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Currarino Syndrome in an Adult Male

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Currarino syndrome (CS) is rare in clinical practice. We describe a 28-year-old man who presented with abdominal pain. He had a past operative history of colostomy for an anal problem as a newborn. Plain film of his abdomen disclosed intestinal obstruction and a sacral defect. Computed tomography revealed a presacral mass protruding from the bony defect of sacrum, which was compatible with later findings of magnetic resonance imaging. Total excision by a sacral laminectomy was accomplished smoothly. Pathological examination showed a mature cystic teratoma. The postoperative course was uneventful. We report on this adult patient to demonstrate this rare syndrome, which includes the classic triad of anorectal malformation, presacral mass, and bony defect of the sacrum. Chronic constipation should raise suspicion of this syndrome as a differential diagnosis.

Key words: Currarino syndrome, constipation, anorectal malformation, sacral defect, presacral mass

INTRODUCTION

Constipation is a common presenting complaint in young children and some adults. Currarino syndrome (CS) is a rare disease that can cause chronic constipation without being recognized. It was first described by Currarino et al. in 1981[1]. Since then, fewer than 250 cases have been documented worldwide[2]. The incidence is unknown. Most cases are diagnosed in children and predominantly in females[3]. The classic triad of CS includes anorectal malformation, a sacral bony defect, and a presacral mass. Herein we report a rare case of CS occurring in a man, and we review the relevant literature.

CASE REPORT

A 28-year-old man, married with one child, presented with progressive abdominal fullness. Personal bowel habit included one bowel movement per week since childhood. He denied fever, unusual body weight loss, or any history of bladder or sexual dysfunction. No significant cardiopulmonary, genitourinary, neurospinal, or family histories were noted except a notable history of temporary colostomy in 1976, at 7 days of age, because of an imperforate anus. Physical examination revealed a distended abdomen, diminished bowel sounds, and an old scar over the right upper quadrant of the abdomen. Digital rectal examination showed no palpable mass but stenosis. Neurological examination had no remarkable findings. A plain film of the abdomen disclosed a low intestinal obstruction (Fig. 1A), and a bony defect at the caudal part of the sacrum was also found, surprisingly. Laboratory tests of blood, urine, and stool were normal except for mildly elevated lactate dehydrogenase (298 U/L) and hypokalemia (2.7 mmol/L) concentrations. Colonoscopy showed no intraluminal lesions but stenosis of the sigmoid colon, suggesting external compression as the etiology.

Computed tomography (CT) of the pelvis (Figs 1C & 1D) showed agenesis of the sacrococcygeal vertebrae with a large presacral cystic lesion, about 10×12×9 cm in size, protruding from the ventral sacral defect. The mass caused severe compression of the adjacent sigmoid colon and subsequent edematous wall thickening and bowel obstruction. Magnetic resonance imaging (MRI) revealed a presacral mass with mainly cystic components of mostly homogeneous intensity on T2-weighted image (Fig. 1B). No thickened filum terminale or tethered cord was found.

After interdisciplinary evaluations, surgical excision was performed successfully using a sacral laminectomy without violating the colon or dura. The intraoperative view demonstrated an absent sacrum below the first level and no communication between the tumor and intrathecal space. The cystic tumor had variable wall thickness and contained pectinaceous material and yellowish fluid, and

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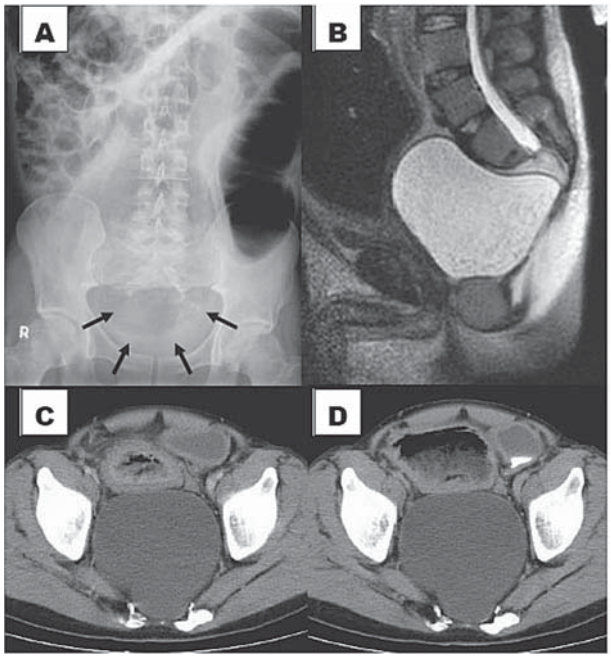

Fig. 1 (A) Plain film of the abdomen disclosed the absence of the caudal part of the sacrum and a pathognomonic "Scimitar sign" (arrows). Large bowel obstruction was also noted. (B) Sagittal T2-weighted MRI showed a hyperintense, homogeneous cystic tumor with a dotted scattered content protruding from the sacral defect and compressing the adjacent colon. Note the thickened, nodular part of the superior-posterior wall of the tumor and the connection to the caudal part of the thecal sac. No tethered cord was noted. (C) Nonenhanced and (D) contrast-enhanced CT of the pelvis showed a cystic tumor, without enhancement, arising from the sacral defect and compression of the sigmoid colon and urinary bladder.

adhered to the posterior wall of the sigmoid colon. Pathological studies showed a mass with a major cystic and a minor nodular component (Fig. 2). The cystic part contained multiloculated cysts with keratin and lined by squamous or cuboid cells. The nodular part included various pathologies of neural ganglia, brain tissue, blood vessels, smooth muscle, and fibrous tissue. These findings are compatible with a mature cystic teratoma.

The postoperative course was uneventful. Our patient was doing well at the 18-month follow-up and had no complaints. His bowel habit changed to 3 to 5 bowel movements per week without requiring stool softeners or laxatives.

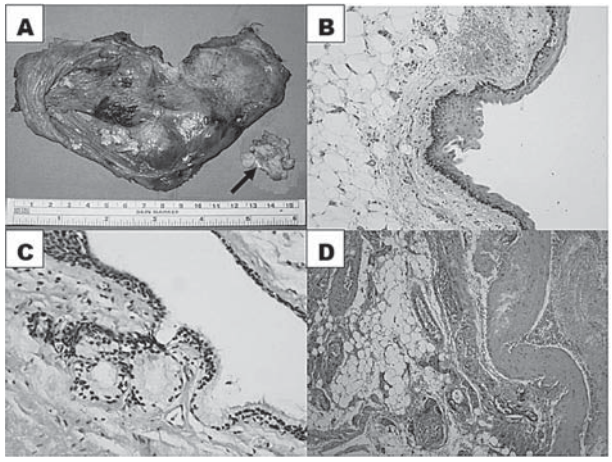

Fig. 2 (A) Gross appearance of the specimen showed a tumor with a cystic portion and nodular portion. The cystic mass contained much pectinaceous material (arrow), which was proven later to be keratin pathologically. (B) Tumor wall included keratinized, stratified squamous cells of epithelial tissue, indicating ectodermal origin. Hematoxylin and eosin (H & E) stain, original magnification 400×. (C) Some parts of the tumor showed a ciliated, cuboidal cell lining and appeared as endoderm in origin. H & E stain, original magnification 400×. (D) The nodular part included various pathologies of nerve tissue, blood vessels, smooth muscle, and fibrous tissue. H & E stain, original magnification 200×.

DISCUSSION

CS has three features: anorectal anomalies, presacral mass, and sacral dysgenesis. Previous studies show that presacral mass is a teratoma (18-40%), a meningocele (47-68%), or, rarely, an enteric cyst, lipoma, dermoid cyst, or any combination of these[3,4]. The anorectal malformations associated with CS are usually imperforate anus or anal stenosis[3,4]. A sacral defect is one possible caudal regression anomaly that may be associated with a number of neurological abnormalities. Its typical picture on radiography illustrates the absence of the caudal sacrum but preservation of the first sacral vertebra with a remnant of S2-S5, which causes a pathognomonic sickle-shaped bony defect.

It has been postulated that the pathogenesis of CS is caused by failure during the process of separation of the endodermal and ectodermal layers during embryogenesis[1]. Genetic studies show that CS is an autosomal dominant disorder with gene linkage to the 7q36 region where neurological anomaly of holoprosencephaly is also located[5]. Terminal deletion of the long arm of chromosome[7] and

mutations in the homeobox gene *HLXB9* have been reported[6,7]. More than 50% of cases are related to familial inheritance. Although CS has been accepted as an autosomal dominant disease, incomplete penetrance leads to phenotypically variable types. Martucciello et al.[8] categorized CS into three types–complete, mild, and minimal forms–according to the degree of involvement of the triad of abnormalities. Complete CS involves all three abnormalities. Mild CS comprises the hemisacrum and one of the other two criteria. Minimal CS involves only the hemisacrum without anorectal anomaly or presacral mass. Clinically, the complete type is usually diagnosed before the teenage years (80%)[3], whereas mild or minimal CS is diagnosed predominantly in adults. Our patient had anal atresia and had been operated on, but the CS was unrecognized because he was born before 1981, when Currarino reported this entity.

Chronic constipation after birth is the most frequent complaint (68-95%) in most patients with CS[1,3,4,9,10]. The cause of constipation in CS is still unknown. The proposed pathogenesis includes the compressive effect of the presacral mass and neurological anomalies associated with the tethered cord or anterior myelomeningocele[11]. Emans and colleagues[10] believe that constipation is not caused by a myelomeningocele. Other manifestations may include bowel obstruction, progressive pollakiuria, perianal sepsis, recurrent meningitis, and local neurological presentations. In patients presenting for neurosurgical consultation, CS is usually associated with some congenital anomalies such as a tethered cord (18%), anterior myelomeningocele, type 1 Chiari malformation, or a fistula between the colon and spinal canal, which may cause frequent central nervous system infection[3,9,10,12]. The fistula-related infectious problems such as meningitis are very rare complications of CS[3,12].

We speculate on the natural history of CS as follows. The patient is usually found with imperforate anus after birth and needs surgical diversion of stools. Anal stenosis is otherwise hard to discover unless it causes young children to cry in discomfort, especially during defecation. As the patient matures, chronic constipation is not usually a reason for a clinical visit and, even if he or she visits the physician, CS is difficult to recognize. Clinically, CS patients are usually diagnosed because of unrelieved abdominal pain, which leads to subsequent plain film and identification of the defective sacrum. Further studies using CT or MRI show the presacral mass and, finally, the CS triad is matched to symptoms. Specialist physicians, such as pediatricians, gastroenterologists, and neurosurgeons, should be aware of CS as a differential diagnosis for a newborn with anorectal anomalies, a child with severe constipation after birth, or a young adult with a chronic history of constipation.

Diagnosis is easily approached by plain film, which discloses the various bony defects of sacrum. The plain film has a typical finding of a "Scimitar sign" of the sacrum shape. Further radiological study with MRI can delineate the details of tumors and associated neurospinal abnormalities[10]. The relationship between the presacral mass, the thecal sac, and possible filum terminale of a tethered cord should be studied in detail for surgical planning to untether the cord or avoid infection. For patients with associated anterior myelomeningocele or a tethered cord, MRI investigation should include the head and entire spine to rule out possible intramedullary syrinx formation or Chiari malformation. With the increasing availability of MRI for other problems, asymptomatic CS may be detected on occasion.

The decision about management should involve interdisciplinary consultation and contributions from various subspecialties. Absence of part of the sacral bone alone rarely needs treatment. Anorectal malformations can produce gastrointestinal symptoms after birth, and surgical correction is mandatory to relieve these symptoms. Our patient had significant anal atresia immediately after birth and received loop colostomy for stool diversion as a newborn. The presacral mass raised concerns whether it was benign or malignant. Total excision to obtain pathological evidence is a reasonable surgical indication. The mass may compress the surrounding organs and may need resection for decompression. A persistent fistula between the colon and intraspinal space that causes meningitis should be operated on as soon as possible. In contrast, an anterior myelomeningocele or tethered cord associated with CS should not be treated during infancy because 60% of patients develop iatrogenic injury[10].

Several surgical options have been studied. The surgical approach through the sacral laminectomy is the procedure most preferred[13]. In our patient, anorectal anomalies had already been corrected as a newborn, and the surgical goal was to remove the presacral mass and dissect the adhesions between the tumor and thecal sac. From the neurosurgical viewpoint, one does not wish to risk violating the relatively contaminated anal region. The posterior sagittal approach by dividing the striated sphincter complex has been described recently as the option of choice because it provides good exposure for safe repair of the dura and excision of the teratoma without injury to the rectum[4,14]. The transabdominal approach is a poor choice because it is prone to causing leakage of cerebrospinal fluid and bowel injury[13].

In summary, adult patients with CS have a history of

chronic constipation since birth. Physicians usually first approach the diagnosis after finding a sacral bony defect on plain film. Further MRI study can confirm the nature of the mass and help guide treatment. The surgical corridor is usually through the posterior sacral region. A multidisciplinary approach is needed for the best outcome. It is a rare disease, but one that pediatricians, gastroenterologists, and neurosurgeons should be aware of.

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