Gorham-Stout disease of skull base

Lesions of the skull base are extremely rare and entail an even more devastating prognosis due to cervical spine instability and cerebrospinal fluid fistula. Due to the scarcity of this condition, the aim of a study was to give an overview of skull base Gorham-Stout disease and review the cases with such conditions reported in the literature.

In this case-based review, different aspects of skull base GSD are discussed, and a sample clinical case of GSD leading to cranial settling and rhinorrhea is presented. The characteristics, symptoms, and management of all English-language PubMed-reported cases were reviewed, and different features of presentation and methods of treatments were analyzed.

Based on the literature review, most of the cases encountered serious problems in the course of the disease. Meningitis/Cerebrospinal fluid fistula was detected in 12 of 26 collected cases, followed by hearing loss/tinnitus/otitis media in 10 cases, headache in 8, and neck pain/stiffness in 8 patients. Despite a variety of treatments, improvement was only observed in 8 of 26 collected cases. The reminders showed either stable condition or worsening and death.

All cases of GSD of the skull base should be evaluated for rhinorrhea/otorrhea and cranial settling, both of them being among the most life-threatening conditions. Since definite treatment, in order to stop disease progression, is sometimes impossible, symptomatic and supportive treatment should be started as possible. [1)].

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6 patients (5 males, 1 female) were included. The mean age at diagnosis was 3.5 years (range 0-10). Follow-up was of 5.2 years. Patients were divided into Naso-temporal (NT) and Vertebro-temporal (VT) groups following anatomical location. NT patients (4 patients) all had petrous defects extending anteriorly, including sphenoid, ethmoidal, and mandibular defects. They all had cerebrospinal fluid fistula (CSF) and recurrent meningitis (range from 3 to 7). Two of those patients had sequelae including deafness, paralysis, and epilepsy. VT patients (2 patients) all had temporal, occipital bone and cervical vertebrae defects. None had Cerebrospinal fluid fistulas but both died from medullar compression (preceded by tetraparesis in one case). Overall, five out of six patients had type I Chiari malformation. Interferon seemed to be the most efficient medical treatment. Surgery included petrectomy, endonasal surgery for Cerebrospinal fluid fistula management, and neurosurgery for medullar management but could not guarantee long-term effects.

The main issues in skull base defects are cerebrospinal fluid fistulas leaks and medullar compressions. Surgical treatment is necessary in both cases but can only be satisfactory if general medical treatment can stabilize the disease [2)].

Case reports

A 27-year-old man was diagnosed with GSD with the involvement of the maxillofacial bones and skull base. The patient developed SBO; LMS resulted from progressive osteolysis, and the patient died of an associated brainstem stroke. Careful follow-up with special emphasis on the early detection of intracranial complications is critical in patients presenting with progressive GSD with involvement of the skull base [3)]

A two-year-old female child with radiological signs mimicking those of raised intracranial pressure is discussed. The differential diagnosis consists of skull base tumors, meningitis, osteomyelitis of the skull base, congenital hydrocephalus, and congenital syndromes involving the skull base. Pathologically it can be very difficult to differentiate it from lymphangioma of the bone. Difficulty in establishing the diagnosis is discussed along with the failure of radiotherapy and palmidronate therapy to cause the arrest of the disease process and failure of surgery to provide stabilization. Girn et al. described the course of the disease in this child over the period of last eight years. This was the youngest case of Gorham's described so far [4].

A 25-year-old woman with Chiari I malformation associated with Gorham's syndrome presented with aggressive paresthesia following bacterial meningitis. Axial magnetic resonance imaging (MRI) and computed tomography (CT) cisternography revealed Cerebrospinal fluid fistula in the right petrous apex. A presyrinx state was diagnosed based on the clinical symptoms and MRI findings. With the resolution of bacterial meningitis, the spinal edema and tonsillar ectopia also improved. Surgical repair of the Cerebrospinal fluid fistula was performed by an endoscopic endonasal transsphenoidal approach to prevent recurrence of meningitis. The postoperative course was uneventful.

Skull base osteolysis in Gorham's syndrome may induce Chiari I malformation and Cerebrospinal fluid fistula. We should pay attention to the acute progression of clinical symptoms because Gorham's syndrome may predispose to the development of Chiari I malformation and may be complicated by Cerebrospinal fluid fistula [5].

A case of spinal and skull base Gorham's disease that was reversed by radiation therapy administered while the spine was supported by a halo-vest [6].

1)
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