Case Report

A RARE FORM OF CANTRELL SYNDROME WITH EXTRACORPORAL ECTOPIC HEART, ANENCEPHALY AND SPINAL DEFECT.

CASE REPORT AND LITERATURE REVIEW

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ABSTRACT

A rare case of Cantrell syndrome with heart evisceration in combination with anencephaly, spinal defect and normal extremities is presented. The Thoracic – abdominal syndrome (TAS), also known as Cantrell syndrome is a rare fetal anomaly, which includes defects of: the abdominal wall, sternum, diaphragm, pericardium and the heart. We describe a case of Cantrell syndrome with cardiac evisceration in combination with anencephaly and spinal bifida of the fetus diagnosed by ultrasound at 24 gestational weeks. We found large omphalocele with fully functioning heart in it, without intracardiac anomalies, which is a very rare finding. The pregnancy was interrupted. We believe that Cantrell syndrome could be a part of a greater fetal anomaly, which expresses with different severity in different cases. In our opinion the sonographers should be aware and looking for these anomalies in order to diagnose it earlier in the pregnancy.

Key words: Cantrell syndrome, anencephaly, meningocele, omphalocele, cardiac evisceration, spina bifida,

CASE REPORT

A 25 year old primigravida (S. H. I.) presented at our clinic, directed by her observing gynecologist, for consulting of pregnancy at 24 gestational weeks, because of obvious fetal anomalies. We did ultrasound and Doppler diagnostics and found several serious malformations:

1. A large defect of the abdominal wall with total enterocele, including liver, spleen, stomach and heart (Figures 1, 3). The heart was fully functional and without obvious intracardiac anomalies, which is a very rare finding.
2. Anencephaly with meningocele 5-6 cm. (Figure 2)
3. Large spinal bifida
4. Mild hydramnion (Figure 4)

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Figure 1. Cantrell syndrome. Arrow pointing at the ectopic heart
Figure 2. Anencephaly

Figure 3. Cantrell syndrome. Magnified. Arrow pointing at the ectopic heart

Figure 4. Mild hydramnion
We established gestational age of 24 weeks by measuring the femur length. After ultrasound and Doppler diagnostics decision was taken and the patient was directed for termination of the pregnancy on medical indications, because of the severe fetal abnormalities. Two days later the pregnancy was interrupted. Postoperative course was uneventful. The patient was discharged in good clinical condition, without any complications. The postmortem examination of the fetus confirmed the ultrasound findings: Fetus with female genitalia, large omphalocele, partial sternal dysplasia, evisceration of small and large intestines, liver, spleen, stomach and heart, anencephaly and opened spinal bifida. The four extremities are without any structural defects (Pictures 1, 2, 3, 4 and 5).

**Picture 1.** Cantrell with anencephaly

**Picture 2.** Cantrell with anencephaly. Arrow, pointing the heart
Picture 3. Cantrell with anencephaly.


Picture 5. Cantrell with anencephaly.
DISCUSSION
A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart is described for the first time by Cantrell JR, Haller JA, Ravitch MM., 1958(1). The typical pentalogy of Cantrell syndrome includes anomalies, considering not just the position of the heart, but also intracardiac structural defects, which we did not discover in our case. Reviewing the literature, we found several reports of different forms and combinations of Cantrell syndrome with other fetal anomalies like OEIS complex (omphalocele, extrophy of the bladder, imperforate anus, and spine defects) (2), ADAM syndrome (amniotic band syndrome)(3), etc. Polat I, Guel A, Aslan H, et all., reported in 2005 three cases very similar to ours. They describe a combination of Cantrell pentalogy with craniorachischisis, completely open spine, plus clubhand, clubfoot and polyhydramnion (4). The difference of our report is in the lack of intracardiac anomalies and entirely normal arms, hands, legs and feet, as visible at the pictures. There is one case report from Jeroen H. L. van Hoorn, Rob M. J. Moonen (5), 2007 that describes large omphalocele with ectopia cordis, and without intracardiac defects. In their case however the fetus doesn’t have an anencephaly. Atis A, Demirayak G, Saglam B, Aksoy F, Sen C(6), 2011, reported a case of craniorachischisis with a variant of pentalogy of Cantrell, with lung extrophy.

CONCLUSION
We believe that Cantrell syndrome could be a part of a more complex fetal anomaly, which expresses with different severity in different cases. In our opinion, the sonographers should be aware and looking for these anomalies in order to diagnose it earlier in the pregnancy.

REFERENCES