LETTER TO THE EDITOR

Prenatal Diagnosis of Limb–Body Wall Complex With Craniofacial Defects

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A 42-year-old, gravida 4, para 0 woman was referred to the hospital at 17 weeks of gestation to evaluate fetal structural abnormalities. The father was aged 42 years. The mother reported no illness or recent infections. She had neither a history of prenatal exposure to teratogenic agents nor any family history of congenital malformations. She had not undergone any assisted reproductive technology for this pregnancy.

Prenatal ultrasound at 17 weeks of gestation demonstrated a live fetus with cranioplacental attachment, scoliosis, and abdominal wall defects but no limb deficiency (Fig. 1). The pregnancy was subsequently terminated, and a 180-g male fetus was delivered with exencephaly, acranium, abdominoschisis, craniofacial deformity, partial deficiency of the second and third fingers of the left hand, left club foot, and extracorporeal intestines and liver, but normal male external genitalia and anus (Fig. 2). A diagnosis of limb–body wall complex (LBWC) with craniofacial defects was made. Cytogenetic analysis of the fetus revealed a karyotype of 46,XY.

LBWC occurs in approximately 1:7000 to 1:42,000 births [1–3]. LBWC is characterized by lateral body-wall defects, limb reduction abnormalities, and/or craniofacial defects [4–12]. The present case is associated with exencephaly, abdominoschisis, club foot, and deficiency of the digits, and belongs to the category of LBWC with craniofacial defects. It has been suggested that LBWC with craniofacial defects is caused by early vascular disruption [13]. Recently, Hunter et al [14] hypothesized that a primary defect/deficiency of the ectoderm of the embryonic disc may explain the key malformations seen in LBWC with craniofacial defects. Prenatal ultrasound diagnosis of concomitant neural tube defects and abdominal wall defects in association with cranioplacental attachment and scoliosis should include a differential diagnosis of LBWC with craniofacial defects.

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Fig. 1 (A) A deformed skull with acranium and attachment of the brain to the placenta. (B) Scoliosis with a curved spine and craniofacial deformation with acranium, a small skull, and attachment of the brain to the placenta. (C) Extracorporeal intestines and a deformed spine. (D) Corresponding three-dimensional ultrasound shows a curved spine, extracorporeal liver and intestines, and attachment of the brain to the placenta. b = brain, i = intestines, l = liver, p = placenta, s = skull, sp = spine.

Fig. 2 (A) Postnatal illustration of a fetus and the attached placenta associated with limb–body wall complex (LBWC) with craniofacial defects. (B) Corresponding X-ray illustration of LBWC with craniofacial defects. (C) Craniofacial abnormalities of acranium, exencephaly, and orbital and nasal deformations. (D) Extracorporeal liver and intestines. (E) Partial deficiency of the second and third fingers of the left hand. (F) Left club foot.
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