Prenatal diagnosis of a case of pentalogy of Cantrell with spina bifida

Sir,

In 1958, Cantrell described a syndrome in which a ventral diaphragmatic hernia occurred in association with
omphalocele. This rare syndrome is now called the pentalogy of Cantrell and it consists of the following five anomalies: a deficiency of the anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, various congenital intracardiac abnormalities and a defect of the lower sternum. We report a case of pentalogy of Cantrell associated with thoraco-lumbar spina bifida, hydrocephalus and lemon sign diagnosed prenatal in the second trimester, a finding not previously recorded in the literature.

An 18-year-old woman (gravida 1, para 0), presented for routine ultrasound examination at 20 weeks gestation. Detailed ultrasonography demonstrated omphalocele with abdominal visceral eventration (liver, stomach and intestines), ectopia cordis and hydrocephalus (lemon sign), thoraco-lumbar spina bifida with a femur length corresponding to the menstrual age. An anterior thoraco-abdominal wall defect was seen with the contents herniating into the amniotic cavity covered by a thin sac. The fetal heart was seen over the omphalocele and clearly out of the thorax [Figures 1, 2]. On the basis of the sonographic findings, we diagnosed the pentalogy of Cantrell with thoraco-lumbar-rachischisis. After we discussed the diagnosis with the parents, the pregnancy was terminated. There was an upper abdominal omphalocele defect with covering membranes getting ruptured during extraction at labor, due to which the liver and intestines were seen protruding out [Figure 3].

The incidence of this syndrome is less than 1 in 100 000, with a 2:1 male predominance. Based on a review of 61 cases of Cantrell's pentalogy, Toyama suggested the following classification for the syndrome: Class 1, certain diagnosis, with all five defects present; Class 2, probable diagnosis, with four defects (including intracardiac and ventral abdominal wall abnormalities) present; and Class 3, incomplete, with various combinations of defects present (but always including a sternal abnormality). A ventricular septal defect was not observed in our case, although Cantrell noted that congenital intracardiac anomalies are constant elements of the pentalogy, with ventricular septal defects seen in every case (100%), atrial septal defects in 53%, pulmonary stenosis in 33%, tetralogy of Fallot in 20% and left ventricular diverticulum in 20%. Only seven of the 60 reported cases of Cantrell's pentalogy involved the central nervous system and craniofacial malformations, which included anencephaly, meningocele, cephalocele, hydrocephalus and exencephaly. To our knowledge, Cantrell's pentalogy with thoraco-lumbar spina bifida, hydrocephalus and lemon sign was not previously reported in this association. Our case is a variant form of the classic pentalogy of Cantrell and extends the spectrum of Toyama's classification, particularly Class 3. Termination of pregnancy can be offered before viability. After viability, a periodic ultrasonographic evaluation of the lesions, fetal growth and delivery in a tertiary center is recommended.
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References