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Clinical vignette

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A case of vanishing skull: Gorham's disease

Gorham's disease (GD) is a rare form of lymphangiomatosis associated with profound osteolysis. Outcome depends on the site affected. Small limb lesions can cause fractures, while spinal involvement or chylothorax associated with rib lesions can be life threatening. There is no consensus regarding treatment. Surgical resection and/or radiotherapy are most commonly used, but often unsuccessfully [1].

We report the case of an otherwise healthy female who presented at the age of 40 years with a small, painless skull indentation. Over the following 6 years the indentation evolved into an 11 $\times$ 11 cm cranial vault defect. There were no other abnormal findings on examination. Investigations were negative for malignancy, infection, biochemical and immunological abnormalities. CT demonstrated a defect in the occipital region (Fig. 1) but no abnormalities elsewhere in the skeletal system. A skull and dural biopsy confirmed GD.

Radiotherapy proved ineffective. Monthly 5 mg i.v. infusion of zoledronic acid was therefore commenced, with weekly s.c. pegylated IFN-α2b 35 μg. Clinical and radiological follow-up over 3 years demonstrated suppression of the osteolytic process with stabilization of the calvarial defect.

Lymphangiomatoses are non-malignant processes involving abnormal lymphatic proliferation. In GD, an increased number of osteoclasts are often present, which is thought to be promoted by elevated levels of IL-6 [2]. While IFN and bisphosphonate can provide effective therapy, cases with poor response may benefit from trials of anti-cytokine therapy or other biologic modulators in rheumatologic research settings.

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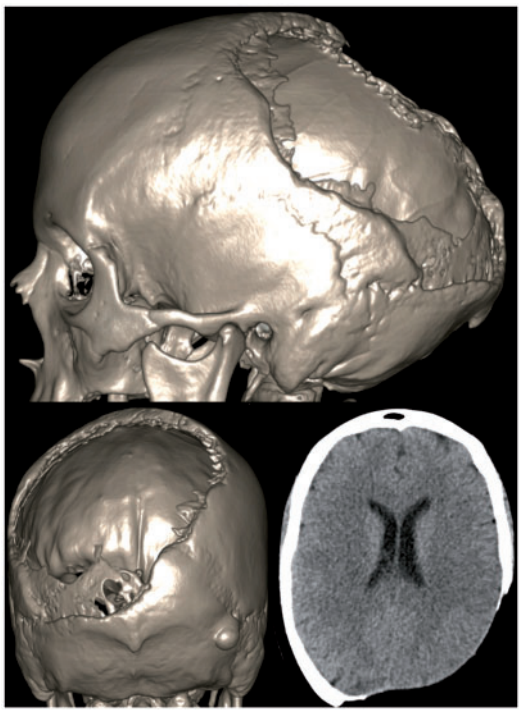
FIG. 1 CT head with 3D rendering of the skull defect



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