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Cantrell Pentalogy Associating Acrania and Iniencephaly: Antenatal Ultrasound Diagnosis at the CHU Pr BSS of Kati

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Abstract **Case Report**

Introduction**:** Cantrell's pentalogy is a rare malformation, which associates 5 cardinal malformations: supraumbilical hernia of the abdominal wall, defect of the anterior part of the diaphragm and diaphragmatic pericardium, abnormality of the lower part of the sternum, heart defects. Ultrasound is essential in antenatal diagnosis. We report a case of polymaformative syndrome, including Cantrell pentalogy, acrania and iniencephaly. ***Observations*****:** Mrs. BS, 24 years old, housewife, resident in rural areas, 3rd pregnancy, two live children, no medical-surgical history or known fetal malformation, no notion of teratogenic drug intake. Coming for his first ultrasound, which, performed by two radiologists concluded to a polyformative syndrome associating cantrell pentalogy, acrânie and iniencephaly: by the presence of a defect of closure of the anterior thoraco-abdominal wall resulting in an evisceration of the intra-abdominal organs, a cardiac malformation, the absence of formation of the bones of the skull, and absence of cervico-occipital bone formation with neck shortening and hyper extension of the head. The pregnancy was estimated at 27 weeks, the couple was informed of the fate of the future baby and the pregnancy intervention was proposed. After their agreement the labor was triggered, after 8 hours, she expelled a live newborn male, polymalformed confirming the ultrasound diagnosis, weighing 1900grams and lived only thirty minutes after birth. ***Conclusion*****:** The polymalformative syndrome associating cantrell pentalogy, acrania and iniencepahlia is exceptional. Ultrasound is essential in diagnosis. Termination of pregnancy is the rule.
Keywords: Cantrell pentalogy, acrânie, iniencephaly-ultrasound-CHU Kati.

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INTRODUCTION

Cantrell's pentalogy is a rare malformation, which associates 5 cardinal malformations: supraumbilical hernia of the abdominal wall, defect of the anterior part of the diaphragm and diaphragmatic pericardium, abnormality of the lower part of the sternum, heart defects. It was first described by Cantrell in 1958 [1, 2]. This pathology is an extremely rare birth defect (5.5 to 7.9 per million live births) [3]. Other rare malformations include acrania and inicencephaly. Acrânie is a rare and lethal malformation, characterized by a defect in the development of the flat bones of the scalp [4]. Iniencephaly is a major extension of the head, continuity between the head and trunk, a shortening of the cervical spine and cervical vertebrae with non-closure of the posterior arch [5]. Over the years, as part of the prenatal check-up, fetal ultrasound has gradually become the obstetrician's most effective diagnostic tool [6]; Because it is a less expensive, non- traumatic and essential examination in the follow-up assessment of pregnancies and the antenatal diagnosis of fetal malformations.

OBSERVATION

Mrs. BS, 24 years old, housewife, resident in a rural area, she is on her 3rd pregnancy, with two live children. It has no known medical-surgical history or fetal malformation. There is no notion of teratogenic drug intake during this ongoing pregnancy, nor of consanguineous marriage. She was admitted by a midwife for her first ultrasound for this pregnancy, which was performed by two radiologists. Morphological exploration has highlighted a polyformative syndrome associating cantrell pentalogy, acrânie and iniencephaly: by the presence of a defect of closure of the anterior thoraco-abdominal wall leading to an evisceration of the intra-abdominal organs, a cardiac malformation. There was no formation of the bones of the skull but the brain was intact, and the absence of cervico-occipital bone formation with shortening of the neck and hyper extension of the head. The age of pregnancy was estimated at 27 weeks. The couple was informed of the fate of the unborn baby and medical termination of the pregnancy was proposed. After their agreement, labor was triggered, after 8 hours she expelled a live male newborn, polymalformed confirming the ultrasound diagnosis, the other parts of the body were unremarkable. The newborn weighed 1900grams and lived only about thirty minutes after birth.

Figures 1 and 2 are iconographic illustrations testifying to the polymalformative syndrome.

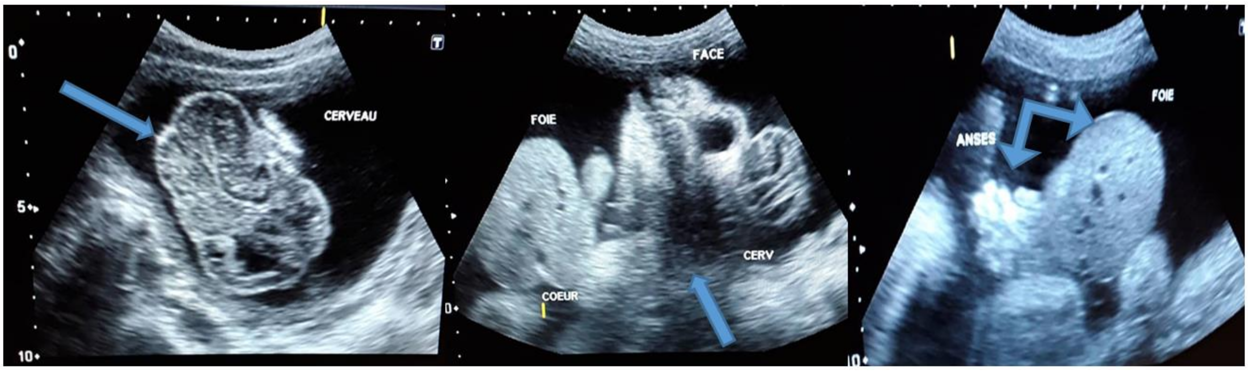

Figure 1: Morphological ultrasound in mode B objectifying the absence of the bones of the skull, the shortening of the cervical spine and evisceration of the digestive loops (arrows)

Figure 2: Photos of the newborn, confirming the ultrasound diagnosis

DISCUSSION

About Cantrell Pentalogy

Cantrell pentalogy is an extremely rare birth defect, first described by Cantrell in 1958 [1].

The cause of Cantrell's pentalogy is unclear and it is not known whether it represents an extreme spectrum of midline malformations, as it shares some characteristics with a midline defect and coexists with other midline malformations [7]. The diagnostic criteria for ultrasound are the same as those for in vitro diagnosis. However, the unique presence of a defect of the anterior abdominal wall cannot be considered as the pentalogy of Cantrell; It is taken as omphalocele or laparoschisis. Note that ultrasound has a good sensitivity and specificity in general; But the sensitivity of antenatal ultrasound in the diagnosis of heart defects is apparently quite disappointing, of the order of 60% for major heart disease [8]. Our observation was a case of complete Cantrell pentalogy through the presence of 5 malformation entities with other malformations not part of this polymalformative syndrome. A complete Cantrell pentalogy case was brought by Sidibe N *et al.*, [9] in 2021. However, the difference between this observation and ours, was that the presence of other malformations, which were absent in Sidibe N *et al.*, Moreover, in Cameroon in 2010, a case of pentalogy was described that presented four of the five elements that define the complete form of this anomaly [10]. This pathology shares some characteristics with a midline defect and can coexist with other midline malformations [7, 9]. In addition to the classic abnormalities described by Cantrell *et al.*, a few cases have been reported with the coexistence of other syndromes such as Edwars syndrome and Goltz-Gorlin. Similarly, other structural abnormalities, including craniofacial (Like what: cleft palate, supernumerary nostrils), central nervous system (hydrocephalus and neural tube defects), skeletal and abdominal abnormalities have been reported [1, 9]. Which was different from our observation.

Gender: Our observation was male, consistent with the literature data; where there is a male predominance with a sex of 2.7/1 [9].

Notion of polymaformative child birth in the lineage and diagnosis of Cantrell syndrome: The appearance of pentalogy is considered sporadic [9], which is our case. Diagnosis can be made antenatally with prenatal ultrasound during the first trimester of pregnancy [9, 11] or even through prenatal magnetic resonance imaging (MRI) which can improve the visualization of fetal abnormalities and aid in planning surgical management [12]. In our case there was no history of malformations or notion of abortion in the mother of the polymalformed. On the other hand, in Sidbe N *et al.*, there was a lack of history of fetal malformations but the mother of the polymalformed presented 4 spontaneous abortions in the past that have not been explored. Age of discovery: Regarding the age of discovery, some cases are diagnosed after childbirth or in childhood and others in the prenatal period. In our case, the diagnosis was made at 27 SA; This is close to the antenatal diagnostic age for the cases observed by Issam Ben A *et al.*, 2010 [13] in Tunisia, Leno DWA *et al.*, [14] in Guinea Conakry, and Flavio Hernández Castro *et al.*, [2] in Mexico in 2006 who found respectively 19 SA, 24 SA and 25 SA as the age of discovery. However, this result differs from that of Sidibe N *et al.*, [9] and A Ngaha [10] who discovered their observations after the birth of the fetus. This can be explained by the non-realization of prenatal ultrasounds especially in the 1st trimester and the 2nd trimester. The survival rate of the complete form of Cantrell pentalogy is less than 20% [15], and according to Vasquez [16] the survival rate reaches 37.3% after surgery. Recent studies have reported normal growth up to age six in children who received early surgical correction [15].

About other malformations associated with Cantrell pentalogy (acrânie, exencephaly and iniencephaly).

As for the acrânie which was the main malformation associated with the pentalogy of Cantrell is a rare lethal malformation, characterized by a defect in the development of the flat bones of the scalp while the brain tissue often present is abnormal; In 2004 Resgui-Marhoul *et al.*, [4] reported a case of antenatal ultrasound diagnosis. The difference with our observation is that his case was associated with other types of malformations which are: spinal and visceral malformations as well as cord abnormalities that had only two vessels. However, in our case, acrânie was associated with Cantrell pentalogy and iniencephaly. Regarding iniencephaly: it is a rare pathology, its incidence is 0.1-10 per 10,000 live births, of unknown etiology [17]. The diagnosis is possible from 12-13SA, it is a malformation that mainly affects the female sex [5]. Hanane S and Abdelaziz B [7], reported a case of antenatal diagnosis of iniencephaly by ultrasound at 22 SA in a 30-year-old patient; without a notable pathological history. It is usually accompanied by other birth defects and has a poor fetal prognosis [18]. Its association with encephalocele, myelomingocele, anencephaly and visceral abnormalities have been reported in the literature [5, 17, 18]. In our case, the diagnosis was made at 27 SA (this can be explained by the fact that she came late for her first antenatal consultation), in a male fetus she associated acrania and Cantrell pentalogy. Fetal MRI is a diagnostic tool to confirm fetal abnormalities. It has allowed the confirmation of malformations with precision in some similar studies [7]. However, it is a means of imaging that is difficult to access in our country currently. Ultrasound was sufficient for diagnosis in our case.

CONCLUSION

Cantrell's Pentalogy is rare, its association with acrania and iniencephaly is exceptional. Ultrasound is the key to antenatal diagnosis, it was sufficient in our case for the therapeutic decision. Medical termination of pregnancy is the rule.

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