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ORIGINAL ARTICLE

Visualization of the fetal anus by prenatal ultrasound for the diagnosis of anorectal malformations: is it feasible?

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Abstract

Purpose The goal of this study was to determine the feasibility of identifying the anal dimple (AD) on routine prenatal ultrasound. Using the presence, absence, appearance, and location of the anal dimple as an indirect sign for possible underlying anorectal malformations (ARM), we hypothesize that evaluation of the anal dimple as part of the fetal anatomic survey may increase the sensitivity in detecting less severe ARMs.

Methods In a prospective longitudinal observational study, pregnant women who underwent prenatal ultrasound (US) at the Colorado Fetal Care Center between January 2019 and 2020 were enrolled. The variables recorded included gestational age, singleton versus multiple pregnancy, gender of the fetus, visualization of the AD, and reason for non-visualization of the AD.

Results A total of 900 ultrasounds were performed, evaluating 1044 fetuses, in 372 different pregnant women. Gestational ages ranged from 16 to 38 weeks. The AD was visualized in 612 fetuses (58.6%) and not seen in 432 (41.4%). The two most common reasons for non-visualization were extremes in gestational age ($n = 155$; 36%) and fetal position ($n = 152$; 35.3%). The optimal gestational age range for AD visualization was 28–33 weeks + 6 days, with 78.1% visualization rate.

Conclusion Visualization of the anal dimple by ultrasound is feasible and may aid in the detection of less severe ARMs, ultimately impacting pregnancy management and family counseling. The optimal timing for anal dimple visualization is late second and third trimester.

Keywords Anorectal malformation · Prenatal ultrasound · Prenatal diagnosis · Fetal anus · Anal dimple

Introduction

Prenatal diagnosis of anorectal malformations (ARMs) remains challenging, with mainly the severe forms, such as cloaca and cloacal exstrophy, commonly identified in-utero due to intra-abdominal findings such as hydrocolpos, omphalocele, bowel dilation/calcification, hydronephrosis or absent kidney [1, 2]. Identification of less severe types of ARMs may be possible through the detection of an anomalous anal opening or absence of one (Fig. 1). These include recto-urethral bulbar or prostatic fistula, recto-bladder-neck fistula, and anorectal malformation without fistula in males, and recto-vestibular fistula and anorectal malformation without fistula in females.

The typical appearance of the anal dimple by prenatal ultrasound has been described as a predominantly rounded area of echogenicity surrounded by a rim of hypoechogenicity posterior to the fetal genitalia, resembling a "target sign" [3–5]. Using the presence or absence, appearance, and

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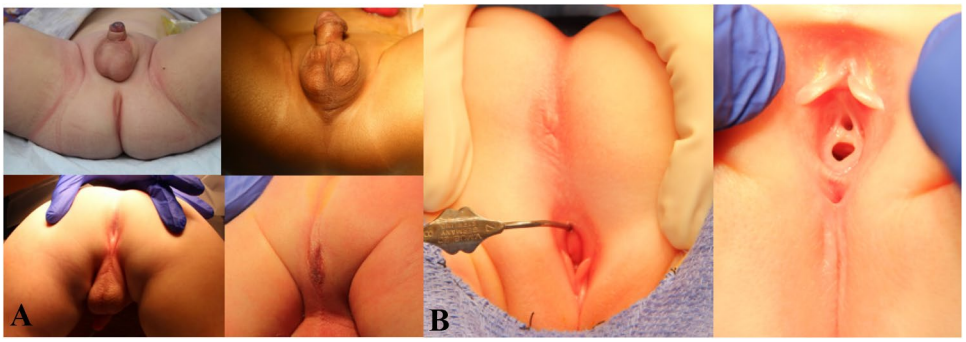

Fig. 1 Perineum appearance in patients with anorectal malformation. **a** Male patients and **b** female patients with recto-vestibular and recto-fourchette fistula

location of the anal dimple as an indirect sign for possible underlying ARM, we hypothesize that evaluation of the AD as part of the fetal anatomic survey may increase the sensitivity in detecting less severe ARMs.

The goal of this study was to determine the feasibility of identifying the anal dimple on routine prenatal ultrasound.

Methods

A prospective longitudinal observational study was designed. Prior to commencing our work, institutional review board approval was obtained (IRB#19-2290).

Pregnant women undergoing prenatal ultrasound at the Colorado Fetal Care Center between January 2019 and October 2020 were enrolled in the study. Fetuses with suspected ARM were excluded. Prospectively collected data included gestational age, singleton versus multiple pregnancy, gender of the fetus, visualization of the anal dimple, and reason for non-visualization of the anal dimple. The gestational age was stratified in four groups: group 1 from 16 weeks until 21 weeks + 6 days, group 2 from 22 weeks until 27 weeks + 6 days, group 3 from 28 weeks until 33 weeks + 6 days, and group 4 from 34 weeks and beyond. All ultrasound images were acquired by three, obstetric sonographers certified by the American Registry for Diagnostic Medical Sonography, with 4–20 years of maternal fetal medicine experience. Images were obtained using three similar GE Voluson E10 machines (GE Healthcare, Milwaukee, WI) using C2-9-D, C1-6D and RM6C (3D) probes. Transverse plane images of the fetal perineal region showing the AD were saved in the patient's study and annotated as "AD" (Fig. 2). If the AD was not visualized, an image of the fetal perineal area was saved with the annotation "ADSO" (AD suboptimal). Data on the reason for lack of visualization were kept on a separate work sheet. Each ultrasound (including repeated exams at different gestational ages) was recorded as an individual opportunity to visualize the anal dimple but each fetus was later evaluated to determine if the anal dimple was visualized in any ultrasound encounter. Data were analyzed with crosstabulation to obtain a Chi square test and statistical analysis was performed using SPSS version 22.0.

Results

A total of 900 ultrasounds were performed, evaluating 1044 fetuses, in 372 different pregnant women. One patient was excluded due to the suspicion for underlying cloacal anomaly that was confirmed postnatally. Gestational ages ranged from 16 to 38 weeks. Each pregnant woman had between 1 and 11 ultrasounds, with a median of 2 ultrasounds. Of the 900 ultrasound studies, 764 were singleton pregnancies,

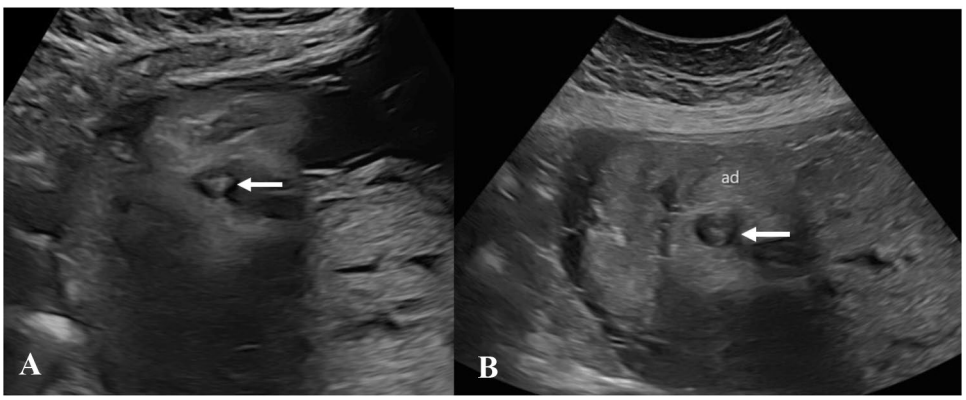

Fig. 2 Transverse plane ultrasound images of the normal anal dimple showing the central area of hyperechogenicity and the surrounding rim of hypoechogenicity. **a** 29 weeks 3 days pregnancy; **b** 30 weeks 3 days pregnancy

127 were twins, 8 were triplets, and 1 was a quadruplet pregnancy. The anal dimple was visualized in 612 fetuses (58.6%) and not seen in 432 (41.4%) (Fig. 3). The two most common reasons for non-visualization were extremes in gestational age in 156 (35.5%), and fetal position in 154 (35.1%) (Fig. 4).

Regarding gestational age, the anal dimple was visualized most often in group 3 (28–33 weeks gestational age + 6 days), successful in 78.1%. This difference was statistically significant $p < 0.001$ (Figs. 5 and 6) when compared to other groups and even when group 1 was eliminated.

Not surprisingly, the anal dimple was more frequently identified in singleton pregnancies than in multiple pregnancies ($p < 0.001$) (Fig. 7), presumably related to the difficulty in obtaining an adequate plane of imaging in multiple gestation pregnancies due to fetal positioning. The anal dimple was visualized in 506 (66.4%) of single pregnancies and in 106 (37.6%) of multiple pregnancies.

Gender did not play a role in the ability to visualize the anal dimple (Fig. 8).

Two hundred and three fetuses had more than 1 ultrasound study and 412 fetuses had only one ultrasound study. In the fetuses with more than one ultrasound, the AD was visualized at least once in 164 (81%) cases, and not visualized in any ultrasound in 39 (19%).

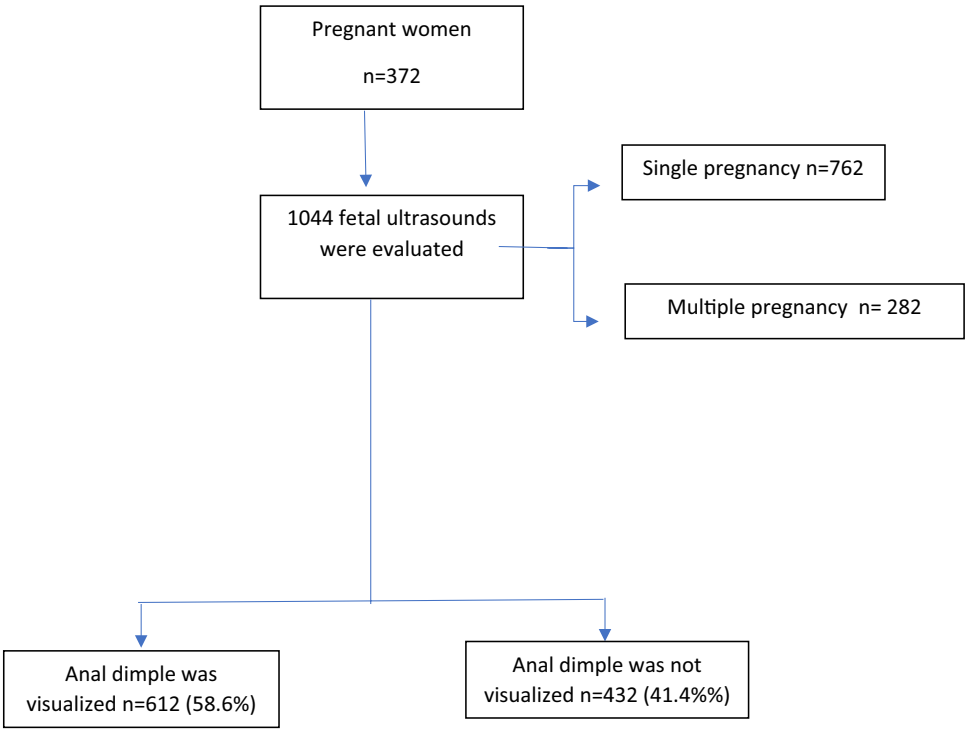

Fig. 3 Patient flowchart

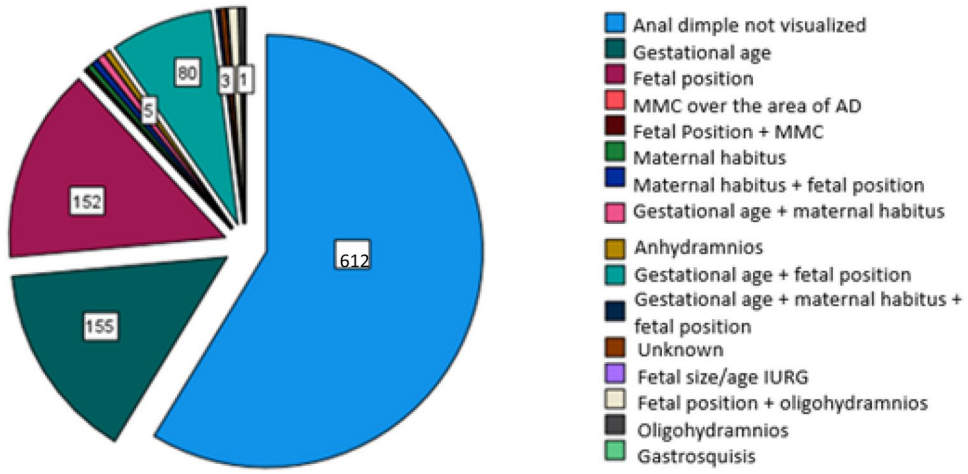

Fig. 4 Reason for non-visualization of the anal dimple

Crosstable of Anal Dimple Visualization on static*Stratification (age)

			Stratification (age)				
			16-21W+6d	22-27w+6d	28-33w+6d	34 w or more	Total
Anal Dimple Imaged on static	no		174	86	79	93	432
		% of total	16.7%	8.2%	7.6%	8.9%	41.4%
	yes		4	132	281	195	612
		% of total	0.4%	12.6%	26.9%	18.7%	58.6%
Total			178	218	360	288	1044
		% of total	17.0%	20.9%	34.5%	27.6%	100.0%

Fig. 5 Table showing stratification according to gestational age and anal dimple visualization

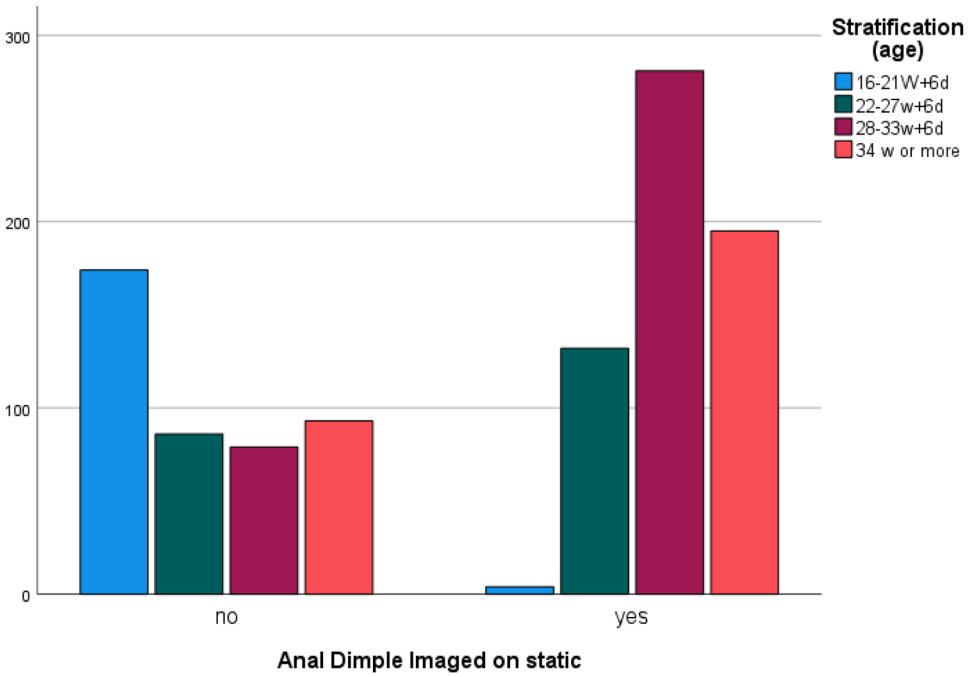

Fig. 6 Anal dimple visualization according to gestational age group

Two hundred and five babies were delivered at Children's Hospital Colorado. Only one was found to have the most benign type of anorectal malformation, a recto-perineal fistula, all the others had a normal anus.

Discussion

In-utero recognition of anorectal malformations allows for proper parental counseling, and prenatal and perinatal management. Patients with ARM should be born in a tertiary center with a level III neonatal intensive care unit and pediatric surgical availability, since most will require surgery shortly after the first 24 h of life. Unfortunately, ARMs remain a challenging diagnosis to make prenatally, especially less severe forms where bowel dilatation may not be present prenatally. The reported incidence of imperforate anus is of approximately 1–4 cases per 5000 live births [4]. However, its prenatal detection has been described as low as 16–42% [6, 7] when relying on bowel dilatation or calcified intraluminal meconium. The lack of a formalized screening of the anal dimple on routine anatomic fetal ultrasound studies may account for this suboptimal detection rate, given the typical appearance of the newborn perineum in ARM (Fig. 1). Evaluation for fetal gender and the anal dimple are considered optional by national guidelines [8–10]; though the former is more routinely performed to accommodate parental desire, as well as to assess for potential genetic or syndromic conditions specifically associated with male or female gender. Therefore, the anal dimple is not routinely examined on prenatal imaging in the United States.

In this single-center study, we demonstrate that AD visualization is feasible. Our identification rate was 58% when assessed by ultrasound encounter, though 81% when

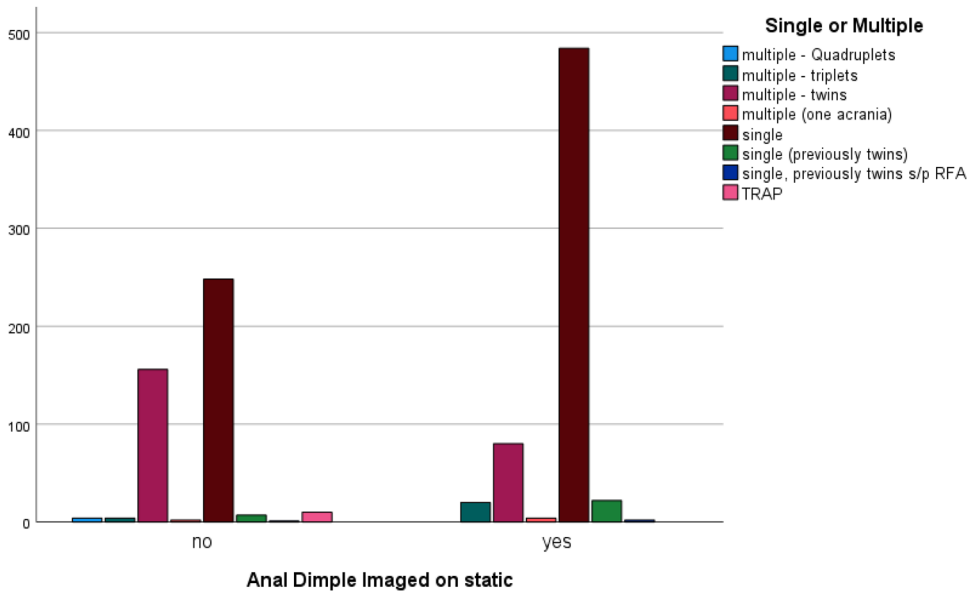

Fig. 7 Visualization of the anal dimple according to the number of fetus (singleton vs. multiples)

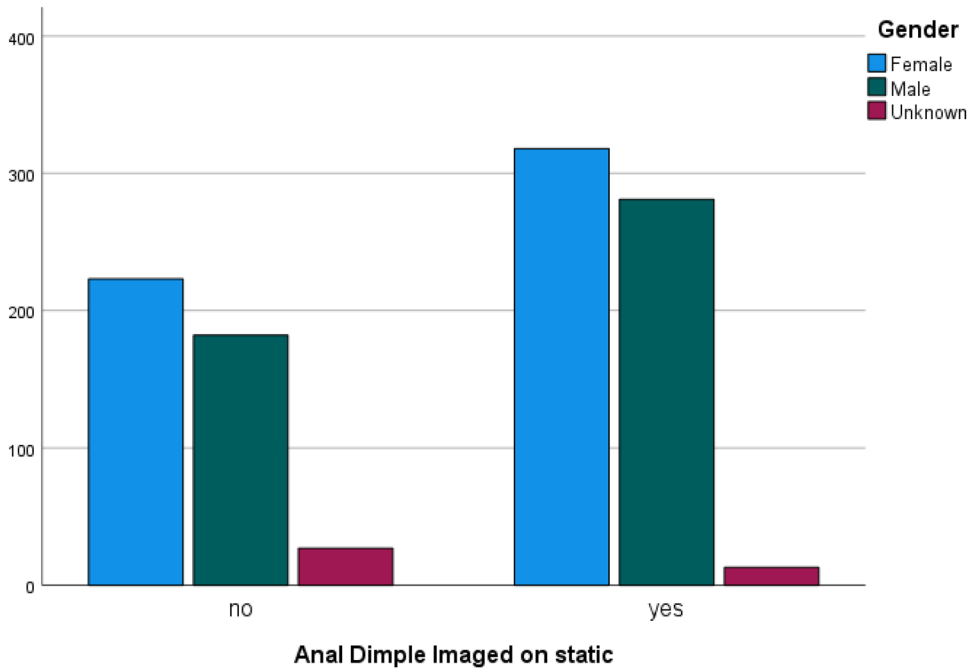

Fig. 8 Visualization of the anal dimple according to the gender of the fetus

assessed by individual patient (fetus). Similar studies, performed in other countries [3–5, 11], have also demonstrated feasibility and consequent increased detection rate of ARMs during fetal life. A prior study from South Korea demonstrated consistent high rates of visualization of the AD between 23 and 34 weeks of gestation and a predictable pattern of anal sphincter growth with gestational age [5]. On our study, the optimal gestational age for the visualization of the AD was between 28 and 33 weeks. In a histological evaluation of fetal anal sphincter development, Arakawa et al. demonstrated a rapid increase in size of the anal sphincter on late gestation fetuses and a slower growth on mid-gestation fetuses [12]. These data help to explain the increased visualization of the AD in the late second and third trimesters on recent studies, including ours. This also supports follow-up scan in cases where the AD was not initially identified [3].

ARMs are associated with other congenital anomalies in up to 70% of cases [13], especially urogenital anomalies. There is a high incidence of ARM in cases of VACTREL syndrome, caudal regression syndrome, and Trisomy 21 [3, 14]. Therefore, a high index of suspicion should be present when additional fetal abnormalities are seen in association

with bowel dilatation. The visualization of a normal AD can aid in the exclusion of ARM in cases of bowel dilatation due to other etiologies [4, 14].

Of the 205 babies that we had access postnatally to confirm a normal anus, only 1 was found to have anorectal malformation. She was born with the most benign type of anorectal malformation, a recto-perineal fistula. This is probably the most difficult to identify prenatally, since it is usually an isolated anomaly, and only the most distal part of the rectum (fistula) is abnormally located, anterior to the center of the sphincter.

To our knowledge, this is the first prospective study accessing the feasibility of prenatal AD visualization performed in the United States. Larger, multi-institutional studies evaluating the incidence of visualization of the AD should follow these preliminary results.

Limitations of our study include the lack of interobserver variability data between different ultrasound technologists performing the ultrasounds, as well as the single institution nature of the study.

Conclusion

Visualization of the anal dimple by prenatal ultrasound is feasible and may aid in the detection of less severe ARMs, ultimately impacting pregnancy management and family counseling. The optimal timing for anal dimple visualization is late second and third trimester.

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