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Currarino syndrome or Hirschsprung disease: how to prevent diagnostic dilemma in chronic constipation

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Abstract

Background: Currarino syndrome is a rare congenital malformation having autosomal dominant inheritance. It comprises of anorectal malformation, presacral mass, and sacral vertebral defect occurring in variable proportion. The most common presentation is chronic constipation which is usually due to compression of rectum by anterior sacral mass. If clinical examination is not properly done and digital rectal examination is excluded from the examination, it can be misdiagnosed as other common cause of constipation like Hirschsprung disease.

Case presentation: We are reporting one such case of one-and-half-year-old female child with chronic constipation which was initially managed as Hirschsprung disease, but later on, after a repeat clinical examination with digital rectal examination, it was evaluated on the line of Currarino syndrome. The diagnosis was confirmed by contrast-enhanced computed tomography of abdomen with 3 dimensional reconstruction. It was then managed by posterior sagittal approach with excision of mass and anorectoplasty.

Conclusion: A proper protocol for clinical evaluation of patient with constipation prevents diagnostic dilemma between surgical causes of constipation in pediatric age group. Digital rectal examination must be included in the protocol for evaluation of chronic constipation. In pediatric age group, clinical workup should be done with keeping in mind the rare diagnosis of Currarino syndrome along with common cause of constipation like Hirschsprung disease.

Keywords: Colostomy, Constipation, Hemi-sacrum, Pediatric, Pre-sacral mass

Background

Currarino syndrome is a rare congenital malformation characterized by anorectal malformation, anterior sacral mass, and sacral defect. The anterior sacral mass may be anterior sacral meningomyelocele, teratoma, enteric cyst, or combination of these [1]. Anorectal malformation may be in the form of anal stenosis, recto-vestibular or rectovaginal fistula, or imperforate anus [2]. The usual presentation is chronic constipation. Sometimes, when the anterior sacral mass is not obvious and presentation is chronic constipation, this entity can be mistaken as Hirschsprung disease which is the common cause of constipation in pediatric age group. We are reporting one such case of chronic constipation which was initially interpreted as Hirschsprung disease (HD) but was confirmed to be Currarino syndrome only after a repeat per rectal examination and contrast-enhanced computed tomography (CECT) of abdomen and pelvis with 3D reconstruction.

Case presentation

A one-and-half-year-old female child presented with complains of abdominal distension for 5 days. She was also having history of chronic constipation since birth. She was passing stools with suppositories and enemas. She also had history of delayed passage of meconium. With history of delayed passage of meconium and

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chronic constipation, we proceeded further with suspicion of Hirschsprung disease. On clinical examination, there was presence of soft and mildly distended abdominal. On perineal examination, anal opening was present, but it was narrow, located slightly anteriorly. She underwent divided sigmoid colostomy after routine workup. In view of uniform dilatation of colon till peritoneal reflection, multiple biopsies were taken. The biopsy was showing presence of ganglion cells but with hypertrophied nerve bundles suggesting a diagnosis of Hisrchsprung disease. Patient did well in post-operative period and was passing stool per stoma normally. After 4 months of follow-up, she was then planned for Duhamel pull through with intra operative frozen section biopsy. One day prior to surgery, per rectal examination was done in which anal canal could not be negotiated. So, diagnosis of Hirschsprung disease was questionable. She was planned for further evaluation with CECT pelvis with 3D reconstruction in view of rectal atresia or any other pelvic pathology. CECT revealed presence of anterior sacral cystic lesion, likelihood of anterior meningomyelocele (Fig. 1). 3D reconstruction of CECT also revealed presence of sacral boney defect as scimitar sacrum (Fig. 2). With the above findings, a diagnosis of Currarino syndrome was made. Patient was then planned for excision of mass through posterior sagittal approach. Intraoperatively, the findings of CECT were confirmed. After carefully removing the anterior sacral meningomyelocele, the rectum was clearly visualized. It

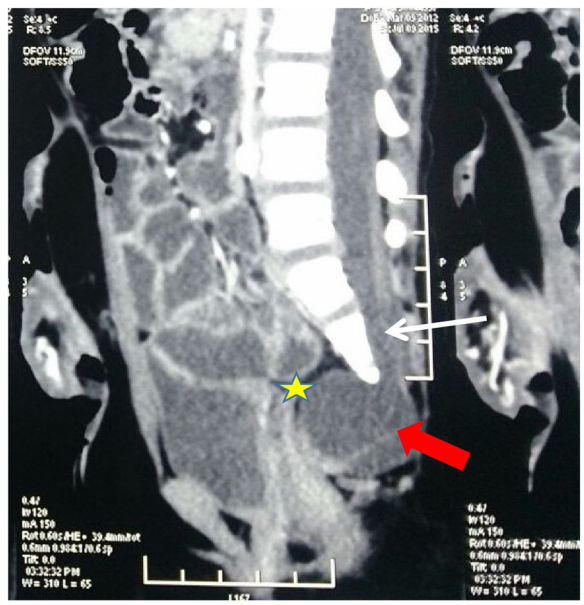

Fig. 1 CECT pelvis (sagittal view) showing the cystic lesion [red arrow] continuous with the spinal CSF space with cord tethering [white arrow] consistent with myelomeningocele and anteriorly compressed rectum [yellow asterisk]

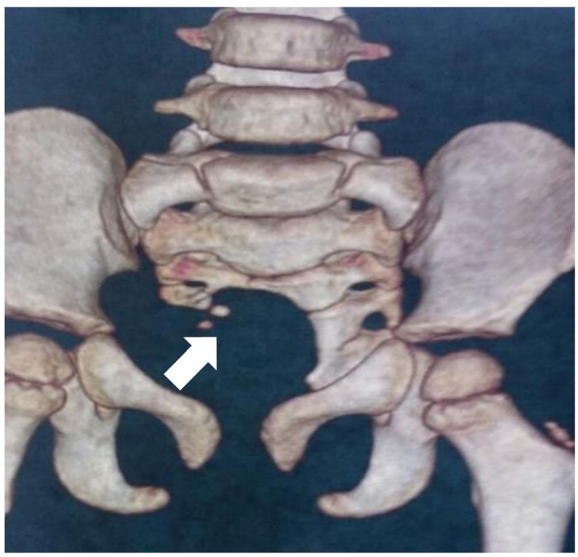

Fig. 2 3D reconstructed view of CECT pelvis showing sacral bony defect (scimitar sacrum) [white arrow]

was then decided to do posterior sagittal anorectoplasty (PSARP) which was completed with usual steps. Post-operatively, patient recovered well. Three months after PSARP, stoma was closed to restore bowel continuity. In post-operative period patient was put on bowel management program with saline rectal washes. At 2-year follow-up, patient is socially continent with saline rectal washes.

Discussion

Currarino syndrome is a rare congenital anomaly characterized by triad of anorectal malformation, pre sacral mass, and sacral defect [3]. These three entities can be present in different proportion. The anorectal malformation may be anal atresia, anal stenosis, recto-vestibular or rectovaginal fistula, or imperforate anus. The anterior sacral mass may be anterior sacral meningocele, teratoma, or an enteric cyst. The sacral defect may be in the form of sickle-shaped sacrum, scimitar shaped sacrum, crescent deformity, or sacral agenesis.

Currarino syndrome is an autosomal dominant condition and its locus is HLXB9 homeobox gene [4]. Variable gene penetrance has been explained as the possible cause of incomplete form of syndrome with absence of one or two clinical features [5, 6]. This syndrome has been described to run in families in few case reports further supporting the genetic association [7, 8]. Various theories have been proposed for embryogenesis of this syndrome. Currarino et al. [1] explained the embryogenesis based on split notochord syndrome in which there are abnormal endo-ectodermal adhesions and notochord defects in fetal life leading to fistula between gut and

spinal canal. The anorectal anomaly results as a consequence of these abnormal adhesions.

Chronic constipation is the most common presentation. It is usually present since early life. The constipation is usually due to the external compression by anterior sacral mass but anal stenosis or tethered cord may also lead to this. In our case, the cause of constipation was the presacral mass which was compressing the rectum. The absence of right hemi-sacrum was also a contributing factor. However, it was misunderstood as Hirschsprung disease because on clinical examination, there was presence of anal opening, no obvious mass was seen and patient had history of chronic constipation. Moreover, the biopsy which was sent intra-operatively was showing the presence of hypertrophied nerve trunks in the presence of ganglion cells which also added to confusion. Association of Currarino syndrome with Hirschsprung disease has also been reported. Baltogiannis N reported first case of such association in 2003 [9]. However, in our case, it was not an association; rather it was a misdiagnosis as HD due to a poorly performed clinical examination. The possible reason of presence of hypertrophied nerve bundles in biopsy may be chronic constipation.

Digital rectal examination is mandatory in cases of chronic constipation so as to have better picture of differential diagnosis. In Currarino syndrome, there will be presence of anal stenosis with opening not admitting the examiner's finger. Presacral mass can also be palpated by digital rectal examination. Pelvic and spinal radiographs and ultrasonography of pelvis further support the diagnosis by identifying sacral vertebral defects and presacral mass. The diagnosis is confirmed by CECT pelvis or MRI [10].

To decrease the diagnostic dilemma between surgical causes of constipation, the usual protocol for evaluation should always be followed. These are as follows:

a. Digital rectal examination must be done with caution during initial clinical workup to differentiate between Hirschsprung disease, anal stenosis, and any other pelvic pathology compressing rectum.
b. Plain abdominal radiograph along with spine radiograph should be done to see for fecal loading of colon as well as any vertebral defect particularly that of sacrum. Sacral defect in the form of agenesis or hemi-sacrum is an important cause of constipation in pediatric age group.
c. Ultrasound of pelvis should be done to rule out mass lesion in pelvis that may be causing compression of rectum.
d. In the presence of sacral defect or pelvic mass lesion, CECT pelvis or MRI pelvis should be obtained to know the exact location of mass, nature of mass, relationship with rectum as well as to know the nature of vertebral defect.

The treatment of Currarino syndrome is excision of presacral mass and correction of anorectal malformation through PSARP approach. However, if the mass is meningocele, the two surgeries should not be carried out simultaneously. Staged surgery should be done to decrease the risk of meningitis. In our case, we initially planned for excision of mass only. However, after excision of mass, we found that rectum has got mobilized. So, we performed anorectoplasty simultaneously. Fortunately, patient did not develop any signs of meningitis in post-operative period.

Conclusions

Following proper protocol for clinical evaluation of patient with constipation prevents diagnostic dilemma between surgical causes of constipation in pediatric age group. Digital rectal examination must be included in the protocol for evaluation of chronic constipation. In pediatric age group, clinical workup should be done with keeping in mind the rare diagnosis of Currarino syndrome along with common cause of constipation like Hirschsprung disease.

Abbreviations

3D: 3 Dimensional; CECT: Contrast-enhanced computed tomography; HD: Hirschsprung disease; MRI: Magnetic resonance imaging; PSARP: Posterior sagittal anorectoplasty

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Declarations

Authors' contributions

MMA- collection of patient record and intra-operative pictures and writing the manuscript; RJS, RR, and AK- proof reading of manuscript; AKS- editing of manuscript texts and pictures; BK- approval of manuscript and overall guidance. The authors read and approved the final manuscript.

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Consent for publication

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Competing interests

None

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