Skull Base Manifestations of Camurati-Engelmann Disease

Matthew L. Carlson, M.D.1, Charles W. Beatty, M.D.1, Brian A. Neff, M.D.1, Michael J. Link, M.D.2, Colin L. W. Driscoll, M.D.1

1Department of Otolaryngology-Head and Neck Surgery, 2Department of Neurologic Surgery, Mayo Clinic School of Medicine, Rochester, MN

ABSTRACT

Camurati-Engelmann Disease (CED), also known as progressive diaphyseal dysplasia, is a rare inherited disorder characterized by progressive symmetric distal diaphyseal deformities of the long bones. The disease is caused by mutations in the BMP15 gene, which results in increased bone remodeling and increased bone density. CED can result in a variety of symptoms, including osteopetrosis, craniodiaphyseal dysplasia, and Van Buchem disease. Since the first description in 1928, over 200 cases have been described, with the diagnosis being confirmed by radiographic findings. The disease is characterized by radiographic evidence of skull base involvement, which is quite variable and results from bony overgrowth leading to foraminal stenosis and diminished cranial vault volume resulting in neurovascular compromise and increased intracranial pressure (ICP).

Objectives: Surgical decompression remains the primary management of progressive symptomatic skull base manifestations. It is typically necessary with cranial bone changes, minimal cranial neurovascular compromise, and subsequent stimulation of osteoblastic bone formation and suppression of osteoclastic resorption.

Methods: A retrospective chart review from 1968-2008 was performed using the electronic medical record and all patients diagnosed with CED were included. Of these, eight (66%) were found to have radiographic evidence of skull base thickening with concurrent cranial signs and symptoms of disease. The average age of onset was 24 years (15-36). Symptomatic surgical intervention (two transcranial, three transoral) was performed for neurovascular compromise in 17% (two) of patients, and for foraminal stenosis and increased ICP in 25% (two) of patients. A total of six decompression surgeries were performed for aggressive disease and one additionally received cochlear implantation for profound bilateral SNHL.

Results: In the last 40 years twelve patients carrying the diagnosis of CED were evaluated and treated at our institution. These included four males and eight females with a mean age of symptom onset at 9.4 years (3-30). Of these, eight (66%) were found to have radiographic evidence of skull base thickening with concurrent cranial signs and symptoms of disease. The average age of onset was 24 years (15-36). Symptomatic surgical intervention (two transcranial, three transoral) was performed for neurovascular compromise in 17% (two) of patients, and for foraminal stenosis and increased ICP in 25% (two) of patients. A total of six decompression surgeries were performed for aggressive disease and one additionally received cochlear implantation for profound bilateral SNHL.

Conclusions: CED is a rare disease that results in symptomatic cranial base hypotranslucency in less than one fourth of patients. In asymptomatic and mild forms of disease, patients should be followed with serial examination, audiometric testing and imaging. Those with more symptomatic skull base involvement benefit from wide decompression of involved neurovascular structures.

DISCUSSION

Despite that over 85% of patients with CED will have radiographically confirmed skull base involvement, less than 25% experience cranial nerve dysfunction. Changing to the symptomatic but often irregular cranial base to the non-symptomatic, progressive cranial bone changes may not reflect subtle unilateral hearing loss, facial nerve paresis, or vision loss, while others may experience gradual or fluctuating unilateral or bilateral partial or complete visual field loss. Increased intracranial pressure (ICP), which may occur in the anterior and middle fossae, can be caused by an arachnoid granulation, a subdural hematoma, or a neoplasm. Unfortunately, there remains no known pharmacologic or surgical strategy for disease reversal. In contrast to lower extremity progression, surgical therapy has not been successful in treating skull base disease. Surgeries with aggressive wide decompression are technically challenging but remain the only means to preserve cranial nerve function for patients with advanced disease.

REFERENCES


CONTACT

Matthew L. Carlson, M.D.
Mayo Clinic, Dept. of Otolaryngology
Phone: 507-255-5123