Gorham Stout syndrome

Gorham's disease is a rare osteolytic disorder characterized by progressive resorption of bone and replacement of osseous matrix by a proliferative non-neoplastic vascular or lymphatic tissue. A standardized treatment protocol has not yet been defined due to the unpredictable natural history of the disease and variable clinical presentations.

Case series

6 patients (5 males, 1 female) were included. 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 cerebro-spinal fluid leak (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 CSF leaks 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 CSF leak management and neurosurgery for medullar management but could not guarantee long-term effects.

Main issues in skull base defects are CSF leaks and medullar compressions. Surgical treatment is necessary in both cases but can only be satisfactory if general medical treatment can stabilise the disease [1)].

No single treatment has proven to be superior in arresting the course of the disease. Trials have included surgery, radiation and medical therapies using drugs such as calcium salts, vitamin D supplements and hormones. We report on our advantageous experience in the management of this osteolyic disorder in a case when it affected only the skull vault. A brief review of pertinent literature about Gorham's disease with skull involvement is provided.

Gorham-Stout syndrome in the spine is extremely rare, and there is no standard curative management thus far. The objective of this article is to report a very rare case of Gorham-Stout syndrome of the lumbar and sacral spine with chylothorax and chyloperitoneum successfully treated by combination of vertebroplasty with cement augmentation and medication treatment. We described the clinical characteristics and postoperative therapy of the patient, and reviewed all of the published cases of Gorham-Stout syndrome of the lumbar and sacral spine.

A 31-year-old man presented with increasingly serious abdominal distention and back pain. MRI showed massive bony destruction of the spine and pelvis. CT and ultrasonography demonstrated massive ascites and mild hydrothorax.

We believe this is the first report of a case of Gorham-Stout syndrome with both chylothorax and chyloperitoneum.

Chest and abdominal cavity puncture was performed for symptomatic relief and the test results

confirmed chylothorax and chyloperitoneum. Tissue biopsy and percutaneous vertebroplasty at L5 were performed and the postoperative pathology together with symptoms and examinations were reported to be consistent with Gorham-Stout syndrome. Subsequently, we administered combination medical treatment consisting of interferon-α-2b, zoledronic acid and calcitriol.

At the 1-year and 2-year follow-up visit, he had nearly full complete remission and reported palliative back pain. Moreover, the amount of pleural and peritoneal fluid was successfully reduced gradually.

Vertebroplasty by cement augmentation may be a treatment option for patients with Gorham-Stout Syndrome in the spine who cannot undergo appropriate surgery or decline open surgery. This represents a safe and minimally invasive approach to sustainably relieve pain and stabilize vertebral bodies with Gorham-Stout syndrome in the spine [2)].

Case reports

A 25-year-old Caucasian male presented with a skull depression over the left fronto-temporal region. He noticed progressive enlargement of the skull defect associated with local pain and mild headache. Physical examination revealed a tender palpable depression of the fronto-temporal convexity. Conventional X-ray of the skull showed widespread loss of bone substance. Subsequent CT scans showed features of patchy erosions indicative of an underlying osteolysis. MRI also revealed marginal enhancement at the site of the defect. The patient was in need of a pathological diagnosis as well as complex reconstruction of the afflicted area. A density graded CT scan was done to determine the variable degrees of osteolysis and a custom made allograft was designed for cranioplasty preoperatively to allow for a single step excisional craniectomy with synchronous skull repair. Gorham's disease was diagnosed based on histopathological examination. No neurological deficit or wound complications were reported postoperatively. Over a two-year follow up period, the patient had no evidence of local recurrence or other systemic involvement.

A single step excisional craniectomy and cranioplasty can be an effective treatment for patients with Gorham's disease affecting the skull vault only. Preoperative planning by a density graded CT aids to design a synthetic bone flap and is beneficial in skull reconstruction. Systemic involvement is variable in this patient's population

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2)
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