Available online at www.sciencedirect.com

SciVerse ScienceDirect

Taiwanese Journal of Obstetrics & Gynecology 52 (2013) 131–132

www.tjog-online.com

Research Letter

Prenatal diagnosis of pentalogy of Cantrell using three-dimensional ultrasound

Ting-Yu Yang, Pei-Yin Tsai, Yueh-Chin Cheng, Fong-Ming Chang, Chiung-Hsin Chang*

Department of Obstetrics and Gynecology, National Cheng Kung University Medical College and Hospital, Tainan, Taiwan

Accepted 18 May 2012

In 1958, Cantrell et al [1] described a syndrome called the pentalogy of Cantrell (POC), consisting of deficiency of the anterior diaphragm, midline supraumbilical abdominal wall defect, defect in the diaphragmatic pericardium, congenital intracardiac abnormalities, and defect of the lower sternum.

Although prenatal detection of POC has been reported previously, to date no prenatal illustration of POC by three-dimensional (3D) ultrasound (US) has been described [2]. The application of 3D US in malformations, especially in the evaluation of small parts, appears to assist in prenatal diagnosis and to be clinically useful. The multiplanar planes proved most helpful in delineating the exact nature and anatomic level of the defect [3].

Here, we reported two cases of POC with different presentation diagnosed using two dimensional (2D) and three dimensional (3D) ultrasound (US).

Case 1: A 19-year-old woman, gravida 1, para 0, was transferred to our hospital because of fetal congenital anomaly at 15 weeks of gestation. US revealed a crown–rump length of 87.3 mm and the fetal heart outside the thorax by Doppler US. Omphalocele and marked spinal curvature were also observed (Fig. 1). After discussion with the patient and family, they decided on termination of the pregnancy. The thoracoabdominal ectopia cordis, left cleft sternum and abdominal wall defect, omphalocele, scoliosis, umbilical cord cyst, and polydactyly of the left hand were grossly observed in the arbutus (Fig. 2). The histopathologic examination at autopsy demonstrated the previously mentioned findings and left diaphragm deficiency with internal organs exposure. Microscopically, the internal organs were well developed. Moreover, the abortus revealed normal female karyotype.

Case 2: A 32-year-old woman, gravida 1, para 0, was also transferred to our hospital for fetal abdominal wall defect at 13 weeks of gestation. US demonstrated the fetal crown–rump length to be 70 mm with associated defects including cystic hygroma, scoliosis, and omphalocele. Moreover, Doppler US showed ectopia cordis. We performed 3D US surface-rendering mode reconstruction for better comprehension. Due to poor prognosis, the patient and family decided to terminate the pregnancy. After termination, the gross appearance was consistent with the previously mentioned findings and there was normal male karyotype.

POC is a rare congenital anomaly. The associated defect of omphalocele and ectopia cordis is the major hallmark of this syndrome. The survival of the fetus is extraordinary and the prognosis depends on the size of the abdominal wall defect, extent of the cardiac defect and presence of associated anomalies. It is possible to make a definitive diagnosis at an early stage of gestation by US. The diagnosis of ectopia cordis relies on demonstration of a displaced heart and it may be partially or completely outside the thorax. Four types of POC by the cardiac location were proposed: thoracic (60%), abdominal (30%), thoracoabdominal (7%), and cervical (3%). The incomplete expression of the syndrome is well recognized, and full POC is a rare occurrence. Notably, ectopia cordis could be part of other conditions, such as POC, limb-body wall complex, or amniotic band syndrome [4].

The hypothesis underlying this condition is developmental failure of a segment of the lateral mesoderm between 14 and 18 days after conception, resulting in failure of ventral wall closure and incomplete external primordial bands fusion. It will become isolated ectopia cordis, POC, or one of its variants. However, most cases are sporadic. A few cases associated with trisomy 18 and X-linked inheritance have also been described previously [5,6]. The associated anomaly with POC, including multicystic dysplastic kidneys, limb defects, cystic hygroma, exencephaly, craniorachischisis, and intact diaphragm have been variously described [7,8]. Prenatal diagnosis is extremely important because the prognosis is poor and the disease is lethal. The diagnosis is usually made at the

* Corresponding author. Department of Obstetrics and Gynecology, National Cheng Kung University Medical College and Hospital, 138 Victory Road, Tainan 70428, Taiwan.

E-mail address: ahsin@mail.ncku.edu.tw (C.-H. Chang).

http://dx.doi.org/10.1016/j.tjog.2013.01.017

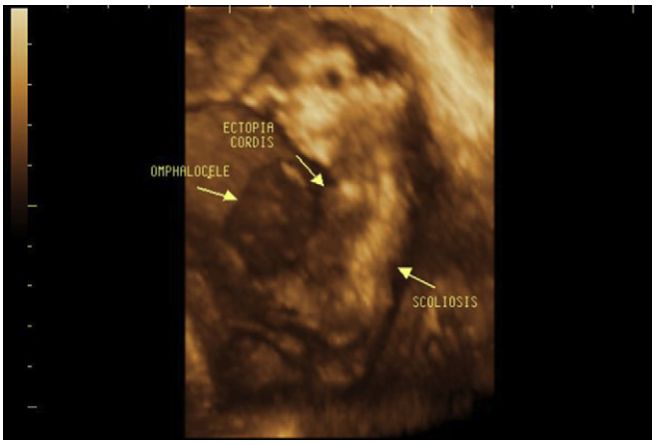

Fig. 1. The ectopia cordis, omphalocele, and marked spinal curvature were clearly observed by 3D US with surface mode.

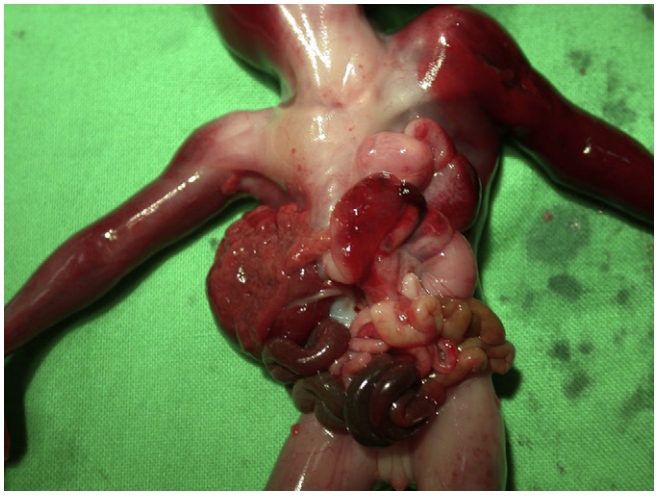

Fig. 2. The ectopia cordis with left cleft sternum, omphalocele, and scoliosis were noted in the gross appearance of the abortus.

beginning of the second trimester; only a few cases have been confirmed in the first trimester [9]. The 2D US with Doppler imaging may be the first choice for evaluation of abdominal wall defects, and 3D surface reconstruction provides a precise demonstration of complex malformations [10–12]. However, there were few cases that presented with POC born at term and treated by surgery after birth [2,13]. However, the prognosis of most cases of POC is still poor. We reported two cases of POC with different associated development. US plays an important role in the prenatal diagnosis of POC and the assistance of further management.

References

[1] Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and heart. Surg Gynecol Obstet 1958;107:602–14.
[2] Zidere V, Allan LD. Changing findings in pentalogy of Cantrell in fetal life. Ultrasound Obstet Gynecol 2008;32:835–7.
[3] Mueller GM, Weiner CP, Yankowitz J. Three-dimensional ultrasound in the evaluation of fetal head and spine anomalies. Obstet Gynecol 1996; 88:372–8.
[4] Liang RI, Huang SE, Chang FM. Prenatal diagnosis of ectopia cordis at 10 weeks of gestation using two-dimensional and three-dimensional ultrasonography. Ultrasound Obstet Gynecol 1977;10:137–9.
[5] Fox JE, Gloster ES, Mirchandani R. Trisomy 18 with Cantrell pentalogy in a stillborn infant. Am J Med Genet 1988;31:391–4.
[6] Parvari R, Carmi R, Weissenbach J, Pilia G, Mumm S, Weinstein Y. Refined genetic mapping of X linked thoracoabdominal syndrome. Am J Med Genet 1996;61:401–2.
[7] Pollio F, Sica C, Pacilio N, Maruotti GM, Mazzarelli LL, Cirillo P, et al. Pentalogy of Cantrell: first trimester prenatal diagnosis and association with multicystic dysplastic kidney. Minerva Ginecol 2003;55: 363–6.
[8] Polat I, Gül A, Aslan H, Cebeci A, Ozseker B, Caglar B, et al. Prenatal diagnosis of pentalogy of Cantrell in three cases, two with craniorachischisis. J Clin Ultrasound 2005;33:308–11.
[9] Sarkar P, Bastin J, Katoch D, Pal A. Pentalogy of Cantrell: diagnosis in the first trimester. J Obstet Gynecol 2005;25:812–3.
[10] Chen CP, Hsu CY, Wu PC, Tsai FJ, Wang W. Prenatal ultrasound demonstration of limb-body wall complex with megacystis. Taiwan J Obstet Gynecol 2011;50:258–60.
[11] Chen CP, Chang TY, Chern SR, Wang W. Third-trimester 3D ultrasound evaluation of thanatophoric dysplasia type I. Taiwan J Obstet Gynecol 2007;46:281–3.
[12] Lin IW, Chueh HY, Chang SD, Cheng PJ. The application of three-dimensional ultrasonography in the prenatal diagnosis of arthrogryposis. Taiwan J Obstet Gynecol 2008;47:75–8.
[13] Takaya J, Kitamura N, Tsuji K, Watanabe K, Kinoshita Y, Hattori Y, et al. Pentalogy of Cantrell with a double-outlet right ventricle: 3.5-year follow-up in a prenatally diagnosed patient. Eur J Pediatr 2008;167: 103–5.