Cerebrospinal fluid leakage diagnosis in Marfan syndrome

- Pituitary pseudo-adenoma from cerebrospinal fluid leak in patient with Marfan syndrome
- Spontaneous Intracranial Hypotension Associated with Marfan Syndrome: A Case Report
- Intracranial hypotension syndrome in a pediatric patient with Marfan syndrome
- Factors Associated with Nontraumatic Spontaneous Subdural Hematomas in Pediatric Patients
- Dural ectasia and intracranial hypotension in Marfan syndrome
- Spontaneous, Intrasphenoidal Rupture of Ecchordosis Physaliphora with Pneumocephalus Captured During Serial Imaging and Clinical Follow-Up: Pathoanatomic Features and Management
- An Unusual Cause of Chronic Headache in an Adolescent Boy: A Case Report
- Prevalence of temporal bone tegmen defects among patients with Marfan syndrome

Diagnosing cerebrospinal fluid (CSF) leakage in individuals with Marfan syndrome or any other condition involves a combination of clinical evaluation, medical history, and diagnostic tests. CSF leakage can be a serious issue and should be promptly diagnosed and treated. Here are the steps typically involved in diagnosing CSF leakage in the context of Marfan syndrome:

Clinical Evaluation:

Symptom Assessment: The process often begins with a thorough assessment of the individual's symptoms. Common symptoms of CSF leakage may include:

Persistent headaches, especially when upright and relieved when lying down. Clear nasal discharge (rhinorrhea) or drainage from the ear (otorrhea). Nausea or vomiting. Neck stiffness. Visual disturbances. Medical History: The healthcare provider will take a detailed medical history, including any known medical conditions such as Marfan syndrome and previous head or spinal surgeries or injuries.

Physical Examination:

Neurological Examination: A neurological examination may be conducted to assess cranial nerve function, reflexes, and signs of increased intracranial pressure, which can be associated with CSF leakage. Imaging Studies:

MRI (Magnetic Resonance Imaging): MRI of the head and spine may be performed to visualize the brain, spinal cord, and surrounding structures. MRI can sometimes reveal structural abnormalities, such as dural tears or CSF fistulas, that could be causing the leakage.

CT Myelography: This specialized imaging technique involves injecting a contrast dye into the spinal fluid followed by a CT scan. It can provide detailed images of the spinal cord and CSF spaces, making it a valuable tool for detecting CSF leaks.

Lumbar Puncture (Spinal Tap):

A lumbar puncture may be performed to analyze the cerebrospinal fluid directly. This procedure involves inserting a thin needle into the spinal canal in the lower back to collect a sample of cerebrospinal fluid. The fluid can be examined for abnormalities, such as an elevated opening pressure (indicating increased intracranial pressure) or the presence of blood cells or markers of CSF

leakage. Nasal Endoscopy: In cases of suspected CSF rhinorrhea (nasal CSF leakage), a nasal endoscopy may be performed. This involves inserting a thin, flexible tube with a camera into the nasal passages to visualize any leakage or defects in the skull base.

Confirmatory Tests: In some cases, confirmatory tests, such as a beta-2 transferrin assay, may be performed on the nasal or ear drainage to definitively identify the presence of CSF.

Once CSF leakage is diagnosed, the next steps typically involve determining the cause of the leak and planning appropriate treatment. Treatment may involve conservative measures (e.g., bed rest, hydration) for mild cases or surgical intervention to repair the leakage source for more severe cases.

Individuals with Marfan syndrome should receive comprehensive medical care from healthcare providers familiar with the condition, as they may be at a higher risk of certain complications, including CSF leakage. Early diagnosis and appropriate management are crucial for the well-being of affected individuals.

The prevalence of radiological evidence of a temporal bone defect among patients with MFS is high. This is a new, important, and potentially life-threatening association with the syndrome [1)]

Case reports

An 11-year-old boy, with medical history of Marfan syndrome, with orthostatic headache and persistent vomiting (12 hours) following a fall on the sacrococcygeal region. Magnetic resonance showed extradural fluid collections at dorsal and lumbosacral levels, compatible with CSF leak. The condition was resolved with treatment, but the patient had two new episodes during the follow-up period. Thus, an epidural blood patch was performed two years after the first episode. Although HIS is uncommon in children, it should be suspected in patients with orthostatic headache, particularly if the patient presents a connectivopathy. Few studies have assessed the management of HIS in paediatric age. The case presented here and the reviewed available literature provides further data for these type of cases [2)].

A 13-year-old female carrier of Marfan syndrome, clinically diagnosed according to the 2010 Ghent criteria, who consulted due to a 6-month history of severe orthostatic headache. Head magnetic resonance imaging (MRI) showed multiple signs of intracranial hypotension, while whole-spine MRI showed dural ectasia that caused the thecal sac dilation and subsequent remodeling of vertebral bodies, especially the sacral ones. Treatment with an autologous epidural blood patch was administered with good clinical response.

Dural ectasia, frequent in Marfan syndrome, is a predisposing cause of cerebrospinal fluid (CSF) leakage, which could cause orthostatic headache secondary to intracranial hypotension syndrome (IHS). [3)].

Two female patients presented with postural headaches. Magnetic resonance imaging revealed multiple leakages of CSF and both patients had a genetically confirmed diagnosis of Marfan syndrome. The initial symptomatic treatment did not result in a significant relief of the headaches. Epidural blood patching was performed and the intervention was successful in both patients. Finally, the most important epidemiological, diagnostic and pathophysiological aspects are demonstrated and the therapeutic procedures are presented [4)]

A 36-year-old patient with Marfan's syndrome who presented with rhinorrhoea, occipital headache and vomiting. Physical examination revealed typical Marfan's syndrome features including dolicocephalous, mandibular micrognathia, tall stature, disproportionately long limbs and digits, and hypermobility of the joints. A high-resolution CT scan demonstrated pneumoencephalous, cerebrospinal fluid (CSF) filling the sphenoidal sinus, and a small bone defect of the clivus. Surgery performed through a transsphenoidal approach revealed the sphenoid sinus to be filled with CSF and a small fenestration in the clivus. The arachnoid diverticulum and the fenestration were repaired and covered with a graft of abdominal fat. In this patient, a deficiency in bone development associated with Marfan's syndrome gave rise to a clival fenestration and a transclival CSF fistula. Although abnormalities of the spinal meningeal membranes have been reported in Marfan's syndrome, to our knowledge, this is the first report of a fistula located in the cranial base in this condition [5)]

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