Review Article

Ultrasound Evaluation of Perianal Diseases in Infants

Ya Ma*, Zhengrong Wang
Department of Ultrasound, Capital Institute of Pediatrics, Beijing, China

Abstract

Organs and tissues comprising the perianal area include the caudal segments of the digestive system and reproductive system, the perianal soft tissues, and the terminal part of the spinal cord. Diseases originating in one part of the perianal region may have connections with others. Incomprehensive and disorganized ultrasound (US) scanning may lead to insufficient diagnostic information, potentially impacting the effectiveness of subsequent treatment. The purposes of this paper are (a) to describe our technique of performing US of perianal area; and (b) to explore the diagnostic strategy of US in infant perianal diseases by outlining the sonographic characteristics of these conditions.

Keywords: Anorectal malformations, hirschsprung disease, neonate, teratoma, ultrasonography

Introduction

Perianal lesions in infants include various anomalies, ranging from congenital malformations, tumors, and inflammatory disorders. Early diagnosis depends on careful clinical examination and comprehensive imaging assessment. At present, there are limited imaging modalities applicable to this condition. Ultrasound (US) is an ideal imaging modality for infants because of its convenience, noninvasiveness, and lack of radiation or sedation.[1] The small size of newborns, thin subcutaneous fat layer and incomplete ossification of vertebrae provide an appropriate acoustic window for the application of high-frequency US in perianal lesions in infants.[1,2] Since various perianal lesions may be associated with other urogenital and gastrointestinal tract abnormalities, as well as the potential to compress or invade surrounding structures, systematic and comprehensive US scanning is required, usually including the anus, rectum, urethra, vagina (female), sacrococcygeal regions, and spinal cord. Tansperineal US can serve as a valuable complementary technique to transabdominal US given its advantages in detecting superficial lesions.[3]

Technique and Protocol

Strategies to reduce infant discomfort and improve imaging quality

Infantile irritability can pose a significant challenge during US examinations, potentially affecting both the comfort of the infant and the quality of the images. Several strategies can be employed to minimize discomfort and improve the success of the procedure. (a) Offering a feeding bottle or sucking comfort: Infants often find comfort in sucking during stressful situations. Offering a feeding bottle or pacifier can help to soothe the infant, reduce distress, and encourage a more cooperative state. (b) Parental presence: Having a familiar face nearby can significantly reduce anxiety, making the infant more comfortable. Allowing a parent to hold or gently soothe the infant during the US can help ensure that the infant remains still for optimal image acquisition. (c) Environmental control: Creating a quiet, warm, and dimly lit environment can help keep the infant calm and reduce distractions during the procedure. (d) Sedation considerations: While sedation may be considered in cases where the infant is particularly uncooperative or agitated, it is not routinely recommended. The use of sedation can interfere with dynamic observations that may be crucial for diagnosing conditions such as occult spinal dysraphism, where the dorsal protrusion of the spinal cord or cauda equina into subcutaneous tissues can be an important finding.

Assessment of anus, rectum, urethra, and vagina (female) through transperineal approach

A linear high-frequency transducer, typically ranging from 7 to 15 MHz, is preferred for obtaining high-resolution detailed

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Address for correspondence: Dr. Ya Ma, Department of Ultrasound, Capital Institute of Pediatrics, No. 2 Yabao Rd, Chaoyang District, Beijing 100020, China.
E-mail: my9374@163.com

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Abbreviations

CE	Contrast enema
HD	Hirschsprung disease
US	Ultrasound

images of perineal area located several centimeters from the rectal lumen. In the midsagittal plane, the transducer is placed longitudinally on the perineum, perpendicular to the anal canal with the patients in the lithotomy position to provide detailed information of the anal canal, rectum, puborectal muscle, urethra, and vagina (female) [Figure 1]. To improve the identification of associated fistulas in cases of anorectal malformations, the transducer is shifted laterally to capture the lower part of the rectum, which is usually extremely narrowed.

Assessment of the sacrococcygeal region

Combining prone position scan with transperineal approach proposed above allows for clear visualization of the sacrum and coccyx. Moreover, careful examination of the area surrounding the tip of the coccyx is conducted to confirm the presence of any cystic or solid masses. When a mass is identified, it is crucial to determine its relationship with the coccyx. The presacral space should also be scanned to exclude presacral mass, which may be associated with Currarino syndrome.

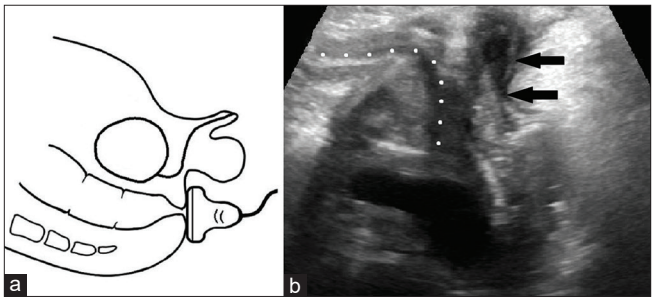

Figure 1: Structures visualized through the perineal approach. (a) This schematic demonstrates the placement of the linear transducer with the transperineal approach. (b) A transperineal ultrasound image in the sagittal plane displays the normal rectum (arrows) and urethra (dashed line)

Assessment of the spinal cord

US examination is performed with the infant lying in a prone position. The spine is scanned in both sagittal and transverse planes from the lumbosacral region to the tip of the coccyx. The following items should be evaluated by US: the location of the conus medullaris, structural changes and pulsatility of the filum terminale and cauda equina, as well as the presence of intraspinal mass or posterior spinal defect.

Transabdominal scan

The course of the bowel loops is followed with a focus on assessing the presence of luminal narrowing or dilatation, as well as thickness and vascularization of the intestinal wall.

Major Applications

Sacrococcygeal teratoma

Sacrococcygeal teratoma originates from Hensen's node at the base of the coccyx. It is the most common germ cell tumor in neonates, with an incidence of 1 in 27,000 live births.[4] Sacrococcygeal teratoma shows a female predilection, with a male-to-female ratio ranging from 1:4 to 1:3. The vast majority of sacrococcygeal teratoma is benign, and the probability of malignancy increases with age. The growth patterns of sacrococcygeal teratoma can be divided into four types according to their morphologic characteristics [Figure 2]. Generally, these tumors start from the base of the coccyx and protrude posteriorly, with some extending into the pelvis from

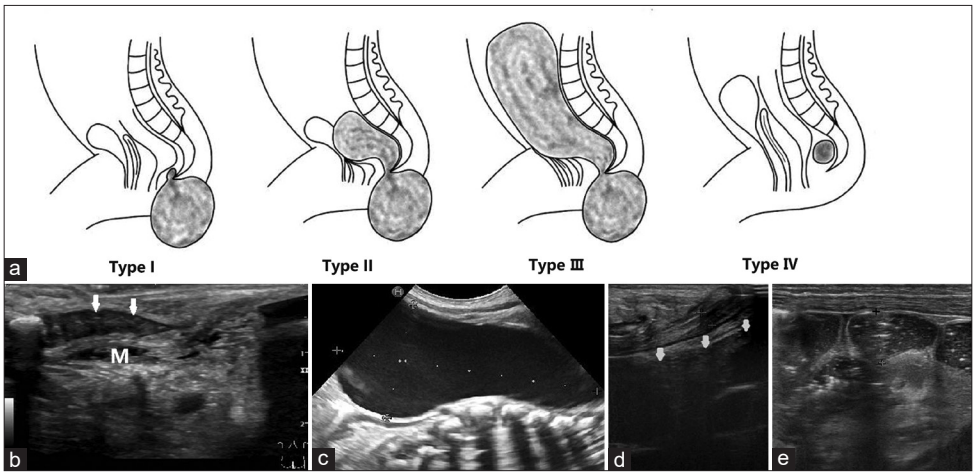

Figure 2: Sacrococcygeal teratoma in neonates. (a) Morphologic classification of sacrococcygeal teratoma. Type I: Primarily external, with minimal presacral growth; Type II: Equal internal and external components; Type III: Mainly located in the pelvic and abdominal cavities, with small external components; Type IV: Entirely internal anterior sacral without external presentation. (b) Sacrococcygeal teratoma seen by ultrasound (US) in a 1-day-old female neonate. Longitudinal US scan of the coccyx reveals the mass (M) located anterior to the coccyx (arrows) with the infant in a prone position. (c) Sacrococcygeal teratoma in a 0-day-old male neonate. Midsagittal scan of the lower abdomen shows the tumor (between electronic calipers) extending upward into the pelvis and abdomen. (d) Longitudinal sonogram of the same patient shows the tumor (arrows) displaces the rectum (between electronic calipers) anteriorly. (e) Transverse sonogram of the left abdomen shows diffuse bowel gaseous dilatation due to extrinsic rectal compression

the anterior surface of the coccyx, compressing and distorting the anus, rectum, vagina (female), and bladder [Figure 2].

Imaging studies are required to confirm the morphologic classification of the tumor as well as its relationship with surrounding tissues (such as the coccyx, rectum, urethra, vagina, and bladder), which help to guide therapeutic decisions. Similar to teratomas arising elsewhere in the body, sacrococcygeal teratomas are pathologically categorized into three types: mature teratoma, immature teratoma, and malignant teratoma. On US, most masses appear cystic or heterogeneous with mixed cystic and solid components, well-defined, and faint vascular signals can be observed. Although US has limited ability to distinguish between benign and malignant teratomas, some signs may suggest specific histologic subtypes. Benign sacrococcygeal teratomas are prone to exhibit more cystic components, extensive calcification or ossification, and homogeneous fat components, whereas heterogeneous solid components, abundant blood supply, noticeable necrosis, or hemorrhage indicate malignancy.[5] Regular US examinations are still needed to monitor recurrence after tumor resection. It is worth noting that some benign teratoma may recur as a malignant type after resection.

Occult spinal dysraphism

Besides sacrococcygeal teratoma, the possibility of occult spinal dysraphism should be considered in newborns presenting with a sacrococcygeal mass, especially when the lesion size changes with pressure. Unlike sacrococcygeal teratoma, which is mostly located anterior to the coccyx, sacrococcygeal region masses caused by occult spinal dysraphism typically occur behind the coccyx or sacrum. Dynamic observation may show that the spinal cord or cauda equina protrudes dorsally to the subcutaneous tissues [Figure 3]. Some cases may be accompanied by intraspinal or subcutaneous lipoma or hemangioma. The location of the conus medullaris should be assessed to determine whether there is an accompanying tethered cord. US can serve as the initial imaging modality of choice in infants <6 months duo to the posterior cartilaginous portions of the vertebrae (ossification begins from the posterior side of the spinal canal 3–4 months after birth).[6-8] Anatomic characteristics of the neonatal spinal cord and spine on US include: (a) Movement of the filum terminale and cauda equina: reduced or absent movement due to respiration and pulsation indicates a tethered cord. (b) Level of the conus medullaris tip: typically located in the center of the spinal canal between L1 and L2. Deviation below the superior margin of L3 or displaced dorsally within the spinal canal raises suspicion of a tethered cord. (c) The shape of the conus medullaris: normally pointed, whereas a blunted termination of the conus medullaris prompts a search for an intraspinal lipoma or a tethered spinal cord. (d) Thickness of the filum terminale: typically ≤2 mm, with thickening suggesting tethering or lipoma. (e) Bone fusion of the vertebral column is observed in the sagittal section of the spine. (f) Detection of intraspinal tumor such as cyst or lipoma. (g) Syringomyelia: manifested as a dilated central canal in the spinal cord. (h) Lumbosacral skin dimples or dermal sinus tracts (distinct from pseudosinus tracts commonly found in the sacrococcygeal region and terminating within the cutaneous layer) should be explored to assess potential communication between the sinus tract and subarachnoid space.

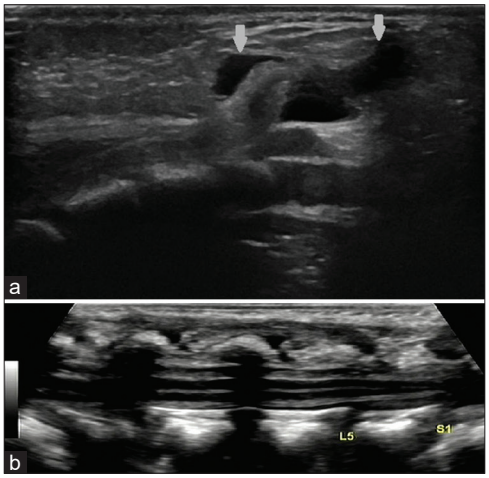

Figure 3: Longitudinal ultrasound image of the sacrococcygeal region demonstrates occult spinal dysraphism in a 3-day-old male neonate. (a) The meninges, filum terminale and cauda equina protrude dorsally and caudally (arrows). (b) The tip of the conus is elongated and located at S1, indicating a tethered cord

Anorectal malformations

Anorectal malformations are the most common congenital abnormalities of the digestive system in neonates, with an incidence of 1/5000 live births.[9] According to the position of atresia (with the puborectalis muscle as a reference), they can be divided into three types: high, intermediate, and low type. The classification helps establish the surgical plan. There are three indicators for US classification of anorectal malformations: [10-12] (a) The distance from the end of rectum to the perineum, (b) the location of the fistula, and (c) the relationship between the terminal rectum and the puborectalis muscle. Previous reports have mainly focused on the distance from the end of the rectum to the perineum. Generally, a distance of <1 cm indicates the low type, whereas a distance >1.5 cm indicates a high or intermediate type. However, factors such as the location and oblique angle of the transducer, as well as the timing of examination, can affect the measurement results. Considering the slow transmission of fetal stool in some neonates, it is suggested that the distance measurement should be obtained 24 h after birth.[13] Minimal pressure should be applied during scanning to avoid underestimating the distance between the perineum and the distal rectal pouch. The cutoff for rectum-perineum distance in distinguishing between low, intermediate, and high types of anorectal malformations is still undefined and often overlaps. Therefore, it is important to identify the location of fistulas. Some beak-shaped distal rectal pouches and the fistulas are too thin to be detected, and repeated dynamic sweep and careful observation are needed to obtain valuable image information [Figure 4]. Contrast-enhanced

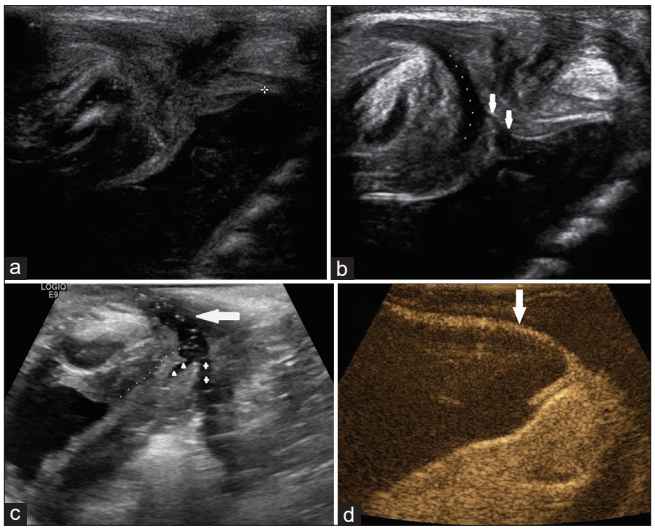

Figure 4: Sonographic manifestations of anorectal malformation. (a) Ultrasound (US) appearance of anorectal malformation seen by the transperineal approach in a 1-day-old male neonate. The rectum-perineum distance is 1.2 cm (asterisk shows the end of the rectum). (b) Transperineal US depicts the beak shape of the distal rectum in the same neonate. A recto-bulbar urethral fistula (arrows) communicated with the bulbar urethra (dashed line) is clearly depicted. (c) Cloaca seen by transperineal US in a 0-day-old female neonate. Midsagittal transperineal US shows a common channel (arrow) connected to the urethra (dashed line), vagina (triangles), and rectum (asterisks). (d) Contrast-enhanced colosonography identifies the bulbar rectourethral communication and the urethra (arrow)

US is more sensitive in imaging fistulas associated with anorectal malformations [Figure 4]. Patients with anorectal malformations often have accompanying anomalies involving cardiovascular, gastrointestinal, genitourinary, and central nervous systems. Radiologists should be aware of these anomalies to establish a complete US evaluation.

For a newborn female, a single perineal orifice indicates a special type of anorectal malformation, specifically cloaca, accounting for 5% of all anorectal malformations. Although cloaca can be diagnosed through physical examination, US can provide more detailed and comprehensive information regarding the surgical plan and prognosis[14,15] [Figure 4]. For example, the length of the common channel can be assessed using a combination of transabdominal and transperineal US to guide future surgical strategy. Complex cloaca with a common channel longer than 3 cm should be repaired by surgeons fully dedicated to reconstruct the urethra, vagina, and rectum.[16] Accompanying anomalies, such as hydrocolpos, hydronephrosis, genitourinary malformations, tethered cord, and duodenal atresia, should also be evaluated by US examination. In infants with severe hydrocolpos or hydrometrocolpos, the bladder, urethra, and rectum can be distorted and displaced, resulting in failure US delineation of the common channel. Repeated US examinations should be performed after sufficient drainage of hydrocolpos.

Anal stenosis is a relatively rare anorectal malformation. It may manifest as dilatation of the colon or rectum, which can resemble Hirschsprung disease (HD). However, anal stenosis differs from HD in that it involves a relatively limited narrow segment [Figure 5], and digital rectal examination can reach the stenosis ring. On US, a transperineal scan can reveal the focal narrowing of the anal canal. A thorough examination is necessary to rule out the presence of an associated presacral mass.

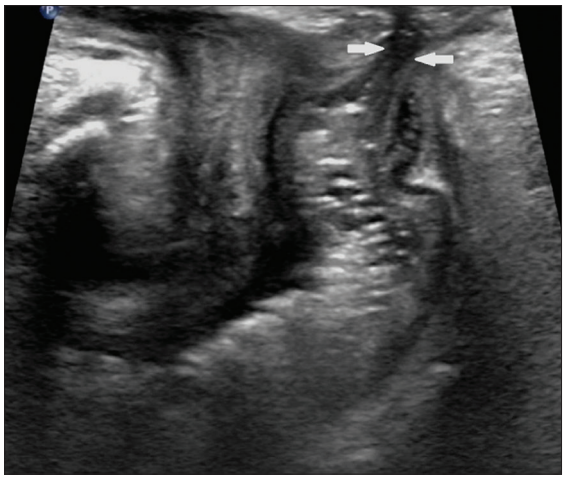

Figure 5: Sonographic manifestations of anal stenosis in a 6-month-old male infant. Transperineal scan shows localized narrowing of the lower anal canal (arrows)

Hirschsprung disease

HD is a common congenital disorder with a global incidence of approximately 1 in 5000 in newborns.[17-20] HD is characterized by the absence of ganglion cells in the submucosa and myenteric nerve plexus in the distal bowel, resulting in delayed passage of meconium and refractory constipation. Affected patients typically present early in infancy or childhood, and surgery is recommended once the diagnosis has been established.

Contrast enema (CE) is commonly utilized for HD assessment, with sensitivity ranging from 76% to 88.9% and specificity ranging from 84.2% to 98%.[21] Some pathological changes of HD can be identified by US with retrograde infusion of saline into the colon (hydrocolonic sonography).[22] US characteristics of HD include the relatively narrowed aganglionic segment, the dilated proximal bowel, transition zone, thickening, and hypervascularity of the upstream bowel. Similar to anorectal malformations, transperineal US also plays a crucial in the diagnosis of HD. When used in conjunction with transabdominal US, transperineal US provides additional information of the distal rectum, which is commonly involved in HD and can be challenging to detect with transabdominal US due to its deep location. Furthermore, transperineal scanning has advantages over CE in detecting extraintestinal diseases, such as presacral masses in Currarino syndrome, which is another differential diagnosis of constipation. HD needs to be distinguished from temporary constipation in some preterm infants due to immature intestinal development, which rarely has a typical narrowed segment, and repeated US scan is

advocated if necessary. Negative imaging finding does not completely exclude the possibility of HD, and rectal biopsy is still required in some indeterminate cases to make a definite diagnosis.

Currarino syndrome

Currarino syndrome refers to a rare congenital condition characterized by sacral agenesis, anorectal malformation, and presacral mass (which may be an anterior sacral meningocele or presacral teratoma). Although it can occur in any age group, it is more commonly seen in young children.[23] It can present with various clinical manifestations. It usually manifests as abdominal distension, severe and recurrent constipation and may be associated with anorectal malformations or meningitis in the neonatal period.[24,25]

Patients with suspected Currarino syndrome commonly undergo multiple radiological examinations, including radiography, US, magnetic resonance imaging (MRI), CE. In infants with anorectal malformations, the retrorectal and presacral space should be scrutinized by US. If a presacral mass is detected, Currarino syndrome should be highly suspected, but it should not be ruled out if no presacral mass is visualized due to the delayed growth of the masses in some cases. On sonography, meningoceles usually appear as anechoic lesions without vascular signals on color-Doppler evaluation [Figure 6]. Presacral teratomas typically present as heterogeneous masses, suggesting variable amounts of cyst (anechoic), fat (hypoechoic or hyperechoic), and calcification (hyperechoic). Most teratomas exhibit focal or dispersed intralesional vascular signals, whereas malignant tumors may display increased vascular signals. The mass can compress the rectum, resulting in dilation of the upstream colon. In addition, the sacrococcygeal region and spinal canal should be further examined to assess for the presence of a tethered cord [Figure 6].

Rectal duplication

Rectal duplication accounts for 5% of all enteric duplications.[26] It usually presents with various clinical manifestations, which are related to lesion size, the presence of fistula, infection, intussusception, or volvulus.[27] Rectal duplications are usually asymptomatic in the neonatal period. Some may manifest as constipation, perianal swelling, perianal fistula or hemorrhage, potentially leading to missed or misdiagnosed cases. Meticulous perianal US helps recognize the lesion and avoid unnecessary radiation and invasive examinations. Typically, it presents as an anechoic mass with a slightly thickened wall beside the rectum. Rectal duplication needs to be differentiated from anterior sacral meningocele, cystic teratoma, hydrocolpos, hydrometra,

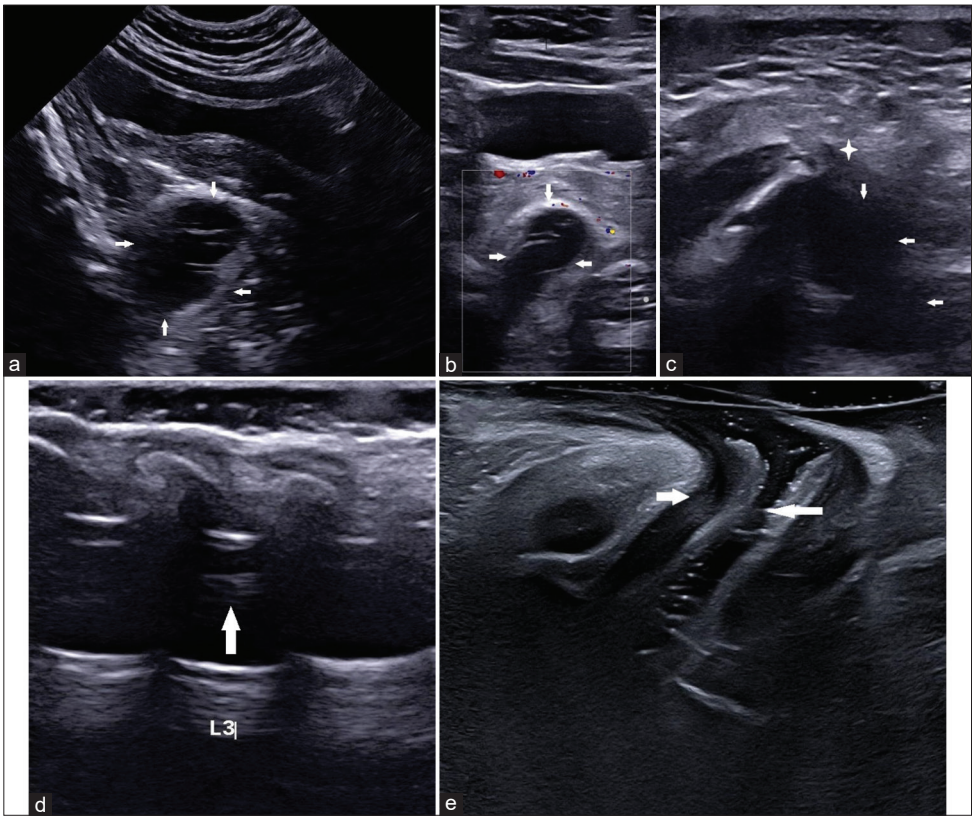

Figure 6: Sonographic findings of Currarino syndrome in a 5-month-old female infant. (a) Longitudinal pelvic scan shows an anechoic mass (arrows) located posterior to the uterus. (b) Transverse pelvic section reveals absence of internal vascularization (arrows) on color Doppler ultrasound (US). (c) Longitudinal scan of the sacrococcygeal region demonstrates the absence of the coccyx (asterisk). The mass (arrows) arises from the anterior aspect of the spine and protrudes into the presacral space, suggestive of anterior sacral meningocele. (d) Longitudinal scan of the lumbosacral region indicates a low-lying cord (conus tip at L3, arrow). (e) Transperineal US depicts narrowing of the anal canal (long arrow) posterior to the urethra (short arrow)

and abscess. US may demonstrate the characteristic appearance of duplication: the lesion is adjacent or adherent to the rectum, and the wall is stratified with an inner echogenic mucosa and a hypoechoic smooth-muscle layer externally.[28] Rectal duplications are morphologically classified into cystic and tubular types. The cystic type is more common and typically lacks communication with the rectum, whereas tubular duplications usually communicate with the lumen of the adjacent intestine. In addition, rectal duplications can be categorized anatomically as complete (with separate luminal openings) or incomplete (characterized by a blind ending or an abnormal connection). Rectal duplications communicating with the perianal skin are extremely rare. Rectal duplication can be associated with abnormalities of the genitourinary system, occasionally combined with bladder duplication [Figure 7]. Therefore, it is necessary to optimize visualization of the genitourinary system with a full-bladder.

Neonatal perianal abscess

Neonatal perianal abscesses show overwhelming male predominance, with a male-to-female ratio of 9:1. The infection commonly derives from the anal crypt and then invades into the perianal subcutaneous tissue, leading to the formation of a perianal abscess. Some perianal abscesses eventually develop into fistulas with external or internal openings. Fistulas usually cannot be diagnosed based on clinical exam only, particularly in awake infants. Transperineal US has proven to be an excellent noninvasive technique to assist in the evaluation of perianal abscesses and fistulas.[3] High-frequency US provides detailed insights into the size and location of the lesion, its relationship with the anal sphincter, as well as clarifies the presence of a fistula and internal opening. An abscess is identified as an irregular heterogeneous hypoechoic area with internal anechoic fluid collections. A fistula appears as an avascular hypoechoic linear area surrounded by echogenic fat. The fistula can be tracked to identify the internal opening. The presence of gas outside the intestinal wall indicates the existence of a fistula.

Role of other imaging modalities

While US is a valuable initial imaging tool for assessing perianal diseases in infants, it may not always provide a definitive diagnosis. In many cases, further imaging studies are necessary to fully evaluate the extent of the disease and obtain a comprehensive diagnosis. Modalities such as computed tomography (CT), MRI, and contrast imaging techniques play crucial roles in these situations.

For the evaluation of sacrococcygeal teratomas, CT provides a rapid and detailed assessment of the tumor's size, location, and its relationship with adjacent structures, such as the sacrum, coccyx, and surrounding soft tissues. It is particularly useful for detecting calcifications within the tumor and assessing any potential involvement of pelvic structures or adjacent bone. However, CT is less adept than MRI in delineating the tumor's soft tissue components or in assessing the spinal cord and neural structures.[29]

MRI plays a critical role in the diagnosis and evaluation of both Currarino syndrome and occult spinal dysraphism,[30,31] offering unparalleled soft tissue contrast and the ability to assess presacral masses. MRI allows for clear visualization of the spinal cord and its attachments, helping to confirm the presence of tethering and evaluate the degree of traction on the spinal cord.

Techniques such as voiding cystourethrogram, distal colostogram, and fistulography are important diagnostic tools for anorectal malformations. These imaging modalities offer a visual map of the blind-ending rectal pouches, fistulas and their potential impact on surrounding structures, which is crucial for surgical planning.[32,33]

Thus, combining US with these additional imaging techniques allows for a more comprehensive evaluation and appropriate management of perianal diseases in infants. A multimodal imaging approach ensures that clinicians have a clear understanding of the extent of the malformation and its potential effects on surrounding structures.

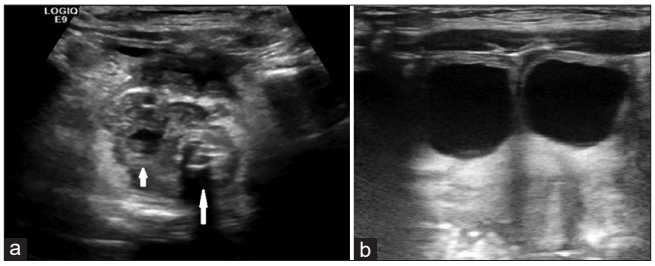

Figure 7: The transverse sonogram of the pelvis reveals the ultrasound appearance of rectal and bladder duplication in a 1-month-old girl. (a) On the left is a gas and feces filled rectum (long arrow). On the right is a tubular structure (short arrow) with a thick and stratified wall, sharing a common wall with the rectum. (b) The transverse sonogram of the pelvis shows bladder duplication

Conclusion

Here, we present a review of the sonographic technique and imaging characteristics of neonatal perianal lesions. US can offer detailed information regarding the recognition and localization of neonatal perianal tumors, malformations, and infections. Given the growing concerns regarding radiation exposure and invasiveness in children, US can be used as the preferred imaging modality to realize safe and effective evaluation of perianal diseases in infants. To ensure accurate characterization of the perianal lesions in infants, radiologists need to be familiar with the normal perianal anatomy and pathology, and pay extra attention to thorough scanning of the sacrococcygeal region, spinal cord, and genitourinary system.

Ethics statement

The study was conducted in accordance with the Declaration of Helsinki and was approved by the Capital Institute of Pediatrics Institutional Review Board (approval code: SHERLLM2024028; approval date: August 15, 2024). The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients have given their

consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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