BRIEF COMMUNICATION

Trisomy 13 with the Absence of Gallbladder

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1. Introduction

Trisomy 13, also known as Patau syndrome, is a severe condition first described by Patau in 1960. Eighty-two percent of the patients die within the 1st month and 85 percent within the 1st year. The frequency of the syndrome is 1:3000-8000 live births. It is the third most frequent trisomy after trisomy 21 and 18 at birth. Although the features of trisomy 13 can vary, in general the syndrome is associated with numerous malformations, including holoprosencephaly, microcephaly, scalp defects, ocular abnormalities such as anophthalmia, micrognathia, and colobomata of the iris, cleft lip and palate, low-set ears, dysplastic auricles, micrognathia, polydactyly, overlapping fingers, congenital heart defects, omphalocele, as well as renal and genital abnormalities.1 Trisomy 13 is also associated with a high rate of spontaneous abortion and intrauterine death.2 To the best of our knowledge, this is the first presentation that is related to absence of gallbladder (GB) in case with trisomy 13.

2. Case Report

A 20-day-old girl was admitted to the Department of Pediatrics because of multiple congenital abnormalities. The child was born at term by cesarean section, weighed 2835 g (10-25 centile), and was 49 cm (25-50 centile) of length and had 33 cm (25 centile) of cephalic perimeter. It was the first pregnancy and child. The following abnormalities were detected on the physical examination: complete form of cleft lip and palate, large nose, bilateral microphthalmia, large ears with abnormal helixes, small attached earlobe, polydactyly only on the right hand, flexion contractures on both hands, bilateral hallux valgus, spina bifida occulta, and hemangioma on the back and ears. The parents were healthy, 31 (mother) and 30 (father) years old, and unrelated. There was no family history of congenital malformations. The total blood level and levels of direct bilirubin were 4.45 mg/dL and 0.72 mg/dL, respectively. Levels of aspartate transaminase and alanine transaminase were 90 U/L (8-33 U/L) and 20 U/L (5-40 U/L). Gallbladder was not seen in abdominal ultrasound or magnetic resonance imaging (MRI) (Figure 1). Intrahepatic biliary tracts could not be demonstrated, but it is known that magnetic resonance cholangiopancreatography (MRCP) in newborn period is difficult to detect on unless it is dilated. Grade two hydronephrosis was also detected on MRI. Cranial MRI showed the...
minimal dysgenesis of corpus callosum (Figure 1). A small muscular ventricular septal defect and patent ductus arteriosus were seen on echocardiography. The clinical diagnosis of trisomy 13 was confirmed by cytogenetic analysis as $47,XX,+13$. Fluorescence in situ hybridization (FISH) analysis excluded mosaicism. The parental karyotypes performed on lymphocytes were normal.

3. Discussion

Trisomy 13, considered a rare chromosomal disease, is a severe form of chromosomal disorder with a poor prognosis. Although it has several malformations, patients can show variable combinations of features. Hydronephrosis is a rare condition in trisomy 13. Spina bifida may be involved in the clinical manifestation of trisomy 13. Malformations of the gastrointestinal system, including esophageal, tracheoesophageal, and rectal malformations and abdominal wall defects are often seen in the patients with trisomy 13. However, to the best of our knowledge, GB in trisomy 13 patients has not been reported previously. GB is also a rare condition in general population; it is often asymptomatic and probably results from failure of the gallbladder bud to develop or vacuolize in utero. GB is almost always misinterpreted as cholecystitis with cystic duct obstruction or a scleroatrophic gallbladder, therefore leading to unnecessary surgery, which can lead to iatrogenic injuries and contribute to the mortality and morbidity in the patients with long survival of trisomy 13. Although the diagnosis of GB may be made by ultrasound (US), it should be confirmed by computed tomography (CT), endoscopic retrograde cholangiopancreatography (ERCP), or MRCP. The genetic basis of the development of gallbladder is still unknown. Bronstein et al have reported that GBA might be associated with chromosomal aneuploidy and other malformations. They have also reported GBA in trisomy 18 but not in trisomy 13.

There were no reports of trisomy 13 being involved in the pathogenesis of GBA by the possible alterations of gene dosages or genetic interactions. We think that absence of GB may be playing a part in trisomy 13. Further studies are required to elucidate the genetic etiopathogenesis of GB anomalies.

References