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Decompression of the internal auditory canal via the retrosigmoid approach in a patient with Camurati-Engelmann disease: illustrative case

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BACKGROUND Camurati-Engelmann disease (CED) is a rare condition characterized by hyperostosis of the long bones and skull base. Symptoms include contractures and pain in affected extremities but can also include manifestations of cranial hyperostosis such as intracranial hypertension, Chiari malformation, exophthalmia, frontal bossing, and several cranial neuropathies due to cranial foraminal stenosis.

OBSERVATIONS This report describes a 27-year-old patient with suspected CED who developed progressive intermittent facial nerve paresis, hemifacial spasms, and a decrease in hearing. There were no symptoms of increased intracranial pressure or vertigo. Radiological evaluation showed a significant thickening of the skull base with serious bilateral internal auditory canal stenosis. Because of the progressive nature of the aforementioned cranial neuropathies in combination with the correlating severe radiological compression, a surgical decompression of the facial nerve and vestibulocochlear nerve was performed via a retrosigmoid approach with intraoperative monitoring. Postoperative facial nerve function was intact. Hearing and vestibular function were unchanged. There were no more episodes of facial nerve palsy or spasm.

LESSONS To the authors' knowledge, this is the first report to describe decompression of the internal auditory canal via a retrosigmoid approach for symptomatic facial and cochlear nerve compression in a patient with CED.

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KEYWORDS Camurati-Engelmann disease; hyperostosis; internal auditory canal; cranial nerve palsy; retrosigmoid approach

Camurati-Engelmann disease (CED) is a rare condition characterized by hyperostosis of the long bones and skull. It is classified as a progressive craniodiaphyseal dysplasia within the group of sclerosing bone dysplasias.[1] Camurati[2] and Engelmann[3] were, separately, among the first to describe the condition in the early 1920s. It is also known as "progressive diaphyseal dysplasia," which emphasizes the pathophysiological epifocus. The disease mainly affects the diaphysis of the long tubular bones.

In addition to specific clinical features and hypersclerotic radiological abnormalities, in most cases, molecular genetic testing shows a mutation in the *TGFB1* gene located on chromosome 19q13.[4]

Symptoms can comprise musculoskeletal and/or neurological manifestations. Musculoskeletal symptoms mostly consist of muscle weakness, fatigue, contractures, and pain. Neurological symptoms can be due to hyperostosis of the skull with a decreasing size of the cranial vault and secondary intracranial hypertension. Another, and perhaps more important, mechanism is hyperostosis of the skull base with foraminal stenosis and secondary cranial neuropathies.[5]

Illustrative Case

The initial presentation of our 27-year-old patient consisted of gait disturbances in early childhood with a progressive course, eventually leading to orthopedic consultation and radiographic evaluation of the skeleton. Radiographs showed hyperostosis of the bilateral humerus, femur, and tibia with uneven cortical thickening of diaphysis and, furthermore, a distinct sclerosis of the skull base. There was no involvement of facial bones, clavicles, scapulae, ribs, or other short tubular bones. There were no significant hematological or hormonal problems.

This specific combination of clinical and radiological manifestations raised the suspicion for a bone dysplasia, more specifically CED. Genetic testing did not show a mutation in the gene encoding *TGFB1*.

ABBREVIATIONS CED = Camurati-Engelmann disease; CT = computed tomography; IAC = internal auditory canal.

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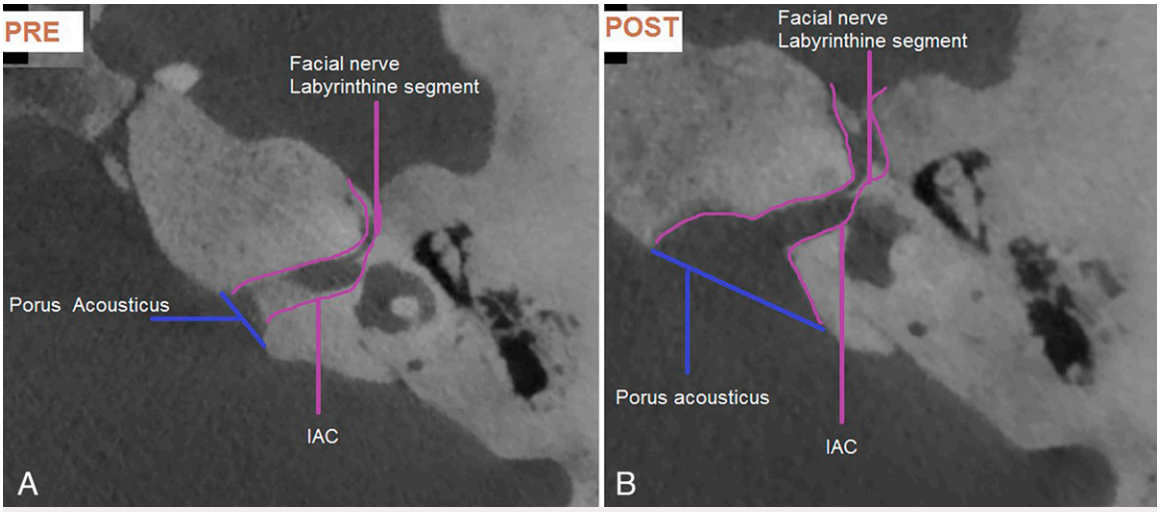

FIG. 1. Axial cone-beam CT at the level of the IAC. **A:** Preoperative left IAC. **B:** Postoperative left IAC.

After the initial diagnosis in 2005, our patient had multiple routine audiograms and ophthalmic evaluations. In the first years, there were no significant auditory deficits, and besides an asymptomatic exophthalmia, there were no visual deficits. The weakness in the proximal part of the legs was progressive, though not yet invalidating.

In 2016, in addition to the aforementioned symptoms, she developed intermittent left peripheral facial nerve palsy, left-sided facial spasm, and progressive left-sided hearing loss. The latter new symptoms all responded well to corticosteroid therapy. The episodes of facial weakness and spasm were nevertheless more frequent and lasted longer. A typical episode of facial palsy consisted of a gradual left-sided peripheral facial-nerve paralysis up to House-Brackmann grade 2 (slight) and lasted about 2 days in the beginning to full recovery after initiation of corticosteroid therapy. Later in the clinical course, the partial peripheral facial-nerve paralysis was more pronounced, with a House-Brackmann grade 3 (moderate), and it lasted longer, up to 4 weeks to full recovery after corticosteroid therapy. The spasms consisted of involuntary muscle contractions in the left side of her face, usually lasting a few hours to a few days. There were no symptoms of increased intracranial pressure or vertigo.

On examination, in an interval between episodes of facial palsy or spasm, there was a marked bilateral exophthalmia with normal eye movements and vision. There was no facial weakness and no vestibular problems. Tympanic membranes were normal. Hearing on the left was diminished. There was no dysphagia and there was a normal tongue protrusion. There was no significant sensorimotor deficit in the extremities, although there was a significant gait disturbance mainly due to arthralgia and upper leg pain in the left leg. A Romberg's test did not show significant instability. An audiogram confirmed left-sided high-frequency sensorineural hearing loss. A cranial computed tomography (CT) scan showed significant thickening of the skull, most prominently in the skull base with serious stenosis of the bilateral internal auditory canal (IAC) (Fig. 1A).

Because of the progressive nature and frequency of the cranial neuropathies in combination with the correlating severe radiological compression, we decided to perform a surgical decompression of the

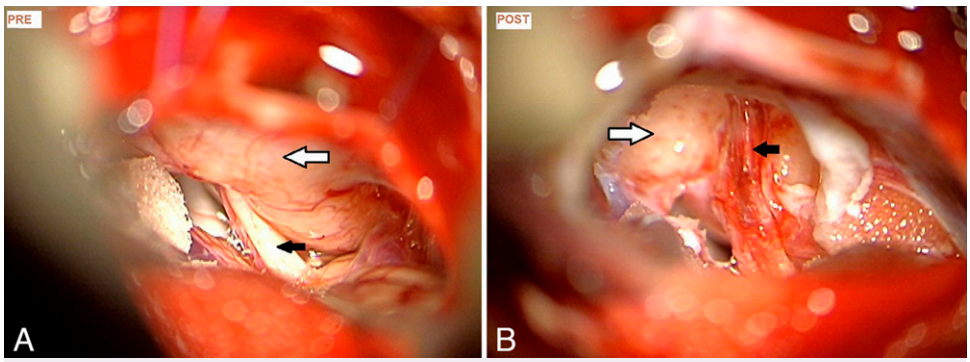

FIG. 2. Intraoperative microscopic view. **A:** Image before decompression showing the hyperostotic petrosal bone (*white arrow*) with the vestibulocochlear nerve (*black arrow*) entering the narrowed porus of the IAC. **B:** Image after decompression showing the thinned sclerotic petrosal bone (*white arrow*) and a proximally opened IAC with vestibulocochlear and facial nerve complex (*black arrow*).

left facial nerve and vestibulocochlear nerve. A 270° decompression of the severely sclerotic IAC was performed via a left retrosigmoid craniotomy with intraoperative monitoring of facial nerve and auditory nerve functions. The drilling was performed using a surgical microscope and diamond burs with continuous irrigation and suction. The drilling was performed from the petrosal bone back wall medially of the meatus into the IAC with bony removal up to the labyrinth, respecting the endolymphatic sac and duct as lateral limit landmarks.

There were no significant complications. A postoperative CT scan of the middle and posterior fossa showed a satisfactory result (Fig. 1B). Perioperative and postoperative facial nerve function was intact. Hearing and vestibular function was unchanged at the follow-up, i.e., the 1-month, 10-month, and 12-month audiological evaluations. To date, more than 1 year after the surgery, there are no more episodes of facial palsy or spasm. The extensive hyperostosis was also present at the right side of the skull base with obliteration of the right IAC. In consultation with the patient, to prevent deterioration of right vestibulocochlear and facial nerve function, decompression of the right IAC was performed (Fig. 2) 1 year after the surgery for the left side. The approach for this surgery was also retrosigmoidal, with a similar 270° bony decompression of the IAC under perioperative monitoring of facial and vestibulocochlear nerve function. There were no perioperative or postoperative complications, especially concerning hearing and facial nerve function. Routine radiological and audiological follow-up is continued.

Discussion

Observations

CED is a rare form of sclerosing bone dysplasia with involvement of the cranial skeleton. It is an autosomal dominant condition with an associated mutation in the *TGFB1* gene, giving rise to a misbalance in bone formation versus resorption in favor of the first.[6] A few cases have been described in which there is no known mutation in the gene encoding the *TGFB1* molecule, suggesting genetic heterogeneity.[7] The patient presented in this case report has all the clinical characteristics of CED, though lacking the mutation in the *TGFB1* gene. To date, after extensive clinical and genetic evaluation, no diagnosis other than CED can be made.

CED can give rise to a wide range of musculoskeletal problems from a waddling gait and pain in the extremities to muscle weakness.[8] CED can also have important neurological manifestations due to the association of cranial hyperostosis and, more specifically, hyperostosis/sclerosis of the skull base with subsequent stenosis of cranial nerve foramina.[8]

Skull base involvement occurs in around 50% of patients.[8,9] In a retrospective study of 306 published cases, Carlson et al.[9] summarized the prevalence of the diverse cranial nerve involvement: hyposmia in 0.3% of cases, visual deficits in 5.6%, papilledema in 4.6%, facial numbness in 0.7%, facial paresis/paralysis/spasm in 4.2%, hearing loss in 19%, vertigo in 4.2%, and dysphagia in 1%. The most frequent neurological complication is hearing loss, of which 7.2% of cases consist of sensorineural hearing loss due to bony overgrowth and critical stenosis of the IAC resulting in significant neurovascular compression. Facial nerve palsy and spasm is the most frequently encountered motor cranial nerve involvement and results from similar neurovascular compression at the IAC.

The nature of symptoms, when present, is usually progressive and requires an active therapeutic approach. From the early 1970s, there has been a prominent role for corticosteroid treatment in CED.[10,11] Despite its still unclear mechanism of action, it gives good alleviation of symptoms. However, it does not alter the clinical course and, like analgesics, should be considered a symptomatic treatment.[11,12] With progressive neurological symptoms secondary to the increase of intracranial pressure or progressive nerve palsy, eventually some form of surgical decompression is indicated. Mocco et al.[13] presented two patients with hyperostosis of the cranium secondary to CED who underwent successful cranial vault decompression by multiple craniotomies and cranioplasties using thinned bone flaps. Both patients suffered mainly from progressive headaches and vision abnormalities. The goal of surgery was management of raised intracranial pressure, which was achieved and maintained during the long-term follow-up. The right-eye vision abnormalities in the first patient did not improve after surgery, and this was attributed to a progressive foraminal stenosis with optic nerve compression. In their discussion, the authors emphasize that symptoms related to foraminal stenosis are less likely to be resolved with their "aggressive" cranial vault decompressive surgery. In the treatment of nerve compression syndromes in cranial hyperostosis, a targeted foraminal decompression should be considered.

There are different approaches to facial and vestibulocochlear nerve decompression. Selection of the approach is determined by the epifocus of the stenosis and hearing status of the patient as well as the expertise or preference of the surgical team.

Transmastoid Approach

A transmastoid approach gives adequate exposure to the mastoid and tympanic segments, and limited access to the geniculate ganglion, labyrinthine segment, and IAC. Yarington and Sprinkle[14] in 1967 and Hamersma[15] in the early seventies describe a decompression of the facial nerve via the transmastoid approach in separate cases of facial nerve palsy secondary to hyperostosis. There was no long-lasting effect on the facial nerve palsy.

Middle Fossa Approach

A middle fossa approach gives a broader exposure from the IAC to the tympanic segment. However, this approach also has limitations and risks of significant complications, for example, edema of the temporal lobe. Nevertheless, it is the most frequently described approach for decompression of the IAC in cases of neurovascular compression by hyperostosis. In 1980, Miyamoto et al.[16] described three cases with hearing loss with and without facial palsy due to hyperostosis of the IAC. Two of the three cases were treated surgically using a middle cranial fossa approach. The first case was a 26-year-old patient with CED with bilateral hearing loss, facial paresis of the right side, and chronic unsteadiness. The patient was already deaf on the right side. A right middle cranial fossa approach was performed for decompression of the facial nerve, with subsequent gradual but incomplete improvement of facial nerve function. Because of intermittent left-sided hearing loss, an additional decompression was performed on the left meatus structures. After an initial improvement of hearing, over the ensuing years there was a gradual and serious deterioration. The second case was a 30-year-old woman with CED who presented with bilateral sensorineural hearing loss due to hyperostosis of the IAC. Again, a middle cranial fossa procedure was performed on the right side for decompression of the IAC. Following surgery, there was stabilization of the hearing.

In 2004, Tibesar et al.[17] presented a case of a 58-year-old patient with CED with long-standing left profound hearing loss, progressive right-sided sensorineural hearing loss, and mild tinnitus. Electrophysiological audiometry and radiological imaging confirmed IAC

stenosis as the likely cause. They performed decompression of the cochlear nerve, also via a right middle cranial fossa approach to the IAC. There was no mention of intraoperative neuromonitoring and the dural sheath was left intact. At follow-up, the tinnitus resolved and the hearing loss stabilized but did not improve. Afterwards, the patient received a right cochlear implant. In cases of isolated facial nerve palsy in generalized temporal bone dysplasia, a middle fossa approach is preferred because of the possibility of decompression of the facial nerve along its meatal and labyrinthine trajectory, which can also be markedly stenotic.

Translabyrinthine Approach

A translabyrinthine approach allows for an exposure of the IAC in its entirety, and the complete infratemporal course of the facial nerve is accessible. However, it can only be applied in cases in which cochleovestibular function is already lost or worth sacrificing. Most of the time, this is not the case for decompressive surgery in temporal bone dysplasia in which hearing preservation should be a main objective. To our knowledge, there are no reports on a translabyrinthine approach for nerve decompression in sclerosing bone dysplasia.

Retrosigmoid Approach

The retrosigmoid approach gives a nice exposure of the proximal part of the IAC and has several other advantages. Especially in cases of changed bony anatomical landmarks, it allows for relatively easy nerve recognition and, consequently, more efficient and safer drilling. It allows for more proximal neurostimulation during intraoperative neuromonitoring. Because of the space-occupying effect of the temporal bone hyperostosis, for the middle cranial fossa approach, increased cerebral lobe retraction could be necessary with an increased risk of temporal lobe edema and epileptic sequelae. In our opinion, regarding retraction on the cerebellum, if the cisterna magna and cerebellopontine cisterns are adequately opened, no retraction on the cerebellum is required in the retrosigmoid approach.

Hellier and Brookes[18] described a case of a patient with CED with dizziness as the main symptom. This was also associated with headache, bilateral moderate fluctuating hearing loss, and left tinnitus. A cranial CT scan showed hyperostosis of the skull base with markedly narrowed IACs. Eventually, they performed an exploration and decompression of the left vestibular nerves via a retrosigmoid approach. Postoperatively, there was an immediate and sustained improvement of the vestibular symptoms. Initially, there was some increased high-frequency hearing loss in the left ear, which improved spontaneously. According to the authors, the slight increase of the high-frequency hearing loss can be explained by either thermal damage or acoustic damage to the cochlear nerve caused by the prolonged drilling. Concerning the latter explanation, acoustic energy from the drill is more efficiently transmitted to the cochlea by denser bone. The authors suggest that a retrosigmoid approach is preferred in cases of primary vestibular symptoms because the vestibular nerves occupy the posterior compartment of the IAC. Baik et al.[19] describe two cases of IAC exostoses accompanied by vestibular symptoms. In one patient, a decompression of the posterior lip of the IAC was performed via a retrosigmoid approach. However, in this case, the bony overgrowth was limited to the porus of the IAC and there was no apparent diagnosis of bone dysplasia.

To our knowledge, this is the first report describing decompression of the IAC via the retrosigmoid approach for symptomatic facial and cochlear nerve compression in a patient with a diagnosis of CED. In cases of predominant or isolated facial nerve neuropathy, IAC decompression with a favorable outcome via a middle fossa approach has been described. This case shows that the stabilization of symptoms may be achieved for both hearing and facial nerve neuropathy when using a retrosigmoid approach for IAC decompression in cases of skull base hyperostosis. This treatment does not alter the progressive pathophysiological mechanisms associated with CED; hence, the recurrence of symptomatic stenosis is possible. Repeated surgeries may be necessary. This is reflected by the case report of Wong et al.,[20] who describe a 46-year-old patient with CED who required multiple surgeries because of the rethickening of cranial bones causing intracranial hypertension.

In any surgery focusing on the salvation of nerve function, the risks of further nerve damage should be minimalized. Manipulation of already compromised nervous tissue, especially in combination with changed anatomical landmarks, poses additional risks for nerve damage. Furthermore, the required use of prolonged drilling presents a greater risk of thermal and acoustic damage to the nervous tissue. Therefore, these kinds of surgeries should be performed in a skull base center by an experienced multidisciplinary team with the use of perioperative neuromonitoring.

Lessons

In selected patients with significant facial or vestibulocochlear neuropathy due to hyperostotic foraminal stenosis, focused skull base decompression via the retrosigmoid approach proves to be a good therapeutic option. However, decompressive skull base surgery in patients with hyperostosis poses additional risks of iatrogenic complications due to changed anatomical landmarks and sclerotic tissue. Therefore, intraoperative neuromonitoring is indispensable.

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Disclosures

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author Contributions

Conception and design: all authors. Acquisition of data: all authors. Analysis and interpretation of data: all authors. Drafting the article: Achahbar. Critically revising the article: all authors. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Achahbar. Administrative/technical/material support: Achahbar. Study supervision: all authors.

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