Management of trigeminal neuralgia in sclerosteosis

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Abstract

Background: Sclerosteosis is a rare bone disorder characterized by a progressive craniotubular hyperostosis. The diagnosis of sclerosteosis is based on characteristic clinical and radiographic features and a family history consistent with autosomal recessive inheritance. The skull overgrowth may lead to lethal elevation of intracranial pressure, distortion of the face, and entrapment of cranial nerves, resulting in recurrent facial palsy or secondary trigeminal neuralgia.

Cases Description: The authors reported cases of two siblings who were diagnosed with familial sclerosteosis and presented with secondary trigeminal neuralgia. The patients were 28 and 40-year-old and presented with pain in the right V2-V3 and V3 distributions, respectively. The facial pain was resistant to medications and was treated with percutaneous techniques. The foramen ovale puncture was complicated initially and the difficulty increased over the years due to stenosis of the foramen.

Conclusion: The treatment of the trigeminal neuralgia secondary to hyperostosis and resistant to medications presents a dilemma. The narrowing of the foramen ovale and difficulty in the identifying and approaching of the foramen makes the percutaneous technique a challenge for the neurosurgeon in patients harboring sclerosteosis. Microvascular decompression should not be considered since the primary cause of the trigeminal neuralgia is the nerve entrapment by the narrowing of neurovascular foramina and not the neurovascular conflict related to essential trigeminal neuralgia. Stereotactic radiosurgery may be a good treatment option, but there is a lack of published data supporting the use of this method in cranial hyperostosis.

Key Words: Percutaneous balloon compression, pain control, sclerosteosis, trigeminal neuralgia

INTRODUCTION

Sclerosteosis is a rare bone disorder characterized by a progressive craniotubular hyperostosis. It is caused by mutations in SOST gene encoding sclerostin protein, an osteocyte-specific secreted protein that is likely involved in the suppression of bone formation.[2,6,13,27,30] The diagnosis of sclerosteosis is based on clinical characteristics, radiographic features, and a family history.
consistent with autosomal recessive inheritance. The main features are a significant sclerosis of the long bones, ribs, pelvis, and skull.

The skull overgrowth may lead to lethal elevation of intracranial pressure (ICP), distortion of the face, and entrapment of cranial nerves. The clinical result is conductive hearing loss in childhood followed by additional entrapment of the eighth cranial nerve and closure of the oval and round windows, leading to sensorineural hearing loss in adulthood.[5,28] Recurrent facial palsy that is initially intermittent and eventually results in constant impaired of facial movements in adulthood, and paroxystic facial pain similar to trigeminal neuralgia (TN) secondary to stenosis of the foramina of its branches has also been described.[3]

The authors present here the management of TN in two siblings with diagnosed familial sclerosteosis and discuss the possibilities and potential complication of treatment modalities.[18]

CASES REPORT

Case 1
A 40-year-old male patient started his symptoms at the age of 10. At the beginning, he presented intermittent right side facial muscle paralysis and jaw pain. Over the years the symptoms progressed to bilateral facial muscle paralysis, bilateral hypoacusia, headaches, and right side facial pain (jaw, teeth, and tongue).

In 2002, he underwent a dental procedure in another institution because of his jaw pain. In 2004, at the age of 30, the patient moved to São Paulo where he was diagnosed with sclerosteosis and TN affecting the right mandibular branch - V3 [Figure 1]. The facial pain was controlled for one year with carbamazepine 1200 mg per day, but side effects limited the maintenance of this medication. Gabapentin was also tried but there was a poor response, so surgical treatment was advised. A percutaneous balloon compression (PBC) was performed in 2005 and the patient had pain control for one year without the use of medications. In 2006, due to recurrence of pain, gabapentin and phenytoin were tried with partial improvement, when a percutaneous radiofrequency rhizotomy (RFR) of the right V3 was performed.

The patient had a 2-year period with no facial pain and another 2 years with the pain under control with the use of gabapentin. The control of the TN became worse, despite the increasing doses, and combination of medications. Another percutaneous RFR was performed to treat his right V3 pain in 2012 [Figure 2]. It should be noted that the difficulty in the percutaneous procedures increased over the years due to stenosis of the foramen ovale. However, today his neuralgia is controlled with no medications and he presents with hypoestesia in the right V3 territory.

Case 2
A 28-year-old woman started her symptoms at the age of 1. Initially she presented an episode of right facial palsy that evolved with spontaneous remission after 3 months. After 1 year, she had new-onset facial palsy that affected her left facial side, with complete resolution after 3 months. Over the next years, she experienced recurrent facial palsies and suffered from several headaches. A generous bilateral bifrontotemporal decompressive craniectomy was then performed to treat the symptoms of raised ICP that improved after the surgical procedure [Figure 3].

At the age of 21, the patient was diagnosed with sclerosteosis and TN in the right V2 and V3 distributions. The patient was primarily treated with carbamazepine and gabapentin until presented with an adverse reaction, before being considered for surgery. She underwent a PBC for treatment of the right facial pain in 2007. After surgery, the patient had an immediate and complete resolution of her V2 and V3 pain but experienced right facial numbness in these trigeminal distributions. After 3 years, the pain recurred with the same intensity and a new PBC was performed. She was again placed on medical therapy with carbamazepine and the pain improved in the immediate postoperative period.

After 2 years, in 2012 the pain recurred with the same initial characteristics and another PBC was performed [Figure 4]. Postoperatively, the patient was free of pain and the distribution of facial numbness was unchanged. The patient was able to stop taking any pain medication within 3 weeks after surgery. Six months after surgery, she continued to be free of facial pain. It is important to note, similar do case 1, the increasing difficulty of the foramen ovale puncture over the years due to its stenosis.

DISCUSSION

TN may be classified as idiopathic or symptomatic according to the etiology. In the idiopathic or essential TN the etiology is unknown. Patients describe a short, intermittent, and sharp or electric shock like one-sided pain affecting one or more division of the trigeminal nerve. In the symptomatic TN, also called secondary, an etiology for the symptoms is identified. Multiple sclerosis and posterior fossa tumors are the most frequent cause of symptomatic TN.[9,14] Recently, Buerchiel proposed a new classification system for facial pain syndromes with seven categories based on the patient’s history and quality of pain. The idiopathic TN is divided in type 1 (spontaneous and episodic) and type 2 (constant pain). The facial pain syndromes are further divided in
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trigeminal neuropathic pain resulting from unintentional injury to the trigeminal nerve from trauma or surgery; trigeminal deafferentation pain as a result of injury to the nerve by peripheral nerve ablation, gangliolysis, or rhizotomy in an intentional attempt to treat either TN or other facial pain; postherpetic neuralgia follows a cutaneous herpes zoster outbreak in the trigeminal distribution; and symptomatic TN results from multiple sclerosis. The final category, atypical facial pain, is synonymous with facial pain secondary to a somatoform pain disorder. Regardless of the classification system, the cases described above account for rare causes of symptomatic TN due to compressive effect of bone overgrowth.

The molecular genetic studies of patients with the sclerosing bone dysplasias revealed that the loss of function of SOST gene results in an osteoblast hyperactivity and is involved in the pathogenesis of this condition, as well as in the van Buchem disease. Kim et al. previously reported the molecular aspects of the familial sclerosteosis described in the present article. The analysis showed that both patients were homozygous for the same nonsense mutation (Trp124X) in exon 2 of the SOST gene. At the time of the previous report, only the male patient (case 1) presented with facial pain.

Tacconi et al. described a case of sclerosteosis in a Black African individual who suffered from headache and TN. The patient presented with recurrent episodes of facial palsy and reported diminished vision in his left eye since childhood. On neurological examination, there was a diminution of touch and pin-prick sensation affecting the three divisions of the left trigeminal nerve, and the left corneal reflex was reduced. The radiologic examination showed an increased bone density and narrowing of the neurovascular foramina. There was no description about treatment of the TN.

Hofmeyr et al. reviewed the literature regarding the neurologic evaluation and management of the more frequent sclerosing bone dysplasias, including sclerosteosis and Van Buchem disease. Hearing loss, recurrent facial palsy, headache, proptosis, and facial pain seem to be the more frequent neurological complaints. In a series of 14 patients with sclerosteosis seen over a period of 14 years, examination revealed changes in the optic disc in five patients, but only one patient presented with severe visual impairment. All patients had bone overgrowth.

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Tissue samples were collected for genetic analysis, and DNA was extracted from peripheral blood leukocytes. The analysis showed that both patients were homozygous for the same nonsense mutation (Trp124X) in exon 2 of the SOST gene. At the time of the previous report, only the male patient (case 1) presented with facial pain.
overgrowth of the calvarium and mandible associated with proptosis and some form of nail dystrophy of the fingers or toes. All patients underwent decompressive craniotomies because of the increase in skull thickness demonstrated in the computed tomography (CT) and the signs of raised ICP. Sometimes repeated decompressive craniotomy may be required in cases presenting with increased ICP due to the natural progression of the bone sclerosis. There was no description about facial pain or TN in this series.

Facial nerve palsy usually presents as an acute and recurrent paralysis, and many patients are frequently diagnosed erroneously as suffering from Bell Palsy. Dort et al. measured the surface area of the fallopian canal and facial nerve in sclerosteosis of the temporal bone and found narrowing of the fallopian canal and facial nerve in the labyrinthine, distal tympanic and mastoid segments, demonstrating that ischemia and bony compression are the underlying causes of recurrent facial palsy in this disease. The narrowing of the internal auditory canal can also compress the acoustic nerve inside the internal auditory meatus causing sensorineural hearing loss. The reduced intracranial volume due to the overgrowth of the calvarium can produce a raise in the ICP causing chronic headache, papilledema, and others symptoms.

TN in the related cases may be attributed to the bony growth that occurs in the superior orbital fissure and in foramen ovale and rotundum. The slow compression of the trigeminal branches can induce a progressive degeneration of the nerve. As we observed in the cases reported in this article, the narrowing of the foramen ovale and rotation of TN making this modality of approach a challenge for the neurosurgeon.

Percutaneous techniques for treatment of TN include RFR, PBC, and percutaneous retrogasserian glycerol rhizolysis (PRGR). According to a systematic review, RFR is the second most effective surgical treatment, but the pain control is associated with the presence of facial numbness and a high risk of side effects comparable to the other percutaneous techniques, including anesthesia dolorosa, corneal hypoesthesia, and keratitis. PBC produce an acceptable pain relief time and is associated with a lower incidence of complications compared with RFR and PRGR, although a transient masseter weakness may be expected in almost all cases during the immediate postoperative period. The balloon shape may have an important role in the clinical results of PBC, and the pear-shaped balloons are associated with a superior pain relief compared with other balloon shapes. In patients harboring sclerosteosis the percutaneous techniques are hindered due to the narrowing and difficulty in identifying the foramen ovale. In such cases, the use of CT-guided percutaneous rhizotomy with three dimensional imaging reconstructions, or a neuronavigation system can facilitate the needle placement and reduce the risk of complications.

The unintentional puncture of vascular and other vital structures around the foramen can result in cranial nerve pareses, cerebrospinal fluid (CSF) leakage, carotid-cavernous fistula, intracranial hemorrhage, and others morbidities. The understanding of the skull base anatomy and the relationships of the foramen ovale is mandatory to keep away from complications during puncture. The more frequent and severe puncture upsets are related to errors in the needle placement both intra- and extracranial. The correct angle and safety distances in addition to better radiographic identification of the FO may improve the safety of this procedure.

Microvascular decompression (MVD) leads to the highest rate of pain control and is the most durable surgical treatment, however, is more invasive than other methods and requires general anesthesia and a craniotomy. Several authors recommend that MVD must be the method of choice even in elderly patients, since they present with good health conditions. The MVD has low rates of sensory dysfunction, but the complications have a propensity to be more severe as meningitis, CSF leakage, stroke, and even death. Although patients underwent to MVD presents with long-term pain relief, this procedure must be avoided in sclerosteosis patients. There is a marked difficulty in the surgical approach due to the prominent hyperostosis near the skull base and this procedure can be dangerous when performed in patients with markedly increased ICP secondary to a reduce cranial capacity. In case with diagnosed sclerosteosis MVD should not be considered, because the primary cause of the TN is the nerve entrapment by the narrowing of neurovascular foramina and not the neurovascular conflict related to essential TN.

Stereotactic radiosurgery (SRS) can be an effective alternative in the treatment of TN. In the SRS the target is localized using three-dimensional image guidance and a high-energy radiation dose is delivered at the root entry zone (REZ) at the level of the pons. The REZ is the transition between the central and peripheral myelin produced by the Schwann cells and oligodendrocytes, respectively. It is located nearly 2-3 mm away from the anterior surface of the pons and is the place more susceptible to injuries that leads to the pain like the produce by vessels contact. The maximal doses are usually of 70-90 Gy, and doses superior than 90 Gy is associated with potential complications such as facial numbness, anesthesia dolorosa, and radiation-induced neoplasia.

Compared with open surgery and percutaneous techniques, the SRS is less invasive, more precise and has
a reduced amount of postoperative complications. The mean disadvantages of radiosurgery are the high rate of recurrence and the time latency to provide pain relief after radiation treatment.\textsuperscript{[11,21,24]} According to Maesawa \textit{et al.}\textsuperscript{[20]} more than 50% pain relief can be achieved in 71% of patients in 2 years, and only 56% sustained the pain relief at 5 years. In patients with no previous surgery the latency interval for the predictable response has been noted to be of about 3-5 weeks after radiosurgery, so patients under acute pain attack are not candidates for this modality of treatment.\textsuperscript{[20,24]} We presume that SRS to be a good alternative in patients harboring sclerosteosis and presenting with no acute pain, since there is no need for foramen oval puncture or craniotomy, but no evidence has been published about the results of this technique in patients with cranial hyperostosis.

**CONCLUSION**

The treatment of the TN secondary to hyperostosis and resistant to medications presents a dilemma. The percutaneous technique is a reasonable treatment option in sclerosteosis patients, but the narrowing of the foramen oval and difficulty in the identifying and approaching of the foramen makes this modality of approach a challenge for the neurosurgeon. Although MVD is recognized as the “gold standard” treatment for patients with TN, there is no role for this surgical approach in patients harboring sclerosteosis since the etiology of pain is diverse and not related to the neurovascular conflict. SRS provide a minimally invasive option for management of TN in patients who are not able or reluctant to undergo surgery, but no evidence has been published about the results of this technique in patients with cranial hyperostosis.

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