Derleme JCAM Review

Pentalogy of Cantrell

Cantrell Pentalojisi

Cantrell Pentalojisi / Pentalogy of Cantrell

Onur Isik[1], Muhammet Akyuz[1], Mehmet Fatih Ayik[1], Ozge Altun Koroglu[2], Arif Ruhi Ozyurek[3], Yuksel Atay[1]
[1]Department of Cardiovascular Surgery, [2]Department of Pediatrics, [3]Departments of Pediatric Cardiology,
Ege University, Faculty of Medicine, Izmir, Turkey

Özet

İlk olarak 1958 yılında Cantrell, Haller ve Ravitch tarafından tanımlanan Cantrell sendromu, ektopia kordis ile birlikte diyafragma, orta hat karın duvarı ve perikardı etkileyen nadir görülen bir anomalidir. Cantrell sendromu sıklıkla intrakardiyak defektler ile birliktelik göstermektedir. Kompleks kardiyak anomali prognozu belirlemede önemli bir role sahiptir. Çeşitli derecelerde birçok sistemi etkileyen bu sendromun tedavi süreci fetal tanı başlamaktadır. Bu aşama itibariyle, farklı disiplinlerden tarafından paylaşılan görüş ve deneyimler hasta yönetimini planlamasında önemlidir. Ancak, tüm bu çabalara rağmen, tedavi şansı bu sendromun ağır formları için düşüktür.

Anahtar Kelimeler

Cantrell Pentalojisi; Ektopia Kordis; Perikardial Defekt

Abstract

First described in 1958 by Cantrell, Haller and Ravitch, Cantrell's Syndrome is a rare anomaly which affects the ectopia cordis together with the diaphragm, the midline abdominal wall and the pericardium. Cantrell's Syndrome is often associated with intracardiac defects. Concomitant complex cardiac anomalies have an important role in determining the prognosis. The diagnosis of this syndrome, which affects numerous systems to varying extent, begins with fetal diagnosis. Starting from this stage, the views and experiences shared by different disciplines are important for the planning of disease management. However, despite the best efforts, the chance of effective treatment for the more severe forms of this disease is fairly low.

Keywords

Pentalogy of Cantrell; Ectopia Cordis; Pericardial Defect

DOI: 10.4328/JCAM.3144 Received: 08.12.2014 Accepted: 05.01.2015 Printed: 01.06.2014 J Clin Anal Med 2014;5(suppl 3): 427-9
Corresponding Author: Muhammet Akyuz, Ege Univ. Cardiovascular Surgery Dept. 35100, Bornova, İzmir, Turkey.
GSM: +905059437989 F.: +90 2323881115 E-Mail: drmak100@gmail.com

History and Definition

Cantrell's Syndrome was first defined by Cantrell, Haller, and Ravitch in 1958. The components of Cantrell's Syndrome are defined as: external herniation of the heart, pericardial diaphragmatic defects, lower sternal defects, anterior diaphragmatic defects, intracardiac defects, and malrotation of the heart [1]. The pathognomonic characteristic of this syndrome is the coexistence of omphalocele and ectopia cordis [2]. Identification of all components of the syndrome is rare in the patients [3].

Incidence and Typology

Cantrell's Syndrome has an incidence of 5.5-7.9 per 1 million live births [4]. The etiologic factor is not well established and familial occurrence of both isolated ectopia cordis and pentalogy of Cantrell has been reported. [5]. There is male dominance with a male to female ratio of 2.7:1 [6]. There are no publications indicating recurrence in the literature. Cases with Trisomy 13 and 18 are reported to have an increased incidence [7,8].

Ectopia cordis has cervical, cervicothoracic, thoracic, thoracoabdominal, and abdominal types. The most common form is the thoracic or thoracoabdominal type [9,10]. The abdominal wall defects include omphalocele, diastasis recti, umbilical hernia, or a combination of these defects. However, the most common form is the omphalocele [11]. In case an omphalocele is identified in the fetus, a detailed ultrasonographic examination should be performed, and the possibility of a Cantrell pentalogy it should be kept in mind [12].

Toyama et al. classified the Pentalogy of Cantrell in three separate classes. According to this classification, Class 1: definite diagnosis with all five defects present; Class 2: with four defects including intracardiac and ventral abdominal wall abnormalities present; and Class 3: incomplete expression with various combinations of defects present (but always including a sternal abnormality) [13].

Embryology

During the embryonic development; the sternum, abdominal wall, pericardium and part of the diaphragm arise from somatic mesoderm, while the myocardium arises from the splanchnic mesoderm.

The abnormal development of the septum transversum results in pericardial and diaphragmatic anomalies, while defects relating to the migration of mesodermal structures represents the possible cause of abdominal wall and sternum anomalies [5] While the specific pathogenesis of the disease has not yet been elucidated, it is known that the differentiation occurs at a time between the 14th and 18th day after conception. It has been suggested that, during this period, the lateral mesodermal folds' failure to migrate towards the midline results in abdominal and sternal defects, while problems relating to the development of the septum transversum results in pericardial and anterior diaphragm defects [14]. Cantrell's Syndrome patients without intracardiac defects can be explained by the teratogenicity developed after the differentiation of splanchnic mesoderm [1].

It is believed that the ventricular diverticulum arises from the abnormal fusion of cardiac loop to the yolk sac in the embryonic development. Therefore, the patients with diverticulum detected should be investigated for the other components of Cantrell's Syndrome [15].

Pathology

The malrotation of the heart is the failure to complete the levorotation due to the impaired differentiation and the heart being remained in the dextraposition. Cardiac herniation results from the absence of lower sternum, the absence of abdominal supra-umbilical structures, and the absence of a pericardial diaphragm [1].

The vast majority of the patients with Cantrell's Syndrome have intracardiac defects. The complicated cardiac anomaly has a significant role in the prognosis. There is a wide spectrum of intracardiac defects. The most common form is the ventricular septal defect (VSD), followed by atrial septal defect (ASD) [16]. The cases in the literature were reported to be associated with the left or right ventricular diverticulum, Tetralogy of Fallot (TOF), pulmonary stenosis or atresia, transposition of the great arteries (TGA), tricuspid atresia, truncus arteriosus, anomalous pulmonary venous return, and double outlet right ventricle [17].

The most common forms of the sternal malformations are the absence of the lower third portion, bifid sternum, absence of xiphoid process and short sternum. The most common forms of the pericardial and diaphragmatic abnormalities are the absence of pericardium and the absence of the anterior part of the diaphragm. The absent pericardium establishes the differential diagnosis of Cantrell's Syndrome with ectopia cordis [1].

The Pentalogy of Cantrell may be associated with various organ and system abnormalities [18]. The cases in the literature were reported to have many abnormalities involving various systems such as frontonasal dysplasia, cleft lip and palate, hydrocephalus, exencephalia, neural tube defect, polysplenia and gallbladder agenesis, imperforate anus, cystic hygroma, agenesis of tibia-radius, foot drop, hypodactylia and calcaneovalgus abnormalities [5,18].

The diagnosis of the Pentalogy of Cantrell may be established by fetal ultrasonography scheduled for the first trimester of the pregnancy. Attempts should be made exclude this syndrome in patients with omphalocele, diaphragmatic hernia, and pericardial defect accompanied by pericardial effusion detected in the prenatal assessment [19]. Fetal echocardiography and MRI will provide more detailed assessments as well as a more comprehensive examination of the cardiac structure, which is important for the pediatricians and surgeons providing care and treatment to the infant following delivery

[20,21]. Three-dimensional sonography, in particular, is being increasingly preferred and used by physicians for these patients. We have observed that this method does indeed provide more detailed assessments and information for such cases [19].

Ectopia cordis is not considered an indication for cesarean sections in the literature, and the patients are followed-up under normal obstetric indications. It is reported that there is not any prognostic difference between normal delivery and cesarean section in patients with ectopia cordis. However, some authors highlight the significance of sterility for the postpartum surgery and prefer cesarean section [21].

The team to plan the follow-up and treatment of the patient in the prenatal and neonatal periods should include a gynecologist, neonatologist, pediatric cardiologist, geneticist, pediatric surgeon, and congenital heart surgeon, and every intervention should be planned in a multidisciplinary approach.

Surgical Treatment

The treatment and the prognosis of the patients will depend on their type of EC, the size of their abdominal defect, and their

concomitant defects and disorders [5]. The surgical repair for the syndrome requires a complicated and generally a stepwise surgical intervention [22]. The surgical plan can be made as stepwise or a single operation depending on the patient clinical data, hemodynamic compatibility, thoracic cavity compatibility and the type of intracardiac defect. The primary objectives of the surgical treatment are to cover the heart with tissue, to place the heart into the thoracic cavity, to repair the intracardiac defects and the sternal-thoracic reconstruction [23].

The first choice for covering the heart with tissue should be the primary approximation [1,17]. The primary approximation is a good barrier for infections; however, it may cause a hemodynamic disturbance. A skin autograft, skin allograft, or a prosthetic material can be used for secondary closure. The advantage of using a skin autograft is the presence of a good barrier and the potential of the graft to grow with the patient [1,23,24]. The placement of the heart into the thoracic cavity generally results in low cardiac output and hypotension. The reasons of the low cardiac output include small thoracic cavity, folding of the great vessels, myocardial compression, and the disorientation of the apex. There have been several attempts to overcome these issues. These include opening and freeing both pleurae, separating the costae from the sternal connection (costal shift maneuver), orienting the apex to the left or if dextracardiac, to the right thorax by placing a traction suture on the apex and freeing and lowering the diaphragm [1,24,25].

The timing of the surgical repair for omphalocele depends on the sac content, width and the surface epithelialization. An urgent surgical intervention may not be required for the omphalocele if lined by epithelium well. However, the repair should be scheduled in a short time considering that a rupture will make the surgery difficult and complications may occur such as peritonitis and mediastinitis [26].

Sternal reconstruction may lead to hemodynamic instability and the repair may require tissues or prostheses. For this purpose, periosteal flap, pectoralis muscle flap, rectus abdominis muscle flap, and rib segment dissected from the separate sternum may be used [9,23].

The ventricular diverticulum may cause fatal complications such as cardiac rupture, tamponade, sudden death, endocarditis, embolism, cardiac failure and arrhythmia, and therefore, surgical resection is required even though the diverticulum is asymptomatic [26].

Cantrell's Syndrome has a poor prognosis. There is no consensus on the prognosis. Some authors consider the type of intracardiac defect as the most important factor determining the prognosis; whereas some authors argue that the class of the syndrome determines the prognosis [17].

Mainly due to complications such as bradycardia, tachyarrhythmias, diverticulum rupture, heart failure and low blood pressure, only a few children are able to survive following surgical repair [5,27]. However, it is known that a certain number of children whose thoracoabdominal defects are limited are able to survive into their adult years [28].

EC is associated with high mortality for children, with a survival rate of less than 5% [26]. When the very mild or incomplete forms of the Pentalogy of Cantrell are included in the series, the survival rate is reported to be lower than 40 % [13,29]. Most of the patients with a surgical success achieved are the cases without cardiac anomalies and with partial anterior wall defects [22,23,27].

The treatment process of this syndrome affecting many systems to the various extents starts from the fetal diagnosis. As of this stage, it is important to plan the patient management through the opinions and experiences shared by different multiple disciplines. However, despite all these efforts, the chance of treatment is low for the severe forms of this syndrome.

Competing interests

The authors declare that they have no competing interests.

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(5):602–14.
2. Sarkar P, Bastin J, Katoch D, Pal A. Pentalogy of Cantrell: diagnosisin the first trimester. J Obstet Gynaecol 2005;25(8):812–3.
3. Chen CP. Syndromes and Disorders Associated with Omphalocele (III): Single Gene Disorders, Neural Tube Defects, Diaphragmatic Defects and Others. Taiwan J Obstet Gynecol 2007;46(2):103–10.
4. Khoury MJ, Cordero JF, Rasmussen S. Ectopia cordis, midline defects, and chromosomal abnormalities. Am J Med Genet 1988;30(3):811-7.
5. Morales JM, Patel SG, Duff JA, Villareal RL, Simpson JW. Ectopia cordis and other midline defects. Ann Thorac Surg 2000;70(1):111 4.
6. Bittmann S, Ulus H, Springer A. Combined pentalogy of cantrell with tetralogy of fallot, and gallbladder agenesis, and polysplenia: a case report. J Pediatr Surg 2004;39(1):107-9.
7. Ghidini A, Sirtori M, Romero R, Hobbins JC. Prenatal diagnosis of pentalogy of Cantrell. J Ultrasound Med 1988;7(10):567-72.
8. Baker ME, Rosenberg ER, Trofatter KF, Imber MJ, Bowie JD. The utero findings in twins pentalogy of Cantrell. J Ultrasound Med 1984;3(11):525-7.
9. Dobell AR, William HB, Long RW. Staged repair of ectopia cordis. J Ped Surg 1982;17(4):353-8.
10. Kim KA, Vincent WR, Muenchow SK, Wells WJ, Downesy SE. Successful repair of ectopia cordis using alloplastic materials. Ann Plast Surg 1997;38(5):518-22.
11. Borade A, Palliana RR, Singh KD. Pentalogy of Cantrell. Pediatric Oncall 2011:8(6);39.
12. Karateke A, Kurt R, Celik C. A Rare Syndrome, Pentalogy of Cantrell: Case Report. 2014: DOI: 10.4328/JCAM.2823
13. 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(5):778-92.
14. Haynor DR, Shuman WP, Brewer DK: Imaging of fetal ectopia cordis: roles of sonography and computed tomography. J Ultrasound Med 1984:3(1):25-7.
15. Nanda S, Nanda S, Agarwal U, Sen J, Sangwan K. Cantrell Syndrome – report of two cases with one atypical variant Arch Gynecol Obstet 2003;268(4):331-2
16. Bryke CR, Breg WR: Pentalogy of Cantrell. from Buyse, M.L., Birth Defects Encyclopedia. Blackwell Scientific Publications 1990:1375-76.
17. Ferna´ndez IS, Lo´ pez A, Vila JJ, Lluna JM. Cantrell's pentalogy. Report of four cases and their management. Pediatr Surg Int 1997;12(5-6):428–31.
18. Carmi R, Boughman JA. Pentalogy of Cantrell and associated midline anomalies: a possible ventral midline develop- mental field. Am J Med Genet 1992;42(1):90–5
19. Rodgers EB, Monteagudo A, Santos R, Greco A, Timor Tritsch IE. Diagnosis of pentalogy of Cantrell using 2 and 3 dimensional sonography. J Ultrasound Med 2010;29(12):1825 8.
20. Doganay S, Kantarci M, Tanriverdi EC. Pentalogy of cantrell with craniorac¬hischisis: MRI and ultrasonography findings. Ultraschall Med 2008;29(5):278-80.
21. 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(3):172–5.
22. Sakasai Y, Thang BQ, Kanemoto S, Takahashi-Igari M, Togashi S, Kato H, et al. Staged repair of pentalogy of Cantrell with ectopia cordis and ventricular septal defect. J Card Surg 2012;27(3):390-2.
23. Zhang X, Xing Q, Sun J, Hou X, Kuang M, Zhang G. Surgical treatment and out¬comes of pentalogy of Cantrell in eight patients. J Pediatr Surg 2014;49(8):1335-40.
24. Lampert JA, Harmaty M, Thompson EC, Sett S, Koch RM. Chest wall reconstruction in thoracoabdominal ectopia cordis: using the pedicled osteomuscular latissimus dorsi composite flap. Ann Plast Surg 2010;65(5):485–9.
25. Chia HL, Rasheed MZ, Ong KK, Yeow VK. Repair of ectopia cordis using a resorbable poly-L-lactic-polyglycolic acid plate in a patient with pentalogy of Cantrell. J Pediatr Surg 2012;47(6):1-4
26. Halbertsma FJ, van OA, Staak F. Cardiac diverticulum and omphalocele: Cantrell's pentalogy or syndrome. Cardiol Young 2002;12(1):71-4.
27. Diaz JH. Perioperative management of neonatal ectopia cordis: report of three cases. Anesth Analg 1992;75(5):833-7.
28. Anderson HR. Abnormal positions and relationship of the heart. In: Anderson HR, Beaker JE, MaCartney JF, Rigby LM, Shinebourne AE, Tynan M, editors. Paediatric cardiology. Vol II. 2nd ed. London: Churchill-Livingstone; 2002. p. 1577–80.
29. O'Gorman CS, Tortoriello TA, McMahon CJ. Outcome of children with pentalogy of Cantrell following cardiac surgery. Pediatr Cardiol 2009;30(4):426–30

How to cite this article:
Isik O, Akyuz M, Ayik MF, Koroglu OA, Ozyurek AR, Atay Y. Pentalogy of Cantrell. J Clin Anal Med 2014;5(suppl 3): 427-9.