Short report

Fatal cerebellar herniation secondary to Camurati-Englemann’s disease

RICHARD K SIMPSON JR,* DUNCAN K FISCHER,* GARY K S E GALL,†
JAMES E ROSE*†

From the Departments of Neurosurgery* and Pathology,† Baylor College of Medicine, Houston, Texas, USA

SUMMARY Suboccipital craniotomy and cervical laminectomy were performed in a patient with Camurati-Englemann’s disease to relieve symptoms of medullary compression. In spite of surgical decompression, the patient expired on the fourth postoperative day from cerebellar herniation.

Camurati-Englemann’s disease, also known as progressive diaphyseal dysplasia, is an unusual disorder of bone metabolism.1 The lesions are primarily characterised by symmetrical, fusiform hyperostosis and sclerosis of long bones and of the cranial base.2 Pain in the lower extremities, general muscle weakness, and a waddling gait are the most common neurological disturbances.3 Cranial nerve deficits can occur with involvement of the skull.4 Surgical treatment has included decompression of the optic or auditory nerves.2 4 Rarely, a patient will have signs and symptoms of increased intracranial pressure (ICP).5 Both the aetiology and surgical management of increased ICP in Camurati-Englemann’s disease have yet to be clarified.

Case reports

A 25 year old chemical engineer with Camurati-Englemann’s disease was admitted to the Methodist Hospital after a 3 week history of progressive occipital headaches accompanied by neck and shoulder stiffness. He also complained of bilateral hand numbness and difficulty with lower extremity coordination. He occasionally experienced dysphagia, diplopia and loss of taste sensation, yet had no nausea or vomiting. His past history was unremarkable. His family history was negative for congenital bone disorders.

Physical examination revealed a severely macrocephalic man with a prominent maxilla and mandible, and hypertelorism. He had a flattened, broad nasal bridge and large turbinates. His external auditory canals were nearly occluded by bony overgrowth. He had broad, thick extremities and digits. Neurological examination revealed diminished auditory acuity bilaterally and weakness of facial muscles, and nystagmus on lateral gaze. He had a wide based gait, mild dysmetria, hyperactive deep tendon reflexes in his legs, and Babinski’s sign was present bilaterally. Fundi were normal.

Methods and results

Laboratory investigation revealed an alkaline phosphatase of 657 IU/100 ml (normal = 25–100 IU), a parathormone level of 59 pg/ml (normal = 230–630 pg/ml). Serum calcium, full blood count, urinalysis and electrocardiogram were normal. A chest radiograph was normal except for thickened ribs and clavicles. A metabolic bone survey showed a diffuse, symmetrical increase in cortical bone thickness with loss of intramedullary cavities. Plain radiograph skull, CT and cerebral angiography were performed but were difficult to interpret because of attenuation caused by thick, dense bone. Magnetic resonance imaging revealed diffuse cranial hyperostosis with no pneumatization of the paranasal or temporal sinuses (fig a). The cerebral hemispheres were normal. There was tonsillar and vermillion herniation caudal to C1.

A decompressive suboccipital craniotomy and Cl–C2 laminectomy were performed to decompress the posterior fossa and cervico-medullary junction. The operation was lengthy and technically difficult because of the thickness and density of the bone. Gross inspection of an intraoperative, nondecalcified specimen revealed apparently normal bone. A thick dense dural band between the arch of C1 and the foramen magnum was identified and divided. The medulla and spinal cord pulsed normally. As the dura was opened, the patient’s temperature rose sud-
Fig MRI of head and upper cervical spine (a). Sagittal image (TR = 0.4, TE = 25) reveals thickened skull, normal cerebrum, and midline cerebellar herniation. Axial image (TR = 2.8, TE = 105) reveals thickened skull and relatively normal ventricular system. Necropsy specimens (b). Extremely thickened cranium but normal cerebral surface. Shallow, thickened skull base with a suboccipital craniotomy defect.
Diaphyseal dysplasia

nerve
corticis deformans), line phosphatase and enostosis Schonberg's disease bone.7

decades.3

in childhood mented.24

variable is dysplasia thickening
diameter, and narrowed bones.6

Common features are diaphyseal dysplasia, Camurati-Englemann's disease,

Enzlen's disease osteosclerosis, Paget's disease osteoporosis, and Albers-Schonberg's diseases have increased ICP.2 10 The precise mechanism responsible for increased ICP has remained elusive for over a century.11 Several patients with these disorders have succumbed from sudden brainstem compromise.12 The surgical procedures that have been used to prevent herniation, have included ventricular shunting, bitemporal craniotomy, and suboccipital craniotomy.8 13 14 Bitemporal craniotomy and cranial nerve decompression have been reported to be less useful in some cases owing to the rapid regrowth of bone. Ventricular shunting has been mildly successful if hydrocephalus is present.14 Suboccipital decompression is frequently performed in order to obviate cerebellum and brainstem impaction into the foramen magnum.13 14 These operations are technically difficult owing to the thickness and density of the skull.5 12 14 Successful surgical outcome is temporary and limited, at best. After operation, patients may suffer fatal progression of increased ICP.13 Although our case was complicated by malignant hyperthermia, known to cause an increase in ICP,15 there was a fatal outcome from progressive herniation despite extensive bony decompression.

Cranial surgery has a limited role in treatment of sclerosing bone dysplasias. A beneficial surgical procedure for treating increased ICP has yet to be devised. Although these diseases are individually rare, taken together they constitute a significant number of patients. Systemic therapy aimed at halting or reversing abnormal bone production is required. Until such treatment becomes available, operative intervention should be exercised with caution.
References

Fatal cerebellar herniation secondary to Camurati-Englemann's disease.

R K Simpson, Jr, D K Fischer, G K Gall and J E Rose

*J Neurol Neurosurg Psychiatry* 1988 51: 1349-1352
doi: 10.1136/jnnp.51.10.1349

Updated information and services can be found at:
http://jnnp.bmj.com/content/51/10/1349

**Email alerting service**

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

**Notes**

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/