Case Report

First Trimester Diagnosis of Pentalogy of Cantrell (POC) by Transabdominal 2-Dimentional (2D) Ultrasonography, First Case Report in Thailand

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Background: *Pentalogy of Cantrell (POC) is an extremely rare and usually lethal congenital anomaly; to the best of our knowledge, less than 10 cases of first trimester diagnosis have been reported by using only transabdominal 2-dimensional (2D) ultrasonography.*

Case Report: *A 37-years-old Thai woman, gravida 3, para 2, 13 weeks of gestation presented at early pregnancy clinic for dating confirmation. Ultrasonography revealed a fetus with multiple anomalies, so she was referred to Maternal Fetal Medicine (MFM) unit. Transabdominal 2D ultrasonography demonstrated a single alive fetus with ectopia cordis and evisceration of liver and bowel loops. Then, the diagnosis of pentalogy of Cantrell was made. After counseling about poor prognosis of POC, the patient and her husband decided termination of pregnancy. The autopsy revealed scoliosis, herniation of chest and abdominal viscera organs including heart, lungs, liver, spleen, small and large intestines through a paramedian abdominal wall fusion defect represented the diagnosed POC.*

Conclusion: *The authors represent the first case of POC in Thailand diagnosed by transabdominal 2D ultrasonography in first trimester. Therefore, careful examination ultrasonography possibly provide an excellent view diagnosed lethal congenital anomaly in the first trimester which reduce mortality and morbidity to the patient with late termination.*

Keywords: *Pentalogy of Cantrell, Ectopia cordis, Evisceration of bowel loops*

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Pentalogy of Cantrell (POC) is an extremely rare and usually lethal congenital anomaly consisting of the following 5 defects: 1) supraumbilical abdominal wall defects; 2) anterior diaphragmatic defects; 3) diaphragmatic pericardial defects; 4) lower sternal defects; and 5) intracardiac defects. The prevalence is 5.5 to 7.9 per million live births[1]. It was first recognized in 1958[2] and revised by Toyama later in 1972[3]. Until now, the pathogenesis is still unknown.

Because the diagnosis of this bizarre condition is usually made in the second trimester; to the best of our knowledge, less than 10 cases of first trimester diagnosis have been reported by using only transabdominal 2-dimensional (2D) ultrasonography[4-6]. Thus, we aimed to present the first case of POC in Thailand diagnosed by 2D ultrasonography in first trimester at Thammasat University Hospital.

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Case Report

A 37 years old Thai woman, gravida 3, para 2 visited at Thammasat University Hospital for first antenatal care. She had no previous family or medical history. Her previous pregnancy were delivered vaginally at term without any complication.

At the hospital, a routine first trimester ultrasonography was performed at early pregnancy clinic for dating confirmation and prenatal counseling. Ultrasonography revealed a fetus with multiple anomalies, so she was referred to Maternal Fetal Medicine (MFM) unit. Transabdominal 2D ultrasonography using GE Voluson E8 demonstrated a single alive 13 weeks of gestational fetus with ectopia cordis (Fig. 1) and evisceration of liver and bowel loops (Fig. 2). The diaphragm was not visualized. Then, the diagnosis of pentalogy of Cantrell was made.

After counseling about poor prognosis of POC, the patient and her husband decided termination of pregnancy. Misoprostol, a prostaglandin analogue, was prescribed for transvaginal administration. A fetus

Fig. 1 Two-dimensional (2D) ultrasound images at 13 weeks of gestation showing an ectopia cordis.

Fig. 2 Two-dimensional (2D) ultrasound images at 13 weeks of gestation showing evisceration of liver and bowel loops (liver; yellow arrow and bowel loops; white arrow).

was delivered on that day. Postnatal examination showed total exposure of the heart and a supraumbilical abdominal wall defect with herniated liver and intestines, which was compatible with the prenatal ultrasonographic finding. The autopsy revealed scoliosis (Fig. 3), herniation of chest and abdominal viscera organs including heart, lungs, liver, spleen, small and large intestines through a paramedian abdominal wall fusion defect represented the diagnosed POC (Fig. 4). The placenta and umbilical cord were unremarkable.

Discussion

Pentalogy of Cantrell (POC) is a rare syndrome

Fig. 3 The autopsy of the fetus demonstrated scoliosis.

which associated with high mortality rate[7]. The survival rate is quietly low depending on spectrum of congenital defects including diaphragm, abdominal wall, pericardium, heart and lower sternum[8-10]. In 2014, Zhang X reported a mortality rate of 61%[11].

Cantrell JR suggested that the pathogenesis of POC may involve a failure in migration of lateral mesoderm to midline between 14 and 18 days after conception[2]. This failure causes defect of sternum and upper abdomen so the visceral organ eviscerated. And this also creates anterior diaphragmatic and pericardial defects from failed development of septum transversum. However, most cases are sporadic and few cases associated with aneuploidy[12]. Until now, the pathogenesis is still not clearly known.

Recently, multiple modalities have been used to diagnose POC prenatally. The 2-dimensional (2D) transabdominally and transvaginally, 3-dimensional (3D) and color Doppler ultrasonography are commonly used in diagnosis[13-16]. Others are fetal magnetic resonance imaging (MRI) and embryofetoscopy which were not routinely used due to invasiveness and high cost[17]. In our case, the patient was 13 weeks of gestation. We first observed ectopia cordis via 2D

Fig. 4 The autopsy of the fetus revealed herniation of chest and abdominal viscera organs including heart, lungs, liver, spleen, small and large intestines.

transabdominal ultrasonography. Then, we clearly identified evisceration of liver and bowel loops.

At this gestational age, there are no physiologic herniation in normal fetus. Therefore, we didn't performed further color Doppler, 3D ultrasonography, or fetal MRI because the 2 existed finding already guided to the diagnosis of POC.

Conclusion

The authors represent the first case of POC in Thailand diagnosed by transabdominal 2D ultrasonography in first trimester. Therefore, careful examination ultrasonography possibly provide an excellent view diagnosed lethal congenital anomaly in the first trimester. Early diagnosis and reasonable cost-effectiveness reduce mortality and morbidity to the patient with late termination.

What is already known on this topic?

POC is an extremely rare, complex and usually lethal congenital anomalies. It was diagnosed by multiple modalities. Less than 10 cases of first trimester diagnosis have been reported by using only transabdominal 2D ultrasonography.

What this study adds?

This study represents the first case of POC in Thailand diagnosed by transabdominal 2D ultrasonography in first trimester.

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Potential conflict of interests

None.

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การวินิจฉัยโรคเพนตาโรจีออฟแคนเทรลของทารกในครรภ์ด้วยการตรวจคลื่นเสียงความถี่สูงสองมิติทางหน้าท้องในไตรมาสแรก: รายงานผู้ป่วย

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ภูมิหลัง: *โรคเพนตาโรจีออฟแคนเทรลเป็นโรคที่พบได้น้อยมากและทารกมักเสียชีวิตหลังคลอดจากการทบทวนวรรณกรรมมีรายงานผู้ป่วยไม่เกิน 10 ราย ที่วินิจฉัยโรคเพนตาโรจีออฟแคนเทรลด้วยการคลื่นเสียงความถี่สูงสองมิติทางหน้าท้องในไตรมาสแรก*

รายงานผู้ป่วย: *สตรีไทยอายุ 37 ปี ตั้งครรภ์ครั้งที่ 3 อายุครรภ์ 13 สัปดาห์ มาพบแพทย์เพื่อฝากครรภ์ครั้งแรก ตรวจพบความผิดปกติหลายอย่าง ขณะที่ตรวจคลื่นเสียงความถี่สูงเพื่อยืนยันอายุครรภ์ จึงได้รับการตรวจเพิ่มเติมที่หน่วยเวชศาสตร์มารดาและทารกในครรภ์ การตรวจคลื่นเสียงความถี่สูงสองมิติทางหน้าท้องพบทารกมีชีวิตในครรภ์มีหัวใจอยู่นอกช่องอก และมีตับและลำไส้ยื่นออกจากผนังหน้าท้องจึงได้รับการวินิจฉัยเป็นโรคเพนตาโรจีออฟแคนเทรล ผู้ป่วยและสามีได้รับคำปรึกษาจากแพทย์ผู้ดูแลจึงตัดสินใจยุติการตั้งครรภ์ ผลชันสูตรพลิกศพพบทารกมีกระดูกสันหลังโค้งงอ มีอวัยวะภายใน ได้แก่ หัวใจ ปอด ตับ ม้าม ลำไส้เล็กและลำไส้ใหญ่อยู่นอกตัวทารกเข้าได้กับโรคเพนตาโรจีออฟแคนเทรล*

สรุป: *การวินิจฉัยโรคเพนตาโรจีออฟแคนเทรลของทารกในครรภ์ด้วยการตรวจคลื่นเสียงความถี่สูงสองมิติทางหน้าท้อง ในไตรมาสแรกในผู้ป่วยรายนี้เป็นครั้งแรกในประเทศไทย ดังนั้นเราสามารถวินิจฉัยความผิดปกติแต่กำเนิดชนิดรุนแรงได้ด้วยการตรวจคลื่นเสียงความถี่สูงสองมิติทางหน้าท้องตั้งแต่ในไตรมาสแรก*