CASE REPORT

Pentalogy of Cantrell

Sally DE Mohmmed[1], Nadia Elrayah[2], Helmi Noor[3], Badreldeen Ahmed[4]

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This is a 24-year-old primigravida patient married to her first cousin. The patient was referred from a rural hospital to the Ian Donald teaching center in Khartoum for the second opinion and for further management. The indication for referral was suspected abnormal fetus at 29 weeks. We did not have the full record of early pregnancy.

At the Ian Donald teaching center, the ultrasound examination revealed the following: the estimated fetal weight was found to be below the 10th centile. There was a marked polyhydramnios, and the deepest vertical pool measured 12 cm. The anterior abdominal was absent with protrusion of stomach, small bowel, and the liver (Fig. 1).

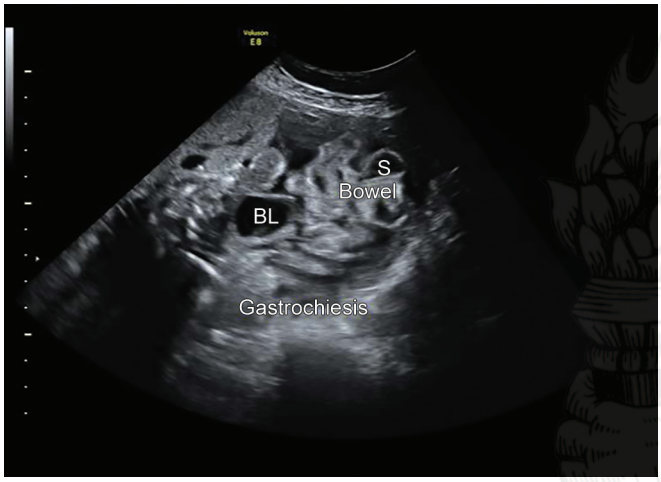

Fig. 1: Twenty eight weeks fetus with complete absence of anterior abdominal wall and chest wall. Bowel, liver and urinary bladder are seen outside the fetal abdomen

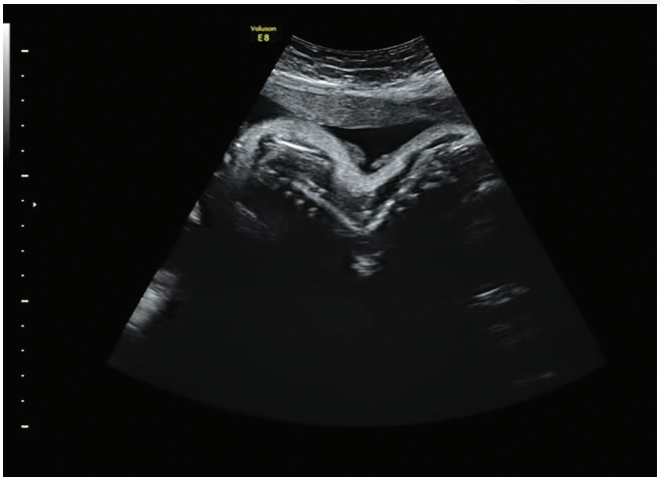

Fig. 2: Spine showed severe deformity in the form of severe scoliosis

[1–3]University of Medical Science and Technology, Khartoum, Sudan
[4]University of Medical Science and Technology, Khartoum, Sudan; Weill Medical College, Doha, Qatar; Qatar University-Medical School, Doha, Qatar; Feto-Maternal Center, Doha, Qatar
Corresponding Author: Badreldeen Ahmed, University of Medical Science and Technology, Khartoum, Sudan; Weill Cornell Medical College; Feto-Maternal Centre, Doha, Qatar, Phone: +974 55845583, e-mail: profbadreldeen@hotmail.com

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The thoracic wall was open with the fetal heart completely outside the chest. The diaphragm could not be visualized. The four-chamber view and the connecting vessels were seen outside the thoracic cavity (Fig. 2). The cardiac examination was limited because of late referral; however, there was an obvious large ventricular septal defect, we could not do a full echocardiography. Examination of the central nervous system was normal. The spine showed severe angulation (Fig. 3). Based on the above, we made the diagnosis of complete pentalogy of Cantrell,[1] the fetal condition is deemed as fatal and no place for surgical correction. At 32 weeks of gestation, the patient was induced with misoprostol. She delivered a female baby weighing 1200 g. The couple declined autopsy, however.

The baby was inspected and examined by the neonatologist. The postnatal findings were similar to the antennal findings.

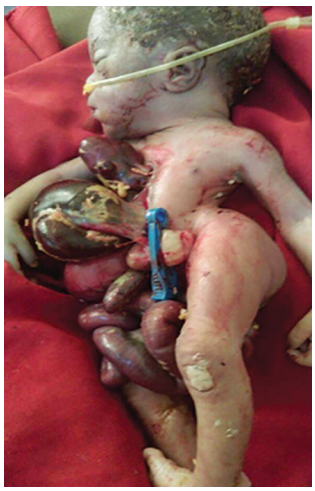

Fig. 3: Postpartum picture of the neonate showing, complete defect of thoracic and abdominal wall. Heart is completely outside and attached only by great vessels. Liver, small and large bowel are seen

There was a large abdominal and thoracic defect. The liver, the spleen, the stomach, the small, and the large bowel were seen outside the baby. The heart was seen outside the chest and connected to the baby only with the great vessels. The diaphragmatic defect was noted by the neonatologist. Autopsy result is not viable.

Discussion

We present here a rare case of complete pentalogy of Cantrell. The etiology of this condition is not known. It is often described as being sporadic in nature.[2] This condition involves disruption of the abdominal wall and the thoracic wall. In our case, all the features of pentalogy of Cantrell were seen, including, lower sternal defect, midline supraumbilical thoracic and abdominal wall defects, diaphragmatic defect, cardiac defect, and ectopia cordis.[3,4]

Pentalogy of Cantrell can include other fetal malformations away from the midline defect. Conditions like craniofacial anomalies, cleft lip and palate, central nervous system anomalies, skeletal anomalies and clubfoot, polysplenia, and gallbladder agenesis can be seen.[5]

The exact etiology of pentalogy of Cantrell is unknown. The general belief is that the problem started in very early embryonic life, mostly likely within the first 3 week of the embryonic life where there is a failure of the development in the lateral mesoderm.[6] The failure in development could be due to a number of factors which include gene mutation, chromosomal anomaly, or disrupted blood vessel.[7,8] In some cases of pentalogy of Cantrell, there are encephalocele and facial defects such as cleft lip and palate.[9] It is also possible in cases of pentalogy of Cantrell, to have the accumulation of fluid leading to pleura this could be due to cardiac failure.[10–12] Skeletal malformation is not very often seen.[13,14] The diagnosis of pentalogy of Cantrell can be made by ultrasound with great degree of accuracy after 12 weeks of gestation. In the first trimester, however, physiological herniation of the fetal bowel can make the diagnosis difficult especially in the mild form of pentalogy of Cantrell.[15] 2D ultrasound is adequate to make the diagnosis in this condition. 3/4D ultrasound is hardly needed in this situation, may it can help to counsel the parents and the couple may understand the 3/4D picture better. Other diagnostic modalities such as magnetic resonance imaging (MRI) and computed tomography (CT) scan are very rarely needed.[16–18] The prognosis in this condition is very poor. The survival rate even after a very complex medical and surgical intervention is very poor. The mean survival rate in most cases is hours rather than days and years.[19]

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