Currarino Triad—Diagnostic Dilemma and a Combined Surgical Approach

By M. Samuel, G. Hosie, and K. Holmes
London, England

Purpose: The authors present 2 families with 3 cases of Currarino triad, diagnostic difficulties, their familial occurrence, and genetic mapping, with emphasis on a combined pediatric surgical and pediatric neurosurgical approach in managing these children.

Results: The main presentation was intractable constipation. In the first family there was a 4-generation pedigree with recurrence of Currarino triad. The mother and the child have the condition. Family 2 screening showed a 3-generation pedigree with presence of Currarino triad in 3 members. Patients 2 and 3 are cousins whose fathers are affected by spina bifida occulta and Currarino triad, respectively. In patient 1, the diagnosis was made after inadvertent rupture of an anterior meningocele during posterior myectomy. In patient 2, the presacral mass was found on examination under anesthesia, and the planned anorectal myectomy for intractable constipation was abandoned. Patient 3 was a cousin of patient 2, and the diagnosis was considered when she presented with intractable constipation at the age of 7 months. Magnetic resonance scan was useful in showing the presence of presacral mass, spinal abnormalities, and tethered cord. A combined pediatric and neurosurgical approach optimized the extirpation of the presacral mass with minimal complications. Surgical treatment was individualized according to the estimation of the operative risk factors. All patients have a normal bladder function. Patient 1 has required laxatives and enemas for intermittent constipation. She has associated learning difficulties but is otherwise well. Patient 2 and 3, aged 10 and 2 years, respectively, are awaiting closure of colostomy. They are thriving and well.

Conclusions: The authors recommend a combined pediatric and neurosurgical assessment and management for all cases of Currarino triad. Family screening is obligatory. The authors suggest the use of a magnetic resonance scan or computerized axial tomography myelogram to define the presence of anosacral and spinal cord anomalies in patients with intractable constipation.

INDEX WORDS: Currarino triad, constipation, anal stenosis, presacral mass, familial occurrence.

THE TRIAD of an anorectal malformation, a sacrococcygeal bony defect, and a presacral mass was first described by Kennedy in 1926.[1] Currarino et al[2] recognized these disorders as a syndrome, and postulated the embryogenesis of this triad in 1981.[2] The Currarino syndrome belongs to the group of persistent neurenteric malformations and is associated with chronic constipation, a bony sacral defect, a presacral mass, and anorectal stenosis or agenesis. The typical radiologic signs of the Currarino triad are represented by an hemisacrum with preserved first sacral vertebra. The presacral mass generally is a presacral teratoma or an anterior meningocele. The anorectal malformation either is anal stenosis or agenesis.

The association is autosomal dominant with incomplete penetrance and variable expressivity. The 3 anomalies characterizing this syndrome can be ascribed to a common developmental defect or anomaly of the notochord in the early phases of embryogenesis. More recently, it has been proposed that the malformations of Currarino triad arise through a failure of dorsoventral separation of the caudal eminence from the hindgut endoderm during late gastrulation.[3] We present 2 families with 3 cases of Currarino triad, their familial occurrence, and genetic mapping. We emphasize the diagnostic difficulties and recommend a combined pediatric surgical and neurosurgical approach in managing these children.

From the Department of Paediatric Surgery, St George's Hospital, London, England.

Address reprint requests to M. Samuel, Department of Paediatric Surgery, 5th Floor, Lanesborough Wing, St George's Hospital, Blackshaw Rd, London SW17 OQT, England.

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CASE REPORTS

Family 1, Patient 1

A white baby girl was born by lower segment cesarean section for breech presentation at 38 weeks' gestation with a birth weight of 2,520 g to a 28-year-old-multigravida with Currarino triad (anal stenosis, partial sacral agenesis, and anterior meningocele). Antenatal ultrasound scans were normal. She passed meconium in the first 24 hours of life and had normal bowel function up to the age of 10 weeks. She was referred at 13 weeks of age for poor weight gain (3,970 g, less than third percentile), abdominal distension, and constipation with overflow incontinence. Abdominal examination found a hard large fecal mass. Anorectal examination under general anesthesia found a normally positioned anus with associated anal stenosis, an irregular natal cleft, and a short sacrum. Sacral dimples were present. Manual evacuation was unsuccessful because the anus could be dilated only up to Hegar 13. Rectal biopsy findings were normal. An anteroposterior radiograph

of the pelvis showed a short sacrum and an ultrasound scan was normal. She had difficulty passing feces despite the use of stool softeners, laxatives, and enemas. She was examined 4 times under anesthetic, the results of which showed anal stenosis needing dilatation, a posterior rectal mass that was not obvious on palpation, and 2 ultrasound scans showed no evidence of a mass or collection between the rectum and the spine. She had grade 1 vesicoureteric reflux with normal upper tracts and kidneys. At 10 months of age a posterior myectomy was performed. During the procedure a small presacral mass was identified with a cerebrospinal fluid leak confirming an anterior meningocele. The pediatric neurosurgeons closed the meningocele through a posterior sagittal approach. A sigmoid loop colostomy and an anoplasty were performed. During the formation of the colostomy a Meckel's diverticulum was found that was excised followed by ileoileal anastomosis. Magnetic resonance (MR) scan confirmed the presence of a repaired anterior meningocele involving the sacral segment S3 to S5. The filum terminale was thickened, and the spinal cord terminated at the level of L2. The colostomy was closed at 2 years of age. In the past 10 years, the child has been treated with laxatives and enemas for intermittent constipation. She is thriving but has learning difficulties. Her vesicoureteric reflux has resolved, and she has a normal bladder as assessed by urodynamics.

Family screening showed a 4-generation pedigree with recurrence of the Currarino triad in which 4 affected members show hemisacrum as the only feature of the disease. The mother of this child has Currarino triad consisting of anal stenosis, partial sacral agenesis and an anterior meningocele, and learning difficulties. The elder sister of the patient has hemisacrum, constipation, and learning difficulties. The patient also has a younger sister who has spina bifida and constipation, and a brother with trisomy 21. The Currarino gene 7q36 is present in the mother and the child.

Family 2, Patient 2

An 8-month old white girl presented with a history of intractable constipation since the age of 6 months with associated overflow incontinence. On examination she had a distended abdomen with a large fecaloma. She had passed meconium in the first day of life after a normal vaginal delivery. Antenatal ultrasound scans had been normal, and the 29-year-old mother reported a regular once daily bowel action up to the age of 6 months. An abdominal x-ray showed gross fecal loading, and an ultrasound scan was normal. Manual evacuation, anal dilatation and a rectal biopsy were performed. She had a normal perineum and anus, and sacral dimples were present. Rectal biopsy was normal.

Constipation persisted despite all medical measures, and the child needed a further manual evacuation at 2 years of age. At the age of 4 years an examination under anesthesia showed a posterior anorectal pit 3 cm from the dentate line, and a soft fluctuant mass was palpable just above the pit between the posterior rectal wall and the spine. An MR scan showed a lipoma at the lower end of the spinal canal, a low-lying cord with a dilated central canal. There was absence of multiple sacral segments, with a 2- $\times$ 2.5-cm anterior sacral meningocele and tethering of the spinal cord.

In association with the pediatric neurosurgeons, a posterior sagittal midline repair of the anterior myelomeningocele was performed. The lipoma was excised, and the cord was untethered. A midsigmoid end colostomy was fashioned. The child had grade I vesicoureteric reflux with normal upper tracts and kidneys. At 10 years of age her vesicoureteric reflux has resolved, and she has a normal compliant bladder on urodynamic study. An endoanal ultrasound scan showed an internal anal sphincter, which was disrupted in several places, and a normal external anal sphincter. Anorectal physiology findings showed low resting pressures at all levels and normal anal sensation. The squeeze pressure was normal in the upper anal canal and was low at all other levels. Rectal volume sensation was normal at threshold and was impaired at constant and maximum tolerated volume. Rectoanal inhibitory reflex was absent. She has flat feet and an unusual gait but no spasticity. Colostomy closure was delayed at the family's request in view of the traumas associated with constipation in early childhood.

Family 2, Patient 3

A 7-month old white girl presented with progressive abdominal distension and severe constipation associated with difficulty in defecation. She had passed meconium on the first day of life after a normal term vaginal delivery. Antenatal ultrasound scans were normal. The 32-year-old mother had observed that the infant suffered from pain during defecation, decreased stool frequency (from once daily to once a week to once a month), and worsening of symptoms despite taking stool softeners and laxatives since cessation of breast feedings. Her first cousin (patient 2) had previously been treated for Currarino triad at our institution. On examination she had a distended tender abdomen with a large fecal mass palpable per abdomen. Anorectal examination showed a normally positioned anus with associated anal stenosis and an irregular natal cleft. Sacral dimples were present. An abdominal x-ray showed severe fecal loading and a typical anterior crescent-shaped sacral bony defect with an intact first sacral vertebra (Fig 1). Ultrasound scan of the spine showed an anterior heterogeneous mass. The renal system and bladder were normal. An upper gastrointestinal contrast study found malrotation and midgut volvulus. At operation, a 360° midgut volvulus was present in association with malrotation. There was a large fecaloma in the sigmoid colon. A Ladd's procedure after derotation of the volvulus and an anterior rectal enterotomy for evacuation of the fecaloma was performed. An MR scan of the pelvis and spine showed a saber-shaped sacrum with an anterior heterogeneous mass measuring 4 $\times$ 3.5 $\times$ 3.5 cm. The mass was found to extend into the filum terminale, into a low tethered cord (Fig 2). A computerized axial tomography myelogram confirmed a heterogeneous mass extending into and distorting the distal spinal canal with associated cord tethering (Fig 3).

In association with the pediatric neurosurgeons, a posterior sagittal midline excision of the tumor was performed. The right levator muscle floor was deficient, and the tumor was adherent to the anterior aspect of the sacrum and walls of the pelvic floor. The tumor was dissected free of the pelvic floor to be left attached to the spinal cord by a stalk (Fig 4). A partial laminectomy of the first and second sacral vertebrae was performed, the theca was opened, and the tumor was dissected free of the distal spinal cord and sacral roots. Histopathology showed a benign teratoma. The child is awaiting closure of colostomy. She has grade 1 vesicoureteric reflux, normal upper renal tracts and kidneys, and a

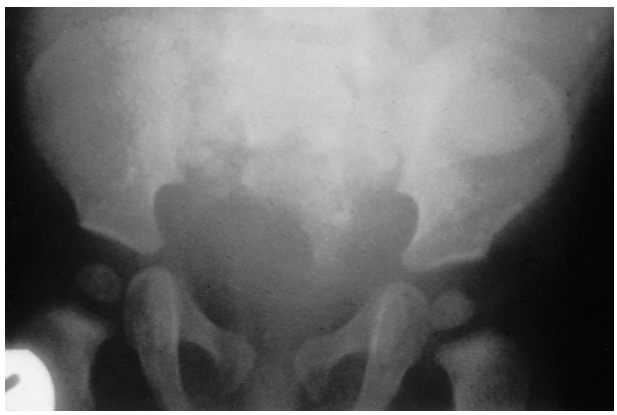

Fig 1. X-ray of the pelvis shows a typical anterior crescent-shaped sacral bony defect with an intact first sacral vertebra.

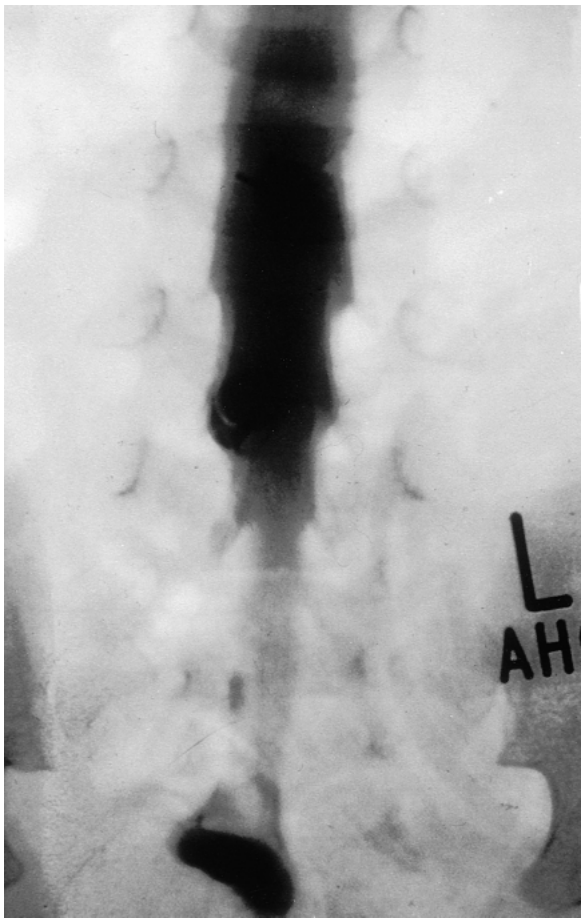

Fig 2. Magnetic resonance imaging shows a high filum terminal and tethered cord.

normal compliant bladder on urodynamics. At 2-year follow-up she is thriving.

Family screening found a 3-generation pedigree with presence of Currarino triad in 3 members. Patients 2 and 3 are cousins and both have Currarino triad. The fathers of these 2 patients are brothers and both suffer from constipation. The father of patient 2 has spina bifida occulta, and the father of patient 3 has Currarino triad (hemisacrum, anal stenosis, and presacral tumor). Molecular genetic testing results showed the presence of the Currarino gene 7q36 in the patient and her cousin (Fig 5).

DISCUSSION

The incidence of myelomeningocele is roughly 1 in 1,000 live births. Anterior meningocele is uncommon. Sacrococcygeal teratoma occurs in 1 in 40,000 live births with a female preponderance,[4] and the incidence of anorectal malformation varies from 1 in 2,000 to 1 in 5,000 live births with an even gender distribution for low anorectal anomalies.[5] Occult myelodysplasia with associated sacral deformities occurs in 20% to 50% of the patients with anorectal malformation, being common in high (48% to 54%) and less common in low anorectal malformation (15% to 27%).[6,7] The concurrence of a presacral mass, most often a teratoma or ventral meningocele, an anorectal anomaly, and an anterior sacral bony defect (scimitar sacrum) should be extremely rare. However, in the past 35 years (1963 to 1998) 230 cases of complete or incomplete Currarino syndrome have been reported.[1-3,8-29] Eighty-one percent of the patients were less than 12 years of age at diagnosis with a wide range of 1 day to 89 years. Girls are more commonly affected as seen in our 2 families, and 60% of the reported cases have a family history of triad-associated anomalies. Antenatal diagnosis of the Currarino triad has been reported in 3 cases, in which an obstetric ultrasound has been able to show spinal abnormalities and associated presacral mass.[30] In our cohort, the antenatal scans were normal. Chronic constipation from early life was the common symptom in all patients, and 60% presented with anorectal stenosis.[1-3,9-29]

Variable sacral bony defects were detected (100%), and the most frequently reported presacral masses were

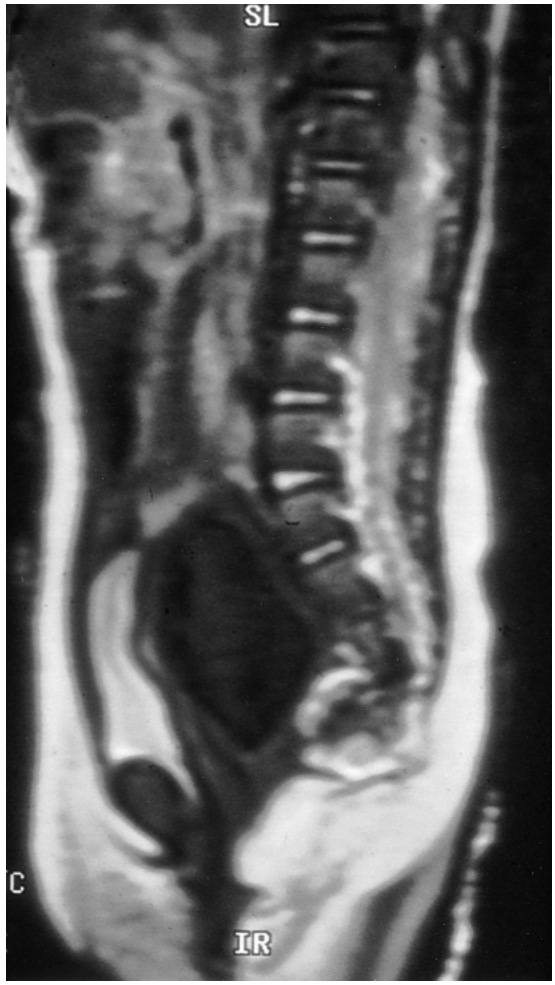

Fig 3. Computerized axial tomography myelogram in the sagittal plane shows a heterogeneous mass extending into and distorting the distal spinal canal with associated cord tethering.

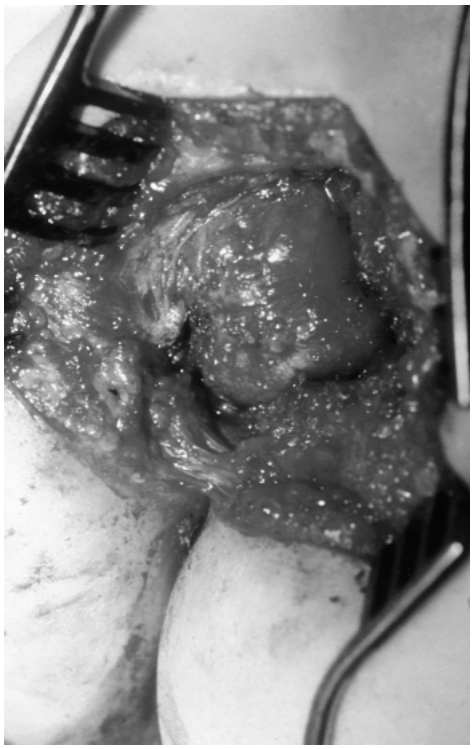

Fig 4. Tumor has been dissected free of the pelvic floor to be left attached to the spinal cord by a stalk.

ventral meningocele (60%), benign teratoma (23%), dermoid cyst (6%), unclassified tumors (4%), lipoma (2%), neurofibromatosis (1%), enteric cyst (1%), and hamartoma (1%). Malignant teratoma was found in 2 patients (1%)[15,18] and a leiomyosarcoma in 1 patient.[26] The other associated abnormalities are tethered cord with a fixed filum terminale and urogenital abnormalities such as rectovaginal fistula and partial duplication of the vagina, uterus, or the urinary tract. Constipation is a pathognomonic feature of Currarino triad, and the dangerous complication is an anal or rectal fistula, which communicates with the spinal cord predisposing the patient to the risk of meningitis. Prenatal diagnosis may aid in the early recognition and surgical management of persistent spinal-rectal fistulas and may help avoid life-threatening bacterial meningitis.[30]

A gene responsible for the Currarino syndrome recently has been mapped at the level of the terminal portion of the long arm of the chromosome 7 (7q36).[8] In our families there is a 2- and 3-generation pedigree with recurrence of the condition, in which 5 affected members all have features of the Currarino triad. Moreover, presacral teratomas are autosomal dominantly inherited, and anterior sacral meningoceles are described as an X-linked dominant condition. Therefore, an anterior meningocele with an associated anterior sacral defect could be an autosomal inherited condition. A single gene mapping in 7q, presenting incomplete penetrance and variable expressivity could cause anorectal malformation in the Currarino syndrome or anorectal anomaly associated with sacral deformities and presacral mass. The first family has members with learning difficulties. The Currarino triad has been shown to be genetically very close to the holoprosencephaly locus on chromosome 7q. There is a strong possibility that there is holoprosencephaly within this family.

To optimize the diagnosis of the Currarino syndrome, a rectal examination is mandatory in all cases of persistent severe constipation. Stenosis is located distally in these patients, and a presacral mass often is palpable. A pelvic x-ray and ultrasound scan of the sacrum is recommended for the detection of the anterior sacral bony defect and presacral mass. However, as shown by our case reports, the diagnosis may be difficult. The presacral mass may not be palpable even under an anesthetic, and an abdominal x-ray or an ultrasound scan may not be helpful. Hence, we recommend a MR scan or computerised axial tomography myelogram to specify the presence of anosacral and spinal cord anomalies in patients with intractable constipation. Investigation of the bladder and the renal system is essential in all cases of Currarino triad to establish or rule out vesicoureteric reflux, which has a familial occurrence, especially when the girls are affected. We recommend routine urodynamics in these

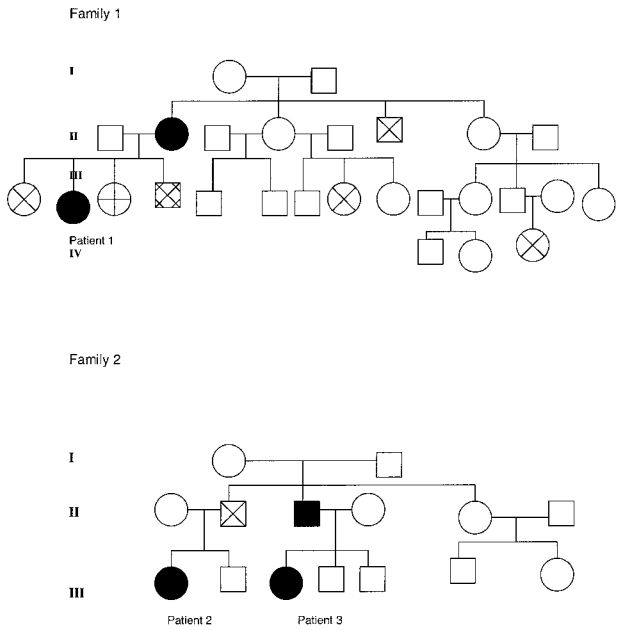

Fig 5. Our families had a 3- and 4-generation pedigree with recurrence of the Currarino triad, in which 5 affected members have all features of the Currarino triad. ○/□ indicate healthy female and male; ●/■ indicate complete Currarino triad; ⊗/⊠ indicate hemisacrum, constipation; ⊕ indicates lumbar spina bifida, constipation; and ⊠ indicates trisomy 21.

patients to document the absence or presence of a neuropathic bladder. A combined pediatric and neurosurgical approach would optimize the extirpation of the presacral mass with minimal complications. A posterior sagittal midline incision provides good exposure and ease of access to the presacral mass. Surgical treatment should be individualized according to the estimation of the operative risk factors. A presacral fistula must be excised immediately to prevent the risk of meningitis. Presacral masses should be evaluated with the neurosurgeons to minimize nerve damage and its sequele of neurogenic bladder dysfunction and stool incontinence. Neurosurgical exploration should precede intestinal surgery, and excision of the coccyx in cases of teratomas is advised because of the reported risk of malignancy. Patients with presacral teratomas must have follow-up ultrasound scans and alpha-fetoprotein estimations. The postoperative continence of these patients does not differ from that of a patient with solitary anorectal malformation. Family screening is obligatory in all cases of Currarino triad.

We recommend a combined pediatric and neurosurgical assessment and management for all cases of Currarino triad. Family screening is obligatory. We suggest the use of an MR scan or computerized axial tomography myelogram to specify the presence of an anosacral and spinal cord anomalies in patients with intractable constipation.

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