Neurochirurgie xxx (2017) xxx–xxx

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Clinical case

Multiple neurosurgical treatments for different members of the same family with Currarino syndrome

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ARTICLE INFO

Article history:
Received 26 September 2017
Received in revised form 2 December 2017
Accepted 27 January 2018
Available online xxx

Keywords:
Currarino syndrome
Functional digestive and urinary disorders
Anterior sacral meningocele
Tethered spinal cord syndrome
Lipoma of the filum terminale
Neurosurgery

ABSTRACT

Introduction. – 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 sacral bony defects. Mutations of the *HLXB9* gene have been identified in most CS cases, but a precise genotype-phenotype correlation has not been described so far. Family screening is obligatory. 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. Surgical management is provided by pediatric surgeons or neurosurgeons.
Familial case report. – Here, we present a family case report with CS requiring different neurosurgical management. The son, a 3-year-old boy, developed a tethered spinal 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 thyreoglossal tract 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. The ligation of the neck of the cyst and aspiration of the liquid inside in full through a posterior partial approach permit a complete collapse of the cyst with an instantly satisfactory clinical outcome.
Conclusion. – In these cases, cooperation between pediatric surgeons and neurosurgeons was crucial. The follow-up of these patients should be done in a spina bifida clinic. 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).

1. Introduction

In 1926, Kennedy was the first to report an association of sacral bony defect, anterior sacral meningocele and rectal polyp [1]. But it was only 50 years later that Currarino et al. described the triad as a complex syndrome [2]. Since then, approximately 300 pediatric cases and about 60 adult cases have been reported in the literature. Incidence is probably higher, because this pathology is little known by clinicians and many patients still go undiagnosed [3].

Currarino syndrome (CS) belongs to the group of neural malformations [4,5]. It results from an abnormal separation between the neuroectoderm and the endoderm [4]. A common embryological abnormality in the triad was suggested by Currarino, in which he was followed by several authors [6,7]. It is characterized by anorectal malformations, abnormalities in the sacral bone and presence of a presacral mass. It is complete if all 3 anomalies are present, and partial or incomplete in the presence of a hemisacrum associated with 1 of the other 2 malformations (presacral mass or anorectal abnormalities) [3–5].

CS is the only known form of hereditary sacral agenesis. The mode of inheritance is autosomal dominant in nearly half of the cases. Mutations of the HLXB9 homeobox gene (which encodes a transcription factor, the HB9 protein), located in the chromosomal 7q36 region, have been demonstrated in many cases of CS and in only 30% of sporadic cases. This suggests that other genes are involved in the syndrome [8,9]. Disease penetrance can be was

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https://doi.org/10.1016/j.neuchi.2018.01.009
0028-3770/

incomplete and expressivity may vary, even within a given family. No correlation was found between phenotype and genotype [9–11].

Here, we present a family case report of CS, requiring multiple neurosurgical management.

2. Family case report

The first case was a 3-year-old child followed for sacral agenesis (Fig. 1A). He was operated on with a multidisciplinary pediatric surgical approach:

- pediatric neurosurgery for tethered spinal cord syndrome associated with lipoma of the filum terminale (Fig. 1B);
- pediatric surgery for a sacrococcygeal teratoma and anal adhesion (Fig. 1C).

The pathology confirmed the diagnosis of lipoma of the filum terminale. The rectal tumor comprised was adipose tissue, with no malignant aspect. Progression was satisfactory. The child had acquired cleanliness 3 months before surgery. Postoperatively, he developed disorders of urination (interruption of the urinary jet and drops at the end of miction), and diurnal urine leaks. The parents noticed that since the operation he no longer asked to use the toilet, but and urinated in his diaper. This is actually very unusual after surgery for a fibromatous lipoma. Renovesical ultrasound and cystography had returned to normal values. The child had urological follow-up due to the infectious risk in the urine. He developed fecal incontinence, with megarectum and megasigmoid on X-ray opacification. The Peristeen® transanal irrigation system was introduced at a frequency of twice daily, with 350 mL water; this was very productive, and very well-tolerated, allowing discharge home with instructions to continue the procedure. Determination of blood tumor markers found alpha-fetoprotein at 1.94 ng/mL and beta-HCG at < 0.04 ng/mL.

The second case was the daughter, who was 2 years old when diagnosed. The situation was much more reassuring. The conus of the spinal cord was in a physiological position. There was a very small lipoma of the filum terminale that required neurosurgical management as a precaution (Fig. 2A and B). She also presented a similar image at the sacrum (Fig. 2C). The only manifestation was a cyst of the thyroglossal tract, infected and fistulized to the skin. Resection was completed by a Meunier stitch in the tongue base after a Sistrunk procedure. Pathology diagnosed a benign lesion. In the blood, alpha-fetoprotein was at 8.62 ng/mL and beta-HCG at < 0.04 ng/mL.

The third case was the father of the children, who was 44 years old, with surgical history of anal imperforation at birth operated on during the neo-natal period. He was followed for CS after the diagnosis in his son and due to the development of a very large pelvic mass. There were signs of pelvic organ compression with urinary disorder (pollakiuria), digestive disorder (chronic constipation treated with laxatives such as Macrogol) and poorly systematized root pain. There was also intracranial hypotension during certain efforts. MRI (Fig. 3A) found a voluminous anterior cystic tumor mass with meningocele and communication under the fourth sacral portion. A posterior approach was performed via partial lumbosacral laminectomy (Video). The giant anterior sacral meningocele was exposed extradurally. Levels S3, S4 and S5 were approached, and the collar of the cyst was found to be in communication with the dura, and was ligated with non-absorbable thread. Liquid inside the cyst was aspirated in totality. After centrifugation and MGG staining, the cystic fluid appeared hematic, with predominant red blood cells without any other identifiable abnormal cellular elements. *Staphylococcus pasteuri*, a multi-susceptible coagulase-negative Gram-positive organism, was detected in the liquid. The patient never presented clinical or biological signs of meningococcus or infectious syndrome, and did not receive any antibiotics. Pathology was consistent with meningocele, showing fibrous meningo-epithelial tissue. Postoperative MRI (Fig. 3B), early after surgery and 3 months later, showed complete regression of the meningocele. Neurological examination was normal. The patient no longer complained of constipation.

3. Discussion

Currarino syndrome results from an abnormal separation between the neuroectoderm and the endoderm during embryogenesis [4]. In the normal state, the endodermal leaflet (future gastrointestinal tract) closes at about the same time as the neuroectoderm (future neural tube). The notochord and somites form the future vertebral bodies, thus isolating the neural tube from the primitive intestine. The persistence of an abnormal adhesion between the endoderm and the neuroectoderm prevents anterior fusion of the vertebral body, resulting in a “fistula” between the digestive and neural elements. Partial resorption of this “fistula” on the dorsal side leads to meningocele, and on the ventral side to an enteric cyst. Teratoma forms if the enteric and neural elements remain bound by elements of the mesoderm which have migrated into the presacral space during the development of the somites [2,6].

Anorectal malformations in CS comprise anal atresia, anorectal stenosis, anal ectopia, anal imperforation, rectourethral fistula or rectovaginal fistula [12].

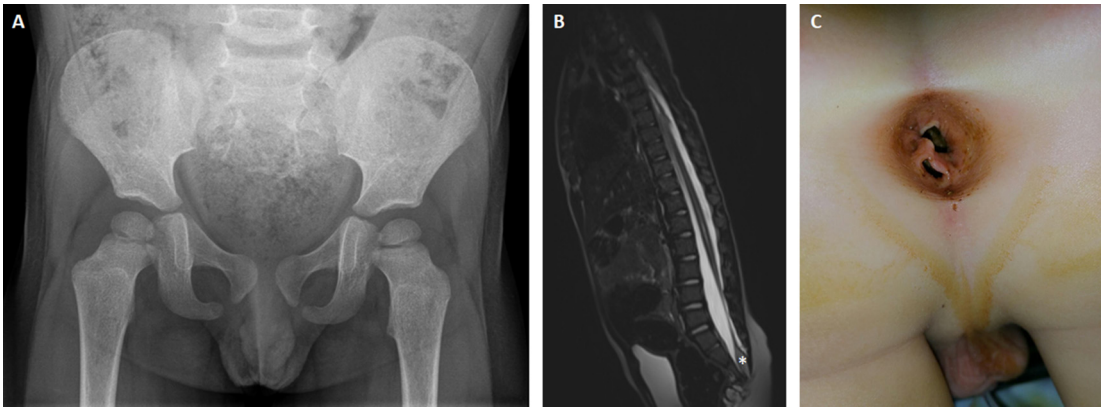

Fig. 1. A. Initial radiograph of the pelvis: sacral agenesis. B. Tethered spinal cord syndrome associated with lipoma of the filum terminale (white asterisk) (MRI before surgery, sagittal view, T2 sequence). C. Anal adhesion.

Please cite this article in press as: Serratrice N, et al. Multiple neurosurgical treatments for different members of the same family with Currarino syndrome. Neurochirurgie (2017), https://doi.org/10.1016/j.neuchi.2018.01.009