

Research Letter

Prenatal ultrasound demonstration of limb–body wall complex with megacystis

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A 35-year-old, primigravid woman was referred to the hospital at 14 weeks of gestation to evaluate fetal structural abnormalities. The father was aged 39 years. The mother reported no illness or recent infections. She neither had

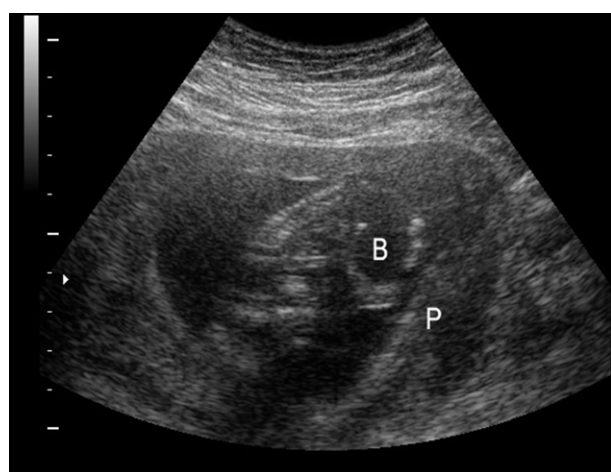


Fig. 1. Prenatal ultrasound at 14 weeks of gestation showing abdominoplacental attachment and megacystis but no scoliosis. B = bladder; P = placenta.

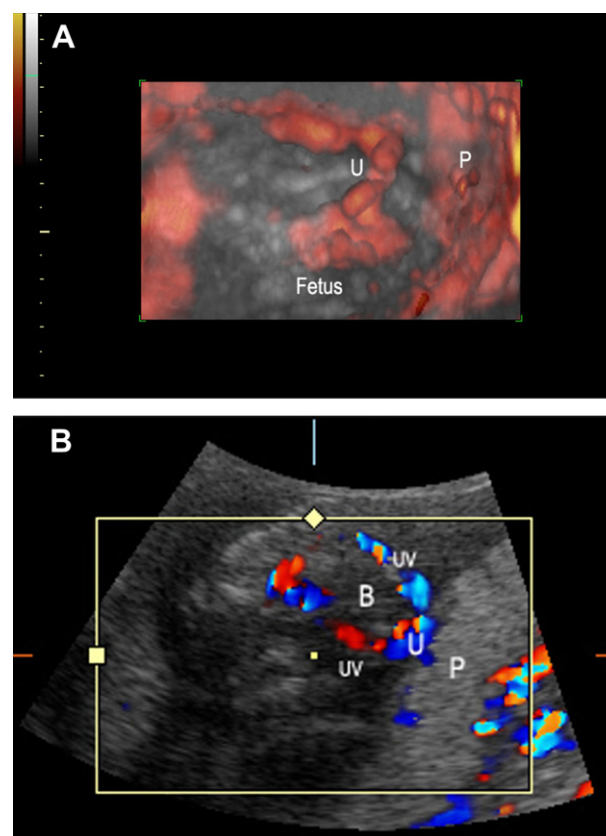


Fig. 2. Color Doppler angiography showing (A) a short umbilical cord and (B) umbilical vessels surrounding the enlarged urinary bladder. B = bladder; P = placenta; U = umbilical cord; UV = umbilical vessels.

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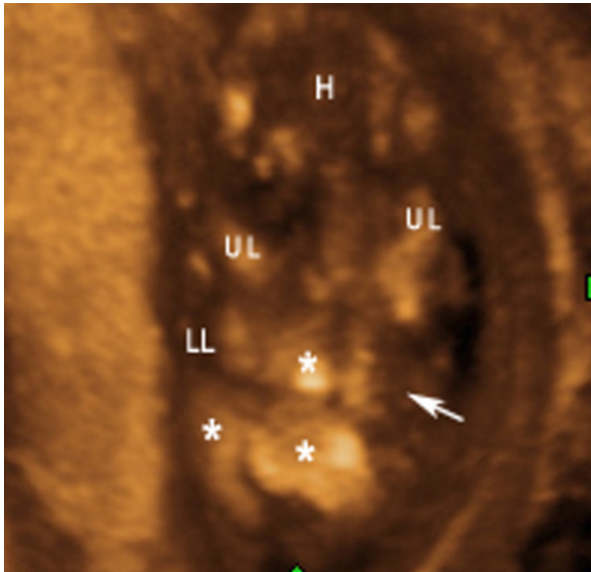


Fig. 3. Three-dimensional ultrasound showing the absence of the left lower limb (arrow). LL = lower limb; UL = upper limb. *Eviscerated internal organs.

a history of prenatal exposure to teratogenic agents nor any family history of congenital malformations. She did not undergo any assisted reproductive technology for this pregnancy. Prenatal ultrasound at 14 weeks of gestation demonstrated a live fetus with abdominoplacental attachment; abdominal wall defects; eviscerated liver, bowel, and giant urinary bladder, but no scoliosis (Fig. 1). Color Doppler angiography showed a short umbilical cord with umbilical vessels surrounding the enlarged urinary bladder (Fig. 2). Three-dimensional (3D) ultrasound showed the absence of left lower limb (Fig. 3). The pregnancy was subsequently terminated, and a 50-g fetus was delivered with a normal head, abdominoschisis, anal atresia, amelia of left lower limb, hyperextension of right lower limb, and a very short umbilical cord (Fig. 4). A diagnosis of limb–body wall complex (LBWC) was made. Cytogenetic analysis of the fetus revealed a karyotype of 46,XX.

LBWC occurs in approximately 1:7,000 to 1:42,000 births [1–3]. LBWC is characterized by lateral body wall defects, limb reduction anomalies, and/or craniofacial defects [4–6]. The present case of LBWC was not associated with craniofacial defects. Early amnion rupture [7], vascular disruption [1,2], and early embryonic maldevelopment [8,9] have been implicated as the pathogenesis of LBWC. Russo et al [10] suggested that LBWC without craniofacial defects is the result of a defective lateral and caudal folding process of the embryonic disk, and LBWC with craniofacial defects is caused by an event of early vascular disruption. In the present case, 2D ultrasound revealed abdominoschisis and extracorporeal megacystis; 3D ultrasound demonstrated a deficiency of the left lower limb; and color Doppler angiography provided a perfusion-like display of the vasculature of the flow in the umbilical vessels. The present case was associated with extracorporeal urinary bladder, intestines, and liver; megacystis; and a short umbilical cord. Chen et al [11] previously

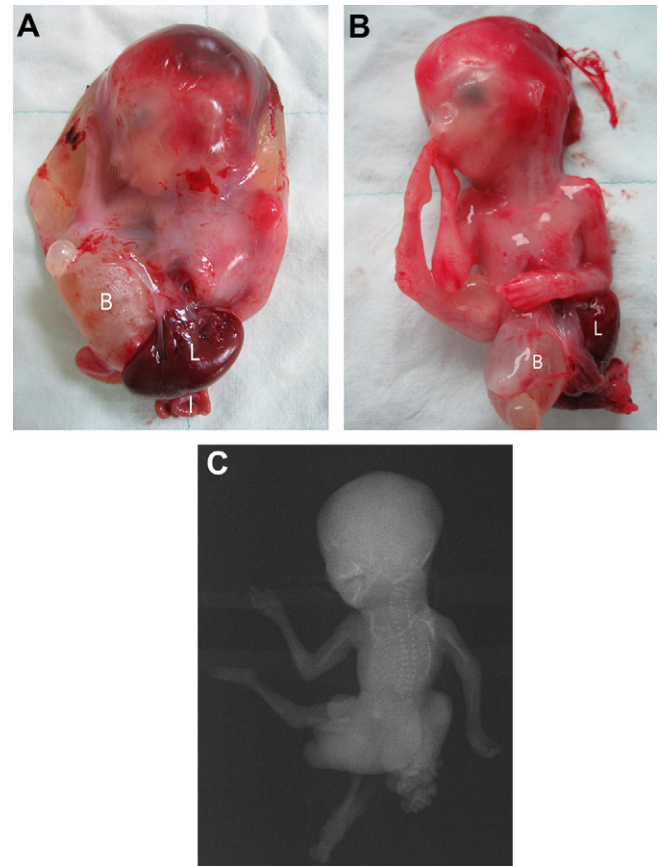


Fig. 4. (A) A fetus with limb–body wall complex wrapped up in the amnion with extracorporeal internal organs. (B) Extracorporeal giant urinary bladder, intestines, and liver; a deficiency of left lower limb; and hyperextension of right lower limb. (C) X-ray of the fetus. B = bladder; I = intestines; L = liver.

described a second-trimester fetus with LBWC; right lower-limb deficiency; umbilical cord dysgenesis; extracorporeal enlarged urinary bladder, intestines, and liver; ambiguous external genitalia; and anal atresia. The present case additionally shows that LBWC can be associated with extracorporeal enlarged urinary bladder on prenatal ultrasound.

Acknowledgments

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