Novel Approach for the Sonographic Evaluation of the Anorectal Anatomical Structure of Fetuses

Takashi Kaji

ABSTRACT

Imperforate anus (IA) is a relatively common gastrointestinal anomaly, usually requiring prompt assessment and treatment after birth. Moreover, >50% of IA cases are associated with other congenital or chromosomal abnormalities. Therefore, the prenatal diagnosis of IA is considered ideal for optimizing neonatal treatment. Imperforate anus had rarely been diagnosed prenatally because of the absence of effective screening and diagnostic methods. Although the prenatal diagnosis of IA remains uncommon, cases of IA diagnosed prenatally have been increasing due to recent advances in the ultrasonographic evaluation of the anorectal anatomical structure of fetuses. A specific sonographic sign, absent or abnormal appearance of the anal sphincter muscles and anal canal mucosa in the tangential view of the fetal perineum, allows for the prenatal diagnosis of IA. High- and intermediate-type IAs are increasingly detected prenatally by this sign, while the prenatal diagnosis of low-type IA remains challenging. Several new sonographic techniques showing the precise anorectal anatomy, such as the sagittal view of the fetal pelvis, transvaginal approach, high-frequency linear transducer, and three-dimensional ultrasound, seem promising to further improve the diagnostic accuracy.

Keywords: Fistula, HDlive, Imperforate anus, Sagittal, Three-dimensional, Ultrasound.

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INTRODUCTION

Imperforate anus (IA) is a relatively common congenital anomaly, with an estimated incidence of 1:1,500–1:5,000 live births.^{1–3} Imperforate anus usually necessitates prompt assessment and treatment after birth.^{1,4,5} Moreover, >50% of cases with IA are associated with other congenital or chromosomal abnormalities, such as VATER association and trisomy 21.^{1,5} Accordingly, the prenatal diagnosis of IA is considered ideal for optimizing neonatal treatment.

Imperforate anus had rarely been diagnosed prenatally because of the absence of effective screening and diagnostic methods.⁶ Although the prenatal diagnosis of IA remains uncommon, cases of IA diagnosed prenatally have been increasing due to advances in the sonographic evaluation of the anorectal anatomical structure of fetuses. In this review article, we present recent advances in the sonographic diagnosis of fetal IA.

Nonspecific Sonographic Sign for the Prenatal Diagnosis of IA: An Abnormally Dilated Distal Bowel or Rectum with or without Calcified Intraluminal Meconium

An abnormally dilated distal bowel or rectum with or without calcified intraluminal meconium has been used as an ultrasonographic sign for detecting fetal IA (Fig. 1).^{6–8} The accuracy of such findings for the prenatal diagnosis of IA is severely limited due to the high false-negative and false-positive rates. The distal bowel or rectum in normal fetuses often seems somewhat dilated in the third trimester, which makes it rather difficult to differentiate fetuses with IA from normal ones. Although the etiology of calcified intraluminal meconium has not been fully elucidated, it has been suggested that the admixture of fetal urine and meconium results in calcification.^{7–9} In other words, calcification formation demands the passage of urine into the bowel/rectum through a fistula. Therefore, in the absence of a fistula connecting the bowel/rectum

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and urinary system, calcification does not occur, even in fetuses with IA. Therefore, only 15.9% of IA cases were diagnosed prenatally according to this nonspecific sign: the abnormally dilated distal bowel or rectum with or without calcified intraluminal meconium.⁶

Specific Sonographic Sign for the Prenatal Diagnosis of IA: Absence or Abnormal Appearance of the Anal Sphincter Muscles and Anal Canal Mucosa in the Tangential View of the Fetal Perineum

Recently, visualization of the fetal perineum using two-dimensional (2D) sonography is highly useful for the prenatal diagnosis of IA.^{10–14} The 2D sonography of the fetal perineum can show the ASCM in normal fetuses. The anal sphincter muscles and anal canal mucosa (ASCM) is depicted as a hypoechoic ring with a hyperechoic center in the tangential view of the fetal perineum (Fig. 2).^{10–14} The hypoechoic ring and hyperechoic center are considered to represent the sphincters and mucosa, respectively.^{11,12} The absence or abnormal appearance of the ASCM strongly indicates IA (Fig. 3).^{10–14}

Fig. 1: An abnormally dilated distal bowel (arrows) with calcified intraluminal meconium in a fetus with an imperforate anus

Fig. 2: Tangential view of the fetal perineum in a normal fetus. The anal sphincter muscles and anal canal mucosa are depicted as a hypoechoic ring with a hyperechoic center (arrows)

Figs 3A and B: (A) Tangential view of the fetal perineum in a fetus with an imperforate anus. The anal sphincter muscles and anal canal mucosa are not visualized; (B) Neonatal perineum showing an imperforate anus

PRENATAL DIAGNOSIS AND THE TYPE OF IA

Imperforate anus is classified into three types (high-, intermediate-, and low-type IA) according to the relationship between the distal rectal pouch and the puborectalis muscle.^{4,5} The types of IA are now considered to affect the accuracy of the prenatal diagnosis of IA.^{13,14} Prenatal diagnosis of low-type IA is much more difficult than that of high- and intermediate-type IAs. Several factors, including the presence of an anocutaneous or anovestibular fistula and well-developed anal sphincters, which can be visualized as an image similar to the normal ASCM (Fig. 4), may account for difficulties in diagnosing low-type IA.^{13–15}

In light of the neonatal treatment, the classification of IA is critical because it determines the surgical approach. A low-type IA is generally treated by transperineal anoplasty, whereas high- and intermediate-type IAs are treated initially by diverting colostomy, followed by posterior sagittal anorectoplasty.^{16,17} Regarding the classification of the type of IA, sonographic assessment of the fistula location, which is closely related to the IA type, is now favored for use in neonates with IA because of its high accuracy and the absence of radiation exposure.^{18–21} Kaji et al. recently

reported that the fistula location can be assessed sonographically, even in fetuses.¹⁵

HIGH-FREQUENCY LINEAR TRANSDUCER

We have been using a 3–8-MHz linear transducer that generates images of higher resolution than conventional convex transducers.¹⁵ The high-frequency linear transducer facilitates a clear demonstration of the ASCM. This enables us to differentiate between the internal and external sphincters (Fig. 5). The internal sphincter is depicted as a very hypoechoic and narrow ring, while the external sphincter is depicted as an echogenic ring surrounding the internal sphincter (Fig. 5A). We expect that the precise image of ASCM will lead to an improvement in the prenatal diagnosis of IA, even low-type IA.

Sagittal View of the Fetal Pelvis

As mentioned above, the absence or abnormal appearance of ASCM in the tangential view of the fetal perineum does not always diagnose IA prenatally. Additional visualization of the sagittal or coronal view of the fetal perineum showing the longitudinal

Figs 4A and B: (A) Tangential view of the fetal perineum in the fetus with low-type imperforate anus. Although the hypoechoic region is visible (arrows), the hyperechoic center seems unclear; (B) Neonatal perineum showing an imperforate anus with an anocutaneous fistula

Figs 5A and B: Sonographic images of the anal sphincter muscles and anal canal mucosa using the high-frequency linear transducer in a normal fetus. (A) Tangential view of the fetal perineum. The internal sphincter is depicted as a very hypoechoic and narrow ring, while the external sphincter is depicted as an echogenic ring surrounding the internal sphincter; (B) Sagittal view of the fetal pelvis obtained using the fetal infracoccygeal approach, showing the rectum, anal canal, and internal and external sphincters. ACM, anal canal mucosa; ES, external sphincter; IS, internal sphincter; R, rectum

aspect of the ASCM may further improve the diagnostic accuracy for IA.^{14,15} In particular, the sagittal view obtained using the fetal infracoccygeal or perineal approaches allows us to define the precise anatomy of the rectum, anal canal, internal and external sphincters, and perineum (Fig. 5B). The sagittal view may be the best way to locate fistulas in fetuses with IA.¹⁵

HDLIVE IMAGES OF THE FETAL PERINEUM

Inspection of the perineum is the first and most important step in the diagnosis and assessment of IA after birth. However, 2D ultrasound has difficulties in revealing the surface appearance of the fetal perineum and anus. HDlive is a surface-rendering mode in three-dimensional (3D) ultrasound imaging, which uses an adjustable light source to increase depth perception.^{22,23} Kaji et al. reported that HDlive images can show a realistic appearance of the fetal perineum, similar to those obtained after birth, due to the increase in depth perception.²⁴ HDlive can depict a normal anus as

a dimple (Fig. 6), and also show the absence of the anal dimple in most fetuses with IA (Fig. 7).²⁴

TRANSVAGINAL 2D AND 3D ULTRASOUND

In breech presentation, transvaginal ultrasound can show clear 2D and 3D images of the anal canal and sphincters. In particular, 3D ultrasound can show three orthogonal planes of the anus, which helps us to understand the spatial anatomy of the anal canal and sphincters (Fig. 8).

CONCLUSION

Imperforate anus has been regarded as one of the most difficult gastrointestinal anomalies to diagnose before birth due to the absence of effective screening and diagnostic methods. A specific sonographic sign, an absent or abnormal anal image in the tangential view of the fetal perineum, now allows us to diagnose

Figs 6A and B: HDlive images of the normal perineum in male (A) and female (B) fetuses showing the anal dimples (arrows)

Figs 7A and B: (A) HDlive image of the perineum of a fetus with an imperforate anus showing the absence of the anal dimple; (B) Neonatal perineum showing an imperforate anus

IA prenatally. However, the prenatal diagnosis of some IA, mainly low-type IA, remains challenging. Several new sonographic techniques showing precise anorectal anatomy, such as the sagittal view of the fetal pelvis, transvaginal approach, high-frequency linear transducer, and 3D ultrasound, seem promising in terms of improving the diagnostic accuracy further, which may lead to better treatment planning and parental counseling.

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Figs 8A to C: Transvaginal three-dimensional ultrasound showing three orthogonal planes of the anal sphincter muscles and anal canal mucosa: (A) Sagittal plane; (B) Axial plane; (C) Coronal plane. ACM, anal canal mucosa; ES, external sphincter; IS, internal sphincter; R, rectum

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