PRENATAL DIAGNOSIS OF ECTOPIA CORDIS : CASE REPORT

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DIAGNOSTIC PRENATAL DE L'ECTOPIE CARDIAQUE : A propos d'une observation.

LA TUNISIE MEDICALE - 2008 ; Vol 86 (n°02) : 171 - 173

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RÉSUMÉ

L'ectopie cardiaque est une malformation rare et impressionnante se présentant comme une lésion isolée ou faisant partie de la pentalogie de Cantrell. Elle est définie comme une anomalie dans laquelle le cœur fœtal se trouve à l'extérieur de la cage thoracique.

Le but du travail est de rapporter les principaux éléments de diagnostic prénatal et de prise en charge de l''ectopia cordi''.

Observation : Nous rapportons un cas d'ectopie cardiaque diagnostiqué à 19 semaines d'aménorrhée lors d'une échographie bidimensionnelle. Des anomalies multiples congénitales ont été trouvées. La plus importante était la présence d'un défect ventral et thoracoabdominal avec une éviscération du cœur, du foie, de l'estomac et des intestins. L'examen autopsique a confirmé les constatations échographiques. La décision d'interruption médicale de la grossesse a été prise

SUMMARY

Ectopia cordis is a rare and impressive malformation presenting as an isolated lesion or as part of the Cantrell's pentology syndrom. It is defined as an anomaly in which the fetal heart lies outside the thoracic cavity.

The aim of the study is to report the prenatal diagnostic features and management of ectopia cordis.

Case report : We report a prenatal diagnosis case of ectopia cordis using two-dimensional ultrasound at 19 weeks of gestation. Multiple congenital anomalies were found. The most important one was the presence of a ventral thoraco-abdominal wall defect with exstrophy of the heart, liver, stomach and intestines. Histopathological examination confirmed the ultrasound findings. Due to severity of the malformations, termination of pregnancy was made.

MOTS-CLÉS

Ectopie cardiaque, échographie, diagnostic prénatal.

KEY-WORDS

Ectopia cordis, ultrasonography, Extrathoracic heart, prenatal diagnosis.

التشخيص قبل الولادة الأنتباذ القلبي . دراسة حول حالة واحدة

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الإنتباذ القلبي هو تشوه نادر يتمثل في تواجد قلب الجنين خارج القفص الصدري . الهدف من هذه الدراسة هو آستعراض أهم العناصر التي تسمح بالتشخيص قبل الولادة و الإحاطة بهذه الإصابة نستعرض حالة واحدة وقع تشخيصها في الأسبوع التاسع عشر للحمل أثناء إجراء تخطيط بالصدى . لاحظنا وجود العديد من التشوهات الخلقية أهمها أنتقاف القلب و الكبد و المعدة و الأمعاء . أكد التشريح نتائج التخطيط بالصدى و أتخذ قرار التوقيف الطبي للحمل

الكلمات الأساسية : أنتباذ قلبي – تخطيط بالصدى – تشخيص قبل الولادة

Ectopia cordis is defined as an anomaly in which the fetal heart lies outside the thoracic cavity. It is a rare and impressive congenital abnormality with an incidence of 5.5 to 7.9 per 1 million live births (1). It was first observed 5000 years ago (2) .To date, the cause of ectopia cordis is unknown, and most cases are sporadic. It may occur as an isolated malformation or associated with a larger category of ventral body wall defects that affect the thorax, abdomen or both.The aim of the study is to report the prenatal diagnostic features and management of ectopia cordis.

CASE REPORT

A 23- year-old woman, gravid 1, para 0, was referred after a routine dating ultrasound which revealed a ventral wall defect. The patient had no prior prenatal care. The family history was negative for congenital or genetic abnormalities and the patient denied exposure to drugs or toxins.Ultrasonographic examination revealed a single fetus with measurements consistant with 19 weeks gestational age. Multiple congenital anomalies were found. The most important one was the presence of a ventral thoraco-abdominal wall defect with exstrophy of the heart, liver, stomach and intestines.No craniofacial or intracardiac abnormalities were detected. (Figures 1,2,3, 4)The umbilical cord and placenta were normal in appearance and location.Diagnosis of ectopia cordis associated with ventral body wall defect was made.Genetic amniocentesis was performed and revealed a 46 XX, karyotype. Owing the poor prognosis, the patient elected to terminate the pregnancy following counseling.Under prostaglandins induction,the fetus and the placenta were vaginally expelled two days later.

Figure 1 : Sonogram showing the fetal heart lying completely outside the chest cavity (circle)cavity (circle)
a : Axial view. b : Sagittal view

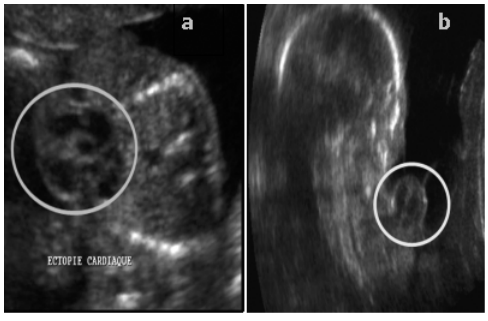


Figure n°2 : Ventral thoraco-abdominal wall defect with exstrophy of the heart (a)liver (b), intestines (c) and stomach (d).1: Sagittal view. 2: Axial view.

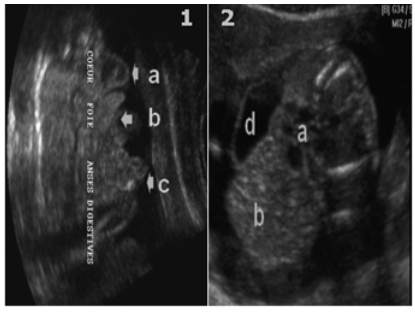
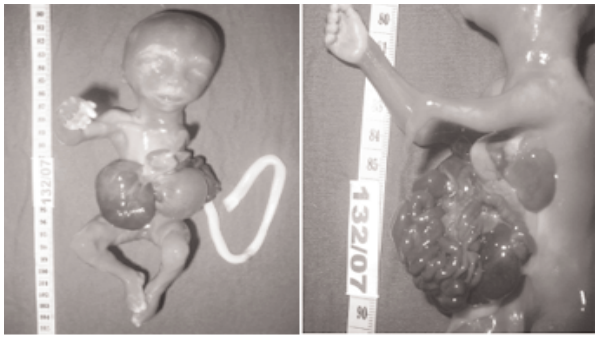


Figure n° 3 : Photographs of the abortus demonstrate: the midline thoraco-abdominal defect (accolade) and an abnormal position of the left arm with partial absence of the forearm and hand (clear arrows).Evisceration of heart (a) , left lung (b), stomach (c) , small and large intestines (d) and liver (e).



The body weight of the female abortus was 320 g. The measurements were compatible with a fetus at 19-20 weeks of gestation.The autopsy confirmed thoraco-abdominal wall defect with evisceration of the heart, left lung, liver stomach, small and large intestines and of the lower part of the esophagus. (Figure 5).
We also found an abnormal position of the left arm with partial absence of the forearm and hand.
Furthermore, hypertelorism, macrocephaly and retrognathy were noted.The intracardiac structures and outflow tracts were normal.The umbilical cord was unremarkable.

DISCUSSION

Ectopia cordis is a rare and impressive congenital malformation which was observed thousands years ago (2). Abbot was the first author who defined the '' extra thoracic heart'' (3). Subsequently, the term "ectopia cordis" has been used to describe all anomalies in which the heart was not located within the thorax (4).Clinically, ectopia cordis has been classified into four types: cervical, thoracic, abdominal and thoracoabdominal whether the heart is respectively in the neck, anterior to the sternum, within the abdomen or between the thorax and the abdomen.(4,5) Kabbani and al. classified this malformation into five types adding the cervicothoracic type (6).The two most common forms of ectopia cordis are the thoracic and the thoracoabdominal type (7,8). The latter can be part of a distinct syndrome known as Cantrell's penology which has a variable expression. (7,9,10,11,12)Embryologically, in about the third week, early disturbance in the formation of the cephalic fold will result in defective formation of the thoracic and epigastric walls, finally resulting in ectopia cordis with anterior defect of the sternum and diaphragm and an omphalocele (13, 14, 15, 16). Until 1981, ectopia cordis was diagnosed only in the delivery room (13)and prior to 1988, prenatal diagnosis of this malformation before the third trimester of gestation had never been reported (11). With the development of ultrasound equipment, diagnosis is possible at a very early stage of gestation. Prenatal diagnosis in the first trimester has been has

been reported (17,18,19). In our case, the diagnosis has been made in the second trimester by two-dimensional ultrasonography. The key features in our diagnosis were a thoracoabdominal wall defect with an extrathoracic pulsating mass containing Doppler waveforms typical of intracardiac flow. According to M Burn (20), the prenatal diagnosis is easily made by visualizing the evisceration of the heart through a parietal defect. The heart can be partially or completely outside the thorax. Pericardic effusion and omphalocele would be elements of the positive elements (20,21). Liamg and al (11) repoted that 3-dimensional reconstructed images will provide a more complete view of the anomaly prenatally.It is known that a few cases of ectopia cordis are associated with chromosomal abnormalities such as triploidy or trisomy, and cases of a familial X-linked inheritance have been reported. (1,21,22,23,24) The risk of recurrence is generally considered minimal. (25)The prognosis of ectopia cordis is generally poor, with death commonly occurring before or immediately after birth, usually owing to associated malformations. Infants with no or mild associated congenital heart disease may survive after successful surgical intervention (6,19).The procedures are complex. Primary repair in the neonatal period is the best type of management (26).

CONCLUSION

Ectopia cordis is a rare congenital malformation with a poor prognosis. Ultrasonography is of great value in the prenatal assessment. The ectopia should be precisely localized and its classification accurately determined. Obstetrical management should include a careful search for associated anomalies, especially cardiac, and assessment of fetal karyotype. Pregnancy termination prior to viability and a non aggressive management in the third trimester should be considered and discussed with the parents.

REFERENCES

1 Hornberger LK, Colan SD, Lock JE, et al. Outcome of patients with ectopia cordis and significant intracardiac defects. Circulation 1996 ; 94 : 32-7.

2 Taussing HB. World survey of the common cardiac malformation: developmental error of genetic variant . Am J Cardiol 1982 ; 50 : 544-59.

3 Abbott FC. Congenital abnormality of sternum and diaphragm; protrusion of the heart in the epigastric region. Tram Pathol Soc London 1898 ; 69 : 57-59

4 Byron F. Ectopia cordis: report of a case with attempted operative correction.J Thorac Surg 1949 ; 17:717-722.

5 Leca F, Thibert N, Khoury W, Fermont L, Laborde F, Dumez Y. Extrathoracic heart (ectopia cordis) : report of two cases and review of literature . Int J Cardiol 1989 ; 22 : 221-8.

6 Kabbani M S, Rasheed K,Mallick MS,Abu-Hassan H, Al-Youssef S. Thoraco-abdominal ectopia cordis. Case report. Annals of Saudi Medicine 2002 ; 22 : 5-6.

7 Amato JJ, Zelen J, Talwakar NG. Single stage repair of thoracic ectopia cordis. Ann Thorac Surg 1995;59:518-20

8 Hochberg J, Ardenghy MF, Gustafon RA, Murray GF. Repair of thoracoabdominal ectopia cordis with mucocutanuous flaps and intraoperative tissue expansion. Plast Reconstr Surg 1995 ; 95 : 148-51.

9 Cantrell JR, Haller JA and Ravitch MM. A syndrome of congenital defects involving the abdominal wall,sternum, diaphragm, pericardium and heart. Surg Gynecol Obstet 1980 ; 107 : 602-14

10 Abdallah HL, Marka LA, Balsara RK Davis DA, Russo PA. Staged repair of pentalogy of Cantrell with tetralogy of Fallot. Ann Thorac Surg 1993 ; 56 : 979-80.

11 Liang RI, Huang SE, Chang FM. Prenatal diagnosis of ectopia cordis at 10 weeks of gestation using two-dimensional and three dimensional ultrasonography. Ultrasound Obstet Gynecol 1997; 10: 137-139.

12 Desselle C, Herve P, Toutain A, Lardy H, Sembely C, Perrotin F. Pentalogy of Cantrell : Sonographic assessment. J Clin Ultrasound 2007; 35 : 216-20.

13 Achron R, Schimmel M, Farber B, Glaser J. Prenatal sonographic diagnosis and perinatal management of ectopia cordis. Ultrasound Obstet Gynecol 1991 ; 1 :431-434.

14 Firmin RK., Fragomeni LS ,Lennox SC. Complete cleft sternum. Thorax 1980 ; 35 : 303-306.

15 Patten BM. Human embryology. 3rd Edn. McGraw-Hill Book Company, New York,1969.

16 Spitz L, Bloom KR. Milner S, Levin SE. Combined anterior abdominal wall defects : a report of five cases and their management. J pediatr Surg 1975 ; 10 : 491-497.

17 Sepulveda W, Weiner E, Bower S,Flack NJ, Bennet PR, Fish NN. Ectopia cordis in a triploid fetus : first trimester diagnosis using transvaginale color Doppler ultrasonography and chorionic villus sampling. J Clin Ultrasound 1994 ; 22 : 573-5.

18 Bennett TL, Burlbaw J, Drake CK, Finley BE. Diagnosis of ectopia cordis at 12 weeks gestation using transabdominal ultrasonography with color flow Doppler. J ultrasound Med 1991 ; 10 : 695-6.

19 Thongsong T, Wanapirak C, Sirivatanapa P. Prenatal sonographic diagnosis of ectopia cordis. J Clin Ultrasound 1999 ; 27 : 440-445.

20 Burn M, Maugey Laulem B, Rauch Chabrol F. Diagnostic échographique anténatal des malformations de la paroi antérieure du fœtus. J Radiol 1998 ; 79 : 1461-8.

21 Ben kahfallah A, Annabi N, Ousji M, Hadrich M, Najai A. Ectopia Cordis thoracique associée à une tétralogie de Fallot. Tunis Med 2003 ; 81 : 661-665

22 Soper SP, Roe LR, Hoyme HE et al. Trisomy 18 with ectopia cordis, omphalocele and ventricular septal defect : case report. Pediatr Pathol 1986 ; 5 : 481.

23 Bick D, Markowitz RI, Horwich A. Trisomy 18 associated with ectopia cordis and occipital meningocele. Am J Med Genet 1988 ; 30 : 805.

24 Fox JE, Gloster ES, Mirchandani R. Trisomy 18 with Cantrell pentalogy in a stillborn infant. Am J Med Genet 1988 ; 31 : 391.

25 Wen Shaw S, Jen Cheng P, Yen Chueh H, Dyh Chang S, Kuei Soong Y. Ectopia cordis in a fetus with trisomy 18. J Clin Ultrasound 2006 ; 34 : 95-98.

26 Kim KA, Vincent WR, Muenchow SK et al. Successful repair of ectopia cordis using alloplastic materials. Ann Plast Surg 1997 ; 38 : 518.