Sirenomelia, characterized by complete fusion of the lower limbs in combination with severe urogenital and gastrointestinal malformations, is a rare and usually lethal disorder. Progression oligohydramnios due to agenesis or dysgenesis of the kidneys is usually the first sign of this lethal malformation during the second trimester. Early prenatal diagnosis of sirenomelia can be achieved by sonographic demonstration of constant simultaneous movements of the lower limbs and/or the identification of a skeletal anomaly like a single femur.

This report provides a case of sirenomelia detected by using three-dimensional (3D) ultrasound at 11th gestational weeks.
CASE REPORT

A 22-year-old woman, gravida 3, para 1, was referred for a targeted ultrasound investigation to our ultrasound unit in the 11th week of gestation because an intraabdominal cyst was detected during a routine scan. B-mode transabdominal and transvaginal ultrasound examinations showed a left-sided intraabdominal cystic structure measuring 9.6 x 9.4 mm in diameter, which was located just beneath the stomach (Figure 1). Meticulous scanning revealed fusion of the lower limbs, an abnormal pelvic bone and a single umbilical artery. The sacrum, both kidneys and bladder were also not visible. These sonographic findings favored the diagnosis of sirenomelia. Presence of lower extremity fusion was further supported by transabdominal 3D ultrasound imaging (Figure 2). After non-directive counseling, pregnancy termination was requested by the couple. Postmortem radiological study revealed a single femur, absent sacrum, and hypoplastic pelvic bones, which is consistent with the prenatal diagnosis (Figure 3). The cystic structure detected on prenatal ultrasound turned out to be dilatation of sigmoid colon during autopsy and the gross appearance of the fetus confirmed the existence of a single lower extremity (Figure 4). A scrupulous dissection showed bladder agenesis and presence of intraabdominally located gonads. Placental examination revealed a single umbilical artery. Fetal karyotype was 46, XX.

DISCUSSION

Sirenomelia sequence combines in all cases fusion and rotation of the lower limbs to various degree and anorectal atresia. Renal agenesis or dysplasia, bladder agenesis and absence of internal and external genitalia except the gonads are noted in nearly all cases. Dysgenesis or even agenesis of the sacrum may be associated with extensive vertebral dysgenesis. The early fusion of the lower extremities confers a siren-like appearance of the fetus. The incidence is about 0.01 to 0.16 per 10000 live births, males are three times more often affected than females and the incidence is higher in one of the iden-
tical twins. The etiology of sirenomelia is not clear, however, vascular disruption or caudal embryo damage was proposed. Additionally, chromosomal abnormalities are rare in cases with sirenomelia.

Although urinary tract dysgenesis or agenesis appears to begin from the end of the first trimester, most cases of sirenomelia were diagnosed early in the second trimester due to oligohydramnios as an alerting sign to the sonographer. As during the early weeks of gestation the amniotic fluid is mainly secreted by the placenta and the umbilical cord, amniotic volume may not be helpful for the prenatal diagnosis of sirenomelia in the first trimester.

In our case, the cystic structure, which was detected at 11 weeks of gestation, reflects the complex nature of sirenomelia, which comprises malformations of the lower abdomen, urogenital system and intestine. Dilatation of the sigmoid colon due to an imperforate anus has been reported as an intraabdominal cystic structure in previous cases with first trimester diagnosis of sirenomelia. Therefore this sonographic finding might be an important indirect sign of serious lower extremity abnormalities, especially sirenomelia.

The differential diagnosis of abdominal cysts during the first trimester includes megacysts, although this is unlikely at 11 weeks of gestation.

3D ultrasound images of two cases with sirenomelia diagnosed in the first trimester were firstly provided by Monteagudo et al, which aided to further characterize the conventional 2D sonographic findings. The prenatal diagnosis with 3D sonography were thought to be the additional information obtained namely was the overall view, the confirmation of 2D findings, and the abnormal fetal movements, which could increase the physician’s confidence and understanding of the fetal condition.

In conclusion, sirenomelia should be suspected when an intraabdominal cystic structure is visible during a routine first trimester ultrasound scan. Subsequent 3D ultrasound may be helpful in confirming the diagnosis of a sirenomelia case.

REFERENCES