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MR of Multiple Cranial Neuropathies in a Patient with Camurati-Engelmann Disease: Case Report

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Camurati-Engelmann disease (progressive diaphyseal dysplasia) is a rare disorder of bone metabolism characterized by hyperostosis along the diaphysis of long bones [1–3]. It may also involve the skull and other membranous bone [1–3]. Cranial nerve deficits can be the most debilitating feature of this disease [3–7]. A narrowed foramen magnum can lead to brainstem compression and death [4, 6, 8]. Radiographic, CT, and MR findings demonstrate severe hyperostosis and sclerosis of the skull with almost complete obliteration of basal foramina and orbital apices. MR can directly demonstrate the course of the cranial nerves with respect to the stenotic foramina.

Case Report

A 46-year-old woman with Camurati-Engelmann disease presented at age 2 with a delay in walking. A biopsy of the left tibia at that time suggested the diagnosis. She developed the radiographic and clinical manifestations of Camurati-Engelmann disease over the next several years, including bone pain, wobbling gait, and cranial nerve deficits. At age 26, an operation to decompress her right seventh nerve failed, leaving her deaf. At age 38, a second attempt to decompress her right seventh nerve was also unsuccessful. At the ages of 43 and 45 she underwent right tarsorrhaphy and later revision, respectively. She has been treated with steroids since the age of 37.

Currently, the patient suffers from numbness of the face and scalp, bilateral facial droop (right greater than left), diminished vision and hearing, difficulty talking, chewing, and swallowing, joint contractures, difficulty walking, and four-pillow orthopnea. Physical examination reveals prominence of the facial bones and a large head. Her speech is difficult to understand, owing to the lack of excursion of her lower jaw, and she has bilateral proptosis (left side greater than right), decreased extraocular muscle motion, and diminished vision. She has diminished fifth nerve function bilaterally, absent right seventh nerve function, diminished left seventh nerve function, decreased gag reflex, decreased hearing on the left, deafness on the right, and two-finger opening of her oral cavity.

Muscle strength is abnormal, with decreased pronator strength of the right upper extremity, decreased hand grip bilaterally, wasting of the right dorsal interosseous muscle, bilateral elbow flexion contracture, bilateral hip flexion, right knee flexion, and bilateral dorsiflexion

of both ankles. She has decreased range of motion of the entire spine, an unsteady wide-based gait, and absent deep tendon reflexes.

Laboratory tests demonstrate normal hematocrit. Alkaline phosphatase is elevated at 434 IU/l, calcium is 9.4 mg/dl, and phosphorous is 2.5 mg/dl. Pulmonary function tests demonstrate restrictive lung disease; nerve conduction tests reveal a mild right ulnar neuropathy.

Plain radiographs of the skull show severe calvarial hyperostosis and skull base sclerosis (Fig. 1A). Thoracic spine plain radiographs demonstrate posterior element and posterior vertebral body sclerosis. Cervical spine plain films demonstrate ankylosis and bilateral encroachment of all intervertebral foramina. Symmetrical diaphyseal hyperostosis involves all of her extremities. A CT scan of the head demonstrates dense hyperostosis and sclerosis of the skull with significant skull base foramina narrowing (Fig. 1B). MR of the brain and skull base shows a mantle of signal void around the brain involving the skull base and the calvaria on short and long TR sequences. There is encasement of the otic capsule and middle and external ear by hyperostotic bone. There are no readily identifiable skull base foramina. The optic canals and optic foramina are narrowed bilaterally (Fig. 1C). MR demonstrates the course of the optic nerves and the seventh and eighth cranial nerves as they pass through the stenotic orifices (Fig. 1D). Short TR, short TE MR images before and after contrast administration show diffuse symmetrical meningeal enhancement and focal areas of intramedullary calvarial enhancement (Figs. 1E and 1F).

Discussion

Camurati-Engelmann disease, inherited as an autosomal-dominant trait, belongs to a group of craniotubular bone modeling diseases that include osteopetrosis and craniometaphyseal dysplasia [4]. It is a rare disorder of unknown cause with approximately 100 reported cases in the literature. Diagnosis is made clinically and radiographically, as there are no specific laboratory findings [1–3, 9–12]. Bone biopsy, in general, demonstrates increased bone growth of mature and immature elements [1, 2, 9]. The clinical triad of bone pain, extremity or muscle weakness, and wobbling gait are frequently cited as the most common features [1, 2, 3, 9, 11].

In advanced cases, cranial nerve deficits can be the most debilitating aspect of the disease, leading to blindness, hear-

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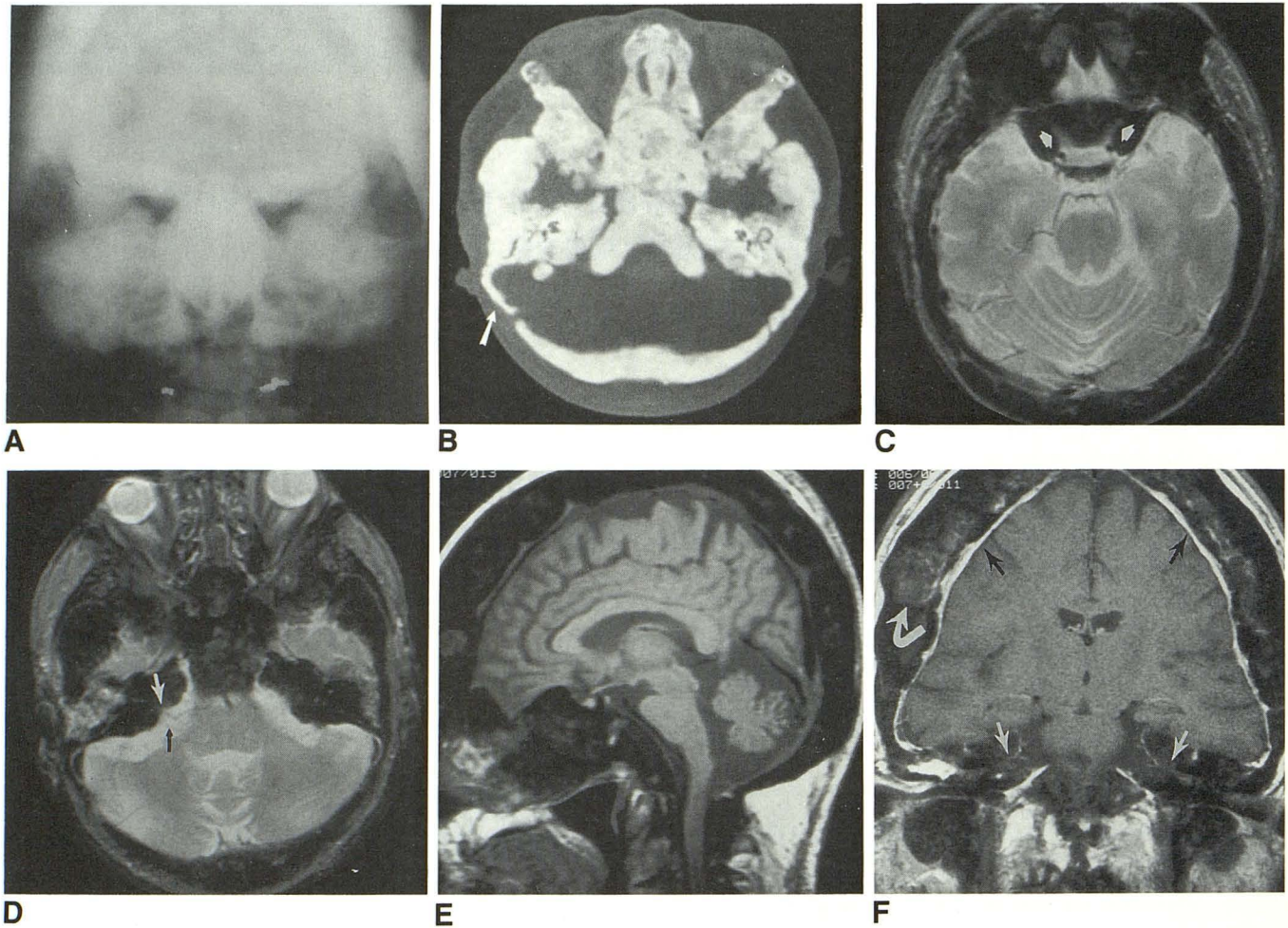


Fig. 1.—46-year-old woman with Camurati-Engelmann disease.

A, Anteroposterior radiograph of skull shows severely thickened and sclerotic facial bones, orbits, and calvaria.

B, Axial CT scan of skull base at level of otic capsule shows diffuse hyperostotic bone. Middle ear cavity has been completely encased by sclerotic bone. Retromastoid surgery was performed to decompress right seventh cranial nerve (arrow).

C, Axial MR image, 2500/80/1, at level of optic chiasm shows bilateral narrowed optic canals (arrows).

D, Axial MR image (2500/80) at level of temporal bones shows cisternal segments of seventh and eighth cranial nerve complex (black arrow) entering stenotic porus acousticus (white arrow). Hyperostotic cortical bone appears as areas of signal void.

E and F, Midline sagittal unenhanced MR image (500/25/2) (E) and postcontrast coronal MR image (500/25) (F) at level of stenotic internal auditory canals (straight white arrows) show diffuse meningeal enhancement (black arrows) and foci of enhancement within medullary portion of calvaria (curved white arrow), thought to represent areas of hematopoiesis. Note postoperative right temporal bone.

ing loss, facial paralysis, and numbness [3–7]. These symptoms are caused by hyperostosis and sclerosis of the skull base, with stenosis of the basal foramina and orbital apices [3, 5–7, 13], which can lead to compression of the cranial nerves directly [3, 5, 13] or of their blood supply, producing venous occlusion and neural edema, eventually leading to occlusion of arterial supply to the nerve [3, 5]. Some patients present with increased intracranial pressure [3, 5, 9], which may be due to decreased venous outflow [5, 6]. The cranial vasculature may be further compromised by arterial wall thickening as part of a generalized vasculopathy seen in many patients with this disease [8, 9, 13]. A few patients have suffered from brainstem compression which leads to herniation and death due to foramen magnum stenosis [4, 6, 8].

In our patient, recent contrast-enhanced MR examinations showed calvarial marrow enhancement and diffuse meningeal

enhancement. Even though the patient has had decompressive surgery in the posterior fossa, we speculate that this enhancement most likely represents meningeal reaction due to increased bone turnover and metabolism.

Treatment of this disease is palliative. Attempts at decompressive surgery of the cranial nerves have been successful albeit difficult because of the thickened and sclerotic bone [3–5, 8, 13]. Although there are periods of quiescence, this is a progressive disease, and the hyperostotic bone usually returns [3, 13]. Decompressive surgery to relieve increased intracranial pressure has also been successful in alleviating cranial neuropathies [6], suggesting further that not only direct compression on the nerve itself but also compression on the venous drainage may be important etiologic factors. Steroids have been helpful [5, 10, 12] in diminishing the elevated intracranial pressure [6] and in alleviating extremity

bone pain [7, 10, 12]. However, there is no treatment to arrest the progressive nature of the disease [5, 7].

MR is as effective as CT in demonstrating hyperostotic bone. Subtle skull base foraminal narrowing may not be as well demonstrated on MR as on CT; however, the effects on specific cranial nerves (in particular cranial nerves two, seven, and eight, which are the most commonly involved in this disease) are better demonstrated by MR imaging, as these nerves can be imaged directly. Caution should be used in suggesting a malignant cause for diffuse meningeal enhancement in this case, as a benign reactive process due to increased metabolism (and blood flow) of overlying bone may be the etiologic factor. Irregular enhancement of the medullary portion of the calvaria most likely reflects intramedullary hematopoiesis.

In conclusion, MR is an excellent technique for evaluating the severity of the diseases that affect cranial bone modeling. This imaging method depicts the effect of these diseases on the cranial nerves and brainstem and may help guide further clinical management.

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