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Currarino syndrome associated with Hirschsprung's disease: Case report and literature review

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ABSTRACT

We present a rare family consisting of a 10-month-old boy with complete Currarino syndrome associated with Hirschsprung's disease. His mother had been diagnosed as having incomplete Currarino syndrome. Only 11 cases of Currarino syndrome associated with Hirschsprung's disease have been reported in the literature. Our report suggests that it is necessary to perform preoperative examinations to rule out Hirschsprung's disease, if the patient presents with a history of severe defecation disorders from early childhood.

Currarino syndrome (CS) was first reported by Currarino et al. [1] in 1981 as a disorder comprising three pathological conditions; anorectal malformation, a sacral defect and a presacral mass. CS is an autosomal dominant inherited disorder caused by a mutation in the *HLXB9* homeobox gene located on chromosome 7q36 [2]. We present a rare pediatric case of CS associated with Hirschsprung's disease (HD). Moreover, the patient's mother had also been diagnosed as having incomplete CS, suggesting that this may be a familial association. We also review the relevant literature.

1. Case report

A 10-month-old boy was referred to us complaining of alternating severe constipation and diarrhea. The boy was born at 37 weeks and 2 days' gestation weighing 3028 g and was transferred to the NICU due to transient tachypnea. Thereafter, from the age of 8 months, he developed alternating diarrhea and severe constipation and was treated by a pediatrician at a local hospital for suspected food allergy. Despite adequate treatment, his symptoms did not improve.

The patient was then referred to our department at 10 months of age. Importantly, his mother had been diagnosed as having incomplete CS with the sacral malformation and a presacral tumor during her pregnancy when she was 34 years old. She subsequently underwent excision of the tumor which, on histopathological examination proved to be an epidermoid cyst.

On admission, the infant's general condition was good with no developmental disorder. He was 71.3 cm long, weighing 9.2 kg. The abdomen was slightly distended but soft, and there was no mass palpable. On perineal inspection, the anal orifice appeared to be displaced anterior to the proctodeum, and examination of the anal site revealed that the anal orifice was stenotic, and did not admit a fifth finger (Fig. 1a). No abnormalities were noted on blood examination including any tumor markers.

A plain abdominal radiogram obtained on admission showed gaseous distension with massive accumulation of feces throughout the whole colon, with a huge fecal mass in the rectum (Fig. 1b). After bowel preparation, anal bougienage and bowel irrigation, the patient underwent various additional examinations. A barium enema revealed significant narrowing of the rectum, which connected to the considerably dilated sigmoid colon. The image was consistent with short segment aganglionosis (Fig. 2a). A pelvic 3D computed tomography revealed dysraphism of the fifth and

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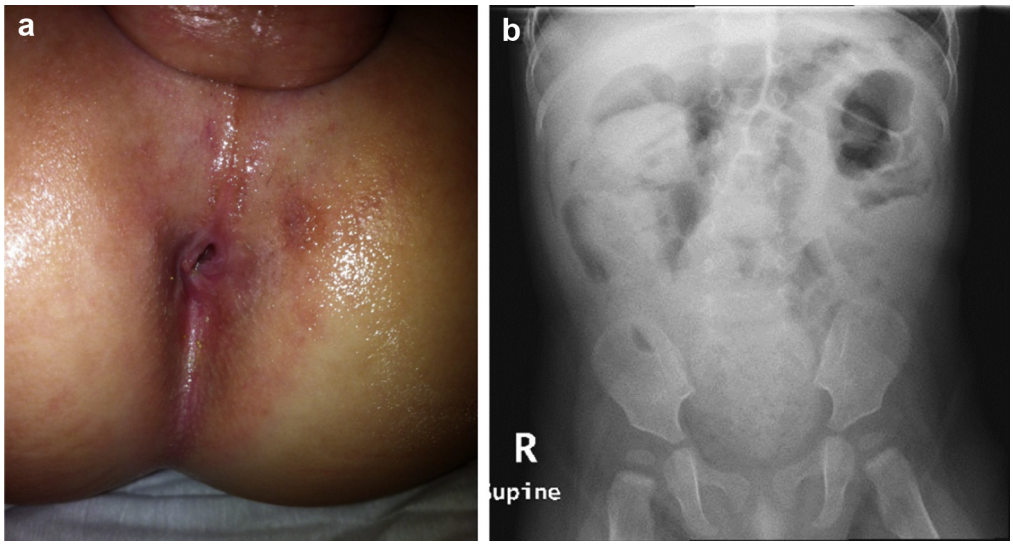

Fig. 1. (a) Appearance of the perineum. A stenotic anal orifice with a skin fold is located slightly anterior to the center of the perineum. (b) Abdominal radiograph shows multiple fecal masses in the dilated colonic loops, with massive accumulation of feces in the dilated rectum.

sixth lumbar spinous processes and sacral dysgenesis (the right hemi-sacrum) (Fig. 2b). Moreover, T2-weighted pelvic magnetic resonance imaging (MRI) depicted a solid and cystic mass (1.5 cm in diameter) in the presacral space (Fig. 2c), leading to the diagnosis of complete CS. For rule out the possibility of HD of the patient, we performed an anorectal manometry, which was positive, showing no relaxation reflex. In addition, rectal mucosal suction biopsy confirmed hyperplasia of acetylcholine-positive nerve fibers in the submucosal layer, leading to the diagnosis of a patient whose CS was complicated by HD (Fig. 3a). After the diagnosis, we decided to perform a single-stage surgery including presacral tumor resection, radical surgery for HD, and proctoplasty for the anal stenosis.

The patient was placed on the jackknife position initially, and he underwent presacral tumor excision using a posterior sagittal approach with a midline skin incision in the sacro-coccygeal region. After dissection of the subcutaneous tissue and muscle layers in the midline around the lower end of the sacrum, the presacral space was entered. The tumor was identified adjacent to the posterior rectal wall which was exposed just anterior to the sacrum. The tumor was dissected sharply, separating from the rough connective tissue on the anterior surface of the sacrum with ease, and was then completely removed from the rectal wall.

The patient's position was then changed to the lithotomy position after a temporary wound closure. Using a laparoscopic approach, approximately 10 cm of the dilated sigmoid colon near the recto-sigmoid junction was resected. The colon immediately proximal to the resection was prepared for a retro-rectal (Duhamel) pull-through. The surgical field was then transferred to the perineum.

Initially, the stenotic anus was opened widely using V and inverted Y-shaped cutaneous incisions. The ganglionated sigmoid colon was pulled down to the anus along the posterior rectal wall. The posterior half of the anal canal was incised circularly just above the dentate line, and the incised distal margin was sutured to the end of the sigmoid colon on the dentate line. Then, a side-to-side anastomosis was performed between posterior half of the rectum and anterior half of the pulled-through sigmoid colon using a stapler.

Histopathological examination of the surgical specimen revealed that the tumor tissue was composed of multiple cysts lined by squamous epithelium, and of a considerable amount of fatty tissue, identical to that found in an epidermoid cyst, and similar to his mother's tumor (Fig. 3b).

After surgery, he was given laxatives and antidiarrheal drugs for the potential transient bowel dysfunction associated with fecal accumulation in the residual rectal area, which is often observed after a Duhamel procedure. The patient was given additional laxatives 1–2 times a week, facilitating bowel movements. Thereafter, spontaneous bowel movements gradually developed. However, one year after surgery, the bowel movements increased to 3–4 times a

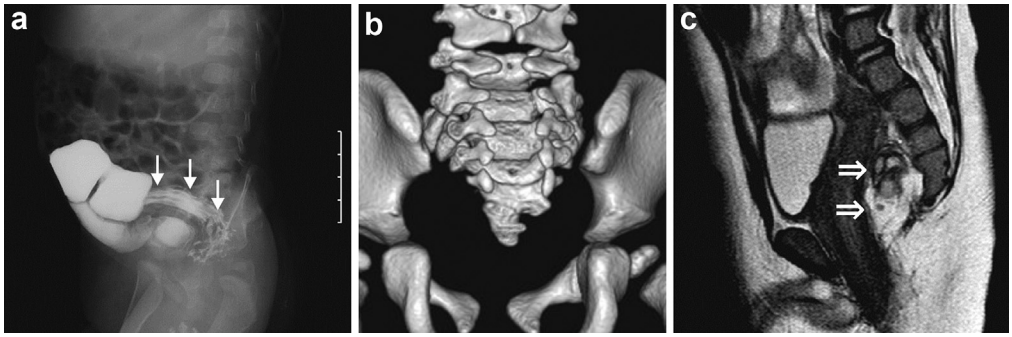

Fig. 2. (a) Barium enema shows the narrowed rectum (arrows) with significant dilatation of the sigmoid colon indicating short segment aganglionosis. (b) A pelvic 3D-CT revealed dysraphism of the fifth and the sixth lumbar spinous processes and the hemisacrum. (c) T2-weighted pelvic MRI (sagittal view) shows a solid and cystic mass (1.5 cm in diameter) in the pre-sacral space (arrows).