Case Report

Intracranial hypotension in a boy with Marfan syndrome

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Abstract

Intracranial hypotension is a rare condition in children and adolescents and the management is not standardized. We report here a child with intracranial hypotension due to Marfan syndrome. A 12-year-old boy presented persistent bilateral frontal headache over two weeks. The patient had no headache when he was in a supine position and it was more severe when he was in an upright position. His physical examination revealed a characteristic marfanoid habitus with tall stature, pectus carinatum, lens dislocation, and joint hyperextensibility. Lumbar puncture was performed and cerebrospinal fluid (CSF) opening pressure was measured below 60 mmH2O. The imaging of the patient showed bilateral subdural hemorrhage, cerebellar tonsillar herniation into the foramen magnum, dural venous distention, pachymeningeal enhancement, dural ectasia, and thoracic CSF leak on the left side. According to these findings, Marfan syndrome and intracranial hypotension syndrome were diagnosed.

Keywords: children, headache, intracranial hypotension, Marfan syndrome

Introduction

Intracranial hypotension is determined to be a rare entity in children and adolescents. It is also difficult to diagnose and the management is not standardized [1]. The patients with intracranial hypotension usually suffer from postural headache. Marfan syndrome is a type of connective tissue disorder with multisystemic involvement resulting in musculoskeletal, ocular, and cardiovascular abnormalities. Marfan syndrome may also lead to dural ectasia and intracranial hypotension [2]. The presentation, diagnosis, and management of spontaneous intracranial hypotension (SIH) are controversial in children with Marfan syndrome. We report here a child with spontaneous cerebrospinal fluid (CSF) leak and SIH due to Marfan syndrome.

Case

A 12-year-old male patient presented persistent bilateral frontal headache over two weeks. There was no headache when the patient was
in a supine position, however, it was more severe when he was in an upright position. There was no history of trauma. His physical examination revealed a characteristic marfanoid habitus with tall stature, pectus carinatum, lens dislocation, and joint hyperextensibility. His limbs were disproportionately long. There were no abnormal neurological examination findings. His echocardiographic examination showed mitral valve prolapse, mitral insufficiency, and aortic dilatation. His family history showed that his father, his uncle, and his brother had died after acute chest pain. All of those relatives had a physical appearance of Marfan syndrome. According to family history, clinical and laboratory findings, our patient was diagnosed with Marfan syndrome.

The diagnosis of intracranial hypotension syndrome in our patient was suspected with the characteristic history of headache. Lumbar puncture was performed, CSF opening pressure was measured below 60 mmH₂O and intracranial hypotension syndrome was also diagnosed. In order to evaluate the etiology of intracranial hypotension syndrome, brain and spinal magnetic resonance imaging (MRI) with gadolinium were performed. Contrast-enhanced brain MRI (Figure 1) showed suggestive findings of intracranial hypotension such as bilateral subdural hemorrhage, cerebellar tonsillar herniation into the foramen magnum, dural venous distention, and pachymeningeal enhancement. Spinal MRI and MRI myelography were also performed. Spinal MRI (Figure 2) demonstrated dural ectasia and MR myelography (Figure 3) revealed thoracic CSF leak on the left side. Postural headache was improved with rest and hydration.

**Figure 1.** (A) shows bilateral frontal subdural hemorrhage, (B) shows mild extensions of the cerebellar tonsils into the foramen magnum associated with dilated superior sagittal sinus and straight sinus, (C) demonstrates bilateral dural enhancement.

**Figure 2.** shows dural ectasia in the lumbosacral region.

**Figure 3.** demonstrates large diverticulum, as the presumed localization of the CSF leak at T7 vertebra level on the left side.
Discussion

We presented here an intracranial hypotension syndrome due to nontraumatic CSF leak because of dural ectasia in a boy with Marfan syndrome. The classic features of headache attributed to SIH are orthostatic headache, low CSF pressure, and diffuse meningeal enhancement on brain MRI [1,3]. In our patient, the presenting complaint was a postural headache. His headache did not respond to painkillers and increased with standing.

The annual incidence of headache attributed to SIH is approximately 5 per 100,000 with a peak incidence around the age of 40. Female to male ratio is reported as 2:1. Unlike adults, intracranial hypotension syndrome due to CSF leak secondary to dural ectasia is very rare among children [4]. Schievink et al. evaluated the patients under the age of 20 who had SIH. They reported only one patient under the age of 20 with Marfan syndrome (19-year-old) [5].

Normally, CSF opening pressure is admitted to be 60 to 200 mmH\textsubscript{2}O. CSF opening pressure is lower than 60 mmH\textsubscript{2}O in patients with SIH [4]. In some cases, it is immeasurably low and CSF may not be obtained. However, we performed lumbar puncture and CSF opening pressure was measured below 60 mmH\textsubscript{2}O in our patient. Most CSF leaks occur at the thoracic or cervicothoracic area. Our patient’s myelography showed that there was a CSF leak at the level of 7th thoracic vertebra.

An underlying connective tissue disorder may play a role in the development of CSF leaks. SIH may be associated with connective tissue diseases such as Marfan syndrome, Ehlers-Danlos syndrome, and type II autosomal dominant polycystic kidney disease. Other potential contributing factors are minor trauma, degenerative disc disease, and osseous spurs [2]. However, these contributing factors were not found in our patient. According to our knowledge, our case is the first male patient aged under 18 years with Marfan syndrome and intracranial hypotension syndrome due to CSF leak secondary to dural ectasia. In the literature, similar cases have been reported, but all of them are female [6-9].

Isolated or multiple cranial nerve palsies may be accompanied with SIH. It has been reported that 3, 4, 5, 6, 7, 8, and 9 cranial nerves may be affected in SIH [10]. However, our case has no additional neurological pathalogy.

Patients with a postural headache and suspected SIH should be first evaluated with contrast MRI [4]. Cranial MRI is one of the most important guides to confirm the diagnosis of SIH. In the current case, the first symptom was an orthostatic headache and the diagnosis of SIH was confirmed with cranial MRI. MRI findings of severe intracranial hypotension are pachymeningeal contrast enhancement, subdural fluid collection and hygroma, pituitary hyperemia, and displacement of brain tissue [4]. Additionally, radionuclide cisternography, CT-myelography, and MR-myelography are performed to determine the localization of spinal congestion [2,11]. We showed cerebellar tonsillar herniation into the foramen magnum, dural venous distention, pachymeningeal enhancement, dural ectasia, and thoracic CSF leak in imaging techniques.

In recent years, a growing number of SIH has been reported. A delay in diagnosis leads to serious neurological complications. On the other hand, the clinical entity may be reversible with early diagnosis and treatment. Therefore, clinicians should be careful about this rare condition.

Conflict of interest

All authors declare that they have no conflict of interest.

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References