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Case Report

Pentalogy of Cantrell: a Case Report

La pentalogie de Cantrell: à propos d'un cas

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ABSTRACT

The Pentalogy of Cantrell is a congenital anomaly characterized by the lower sternum, a midline supra umbilical thoracoabdominal wall defect, a deficiency of the diaphragmatic pericardium, a deficiency of the anterior diaphragm and congenital cardiac anomalies. We report a case of newborn who presented at birth an ectopia cordis associated with an omphalocele type 2 according to the Aiteken classification. After investigations the diagnosis of pentalogy of cantrell was made. The outcome of newborn is unfavorable because he died after 12 hours before surgery. The prevention of Pentalogy of Cantrell goes through early antenatal diagnosis and appropriate counseling to decide the outcome of the pregnancy. The prognosis after birth is poor despite early surgical management by trained teams.

RÉSUMÉ

La pentalogie de Cantrell est une anomalie congénitale caractérisée par le sternum inférieur, une anomalie de la paroi thoraco-abdominale supra-ombilicale médiane, une déficience du péricarde diaphragmatique, une déficience du diaphragme antérieur et des anomalies cardiaques congénitales. Nous rapportons un cas de nouveau-né qui a présenté à la naissance une ectopie cordienne associée à une omphalocèle de type 2 selon la classification d'Aiteken. Après investigations, le diagnostic de pentalogie du cantrell a été posé. L'issue du nouveau-né est défavorable car il est décédé après 12 heures avant l'opération. La prévention de la pentalogie de Cantrell passe par un diagnostic prénatal précoce et des conseils appropriés pour décider de l'issue de la grossesse. Le pronostic après la naissance est sombre malgré une prise en charge chirurgicale précoce par des équipes compétentes.

INTRODUCTION

Pentalogy of Cantrell (PC) is a very rare pathology. Omphalocele and ectopias cordis are the two mains defects that characterizes it. Cantrell is the first author who describe this syndrome in 1958. It is characterized by the lower sternum, a midline supraumbilical thoracoabdominal wall defect, a deficiency of the diaphragmatic pericardium, a deficiency of the anterior diaphragm and congenital cardiac anomalies [1]. A classification of PC is proposed by Toyama : class 1 : defnite diagnosis with all 5 defects; class 2 : probable diagnosis with 4 defects (including intracardiac and ventral abdominal wall abnormalities); and class 3 : incomplete expression [2]. We report the first case of PC at the neonatology unit of Issaka Gazoby maternity.

OBSERVATION

A 25 years old woman at 9 weeks gestation was referred in our department for labor and intrapartum asphyxia. She was gravida 1 para 1 and pregnancy was poorly monitoring. No obstetric ultrasound was performed. Its a cosanguinous couple and no history of congenital anomaly was reported in family. She benefied a cesarean section.

After the birth, the newborn weighted 3100 grams with a length of 52 centimetres and circumference head of 34 centimetres. Apgar's score was 4 then 6 at 5 and 10 mins and had respiratory distress with 76% of saturation. The pulse rate was 121 pulse. He presented a large anterior thoraco abdominal wall defect (omphalocele), at the top of this defect a pulsatile mass was seen and in auscultation we heard a 3/6 graduated systolic murmur(figure 1). We concluded with an ectopias cordis associated with an omphalocele type 2 according to the Aiteken classification. He presented also a trisomic facies with low implant of ears

and weebed neck. An abdomino-pelvic and cardiac ultrasounds showed normal abdominal visera and cardiac malformations such as patent ductus arteriosus and ventricular septal defect.
The diagnosis of Pentalogy of Cantrell was made. The blood investigations were normal. The karyotype was no performed.
Management included oxygenotherapy, resuscitation and recovered mass with saline soaked gauze. He was transferred in pediatric surgery unit and died 12 hours before surgery. The autopsy was not performed.

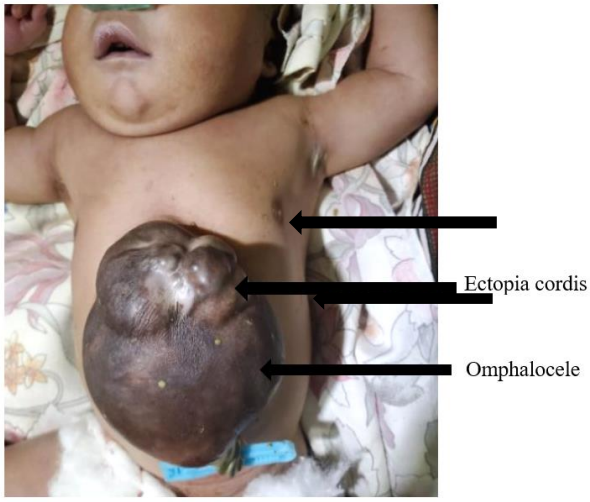

Figure 1: Newborn with ectopia cordis and omphalocele

DISCUSSION

The PC is an extremly rare pathology. William in his review noted 250 cases to date with an incidence of 1 case per 65,000 to 200,000 live births [3]. Most documented cases occurred in the United States and Europe [4]. In Africa, Bagabir report the fist case of complete PC in saudi arabia [5]. In subsaharian countries, no study about this pathology has been reported but a rare case of ectopia cordis associated with anothers abnormalities have been notified [6, 7]. The aetiopathogeny of Pc is unknow. Most theory is avanced these last years but the cantrell theory remains the predominant. Cantrell avanced that an embryologic developmental failure of a segment of the lateral mesoderm around gestational age 14–18 days; consequently the transverse septum of the diaphragm does not develop, and the paired mesodermal folds of the upper abdomen do not migrate ventromedially. Organs may eviscerate through the resulting sternal and abdominal wall defects [1].
Although origin of PC is unknow, many cases remains sporadic and Carmi et al reported in family cases, the implication of gene linked to the X chromosome in the Xq25-26.1 region [8].
Furthermore, the alteration conversion of retinaldehyde to retinoic acid secondary to ALDH1A 2 dysfunction can contribute to the developmental failure in Cantrell's pentalogy. Its due to a novel gain in copy number on an interval of 15q21.3 in chromosome. Retinoid acid arise from vit A which is necessary for organogenesis during embryonic development [9].
The PC is frequently associated with other anomalies such as central nervous system anomalies, cleft lip and palate, limb defects [11, 12]. Association with anomaly genetic such as trisomy 13 and 18 is reported [12]. In our case, the newborn is from cosanginous marriage and he present short neck and low implant of ears as in trisomique syndrom but caryotype is not performed because of the limited ressources.
The management of PC is complex. Its depend to the the type of EC, the size of the defect in the abdominal wall, and the abnormalities in the heart. The prognosis of PC is poor despite of surgery.
The surgical repair consist to : (1) correcting cardiac malformations, (2) restoring cardiac position and anatomy, and (3) repairing the thoraco-abdominal wall and diaphragmatic defects [3]. Nevertheless, after surgery multiples complications may occur such as cardiac and respiratory insufficiency because of the hypoplasia of the rib cage and inhability to enclose the ectopic heart [10]. Hoorn demontrate that mortality remains higher despite of the management and may increase when complete form and associated others anomalies were present [13].
In contrast, the surviving patients with PC and intracardiac anomalies do not increase the prognosis [10, 11]. Finally, when the diagnosis of PC is made, a multidiciplinary must be held including obstetrician, neonatologist, pediatric surgeon, pediatric cardologist and geneticist to choose the best approch during and after delivery.

CONCLUSION

Pentalogy of Cantrell is a rare pathology. The prognosis is poor and depends upon the promptitude of surgical intervention. When the diagnosis is made during the pregnancy, an appropriate prenatal counseling should be done to the parent to choosing or not continue pregnancy. The management of PC is so complex that require an effective team of specialist.

Conflict interst

No.

Contributions of authors

All authors approved the final version of the manuscript.

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
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;50:778-92.
3. Williams AP, Marayati R, Beierle EA. Pentalogy of Cantrell . Semin Pediatr Surg. 2019;28(2): 106–110
4. Jnah AJ, Newberry DM, England A. Pentalogy of Cantrell: Case Report With Review of the Literature. Adv Neonatal Care. 2015;15(4):261–8
5. Bagabir HA, Azhar AS. Pentalogy of Cantrell: frst case reported in Saudi Arabia. Ann Saudi Med 2014; 34(1): 75-77

6. Oumarou H, Amadou MI, Harissou A et al. Ectopia Cordis: A Case Report and Review of Literature. Journal of Case Reports 2017;7(2):143-145
7. Chishugi JB, J Franke TJ. Thoraco-abdominal Ectopia Cordis in Southwest Cameroon. Pan African Medical Journal. 2014; 18:124
8. carmi r, Meizner I, Katz M. Familial congenital diaphragmatic defect and associated midline anomalies: Further evidence for an X-linked midline gene? Am J Med genet. 1990;36:313-5.
9. Steiner MB, Vengoechea J, Collins RT. Duplication of the ALDH1A2 gene in association with pentalogy of Cantrell: a case report. Journal of Medical Case Reports 2013;7:287
10. Nayak S, Dash SP, Khatua M. Fetal Anomaly: Pentalogy of Cantrell. IOSR Journal of Dental and Medical Sciences. 2015;14:52-55
11. Marginean C, Cristina Oana Marginean CO, Gozar L et al. Cantrell Syndrome—A Rare Complex Congenital Anomaly: A Case Report and Literature Review. Front. Pediatr. 2018;6:201
12. Yi-Jen H, Fong-Lin C, Yan-Yan N et al. Trisomy 18 Syndrome with Incomplete Cantrell Syndrome. Pediatr Neonatol 2008;49(3):84−87
13. Hoorn J , Moonen R , Clément J. R. Huysentruyt C et al. Pentalogy of Cantrell: two patients and a review to determine prognostic factors for optimal approach. Eur J Pediatr (2008) 167:29–35