

An Apparent Anus

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Port J Pediatr 2022;53:693-4

DOI: <https://doi.org/10.25754/pjp.2022.24647>

Keypoints

What is known:

- Constipation is a frequent symptom in children and is mostly of functional etiology.
- Awareness should be raised about the signs which indicate the need for exclusion of organic causes.

What is added:

- Although most cases of anorectal malformations are diagnosed during the neonatal period, 25% may be diagnosed later.
- Anorectal malformation must be considered during the evaluation of an infant or child with constipation.

Introduction

A 4-month-old female infant, without remarkable history, except for recurrent perineal candidiasis, presented a change in her defecation pattern. She maintained daily bowel movements but with increasingly harder and difficult to pass stools, associated with painful defecation. Dietary management of constipation was tried which proved ineffective. Following an episode of rectal bleeding, after painful defecation, by 12 months of age, an external hemorrhoid was detected and she was started on osmotic laxatives, with partial improvement. Routine examination by 18 months of age indicated no abnormal abdominal findings and no palpable feces. However, anterior implantation of an apparent anus was identified on the perineal examination (Fig. 1). The child was referred for pediatric surgery consultation which led to the diagnosis of imperforate anus with perineal fistula. Surgical correction was performed after the exclusion of urogenital malformations and congenital heart defects.

Although constipation in children is mostly a functional etiology, detailed history and a thorough physical examination should be performed to exclude an organic cause.¹ Moreover, the identification of an abnormal anal position should raise the suspicion of anorectal malformations and motivate early referral to pediatric surgery.

Anorectal malformations are congenital anomalies with a wide phenotypical spectrum, reportedly occurring in around one in 5000 live births. The etiology is not completely known, although it seems to be multifactorial and frequently associated with other congenital malformations, especially urinary tract malformations.²

Most cases of anorectal malformations are diagnosed during the early neonatal examination, but around 25% of milder forms are diagnosed later.^{2,3} Patients with later diagnosis may present with constipation often unresponsive to laxative treatment, abdominal distention, and rectal bleeding.³ Most anorectal malformations require surgical treatment. A delay in the diagnosis results in a higher risk of surgical and functional complications, with a consequent negative impact on the quality of patient life.⁴

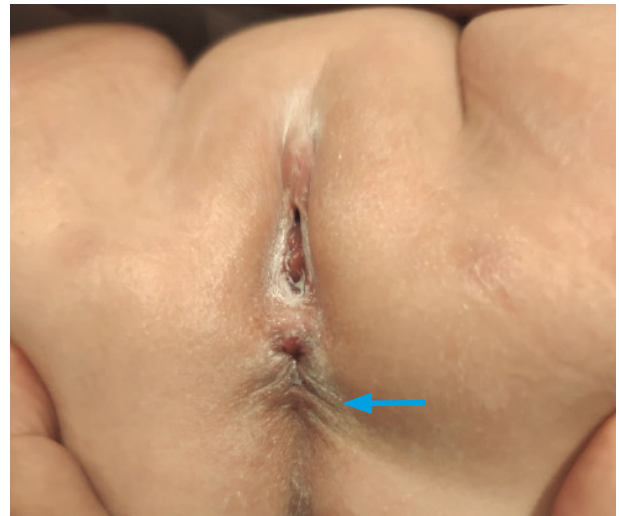


Figure 1. Perineal area presenting an anal dimple (arrow) and anteriorly a cutaneous fistula in close proximity with external genital apparatus. The child was contracting pelvic muscles while crying which allowed to observe anal dimple demarcated by folds from external sphincter muscle contraction, not observed during relaxation.

Keywords: Anus, Imperforate/diagnosis; Infant; Constipation/etiology

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Received: 30/06/2021 | Accepted: 20/10/2021 | Published online: 10/09/2022 | Published: 01/10/2022

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Author Contributions

CF, CA and BG participated in the study conception or design. CF, CA and BG participated in acquisition of data. CF, CA and BG participated in the drafting of the manuscript. NC participated in the critical revision of the manuscript. All authors approved the final manuscript and are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

Funding Sources

There were no external funding sources for the realization of this paper.

Protection of human and animal subjects

The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki 2013).

Provenance and peer review

Not commissioned; externally peer reviewed

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data

Acknowledgements

The authors would like to thank to Dr. Berta Bonet (Pediatric Surgery Department, Centro Materno-Infantil do Norte, Centro Hospitalar Universitário do Porto) for the image and collaboration in the diagnosis.

Awards and presentations

The present article was accepted for e-poster at Update em Medicina 2021 (22-25 April 2021).

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