Atypical Presentation of Duodenal Atresia Concomitant with Type-A Esophageal Atresia in Fetal Life: A Case Report

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Duodenal atresia concomitant with type-A esophageal atresia (DA + TA-EA) is rare. A pronounced enlargement of a closed loop of the upper gastrointestinal tract serves as an early clue for its prenatal detection. We describe an atypical case of DA + TA-EA in which the dilatation of the upper gastrointestinal tract remained mild. Ultrasonographic examination at 28 weeks of gestation showed mild polyhydramnios. Subsequent detailed sonographic and magnetic resonance imaging studies revealed a mildly enlarged stomach and duodenum that resembled a "double bubble," mild ascites, and polydactyly of the right thumb. Fetal abdominal circumference measurements were within normal range. A female neonate born at 36 weeks gestation did not show abdominal distension. DA + TA-EA was diagnosed based on clinical characteristics and X-ray studies of the neonate; the diagnosis was confirmed by surgery. Duodenoduodenostomy and gastrostomy in the first week of life and esophagoesophagostomy at six months of age were performed with satisfactory results, and the infant developed well. Prominent and/or increasing C-shaped fluid collection in the upper abdomen is a highly useful diagnostic sign for DA + TA-EA, but it is not applicable for all fetuses with this disease. Physicians should bear this caveat in mind to avoid diagnostic delays and initiate prompt postnatal therapy.

Key words: Prenatal detection, Duodenal atresia, Type-A Esophageal Atresia, Ultrasonography

INTRODUCTION

Duodenal atresia concomitant with type-A esophageal atresia without tracheoesophageal fistula (DA + TA-EA) is extremely rare, and in most of the limited cases detected prenatally, progressive and marked enlargement of a closed loop of the gastrointestinal tract consisting of the distal esophagus, stomach, and proximal duodenum has been a characteristic finding. Fetal gastric rupture and neonatal respiratory distress occurred in two such cases [1, 2], and in another case, in utero stomach paracentesis was successfully performed to avoid gastric rupture and respiratory distress [3]. Here, we report an atypical case of DA + TA-EA in which the enlargement of the stomach and duodenum remained less than moderate during fetal life.

CASE REPORT

A 33-year-old nulliparous woman underwent an ultrasound examination at another institution at 28 weeks of gestation because of suspected polyhydramnios. In addition to polyhydramnios with amniotic fluid index (AFI) of 28 cm, abnormalities detected on sonography included a small stomach, single umbilical artery, and small amount of ascites. TORCH titers were negative, and oral glucose tolerance test (75 g) was normal. The woman was then referred to our hospital for further evaluation and management at 32 weeks of gestation. She was otherwise healthy, and her family history was unremarkable. Detailed ultrasonography (Fig. 1) revealed mild polyhydramnios with amniotic fluid index (AFI) of 26 cm; a mildly enlarged stomach and duodenum, resembling a "double bubble"; mild ascites; and polydactyly of the right thumb. Such findings were confirmed by fetal magnetic resonance image (MRI) (Fig. 2), which also demonstrated no evidence of a dilated portion of the esophagus. Mild to moderate polyhydramnios was noted. Amniocentesis for karyotyping was offered but declined. Fetal abdominal circumference measurements remained within normal range for gestational age (Table). Preterm premature rupture of the membranes occurred at 36 weeks, and a female neonate weighing 2860 g was delivered via cesarean section, with Apgar scores of 8 and 10 at 1 and 5 min, respectively. Polydactyly of the right thumb was the only external anomaly. The neonate exhibited no phenotypic features of trisomy 21 or other chromosomal abnormalities. Her abdomen was not distended with an abdominal circumference of 26.0 cm. She had excessive salivation in the few hours of life but did not have respiratory distress. Plain X-ray showed a coiled nasogastric tube in the upper portion of the esophagus and a gasless abdomen, suggestive of esophageal atresia (Fig. 3). Postnatal ultrasonography demonstrated a fluid-filled distended stomach and proximal duo-

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Table Comparison between the subacromial spur (SS) absent and SS present groups

Gestational Age	EFW (g)	BPD (mm)	AC (mm)	FL (mm)	AFP (mm)	AFI (cm)
32w4d	1672 (-1.01SD)	78.3 (-0.7SD)	260 (-0.4SD)	57.2 (-0.5SD)	98.3	26.0
33w6d	1893 (-0.9SD)	79.7 (-0.9SD)	270 (-0.3SD)	61.7 (0.2SD)	93.2	29.0
34w5d	2042 (-0.89SD)	84.1 (-0.2SD)	274 (-0.5SD)	62.4 (-0.1SD)	91.5	32.6
35w2d	2201 (-0.7SD)	85.7 (0.0SD)	286 (0.0SD)	62.2 (-0.4SD)	88.7	24.0

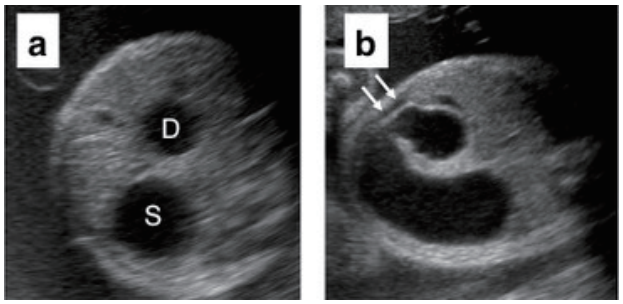

Fig. 1 Ultrasonographic images at 32 weeks of gestation. Axial section presents a dilated stomach (S) and proximal duodenum (D) (**a**, a double-bubble appearance). Continuity was noted between the two bubbles (**b**). White arrows show minimal ascites.

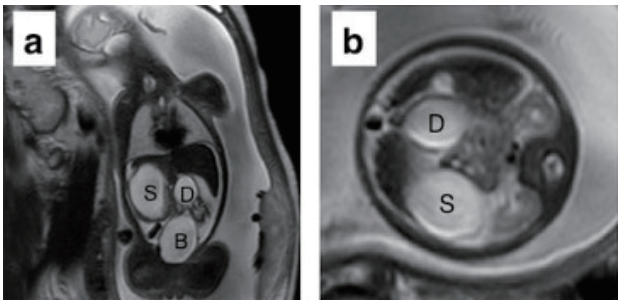

Fig. 2 T2-weighted coronal (**a**) and axial (**b**) MRI images at 32 weeks of gestation. Mild dilatation of the stomach (S) and duodenum (D) with mild polyhydramnios was noted. B, bladder.

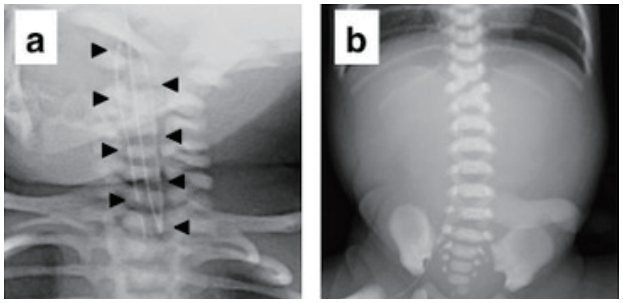

Fig. 3 Plain X-ray images. **a**. A coiled nasogastric tube in the upper portion of the esophagus (arrow heads). **b**. A gasless abdomen was noted.

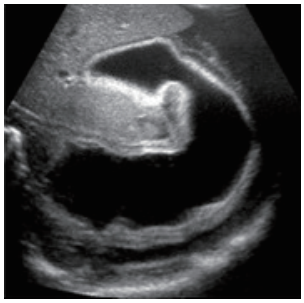

Fig. 4 Ultrasonographic axial image of the neonatal abdomen. Findings similar to those in Fig. 1 were noted.

denum (Fig. 4). Based on these findings, a diagnosis of duodenal and esophageal atresia without tracheoesophageal fistula (Gross type A) was made. No other components of the VACTERL association (Vertebral, Anorectal, Cardiac, Tracheo-Esophageal, Renal and Limb malformations) were detected. On the third day of life, duodenoduodenostomy and gastrostomy were performed. During the operation, a small amount of clear ascites was noted, but no evidence of gastric rupture was noted. No additional gastrointestinal or hepatobiliary abnormalities were found. At 6 months of age, the infant underwent esophagoesophagostomy and did not have major complications in the postoperative course. She developed well without evidence of significant sequelae.

DISCUSSION

Duodenal atresia (DA) concomitant with type-A esophageal atresia (EA) without tracheoesophageal fistula (DA+TA-EA) is rare and accounts for only 10% of type-A EA cases [4]. In the present case, based on sonographic visualization of the characteristic fluid-filled "double bubble" in the fetal upper abdomen with the continuity of the gastric and duodenal bubbles, DA was suspected. However, DA+TA-EA was diagnosed only after the birth when a nasogastric tube was found to be coiled in the esophagus without the presence of air in the stomach.

Through an extensive English literature search, we found 14 prenatally detected cases of DA+TA-EA [1–3, 5–12]. Previous case reports highlight the diagnostic value of a prominent and/or increasing C-shaped fluid collection in the upper abdomen on fetal sonography or MRI for prenatal suspicion of DA+TA-EA. As has been suggested by some authors [1, 2], the stomach and duodenum in DA+TA-EA should be filled with secretions to a much greater extent than what is observed in DA alone, which results in enormous dilatation of the closed loop of the upper gastrointestinal tract, including the distal esophagus, stomach, and proximal duodenum. However, the present case lacked this pathognomonic feature. The reason underlying this difference is unclear. However, considerable interfetal variation in the size of the stomach, the major component of the upper gastrointestinal loop, may be

involved. Moreover, the distribution of normal gastric measurements has been shown to increase noticeably with advancing gestation, particularly after midgestation [13–15]. In a reported series of ten neonates with esophageal atresia with associated DA, DA was initially missed in three infants [16]. Taken together, the production of gastric and probably duodenal juice, seems to differ considerably among fetuses and infants.

Several reports show that detection of the dilated distal esophagus can also be diagnostic of DA + TA-EA [2, 3, 8]. The present case also lacked this sign. Similarly, as discussed earlier, less fluid in the closed upper gastrointestinal loop seems to explain this lack of detection.

Fetuses with DA + TA-EA could potentially develop gastric rupture and neonatal respiratory distress due to extreme dilatation of the closed loop of the upper gastrointestinal tract [1, 2, 12]. Such complications might be avoided by in utero stomach paracentesis, as was reported by Kadohira *et al*. [3]. Theirs is the first and so far only case in which fetal stomach paracentesis was performed. Further studies are needed to justify this moderately invasive method and to define its indications. At least at present, this preventive method should be considered only for DA + TA-EA cases with progressive dilatation of the closed loop that threatens imminent rupture. In this respect, the present case was clearly not a candidate for fetal stomach paracentesis.

Therefore, this case adds to the knowledge on prenatal diagnosis of this rare anomaly. Thus, detection of enormous and/or increasing C-shaped fluid collection in the upper abdomen is a greatly useful, but not universal, diagnostic sign for DA + TA-EA. Mild dilatation of the "double bubble" alone should not exclude the possibility of associated esophageal atresia. Physicians should keep this fact in mind, perform careful antenatal observation of the fetal gastrointestinal tract, and aim to provide an optimal postnatal management of DA + TA-EA.

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