

Currarino triad associated with malrotation of the colon

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ABSTRACT

Currarino triad is a rare congenital condition characterized by a sacral bony defect, presacral mass, and anorectal malformations. We describe an unusual case of complete Currarino triad in a 22-year-old female with sacral bony defect, anal stenosis, recto-vaginal fistula, and dual pathology (meningocele and teratoma) in the pre sacral mass associated with malrotation of the colon. This combination has not been reported previously. The clinical presentation, surgical management and review of literature are discussed.

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Currarino triad is a rare hereditary condition that was reported by Currarino et al.¹ It is characterized by a complex of congenital caudal anomalies: sacral bony defect, presacral mass, and anorectal malformations. A variety of other associated congenital anomalies have been described.²⁻⁵ The triad is a result of mutations in the homeobox HLXB9 gene on chromosome 7, and it is inherited as an autosomal dominant trait.⁶⁻⁸ More than half of the cases reported in the literature have a positive family history for at least one of the components of the Currarino triad.^{2,3,9} In more than 80% of cases, this triad is diagnosed in the first decade.^{3,10} Chronic constipation since early life is the most common symptom, but patients may present with recurrent urinary tract infections, headache, low back pain, meningitis, and perianal infections.^{2,11-13} We present this unusual case of Currarino triad aiming to increase the index of suspicion for this rare condition.

The presence of any of the abnormalities of the triad necessitates appropriate investigations to exclude or to confirm the diagnosis.

Case Report. A 22-year-old single female patient presented with a history of constipation, and passage of stools per vagina since early childhood. She used to pass stool with difficulty every 1-2 weeks. She was admitted to other hospitals several times with attacks of right sided lower abdominal pain, which was usually relieved by the passage of stool. She also complained of recurrent attacks of low back pain, and headache since early childhood, which increased with constipation and straining and relieved by passing stool. She was a product of uneventful pregnancy and normal delivery. There was no family history of a similar condition. Physical examination revealed a healthy and normally developed female. The abdomen was slightly distended and tender in the right iliac fosse. Perineal examination revealed normally located but stenosed anus admitting only the little finger, feces in the vagina, and a wide recto-vaginal fistula which could admit the little finger. Neurological examination was normal.

The radiograph of the pelvis (Figure 1) showed scimitar sacral bony defect and spina bifida sacral segment 1. Abdominal and pelvic ultrasound scan was reported as normal. Spinal magnetic resonance imaging (MRI) scan showed an anterior sacral meningocele (ASM), and 2 soft tissue masses located anterior and caudal to the ASM, which displaces the dilated rectum ventrally (Figure 2). Intravenous urography showed incomplete duplex and slight malrotation of both pelvicalyceal systems (Figure 1). Barium enema revealed dilated rectum and sigmoid colon, and malrotation (sigmoid and descending colon in the right side and cecum and ascending colon to the left side of them in the center of the abdomen) of the colon (Figure 3). The MRI scan of the brain and chest radiograph were normal. She was managed first by a diverting loop ileostomy. Colostomy was not carried out as the sigmoid colon was dilated and fixed in the right iliac fossa without mesentery. After mechanical preparation of the colon and rectum, exploration through a posterior sagittal approach was performed. The neurosurgeon excised the ASM after



Figure 1 - Intravenous urography showing sacral bony defect at the head of the arrow, spina bifida at S1, and incomplete duplex, fullness, and slight malrotation of both pelvicalyceal systems.

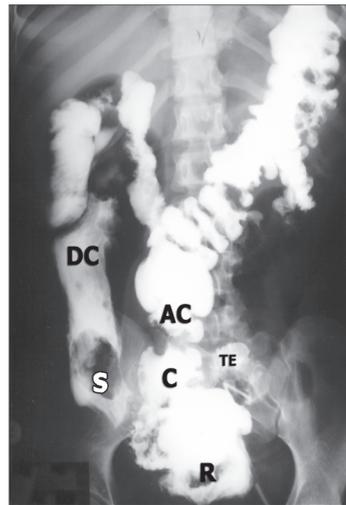


Figure 3 - Barium enema showing colon malrotation (R=rectum, S=sigmoid colon, C=cecum, AC=ascending colon, TE=terminal ileum, DC=descending colon).

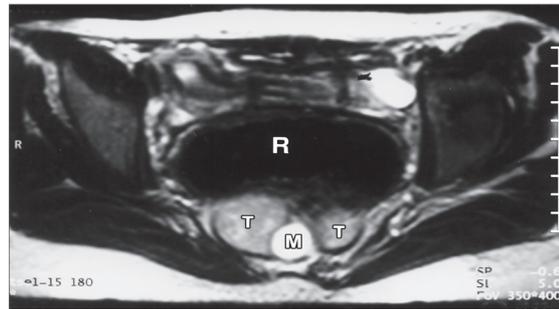
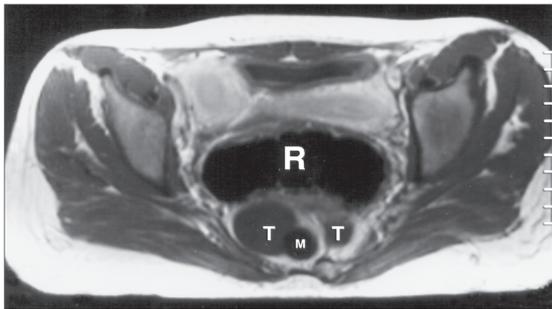


Figure 2 - Spiral MRI: a) T1 MRI of the lumbo-sacral spine showing: M= anterior sacral meningocele (ASM), T=2 teratoma, R= rectum and b) T2 MRI of the lumbo-sacral spine showing: M=ASM, T=2 teratoma, R= rectum.



Figure 4 - Macroscopic pathology specimen of the 3 excised teratomas.

identifying and ligating its neck. The histopathology confirmed the diagnosis of meningocele. Three presacral masses attached together and firmly adherent to the rectum were found. They were dissected and excised completely without breaching the rectum (Figure 4). There were no communications to the rectum or the ASM. The histopathology of the masses was mature cystic teratoma. She had an uneventful smooth postoperative course without any neurological deficit. Two months later anoplasty and repair of the recto-vaginal fistula was performed. The fistula was repaired using a mucosal advancement flap. The loop ileostomy was closed 6 months later after insuring the healing of the recto-vaginal fistula. She had an uneventful post operative period. On a 4 years follow-up she is free of symptoms and has normal bowel habits. The recto-vaginal fistula has healed completely.

Discussion. Currarino triad is a rare hereditary condition with less than 250 patients reported up to the year 2005.¹⁴ The triad is characterized by a sacral bony defect, presacral mass, and anorectal malformations. The sacral bony anomaly which is present in almost all cases varies from spina bifida to hypoplasia of lateral bodies of the distal 3 sacral vertebrae (sickle or scimitar shaped deformities, sacral agenesis, spina bifida, coccygeal defects).^{2,14} The anorectal malformation in most of the reported cases has been either anal stenosis or imperforated anus, but other malformations (recto-vaginal and recto-urethral fistula) have been reported.^{2,3} The presacral mass is more frequently an ASM 60%, teratoma in 25%, and other tumors (dermoid or enteric cyst, lipoma, hamartoma, neurofibromatosis, and a combination of these) in 15%.² Associated congenital anomalies included duplication of vagina and uterus, partial duplication of urinary tract, horseshoe kidney, vesicoureteral reflux, and Hirschsprung disease.²⁻⁵ Our patient has a scimitar sacral bony defect, anal stenosis with recto-vaginal fistula, dual presacral pathologies (ASM and 3 cystic teratomas), and associated malrotation of the colon, which has not been reported before. More than 80% of the cases are diagnosed in the first decade of life.^{3,10} Chronic constipation since early life and sacral bony defect were present in almost all of the patients.^{2,13} Constipation is most probably as a result of anal stenosis, extrinsic pressure from the presacral mass or neurologic abnormality.^{1,2,8,14} Other presentations included urinary tract infection, headache, back pain, meningitis, perianal abscesses, and anorectal fistula.^{2,11-13} The diagnosis in our patient was delayed, and not thought of despite being admitted several times in other hospitals. None of the patients reported by Martucciello et al¹⁵ had an early diagnosis. This indicates that Currarino triad

is frequently not recognized and misdiagnosed. This also suggests that its incidence could be more frequent than currently estimated. Staged operations have been advised by most authors, when the presacral mass is an ASM.^{3-5,10,13} Accordingly, our patient was treated initially with a diverting ileostomy. This was followed by excision of the ASM and the presacral teratomas through the posterior sagittal approach, which has been recommended by most authors.^{3,4,7,13} Six weeks later an anoplasty and repair of the recto-vaginal fistula with mucosal advancement flap was performed. Finally the ileostomy was closed after insuring the healing of the recto-vaginal fistula. No complications were encountered, all symptoms disappeared, and the fistula has not recurred. In a review of 29 cases by Cretolle et al,⁷ 2 patients developed transient cerebrospinal fluid leak, 3 urinary incontinence, one neurological bladder, and one fecal and urinary incontinence. One of the 3 patients reported by Currarino¹ died of meningitis.

In summary, Currarino triad is a rare congenital condition. The triad is frequently missed, and its diagnosis is delayed. The diagnosis of Currarino triad should be suspected in patients with persistent constipation since early life, ano-rectal malformations and presacral mass on rectal examination. A search for associated congenital anomalies is recommended after the diagnosis is confirmed followed by staged surgical approach to alleviate patient's symptoms and to guard against future morbidity and mortality.

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