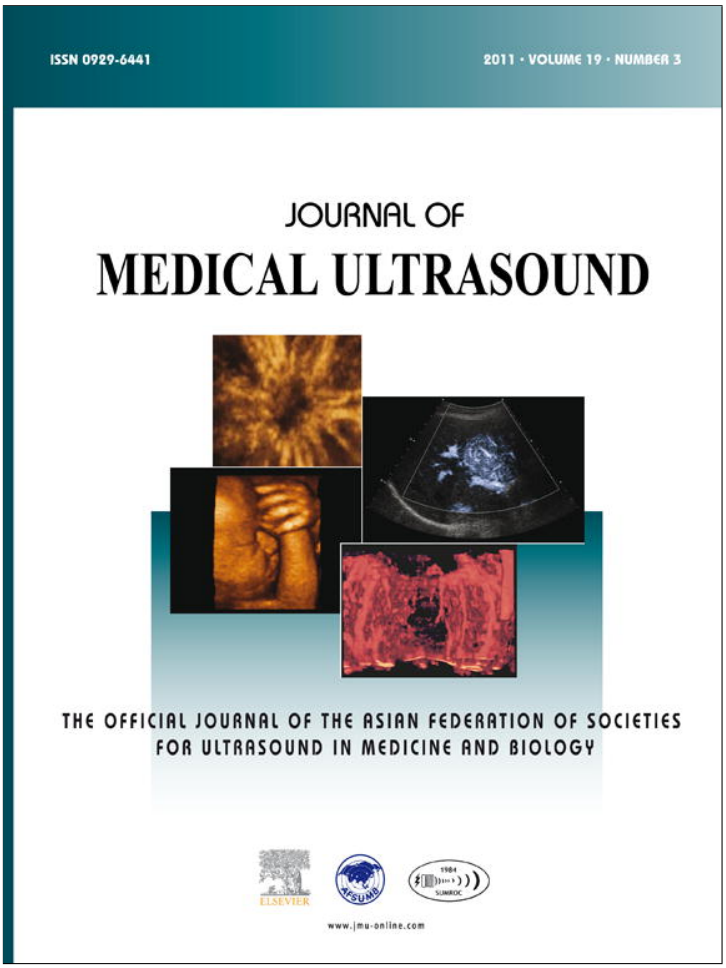

Journal of Medical Ultrasound (2011) 19, 112—114

Available online at www.sciencedirect.com

SciVerse ScienceDirect

journal homepage: www.jmu-online.com

LETTER TO THE EDITOR

Prenatal Diagnosis of Limb—Body Wall Complex With Craniofacial Defects

Chih-Ping Chen [1,2,3,4,5,6*], Yi-Yung Chen [1], Jun-Wei Su [1,7], Wayseen Wang [2,8]

[1] *Department of Obstetrics and Gynecology, Mackay Memorial Hospital, Taipei,* [2] *Department of Medical Research, Mackay Memorial Hospital, Taipei,* [3] *Department of Biotechnology, Asia University, Taichung,* [4] *School of Chinese Medicine, College of Chinese Medicine, China Medical University, Taichung,* [5] *Institute of Clinical and Community Health Nursing, National Yang-Ming University, Taipei,* [6] *Department of Obstetrics and Gynecology, School of Medicine, National Yang-Ming University, Taipei,* [7] *Department of Obstetrics and Gynecology, China Medical University Hospital, Taichung, and* [8] *Department of Bioengineering, Tatung University, Taipei, Taiwan, ROC*

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.

* Correspondence to: Dr Chih-Ping Chen, Department of Obstetrics and Gynecology, Mackay Memorial Hospital, 92, Section 2, Chung-Shan North Road, Taipei, Taiwan, ROC.

E-mail address: cpc_mmh@yahoo.com (C.-P. Chen).

doi:10.1016/j.jmu.2011.09.001

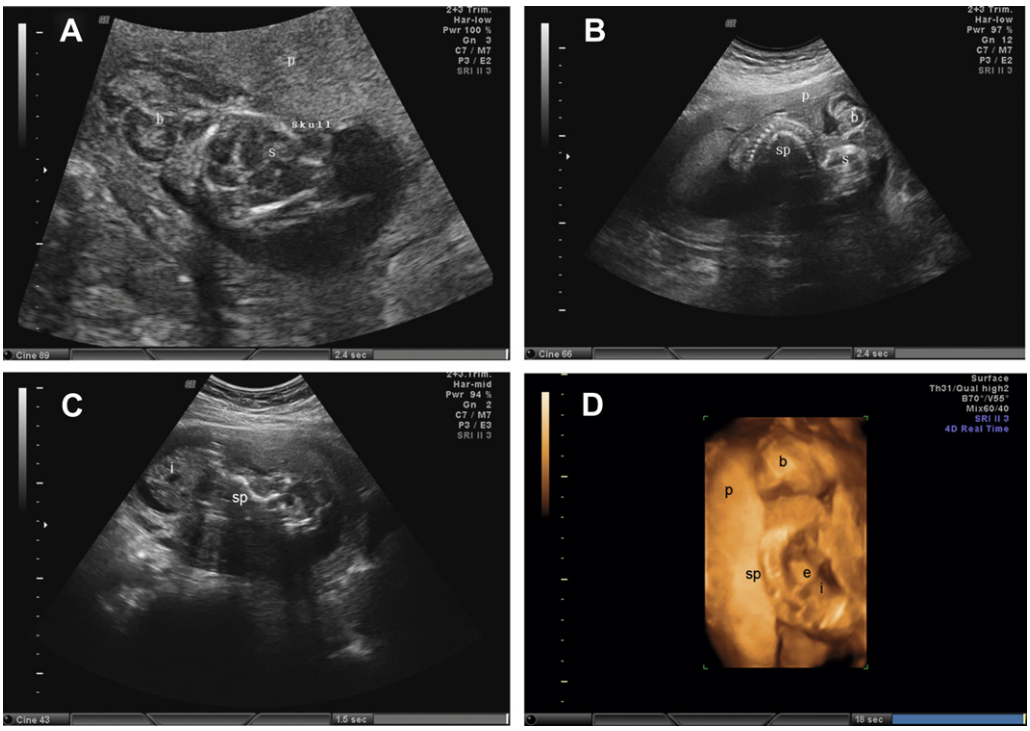

Fig. 1 (A) A deformed skull with acranium and attachment of the brain to the placenta. (B) Scoliosis with a curved spine and craniofacial deformation with acranium, a small skull, and attachment of the brain to the placenta. (C) Extracorporeal intestines and a deformed spine. (D) Corresponding three-dimensional 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.

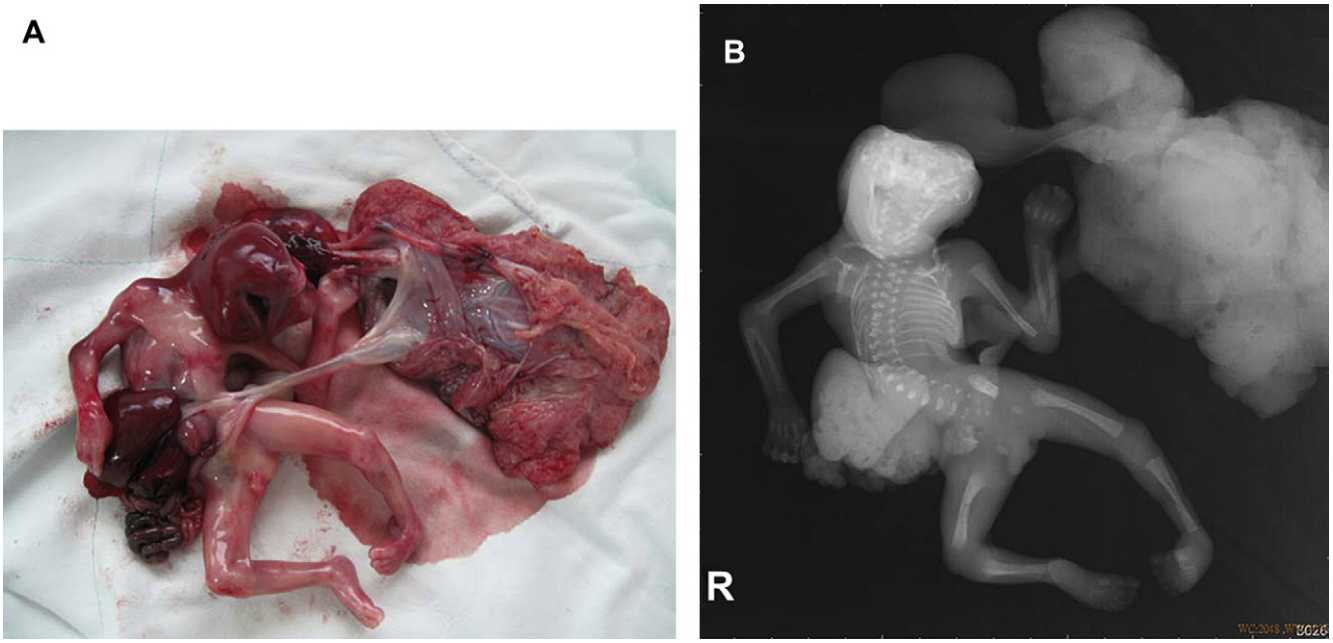

Fig. 2 (A) Postnatal illustration of a fetus and the attached placenta associated with limb—body wall complex (LBWC) with craniofacial defects. (B) Corresponding X-ray illustration of LBWC with craniofacial defects. (C) Craniofacial abnormalities of acranium, exencephaly, and orbital and nasal deformations.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

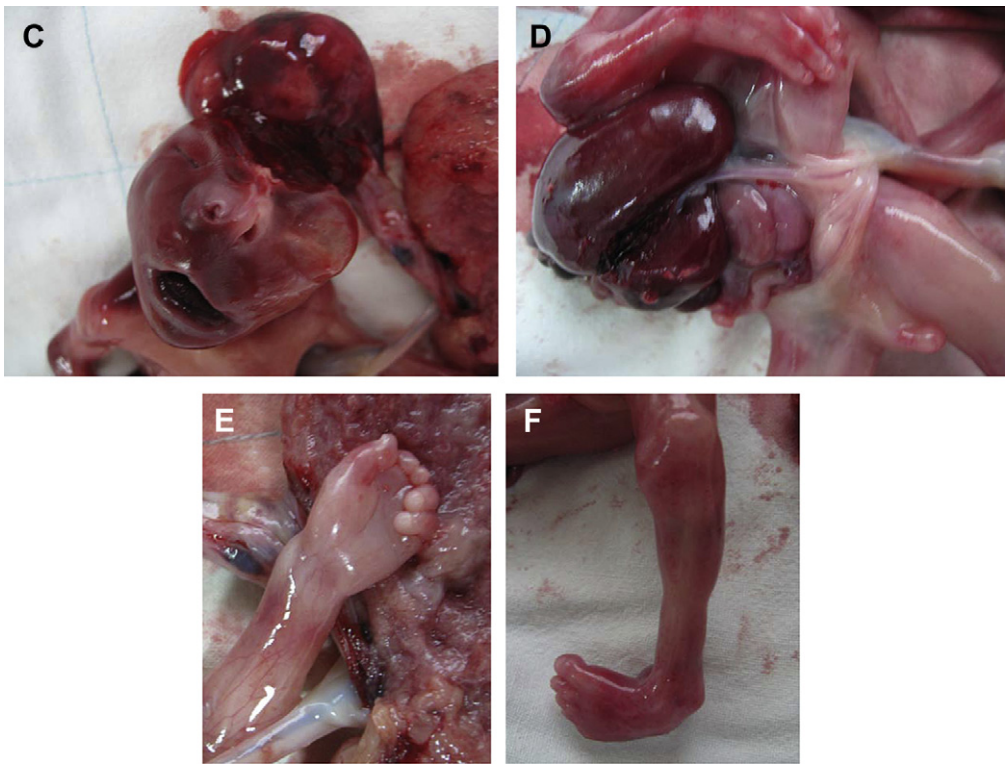

Fig. 2 (*continued*)

Acknowledgments

This work was supported by research grants NSC-97-2314-B-195-006-MY3 and NSC-99-2628-B-195-001-MY3 from the National Science Council (gs1), and MMH-E-100-04 from Mackay Memorial Hospital (gs2), Taipei, Taiwan.

References

[1] Van Allen MI, Curry C, Walden CE, et al. Limb-body wall complex: I. Pathogenesis. Am J Med Genet 1987;28:529—48.
[2] Van Allen MI, Curry C, Walden CE, et al. Limb-body wall complex: II. Limb and spine defects. Am J Med Genet 1987;28:549—65.
[3] Kurosawa K, Imaizumi K, Masuno M, et al. Epidemiology of limb-body wall complex in Japan. Am J Med Genet 1994;51:143—6.
[4] Chen C-P, Shih J-C, Chan Y- J. Prenatal diagnosis of limb-body wall complex using two-dimensional and three-dimensional ultrasound. Prenat Diagn 2000;20:1020 [Letter].
[5] Chen C-P, Tzen C-Y, Chang T-Y, et al. Prenatal diagnosis of acrania associated with facial defects, amniotic bands and limb-body wall complex. Ultrasound Obstet Gynecol 2002;20:94—5.
[6] Chen C-P, Cheng S-J, Lin Y-H, et al. Prenatal imaging of limb-body wall complex by magnetic resonance imaging. Prenat Diagn 2005;25:521—3.
[7] Chen C-P, Lin C-J, Chang T-Y, et al. Second-trimester diagnosis of limb-body wall complex with literature review of pathogenesis. Genet Counsel 2007;18:105—12.
[8] Chen C-P, Tsai F-J, Chen C-Y, et al. Limb-body wall complex with craniofacial defects after ovarian stimulation. Taiwan J Obstet Gynecol 2008;47:474—5.
[9] Chen C-P, Lee M-S, Tsai F-J, et al. Limb-body wall complex in one fetus of a dizygotic twin pregnancy obtained by egg donation, *in vitro* fertilization and embryo transfer: prenatal diagnosis and literature review. Taiwan J Obstet Gynecol 2009;48:446—50.
[10] Chen C-P, Lee M-S, Tsai F-J, et al. Prenatal ultrasound demonstration of limb-body wall complex with megacystis. Taiwan J Obstet Gynecol 2011;50:258—60.
[11] Chen C- P. Prenatal diagnosis of limb-body wall complex with craniofacial defects, amniotic bands, adhesions and upper limb deficiency. Prenat Diagn 2001;21:418—9.
[12] Chen C- P. Prenatal sonographic diagnosis of limb-body wall complex with craniofacial defects. Ultrasound Obstet Gynecol 2003;22:101.
[13] Russo R, D'armiento M, Angrisani P, et al. Limb body wall complex: a critical review and a nosological proposal. Am J Med Genet 1993;47:893—900.
[14] Hunter AGW, Seaver LH, Stevenson RE. Limb-body wall defect: is there a defensible hypothesis and can it explain all the associated anomalies? Am J Med Genet 2011;155A: 2045—59.