

Ectopia Cordis and Pentalogy of Cantrell: Report of Two Cases

Murat Bakacak¹, Mehmet Sühha Bostancı^{2*}, Salih Serin¹, Cemil Goya³, Zeynep Bakacak⁴, Fazlı Avcı¹

Abstract

Ectopia cordis (EC) is the presence of a live, beating heart outside the thorax and is one of the most unique congenital anomalies. EC has been a weak association noted with trisomy 18 and a few cases have been associated with other chromosomal abnormalities. Pentalogy of cantrell (PC) is a rare congenital syndrome of abdominal wall defect, lower sternal defect, diaphragmatic pericardial defect, anterior diaphragmatic defect, and intracardiac abnormalities. It has a rare frequency of about 1/100,000 births. The hallmark of this syndrome is an omphalocele associated with EC. Only a few patients with the full spectrum of the pentalogy have been described. Only very few patients survive attempts at surgical repair, the main causes of death being tachyarrhythmias, bradycardia, low blood pressure, rupture of the diverticulum, and heart failure. Early diagnosis is important about discuss the option of elective termination with the parents if intracardiac anomalies of incompatible with life are noted. We report two cases of PC diagnosed in early second trimester.

Keywords: Pentalogy of Cantrell, Ectopia Cordis, Congenital Heart Defect

Introduction

By definition, ectopia cordis (EC) is the presence of a live, beating heart outside the thorax and is one of the most unique congenital anomalies. Pentalogy of Cantrell (PC) is a rare congenital syndrome of abdominal wall defect, lower sternal defect, diaphragmatic pericardial defect, anterior diaphragmatic defect, and intracardiac abnormalities. In 1958, Cantrell described what today is known as PC, characterized by a midline supraumbilical abdominal wall defect, a defect of the lower sternum, a deficiency of anterior diaphragm, a defect in the diaphragmatic pericardium, and congenital intracardiac defects (1). The hallmark of PC is an omphalocele associated with EC. Only a few patients with the full spectrum of the pentalogy have been described. It has a rare frequency of about 1/100,000 births (2). To our knowledge, approximately 90 cases of PC have been documented in published articles. We report two cases of PC diagnosed in early second trimester.

Case Presentations

Patient 1

A 22-years-old G3P1 pregnant at 15 weeks 1 day gestational age admitted to our fetal medicine centre. She reported taking no medications except folic acid, iron supplement. There was no family history of diabetes and her blood sugar was normal. On examination, her general and abdominal examination did not reveal any significant finding. The maternal serological tests to toxoplasmosis, rubella, and cytomegalovirus were negative. Prenatal ultrasound showed a foetus with bilateral hydrothorax, EC, and a large omphalocele with evisceration of the heart

(Figure 1). Pentalogy of Cantrell with thoracoabdominal EC was diagnosed, and the prognosis was discussed with the parents. Parents were offered termination of pregnancy. Pregnancy was terminated. The parents refused autopsy.

Patient 2

A 32-years-old G1P0 pregnant at 11 weeks 3 days gestational age admitted to our fetal medicine centre. There was no family history of diabetes and her blood sugar was normal. On examination, her general and abdominal examination did not reveal any significant finding. Ultrasound done at our fetal medicine centre revealed absent sternum, ectopia cordis, absent diaphragm, and omphalocele (Figure 2). Cantrell's pentalogy with thoracoabdominal EC was diagnosed. After discussing the diagnosis of pentalogy of Cantrell and related prognosis with the parents, the pregnancy was terminated at 12 weeks and 3 days of gestational age. The parents refused autopsy.

Discussion

EC is a challenging congenital anomaly because of its rarity and associated anomalies. The embryonic problem associated with EC lies in the abnormal migration of the splanchnic and somatic mesoderm which affects the development of the heart and the major vessels, with the premature breakage of the chorion or vitelline sac at around day 14-18 of gestation leading to a mid-line defect (3,4). EC has been a weak association noted with trisomy 18 and a few cases have been associated with other chromosomal abnormalities, such as triploidy and familial X-linked heritage (5,6). In complete EC (as

Received 16 April 2014, Revised 10 June 2014, Accepted 13 June 2014, Available online 20 July 2014

¹Kahramanmaraş Sutcu Imam University, Department of Obstetrics and Gynecology, Kahramanmaraş, Turkey. ²Sakarya University Research and Education Hospital, Department of Obstetrics and Gynecology, Sakarya, Turkey. ³Dicle University, Department of Radiology and Diagnostic Imaging, Diyarbakır, Turkey. ⁴Specialist of Obstetrics and Gynecology, Kahramanmaraş, Turkey.

*Corresponding Author: Mehmet Sühha Bostancı, Tell: + 90 532 514 03 63, E-mail: msuhha@gmail.com



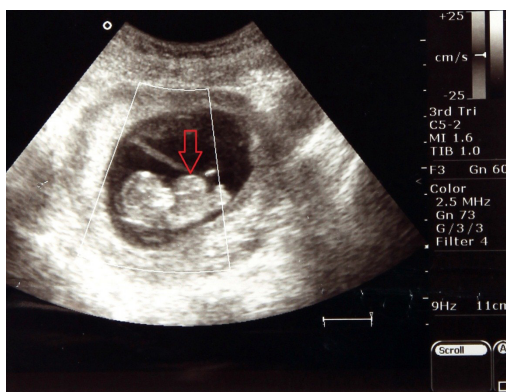


Figure 1. Prenatal ultrasound of first patient showed a fetus with bilateral hydrothorax, EC, and a large omphalocele with evisceration of the heart.

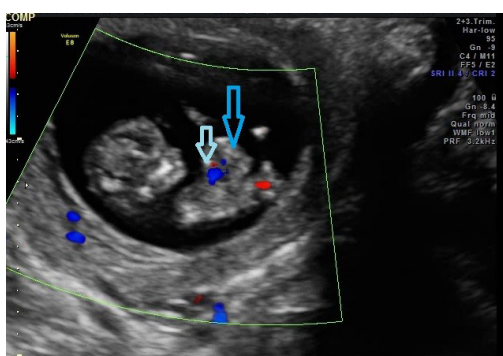


Figure 2. Prenatal ultrasound of second patient showed a foetus with bilateral hydrothorax, Ectopia Cordis, and a large omphalocele with evisceration of the heart.

in our cases) the heart is entirely outside the thoracic cavity with or without a pericardial covering. Complete EC is very rare and presents as a neonatal emergency. It is typically considered fatal with death occurring as a result of infection, cardiac failure, or hypoxemia (3). In partial EC, the heart can be seen to pulsate through the skin (7). Cervical (3%), cervicothoracic, thoracic (60%), thoracoabdominal (7%) and abdominal (30%) types of EC have been described (8). Early ultrasonographic diagnosis of the defect can often be made antenatally and is typically noted at the beginning of the second trimester (9).

In 1958, Cantrell reported a congenital syndrome characterized by: (a) a midline, supraumbilical abdominal wall defect; (b) a defect of the lower sternum; (c) a deficiency of the anterior diaphragm; (d) a defect in the diaphragmatic pericardium; and (e) congenital intracardiac defects (1). The syndrome occurs with various degrees of severity from incomplete to severe expression with involvement of other organ systems. The etiology of the pentalogy is not well established. A widely-accepted theory, which was proposed by Cantrell et al., stated that developmental failure of the mesoderm in early embryonic life between 14 and 18 days of gestation results in a failure in the development of the transverse septum of the diaphragm, and of the ventromedial migration of the paired mesodermal folds of the upper

abdomen (1). Toyama suggested further classification of the syndrome: Class 1, definite diagnosis with all 5 defects present; Class 2, probable diagnosis with 4 defects noted (including intracardiac and ventral abdominal wall abnormalities); and Class 3, incomplete expression (10). Children with Cantrell pentalogy who also have EC, pulmonary hypoplasia or complex heart disease, or who undergo surgery late, all face a poor prognosis (11). Renal involvement is a rarer additional defect in the pentalogy. Fernández et al. reported one patient having agenesis of the right kidney (12). The treatment of patients with pentalogy of Cantrell consists of corrective or palliative cardiovascular surgery, correction of ventral hernia and diaphragmatic defects and correction of the other associated anomalies. Only very few patients survive attempts at surgical repair, the main causes of death being tachyarrhythmias, bradycardia, low blood pressure, rupture of the diverticulum, and heart failure (13). With routine prenatal ultrasonography, the pentalogy of Cantrell usually can be diagnosed in the first trimester of pregnancy. Especially in a fetus with omphalocele, pentalogy of Cantrell should be ruled out. If intracardiac anomalies of incompatible with life are noted, this allows the physician and family to discuss the option of elective termination.

Conclusion

We report an extremely rare and lethal case of a complete EC associated with the Cantrell pentalogy. Pentalogy of Cantrell with ectopia cordis is a lethal anomaly and prompts medical and surgical interventions. The prognosis of having either of these syndromes is poor despite early detection and intervention. But early diagnosis is important about discuss the option of elective termination with the parents if intracardiac anomalies of incompatible with life are noted.

Ethical issues

Written informed consent was obtained from the patient for publication of this Case report and any accompanying images and the local ethics committee approved the study.

Conflict of interests

The authors declare no conflict of interests related to this manuscript.

Acknowledgments

We would like to thank all of our colleagues who helped us in this study.

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. McMahon CJ, Taylor MD, Cassady CI, Olutoye OO, Bezold LI. Diagnosis of pentalogy of Cantrell in the fetus using magnetic resonance imaging and

- ultrasound. *Pediatr Cardiol* 2007; 28:172–5.
3. Amato JJ, Douglas WI, Desai U, Burke S. Ectopia cordis. *Chest Surg Clin North Am* 2000; 10:297–316.
 4. Kaplan LC, Matsuoka R, Gilbert EF, Opitz JM, Kurnit DM. Ectopia cordis and cleft sternum: evidence of mechanical teratogenesis following rupture of the chorion or the yolk sac. *Am J Med Genet* 1985; 21:187–202.
 5. Soper SP, Roe LR, Hoyme HE, Clemmons JJ. Trisomy 18 with ectopia cordis, omphalocele and ventricular septal defect: case report. *Pediatr Pathol* 1986; 5:481–3.
 6. Fox JE, Gloster ES, Mirchandani R. Trisomy 18 with Cantrell pentalogy in a stillborn infant. *Am J Med Genet* 1988; 31:391–4.
 7. Kumar B, Sharma SB, Kandpal DK, Agrawal LD. Unusual variant of Cantrell's pentalogy? *Ann Thorac Med* 2008; 3:106–7.
 8. Mohta A, Bagga D, Anand R. Ectopia cordis: report of two cases. *Indian Heart J* 1993; 45: 133–4
 9. Harrison MR, Filly RA, Stanger P, de Lorimier AA. Prenatal diagnosis and management of omphalocele and ectopia cordis. *J Pediatr Surg* 1982; 17:64–6.
 10. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: a case report and review of the syndrome. *Pediatrics* 1972; 50:778–92.
 11. Theera T, Chanane W, Pannee S, Supreeya W. Prenatal sonographic diagnosis of ectopia cordis. *J Clin Ultrasound* 1999; 27:440–5.
 12. Fernández MS, López A, Vila JJ, Lluna J, Miranda J. Cantrell's pentalogy. Report of four cases and their management. *Pediatr Surg Int* 1997; 12:428–31.
 13. Morales J, Patel S, Duff J, Villarreal R, Simpson J. Ectopia cordis and other midline defects. *Ann Thorac Surg* 2000; 70:11–4.

Copyright © 2014 The Author(s); This is an open-access article distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/4.0>), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.