

Ectopic anus with barrel gun perineum rare type of anorectal anomaly

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ABSTRACT

Perineal ectopic anus in female infants is not a very uncommon congenital anorectal anomaly with opening into the low vaginal or vulvar region. However, ectopic anus with barrel gun perineum is a less common variety. Patients generally present with frequent history of constipation, but may seek medical help for just aesthetic reasons. We present here one such case of an asymptomatic seven years old female with the rare form of anterior ectopic anus with barrel gun perineum without any fistulous communications with an innovative method of demonstration of the anomaly by using ultrasound gel as rectal contrast in MRI pelvis.

CASE REPORT

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A 7 years old female child was referred from the department of surgery for MRI of the pelvis with the complaints of passing urine and stool from the same opening. There are no similar complaints in other siblings. Patient was born as a full term normal vaginal delivery at home. No other significant past history was noted. CNS / Respiratory / Cardiovascular / per abdomen examination revealed no significant abnormality.

Local perineal examination revealed a single opening of approximately 5 x 3cms in the perineal region. Urethra was seen in the anterior aspect of the opening and faecal matter in the posterior aspect of the opening.

Blood and urine investigations showed no significant abnormality. Renal function test was normal.

Patient was referred to the Radiology department for evaluation of the defect. The clinical concern was to demonstrate the anomalous anatomy and any fistulous communication between the urinary and gastro-intestinal tracts.

MRI of the pelvis was performed on a 0.35T MRI (SIEMENS Magnetom C). We performed MRI before (figure: 1a&b) and after (figures: 2a&b, 3a&b) instillation of 20cc of

sterilized ultrasound gel through the anal opening in the vulvar area to demonstrate the anomalous anatomy and to rule out anovestibular fistula. The procedure was well accepted. Informed consent was taken from the parents. Thin slice sagittal (figure: 2a&b) and axial (figure: 3a&b) sections were acquired. The gel was seen to be located within the rectum and also within the endometrial cavity. However, there was no evidence of fistulous communication between the rectum and the uterus. Urinary bladder was normal. No other significant pathology was noted in the pelvis. So, the final diagnosis of ectopic anus with barrel gun perineum (DENIS BROWNE CLASSIFICATION) was given. Surgeons successfully performed posterior sagittal anorectoplasty (PSARP) for aesthetic reasons. She is currently doing well.

DISCUSSION

Anorectal malformations (ARM) are rare birth defects concerning the anus and rectum. Approximately 1 in 2500 to 1 in 5000 new born babies are affected [1]. Ectopic anus with barrel gun perineum is an even rarer form of anterior ectopic anus in females. In this condition a normal anus and vagina lie with their edges touching, without the normal strip of skin intervening. Usually there is a dimple behind the anal opening to mark the normal site for the anal opening [2]. Patients generally presents with frequent history of constipation, but

may seek medical help for just aesthetic reasons [3]. We present here one such case of an otherwise asymptomatic seven years old female with the rare form of anterior ectopic anus with barrel gun perineum without any fistulous communications who sought medical help for aesthetic reasons.

Anorectal malformation (ARM) may present with a wide spectrum of defects, ranging from relatively low (translevator) malformation to very complex high (supralelevator) defects. Previous studies have shown that associated malformations are more frequent in 'High' defects that are complex and difficult to manage with poor functional prognosis than in low defects that are less complex and easily treated with an excellent functional prognosis. Associated malformations mainly include the genito-urinary system, spine and spinal cord, the rest of the gastrointestinal tract and the heart [4].

Although the diagnosis of ARM is made at birth, the situation is different in our country, where a large number of deliveries take place at home. A long term study noticed that all patients with delayed diagnosis had low type of ARM, as in our case [4].

The cause for the low type of ARM has been considered to be due to genetic variation with erratic migration, i.e., when a migrating structure leaves the correct path. A study found a few potential risk factors for ARM, including fever during pregnancy, maternal overweight, use of multivitamins, paternal smoking, and occupational exposures, but a familial component seems important as well [5].

Vestibular ectopic anus with barrel gun perineum (DENIS BROWNE CLASSIFICATION, see Table 3) is a low type of ARM in which the puborectalis sling of the levator ani is usually normally developed around the lower end of the bowel before this takes its abnormal terminal course. Satisfactory continence can be achieved as a result of this sling. In the perineal and vulval ectopic anus, Denis Browne called the 'shot-gun perineum' with both barrels alongside but functioning normally required no further treatment and an experience over many years with some patients having reached married life confirms that it is a fully acceptable situation. However, in cases of the vestibular ectopic anus where the orifice is immediately adjacent to the hymen as in our case and the bowel extends more directly up behind the vagina rather than going backwards for a considerable distance as in the vulval ectopic anus, the final anal orifice, although satisfactorily continent may be awkwardly placed for hygiene. In these cases a perineal transplantation of the anus to the normal site through a posterior sagittal approach (posterior sagittal anorectoplasty) can be carried out more satisfactorily in the older child who has already achieved continence and who understands the object of the operation and will cooperate in aftercare [6].

For such a complicated surgery, one should have a very clear picture of the anatomy, variations and any fistulous communication for a successful surgery. So, radiologists have an essential role to play in the patient management. Although MRI images have a very good tissue contrast, however, use of

ultrasound gel as rectal contrast improves the diagnosis and evaluation of anorectal anomalies, as seen in this case.

Differential diagnosis for ectopic anus with barrel gun perineum, a low type of anorectal malformation includes: rectovestibular fistula and persistent cloaca. In rectovestibular fistula, MRI shows a common wall between anterior wall of the rectum and the posterior wall of the vagina. Urethra is normal. In persistent cloaca, there is single common channel connecting rectum, vagina and urethra [7].

TEACHING POINT

Ultrasound gel can be used as rectal contrast in MRI of the pelvis for the better delineation of anorectal anomalies.

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FIGURES

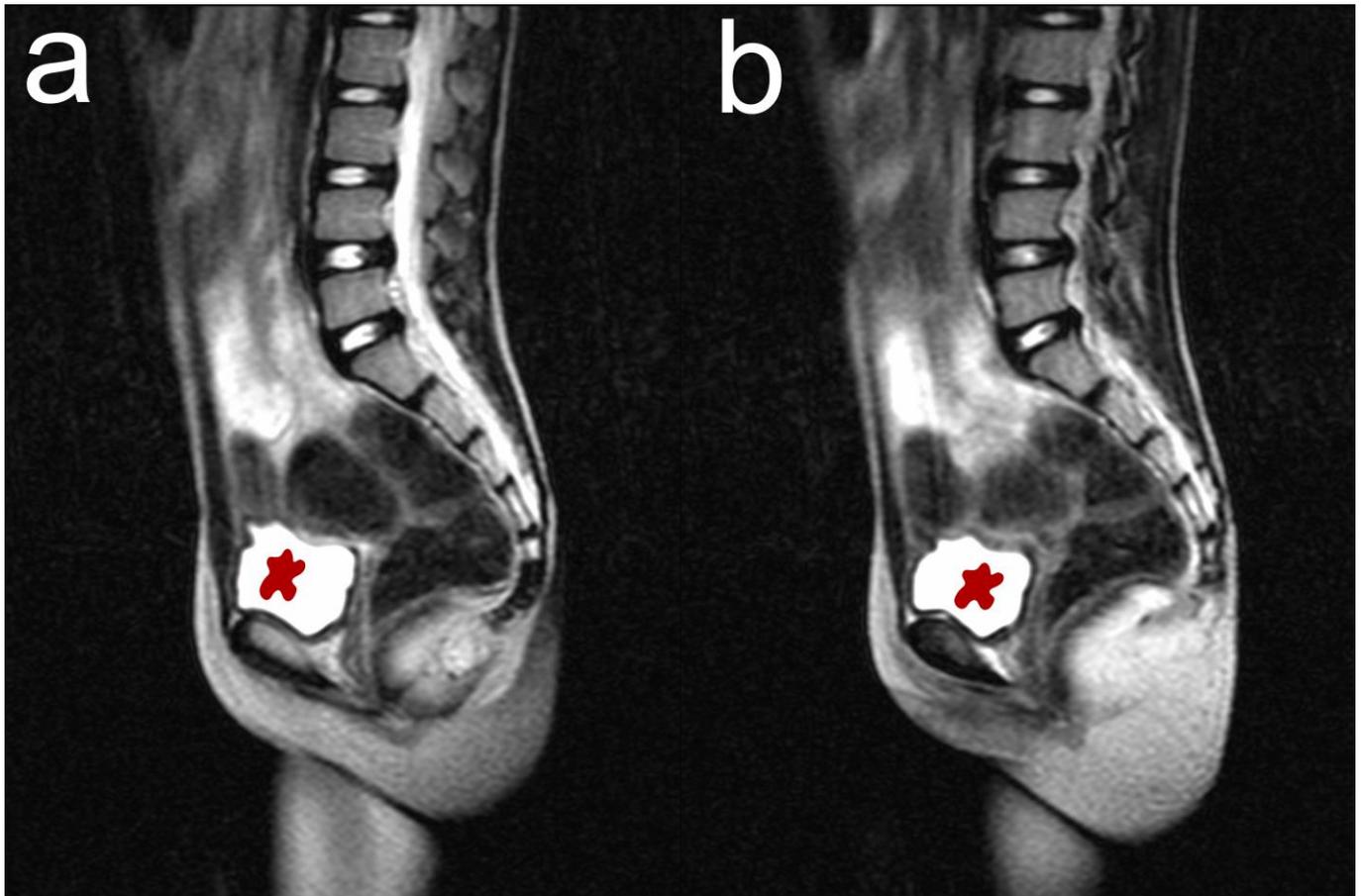


Figure 1: 7 years old female with ectopic anus and barrel gun perineum. MRI of the pelvis, STIR sequence in sagittal plane before instillation of ultrasound gel shows bladder (star) filled with urine acting as natural contrast. There is no abnormal hyperintense area seen in the pelvis. No obvious fistulous communication. (Protocol: 0.35T, TR 7330ms, TE 126ms, slice thickness 3mm, interslice gap 1.8mm)

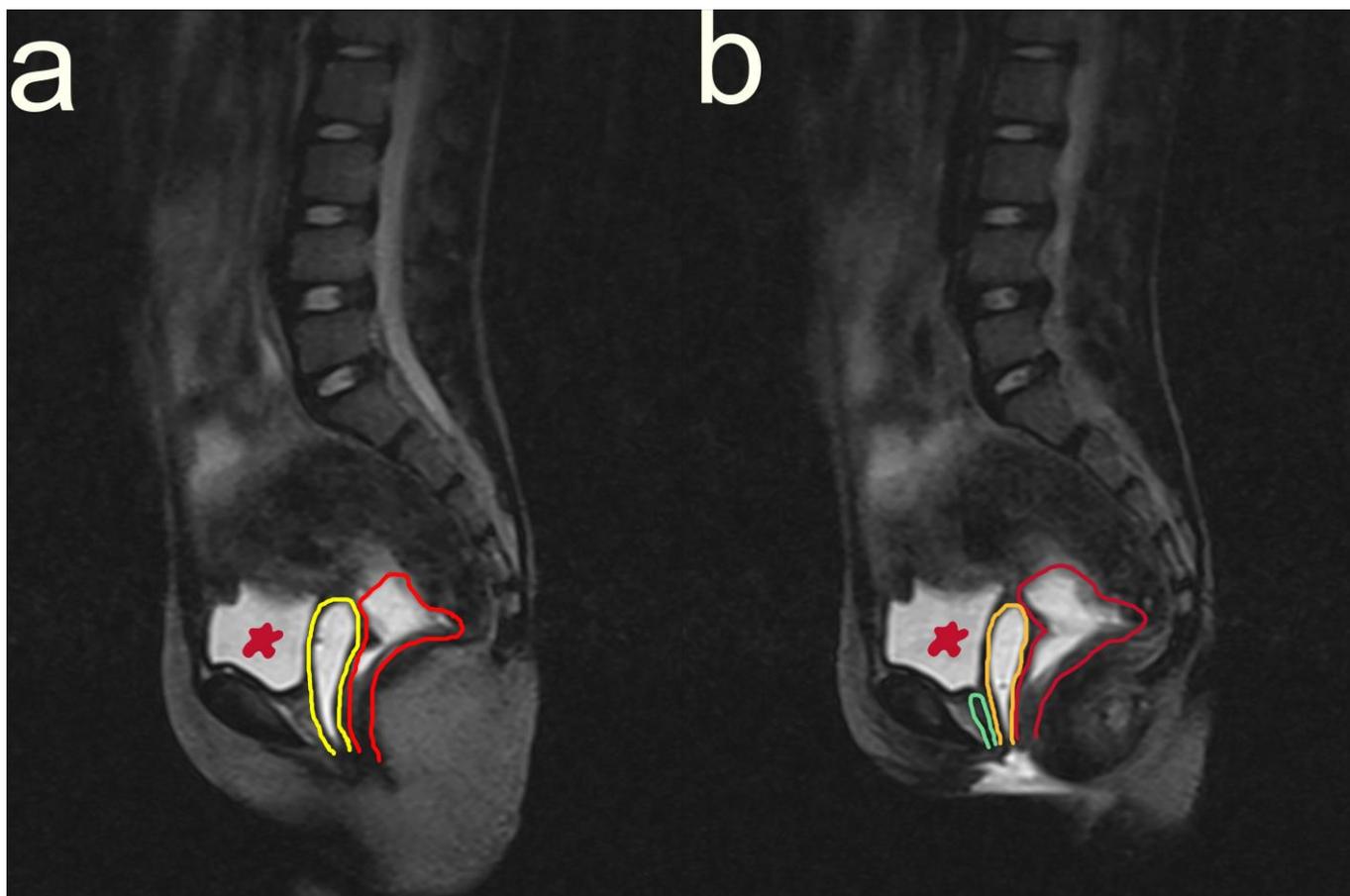


Figure 2: 7 years old female with ectopic anus and barrel gun perineum. MRI of the pelvis, STIR sequence in sagittal plane after instillation of 20cc of ultrasound gel shows gel to be located in the rectum (orange outline) and endometrial cavity (yellow outline). The fat plane between the uterus and rectum is well-maintained. There is no fistulous communication. (Barrel gun perineum). Urethra (green outline) and bladder (star) are normal. (Protocol: 0.35T, TR 7330ms, TE 126ms, slice thickness 3mm, interslice gap 1.8mm)

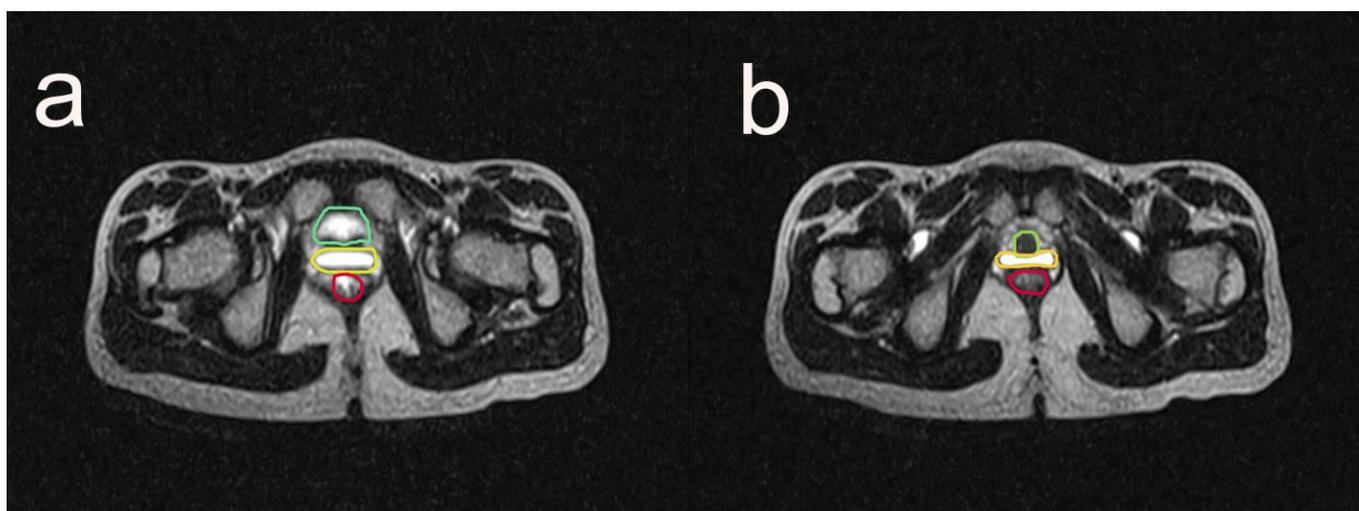


Figure 3: 7 years old female with ectopic anus and barrel gun perineum. MRI of the pelvis, STIR sequence in axial plane showing three separate tracts: urinary (green outline), vaginal (yellow outline) and bowel (orange outline). No fistulous communication. (Protocol: 0.35T, TR 7330ms, TE 126ms, slice thickness 3mm, interslice gap 1.8mm)

Definition	A low type of anorectal malformation in which the puborectalis sling of the levator ani is usually normally developed around the lower end of the bowel before this takes its abnormal terminal course. Satisfactory continence can be achieved as a result of this sling.
Etiology	Remains unclear and is likely multifactorial. Considered to be due to genetic variation with erratic migration, i.e., when a migrating structure leaves the correct path.
Incidence	Approximately 1 in 2500 to 1 in 5000 new born babies are affected with anorectal malformations. Ectopic anus with barrel gun perineum is an even rarer form of anterior ectopic anus in females.
Gender ratio	1: 3
Age predilection	Congenital malformation.
Risk factors	Fever during pregnancy, maternal overweight, use of multivitamins, paternal smoking, and occupational exposures, but a familial component seems important as well.
Treatment	Seek medical help mostly for aesthetic reasons, i.e. Posterior sagittal anorectoplasty.
Prognosis	Prognosis is excellent.
MRI findings	Three separate tracts of urethra, vagina and rectum are seen. The rectum is seen just posterior to the vagina, slightly anterior than the normal posterior position of the rectum. There is no fistulous connection.

Table 1: Summary table for ectopic anus with barrel gun perineum

Differential diagnosis	MRI findings
Ectopic anus with barrel gun perineum	Three separate tracts of urethra, vagina and rectum are seen. The rectum is seen just posterior to the vagina, anterior in position than the normal posterior position of the rectum. There is no fistulous communication.
Rectovestibular fistula	Anterior wall of the rectum and the posterior wall of the vagina share a common wall. Urethra is normal.
Persistent cloaca	Rectum, vagina and urethra meet and fuse, creating a cloaca, a single common channel.

Table 2: Differential diagnosis table for ectopic anus with barrel gun perineum

Anorectal malformations in males	High/intermediate	<ul style="list-style-type: none"> ▪ Anorectal atresia with rectourethral fistula ▪ Anorectal atresia with rectovesical fistula ▪ Anorectal atresia without fistula ▪ Rectal atresia ▪ Cylindrical rectal stenosis ▪ Diaphragmatic rectal stenosis
	Low	<ul style="list-style-type: none"> ▪ Perineal anus ▪ Imperforate , stenotic or Denis Browne microscopic anus, complete covered anus ▪ Incomplete covered anus or anocutaneous fistula to scrotum
Anorectal malformations in females	High	Anorectal atresia with high suprahymenal vaginal fistula
	Intermediate	Subhymenal or vulvar anus or fourchette anus
	Low	Ectopic anus with barrel gun perineum (anal orifice is separated by vaginal opening by a tract of skin)

Table 3: Denis Browne classification of anorectal malformation [8]

ABBREVIATIONS

ARM = Anorectal malformation
CNS = Central nervous system
MRI = Magnetic resonance imaging
PSARP = Posterior sagittal anorectoplasty
STIR = Short tau inversion recovery

KEYWORDS

ectopic anus; barrel gun perineum; anorectal anomalies

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