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Case Report

Surgery Section

Currarino Syndrome: A Silent Challenger to the Hirschsprung Paradigm

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ABSTRACT

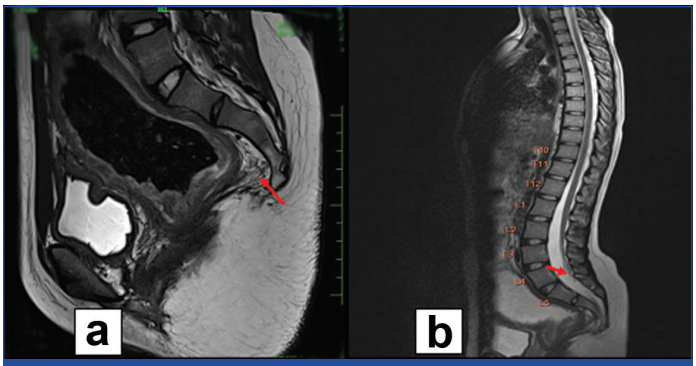
Currarino Syndrome (CS) is a rare congenital anomaly defined by a triad of anorectal malformations, presacral mass, and sacral bony defects. Although it typically presents in early childhood, adolescent cases are increasingly recognised but often missed. Due to significant overlap in clinical features, CS may be mistaken for more prevalent conditions such as Hirschsprung's Disease (HD). This diagnostic ambiguity often leads to prolonged evaluation, repeated testing, higher healthcare costs, and patient dissatisfaction. We present the case of an 18-year-old female with a 12-year history of severe, refractory constipation who was initially evaluated for HD. Cross-sectional imaging ultimately revealed sacral agenesis, a presacral mass, and a tethered cord, leading to a definitive diagnosis of CS. The patient subsequently underwent neurosurgical detethering and mass excision, resulting in substantial symptomatic relief. This case underscores the need to maintain a high index of suspicion for CS in adolescent patients with unexplained constipation, particularly when initial investigations are inconclusive. Early recognition is essential to ensure appropriate management and to avoid the burden of delayed diagnosis.

Keywords: Anorectal malformations, Case report, Chronic constipation, Presacral mass, Sacral agenesis, Tethered cord

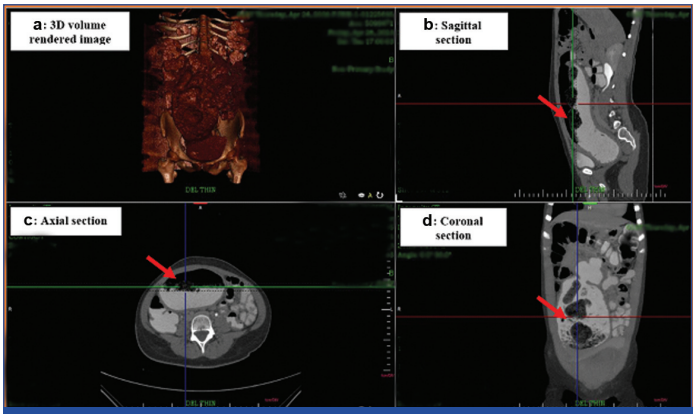
CASE REPORT

An 18-year-old female presented with a long standing history of chronic constipation, persisting for 12 years. She had previously consulted multiple physicians and had been managed conservatively with various treatment modalities, including dietary modifications, laxatives, and repeated enemas, all of which failed to provide lasting relief. Over the preceding three years, she developed generalised weakness, fatigue, and easy fatigability. She was evaluated at a peripheral facility and diagnosed with severe anaemia, for which she received multiple blood transfusions. Notably, there was no associated history of abdominal pain, vomiting, urinary or menstrual complaints, or gait disturbances. On examination, the patient weighed 35 kg and was clinically pale, with pallor evident in the palpebral conjunctivae, nail beds, and palms. Chest auscultation revealed normal bilateral air entry. Abdominal examination demonstrated a soft, non distended abdomen with a vague, palpable mass in the lower abdomen. Bowel sounds were normal. Digital rectal examination revealed a narrowed anal canal with normal tone, although further assessment was limited due to the inability to negotiate the examining finger. Spinal examination was unremarkable, but focused neurological assessment identified diminished to absent perianal sensation. Because of her clinical features, a provisional diagnosis of HD was made, and investigations were directed accordingly.

Initial laboratory evaluation revealed severe anaemia, with a haemoglobin level of 2.8 g/dL. In view of the anaemia, the patient received multiple blood transfusions concurrently as further diagnostic workup was undertaken. Abdominopelvic ultrasonography showed grossly dilated bowel loops in the iliac fossae. Pelvic Magnetic Resonance Imaging (MRI) with Magnetic Resonance (MR) identified a large, heterogeneous, non-enhancing lesion measuring 25×7.5 cm anterior to the lumbar spine, consistent with an anterior sacral meningocele [Table/Fig-1]. Contrast Enhanced Computed Tomography (CECT) of the pelvis revealed marked dilatation of the sigmoid colon and rectum with faecal loading [Table/Fig-2]. Non Contrast Computed Tomography (NCCT) of the lumbosacral spine demonstrated a left-sided sacral scimitar deformity with preservation of the S1 vertebral segment [Table/Fig-3]. No additional urogenital malformations were noted.

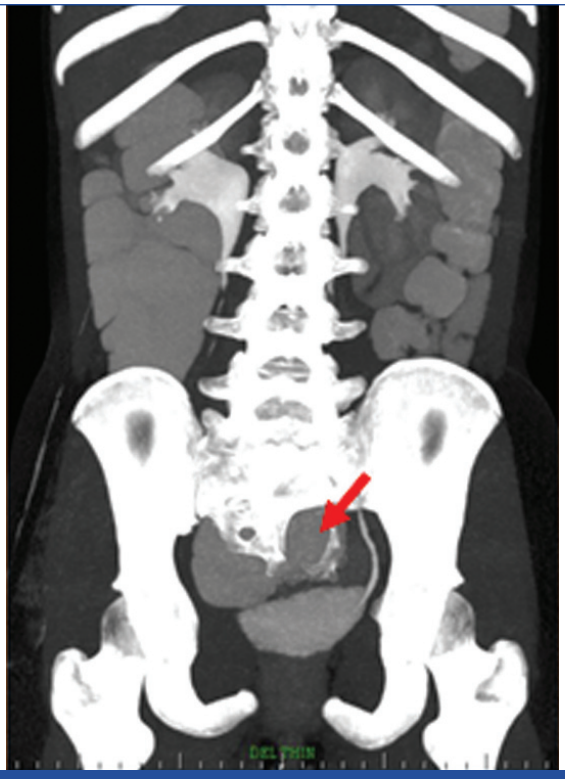


[Table/Fig-1a]: Magnetic resonance imaging of the pelvis demonstrated a presacral lesion, likely representing an anterior meningocele, with proximal colonic dilatation marked by the red arrow.
[Table/Fig-1b]: Magnetic resonance myelography reveals low lying tethered cord terminating at the L5-S1 junction.



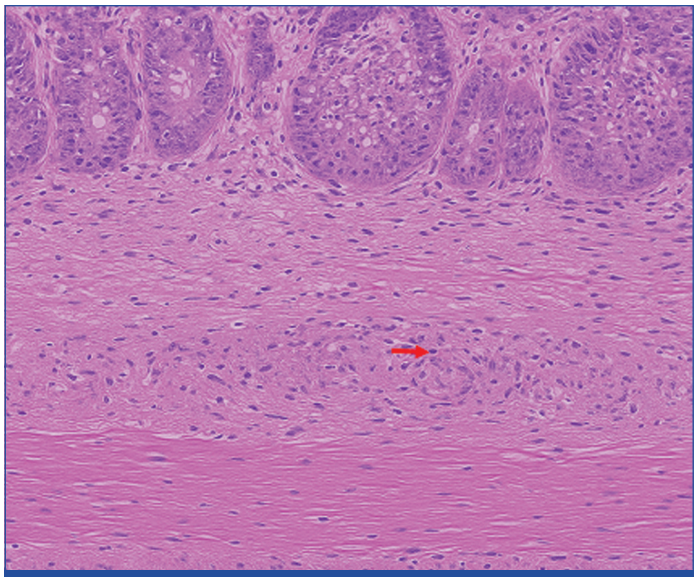
[Table/Fig-2a-d]: CECT-pelvis images showing marked dilatation of sigmoid colon and rectum with faecal impaction marked by the red arrow.

Further evaluation included an echocardiogram, which ruled out associated cardiac anomalies. Sigmoidoscopy revealed a stenotic anal canal. Full thickness rectal biopsy demonstrated the presence of ganglion cells, thereby excluding HD [Table/Fig-4]. Anorectal manometry showed a type II dyssynergic defecation pattern [Table/Fig-5]. The patient was optimised preoperatively and scheduled for surgical intervention. A posterior midline approach was used by



[Table/Fig-3]: NCCT pelvis showing a left sacral scimitar deformity with preservation of the S1 vertebra as indicated by the red arrow.

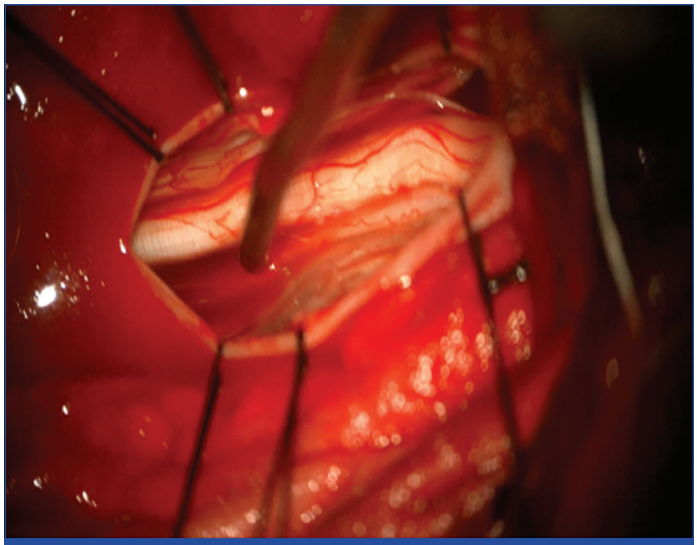
the neurosurgical team to expose the sacrum. Laminectomy of S2 to S4 vertebrae revealed an anterior dural outpouching, herniating through a 2×3 cm sacral defect, consistent with an anterior meningocele. No neural elements were found within the sac. The meningocele was disconnected from the dural sac, marsupialised, and the spinal cord was then detethered as shown in [Table/Fig-6]. The dural tube was then closed in a watertight fashion. Layered muscle and skin closure was performed. Subsequently, a diversion loop sigmoid colostomy was created in the left iliac fossa and matured.



[Table/Fig-4]: High-power photomicrograph (H&E stain, 40×) of a full-thickness rectal biopsy. The mucosa, submucosa, and muscularis layers are clearly seen with presence of ganglion cells within the submucosal plexus (indicated by red arrow), confirming the presence of enteric innervation and ruling out HD.

Pre-procedure Anorectal manometry:	Post-procedure Anorectal manometry:
- Rectal pressure: 19mmHg	- Rectal pressure: 22mmHg
- Dyssenergia type-II	- Dyssynergia type IV
- Absence of urge at 500ml	- Absence of urge at 500ml

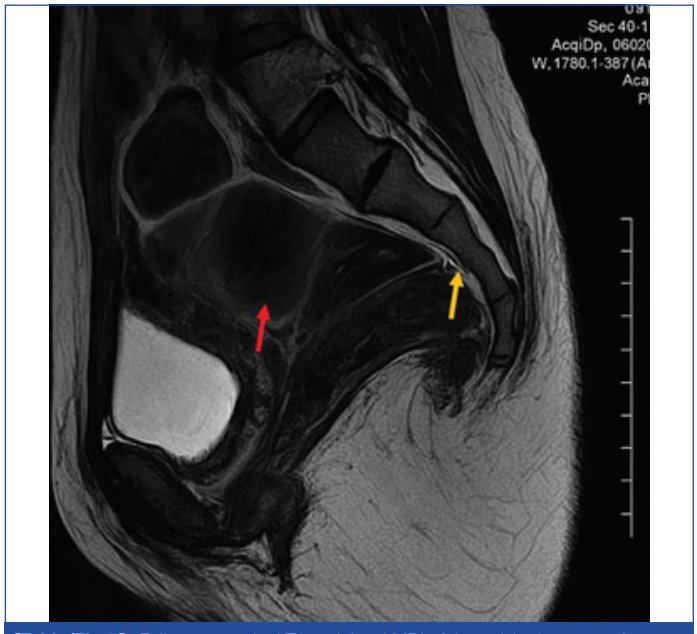
[Table/Fig-5]: Anorectal manometry findings both pre and post procedure.



[Table/Fig-6]: Intraoperative image under magnification showing the dural tube being opened and the cord being detethered.

The immediate postoperative period was uneventful, with no neurological deficits. Bowel function through the stoma resumed by postoperative day two, and the patient was mobilised on the same day. Oral intake was well tolerated by day three. The surgical site healed well, and she was discharged on the fifth postoperative day with instructions for regular anal dilatation. Over the following six weeks, gradual dilatation achieved the desired anal canal diameter. Three weeks postoperatively, colostomy reversal was performed to restore bowel continuity. Post reversal recovery was uneventful; the patient passed stools normally via the anal canal.

On follow-up, she demonstrated clinical improvement with weight gain and increasing haemoglobin levels. A repeat MRI performed at three months postoperatively showed complete resolution of the presacral mass and significant reduction in sigmoid colon dilatation [Table/Fig-7].



[Table/Fig-7]: Follow-up sagittal T2-weighted MRI of the pelvis demonstrating resolution of the previously noted presacral mass (As indicated by the yellow arrow) and reduction in sigmoid colon dilatation (As indicated by the red arrow).

DISCUSSION

Chronic constipation in adolescent females is a common clinical problem that is often addressed symptomatically without adequate diagnostic evaluation. While functional gastrointestinal disorders, dietary factors, and pubertal hormonal changes account for the majority of cases, in the absence of a clear diagnosis, it is crucial to consider less common but clinically significant structural or

syndromic etiologies [1]. Conditions such as delayed-onset HD and CS may present subtly during adolescence and are often overlooked. Reliance on over-the-counter laxatives and home remedies without medical supervision can further obscure the clinical picture, resulting in delayed recognition and appropriate management [2].

Currarino Syndrome (CS) is a rare congenital anomaly defined by a triad comprising anorectal malformations, a presacral mass, and sacral bony defects [3]. First described by Guido Currarino in 1981, the syndrome was previously noted in clinical literature by Kennedy, Ashcraft, and Holder, who observed similar constellations of symptoms without classifying them as a single entity [4]. CS is considered part of the caudal regression spectrum and is typically inherited in an autosomal dominant pattern. The HLXB9 homeobox gene on chromosome 7q36 has been identified as the genetic basis, with variable expressivity and incomplete penetrance accounting for the heterogeneity in clinical presentation [5]. In contrast, HD arises from a failure of neural crest cell migration during embryogenesis, leading to aganglionosis of the distal bowel and subsequent functional obstruction. It may occur sporadically or as part of familial inheritance, with implicated mutations in genes such as RET, EDNRB, EDN3, and SOX10, contributing to its variable phenotype and association with syndromic forms [6].

The clinical overlap between CS and HD presents a significant diagnostic challenge. Both conditions can manifest as refractory constipation, sometimes from infancy. However, CS may feature subtle neurological signs such as perianal hypoesthesia, or symptoms suggestive of spinal dysraphism, which are typically absent in HD. Moreover, while HD results from aganglionosis of the distal colon, CS involves mechanical obstruction from a presacral mass or anorectal narrowing [7].

Similar cases of diagnostic confusion between HD and CS have been well documented in the literature. Saberi A et al. detailed the case of an 18-year-old female who had been managed for presumed HD for over 17 years, in the absence of confirmatory histopathology [8]. It was only after advanced cross-sectional imaging that defining features of CS were revealed, allowing for definitive surgical management. Similarly, Djordjevic I et al. reported a three-year-old male who was initially treated with a colostomy for suspected congenital megacolon [9]. Further radiological evaluation subsequently unveiled the complete Currarino triad, redirecting the clinical course towards appropriate surgical correction. The differentiating features between CS and HD are shown in [Table/Fig-8] [10].

Features	Hirschsprung's Disease (HD)	Currarino Syndrome (CS)
Genetic Basis	Associated with mutations in RET proto-oncogene, EDNRB, and SOX10; often sporadic but can be familial	Typically autosomal dominant with mutations in MNX1 gene (formerly HLXB9); familial clustering common
Primary symptom	Chronic constipation due to functional obstruction	Chronic constipation due to mechanical obstruction
Neurological Signs	Absent	May be present (e.g., perianal hypoesthesia, urinary symptoms)
Aetiology	Aganglionosis of distal colon (neural crest cell migration defect)	Triad of anorectal malformation, presacral mass, sacral anomaly
Key clinical findings	Abdominal distension, explosive stools on Digital Rectal Examination (DRE)	Anal stenosis, anteriorly displaced anus, palpable presacral mass
Imaging modality of choice	Contrast enhanced CT scan, Rectal biopsy	MRI of lumbosacral spine and pelvis
Radiological findings	Transition zone between narrow aganglionic and dilated proximal bowel	Presacral mass, sacral agenesis, spinal cord anomalies
Definitive diagnostic tool	Rectal biopsy showing absence of ganglion cells	MRI and physical findings; biopsy not typically diagnostic
Digital rectal exam	Tight anal sphincter, explosive stool upon withdrawal	Anal stenosis, mass may be palpable anterior to rectum
Surgical management	Pull-through procedures (e.g., Soave, Duhamel)	Excision of presacral mass, anorectal reconstruction, possible spinal detethering

[Table/Fig-8]: Characteristic differentiating features between Currarino Syndrome (CS) and Hirschsprung's Disease (HD).

Diagnostic evaluation for CS must be methodical and multimodal. Plain pelvic radiographs may reveal characteristic sacral defects, including the "scimitar sacrum" sign, a hallmark radiologic finding. MRI is the gold standard, providing detailed anatomical information about the presacral mass, sacral defect, spinal anomalies (such as tethered cord), and potential connections with the neural axis [11]. In contrast, rectal biopsies-central to diagnosing HD may be misleading in CS, particularly when ganglion cells are present [12].

Management of CS is tailored to the specific components identified. Anorectal anomalies are usually addressed surgically via Posterior Sagittal Anorectoplasty (PSARP), while the presacral mass, if non-neural, can often be excised in the same sitting [13,14]. However, if the mass represents a meningocele or is intradural, neurosurgical input is essential, and staged procedures are recommended to avoid complications such as meningitis. In our case, the mass was resected first, followed by creation of a diverting sigmoid colostomy, and eventually a colostomy reversal after clinical stabilisation. The patient tolerated all procedures well and showed marked symptomatic improvement without postoperative neurological compromise. Despite increasing awareness, the diagnosis of CS is often delayed in the Indian healthcare setting due to several factors. These include over-reliance on symptomatic management of constipation, limited access to advanced imaging modalities in peripheral or resource constrained regions, and a lack of familiarity with the syndrome among general practitioners and paediatricians. Societal taboos around anorectal symptoms in adolescent females may further delay timely medical consultation.

CONCLUSION(S)

Currarino syndrome remains a rare but critical differential diagnosis in adolescents presenting with chronic constipation. Its clinical resemblance to HD, particularly in the absence of classical triad features, can lead to delayed or incorrect diagnosis. This case highlights the importance of a high index of suspicion and the indispensable role of imaging in guiding accurate diagnosis. Early recognition and multidisciplinary management are key to improving outcomes and preventing long-term complications.

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