Prenatal Diagnosis of Pentalogy of Cantrell in the First Trimester: Is 3-Dimensional Sonography Needed?

Fernando Maia Peixoto-Filho, MD, Luciana Carneiro do Cima, MD, Marcos Nakamura-Pereira, MD

Fetal Medicine Unit, Department of Obstetrics, Fernandes Figueira Institute, Oswaldo Cruz Foundation (IFF-FIOCRUZ), Avenida Rui Barbosa, 716, 3º Andar, Flamengo, Rio de Janeiro, Brazil

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ABSTRACT: We report the prenatal diagnosis of 2 cases of Pentalogy of Cantrell in the first trimester. In case 1, sonographic evaluation revealed ectopia cordis, omphalocele, and cystic hygroma at 10 weeks’ gestation. In case 2, sonographic assessment during the first trimester detected ectopia cordis and omphalocele at 11 weeks’ gestation. In both cases, the patient opted for elective pregnancy termination, and Pentalogy of Cantrell was confirmed in 2 male fetuses. We discuss the role of Doppler imaging and 3-dimensional sonography as complementary methods to conventional sonographic assessment of abdominal wall defects at early pregnancy.

Pentalogy of Cantrell was first described in 1958 and was characterized as an anterior body wall midline developmental anomaly, the prevalence of which ranges from 1/65,000 to 1/200,000 births. This syndrome consists of a supraumbilical wall defect, a defect of the lower sternum, a deficiency of the anterior diaphragm, and various intracardiac defects. The expression of these defects are variable, and the prognosis depends on the severity of the lesions. However, generally, Pentalogy of Cantrell with ectopia cordis has a very high perinatal mortality rate, characterizing the importance of its differentiation from other abdominal wall defects. The differential diagnoses include isolated thoracic ectopia cordis, amniotic band syndrome, and body stalk anomaly. The precise etiology of Pentalogy of Cantrell remains unknown, and the most accepted hypothesis involves the abnormal development of mesoderm at an early stage of embryonic life. We report the prenatal diagnosis of 2 cases of Pentalogy of Cantrell in the first trimester and discuss the role of Doppler imaging and 3-dimensional (3D) sonography as complementary methods to conventional sonographic assessment.

CASE REPORTS

In case 1, a routine sonographic examination was performed on a 22-year-old woman, gravida 1, para 0, for first-trimester screening of aneuploidy. Transabdominal sonographic examination using an HDI 4000 scanner with a 3.5-MHz curved-array transducer (Philips Medical Systems, Bothell, WA) revealed a live fetus with a crown–rump length (31 mm) consistent with a 10 weeks’ gestation, ectopia cordis, omphalocele with abdominal visceral eventration, and cystic hygroma (Figure 1). In case 2, a 33-year-old woman, gravida 1, para 0, was referred to our fetal medicine unit to undergo first-trimester screening of aneuploidy. A transabdominal sonographic evaluation using a Voluson 730 Expert scanner equipped with a 4- to 8-MHz transducer (GE Medical Systems, Milwaukee, WI) demonstrated a live fetus with a crown–rump length (44 mm) consistent with 11 weeks’ gestation, ectopia cordis, and omphalocele with liver eventration.
In these 2 cases, no intracardiac abnormalities were detected due to early gestational age, and nuchal translucency was not measured because the crown–rump lengths were below 45 mm. After counseling the parents on the poor prognoses of fetuses, both patients opted for elective pregnancy termination and Pentalogy of Cantrell was confirmed in 2 male fetuses. We used 3D sonography and Doppler imaging as complementary tools to obtain better images of the supraumbilical wall defect and to improve counseling of patients.

**DISCUSSION**

Pentalogy of Cantrell is a rare congenital anomaly characterized by midline closure defects, and its survival rate is quite low. In a review of cases published in the last 20 years, van Hoorn et al suggested that the presence of intracardiac defects in patients with ectopia cordis may favor the prognosis, which seemed to be poorer in patients with the complete form of Pentalogy of Cantrell and when associated anomalies were present.

The prenatal diagnosis of Pentalogy of Cantrell in the first trimester was initially described by Bennett et al using 2-dimensional (2D) sonography and Doppler imaging. Recently, various
reports discussed the role of 3D sonography and fetal MRI on Pentalogy of Cantrell assessment.2,4,6,9-11 Although there is some agreement that 3D sonography provides additional information for fetuses with abdominal wall defects4,11,12 and Pentalogy of Cantrell,2,11 the application of this tool is not necessary for diagnostic purposes in most cases. Early prenatal diagnosis of Pentalogy of Cantrell is enabled by the existence of ectopia cordis and omphalocele,3 and 2D sonography with Doppler imaging can clearly demonstrate these defects. In the cases in which the defect is too small to be detected—as in those with partial anterior, transient, or intermittent displacement of the heart—other sonographic markers (eg, transient pericardial effusion associated with omphalocele), which are usually detected in the second trimester, are of great value.2 Various other anomalies have been reported in association with Pentalogy of Cantrell, such as cleft lip/palate, encephalocoele, hydrocephalus, craniarachischisis, clubfoot, gallbladder agenesis, and polysplenia,5 and some of them can be detected in the first trimester. In addition, McMahon et al6 suggested that the association of fetal MRI and prenatal echocardiography allows optimal assessment of fetuses with Pentalogy of Cantrell due to better delineation of the cardiac anatomy and the extent of thoracoabdominal defect, providing prognostic information. However, these tools are used in view of the standpoint of preoperative planning after the first trimester.

In our 2 cases of Pentalogy of Cantrell, the diagnoses were achieved with 2D sonography and Doppler imaging. We believe that 3D sonography is a valuable tool for evaluation of fetuses with abdominal wall defects during the second and third trimesters but is not essential in the first trimester. In the second and third trimesters, the fetal echocardiography may also provide relevant information for preoperative assessment due to the various cardiac anomalies associated with the Pentalogy of Cantrell. 2D sonography with Doppler imaging is the choice for sonography assessment of abdominal wall defects during the first trimester. When possible, complementary 3D sonography can be used to help with parent counseling, because it provides images that are easily understood.

REFERENCES


