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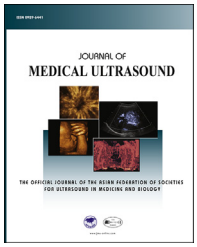

Letter to the editor

Prenatal Diagnosis of Anal Atresia — A Case Report

Yu-Ching Chou*, Wan-Ting Chang

Fetal Medicine Center, Taiji Clinic, Taipei, Taiwan

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KEYWORDS
anal atresia,
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target sign

Abstract Anal atresia can be divided into high type and low type depending on the relationship between the distal rectal pouch and the puborectalis muscle. Prenatal diagnosis of anal atresia is very challenging. Indirect findings include dilated distal bowel segments and calcified intraluminal meconium in 2nd & 3rd trimester. Direct findings include no PAMC (perianal muscular complex) and no target sign (hypoechoic anal sphincter and echogenic anal mucosa). PAMC is intact in low atresia, no PAMC can only be applied to high atresia. A visible echogenic anal mucosa excludes all cases of high atresia and most cases of low atresia, with the exception of the mildest cases with only a thin membrane covering the anal opening.

A 38-year-old (gravida 1, para 0) woman was referred to our clinic because of abnormal findings of fetal ultrasound. Ultrasound examination at 20 weeks' gestation at other clinic revealed an abdominal mass of unknown origin. However, after detailed second-trimester anatomical scanning at 21 weeks' gestation, we found the previously described abdominal mass turned out to be a dilated colon loop. In addition to colon dilatation 1.09 cm (Fig. 1), we also discovered absent anal pit in the uro-genital region (Fig. 2), single umbilical artery and aberrant right subclavian artery (ARSA). Anal atresia was highly suspected then. We followed the fetus at 25 weeks' gestation, and the fetal stomach could not be well expanded at this time. After consultation with geneticist, the patient opted against further genetic testing. The baby was born at 40 weeks' gestation, and high-type anal atresia was confirmed (Fig. 3). Nevertheless, esophageal atresia with trachea-esophageal fistula was also noticed. The newborn underwent esophageal re-anastomosis surgery

Conflicts of interest: The authors have no conflicts of interest relevant to this article.

* Correspondence to: Yu-Ching Chou, Fetal Medicine Center, Taiji Clinic, 162, Section 2, Chung-Shan North Road, Taipei 104, Taiwan. Fax: +886 2 25953710.
E-mail address: yuching.chou@fetalmedicine.tw (Y.-C. Chou).

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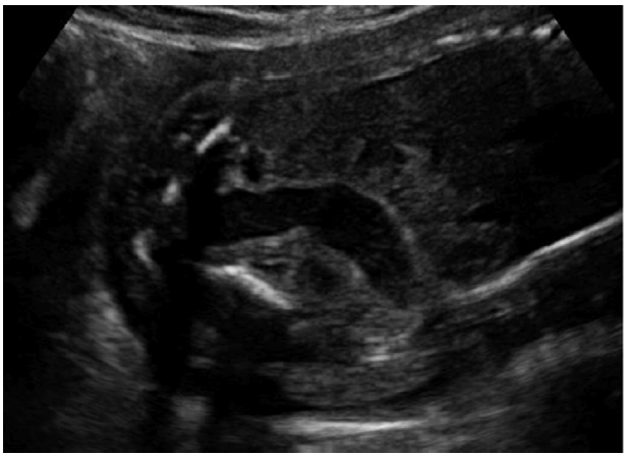

Figure 1 Colon dilatation.

and colostomy soon after birth. The baby is now 11 months old and is doing very well.

Prenatal diagnosis of esophageal atresia is very challenging since more than 90% of esophageal atresia cases are associated with tracheo-esophageal fistula. The fetal stomach can still be filled with amniotic fluid bypassing from the trachea. The only hint to in-utero diagnosis is a "small" gastric bubble. However, there is no defined measurement for fetal gastric bubbles [1]. The fetal stomach appeared to be normally dilated at 21 weeks' scan (Fig. 6). That's why esophageal atresia with trachea-esophageal fistula was missed prenatally in this case.

Anal atresia is a relatively common congenital anomaly, with an incidence of about one in every 1500 to 5000 live births. Most cases are sporadic. Some are associated with trisomy 18 or 21. No associated genetic defects are found.

Anal atresia can be divided into high type and low type depending on the relationship between the distal rectal pouch and the puburectalis muscle [1]. Perianal muscular complex (PAMC), namely the puborectalis muscle, internal anal sphincter and external anal sphincter, is responsible for the continence of the anus [2]. In high anal atresia, the PAMC is poorly developed leaving only internal anal sphincter. In low anal atresia, the entire anal sphincter is intact.

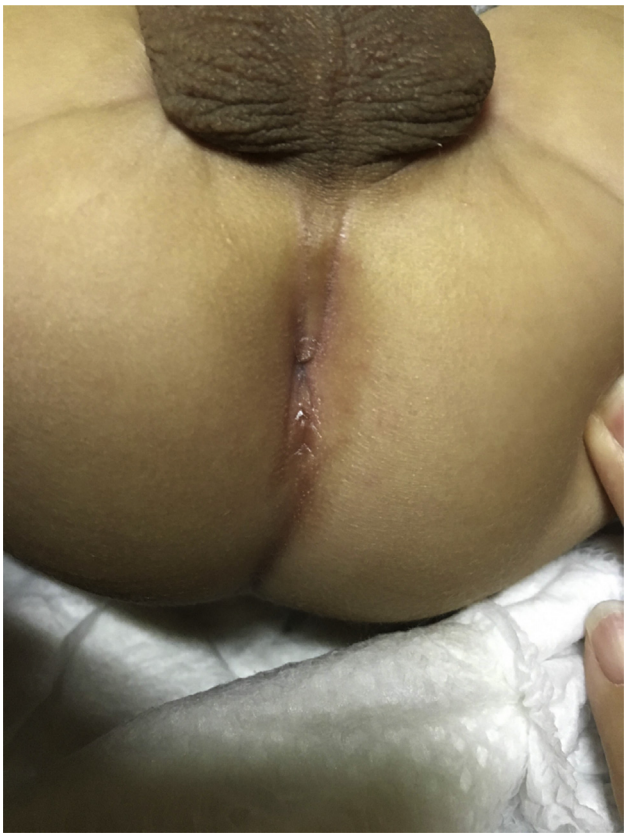

Figure 3 Anal atresia is confirmed postnatally.

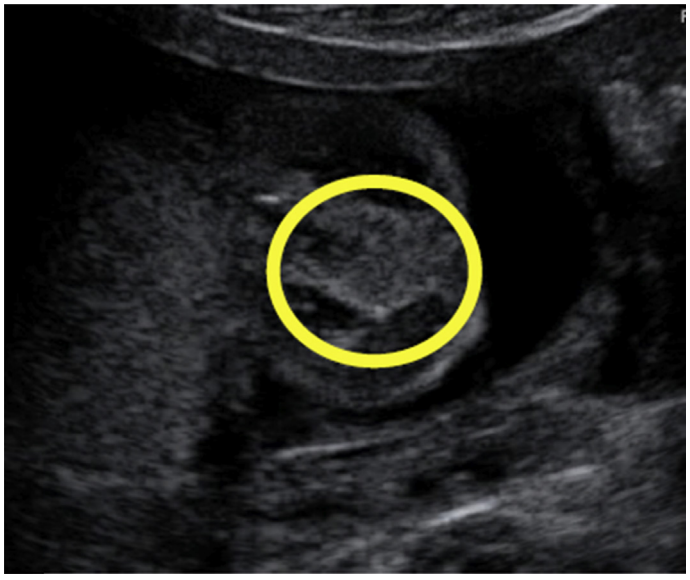

Figure 2 Absent anal pit in the urogenital region.

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