CASE REPORT

A Painless Retroauricular Mass in A Patient with Myotonic Dystrophy: Computed Tomographic Documentation of the Bone Changes that Occur in the Skull Base

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ABSTRACT—A 16-year-old male with a history of neonatal myotonic dystrophy had a painless enlarging left retroauricular mass. Computed tomography showed an enlarged frontal sinus, thickened apparently normal bone in the anterior left clinoid process, dorsum sellae, and all portions of the temporal bones. In particular, there was bone production in the left retroauricular region and along the margins of the left internal auditory canal. The left temporomandibular joint was also dislocated. This is the first case report to document the dynamic aspect of the bone changes in this dystrophy.

Myotonic dystrophy is one of the most common of the dystrophies, with an incidence of 2.5 to 5 per 100,000 people, and it is estimated that between 1/20,000 and 1/7,500 people carry the gene mutation for the disease. It is an inherited autosomal dominant disorder with variable transmission. The abnormality has been mapped to the q13.3 band of chromosome 19, and when the disease is inherited from the mother it tends to be more severe. Paternal transmission is rare.¹⁻³ Typically the disease presents in the second to fourth decades of life; however, children can also be affected. Commonly the small muscles of the hands and the extensor muscles of the forearms are the first to atrophy. However, in other patients, ptosis and facial muscle atrophy are the initial findings. Usually affected are the masseter and sternocleidomastoid muscles, leading to a deformed mandible and a "swan neck." The association of frontal baldness and a wrinkled forehead give the patient a distinctive appearance. Virtually all muscles can be affected, leading to a variety of symptoms, and most patients are confined to either a wheelchair or bed by the age of 15 to 20 years. Myotonia may precede muscle weakness, and most patients have opacities of the lens of the eye. Associated with this disease is a generalized

hyperostotic component that affects the skull and skull base. Only the calvarial disease has been previously documented with computed tomography (CT).^{4,5} The purpose of this article is to describe a patient with known myotonic dystrophy who presented with an enlarging painless retroauricular mass. The computed tomographic (CT) findings of the skull base disease are reviewed.

CASE REPORT

The patient is a 16-year-old boy with neonatal myotonic dystrophy who was first referred for otological evaluation 2 years prior to the current studies. At that time, he had copious bilateral purulent otorrhea. Following a course of systemic antibiotics (amoxicillin/clavulanate) and topical preparation (tobramycin/dexamethasone drops), the otorrhea resolved. The left tympanic membrane was intact with a middle ear effusion, while the right ear had a posterior retraction pocket filled with squamous debris. There was no external abnormality of the mastoid at that time. Audiometry was performed and demonstrated a bilateral high-frequency

sensorineural hearing loss with a low-frequency conductive component. Following repeated microscopic cleaning and topical therapy, the right ear cleared. A left ear effusion persisted, and a pressure equalization tube was placed in the left side with resultant improvement in hearing.

Approximately 1 year after his initial presentation to the Ear, Nose, and Throat service, the patient's myotonic dystrophy progressed to the point of requiring intubation for a pneumonia and respiratory failure. He had a tracheotomy and subsequently was discharged on a home ventilator for intermittent support. Soon after this,

a left firm mastoid/retroauricular prominence was first noted to be slowly enlarging. Given the expectation of imminent end-stage respiratory failure, this mastoid prominence was not pursued and supportive measures were continued. However, the patient's condition stabilized and approximately 2 years after presentation a CT scan was obtained that showed thickened bone in the mastoid portion of the left temporal bone. There was also thickened bone in the left anterior clinoid process, in the dorsum sellae, along the posterior aspect of the petrous pyramid (especially near the margins of the internal auditory canal), and the petrous tip. Similar,

Figure 1. Axial CT scans viewed at wide windows. **(A)** The most cranial image, showing a very large frontal sinus and a small sella turcica. There is also thickened bone in the left anterior clinoid process, the dorsum sellae, the left petrous tip, the dorsal surface of the left petrous pyramid, and the lateral surface of the right temporal bone (arrowheads). **(B)** More caudal than that in A, showing in addition productive bone along the dorsal surfaces of both petrous pyramids, in the right petrous apex, and especially along the margins of the left internal auditory canal. The canal itself was not narrowed. There is also productive bone in the left retroauricular region (arrowheads). **(C)** The most caudal image, showing the productive bony changes in the left retroauricular region (arrowhead) and near the left temporomandibular joint glenoid fossa (arrow). The left mandibular head (m) is slightly dislocated caudally out of the glenoid fossa.

though less pronounced, changes were present in the right temporal bone. In addition on the left side, there was productive bone in the glenoid fossa region, and there was dislocation of the temporomandibular joint. The patient also had a huge frontal sinus and a small sella turcica (Fig. 1). No surgical management of the bone disease was undertaken.

DISCUSSION

Previous studies have discussed the changes that may occur in the skull and skull base of patients with myotonic dystrophy. These changes include thickening of the calvarium, a small sella turcica, and large frontal sinuses.³⁻¹⁰ Less often there can be craniokypnosis, a small nasal angle, narrowing of the internal auditory canals, hypotelorism, enlargement of the mandible, and subluxation of the temporomandibular joints.^{1,3} Most of the studies reporting these findings were based on plane films of the skull,⁶⁻¹⁰ and in the one report that utilized CT the authors stated that they thought that the bones of the skull base were not involved by the bone thickening process that affected the calvarium.⁴ However, this was disputed by others, who noted the occurrence of hyperostotic bone in the clivus and even lytic and nonexpansile sclerotic changes in the temporal bone.^{11,12} In our patient, there are multiple areas of bony overgrowth that affected the mastoid processes, the petrous pyramids, the temporal bone squamae, the left glenoid fossa with temporomandibular joint dislocation, the clivus, left anterior clinoid process, and the dorsum sella. Although most of the changes were bilateral, the left side was more affected in our patient. For the most part, the areas of expanded bone had a normal intact cortex with what appeared to be a normal trabecular pattern. There was no actual hyperostotic bone in the sense that the bone was dense or sclerotic except in the area immediately around the otic capsule, where the dense bone of the capsule may have contributed to the overall increased density of the bone in this region. These osseous changes were dynamic in that clinically the retroauricular fullness was enlarging. There is as yet no known cause for these bony changes. Although there was no ac-

tual narrowing of the internal auditory canals in our patient, the location of the productive bone appeared to be so positioned as to soon encroach upon these canals. Our patient also had a very large frontal sinus and a small sella turcica.

This report documents with CT that bone changes associated with myotonic dystrophy can occur in the skull base as well as in the calvarium and that the previously reported "hyperostotic" bone appears for the most part on CT to be productive, but otherwise normal bone. Our patient also demonstrates that these bony changes are dynamic and can cause a clinically noted "enlarging mass."

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