PONTANEOUS intracranial hypotension with severe headache secondary to a CSF leak is a well-established condition. Recently, dural ectasia has been described as a common finding in patients with connective tissue disorders and it is purported to be implicated in CSF leakage at these dural weak points.

We present a case of a child suffering from headaches in whom we discovered both tonsillar herniation and dural ectasia of the sacral nerve roots. Tonsillar herniation is a known complication of a CSF leak after lumbar puncture and lumboperitoneal shunt placement. To our knowledge, however, this is the first reported case of a patient with Marfan syndrome presenting with an asymptomatic spontaneous CSF leak complicated by tonsillar herniation. In this rare association of SIH and connective tissue disorders, recognition of the clinical signs and typical MR imaging features of SIH may lead to more appropriate and less invasive treatment, potentially avoiding surgery.

KEY WORDS • intracranial hypotension • tonsillar herniation • Chiari malformation • Marfan syndrome • blood patch • pediatric neurosurgery

The authors describe the case of a 12-year-old girl with Marfan syndrome, sacral dural ectasia, and tonsillar herniation, who presented with headache. Initially, it was hypothesized that the headaches were secondary to the tonsillar herniation, and the patient consequently underwent surgical decompression of the foramen magnum. Postoperatively, the patient’s condition did not improve, and additional magnetic resonance (MR) imaging demonstrated evidence of a cerebrospinal fluid (CSF) leak at the level of the dural ectasia. It was surmised that the girl’s symptoms were due to spontaneous intracranial hypotension (SIH) and that the tonsillar herniation was caused by the leakage. The patient responded well to application of a blood patch at the level of the demonstrated leak, and her headache resolved. This appears to be the first reported case of a patient with Marfan syndrome presenting with a symptomatic spontaneous CSF leak complicated by tonsillar herniation. In this rare association of SIH and connective tissue disorders, recognition of the clinical signs and typical MR imaging features of SIH may lead to more appropriate and less invasive treatment, potentially avoiding surgery.

Abbreviations used in this paper: CM-I = Chiari malformation Type I; CSF = cerebrospinal fluid; MR = magnetic resonance; SIH = spontaneous intracranial hypotension.
vomiting, all of these symptoms improving within a few minutes of lying down.

The results of routine blood examinations were normal. A review of the MR images demonstrated tonsillar herniation in the context of small ventricles. A spinal MR imaging study was performed; the images revealed widening of the dural sac and bilateral meningeal diverticula at the level of the first sacral nerve root with pachymeningeal enhancement but no evidence of syrinx formation.

Although the possibility of SIH was discussed, the tonsillar herniation provided adequate explanation for the patient’s intractable headache, and a decision was made to operate on the posterior fossa.

First Surgical Procedure and Postoperative Course. We performed a foramen magnum decompression while the patient was in a prone position, opening the C-1 lamina. The dura mater was opened, but the arachnoid membrane was left intact. On opening the dura, we observed evidence of intracranial hypotension; there was a paucity of CSF such that the subarachnoid space was diminished and the arachnoid mater was very close to the pia mater.

Postoperatively the patient’s condition did not improve; indeed, she reported worsening of her headaches, again with a strong postural component. It was evident from her clinical response and the operative findings that her symptoms were due to SIH and that the tonsillar descent was probably a secondary phenomenon.

The patient underwent MR myelography including $T_1$-weighted sequences (with and without a contrast agent) and $T_2$-weighted sequences, which confirmed the presence of sacral meningeal diverticula (Fig. 2). Additionally there was evidence of an ongoing CSF leak on the left side at the level of the S-1 nerve root with extravasation of contrast medium into the soft tissues (Fig. 3).

Second and Third Surgical Procedures and Postoperative Course. After a postoperative interval of 1 week (a trial of conservative management), the patient was treated with an autologous epidural blood patch at the L5–S1 level. This treatment resulted in marked improvement in her symptoms over the next 5 days. The initial improvement was not fully maintained, and she required application of one more blood

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Fig. 1. Sagittal MR images. **Upper:** Preoperative image of the craniocervical junction showing tonsillar herniation. **Lower:** Posttreatment image (obtained 2 weeks after placement of the second blood patch) demonstrating improvement of the abnormality.

Fig. 2. Coronal $T_2$-weighted MR myelography image of the lumbosacral region showing diverticula at the S-1 nerve root.
The cause for these CSF extravasations may be trauma, weakness of the dural sac, or a combination of both. Recently, Foran and associates described seven cases of acquired dural ectasia in patients with Marfan syndrome. They reported that in all cases treatment with a blood patch led to excellent clinical results and resolution of symptoms. In our case, MR imaging clearly demonstrated the CSF leak complicating Marfan syndrome. These findings, in combination with the operative findings and the patient’s clinical response, indicate that our patient suffered from SIH. The clinical difficulty in this case lay in the fact that the patient also had tonsillar herniation, which in itself could provoke headache through compression of neural structures.

I. Discussion

The syndrome of SIH has been well described by Mokri and is defined as a decrease in CSF pressure to less than 60 mm H₂O associated with occipital headache. On MR images, it is characterized by pachymeningeal enhancement (secondary to dural venous dilatation, descent of the cerebellar tonsils, a decrease in the size of the preptonepine and prechiasmatic cisterns, inferior displacement of the optic chiasm, and crowding of the posterior fossa. Other common findings include subdural fluid collections, small and often slitlike ventricles, and spinal meningeal diverticula.

Intracranial hypotension is a well-recognized sequela of a spontaneous CSF leak, particularly in cases in which the leak involves the thoracic spine. The cause for these CSF leaks remains unclear, but authors have postulated minor trauma, weakness of the dural sac, or a combination of both. Recently, there have also been interesting reports of the use of radiolotope cisternography to demonstrate sites of CSF extravasation.

Dural ectasia is well established as a highly characteristic sign in patients suffering from connective tissue disorders. Fattor and colleagues reviewed 83 cases of Marfan syndrome and found a 92% rate of dural ectasia documented on MR images. Moreover, Schrijver and coauthors reviewed cases in which patients presented with spontaneous CSF leaks and found that 20% of the patients had minor skeletal features characteristic of Marfan syndrome. The pathophysiology of the dural ectasia seen in patients with Marfan syndrome is thought to relate to alterations in the elastin composition of the dura, which allow CSF pulsation to dilate the dural sac, thereby sometimes leading to scalloping of the vertebral bodies. Recently, Foran and associates tried to characterize the symptoms associated with confirmed dural ectasia in 22 patients with Marfan syndrome. They reported low-back pain, headache, proximal leg pain, weakness and paresthesia both above and below the knee, and genital and rectal pain. These symptoms often occurred daily and were commonly exacerbated by upright posture. Other authors reported patients with Marfan syndrome suffering from headache, but few could identify, on MR imaging or cisternography, the CSF leak associated with the dural ectasia.

The pathophysiology of postural headache in intracranial hypotension caused by a CSF leak remains unclear. The headache is postulated to be caused by traction on pain-sensitive structures of the brain, as a consequence of the descent of the brain due to CSF volume depletion. A number of case reports involving patients with Marfan syndrome support this mechanism, with resolution of headache after treatment of the CSF leak.

In our case, MR imaging clearly demonstrated the CSF leak complicating Marfan syndrome. These findings, in combination with the operative findings and the patient’s clinical response, indicate that our patient suffered from SIH. The clinical difficulty in this case lay in the fact that the patient also had tonsillar herniation, which in itself could provoke headache through compression of neural structures.

Tonsillar herniation, which may also be called “acquired” CM-I, can occur after lumboperitoneal shunt placement or the performance of multiple lumbar punctures. Atkinson and colleagues described seven cases of acquired CM-I secondary to a spontaneous CSF leak and chronic SIH. Imaging showed a leak in six cases and an arachnoid diverticulum at L-2 in one case. No syringomyelia could be identified. The authors purport that, in cases of SIH, tonsillar herniation is associated with a characteristic pachymeningeal enhancement, a pattern not seen in idiopathic CM-I. This imaging feature has the potential to facilitate the diagnosis and therefore the treatment of the CSF leak—helping to prevent unnecessary suboccipital craniectomies. Interestingly, in their series, Atkinson et al. describe a patient similar to ours who was treated with craniectomy without resolution of symptoms. There are descriptions of two other cases in the literature, involving an association of postural headache, a CSF leak, and tonsillar herniation. In both cases treatment with a blood patch led to excellent clinical results and resolution of the brain sagging on follow-up MR imaging. In our case, the neurosurgeon noticed intraoperatively an unusual appearance of the arachnoid mater, which was extremely close to the pia mater. These findings
support the diagnosis of subarachnoid space hypotension with secondary tonsillar herniation.

In the reported cases of SIH due to a CSF leak, symptoms were sometimes relieved after bed rest and rehydration. When this conservative management fails, the condition can be effectively treated by placement of an epidural blood patch at the interspace corresponding to the level of the leak,3,12,14 but the procedure may need to be repeated.6,9,10 There are also reports of treatment with epidural saline injection.3,5 In cases of headache and a CSF leak in the context of connective tissue disorders, the recommended treatment is application of an autologous epidural blood patch.1,12,14,21 If this treatment is unsuccessful or if the meningeal diverticulum is too large, it is possible to perform a direct surgical repair of the site of the leak by using a circumferential suture and/or packing the lesion with muscle or fibrin glue or a blood patch.1,12,14,21

Some authors have noted a rebound period of intracranial hypertension following treatment of the leak, however in all cases the hypertension was self-limiting.1,5,9 In our case, the placement of the initial blood patch was followed by bed rest for one week. This treatment may have failed to resolve all of the patient’s symptoms due to insufficient duration of bed rest. The second epidural blood patch procedure was immediately successful without complications. There has been no evidence of any rebound intracranial hypertension or any other symptoms since then.

Conclusions

We present a case of SIH and tonsillar herniation secondary to a CSF leak in a patient with Marfan syndrome. To our knowledge, this is the first published case involving this particular association of clinical and radiological findings. Most patients with SIH and connective tissue disorders have meningeal ectasia, but the site of the CSF leak can be identified in only a minority.

In our case, the tonsillar herniation was undoubtedly a consequence of SIH due to a chronic CSF leak at the site of meningeal ectasia in the sacral nerve roots. Initially the significance of the dural ectasia was not fully appreciated, and the patient was treated for idiopathic CM-I. In rare cases such as this one, the combination of marfanoid features and the characteristic pachymeningeal enhancement on MR imaging can guide us to the correct diagnosis. With appropriate recognition, treatment can be tailored to the patient to provide relief of symptoms without unnecessary surgery.

References
