrotum. Skeletal survey showed aplastic left pelvic bone, absent tibia-fibula and hypoplastic femur. Karyotype was normal.

The finding of a limb reduction defect in an otherwise healthy baby can be very disappointing for the expectant parents. The upper limb is twice as commonly affected by reduction defects than the lower limb.¹ In our case, the baby had a longitudinal defect (absence/severe hypoplasia of the long bones). Most upper limb defects are isolated events but those involving lower limbs are often part of more complex anomalies or syndromes.² Possible causes include chromosomal anomalies, drugs (thalidomide, a classic example) and amniotic bands. Various anomalies can be caused by amniotic bands ranging from minor constrictions of digits to cleft lip and palate. The significance of an association between maternal factors and limb reduction defects is difficult to assess in retrospective studies.³



Figure 1 4D image of the limb reduction defect, in utero.



Figure 2 Postnatal image showing absent lower limb and hypoplastic femur.

Prospective studies would then be required to confirm any hypothesis regarding causation.

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Competing interests None.

Patient consent Obtained.

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