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Complete Pentalogy of Cantrell

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Abstract:

Pentology of Cantrell (PC) is a rare syndrome with unknown etiology presenting with pentad of midline, upper abdominal wall disorder, defect of lower sternum, anterior diaphragmatic defect, pericardial abnormality and congenital abnormalities of the heart as explained by Toyama. Genetic conditions associated with PC, include trisomy 18, 13 and Turner syndrome. Prevalence varies from 1/65,000 to 1/200,000 births. Mortality depends on the size of the abdominal wall defects, ectopia cordis and associated heart defects. Early antenatal diagnosis is very much important in these cases for better management. In our case report we are presenting a neonate born at 34 weeks of gestation to an elderly multipara mother with complete PC with severe form of omphelocele major, ectopia cardis and ventricular septal defect. She had low APGAR score at birth with severe respiratory distress, was managed conservatively and expired at 24 hours of life.

Key words: Ectopia Cordis, Heart Defects, Infants, Sternum, Teratogens.

Introduction

In 1958 Pentology of Cantrel (PC) was described by James R. Cantrell [1]. The cardinal features of the syndrome were anterior abdominal defect (omphalocele) in association with ectopia cordis. The full spectrum of the PC consist of the following 5 characteristics: (i) midline, upper abdominal wall disorder (e.g. omphalocele, gastroschisis); (ii) defect of the lower sternum (i.e. cleft sternum or absent sternum); (iii) anterior diaphragmatic defect (i.e. hypoplastic diaphragm, anterior diaphragmatic hernia); (iv) pericardial abnormality (e.g. ectopia cordis); and (v) congenital abnormalities of the heart (e.g. Tetralogy of fallot, ventricular or atrial septal defect) [2,3].

Complete PC is rare; the estimated prevalence varies from 1/65,000 to 1/200,000 births, affecting male and female fetuses at a ratio of 2:1. Affected females usually present more severe symptoms [4]. The risk of recurrence is quite small. There are no documented teratogens linked to this condition. Less than 90 cases have been reported in the literature.

Case Report

The patient product of non-consanguineous marriage was a female baby born by normal vaginal delivery at 34 weeks of gestation to a

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36 year old gravid 4 para 4 mother. The birth weight of a baby was 2600 grams, length 47 cm and head circumference 31 cm. No antenatal history suggestive of any infections, gestational diabetes, hypertension, radiation exposure could be elicited. Family history was not suggestive of any congenital diseases or unexplained sibling death. One antenatal ultrasound (USG) done day before delivery showed major omphelocele with possible diaphragmatic defect.

APGAR score was 3, 5, 6 at 1, 5 and 10 minutes respectively. Neonate was immediately shifted to neonatal intensive care unit and started on oxygen. Physical examination revealed major omphelocele with pulsatile mass situated at the epigastric part of omphelocele. This was apex cordis located outside the thoracic cavity recognized as thoraco-abdominal type of ectopia cordis (EC) - a severe condition, which was a part of PC [Fig.1]. Further examination showed absence of lower end of sternum, no obvious facial dimorphism, with normal genitalia, spine and extremities.

Baby was having ineffective respiration with low SPO_2, was subsequently put on CPAP. Baby continued to deteriorate; invasive ventilation, ionotropes and prophylactic antibiotics were started. Surgical opinion taken, but due to unstable condition of baby no surgical intervention could be done and conservative management was continued. Routine blood reports were normal. Infantogram showed collapsed lung with possibly downward displaced heart. Ultrasound reveled thoracoabdominal type of ectopic cardia with anterior defect in diaphragm. Echocardiography showed medium sized ventricular septal defect with left to right shunt. Parents were unable to afford karyotyping and TORCH panel. Newborn maintained on ventilator for 24 hours of life, however she expired at 24 hours of age.

Discussion

Toyama described PC in 3 classes: Class 1: Exact diagnosis with all five defects present. Class 2:

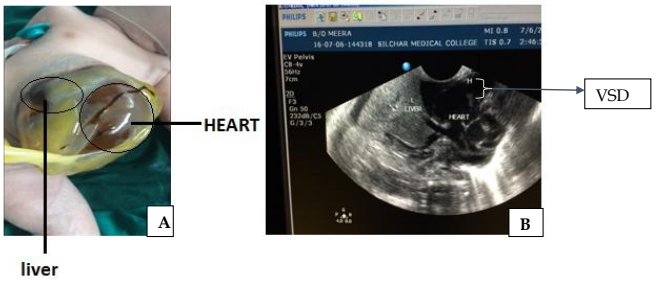

Fig.1(A): Showing major omphelocele and heart as content. **(B):** USG scan showing heart, as abdominal content, at the level of liver with medium sized VSD.

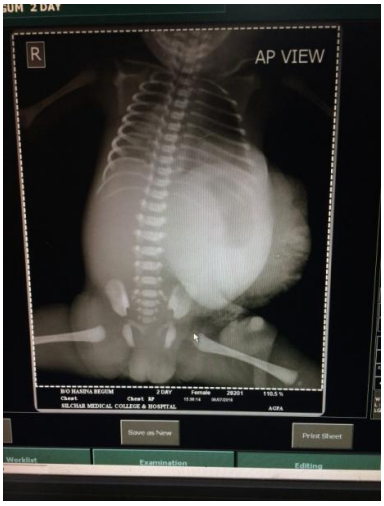

Fig.2: Infantogram of newborn showing collapsed lung with major omphelocele.

Probable diagnosis with four defects present and Class 3: Incomplete diagnosis, with combination of defects where sterna defects is always present [3]. In our case baby exhibited "Toyama Class-1 PC". The constellation of defects observed in PC is thought to result from the abnormalities in the differentiation of the intraembryonic mesoderm at approximately 14 to 18 days after conception. The diaphragmatic and pericardial defects result from abnormal development of the septum transversum whereas the sternum and abdominal wall defects

are probably related to impaired migration of mesodermal structures [5].

Many other associations of congenital anomalies have been found with PC. With prenatal USG, the PC usually can be diagnosed in the first trimester of pregnancy. The visualization of the fetal anomalies can be enhanced by the use of prenatal MRI. If the maximum diameter of the omphalocele is greater than 1 cm in the first trimester or if it persists beyond 14 weeks of gestation, this is regarded as pathological [6,7]. Association with aneuploidy, especially with trisomy 18, 13 and Turner syndrome, has been described and therefore, prenatal chromosomal studies is highly desirable [8,9]. The thoraco-abdominal syndrome including the PC is suggested to be an X-linked dominant disorder, with mapped Xq24-q27 gene [10]. In our case as mother is elderly multigravida chance of chromosomal disease was more.

Depending on the location of the protruding heart, and extend of the abdominal wall defect, ectopia cordis can be divided into cervical, cervicothoracic, thoracic, thoraco-abdominal and abdominal typologies. Here the body wall defect usually extends into the umbilicus. The patient presented in our report had the most common thoraco-abdominal typology. Treatment of PC is challenging and outcome depends on the size of the abdominal wall defects, ectopia cordis and associated heart defects. The treatment is mainly surgical.

Conclusion

In every prenatally diagnosed omphalocele, PC should be excluded. The preferred approach to management still needs to be established in resourced constrained countries like ours.

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References

1. Cantrell JR, Heller RJA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet. 1958;107(5):602-614.
2. 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;50:778-779.
3. Chen CP. Syndromes and disorders associated with omphalocele (II): OEIS complex and pentalogy of Cantrell. Taiwan J Obstet Gynecol. 2007;46:103-110.
4. Zidere V, Allam LD. Changing findings in pentalogy of Cantrell in fetal life. Ultrasound Obstet Gynecol. 2008;32:835-837.
5. Morales JM, Patel SG, Duff JA, Villareal RL, Simpson JW. Ectopia cordis and other midline defects. Ann Thorac Surg. 2000;70:111-114.
6. Chen CP. Syndromes and disorders associated with omphalocele (II): OEIS complex and pentalogy of Cantrell. Taiwan J Obstet Gynecol. 2007;46:103-110.
7. Desselle D, Fellow R, Herve P, Toutain A, Lardy H, *et al*. Pentalogy of Cantrell: Sonographic assessment. J Clin Ultrasound. 2007;35:216-220.
8. Kim KA, Vincent WR, Muenchow SK, Wells WJ, Downesy SE. Successful repair of ectopia cordis using alloplastic materials. Ann Plast Surg. 1997;38:518-522.
9. Cyr DR, Mack LA, Schoenecker SA, Patten RM, Shepard TH, Shuman WP, *et al*. Bowel migration in the normal foetus: US detection. Radiology.1986;161:119-121.
10. Cullinan JA, Nyberg DA. Fetal abdominal wall defects. *In*: Rumack CM, Wilson SR, Charboneau JW, eds. Diagnostic Ultrasound. 2nd ed. St. Louis, MO: Mosby-Year Book, Inc. 1998:1167-1169.