Currarino syndrome

Currarino's syndrome (CS) is an autosomal dominant disorder of embryonic development causing a rare malformating syndrome characterized by a triad of an anorectal malformations, presacral mass (most commonly an anterior sacral meningocele) and sacrum defect.

Tumors like teratoma, hamartoma, epidermoid cysts of the central nervous system have also been associated.

It was originally described by Guido Currarino in 1981 [1].

Till 2013 approximately 300 cases have been reported in the literature [2].

Approximately 60% of the patients with Currarino syndrome reported in the literature have an occult spinal dysraphism. Published studies suggest that the risk of tethered cord syndrome may be higher among patients with a lipoma and lower among those with a teratoma or anterior meningocele [3].

Only four cases of carcinoid transformation of the presacral mass have been described in the literature [4].

Etiology

This syndrome is thought to result from abnormal separation of the neuroectoderm from the endoderm, caused by HLXB9 mutation in chromosome 7q36 in 50% of cases. The disorder is mostly hereditary as it can also be sporadic with a variable expression spectrum.

A precise genotype-phenotype correlation has not been described so far. Family screening is obligatory.

Clinical features

Although it is rarity, the Currarino syndrome might be one of the causes of chronic constipation [5].

Diagnosis

The diagnosis is usually made during childhood and rarely in adulthood. In this context, imaging, and especially MRI plays a major role in the diagnosis of this syndrome.

When an anterior sacral meningocele is encountered, Currarino syndrome should be taken into consideration [6].

A geneticist evaluation must be offered to the patient in the case of a CS as well as a clinical

evaluation of the relatives (parents, siblings) [7].

Treatment

see Currarino syndrome treatment.

Case series

Between 2008 and 2015, 10 patients presented with CS, 9 with constipation. Median age at the time of surgery was 1.3 years. Six of the 10 patients had anorectal malformation consisting of anal stenosis, rectal stenosis, or imperforate anus. Spinal anomalies included anterior meningocele (5 cases), low-lying conus (8), terminal syrinx (4), fatty filum (5), caudal lipoma (3), and intraspinal cyst (1). In all cases, the lumbosacral spinal canal was accessed via a midline approach with laminoplasty, allowing spinal cord untethering and repair of the dysraphic anomalies. Following dural closure, the incision was extended inferiorly to incorporate a posterior sagittal approach to resect the presacral mass. The histopathological nature of the mass was mature teratoma (8 cases), complex hamartomatous malformation (1), or neurenteric cyst (1). There were no new instances of neurological deterioration, with most instances of persisting morbidity related to constipation (6 cases) or neurogenic bladder dysfunction (8). There were no infective complications, no instances of cerebrospinal fluid fistula, no recurrences of the presacral mass, and no cases of retethering of the spinal cord.

Although not part of the original triad, spinal dysraphic anomalies are common in CS and in keeping with a disorder of secondary neurulation. Lumbosacral MRI is an essential investigation when CS is suspected. Children are at risk of sphincter impairment due to the anorectal malformation; however, both spinal cord tethering and compression from the presacral mass may further compromise long-term continence. A combined operative approach to repair the dysraphic anomalies and resect the presacral mass is described with good postoperative and long-term outcomes [8].

A family case report with CS requiring different neurosurgical management. The son, a 3-year-old boy, developed a tethered cord syndrome associated to a lipoma of the filum terminale, a sacro-coccygeal teratoma and an anal adhesion. A combined surgical approach permitted a good evolution on the urinary and digestive functions despite a persistent fecal incontinence. The 2-year-old daughter presented with a cyst of the thyroglossal duct infected and fistulized to the skin. She was also followed for a very small lipoma of the filum terminale that required a neurosurgical approach. The father, 44-year-old, manifested functional digestive and urinary disorders caused by a giant anterior sacral meningocele [9].

A study included 17 patients (13 girls and 4 boys). Their age at presentation ranged from 7 months to 10years. They used posterior sagittal approach to correct anorectal anomalies, and excise presacral cysts that were subjected to histopathological examination. Two cases presented with a pelvic abscess (infected presacral dermoid cyst), which were initially drained followed by excision. The presacral mass consisted of either lipomyelocele (6), lipomyelomeningocele (3), or a developmental

(dermoid) cyst (8). Tethering of the spinal cord was a common association (70%)

Apart from diagnostic challenges, the management of Currarino syndrome is similar to the usual management of ARM regarding the surgical approach and probably the prognosis that mainly depends on degree of associated sacral dysplasia [10].

Duru et al., presented five cases of Currarino triad who underwent surgical intervention. All patients had sacral bony deformity, anorectal malformations, and anterior sacral meningocele. A 40-year-old-male had chronic constipation. He was incidentally diagnosed with Currarino syndrome. A 19-year-old-female suffered from a slight weakness in lower extremities and urinary incontinence. Her past medical history was remarkable for anal atresia. The other three cases were children [11].

Case reports

Hage et al., reported a previously healthy 3-month-old girl with abdominal distension, post-prandial vomiting, obstipation, and anuria of 5 days. Abdomino-pelvic magnetic resonance imaging (MRI) showed a large cystic multilobulated mass in the sacrococcygeal region with a dural communication evident of an anterior sacral meningocele. 1 year later, the child came back with constipation and was found to a have a malignant mixed germ cell tumor in the presacral area, a very rare presentation in Currarino syndrome.

In a child presenting with at least one of the features of Currarino syndrome's triad, a diagnosis should be suspected. After reviewing the literature, the syndrome is usually missed and hence is under diagnosed. MRI is the best imaging modality for diagnostics and follow-up for any mass, benign or malignant, can bring life saving measures. Most masses are benign but can undergo malignant transformation even after resection. De novo malignancy is very rare and is described in our case.

Physicians treating patients with spinal dysraphism should suspect a diagnosis of Currarino syndrome by follow up imaging for any new benign or malignant growth [12].

A 44-year-old man was diagnosed with Currarino triad, with a huge presacral teratoma and meningocele. One-stage surgery via posterior approach was successful [13].

A 18-year-old female with Currarino Syndrome who underwent surgery by a combined abdominal laparoscopic-posterior Kraske approach for the resection of a large retrorectal tumor [14].

A 36-year-old female who presented with late onset of symptoms attributed to CS (e.g., presacral mass, anterior sacral meningocele, and sacral agenesis). She successfully underwent multidisciplinary single-stage approach for treatment of the anterior sacral meningocele and resection of the presacral mass. This required obliteration of the meningocele and closure of the dural defect. One year later, her meningocele had fully resolved.

While late presentations with CS are rare, early detection and multidisciplinary treatment including single-state anterior may be successful for managing these patients [15].

A young woman who presented with recurrent abortion and later with a huge palpable mass of pelvic origin, measuring 14 × 12 cm.

The various radiologic features, associations such as presacral dermoid as a part of incomplete Currarino syndrome are described [16].

A rare case of Currarino syndrome in an infant with tethered cord syndrome and a dorsal lipomyelomeningocele continuous with a presacral intradural spinal lipoma, in addition to an imperforate anus and a scimitar sacrum [17].

Sala et al., described the case of a retained medullary cord (RMC) in a 1.5-year-old child with Currarino syndrome. At surgery, an apparently normal-looking spinal cord, stretched and tethered by a lipoma to the level of S2-S3, was observed. The border between the functional conus and the non functional RMC was defined through neurophysiological mapping. The cord was sharply interrupted at this level and untethered. A specimen was sent for pathology, which confirmed the presence of glial and neural elements. The post-operative neurological exam was normal.

Neurosurgical procedure for RMC should only be rendered with intraoperative neurophysiological mapping, as the anatomical judgment would not suffice to allow a safe cutting of these “normal-looking” neural structures [18].

An adult patient with CS, manifesting by an acute intestinal obstruction. Acute intestinal obstruction in an adult as a presentation of CS was not been reported previously. Colostomy was performed first by the general surgery team to relieve intestinal obstruction caused by the giant cyst. After the final diagnosis of anterior sacral meningocele was established, a second operation was performed for the ligation of the cyst neck through a posterior approach. The size of the cyst gradually reduced over time. A staged approach and the multidisciplinary management, with the collaboration of the general surgery and neurosurgery teams, provided a satisfactory clinical outcome [19].

Unclassified

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