

## Research Letter

# First-trimester two-dimensional and three-dimensional ultrasound demonstration of craniofacial defects, abdominal wall defects and upper limb deficiency associated with limb–body wall complex

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A 35-year-old, primigravid woman was referred to the hospital at 13 weeks of gestation to evaluate fetal structural abnormalities. The father was aged 32 years. The mother reported no illness or recent infections. She had neither history of prenatal exposure to teratogenic agents nor any family history of congenital malformations. She had not undergone assisted reproductive technology for this pregnancy. Prenatal ultrasound at 13 and 14 weeks of gestation demonstrated a live fetus with attachment between the fetal head and the placenta, skull deformation, hydrocephalus, nasal hypoplasia, scoliosis, extracorporeal stomach, liver and intestines, deficiency of the left upper limb, and hypoplasia of the right upper extremities (Figs. 1 and 2). A diagnosis of limb–body wall complex (LBWC) with craniofacial defects was made. The pregnancy was terminated at 14 weeks of gestation, and a 50-g female fetus was delivered with an irregularly shaped skull, facial deformation, adhesive bands over the skull, nasal hypoplasia, scoliosis, absence of the left upper and lower arms, hypoplasia of the right lower arm, and eviscerated stomach, liver and intestines, but normal female external genitalia, anus, lower extremities and umbilical cord (Fig. 3). Cytogenetic analysis of the fetus revealed a karyotype of 46,XX.

LBWC occurs approximately in 1:7000 ~ 1:42,000 births and is characterized by lateral body-wall defects, limb

reduction abnormalities, and/or craniofacial defects [1–6]. The present case is associated with hydrocephalus, skull deformation, facial deformation, amniotic adhesive bands, abdominoschisis, absence of the left upper limb and hypoplasia of the right lower arm, and belongs to the category of LBWC with craniofacial defects. Russo et al [7] hypothesized that LBWC with craniofacial defects is caused by early vascular disruption, whereas Hunter et al [8] proposed that LBWC with craniofacial defects is a primary defect of the ectoderm of the embryonic disc, and some of the malformations are secondary complications of the primary disturbance in embryogenesis. Most cases of LBWC have been diagnosed in the second trimester [4,8]. Prenatal diagnosis of LBWC with craniofacial defects is uncommon. In a review of 12 cases of LBWC, Chen et al [4] found that three-quarters of the cases were LBWC without craniofacial defects, and only one-quarter of the cases were LBWC with craniofacial defects. With the advent of two-dimensional and three-dimensional prenatal ultrasonography, the present case provides evidence that LBWC with craniofacial defects can be correctly diagnosed in the first trimester. We suggest that first-trimester ultrasound diagnosis of craniofacial abnormalities, abdominal wall defects, limb deficiency and scoliosis should include a differential diagnosis of LBWC with craniofacial defects.

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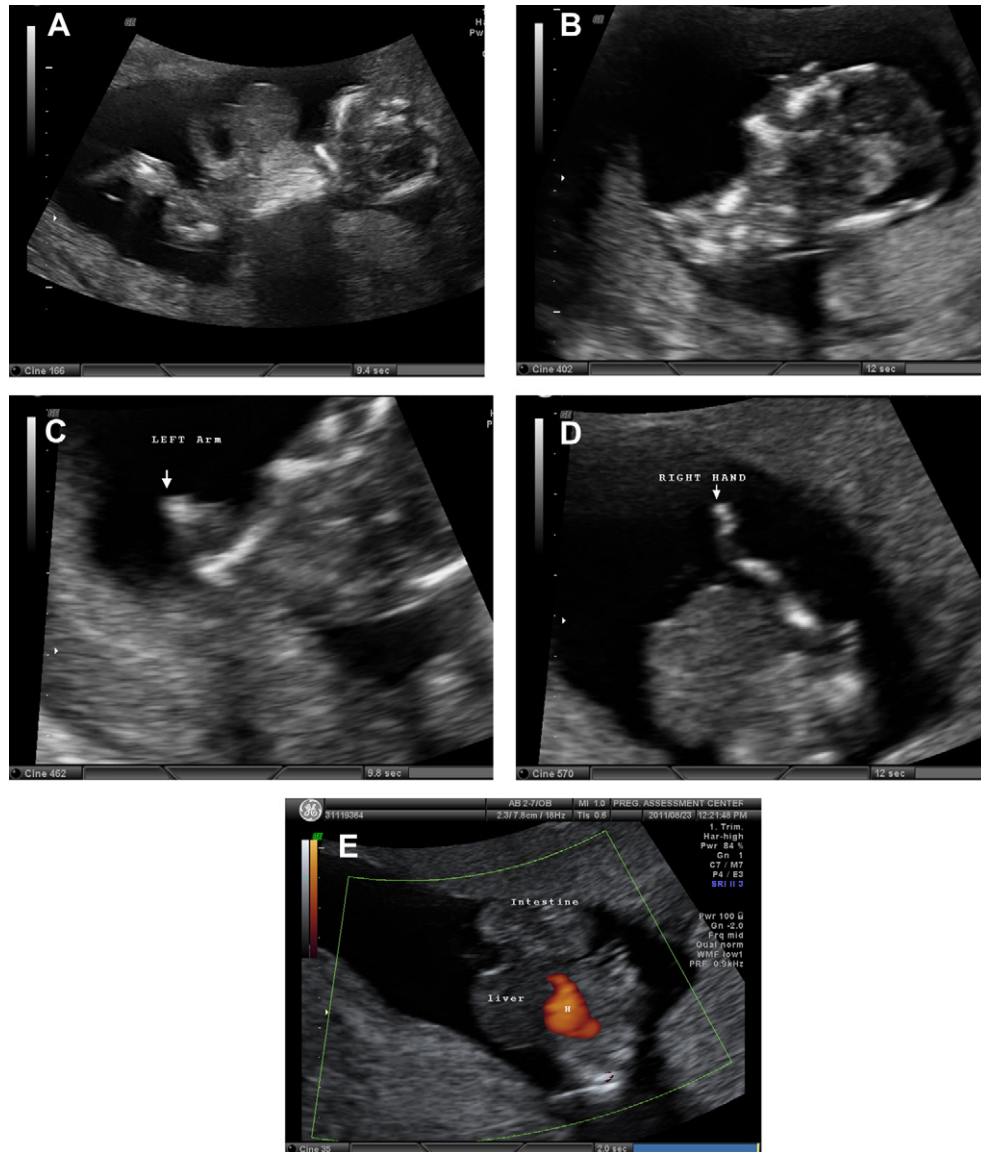


Fig. 1. Prenatal ultrasound at 13 weeks of gestation shows (A) a deformed skull with cranioplacental attachment, eviscerated solid and hollow internal organs, and upper limb deficiency, (B) nasal deformation, (C) left arm deficiency, (D) right arm hypoplasia with absence of the hand, and (E) extracorporeal liver and intestines. H = heart.

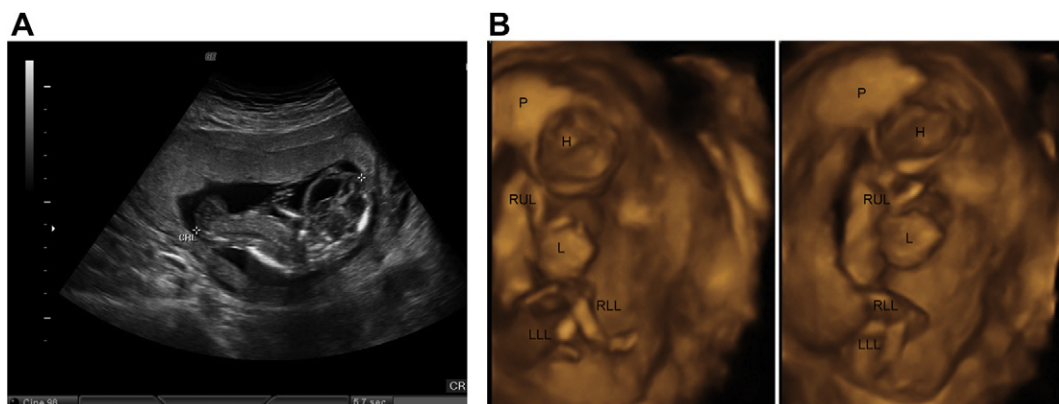


Fig. 2. Prenatal ultrasound at 14 weeks of gestation shows (A) scoliosis, hydrocephalus and hypoplastic right arm on two-dimensional ultrasound and (B) cranioplacental attachment, eviscerated liver, absence of the right hand and deficiency of the left arm on three-dimensional ultrasound. H = head; L = liver; LLL = left lower limb; P = placenta; RLL = right lower limb; RUL = right upper limb.

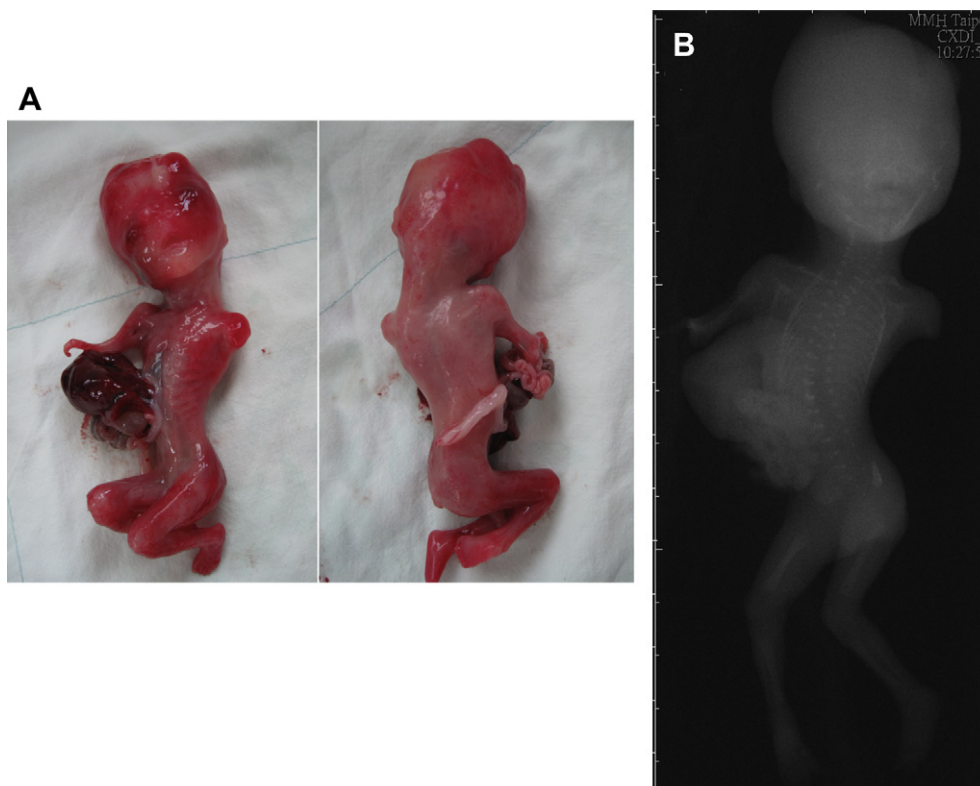


Fig. 3. (A) Anterior view and posterior view of the fetus at birth at 14 weeks of gestation. (B) Corresponding X-ray image of the fetus.

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