Currarino syndrome with intramedullary spinal cord abscess related communication between the tethered cord and a presacral mass: A case report

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A B S T R A C T

We herein report the case of a 21-day-old boy in which the detection of an intramedullary spinal cord abscess led to the diagnosis of Currarino syndrome (CS). He had a complete phenotype of CS, including sacral agenesis, an anorectal malformation, a presacral mass, and spinal cord malformations. In addition, he had an intramedullary spinal cord abscess. Intramedullary spinal cord abscess is rare in CS and is thought to require immediate intervention. Therefore, we additionally reviewed the available literature and discussed the therapeutic approach for CS with an intramedullary cord abscess.

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Currarino syndrome (CS) is a hereditary disorder characterized as the triad of an anorectal malformation, sacral bony defect, and presacral mass. This syndrome was first described by Currarino et al. [1]. And CS is now known to be associated with the genetic mutation of HLXB9 located at 7q36 [2]. The clinical phenotype of CS is variable. Some patients with CS do not exhibit the complete triad [3].

We herein report a rare case of a 21-day-old boy with Currarino syndrome manifesting as an intramedullary spinal cord abscess and review the treatment of CS with an intramedullary spinal cord abscess.

1. Case report

A 21-day-old boy was referred to a previous hospital due to a high fever. He was diagnosed as a urinary tract infection with a neurogenic bladder, and ceftriaxone was administered. Because the high fever persisted, he was transferred to our hospital and was diagnosed as an intramedullary spinal cord abscess by the lumbar puncture and magnetic resonance imaging (MRI) (Fig. 1). The cerebrospinal fluid was cloudy and cell count of the cerebrospinal fluid was 13,356/mm³ (mononuclear cells: 1368/mm³; polymorphonuclear cells: 11988/mm³). Antibiotic therapy, including both panipenem/betamipron and gentamicin, was initiated and gentamicin was administered for two weeks. Panipenem/betamipron was changed to cefotaxime and administered for four weeks. Thereafter the patient's infection improved.

On admission to our department, funnel anus was also identified and neurogenic bladder was suspected, because he required urethral catheterization for urination. The X-ray film findings showed a hemisacrum, also known as a “scimitar sign,” and MRI revealed the presacral tumor which was suspected to be a multicystic lipomatous tumor. In addition, thin slice image computed tomography (CT) was performed to allow for the precise evaluation of the anatomical findings. The CT showed the narrow continuity between the presacral tumor and rectal cavity; the presacral tumor continued to the spinal cord abscess through the Tethered cord. Similarly, a barium enema showed the fistula between the presacral tumor and rectum in addition to anal stenosis (Fig. 2).

We first performed colostomy at the transverse colon to prepare for the repair of the anorectal malformation. We did not select a one-stage treatment, including the resection of the presacral mass...
and neurosurgery, because the treatment was thought to involve a high-risk of the recurrence of the abscess or meningitis. In fact, the colostomy prevented the recurrence of the spinal cord abscess after the presacral abscess improved.

Additionally, the repair of the Tethered cord was performed on the boy at 50 days of age and no recurrence has been observed following the repairing operation. At 6 months of age, the patient received presacral tumor extirpation through the posterior sagittal approach, and the rectal stenosis, 1.5 cm in length, was completely resected. We were not able to maintain his native anal mucosa because the anal stenosis was extremely rigid and the anal canal was less than 8Fr in diameter. The sacral bony defect that was detected preoperatively was not the coccyx bone. The gross specimen consisted of a 3.5 × 2.7 × 2.0 cm soft tumor with multiple cystic components (Fig. 2). The pathological findings showed the lipomatous tissue of the tumor was a mature teratoma with a cystic lesion. He was discharged on postoperative day 8. His colostomy was closed at one years of age. After these treatments, no recurrence of the presacral mass during a 1.5-year follow-up period has been observed. Neurogenic bladder was confirmed by a urodynamic study during the follow-up period. In addition, the patient currently requires the use of a glycerin enema for constipation, because the anal function was affected by the tethered cord.

2. Genetic examination

Informed consent for the genetic examination, only chromosome examinations, was obtained from the patient’s parents. The genetic examination detected a mutation at the chromosomal region 7q34 in the present patient. Therefore, he was diagnosed with complete Currarino syndrome. We did not perform the familial screening because his parents did not consent to the screening genetic examinations.

3. Discussion

CS is a rare complex of congenital caudal anomalies, including the following three features: a sacral bony deformity, anorectal malformations, and a presacral mass. Since it was first described in
1926 by Kennedy RLJ [4] and further defined by Currarino to be the association of sacral agenesis, anorectal malformation and a presacral mass in 1981 [1]. CS has been described in over 200 cases, of which at least 50% were familial [5], and is known to be a genetic autosomal-dominant disorder. Recently, a gene mutation in the HUX89 gene located at the chromosomal region 7q36 in CS was found [2]. The study of HUX89 expression during embryonic sacral development has been previously reported by Ross et al. [6]. However, it remains possible that mutations elsewhere in the HUX89 gene may lead to the clinical phenotype [7]. In the present case, the gene mutation was additionally detected at the chromosomal region 7q34, and thus he was diagnosed with complete Currarino syndrome. CS also has a variety of presentations, depending on the expressivity of the syndrome [3].

Additionally, we reviewed the symptoms and the treatment for CS associated with meningitis and presacral abscesses, including an intramedullary spinal cord abscess. A total of 120 patients was extracted from 9 studies on the major features of CS (Table 1) [1e3,8e13]. A presacral mass was present in 92 (76.7%) patients, a sacral defect in 112 (94.2%) patients, and anorectal malformations in 78 patients (65.0%), including anal stenosis (48 patients: 40%) and an imperforate anus (30 patients: 25%). The most frequent symptom was constipation (65 patients: 54.2%), followed by urogenital symptoms (13 patients: 10.8%), including urinary tract infection and neurogenic bladder. Furthermore, meningitis is one of the most severe complications in CS and occurred in 7e11% of all cases, including familial cases in previous reports [9]. The communication between the spinal canal and the presacral tumor through the tethered cord is considered to be the cause of the patient’s meningitis and spinal cord abscess [1,14,15]. The mortality due to meningitis in CS is surprisingly high at 56% [10]. In our review, 12 patients (10%) presented meningitis and 5 of 12 patients had presacral abscesses. In 12 patients, presacral abscesses were detected and 3 patients died due to meningitis associated with presacral abscesses. According to our review, the mortality of meningitis in CS is 25%.

The onset of neurological complication in this syndrome may also be associated with a Tethered cord in approximately 63% of the patients [8]. It is hypothesized that meningitis is caused by a Tethered cord communicating between the presacral space and intramedullary space [1,14,15]. In severe cases, the cavity of the abscess extends to the intramedullary space and leads to a life-threatening septic condition. Therefore, the patients with a Tethered cord require immediate separation. Twenty-seven patients (22.5%) with CS have a Tethered cord according to our literature review and these patients are a high-risk group due to life-threatening meningitis and presacral abscesses. Therefore, the surgeons must always be alert to the possibility of a communication between the spinal canal and the tumor, which could lead to severe neurological complications if not treated correctly [8].

Another consideration for the treatments of CS is the presence of anorectal malformations. In CS patients, most of the anorectal malformations in the previously reported cases presented as anorectal stenosis and the treatment is typically anorectal dilatation. Our review, including 39 patients from 7 studies in which the record of the treatment was available, showed that dilatation was performed in 19 of 23 patients who had anorectal stenosis. After the dilatation, resection of the presacral mass was performed in almost patients. However, resection alone may not relieve the patients from constipation because constipation may also be caused by spinal cord tethering, in addition to a narrow anal canal.

The conservative management, including dilatation, does not often lead to satisfactory results [16]. In some reports, PSARP is...
indicated to be a well-established procedure for treating anorectal stenosis and an excellent approach for the excision of the presacral tumor [5]. Based on our experience, we additionally believe this procedure is best for the immediate separation of the fistula between the intradural space and the rectum and the prevention of life-threatening meningitis and presacral abscesses.

Conversely, in the present case, colostomy was first performed to prevent the recurrence of meningitis because he had already had severe meningitis due to an intramedullary spinal cord abscess and the severe infection needed to be controlled. Some authors recommend the use of a single-step surgery, however, this procedure should not be performed under an infectious condition. We speculate that, in complete CS with a tethered cord, colostomy should be initially performed for both preventing the infectious complication and managing the defecation. The repair of a tethered cord should be performed as the second treatment to prevent any complications associated with infection. The treatment strategy differs according to the clinical type in CS; therefore, it is important for the surgeons to decide when and which site will be first repaired in a patient with CS. Martucciello et al. showed the diagnostic and therapeutic strategy for CS in 2004 [12]. Their strategy indicated that colostomy and neurosurgery should be initially performed in complete CS with a Tethered cord. In addition, in the cases with anorectal malformations without neural tube defects, they indicated that primary PSARP should be initially performed.

Based on the experience of the present case, their strategy for treatment according to the clinical type of CS is believed to be good for the patients with CS. Furthermore, the present case indicated that CS patients with an intramedullary spinal cord abscess should be managed by antibiotic therapy and colostomy as an initial strategy for treatment. In addition, in CS patients with meningitis or intramedullary spinal cord abscess related to tethered cord, the repair of tethered cord should be considered in order to reduce the risk of the recurrence of life-threatening meningitis or intramedullary spinal cord abscess.

**Conflict of interest**

The authors declare that they have no conflicts of interest.

**References**