Introduction

Omphalocele is a central abdominal wall defect containing abdominal viscera, covered by a membrane of amnion and peritoneum with Wharton’s jelly between the two layers.

The defect is located at the base of the umbilical cord and the cord inserts at the top of the omphalocele sac, which can contain bowel, liver, stomach and bladder accompanied by ascites in the sac or the abdomen.

Omphalocele is frequently associated with chromosomal and structural anomalies, namely Trisomy 13, 18, 21, Turner syndrome, triploidy or some rare chromosomal deletions. Here, we report a fetal omphalocele having 18p deletion.

Case

A 31-year-old woman, gravida 1 was referred at the 20th weeks of gestation due to a suspicion of fetal omphalocele. Her medical history was uneventful. The patient did not have first trimester screening test and her triple screening test was in the low risk area for trisomies.

Ultrasound revealed an omphalocele sac containing liver, bowel and bladder (Figure 1). No additional structural abnormality was detected except for an umbilical cord cyst.

Genetic counseling was recommended and amniocentesis was performed, revealing a 18p deletion. Then termination of the pregnancy was recommended because of the poor prognosis, and medical abortion was done.

Postmortem examination revealed a female fetus with omphalocele.

Conclusion

Further evaluation is mandatory in fetuses with a prenatally identified omphalocele. Genetic counseling and fetal karyotyping should be offered and also, detailed ultrasound examination should be performed to evaluate other possible coexisting structural abnormalities.