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Case Report

Prenatal diagnosis of fetal gallbladder duplication associated with uncommon chromosomal anomaly (46, XX, t(X;10) (p11.2;q24.3) [20])

E.H. Comert[1], H. Sal[1], C. Kart[1], E.S. Guvendag Guven[1], S. Guven[1]

1*Department of Obstetrics and Gynecology, Karadeniz Technical University, School of Medicine, Trabzon (Turkey)*

Summary

Fetal gallbladder duplication is a rare congenital malformation. In the literature only a few cases of fetal gallbladder duplication in utero is reported. A 22-year-old woman was referred to this hospital at 26 weeks of gestation for a routine ultrasound examination. A fetal duplication of gallbladder accompanied was diagnosed with ultrasonography (axial section of fetal abdomen on sonography showed two hypoechoic cyst-like structures). The diagnosis of duplication of gallbladder, was also confirmed with MRI. The result of cordocentesis was reported as 46, XY, t(X;10) (p11.2;q24.3) [20]. Antenatal/natal/postnatal care was unremarkable. A novel chromosomal mutation (46, XX, t(X;10) (p11.2;q24.3) [20]) with duplicated fetal gallbladder which has not been reported previously was described.

Key words: Chromosomal anomaly; Fetal gallbladder duplication; Ultrasonography; MRI.

Introduction

Fetal gallbladder duplication is a rare congenital malformation, occurring in about one in 4,000 births, resulting in two separate gallbladder cavities, each with a cystic duct. Gallbladder anomalies, including cholelithiasis, choledochal cysts, and agenesis, have been observed in utero [1, 2]. In the literature only a few cases of fetal gallbladder duplication in utero (only four cases) were reported. All reported cases were not associated with additional congenital or genetic anomalies. A novel chromosomal mutation (46, XX, t(X;10) (p11.2;q24.3) [20]) with duplicated fetal gallbladder which has not been reported previously was reported.

Case Report

A 22-year-old woman (gravidity 1, parity 0) was referred to the present hospital at 26 weeks of gestation for a routine ultrasound examination. Obstetric and medical histories were all unremarkable. A fetal duplication of gallbladder accompanied was diagnosed with ultrasonography (Figure 1a). The diagnosis of duplication of gallbladder (Figure 1b), was also confirmed with MRI.

In order to rule out fetal aneuploidy, cordocentesis was suggested. The result of cordocentesis was reported as 46, XY, t(X;10) (p11.2;q24.3) [20]. C-banding with trypsin-Giemsa and high resolution banding techniques were used to assess this anomaly. The patient was counseled about the implication of this chromosomal anomaly based on literature findings (infertility and gallbladder diseases). Fetal sonographic follow-up was also confirmed the initial diagnosis. At 39 weeks of gestation a 2,480-gram female was delivered via cesarean section for primiparous breech presentation. Antenatal care was only remarkable for the diagnosis of small for gestational age fetus (without and abnormal fetal well being problems and abnormal prenatal doppler findings). Neonatal abdominal sonography on postnatal day 3 confirmed the prenatal diagnosis of gallbladder duplication (two gallbladders with two cystic canal opening to one common bile duct). The baby was discharged at day 7 without any complications. No additional ultrasound abnormalities either prenatal or postnatal screening were reported. The woman also had the same chromosomal anomaly and abdominal sonography of woman revealed the diagnosis of cystic canal duplication (one gallbladder with two cystic canal opening to one common bile duct).

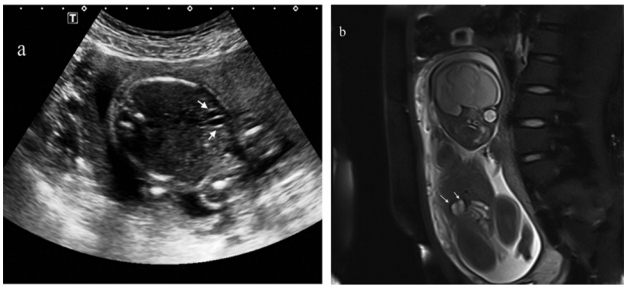

Figure 1. — (a) Axial section of fetal abdomen on sonography shows two hypoechoic cyst-like structures (arrows, fetal gallbladder duplication). (b) Coronal T2 weighed MRI shows two rectangular cyst-like structure located at the lower border of the liver (arrows, fetal gallbladder duplication) at 26 weeks' of pregnancy.

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Discussion

The diagnosis of gallbladder duplication is very rarely carried out during routine obstetric sonography. The gallbladder appears as a small, hypoechoic, oval or rectangular cyst-like structure located at the lower border of the liver, close to the intestinal loops, and to the right of the intrahepatic umbilical vein [2]. The fetal gallbladder is visualized in 65 -82% of fetuses at 24-27 weeks' gestation [3]. Although by using a transvaginal approach at 14-16 weeks' gestation the detection rate may be increased to more than 99% [2].

Gallbladder duplication should be considered when an additional cystic structure is seen in the right upper quadrant. Its specific location in the gallbladder fossa and its characteristic appearance are helpful in distinguishing it from other pathologic entities, particularly choledochal and duodenal duplication cysts, both of which occur in this location. A structure lying parallel to the gallbladder was not identified in neither of these conditions. In particular, duplication cyst of the duodenum is contiguous with the descending duodenum and appears as an anechoic mass with strong back wall echoes. This may be associated with various congenital anomalies [4].

The differential diagnosis of gallbladder duplication from some gallbladder diseases is often difficult in woman or man. It is often found incidentally during the surgery for cholelithiasis [5-7]. The diagnosis duplication of gallbladder in utero may not be easily done since it does not have specific symptoms, only strict sonographic anatomic survey may give the opportunity to make the diagnosis of such rare anomaly. There have been no reported genetic or biochemical marker for such anomaly.

In case of suspicious fetal gallbladder anomaly in pregnancy, fetal sonography reveals two similar cystic structures located in the right upper quadrant, and the woman should be subjected to MRI study to confirm the diagnosis of gallbladder duplication. Following confirmation the diagnosis, fetal and/or parent karyotype analyses should be advised to exclude de novo or inherited genetic anomalies. Patients should be counseled about the clinical implication of such chromosomal anomaly (infertility, gallbladder diseases). Antenatal care should include the risks of small fetus and need of cesarean delivery.

In this case, MRI was planned when duplication of gallbladder was diagnosed on routine prenatal obstetric ultrasonography. The diagnosis was confirmed with MRI. Fetal karyotype analysis revealed 46, XY, t(X;10) (p11.2;q24.3) [20]. Literature review failed to reveal the association of such chromosomal anomaly and fetal gallbladder duplication. When clinicians suspect fetal gallbladder anomaly, MRI for confirming the diagnosis is suggested and fetal karyotype analysis to discover such unreported chromosomal anomaly.

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Corresponding Author:
S. GUVEN, M.D.
Department of Obstetrics and Gynecology
School of Medicine, Karadeniz Technical University
61080 Trabzon (Turkey)
e-mail: drsuleymanguven@yahoo.com