



在线全文

Cantrell综合征15例产前超声表现分析*

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【摘要】目的 总结分析Cantrell综合征胎儿产前超声表现及产后检查结果。**方法** 回顾性分析2018年3月至2023年7月产前超声诊断为Cantrell综合征并经引产标本或产后检查确诊的胎儿15例,分析产前超声图像表现及产后结果。**结果** 15例中有单胎10例及双胞胎之一5例。早孕期超声诊断13例,漏诊1例,漏诊率为7.1%;中孕期超声诊断2例。产前超声检查显示15例胎儿共有表现为异位心和腹部膨出团块。5例行胎儿超声心动图检查,发现心内畸形4例(80%);13例(86.7%)合并其他系统畸形;14例行颈项透明层(nuchal translucency, NT)检查的胎儿中有7例(50%)NT增厚,其中有颈部淋巴水囊瘤5例。10例单胎均被引产,引产胎儿标本外观与产前超声检查结果相符。5例双胞胎之一中有2例胎死宫内,2例行选择性减胎术,其中3例胎儿产后外观与产前超声检查结果相符,1例减胎术后胎儿在分娩时已显示不清;1例失访。4例引产胎儿行遗传学检查,均未查见相关的致病性或可能致病性变异。**结论** Cantrell综合征产前超声共有表现为异位心和腹部膨出团块,心内畸形及伴发畸形多见;绝大多数可在早孕期由超声诊断,但也有漏诊可能,需在中孕期密切随访。

【关键词】 Cantrell五联征 超声检查, 产前 胎儿

Analysis of Prenatal Ultrasound Manifestations in 15 Cases of Cantrell Syndrome LIANG Xiaoling¹, YE Lu^{2△}.

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【Abstract】 Objective Cantrell syndrome, a rare congenital disorder, is characterized by a unique collection of defects on the midline abdominal wall, the lower sternum, the anterior diaphragm, and the diaphragmatic pericardium in addition to some form of intracardiac defect. So far, most of the reports on fetuses with Cantrell syndrome worldwide are either case reports or literature reviews, and few comprehensive studies on fetuses with Cantrell syndrome have been reported, especially in domestic literature. This study aims to provide a detailed analysis of 15 cases of Cantrell syndrome fetuses, focusing on their prenatal ultrasound manifestations and postnatal examination outcomes. **Methods** A retrospective analysis was conducted with 15 cases of fetuses diagnosed with Cantrell syndrome via prenatal ultrasound examinations between March 2018 and July 2023. Ultrasound examinations were performed in accordance with the Guidelines for Obstetric Ultrasound in China, including first-trimester fetal ultrasound scan and routine second-trimester fetal ultrasound scan. Gestational age was evaluated and nuchal translucency (NT) was measured during first-trimester fetal ultrasound scan at 11 to 13+6 weeks. The diagnostic criterion for NT thickening was $NT \geq 3.0$ mm and the screening of severe fetal structural malformations was performed, including the screening of the head, the neck, the thorax, the abdominal content, the abdominal wall, the limbs and other structures. During routine second-trimester fetal ultrasound scan, the fetal biometry was assessed and an anatomy survey was performed. Post-induction and postnatal outcomes of fetuses diagnosed with Cantrell syndrome by prenatal ultrasound were followed up by postnatal observation, inquiries with the electronic medical record system, or telephone follow-up. The prenatal ultrasound imaging manifestations and features of the fetuses with Cantrell syndrome, as well as their post-induction or postnatal examination results were comprehensively summarized and analyzed. **Results** The study involved pregnant women of the average age of 30.1 ± 3.5 years, with ultrasound diagnoses made between 11 to 26 weeks of gestation (mean: 13.4 ± 4.0 weeks). Among the 15 cases, there were 10 singleton pregnancies and 5 cases of one twin in a pair of twins. These twins comprised 3 monochorionic diamniotic twins and 2 dichorionic diamniotic twins, with Cantrell syndrome present in one of the twins in all 5 cases. Thirteen cases were diagnosed by fetal ultrasound scan conducted in the first trimester, with 10 being singleton pregnancies and 3 being twin pregnancies (1 monochorionic diamniotic twins and 2 dichorionic diamniotic twins). One case was missed in the first-trimester ultrasound scan, resulting in a missed diagnosis rate of 7.1%. Two cases were diagnosed in second-trimester fetal ultrasound scan, both involving monochorionic diamniotic twins. One case was a referral from another hospital at 19 weeks, while the other was initially not diagnosed for Cantrell syndrome and was

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diagnosed at 26 weeks. Prenatal ultrasound examinations revealed a consistent pattern of abnormalities across all 15 fetuses, including manifestations of ectopic cordis combined with abdominal protrusion mass. Specifically, 4 cases were diagnosed with omphalocele, 4 with gastroschisis, and the remaining 7 had uncertain coverage of the membrane on the surface of the abdominal protrusion mass. Six fetuses had complete ectopic cordis, while nine had partial ectopic cordis. Fetal echocardiography was performed in 5 cases, revealing intracardiac malformations in 4 cases (80%). Notably, 2 cases were diagnosed in the second trimester, including one with right ventricular hypoplasia accompanied by interventricular septal defect and another with double outlet right ventricle accompanied by interventricular septal defect. Additionally, 2 cases were diagnosed in the first trimester, one with single atrium and single ventricle, and the other with complete transposition of the great arteries. Of the 15 cases of fetuses with Cantrell syndrome, 13 (86.7%) exhibited concomitant malformations in other systems. These included 7 cases of spinal malformations, 4 limb abnormalities, 3 umbilical cord abnormalities, 2 central nervous system malformations, 1 facial malformation, and 2 fetal hydrops. Spinal malformations were the most prevalent concomitant malformation, accounting for 46.7% of all cases. Among the 14 fetuses undergoing NT examination, 7 (50%) had increased NT, and 5 of them had cystic hygroma. All 10 singleton pregnancies underwent induced abortion, and the appearance of the induced fetuses was consistent with the prenatal ultrasound manifestations. In the twin pregnancies, 2 cases experienced intrauterine fetal death, while 2 underwent selective reduction. Notably, 3 of these cases exhibited postnatal appearances consistent with prenatal ultrasound manifestation, while 1 case showed an indistinct appearance after selective reduction during delivery. One case was lost to follow-up. Genetic testing was conducted for 4 induced fetuses, none of which yielded any relevant pathogenic or potentially pathogenic variants.

Conclusion In conclusion, Cantrell syndrome manifests prenatally with ectopic cordis combined with abdominal protrusion mass, often accompanied by intracardiac malformations and other concomitant malformations. While most cases can be diagnosed in the first trimester, there remains the possibility of missed diagnoses, which underscores the importance of close follow-up in the second trimester.

【Key words】 Pentalogy of Cantrell Ultrasonography, prenatal Fetus

Cantrell综合征也被称为Cantrell五联征,是罕见的先天性综合征,表现为腹中线脐上腹壁缺损、胸骨下段缺损、膈肌前部缺损、膈心包缺如及心内畸形等五种畸形^[1]。自1958年Cantrell等首次报道该综合征以来,新生儿科和外科技术的进步为其治疗提供了更好的条件,但是该病的存活率仍仅为61%^[2]。1991年BENNETT等^[3]首次采用超声检查对Cantrell综合征胎儿进行诊断,迄今为止国内外对于胎儿Cantrell综合征的报道多为个案报道或文献复习,国内对胎儿Cantrell综合征的总结报道较少^[4-7]。本研究对15例Cantrell综合征胎儿的产前超声表现及产后检查结果进行回顾性总结分析。

1 资料与方法

1.1 研究对象

回顾性分析2018年3月至2023年7月在四川大学华西第二医院建档或从外院转诊而行产前超声检查诊断为Cantrell综合征并经引产标本或产后检查确诊为Cantrell综合征的胎儿15例。本研究遵守《赫尔辛基宣言》,经四川大学华西第二医院医学伦理委员会批准,批准号:医学科研2022伦审批第(227)号。研究对象在行产前超声检查前均签署知情同意书。

1.2 仪器与方法

超声检查仪器包括GE Voluson E8、GE Voluson

E10、PHILIPS EPIQ 7C彩色超声诊断仪,探头包括C1-6、C4-8、C5-1凸阵探头,频率分别为1~6 MHz、4~8 MHz、1~5 MHz。

超声检查按照《中国产科超声检查指南》^[8]进行,包括妊娠11~13⁺6周超声检查及中孕期Ⅲ级超声检查。在11~13⁺6周超声检查中,进行孕龄的评价及颈项透明层(nuchal translucency, NT)测量(NT增厚的诊断标准为NT≥3.0 mm),并进行胎儿严重结构畸形的筛查,包括头部、颈部、胸腔、腹腔及腹壁、四肢,其中胸腔切面用于观察肺、有无胸腔积液及占位、心脏位置及搏动,腹腔及腹壁切面用于观察脐带插入处、胃泡位置等;在中孕期Ⅲ级超声检查中,进行胎儿生物学径线的测量及解剖结构的检查^[8]。超声检查发现胎儿同时具有心脏异位、高位脐膨出或腹裂即考虑诊断为Cantrell综合征^[4,7]。此外,检查胎儿有无合并其他畸形,若合并其他畸形则对相应部位进行详细扫查。诊断病例留存的所有超声图像均存储于超声工作站中。

对产前超声诊断为Cantrell综合征的胎儿进行随访,直至引产后或产后,随访方式包括产后观察、电子病历系统查询或电话随访。对Cantrell综合征胎儿产前超声表现及特征、引产或产后检查结果进行总结分析。

2 结果

15例Cantrell综合征胎儿的基本资料、产前超声表现

及引产或产后检查结果见表1。

2.1 基本资料

孕妇平均年龄(30.1±3.5)岁, 超声诊断孕周11~26周, 平均(13.4±4.0)周。15例胎儿包括10例单胎和5例双胎, 其中单绒毛膜双羊膜囊(单绒双羊)3例, 双绒毛膜双羊膜囊(双绒双羊)2例, 5例双胎均为双胎之一Cantrell综合

征。早孕期超声诊断Cantrell综合征13例, 漏诊1例, 漏诊率为7.1%; 早孕期诊断的Cantrell综合征中有10例为单胎妊娠, 3例为双胎妊娠(单绒双羊1例, 双绒双羊2例)。中孕期超声诊断Cantrell综合征2例, 均为单绒双羊, 其中1例为19周由外院转诊, 另1例自早孕期在本院建档, 12周和22周被漏诊Cantrell综合征而仅诊断脐膨出, 至26周被

表 1 Cantrell综合征胎儿的产前超声表现及产后检查结果

Table 1 Prenatal ultrasound findings and postnatal results of fetuses with Cantrell syndrome

Fetus number	Maternal age/yr.	GD/week	Number of fetuses	NT	Ultrasound findings	Concomitant malformations	Intracardiac abnormalities	Follow-up result
1	35	11	Singleton	Increased	Abdominal protruding mass and complete ectopic cordis	Cystic hygroma	—	Induced abortion at another hospital
2	28	13	Singleton	Increased	Omphalocele and partial ectopic cordis	Scoliosis, posterior fossa cyst disappearance, stiff morphology of limbs with restricted movement, cystic hygroma, and fetal hydrops	—	Induced abortion at another hospital
3	32	19	MCDA twins	—	Omphalocele and complete ectopic cordis	Anencephaly, cleft lip and palate, one orbit not visible, and single umbilical artery	Right ventricular hypoplasia and ventricular septal defect	20-week spontaneous miscarriage of twin stillbirths: the malformed stillborns had anencephaly, cleft lip and palate with eye deformities, omphalocele, and complete ectopic cordis
4	27	13	MCDA twins	Normal	Gastroschisis and complete ectopic cordis	Abnormal spinal curvature	Double outlet right ventricle and ventricular septal defect (diagnosed in the 2nd trimester)	Selective reduction in the 2nd trimester and Cesarean section at 32 weeks: the malformed stillborns had gastroschisis, complete ectopic cordis, and scoliosis
5	34	11	Singleton	Increased	Gastroschisis and partial ectopic cordis	Cystic hygroma, abnormal spinal curvature, and abnormal morphology of the left hand	—	Induced abortion: swelling of the head and neck skin, gastroschisis, partial ectopic cordis, scoliosis, and left upper limb being small and flexed. Whole-exome sequencing analysis did not reveal any pathogenic or potentially pathogenic variants associated with the condition.
6	30	13	DCDA twins	Normal	Gastroschisis and complete ectopic cordis	Abnormal spinal curvature	—	Selective reduction at 15 weeks and full-term Cesarean section: the malformed fetus after the selective reduction procedure was adhered to the placenta and appeared indistinct.
7	29	11	Singleton	Increased	Abdominal protruding mass and partial ectopic cordis	Scoliosis and disordered vertebral alignment with partial vertebral absence	Single atrium and single ventricle	Induced abortion: gastroschisis, partial ectopic cordis, and scoliosis. Chromosome analysis did not reveal any pathogenic or potentially pathogenic variants associated with the condition.
8	26	26	MCDA twins	Normal	Omphalocele and partial ectopic cordis	Bilateral talipes equinovarus, short umbilical cord, and spinal scoliosis with disordered alignment of thoracolumbar vertebrae	None	Malformed fetus deceased in utero in the 2nd trimester and emergency Cesarean section at 28 weeks due to severe vaginal bleeding: the malformed stillborn exhibited omphalocele, partial ectopic cordis, bilateral talipes equinovarus, and scoliosis.
9	32	12	Singleton	Increased	Gastroschisis and partial ectopic cordis	Bilateral talipes equinovarus	—	Induced abortion: gastroschisis, partial ectopic cordis, and bilateral talipes equinovarus

续表 1

Fetus number	Maternal age/yr.	GD/week	Number of fetuses	NT	Ultrasound findings	Concomitant malformations	Intracardiac abnormalities	Follow-up result
10	33	13	Singleton	Normal	Abdominal protruding mass and complete ectopic cordis	Single umbilical artery	Complete transposition of great arteries	Induced abortion: gastroschisis and complete ectopic cordis. Chromosome analysis did not reveal any pathogenic or potentially pathogenic variants associated with the condition.
11	33	12	Singleton	Increased	Abdominal protruding mass and partial ectopic cordis	Cystic hygroma	—	Induced abortion: gastroschisis, partial ectopic cordis, and swelling of the neck skin. Chromosome analysis and whole-exome sequencing analysis did not reveal any pathogenic or potentially pathogenic variants associated with the condition.
12	27	12	Singleton	Increased	Omphalocele and partial ectopic cordis	Cystic hygroma and fetal hydrops	—	Induced abortion at another hospital
13	23	12	Singleton	Normal	Abdominal protruding mass and partial ectopic cordis	None	—	Induced abortion at another hospital
14	28	12	DCDA twins	Normal	Abdominal protruding mass and partial ectopic cordis	Abnormal spinal curvature	—	Lost to follow-up
15	34	12	Singleton	Normal	Abdominal protruding mass and complete ectopic cordis	None	—	Induced abortion at another hospital

— indicates that the examination was not performed. GD: gestational age at diagnosis; NT: nuchal translucency; MCDA: monochorionic diamniotic; DCDA: dichorionic diamniotic.

诊断为Cantrell综合征。

2.2 产前超声表现

2.2.1 共有畸形

15例Cantrell综合征胎儿产前超声图像均表现为心脏异位合并高位脐膨出或腹裂(图1、图2)。15例胎儿中,腹部膨出团块被诊断为脐膨出4例、腹裂4例,其余7例胎儿的腹部膨出团块表面是否覆盖包膜不明确。15例胎儿中,6例心脏完全异位于胸腔外,9例心脏部分异位于胸腔外。

15例 Cantrell 综合征胎儿中有10例早孕期胎儿未行胎儿超声心动图检查。5例胎儿(3例早孕期和2例中孕期)进行了胎儿超声心动图检查,其中1例未发现明确的心内畸形,其余4例(80%)产前超声明确诊断心内畸形。2例在中孕期诊断,其中1例表现为右室小、肺动脉偏窄、室间隔上份连续性中断2 mm,诊断为右室发育不良伴室间隔缺损(图2);另1例表现为主动脉从右室发出,肺动脉骑跨于室间隔上且大部分由右室发出,室间隔上份连续性中断4 mm,诊断为右室双出口伴室间隔缺损。2例在早孕期诊断,其中1例仅见单心房和单心室,两支大血管连接单心室,诊断为单心房单心室;另1例表现为主动脉增宽、肺动脉狭窄,二者呈前后平行排列,右室与主动脉连接,左室与肺动脉连接,右位主动脉弓,诊断为完全性大动脉转位。

2.2.2 伴发畸形

15例Cantrell综合征胎儿中13例(86.7%)伴发其他畸形,包括:脊柱畸形7例,肢体异常4例,脐带异常3例,神经系统异常2例,颜面部异常1例(图2),全身水肿2例。伴发畸形中以脊柱畸形(46.7%)居多。

13例在早孕期被诊断为Cantrell综合征的胎儿伴发其他系统畸形11例。2例在中孕期被诊断为Cantrell综合征的胎儿均伴发其他系统畸形。

2.2.3 超声软指标异常

15例Cantrell综合征胎儿中有14例胎儿于早孕期行NT检查,发现NT增厚7例(50%),其中颈部水囊瘤5例(图1);1例胎儿转诊检查时为中孕期,未行NT测量。

2例未伴发其他系统畸形的 Cantrell 综合征胎儿早孕期NT正常。

2.3 随访引产或产后检查结果

10例被超声诊断为Cantrell 综合征的单胎均被引产。其中,5例于外院引产,5例于本院引产,引产胎儿外观与产前超声检查结果相符(图1)。

5例双胎之一Cantrell 综合征中,2例胎死宫内,2例行选择性减胎术,具体如下:1例于20周自然流产双死胎,1例中孕期双胎之一(畸形胎儿)胎死宫内、于28周因阴道大出血而行急诊剖宫产,1例中孕期行选择性减胎术后于32周剖宫产分娩,以上3例胎儿产后外观与产前超声表现

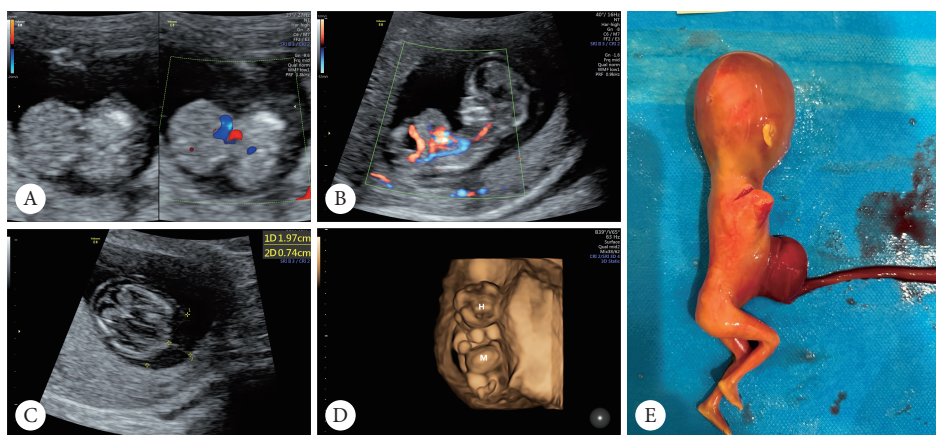


图1 编号11胎儿Cantrell综合征超声声像图及引产后外观

Fig 1 Ultrasonographic image and post-induction appearance of the No. 11 fetus with Cantrell syndrome

This is a singleton pregnancy at 12 weeks. Abdominal transverse section two-dimensional and color Doppler images (A) and sagittal section color Doppler image (B) revealed partial ectopic cordis with an abdominal protrusion mass (liver). The neck transverse section showed cystic hygroma (C). Three-dimensional ultrasound imaging demonstrated the abdominal protrusion mass (D). Post-induction appearance showed gastroschisis and neck skin swelling (E). H: head; M: mass.

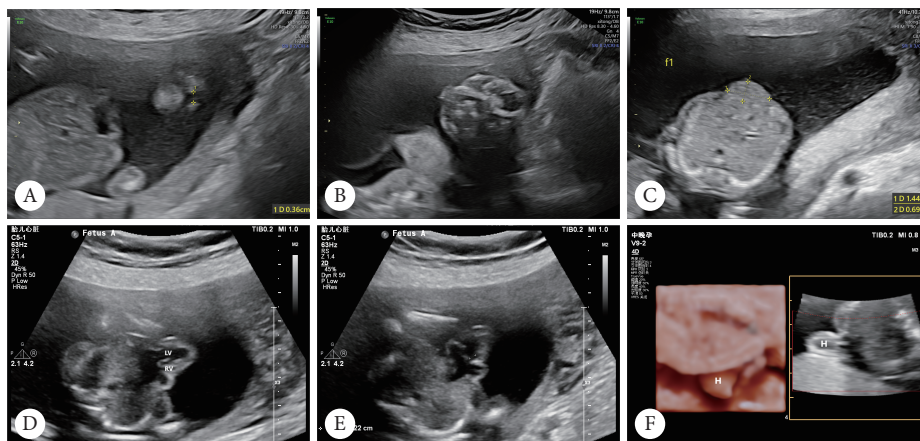


图2 编号3胎儿Cantrell综合征超声声像图

Fig 2 Ultrasonographic image of the No. 3 fetus with Cantrell syndrome

This is one of the twins in a monochorionic diamniotic twin pregnancy at 19 weeks. The fetal face coronal section revealed a cleft lip with a continuous opening in the upper lip (A). In the orbit transverse section, one side was not visible (B). The transverse section of the upper abdomen revealed an abdominal protrusion mass (C). Fetal echocardiography four-chamber view displayed a small right ventricle, a normal-sized left ventricle (D), and a continuous interruption of ventricular septum (E). Three-dimensional imaging showed that the heart was outside the chest cavity (F). LV: left ventricle; RV: right ventricle; H: heart.

相符;1例于15周行选择性减胎术后回当地妊娠至足月剖宫产分娩,减胎术后的胎儿贴附于胎盘且已显示不清。1例失访。

2.4 胎儿遗传学检查结果

4例引产胎儿的组织被送检行遗传学检查,均未查见相关的致病性或可能致病性变异。

3 讨论

Cantrell综合征是罕见的先天性综合征,在活产婴儿中的发病率约为1/65 000至1/200 000^[2]。Cantrell综合征的发生原因尚不明确,多数学者认为Cantrell综合征五种畸形的发生密切相关,是由胚胎期中胚层移行发育缺

陷及变异所致^[9],有学者将Cantrell综合征、泄殖腔外翻综合征和肢体-体壁综合征视为由于两侧褶、头褶、尾褶闭合不全而导致的一系列疾病谱^[10]。

Cantrell综合征分为完全型和不完全型^[9]。由于超声检查具有局限性以及胎儿尚未发育成熟,产前超声无法同时准确诊断出Cantrell综合征的5种结构畸形,尤其是心内结构异常、心包及膈肌缺损在早孕期难以确诊,产前超声难以分型诊断,因此本研究未进行疾病分型。Cantrell综合征的典型产前超声表现为心脏(部分或全部)异位于胸腔外合并脐膨出或腹裂,可在早孕期被诊断,但通常在中孕期更容易被诊断,三维超声可更全面细致显示结构,磁共振成像可用于进一步确诊^[11]。本组

15例胎儿的产前超声诊断最早孕周为11周,与文献报道的诊断孕周^[5-7]相近。本组胎儿中有14例于早孕期(11~13⁺₆周)接受超声检查,其中13例被诊断为Cantrell综合征,这表明早孕期超声检查能检出大多数Cantrell综合征。但是,早孕期超声检查也可能漏诊心脏异位,从而漏诊Cantrell综合征。本研究中1例双胞胎孕妇自早孕期行定期产前检查,但其双胎之一在12周和22周的超声检查中均被漏诊Cantrell综合征而仅诊断脐膨出,直至26周才被诊断为Cantrell综合征。调取12周和22周超声图像显示脐部腹壁前方占位,占位内见肝脏、肠管等腹腔脏器而未见确切心脏。分析漏诊原因如下:早孕期至中孕初期胎儿心脏异位不明显;检查医师对该病认识不足,忽略了对心脏、胸骨、膈肌的观察。王莹莹等^[6]关于Cantrell五联征的回顾性研究也报道了1例漏诊病例,该病例于孕13周⁺时漏诊,仅表现出NT增厚(3.1 mm),于孕17周⁺复诊时得以诊断。因此,当早孕期发现脐部占位或NT增厚等线索后,应再仔细观察心脏位置以及胸骨、膈肌的连续性并密切随访,以免Cantrell综合征被漏诊。反之,也有文献报道Cantrell综合征胎儿在妊娠早期相比妊娠后期心脏异位程度更重,该文献报道的3例胎儿在首次超声检查时均存在严重腹壁缺损并伴有明显的心脏向胸外突出,但在20周和26周时心脏却位于胸腔内,患儿出生后接受手术均存活^[12],这说明Cantrell综合征的心脏异位随孕周增加可能有所改善。综上所述,对于早孕期超声检查发现脐部占位的胎儿,无论是否合并心脏异位,都应密切随访,其原因在于心脏异位程度可能有所变化。

Cantrell综合征胎儿心内畸形复杂多样,产后检查发现的心内畸形主要有室间隔缺损、房间隔缺损、左心室憩室、肺动脉狭窄或闭锁和法洛四联征,其中最常报道的是室间隔缺损,其他心内畸形包括右位心、左侧上腔静脉、肺静脉异位引流、动脉导管未闭、三尖瓣闭锁、大动脉转位、永存动脉干、房室间隔缺损、右心室憩室、右室双出口和左心发育不良综合征等^[1,13-15]。本组15例Cantrell综合征胎儿中有5例行胎儿超声心动图检查,其中4例被诊断为心内畸形,心内畸形发生率高达80%。因此,对于可疑Cantrell综合征者,胎儿超声心动图检查至关重要,可确定心内畸形的有无、类型和严重程度,有助于评估预后。本研究中接受了胎儿超声心动图检查的病例仅占少数,这是本研究的局限性,在未来的临床实践中,对于疑似Cantrell综合征者应行进一步胎儿心脏检查以详细评估心脏结构。

Cantrell综合征胎儿常伴发胸腔或腹腔脏器畸形、颅面部畸形、肢体畸形、脊柱畸形、脐带畸形等^[13,16]。本组

15例胎儿中大多数(86.7%)合并其他系统畸形,这说明Cantrell综合征伴发畸形的情况多见。本组胎儿中1例胎儿合并脊柱畸形、肢体畸形及脐带短,2例合并脊柱畸形和肢体畸形,4例单纯合并脊柱畸形,因此需要与肢体-体壁综合征相鉴别。此外,Cantrell综合征还须与单纯腹裂、单纯胸外异位心、羊膜带综合征等进行鉴别。

本组胎儿中有14例于早孕期行NT检查,其中NT增厚(含颈部水囊瘤)的发生率是50%。NT增厚在Cantrell综合征中并非意外发现,它可被视为心内畸形的间接征象,可能是心内畸形和心力衰竭所致^[16]。在GRIGORE等^[16]对67例Cantrell综合征的病例报道和文献综述中,44例(65.67%)具有NT检查结果,其中39例NT增厚,检出率高达88.63%。在某些早孕期诊断Cantrell综合征的病例中,正是NT增厚引起了检查者对胎儿的早期关注^[12]。因此,在早孕期胎儿超声检查中发现NT增厚时,应进一步关注胎儿各系统结构。

本研究报道的15例胎儿中有5例为双胎之一Cantrell综合征,占比高达1/3,高于国内学者之前报道的比例(2/20、1/21、3/12)^[4,6-7]。这提示Cantrell综合征在双胎中较在单胎中可能更加高发,在对双胎进行超声检查时须更加警惕。

Cantrell综合征预后较差,生存率低至37.3%^[13],其预后主要取决于心内畸形和其他畸形的类型和严重程度,但也取决于异位心脏的位置,部分Cantrell综合征患者可通过手术治疗存活^[2,17]。诊断为Cantrell综合征的患者应接受产前咨询,对于羊水穿刺显示核型异常的严重病例可进行终止妊娠^[2]。双胎之一Cantrell综合征可进行选择性减胎术。此外,有研究者对存在大量心包积液的Cantrell综合征胎儿进行心包-羊膜腔分流术,可能使胎儿肺生长和发育有所改善,这是对该疾病胎儿宫内治疗的首次报道^[18]。

综上所述,Cantrell综合征产前超声共有表现为异位心和腹部膨出团块,心内畸形及伴发畸形多见;绝大多数Cantrell综合征可在早孕期由超声诊断,但也有漏诊可能,需在中孕期密切随访。

* * *

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