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New ultrasound features in diagnosing fetal anal atresia: a multicenter prospective cohort study

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This research aimed to assess the validity of ultrasound scans with new features in detecting fetal anal atresia and verify the effectiveness of these new features. Additionally, we aimed at investigating the perinatal incidence of anal atresia. This multicenter prospective study recruited 94,617 normal gravidas and 84 gravidas with anal atresia fetuses. The gold standard for diagnosing perinatal anal atresia is routine neonatal anus examinations. The incidence calculation was based on the results of the gold standard. The validity of our new approach was evaluated via a diagnostic test involving all 94,701 subjects. The effectiveness of our new features was assessed through an ablation study in a randomly established new dataset, with the ratio of anal atresia to non-anal atresia cases of 1:4. The annual perinatal incidence of anal atresia between 2019 and 2023 ranges from 0.57‰ to 1.29‰. Our new method performed great regarding the Youden index, diagnostic odds ratio (DOR), area under the curve (AUC) of the receiver operating characteristic curve (ROCC), AUC of the precision-recall curve (PRC), F1-score, and Cramer's V. In the ablation study, our new approach surpassed its competitors concerning Youden index, DOR, AUC of the ROCC, and AUC of the PRC. Ultrasound scans show high validity and clinical value in detecting fetal anal atresia. Our new ultrasound features significantly promote the detection of fetal anal atresia.

Keywords Ultrasonography, Anus, Imperforate, Incidence, Diagnostic tests

Abbreviations

AUC	Area under curve
BA	Balanced accuracy
CI	Confidence interval
DOR	Diagnostic odds ratio
FD	First diagnosis
GA	Gestational age
HPMCHCH	Hunan Provincial Maternal and Child Health Care Hospital
LR^+	Positive likelihood ratio
LR^-	Negative likelihood ratio
NPV	Negative predictive value
PPV	Positive predictive value
PRC	Precision-recall curve
ROCC	Receiver operating characteristic curve
YMCHH	Yueyang Maternal and Child Health-care Hospital

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Anal atresia is a congenital anorectal malformation featuring an absence of a normal anus[1]and may accompany a broad spectrum of congenital defects from membranous covering to complex cloacal malformations related to genital or urinary tracts[2–5]. It is rare in low-risk fetuses with a perinatal incidence under 0.40‰[2,6]. However, it can produce numerous poor outcomes, such as high long-term mortality of fetuses and neonates, and urogenital system dysfunction[3]. Commonly, different types of anal atresia need different therapeutic protocols and have different prognoses[7–9]. Nowadays, 84% of the surveyed pediatric surgeons in the Chinese mainland conduct colostomies on high-type anal atresia neonates instead of primary repair[10]. It is crucial to diagnose fetal anal atresia early on and to investigate the perinatal incidence of anal atresia.

Many deficiencies exist in the previous studies on diagnosing fetal anal atresia via ultrasound screening. First, most of the corresponding studies were case reports or case series with low evidence levels[4,11–14]. Second, a few researchers reported non-case series studies pertinent to prenatal ultrasound diagnosis for anal atresia, but the direct ultrasound features they utilized were only "target sign" and "equal sign"[3,15,16]. Third, these studies reported a small fraction of indices for diagnostic tests, and their results cannot be compared owing to the differences in study populations[17,18]. Finally, no such research concerning the anal atresia incidence of Chinese perinatal infants has been published throughout the last 15 years[6].

This research first aimed to assess the validity of ultrasound scans with new diagnostic features in detecting fetal anal atresia via a diagnostic test. It then aimed at comparing new ultrasound features in detecting fetal anal atresia with traditional ones through an ablation study. Additionally, we would like to report the incidence of perinatal anal atresia.

Results

Incidence rates and case numbers of anal atresia

A total of 94,701 gravidas were prospectively enrolled in this research. As depicted in Fig. 1, the annual perinatal incidence of anal atresia between 2019 and 2023 ranges from 0.57‰ to 1.29‰. The overall incidence of perinatal anal atresia between 2019 and 2023 was 0.89‰. The perinatal incidence between 2020 and 2023 is much higher than that of 2019.

Validity assessment of ultrasound scans with new diagnostic features in detecting fetal anal atresia

The prenatal ultrasound screening was feasible for fetal anal canals or anal atresia in 99.41% (95% CI: 99.36-99.46%) of all subjects on both coronal and transversal planes. As shown in Table 1, Table S3, and Fig. 2, the values of validity assessment indices stand high. The point and interval estimations of the Youden index, LR^+, LR^-, DOR, AUC of the ROCC, and AUC of the PRC were 0.940 (95% CI: 0.889–0.991), 22246.259 (95% CI: 8337.129-59360.490), 0.060 (95% CI: 0.025–0.154), 373721.350 (95% CI: 27400.595-5094540.326), 0.970 (95% CI: 0.969–0.971), and 0.904 (95% CI: 0.819–0.951), respectively. The point estimations of F1-score, Cramer's V, accuracy, and BA were 0.946, 0.945, 99.990%, and 97.022%, respectively. The AUC of the ROCC was rated as outstanding diagnostic value, and the Cramer's V as strong association. The LR^+ implied a high diagnostic value.

Ablation analysis on different ultrasound features in diagnosing fetal anal atresia

As indicated in Table 2, our proposed method (major features: "target sign," "equal sign," "funnel sign," "pseudo-target sign," and "line sign"; minor features: colorectal dilatation and enterolithiasis) is the most effective approach with Youden index of 0.86 (95% CI: 0.79–0.93), DOR of 401.63 (95% CI: 142.21-1144.50), AUC of the ROCC of 0.931 (95% CI: 0.902–0.953), and AUC of the PRC of 0.859 (95% CI: 0.767–0.919). Su's and Ochoa's methods are the second best, followed by Lee's method and the indirect signs. Our proposed approach surpassed all the other means significantly in terms of the AUC of the ROCC and the AUC of the PRC.

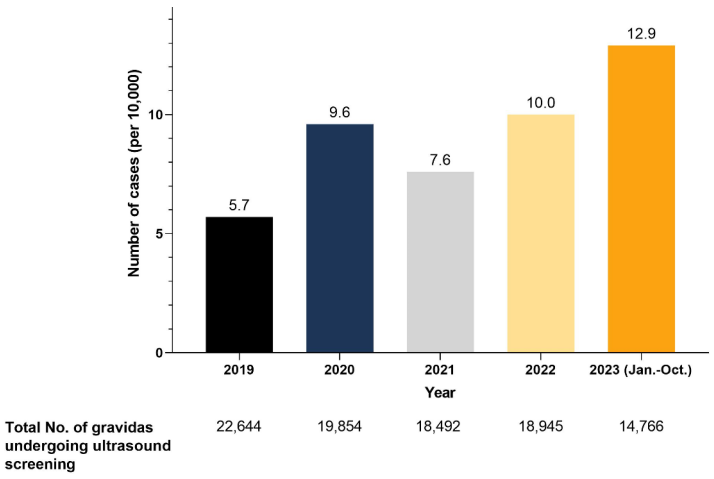

Fig. 1. Annual perinatal incidence of anal atresia.

Evaluation indices	Point estimation	Lower limit of 95% CI	Upper limit of 95 CI
Sensitivity (%)	94.048	86.653	98.039
Specificity (%)	99.996	99.989	99.999
Youden index	0.940	0.889	0.991
LR^+	22246.259	8337.129	59360.490
LR^-	0.060	0.025	0.154
DOR	373721.350	27400.595	5094540.326
AUC of the ROCC	0.970	0.969	0.971
PPV (%)	95.181	88.098	98.138
NPV (%)	99.995	99.988	99.998
AUC of the PRC	0.904	0.819	0.951
F1-score	0.946	-	-
Cramer's V	0.945*	-	-
Accuracy (%)	99.990	-	-
Balanced accuracy (%)	97.022	-	-

Table 1. Validity evaluation of ultrasound scans with new diagnostic features in diagnosing fetal anal atresia through diagnostic tests. AUC, area under curve; CI, confidence interval; DOR, diagnostic odds ratio; LR^+, positive likelihood ratio; LR^-, negative likelihood ratio; NPV, negative predictive value; PPV, positive predictive value; PRC; precision-recall curve; ROCC, receiver operating characteristic curve. * $P < 0.05$.

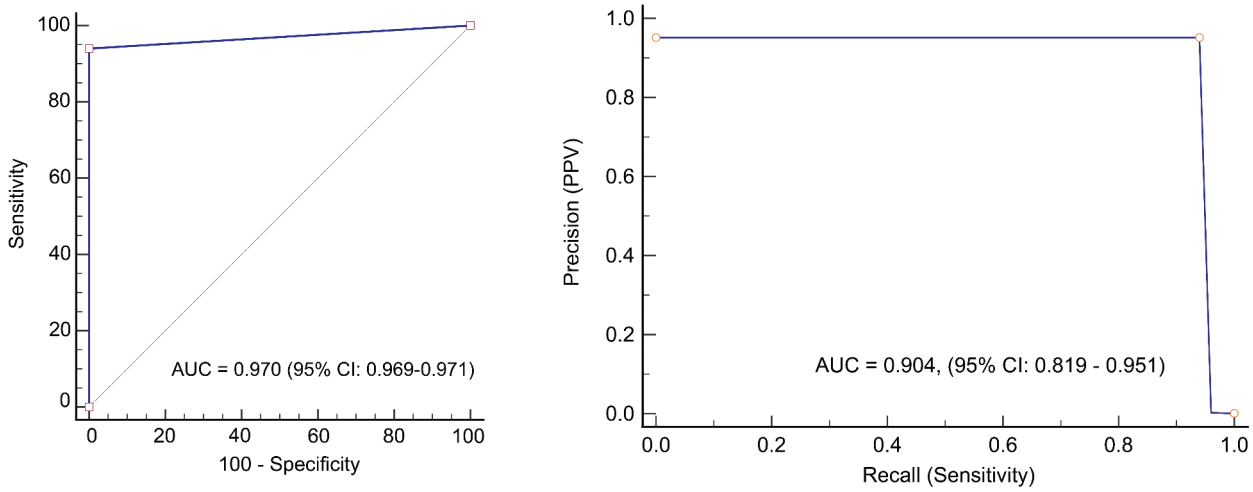

Fig. 2. Validity assessment via receiver operating characteristic curve (**a**) and precision-recall curve (**b**). AUC, area under curve; CI, confidence interval; PPV, positive predictive value.

Discussion

The anal atresia is a rare disease, whether in neonates or fetuses[19,20]. The perinatal incidence of anal atresia approximated 0.32‰ in the Chinese mainland and 0.29‰ in Hunan Province between 2001 and 2005[6]. Our result suggests that the perinatal incidence in Hunan Province from 2019 to 2022 was 0.57‰ to 1.29‰, higher than that between 2001 and 2005. The perinatal incidence between 2020 and 2023 is much higher than that of 2019, which may be associated with COVID-19 infection or the application of the COVID-19 vaccine.

Regarding the validity of ultrasound scans in detecting fetal anal atresia, Su et al[3]. reported in a 63,101-case prospective study, where their sensitivity attained 87.5% by means of the "target sign," "equal sign," colorectal dilatation, and enterolithiasis. Lee's 9,499-case retrospective research[15]documented a sensitivity of 74% and an accuracy of 91% through the "target sign," colorectal dilatation, and enterolithiasis. In a 189-case study of fetuses at high risk for anal atresia, Ochoa et al.[16] acquired a sensitivity of 93%, specificity of 99%, and false positive rate of 7% via the "target sign," "equal sign," colorectal dilatation, and enterolithiasis. Our new approach performed great with a sensitivity of 94.048% (95% CI: 86.653-98.039%), specificity of 99.996% (95% CI: 99.989-99.999%), false positive rate of 5.952%, and false negative rate of 0.004% under the employment of "funnel sign," "pseudo-target sign," "line sign," "equal sign," and "target sign", along with colorectal dilatation, and enterolithiasis. Further, The AUC of the ROCC was rated as an outstanding diagnostic value, the LR^+ as a high diagnostic value, and the Cramer's V as a strong association.

Methods	Ultrasound characteristics							Evaluation indices (point estimation (95% CI))			
	Target sign	Equal sign	Funnel sign	Pseudo-target sign	Line sign	Colorectal dilatation	Enterolithiasis	Youden index	DOR	AUC of the ROCC	AUC of the PRC
Proposed approach [$]	√	√	√	√	√	√	√	0.86 (0.79–0.93)	401.63 (142.21-1144.50)	0.931 (0.902–0.953)	0.86 (0.77–0.92)
Su[3] and Ochoa [17 #]	√	√				√	√	0.74 (0.65–0.83)	99.66 (45.16-219.18)	0.869 (0.833-0.900) [a]	0.74 (0.64–0.82) [d]
Lee [16 #]	√					√	√	0.65 (0.55–0.76)	49.84 (24.83-100.26)	0.827 (0.788–0.862) [b]	0.66 (0.55–0.75) [e]
Indirect signs [#]						√	√	0.43 (0.51–0.57)	13.99 (7.62–25.78)	0.714 (0.669–0.757) [c]	0.47 (0.37–0.58) [f]

Table 2. Ablation analysis of the proposed approach by testing set. DOR, diagnositc odds ratio; AUC, area under curves; ROCC, receiver operating characteristic curve; PRC, precision-recall curve; CI, confidence interval. [$] A case would be considered as anal atresia if any of the "target sign," "equal sign," "funnel sign," "pseudo-target sign," or "line sign" existed. Colorectal dilatation and enterolithiasis are minor features suggesting fetal anal atresia only in coexistence with the aforementioned signs. [#] A case would be considered as anal atresia if any of the ticked features existed. [a] the difference in AUC (ΔAUC) compared to the proposed approach is 0.06, the 95% CI ranges from 0.03 to 0.10, and the P value is below 0.001 according to the DeLong method;. [b] the ΔAUC compared to the proposed approach is 0.10, the 95% CI ranges from 0.03 to 0.15, and the P value is below 0.001 according to the DeLong method;. [c] the ΔAUC compared to the proposed approach is 0.22, the 95% CI ranges from 0.15 to 0.28, and the P value is below 0.001 according to the DeLong method;. [d] the ΔAUC compared to the proposed approach is 0.12, and the 95% CI ranges from 0.07 to 0.19 according to the Bootstrap method;. [e] the ΔAUC compared to the proposed approach is 0.20, and the 95% CI ranges from 0.13 to 0.28 according to the Bootstrap method;. [f] the ΔAUC compared to the proposed approach is 0.39, and the 95% CI ranges from 0.30 to 0.47 according to the Bootstrap method.

The prenatal ultrasound screening was feasible for fetal anal canals on both coronal and transverse planes in 99.41% (95% CI: 99.36-99.46%) of the subjects, which approximates Su's feasibility rate for fetal perinea (99.69%, 95 CI: 99.64-99.73%)[3]and exceed Xu's feasibility rate for the Visualizable intracardiac flow pattern of fetal hearts (81.62%, 95 CI: 73.87-87.54%)[21].

Owing to the subjects' differences, there is little chance that we can directly compare the diagnostic effectiveness of our proposed approach with the other methods via the results from original studies. Thus, we introduced an ablation analysis[22,23] in a randomly established dataset (Testing set), where each anal atresia case was randomly matched with four non-anal atresia subjects according to anal atresia cases' GA at FD, fetal presentation, singleton/ multiple pregnancies, and high-risk factors (concomitant malformation except for colorectal dilatation and enterolithiasis). In this ablation study (Table 2), our new method performed best, followed by Su's and Ochoa's methods, Lee's method, and indirect signs (colorectal dilatation and enterolithiasis). Our approach statistically outperformed all its competitors judging by the DeLong method and Bootstrap method[24]. This is clinically explicable since our proposed method contains more meaningful positive features (say "funnel sign," "pseudo-target sign," and "line sign") than all the other methods. Positive features make ultrasonographers sensitive to anal atresia, so our proposed approach performed better.

The major limitation of our research is that we did not provide a reliability assessment in the diagnostic test part, such as Cohen's Kappa Statistic. Fetal anal atresia is a kind of congenital anorectal malformation whose prenatal diagnosis is of great medical risk to Chinese ultrasonographers. Patients and subsequent ultrasonographers shall be informed once the ultrasound diagnosis of anal atresia is made. Therefore, we were unable to conduct a reliability assessment for the whole process across image collection and image audit.

This large-scale multicenter prospective cohort study finds that ultrasound scan enjoys high validity in detecting fetal anal atresia. Our new ultrasound features significantly promote the detection of fetal anal atresia.

Methods

This prospective cohort research was approved by the ethics committees of the HPMCHCH and YMCHH. The approval number is 2019-S015. Prior informed consent was obtained from all recruited gravidas. This study was designed and carried out according to the STARD guidelines[17] and the Declaration of Helsinki.

Patients

A total of 94,701 gravidas receiving routine prenatal ultrasound screening were prospectively recruited from the HPMCHCH or YMCHH between January 1, 2019, and October 31, 2023, which formed a consecutive series. The inclusion criteria were gravidas receiving routine prenatal ultrasound screening in the hospitals above, gravidas planning to give birth in the two hospitals, and gravidas in their second or third trimesters. The exclusion criterion was a gravida unwilling to participate in this research (Fig. 3). We collected medical information from all participants, such as maternal age at first diagnosis (FD), gestational age (GA) at FD, fetal

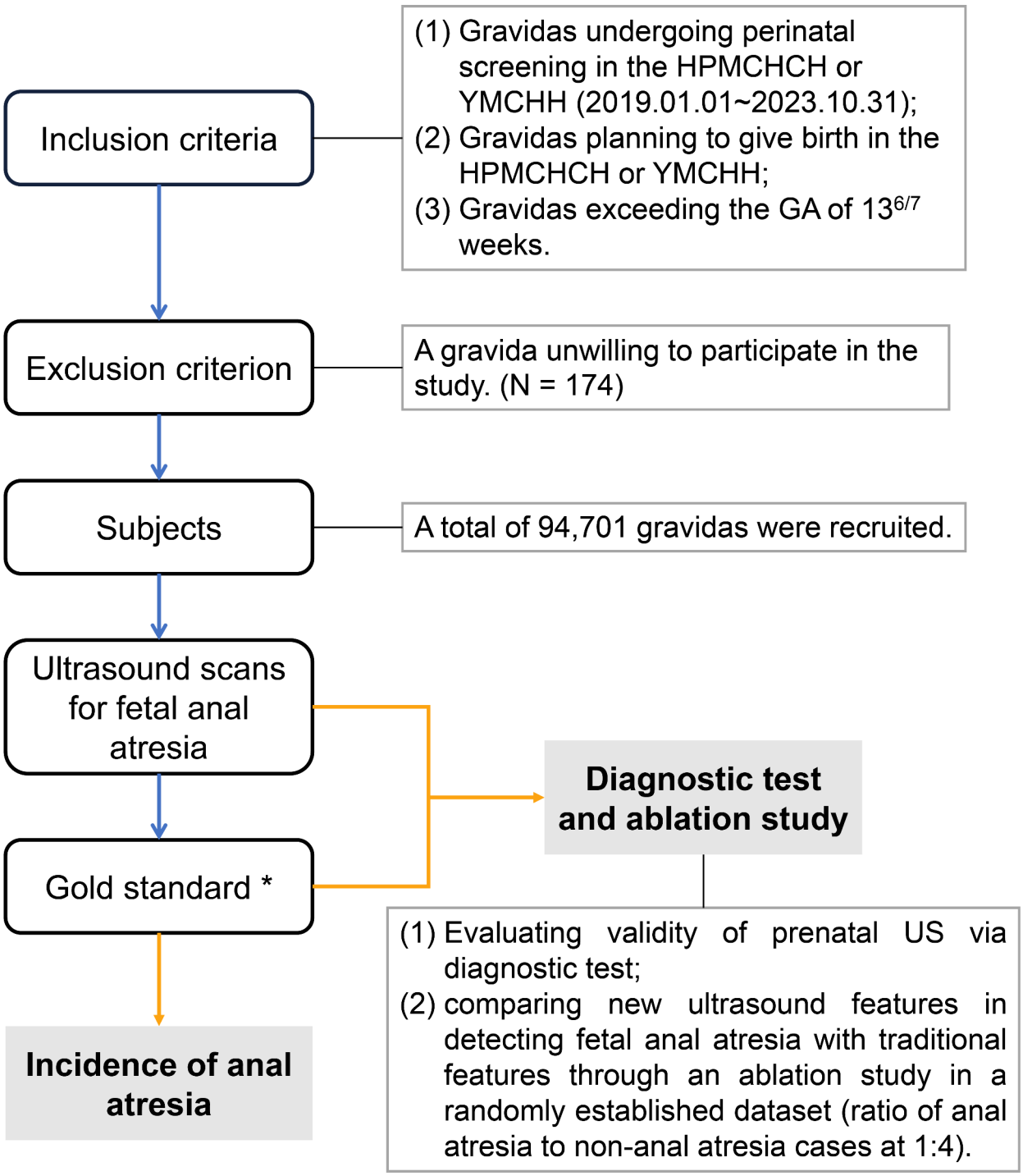

Fig. 3. Flow chart of overall study. HPMCHCH; YMCHH; GA, gestational age; US, ultrasound scan. * The gold standard for diagnosing fetal anal atresia is routine examinations of neonatal anus.

weight at FD, single deepest vertical pocket at FD, service year of diagnosing ultrasonographers at FD, singleton or multiple birth, live birth or stillbirth, concomitant malformation, fetal presentation, and type of operations that perinatal infants need.

Ultrasound screening

A Voluson E10 (General Electric, Bosten, United States) with C1-6-D (4–6 MHz) and RM7C (5–7 MHz) probes and a Voluson E8 (General Electric, Bosten, United States) with C1-5-D (2–5 MHz) and RAB6-D (4–6 MHz) probes received careful fine-tuning before deployment.

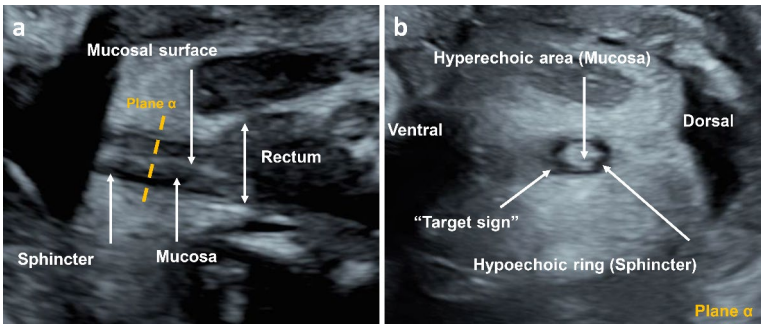

Fig. 4. Illustration for normal fetal anal canal (2D, transabdominal). (**a**) "equal sign" on coronal plane; (**b**) "target sign" on transverse plane α.

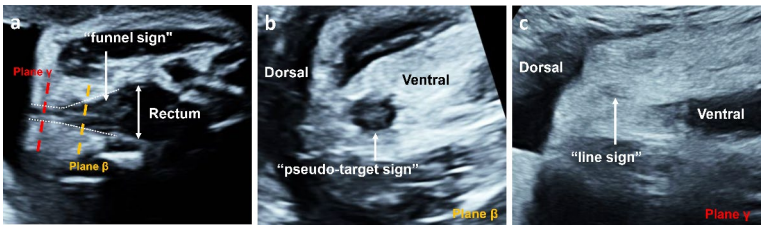

Fig. 5. Partly developed anal canal (2D, transabdominal). (**a**) "funnel sign" on coronal plane; (**b**) and (**c**) "pseudo-target sign" and "line sign" on transverse plane β and γ, respectively.

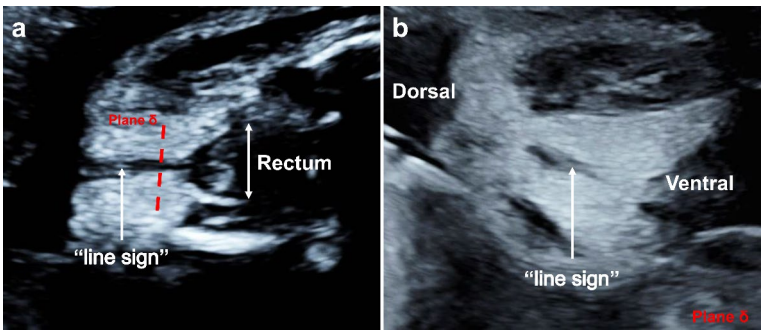

Fig. 6. Completely undeveloped anal canal (2D, transabdominal). (**a**) and (**b**) "line sign" on coronal plane and transverse plane δ, respectively.

Albeit screening for anal atresia is not a compulsory requirement by the Chinese practice guidelines[25], an unfriendly medical environment makes it a routine item in daily prenatal ultrasound screening. Ultrasound scans for fetal anal canals were conducted during routine prenatal ultrasound screening of all enrolled gravidas with their images recorded whether on coronal or transverse planes. If fetal anal canals were not exposed sufficiently, we would advise the gravidas to take a rest to slightly alter fetal position until acquiring clear ultrasound images of fetal anal canals. Additionally, we would record the feasibility of ultrasound scans on both coronal and transverse views of an anal canal in each fetus to calculate the feasibility rate.

The ultrasound diagnostic criteria included several major and minor features. The former contained solely direct signs, such as "funnel sign," "pseudo-target sign," "line sign," "equal sign" (Fig. 4a), and "target sign" (Fig. 4b). A case would be considered as anal atresia if any of these major features existed. The minor features were colorectal dilatation (either colonic or rectal dilatation)[26] and enterolithiasis, which suggested fetal anal atresia only in coexistence with those major features.

As shown in Fig. 5a, the "funnel sign" is typically seen on a coronal plane of an anal atresia case with a partly developed anal canal. The funnel tube and cone denote the atretic and developed parts of the anal canal, respectively. The "pseudo-target sign"[4,26] is often seen on a transverse plane at the funnel cone level in the same case (Fig. 5b). The "line sign" is usually seen on a transverse plane at the atretic anal canal level of an anal atresia case with a partly developed anal canal (Fig. 5c) or on both coronal (Fig. 6a) and transverse planes (Fig. 6b) of an anal atresia case with a completely undeveloped anal canal.

Validity assessment in diagnostic test

A gravida served as a basic unit of this diagnostic test, which meant each multiple pregnancy was regarded as a whole[27]. The predicted results were the prenatal diagnoses of ultrasound screening. The gold standard for diagnosing fetal anal atresia was a routine examination of neonatal anus after birth or termination (SFigure 1)[6].

Validity measures the extent to which the results of prediction models are close to the ground truth[28]. The sensitivity[29], specificity, Youden index, positive likelihood ratio (LR^+), negative likelihood ratio (LR^-), diagnostic odds ratio (DOR), positive predictive value (PPV), negative predictive value (NPV), F1-score, Cramer's V, area under curve (AUC) of the precision-recall curve (PRC), and AUC of the receiver operating characteristic curve (ROCC), accuracy and balanced accuracy (BA)[30] were applied to rate the validity of our new approach.

Youden index[31] is defined by formula 1

$$Youden\ index = Sen + Spe - 1, \tag{1}$$

where Spe and Sen indicate specificity and sensitivity, respectively.

Cramer's V is typically used to evaluate the correlation between the two nominal variables of a two-way contingency table. Cramer's V falling into 0-0.1, 0.1–0.3, 0.30–0.5, or 0.5-1.0 means no association, weak association, moderate association, and strong association between these two nominal variables, respectively[32].

ROCC can illustrate the specificity and sensitivity values of different cutoff points and is not correlated with disease prevalence. AUC of the ROCC under 0.7 suggests poor diagnostic value; AUC of the ROCC between 0.7 ~ 0.8 infers acceptable diagnostic value; AUC of the ROCC between 0.8 ~ 0.9 denotes excellent diagnostic value; AUC of the ROCC over 0.9 indicates the outstanding diagnostic value[33].

PRC and BA are often utilized to contrast imbalanced classification models[30,34], which surpass ROCC and accuracy in imbalanced classifications, respectively.

To sum up, the larger the specificity, sensitivity, Youden index, PPV, NPV, LR^+, DOR, F1-score, Cramer's V, AUC of the PRC, AUC of the ROCC, accuracy, and BA are, the better the model performs. Conversely, the smaller the LR^- is, the better the model does.

Ablation study on different ultrasound features in diagnosing fetal anal atresia

We introduced an ablation study in a randomly established dataset (Testing set) to prove the effectiveness of our new ultrasound features for anal atresia diagnosis. In the Testing set, each anal atresia case (STable 1) was randomly matched with four non-anal atresia subjects (STable 2) according to anal atresia cases' GA at FD, fetal presentation, singleton/ multiple pregnancies, and high-risk factors (concomitant malformation except for colorectal dilatation and enterolithiasis). Three ultrasonographers collaborated in reviewing digital images of all cases in this dataset and diagnosing them through different ultrasound features with proper intervals and blind methods. The baseline was features recommended by Su[3]and Ochoa[16], containing "target sign," "equal sign," colorectal dilatation, and enterolithiasis. Colorectal dilatation and enterolithiasis with or without "target sign" served as two comparative items[15]. Our proposed approach (major features: "target sign," "equal sign," "funnel sign," "pseudo-target sign," and "line sign"; minor features: colorectal dilatation and enterolithiasis) served as another comparative item.

Statistical analysis

RStudio 2024.04.2 + 764 (posit.co) was employed for statistical analysis. All statistical analyses are two-tailed at a significance level of 0.05. Normal continuous data were described by $\bar{X} \pm SD$[35], while non-normal continuous data detected by Q-Q plot and Shapiro-Wilk test were detailed by medians, quartiles, ranges, excess kurtosis, and skewness[36]. Qualitative data were described by absolute and relative numbers[19].

In the validity assessment and ablation analysis, we provided both point and interval estimations of sensitivity, specificity, Youden index, LR^+, LR^-, DOR, PPV, NPV, F1-score, Cramer's V, AUC of the PRC, and AUC of the ROCC to the best of our ability, as well as *P*values compared to null hypotheses. The contrasts in the AUC of the ROCC and the AUC of the PRC among different anal atresia diagnostic methods were based on the DeLong method[24] and Bootstrap method, respectively.

Data availability

The data analyzed in this study will be available from the corresponding author on reasonable request after follow-up studies will have been conducted on these data.

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Conceptualization: H.K. and Y.P.; Methodology: H.K., S.W., and Y.P.; Formal analyses and investigation: Haiyan Kuang, Hui Cao, L.Y., and Y.P.; Writing - original draft preparation: H.K.; Writing - review and editing: H.K., H.C., S.W., Y.L., Y.G., L.Y., J.Y., and Y.P.; Funding acquisition: H.K., Y.L., J.Y., and Y.P.; Resources: H.K., Y.L., S.W., Y.G., and Y.P.; Supervision: H.K., Y.L., L.Y., J.Y., and Y.P. All authors reviewed the manuscript.

Declarations

Competing interests

The authors declare no competing interests.

Ethical approval

Institutional Review Board approval was obtained.

Consent to participate

Written informed consent was obtained from all subjects (patients) in this study.

Additional information

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