


Sonographic Depiction of the Fetal Anus and Its Utility in the Diagnosis of Anorectal Malformations

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Objectives—The purpose of this study was to describe the sonographic appearance of the fetal anus and its usefulness in diagnosis of anal atresia.

Methods—An anomaly scan was performed in 13,150 patients over 8 years. Gestational ages ranged from 16 to 38 weeks. A tangential scan of the fetal perineum was performed. The anus was seen as a hypoechoic ring representing the wall of the anal canal with a central echogenic dot representing the lumen. This appearance was seen posterior to the external genitalia. If there was failure to see this appearance, a coronal scan of the fetal pelvis was done to look for the anal canal in contiguity with the rectum and to confirm its extension up to perineum. Failure to see these two features was considered diagnostic of anal atresia.

Results—On the basis of these criteria, anal atresia was diagnosed in 17 fetuses. Sixteen of these fetuses also had other associated anomalies. The appearance of the anus was useful for ruling out anal atresia in 2 fetuses with a dilated colon containing echogenic meconium balls.

Conclusions—The anus is visible on a tangential scan of the fetal perineum. It can be seen routinely during an anomaly scan to diagnose or rule out anal atresia.

Key Words—anomaly scan; anorectal malformation; fetal anus; sonography

Advances in ultrasound technology and techniques have increased the resolution of fetal anatomy. More and more fetal structures are detected. The fetal anus, the visualization of which is crucial to the diagnosis of anorectal malformations, is one of them.¹ Our aim was to evaluate the performance of an anomaly scan to visualize the fetal anus and its role in the diagnosis of anorectal malformations and associated anomalies.

Materials and Methods

We prospectively evaluated visualization of the fetal anus in all pregnant women at 16 to 38 weeks' gestation who visited our unit for prenatal sonography over 8 years up to January 2010. The gestational age was calculated from the last menstrual period or sonographic biometric measurements as appropriate. This study was approved by the Ethics Committee of our region. At our unit, informed consent was obtained routinely before sonography for future analysis.

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Abbreviations

OEIS, omphalocele, exstrophy, imperforate anus, and spinal defects; URSM, urorectal septal malformation; VACTERL, vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal malformations, and limb defects

Sonography was performed by 3 physicians using HDI 5000 and iU22 (Philips Healthcare, Bothell, WA) and Voluson 730 (GE Healthcare, Milwaukee, WI) scanners equipped with convex 2- to 5- and 4- to 7-MHz and linear 5- to 12-MHz transducers. Visualization of the fetal anus was done as a standard part of the prenatal sonographic examination. An axial section of the fetal pelvis with the urinary bladder and sacral spine was obtained with a convex 2- to 5-MHz probe (Figure 1A). Then the transducer was moved caudally in a sliding fashion until a tangential section of the fetal perineum was seen (Figure 1, B and C). The perineum was seen between the two gluteal bulges. The region posterior to the fetal genitalia was seen in this section, and on finer movements of the probe, the fetal anus was seen as a hypoechoic ring with an echogenic center (Figure 1C and Video 1). When there was any difficulty or variation in visualization of the fetal anus, sagittal and coronal scans of the fetal pelvis and perineum were performed. In these sections, the rectum was seen as a fluid-filled structure with mucosal folds. The anus was seen as a hypoechoic tubular structure with a central echogenic stripe. Rarely, a high-frequency transducer was used in a similar fashion (Figure 2). Failure to visualize the fetal anus in both sections was taken as a sign of an anorectal malformation (Figure 3 and Video 2).

Results

The fetal anus was assessed prospectively in 13,290 fetuses at 16 to 38 weeks' gestation. In 20 patients, the fetal perineum could not be visualized because of severe oligohydramnios or a gross fetal anomaly distorting the perineum such as sirenomelia or sacrococcygeal teratoma. One hundred twenty fetuses were lost to follow-up. These patients were excluded from the study, leaving 13,150 fetuses in whom the fetal perineum was visualized satisfactorily. The distribution of fetuses by gestational age is given in Table 1.

The fetal anus could be visualized in 13,133 fetuses and could not be visualized in 17. There were associated anomalies in 16 of them, which are shown in Table 2. There were 3 fetuses with a dilated colon containing echogenic meconium balls, suggestive of an anorectal malformation with a colourinary fistula. The perinea of the abortuses and neonates were examined for the presence or absence of the anus after termination of pregnancy or delivery. The absence of an anal opening was confirmed in all 17 cases in which prenatal sonography failed to show a normal fetal anus. Among 16 cases in which termination of pregnancy was done, autopsy find-

Figure 1. Visualization of the fetal anus. The transducer is moved down from an axial section of the pelvis (A) through the urinary bladder (BL) and sacral spine to the lower pelvis (B) and then a tangential section of the perineum showing the anus (C, arrows).



Figure 2. A. Sagittal high-resolution scan showing the fetal anus as a hypoechoic tubular structure with a central echogenic stripe (arrowheads). A indicates anterior; arrow, uterus; BL, urinary bladder; P, posterior; R, rectum; S, sacrum; and V, vagina. **B.** Coronal high-resolution scan showing the fetal anus (arrows) and rectum.

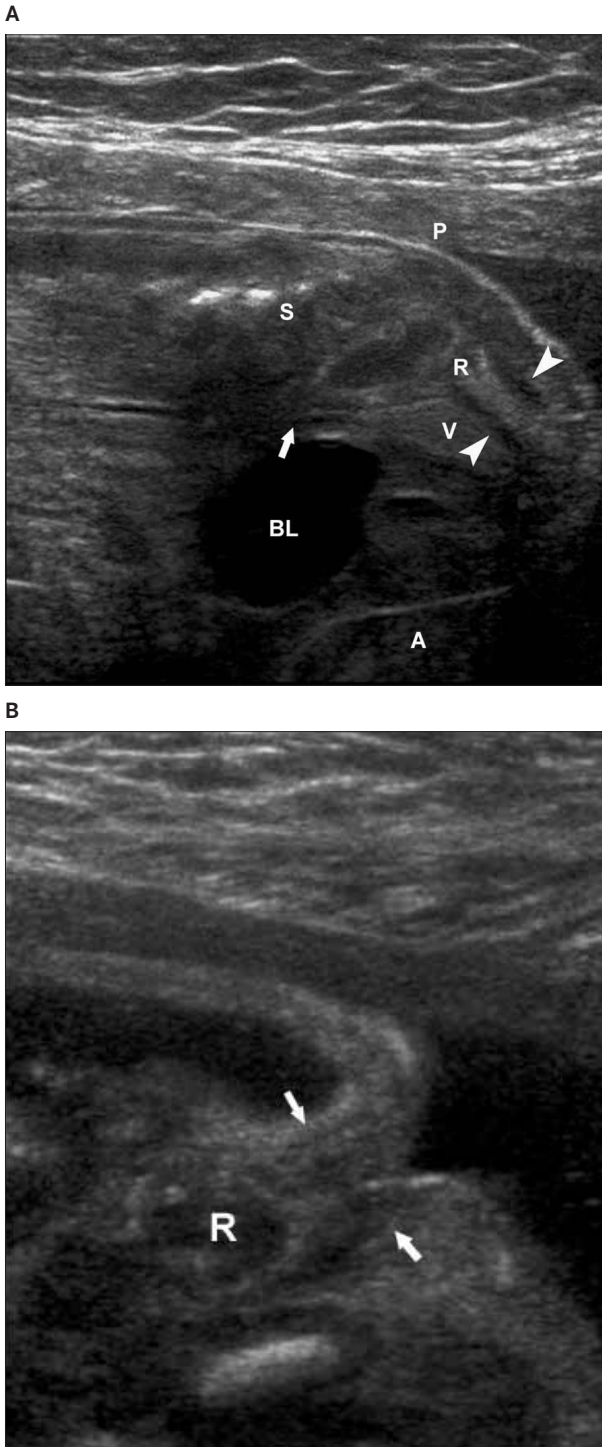
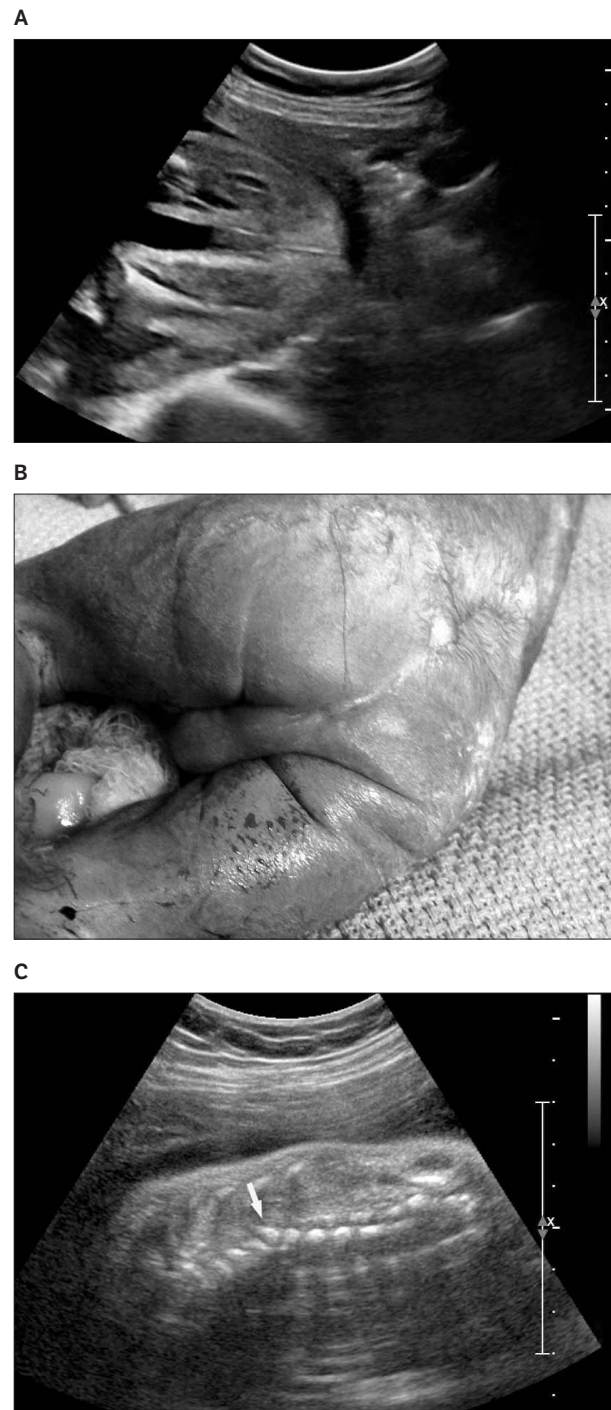


Figure 3. Failure to see the fetal anus in a fetus with the VACTERL association (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal malformations, and limb defects). **A.** Tangential scan of the perineum showing the midline echogenic stripe of the perineum with a nonvisualized anus. **B.** Perineum of the abortus with anal atresia. **C.** Coronal scan of the fetal spine showing the hemivertebra (arrow).



ings were available in 4. In the cases in which a normal anus was visualized on prenatal scans, the normalcy of the anus in the neonate or abortus was confirmed by a telephone conversation with either the obstetrician or the parents. The anus was normal in all such cases except 1, which had a covered anus.

Discussion

An imperforate anus is caused by a defect or an arrest in the development of the hindgut and is a relatively common congenital anomaly, with a frequency of approximately 1 to 4 cases per 5000 live births.² Although the condition is generally treated neonatally by surgery, prenatal recognition of an imperforate anus is important because it is frequently associated with other congenital and chromosomal abnormalities such as trisomy 21. Although its prenatal diagnosis has been reported,^{3,4} an imperforate anus is usually underdiagnosed until after birth, mainly because of the difficulty in sonographic depiction of the fetal anus. Most reports on the prenatal diagnosis of an imperforate anus have focused on the sonographic identification of a dilated distal bowel or rectum. Harris et al³ achieved prenatal diagnosis of an imperforate anus in 42% of cases (5 of 12), whereas Brantberg et al⁴ prenatally diagnosed 15.9% of cases (11 of 69) cases by this means. A possible mechanism for a dilated distal bowel or rectum is the concurrent

presence of an enterourinary fistula, which causes fetal urine reflux into the distal bowel. However, in some fetuses an imperforate anus is not associated with an enterourinary fistula, and in others an enterourinary fistula is present but not patent. This may explain why an imperforate anus is usually underdiagnosed using prenatal sonographic depiction of a dilated distal bowel or rectum. Therefore, the accuracy of prenatal sonographic diagnosis of an anorectal malformation can be improved by visualization of the fetal anus.¹ This approach was attempted by Guzman et al,⁵ who compared the fetal anuses between a healthy twin and the other twin with features of the VACTERL association (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal malformations, and limb defects). Recently, Moon et al¹ described the in utero development of the fetal anal sphincter. They showed that prenatal sonographic depiction of the fetal anal sphincter is consistently possible in healthy fetuses at 23 to 34 weeks' gestation and that the fetal anal sphincter grows in a predictable pattern according to gestational age. They did not study its usefulness in the diagnosis of an anorectal malformation.

In this article, we have shown that the fetal anus is consistently seen in all fetuses from 16 to 38 weeks' gestation. The fetal anus is seen on prenatal sonography as a hypoechoic ring with an echogenic center on a tangential scan of the fetal perineum. This appearance is produced by the circular fibers of the internal anal sphincter. It is similar to the appearance of the anal canal in adult women on transverse images obtained with a transvaginal transducer placed in the distal vagina.⁶ Failure to visualize the fetal anus in axial and sagittal/coronal views is diagnostic of an anorectal malformation, as seen in the 17 of our cases. In 1 case, the anal atresia was isolated, and the remaining 16 had an associated anomaly. The associated anomaly was either a complete or partial urorectal septal malformation (URSM) sequence in 12 cases. Complete URSM consists of the absence of the perineal and anal openings (Figure 4).⁷ In partial URSM, there is a single perineal opening that serves as an outlet for a common cloaca and a conduit for urine and feces to the outside (Figures 5 and 6).⁷ Both are usually associated with ambiguous genitalia. It was not possible to indicate partial or complete URSM on prenatal sonography. In this study, there were 11 cases of partial URSM. Of these, 3 had only a urinary tract anomaly, and 1 had only intracolonic calcification. In these 4 fetuses, URSM could be diagnosed because of nonvisualization of the anus. In 3 cases of anal atresia, an indication of the VACTERL association could be given because of an associated spinal deformity or esophageal atresia. In this study, there were 14 fetuses with a spinal anomaly. The fetal anus was not seen in 2, indi-

Table 1. Distribution of Fetuses by Gestational Age

Gestational Age, wk	n	%
18–22	9,205	70
23–30	2,630	20
>30	658	5
<18	657	5
Total	13,150	100

Table 2. Fetuses in Whom the Anus Was Not Visualized

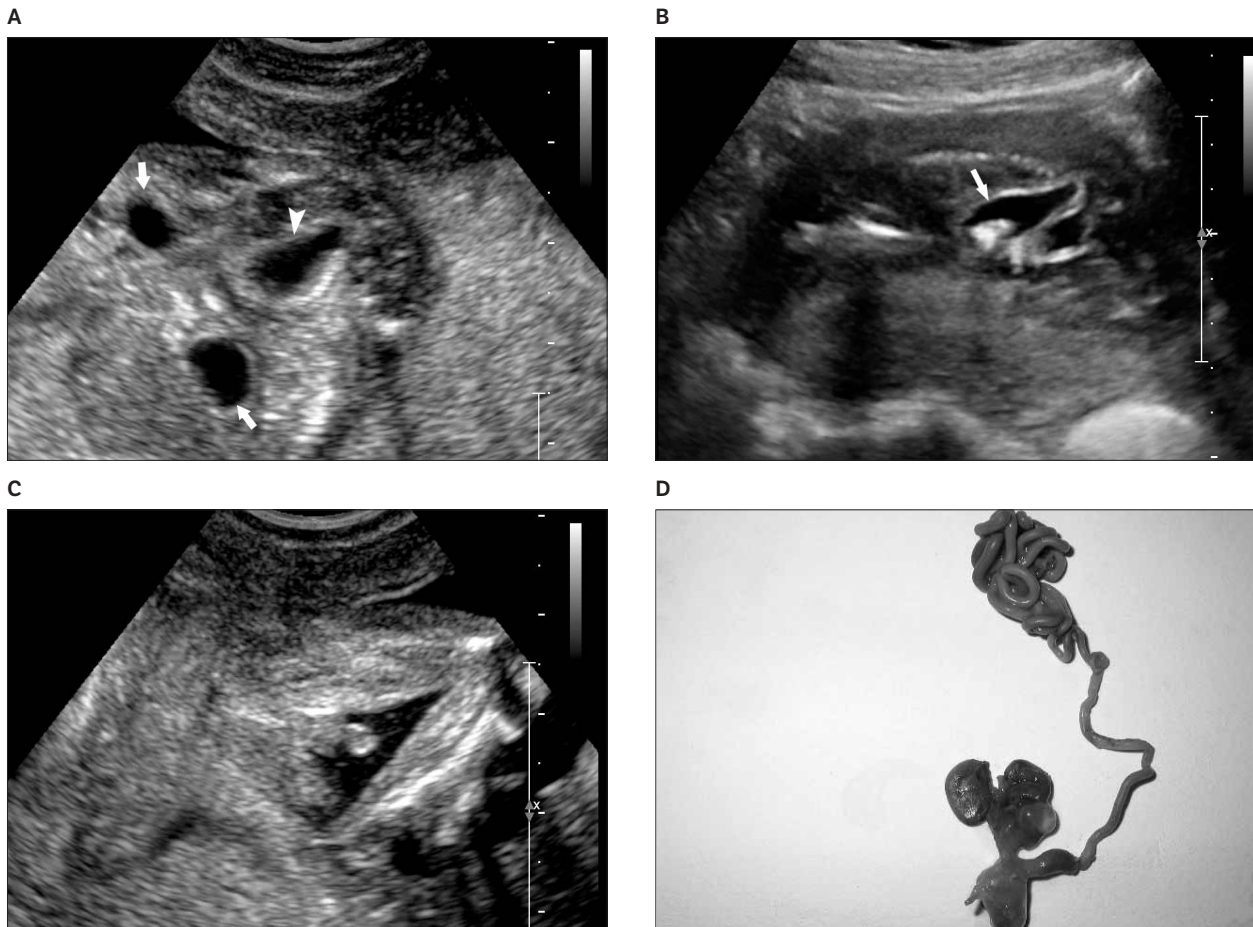
Fetuses	n
Total	17
Isolated nonvisualization	1
Associated anomalies	16
Complete URSM	1
Partial URSM	11
VACTERL	3
OEIS complex	1

OEIS indicates omphalocele, exstrophy, imperforate anus, and spinal defects; URSM, urorectal septal malformation; and VACTERL, vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal malformations, and limb defects.

cating the VACTERL association (Figure 7). In the other 12 fetuses, a normal fetal anus was seen, making this association unlikely. In 1 case with esophageal atresia, the fetal anus was not seen, again indicating the VACTERL association. In 1 case in which the fetal anus was not seen, there were other features of the OEIS complex (omphalocele, exstrophy, imperforate anus, and spinal defects; Figure 8). The OEIS complex is a condition with major criteria of an infraumbilical abdominal wall defect, omphalocele, myelomeningocele, an imperforate anus, and a nonvisualized bladder and minor criteria of lower extremity abnormalities, renal malformations, ascites, hydrocephalus, and a single umbilical artery. In this study, 1 patient had a previous neonate with an anorectal malformation, and the fetus of the subsequent pregnancy showed a normal anus, which was reassuring for the couple.

Intraluminal meconium calcification or enterolith is another prenatal sonographic feature of an imperforate anus. In a summary of cases with prenatal intraluminal enterolithiasis, Pohl-Schickinger et al⁸ found that all 10 had an imperforate anus on postnatal physical examination. The pathogenesis of intraluminal enterolithiasis is not well known. It is possible that prolonged stasis of a mixture of fetal urine and meconium might cause calcification in the bowel of fetuses with an enterourinary fistula. In our study, 5 fetuses had intracolonic meconium calcification. The finding was isolated in 3 fetuses, whereas the others had associated urinary tract anomalies. Among the 3 fetuses with this finding as an isolated feature, the fetal anus was not seen in 1, indicating an anorectal malformation, which was confirmed postnatally (Figure 9). In the other 2 cases, a normal fetal anus was shown on prenatal sonography,

Figure 4. Complete urorectal septal malformation sequence. **A.** Coronal scan of the fetal abdomen showing bilateral gross hydronephrosis (arrows) with a thick-walled conical cystic structure (arrowhead) in the pelvis and oligohydramnios. **B.** Oblique scan of the lower abdomen showing a dilated distal colon (arrow) with echogenic calcified meconium. **C.** Tangential scan of the perineum showing a nonvisualized anus. **D.** Common cloacal pouch to which the ureters and rectum are connected.



excluding a diagnosis of an anorectal malformation, and confirmed as normal postnatally (Figure 10). The meconium was reported to be granular in these cases, and no further investigations were performed. In 1 case in which the fetus showed penoscrotal transposition, a normal fetal anus was visualized on prenatal sonography. Postnatally, the neonate had a covered anus, which is expected in this condition because the sphincter complex is present up to the skin but is covered by the skin.

We have shown that the fetal anus can be consistently depicted on prenatal sonography. Failure to visualize the fetal anus is diagnostic of an anorectal malformation. It is useful to determine whether a spinal anomaly is isolated or part of the VACTERL association and whether omphalocele is isolated or a part of the OEIS complex. Visualization of the fetal anus is useful for reassuring couples who have had a previous neonate with an anorectal malformation. Finally, it helps in determining whether a fetus with colonic intraluminal meconium calcification has an anorectal malformation.

Figure 5. **A** and **B**, Coronal scans of the fetal abdomen showing gross hydronephrosis of the right kidney (**A**) and agenesis of the left kidney (**B**, arrow). **C**, Scan of the perineum showing a nonvisualized anus. **D**, Neonate with anal atresia.

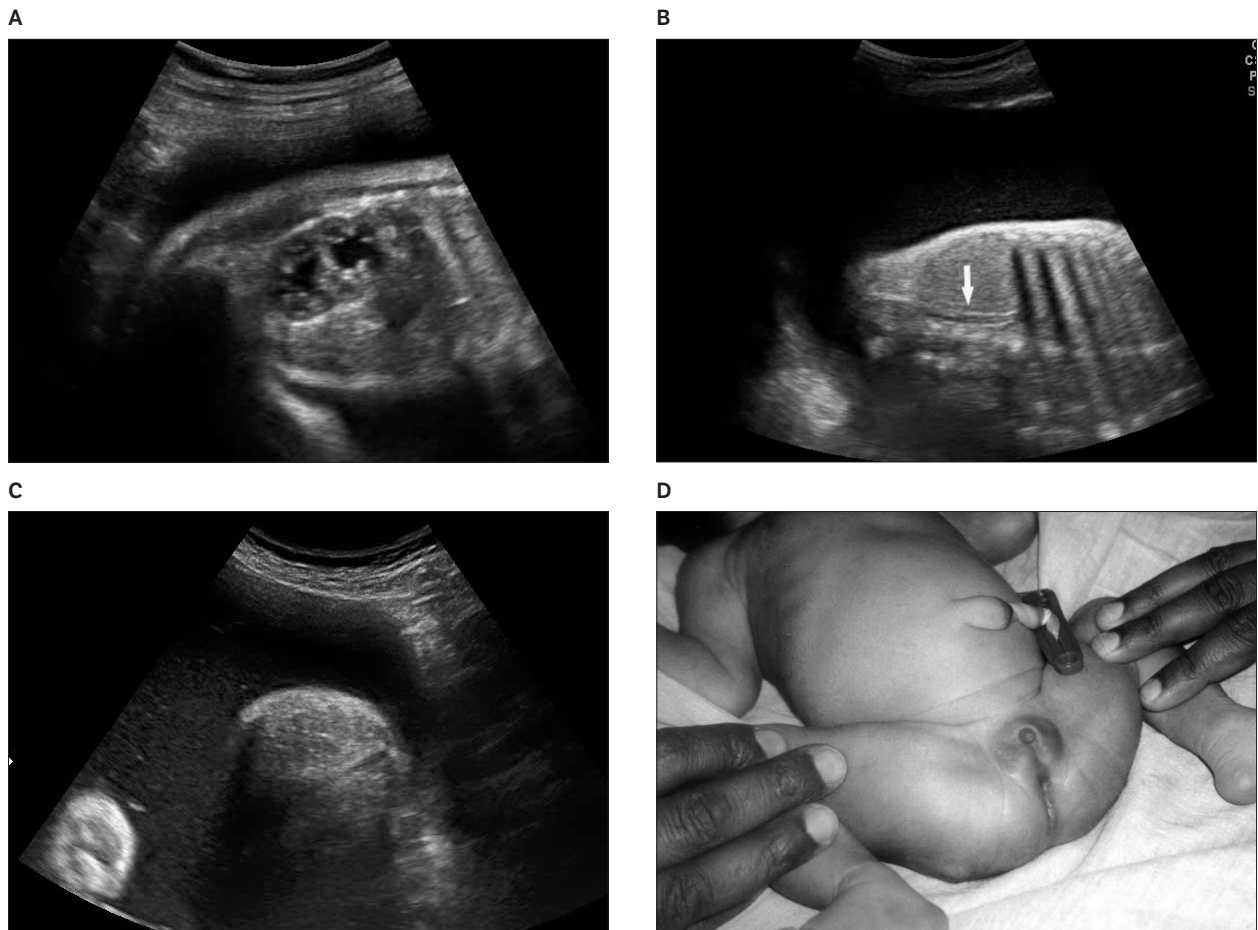


Figure 6. **A**, Axial scan showing bilateral hydronephrosis. **B**, Pelvis and urinary bladder (BL). **C**, Scan of the perineum showing ambiguous genitalia and absence of the anus. **D**, Abortus confirming the findings in **A–C**.

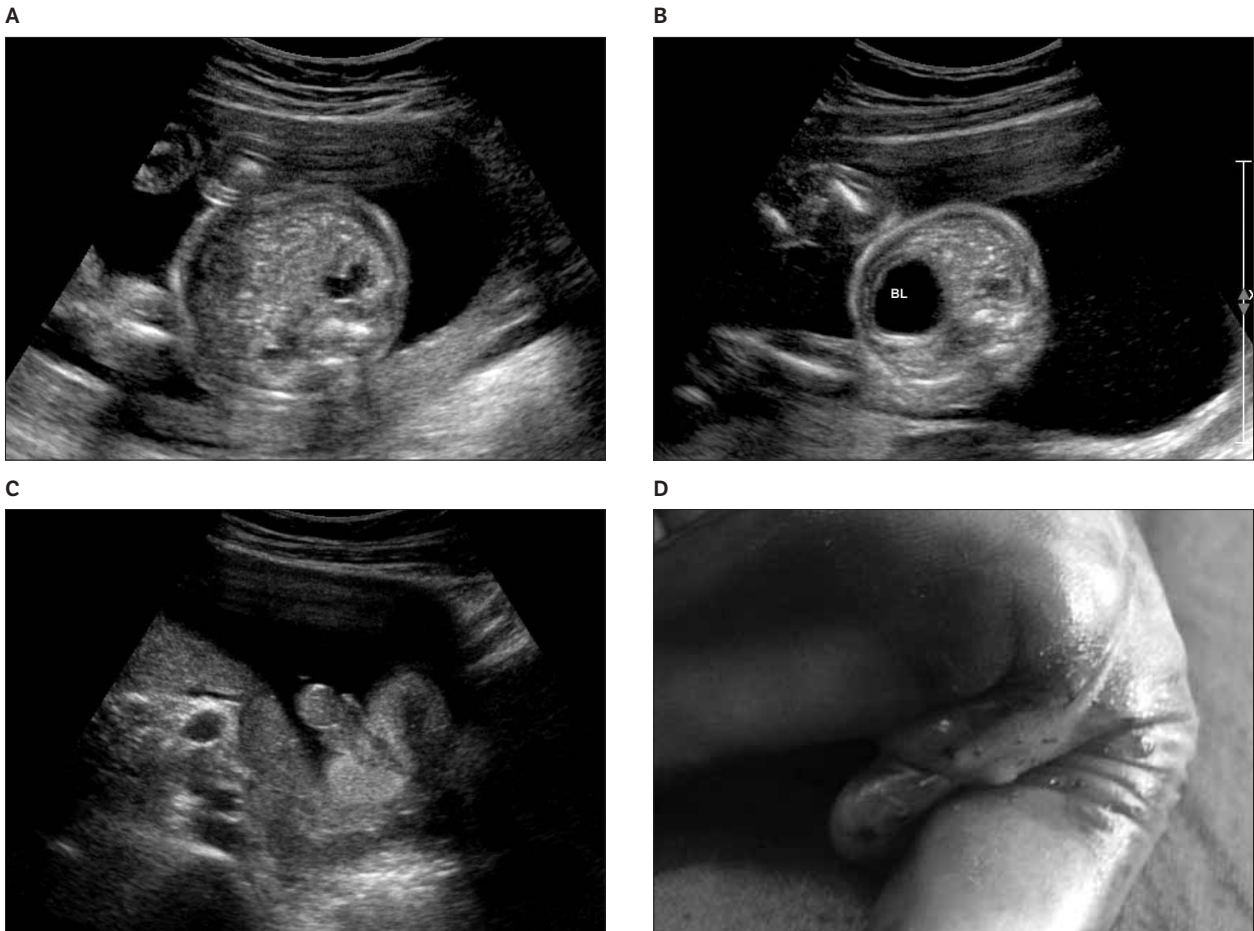


Figure 7. Fetus with the VACTERL association (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal malformations, and limb defects). **A**, Agenesis of the kidney (arrow). **B**, Scan of the perineum showing absence of the anus.

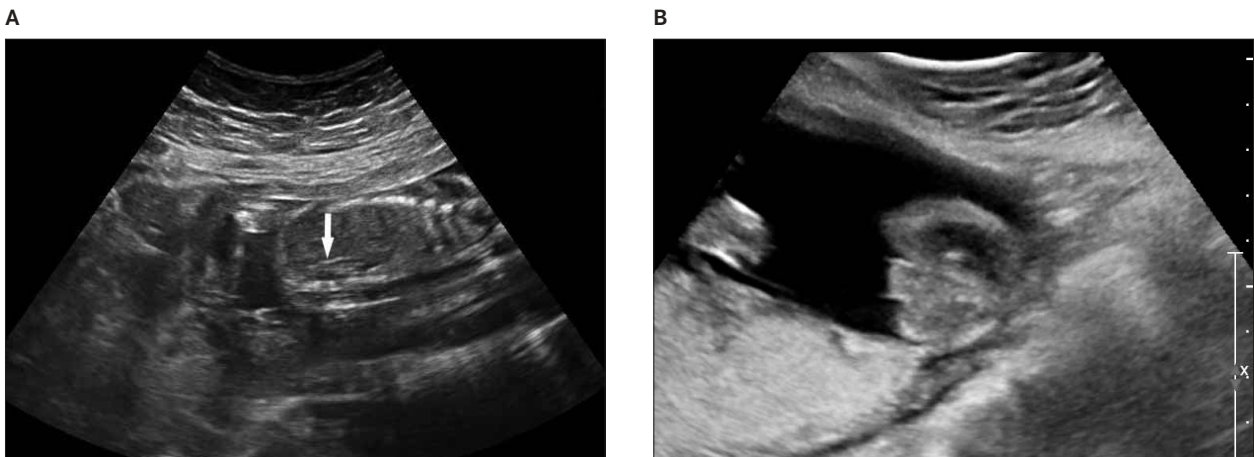


Figure 8. Fetus with the OEIS complex (omphalocele, exstrophy, imperforate anus, and spinal defects). **A.** Omphalocele (arrows). **B.** Spinal anomaly with meningocele (arrows). **(C)** Scan of perineum showing absence of the anus. **(D)** Abortus confirming the features shown in **A–C**.

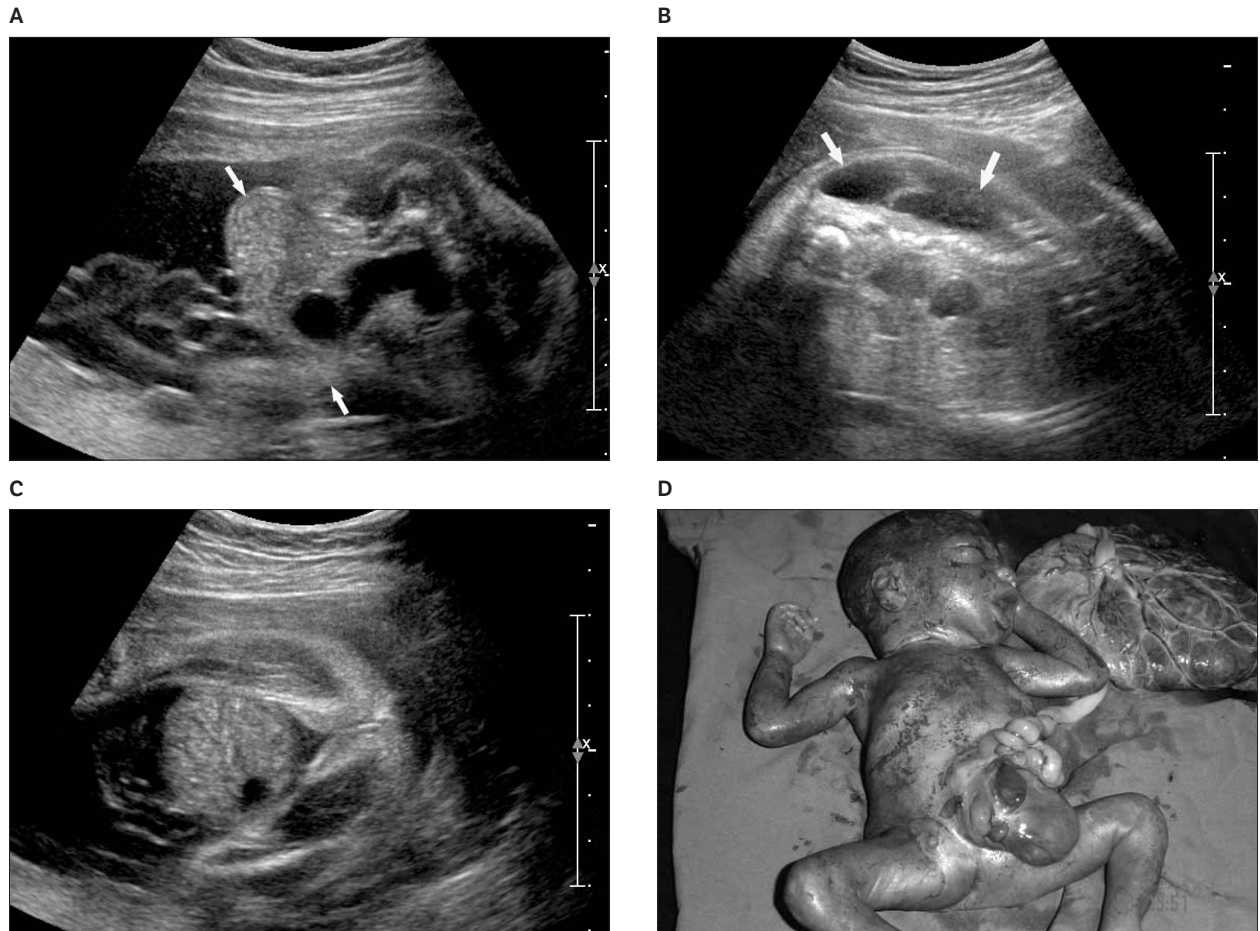


Figure 9. **A.** Axial scan showing a dilated colon with calcified meconium balls (arrows). **B.** Scan of perineum showing absence of the anus, indicating anal atresia.

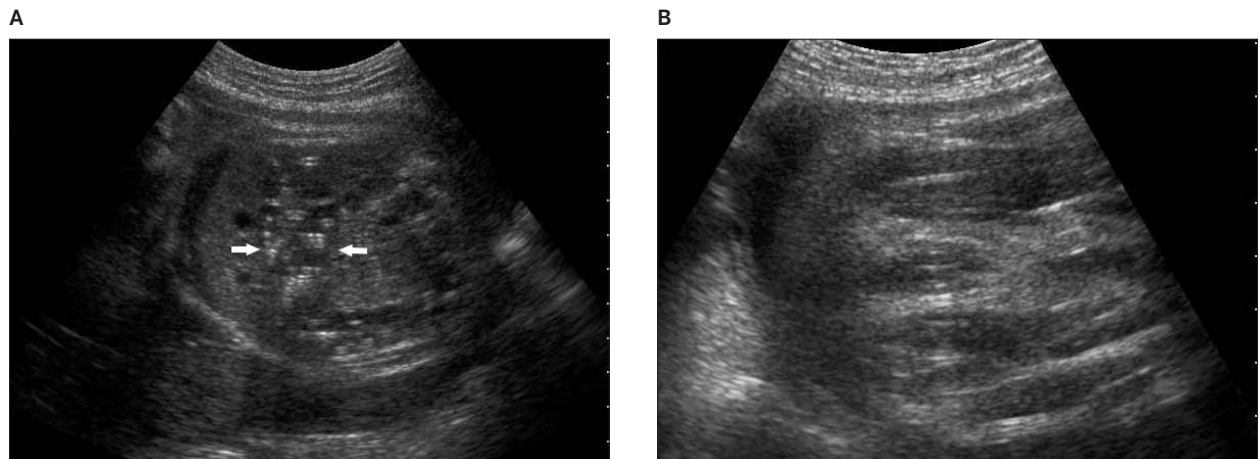
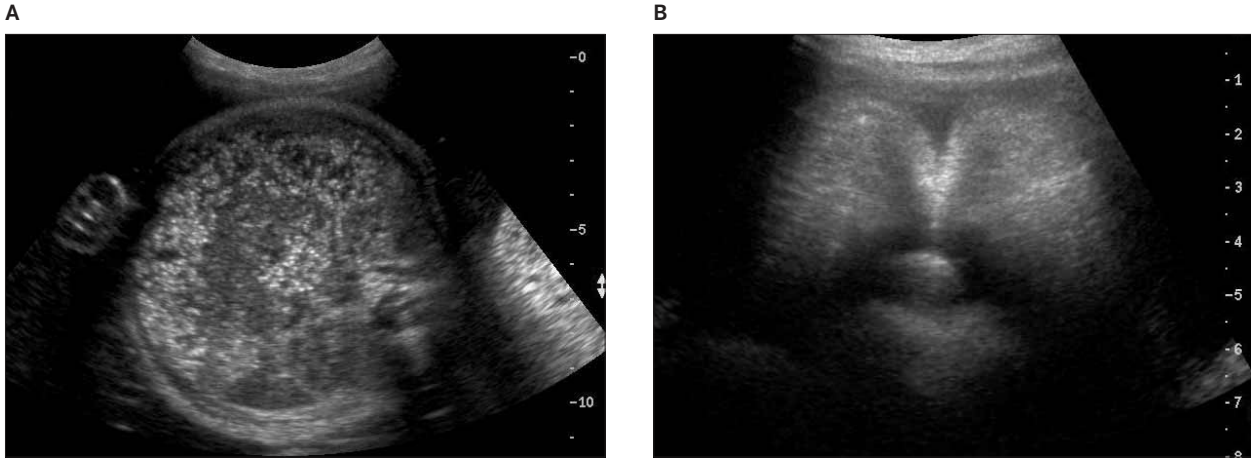


Figure 10. A, Axial scan showing a dilated colon with echogenic meconium balls. **B,** Scan of the perineum showing a normal anus, ruling out anal atresia.



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