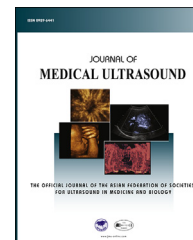


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## LETTER TO THE EDITOR

# Prenatal Diagnosis of Anal Atresia – A Case Report



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### KEYWORDS

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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.

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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.

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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.

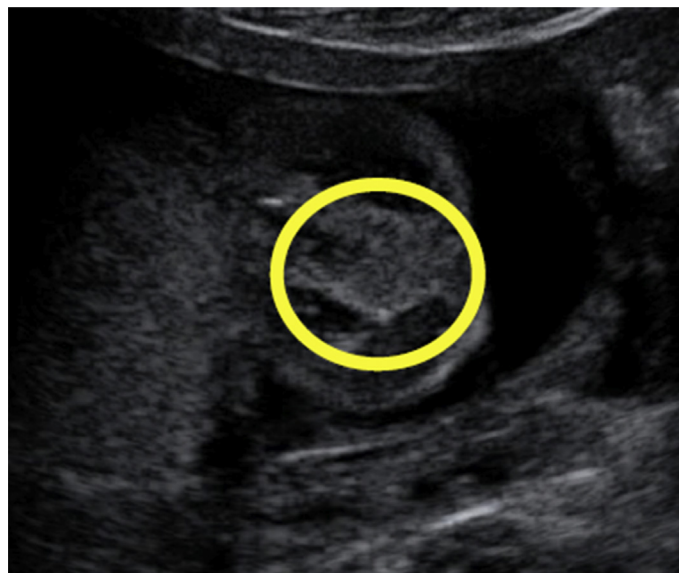
Anal atresia is a relatively common congenital anomaly, with an incidence of about one in every 1500 to 5000 live births. Most cases are sporadic. Some are associated with trisomy 18 or 21. No associated genetic defects are found.

Anal atresia can be divided into high type and low type depending on the relationship between the distal rectal

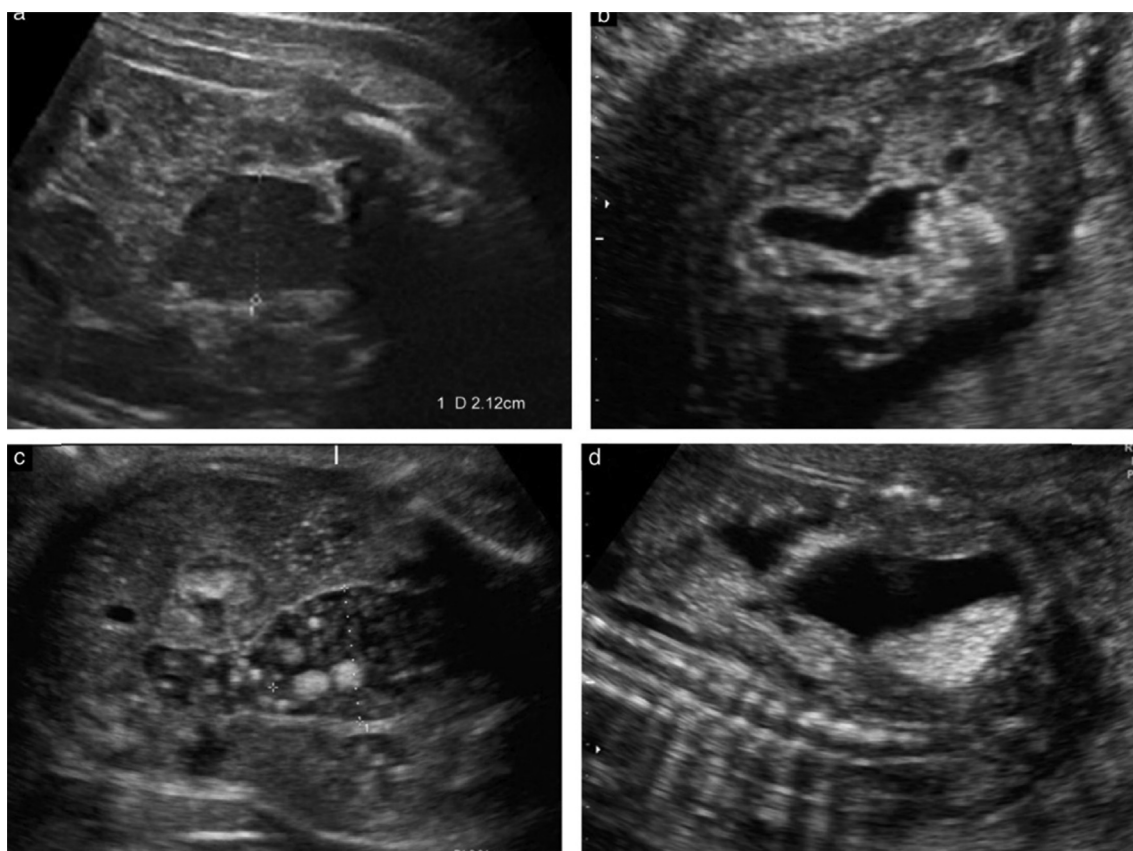


**Figure 3** Anal atresia is confirmed postnatally.

pouch and the puborectalis 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 2** Absent anal pit in the urogenital region.

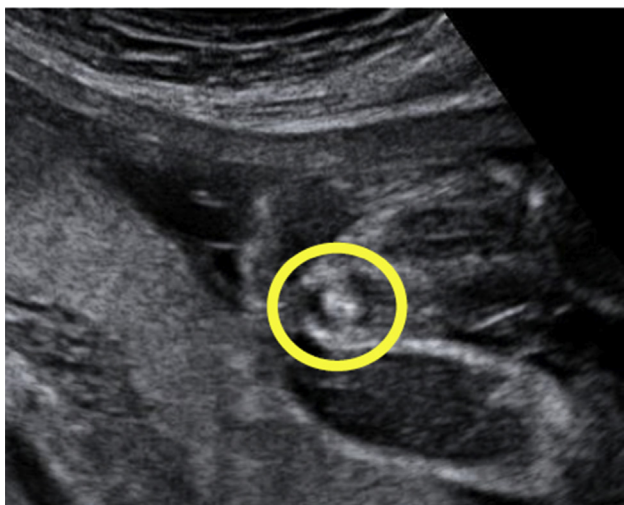


**Figure 4** Echogenic meconium in bowel lumen by Ochoa et al. [2].

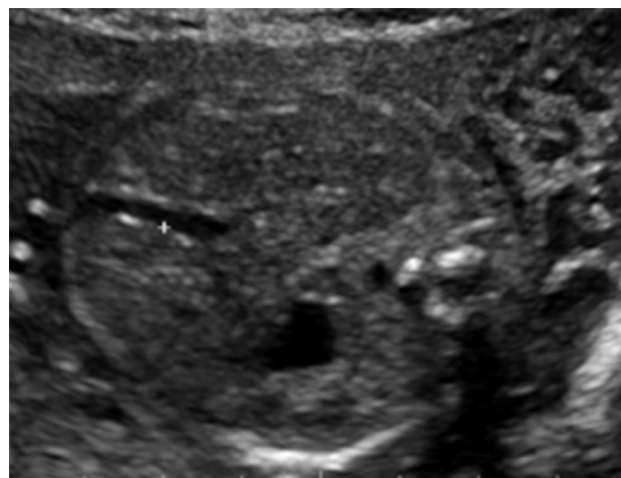
Prenatal diagnosis of anal atresia is very challenging given the wide spectrum of anomalies. There is no golden standard but some indirect as well as direct findings exist.

Indirect findings include dilated distal bowel segments, and calcified intraluminal meconium or enterolithiasis in 2nd & 3rd trimester (Fig. 4) [2]. Direct findings include no PAMC and no target sign (anal sphincter and anal mucosa) (Fig. 5).

The dilatation of distal bowel segment may be transient. Sometimes the bowels may appear completely normal prenatally. Calcified intraluminal meconium is caused by



**Figure 5** Normal anal pit. Yellow circle indicates the typical target sign: hyperechoic anal mucosa surrounded by hypoechoic anal sphincter.



**Figure 6** Normally dilated fetal stomach.

alkalization of fetal meconium by urine flow from rectourethral or rectovesical fistula. However, this finding is not frequently encountered [3].

PAMC is the hypoechoic ring-like structure surrounding the echogenic anal mucosa. PAMC is intact in low atresia, so diagnosing anal atresia by detecting PAMC can only be applied to high atresia. While trying to look for “target sign”, it is important to make sure to scan at a level where neither the pelvic bones nor the femur head should be visible. Ischial tuberosity could be erroneously diagnosed as anal mucosa under wrong scanning planes. 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 [4].

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