Rare disease

Reverse (posterior) cloaca with congenital rectal stenosis and pseudohermaphrodisim: a rare entity with rare association

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Summary

Reverse (posterior) cloaca is itself a rare entity and association of this defect with other congenital syndromes or defects is extremely rare to find. To diagnose this unique anomaly one should be aware about this very infrequently encountered clinical entity. Reverse cloaca is a very rare variant of anorectal malformation in which urethra and vagina fused to form a urogenital sinus that diverts posteriorly and opens in the anterior rectal wall at the anus or immediately anterior to it. We report a neonate with the rare clinical entity of reverse cloaca type C, pseudophallus with rudimentary urethra as a component of female pseudohermaphrodisim and congenital rectal stenosis. Surprisingly, there was no associated urological abnormality on ultrasonography and laparotomy.

BACKGROUND

Cloaca represents the most complex deformity in female anorectal, vaginal and urogenital malformations. A typical cloaca is defined as a defect in which the urethra, vagina and rectum converge into one common channel that opens as a single orifice at the normal location of the urethra.¹ Typical cloaca is also a rare entity and occurs in 1:250 000 newborns.² The clinical entity we reported is far more rare and the opposite of typically described cloaca—that is, a urogenital sinus that has formed by the fusion of the urinary tract and the vagina diverts posteriorly and opens in the anterior rectal wall, at the anus or immediately anterior to it. In these cases, position of the rectum is essentially normal or may be slightly anterior in location. The majority of these patients have associated important urinary tract malformations.³ To the best of our knowledge, this is probably the first reported case of posterior cloaca along with congenital rectal stenosis and female pseudohermaphrodisim in the English language literature.

CASE PRESENTATION

A 3-day-old neonate presented to our department with complaints of genital ambiguity and abdominal distension. The weight of the baby was 2.4 kg. On asking for the detailed clinical history, we found that the patient was passing both urine and fecal matter from a normally placed anal opening. The baby was actively crying and taking oral feeds normally, but there was history of a on and off abdominal distension and constipation. On systemic examination, there was no other apparent congenital anomaly. Perineal examination revealed a normally placed anal opening, and a hypertrophied clitoris (pseudophallus) with a rudimentary urethra, which was present at a location where the urinary tract was supposed to be open (figures 1 and 2). The sacrum was well formed and no abnormality was seen in

the spine. As the patient was having on and off abdominal distension with constipation, we got a lower gastrointestinal contrast study done, which showed a stenosed lower rectum and grossly dilated upper rectum and sigmoid along with an urogenital opening (figure 3).

INVESTIGATIONS

Karyotyping was 46XX. Serum luteinising hormone, follicular stimulating hormone and testosterone hormone levels were normal. Ultrasonography of the abdomen showed a normal urinary tract. Further evaluation of the urinary tract with more sophisticated diagnostic tools could not be done due to financial limitations. Distal colostogram was also done after performing diverting colostomy, which showed a dilated rectal pouch (figure 4).

TREATMENT

Exploratory laparotomy was carried out for the distension of the abdomen with an additional aim of a mullarian structure search as the patient was also having genital ambiguity. Intraoperatively, we found that the patient had a grossly dilated rectum and sigmoid colon along with a normal uterus, bilateral ovaries and urinary bladder (figure 5). The patient had a staged repair due to his poor general condition and a diverting loop transverse colostomy was performed as a first stage procedure. Loop transverse colostomy was done for rectal stenosis.

OUTCOME AND FOLLOW-UP

The post-operative period was uneventful and the patient was discharged on the 7th postoperative day. At 2 months follow-up the patient is growing well with a colostomy. Final definitive repair via posterior sagittal transanorectal approach is the next step.

Figure 1 Clinical photograph showing pseudophallus with rudimentary urethra.

Figure 2 Clinical photograph showing normally located anus.

DISCUSSION

Posterior cloaca was first described by Leditschke and Pena.⁴ The term posterior cloaca was suggested by Pena and Kessler.³ Three anatomic variants of this defect are described: (A) fused urethra and vagina open into the anterior rectal wall, at the anus and is visible only endoscopically; (B) the urogenital sinus opens into the perineum

immediately anterior to the anus; and (C) the urogenital sinus opens either in the rectum or the perineum and there is a rudimentary urethra opening at the tip of a pseudophallus (as in our case).³ By definition, our case fits the type C defect.

We called it reverse cloaca because it is simply the reverse of the classical persistent cloaca. Posterior cloaca is a rare

Figure 3 Lower gastrointestinal tract contrast study showing rectal stenosis and urogenital sinus opening.

Figure 4 Distal colostogram showing dilated upper rectum and stenosed lower rectum.

anomaly but, in this patient, the associated defect made the combination exceptionally rare. Gender assignment and detection of life-threatening conditions (salt-losing crisis due to congenital adrenal hyperplasia or Wilms' tumour) are the main aims when we deal with ambiguous genitalia. The

main criteria for the diagnosis of female pseudohermaphrodisism are: normal female karyotype 46XX, masculinisation of external genitalia and no palpable gonads. One can reveal urogenital sinus and vagina on genitography, and uterus on ultrasound examination or exploratory laparotomy.⁵ In our

Figure 5 Intraoperative photograph showing dilated upper rectum, sigmoid colon, urinary bladder and uterus with bilateral ovaries.

case, there was 46XX female pseudohermaphrodite with normal testosterone and complete masculinisation of external genitalia, which is less frequently reported.⁶

The majority of the patients with posterior cloaca have urinary tract malformations. According to Pena and Kessler, 88% of these patients have urological defects. Hence, urological evaluation to detect these urogenital defects is recommended.³ Surprisingly, there was no urinary tract anomaly on ultrasonography in our patient. The kidneys, ureters and urinary bladder were found normal on sonography and laparotomy.

Further urological evaluation in the form of intravenous urogram, radioisotope renogram and/or MR urogram could not be done because of limited resources and the poverty of the patient. If these investigations would have been done they might have added some additional procedure depending upon the result of tests like nephrectomy for non-functional kidney.

Another associated defect in this patient was congenital rectal stenosis. Congenital rectal stenosis is a rare variant of anorectal malformations. Rectal atresia and rectal stenosis are very rare constituting 1–2% of the anorectal-malformations.⁷ In this congenital defect, the stenosed rectum is found placed within the levator muscle complex, the anal canal is normal in these patients and no fistula are usually found. The exact embryological mechanism for origin of this anomaly is not known, but it is thought to be secondary to vascular accident during gestational period. Genetic and environmental factors may be implacable according to some researchers.⁸

The reverse cloaca can be managed in single or multiple stages. Pena and Kessler had managed five patients by staged procedures and four patients in a single stage. They

have reported the longest series of posterior cloaca patients' management in English language literature.³ We did diverting transverse colostomy in our patient as her general condition was very poor. The next step is final repair via a posterior sagittal transanorectal approach as described by Pena.³

Learning points

- Reverse cloaca is an extremely rare entity and it is very hard to find this anomaly in day-to-day clinical practice.
- To diagnose this defect one should be aware of this anomaly and must examine perineum and external genitalia of the patient meticulously.

Competing interests None.

Patient consent Obtained.

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