Presacral Epidermoid Cyst in a Child with Anorectal Malformation: An Incomplete Currarino Triad

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ABSTRACT

Currarino syndrome is a hereditary disorder characterised by a triad of anorectal malformation (ARM), sacral vertebral defect, and presacral mass. An incomplete triad may also be present due to a variable phenotype. Presacral masses in children often include a number of differential diagnoses. An epidermoid cyst is rarely found in the presacral location. A congenital epidermoid cyst associated with ARM has rarely been reported. We report a case of a seven-month female with rectoperineal fistula, in whom a presacral mass was incidentally found during anterior sagittal anorectoplasty (ASARP). Histopathologic examination confirmed the diagnosis of an epidermoid cyst. This is a rare case of a congenital presacral epidermoid cyst associated with ARM, making it an incomplete Currarino triad.

Key Words: *Presacral mass, Anorectal malformation, Currarino triad, Epidermoid cyst*.

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INTRODUCTION

Presacral cystic masses include several developmental lesions, such as an epidermoid cyst, a dermoid cyst, chordoma, anterior meningocele, hamartoma, or a duplication cyst. Epidermoid cysts are rarely seen in presacral location. They develop from ectodermal remnants from the maldevelopment of adjacent structures.[1] These are rare in children. Till date, only a few cases have been reported.[2-5] While presacral masses are found in children, those with anorectal malformation (ARM) are often seen as a part of Currarino syndrome. We report a case of a female child with a rectoperineal fistula without any sacral defect in whom a presacral mass was found incidentally during surgery which later proved to be an epidermoid cyst on histopathology.

CASE REPORT

A seven-month female child with a weight of 6.5 kg, presented to OPD in April 2024 with the complaint of straining during defaecation since the last one month after weaning was commenced. It was her first visit to the institution. She was born *via* normal delivery at term. She was exclusively breastfed and weaning commenced at the 6th month of life. Developmental milestones were achieved according to age. She was vaccinated up to date and her past history was unremarkable.

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On examination, the abdomen was soft and mildly distended. Perineal examination revealed normal female genitalia, absent anal opening, and a perineal fistula. Buttocks were well-formed. Ultrasound KUB, x-ray of lumbosacral spine, and echocardiography were normal. No record of neonatal spinal ultrasound was available. There was no family history of congenital malformations.

The patient was prepared for a 2-stage repair. According to our hospital's protocol for anorectal malformation (ARM) with perineal fistula, anterior sagittal anorectoplasty (ASARP) was undertaken. A midline incision was made from the fistula to the proposed sphincter site after muscle stimulation confirmation. Fistula was delineated, separated from the vaginal wall and the rectum was mobilised. During posterior mobilisation of the rectum, a well-defined mass was noted in the presacral space, adherent to the rectum (Figure 1). The mass was tubular, 6 × 2 × 2 cm, and gave the impression of a rectal duplication cyst. During mobilisation of the mass, clear fluid was seen draining from its wall. The incision was extended in the posterior midline. The mass was separated from the rectum and excised in total without any breach to the rectal wall. No further fluid drain was observed. Anorectoplasty was done and a covering colostomy was made. Postoperatively, the patient was started on a prophylactic meningitic dose of antibiotics with the suspicion of anterior meningocele. The patient had an uneventful recovery; the wound was healthy and the stoma was functional. MRI of spine was done in the postoperative period which showed no spinal pathology. Histopathology later revealed a cyst wall with stratified squamous epithelium and subepithelial tissue showing flakes of keratin, thus confirming a presacral epidermoid cyst.

The patient was kept on anal dilation according to her age and after 5 months, she underwent colostomy closure. Continence could not be assessed at her age. No complications were noted in the patient.

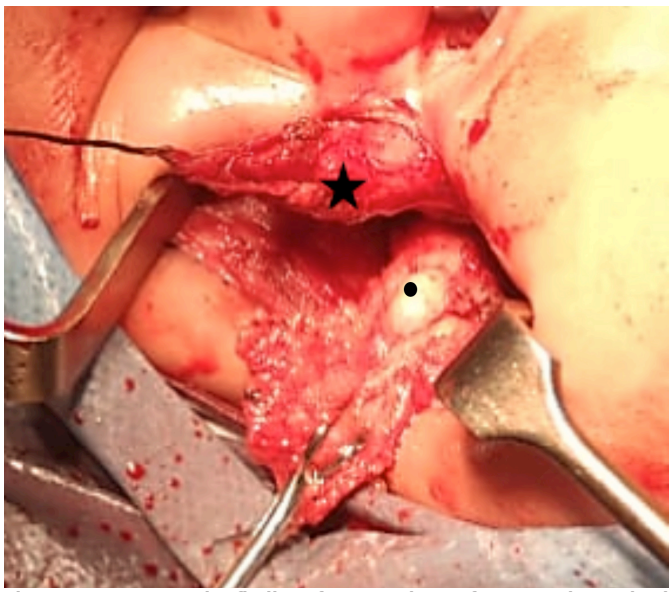

Figure 1: Intra-operative finding of presacral mass from anterior sagittal approach. The rectum (black star) has been retracted anteriorly with the presacral mass (black dot) lying posterior to it.

DISCUSSION

Presacral cysts are often congenital, lying between the rectum and sacrum. They arise from caudal embryological remnants. Presacral cysts include a variety of differential diagnoses, namely, sacrococcygeal teratoma, anterior sacral meningocele, neurogenic cyst, lipoma, rectal duplication, or dermoid cyst.[1] Presacral location is a rare site for epidermoid cyst. These are often asymptomatic, benign, lined with stratified epithelium, and filled with clear fluid. They are often mistaken to be either rectal duplication or anterior meningocele.[6] These cysts may cause symptoms due to compression effects on the rectum, urinary tract, or pelvic nerves. A 22-year study reviewed congenital presacral masses in children. Mature teratoma was seen in 64% and Currarino syndrome in 71%.[7] In another long cohort study, presacral masses were assessed in relation to ARM. Out of 46 patients with presacral masses, 12 had ARM. In this group, the tumour was found to be a mature teratoma, followed by a yolk sac tumour (YST) and lipoma. Presacral masses without ARM included teratoma, YST, ganglioneuroma, neuroblastoma, and one benign epithelial cyst.[8]

While isolated cases of presacral epidermoid cysts in adults have been reported, their incidence in the paediatric population is exceedingly rare. A literature search showed only a few such cases in the paediatric population, but none as associated with ARM.[2-4] We found one case report of a child with a para-rectal epidermoid inclusion cyst who had a past history of recto-vestibular fistula. She presented a gluteal abscess with deep communication with a cyst. However, the authors believed it was an acquired epidermoid cyst rather than congenital, as the cyst was not seen in previous investigations or multiple surgeries and had a significant history of gluteal abscess drainage in the past.[5]

Currarino syndrome is a rare hereditary disorder characterised by a triad of ARM, sacral vertebral defect, and pre-sacral mass. Constipation is a common presenting symptom. This triad is often suspected when x-ray sacrum shows a sacral notch.[9] In this patient, Currarino syndrome was not suspected due to a normal sacrum and no accompanying symptoms. However, it is noted that Currarino syndrome may present as an incomplete triad due to variable phenotype. Such cases may have one of the components missing, therefore, diagnosis is often missed or delayed. One similar case reported was of a male neonate with ARM, having a normal sacrum but a presacral mass diagnosed antenatally. The mass was found to be sacrococcygeal teratoma.[10]

Preoperative MRI of spine in the present patient was not undertaken as the sacrum was normal and the patient was asymptomatic. Postoperative MRI can rule out any underlying spinal defect.

The present case is unique because the presacral mass was found incidentally during surgery in a patient with ARM. This is a rare case of a child with ARM with a congenital presacral mass being an epidermoid cyst, suggestive of an incomplete Currarino triad.

PATIENT'S CONSENT:

Informed written consent was obtained from the parents of the patient to publish the data concerning this case.

COMPETING INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

SV: Data acquisition, manuscript writing, and literature reviewing.
SAB: Literature review, manuscript editing, and reviewing.
MAC: Conception and design of study, supervision, and reviewing.
All authors approved the final version of the manuscript to be published.

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