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● *Original Contribution*

PRENATAL DIAGNOSIS OF FETAL ACRANIA USING THREE-DIMENSIONAL ULTRASOUND

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Abstract—Fetal acrania is uniformly lethal and termination is suggested whenever the diagnosis is made. Traditionally, the diagnostic tool was 2-D ultrasound (US). In this series, we report our work of detecting acrania using 3-D US. We reviewed our medical records of prenatal diagnosis on fetal acrania in National Cheng Kung University Hospital from May 1997 to December 2002. All the cases were scanned by a 3-D US scanner. In total, 29 cases of fetal acrania were diagnosed. The range of gestational age at prenatal diagnosis by US was between 11 and 21 weeks and 44% were depicted in the first trimester. Among them, 93.1% were isolated findings, and one was associated with trisomy 18. Comparing with previous literature, 3-D US can detect fetal acrania as early as 2-D US, and it also can provide additional vivid illustration after various modes of reconstruction, which 2-D US cannot. In conclusion, 3-D US may contribute to early detection of fetal acrania and provide a novel visual depiction of this defect after reconstruction; thus, assists substantially in diagnosis as well as consultation. (E-mail: ahsin@mail.ncku.edu.tw)

Key Words: **Fetal acrania, Prenatal diagnosis, 3-D ultrasound.**

INTRODUCTION

Acrania is a rare congenital anomaly that is defined as the absence of the flat bones of the skull with the presence of significant brain mass. The reported incidence is geographically- and population-dependent and is about 1 per 1000 to 2000 deliveries (Cafici and Sepulveda 2003; Sanders 1996). The prenatal diagnosis of acrania was traditionally made by using of 2-D transabdominal or transvaginal ultrasound (US) and only isolated cases or small series have been reported previously (Chen et al. 1992; Hautman et al. 1995; Johnson et al. 1997; Mannes et al. 1982; Weissman et al. 1997; Yang et al. 1992). The diagnosis has easily been made with the confident sonographic findings during the second trimester for decades and also can be made in the first trimester, after the 10th week of gestation (Johnson et al. 1997). Because acrania is invariably lethal, pregnancy termination can be offered at any time, whenever the diagnosis is made.

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Prenatal diagnosis in the first trimester, however, can be challenging because of the variable amount of brain tissue that may be present. Some ancillary sonographic findings, such as echogenic amniotic fluid in the first trimester, may also be used to help the diagnosis (Cafici and Sepulveda 2003). With the advent of 3-D US, various rendering modes of imaging may help to depict the condition and, actually, we have used this technique in prenatal diagnosis and reported the clinical application of 3-D US in detecting congenital anomalies since 1995 (Chang FM et al. 1997a, 1997b, 1997c; Chang CH et al. 2000a, 2000b, 2000c; Chang LW et al. 2002; Chuang et al. 2000; Kuo et al. 1992; Liang et al. 1997a, 1997b, 1998; Lai et al. 2000; Lin et al. 1998; Liu et al. 2003; Wang et al. 1999). Herein, we collected and analyzed the cases of acrania diagnosed by 3-D US retrospectively. Although using 3-D US to evaluate the fetal head anomalies was previously reported (Mueller et al. 1996; Pooh and Pooh 2001), to the best of our knowledge, this series is the first large one using 3-D US to approach this anomaly.

MATERIALS AND METHODS

From May 1997 to December 2002, the medical records of patients with fetal acrania were reviewed. In

addition to the cases depicted primarily during routine prenatal examinations, as a tertiary medical center in Southern Taiwan, regional clinics and hospitals also referred patients with suspected abnormalities for further US assessment. All the US examinations were done in the Antenatal Ultrasound Unit of Obstetrics and Gynecology, National Cheng Kung University Hospital, Tainan, Taiwan. After the diagnosis of fetal acrania was made, pregnancy termination was suggested and soon was performed in our hospital. The medical records are 3-D and 2-D US imaging records, admission notes, delivery notes and discharge summaries. According to the previous reports, fetal acrania is sporadic and not associated with chromosomal anomalies (Sanders 1996; Weissman et al. 1997). Therefore, karyotyping was not a routine examination in this series, and was done only when indicated.

For 3-D US examinations, we used a 3.5- to 7.0-MHz Voluson US scanner of conventional equipment (Kretz Voluson 530, 530D, Zipf, Austria) to obtain a three-orthogonal multiplanar view and, further, to generate 3-D reconstructive images of acrania by a variety of rendering modes of 3-D US, including surface-rendering mode, transparent mode, x-ray mode, gradient-light mode or as a mixture of various modes. The details of 3-D US scanning have been described previously (Chang FM et al. 1997a, 1997b, 1997c; Chang CH et al. 2000a, 2000b, 2000c; Chuang et al. 2000; Kuo et al. 1992; Liang et al. 1997a, 1997b, 1998; Lai et al. 2000; Lin et al. 1998; Wang et al. 1999).

RESULTS

In total, 29 cases were diagnosed as fetal acrania by 3-D US in 5 years. All the results of this series are summarized in Table 1. Of these, 28 cases (96.5%) were referred from local clinics or other hospitals. The mean gestational age of prenatal diagnosis by US was 15.3 weeks (range from 11 to 21 weeks). Maternal ages ranged from 20 to 40 years. Only one case had karyotyping for advanced maternal age and the result was trisomy 18 (case 29).

Table 1. Prenatal diagnosis of fetal acrania by 3-D us

Case no.	Maternal age (years)	Obstetric history	Gestational age (weeks)	US indication	US findings	Outcome	Associate conditions
1	25	G1P0	11	Referral	AC	TOP	–
2	25	G1P0	12	Referral	AC	TOP	–
3	30	G2P0A1	13	Referral	AC	TOP	–
4	22	G1P0	13	Referral	AC	TOP	–
5	28	G1P0	13	Referral	AC	TOP	–
6	31	G3P2	13	Referral	AC	TOP	–
7	23	G1P0	13	Referral	AC	TOP	–
8	24	G1P0	13	Referral	AC	TOP	–
9	28	G3P0A2	13	Routine	AC	TOP	–
10	25	G2P1	13	Referral	AC	TOP	–
11	28	G3P2	13	Referral	AC	TOP	–
12	23	G4P2A1	14	Referral	AC	TOP	–
13	21	G1P0	14	Referral	AC	TOP	–
14	36	G3P1A1	15	Referral	AC	TOP	–
15	28	G2P0A1	15	Referral	AC	TOP	–
16	20	G1P0	15	Referral	AC	TOP	–
17	35	G3P2	15	Referral	AC	TOP	–
18	20	G2P1	15	Referral	AC	TOP	–
19	26	G2P1	16	Referral	AC	TOP	–
20	32	G3P2	16	Referral	AC	TOP	–
21	27	G1P0	16	Referral	AC	TOP	–
22	32	G4P1A2	17	Referral	AC	TOP	–
23	23	G2P1	17	Referral	AC	TOP	–
24	24	G2P0A1	18	Referral	AC	TOP	–
25	28	G2P1	18	Referral	AC	TOP	–
26	25	G1P0	20	Referral	AC	TOP	–
27	29	G1P0	20	Referral	AC	TOP	–
28	33	G1P0	21	Referral	Twin with one acrania and multiple anomalies (cleft lip, hypoplastic left heart)	Delivered at 36 weeks	–
29	40	G2P1	21	Referral	AC, VSD, abdominal wall defect, club feet, upper limbs defect;	TOP	karyotype: 47, XY, +18

G = gravida; P = para; A = abortion; US = ultrasound; AC = acrania; TOP = termination of pregnancy; VSD = ventricular septal defect.