A 28-year-old woman, gravida 2, para 0, was referred to our hospital because of a fetal anomaly at 12 weeks 6 days of gestation; she had no previous family or medical history. Transvaginal two-dimensional (2D) ultrasound examination showed a single live fetus consistent with a 12-week gestation, with abdominal visceral evagination of the gastrointestinal tract, ectopia cordis, and a defect of the lower sternum. In addition, a cystic lesion in the cranium suggesting exencephaly was noted, but detailed detection of the craniospinal structure could not be achieved. Three-dimensional (3D) ultrasound examination using a Medison ultrasound system (Accuvix XQ; Medison, Seoul, Korea) was then performed for differential diagnosis of the cranial lesion. A transparent image of the fetal skeleton obtained using 3D demonstrated craniorachischisis (Figure 1).

Because of the diagnosis of pentalogy of Cantrell with craniorachischisis, the patient and her family decided to terminate the pregnancy after giving written informed consent. A 15-g male fetus with multiple anomalies was delivered vaginally at 13 weeks 4 days of gestation by transvaginal administration of gemeprost. Gross examination confirmed ectopia cordis, a supraumbilical abdominal wall defect with evagination of the liver, stomach and intestine, a defect of the lower sternum, and craniorachischisis, which was compatible with the prenatal findings (Figure 2). Chromosomal diagnosis by using chorionic villi from the aborted specimen resulted in the finding of a normal karyotype of 46,XY.

Pentalogy of Cantrell is a rare syndrome which is defined by the presence of a supraumbilical abdominal wall defect, various intracardiac defects, a defect of the lower sternum, deficiency of the anterior diaphragm, and a defect in the diaphragmatic pericardium [1]. The prevalence of this syndrome has been estimated as 5.5 in 1 million live births [2]. This syndrome has been reported with complete or incomplete phenotypes in the literature [3], the latter phenotype being observed in 40% of the cases of this syndrome. The central nervous system malformations are associated with the incomplete phenotypes [4]. Only three cases with craniorachischisis in the literature were reported to have been prenatally diagnosed, all of which were diagnosed during 18–26 weeks of gestation by using 2D ultrasound examination or magnetic resonance imaging [5,6].

This is the first case of pentalogy of Cantrell with craniorachischisis diagnosed by 3D ultrasound in the first trimester. There seems to be a rare association of this syndrome with malformations in the central nervous system and, to our knowledge, only three cases with craniorachischisis have been reported as mentioned above [5,6]. Bognoni et al [4] also reported a case with exencephaly in the first trimester, in which spinal dysraphism was confirmed at autopsy. In this case, a cystic lesion detected by 2D ultrasound implied exencephaly, but was not sufficient to establish the diagnosis because of the earlier stage of gestation. Accordingly, the 3D ultrasound examination was useful for detecting the structural malformation of the bone in craniorachischisis.

Although 2D ultrasound examination has played an important role in prenatal diagnosis, the recent development of 3D imaging has provided more detailed anatomic information regarding fetal malformations in cases where 2D imaging has proven insufficient. Furthermore, the 3D transparent mode is more suitable for detecting fetal bone malformations because of the relatively greater contrast difference compared with its neighboring structures [7].

In this case, the combination of the 2D and 3D ultrasound methods provided useful information for prenatal detection of pentalogy of Cantrell with craniorachischisis.
References