

Intraosseous schwannoma of the mandible and schwannoma of the spinal cord: A rare presentation of schwannomatosis – Case report and review of the literature

ABSTRACT

Schwannomatosis is a recently recognized distinct form of neurofibromatosis (NF). It is a rare condition, the incidence of which varies between 1/400,000 and 1/1.7 million. An important feature of schwannomatosis is the presence of multiple intracranial, spinal, and peripheral schwannomas in the absence of acoustic neuromas. Schwannomatosis presenting with intraosseous schwannoma of the mandible is even rarer, and only a few cases have been reported. It usually affects individuals in the third to fifth decade of life. Usually, it is sporadic in origin, but in 20% of patients, it can be familial. As a diagnostic criterion, NF2 gene is not involved in schwannomatosis. We report a case of a 48-year-old male presenting with facial pain and difficulty in chewing, and subsequent development of spastic paraplegia. Magnetic resonance imaging scan of head and neck revealed mass lesion involving infratemporal region on the left side, intraosseous lesion of the mandible, and multiple mass lesions in the neck. Acoustic nerves were not involved. Mutagen-induced chromosome sensitivity analysis test suggested no predisposition for malignancy. His clinical features are suggestive of schwannomatosis, which is a recently recognized distinct form of NF.

Keywords: Intraosseous schwannoma, schwannomatosis, spinal cord schwannoma

INTRODUCTION

Classically, there are two major types of neurofibromatosis (NF), consisting of NF1 and NF2. Schwannomatosis is a recently recognized form of NF with certain distinct features differentiating it from NF2. It is a rare condition, the incidence of which varies between 1/400,000 and 1/1.7 million.^[1] Retrospective studies suggest that 2.4%–5% of resected schwannomas can be attributed to schwannomatosis.

Acoustic neuromas, either unilateral or bilateral, are characteristic of NF2 but are absent in schwannomatosis. Other cranial nerves may be affected in this condition. Segmental subtypes with lesions localized to a single limb or five or fewer contiguous spinal segments are more common in schwannomatosis. NF2 usually affects young adults, whereas schwannomatosis typically occurs in the third to sixth decade of life. NF2 is an autosomal dominant syndrome caused

by mutations in the NF2 gene, whereas schwannomatosis is usually sporadic, though familial cases occur in <20% patients. Schwannomatosis is a genetic condition but may skip generations so that more distant family members with unexplained neurological symptoms and or unexplained pain attributable to swellings resembling schwannoma should be evaluated for the possibility this disease. Genetic basis of

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schwannomatosis is not clearly known. It does not follow the common inheritance pattern. Two genes responsible for this syndrome are SMARCB1 and the LZTR1.^[2] In a case of multiple schwannomas, a diagnosis of schwannomatosis is made when acoustic nerve tumors are ruled out with magnetic resonance imaging (MRI) scan, the patient is older than 30 years, genetic study rules out NF2 gene, and no first-degree relative has NF2.^[3]

CASE REPORT

A 48-year-old male presented with history of numbness over the left side of face along the distribution of his trigeminal nerve and difficulty in chewing of 1-year duration. He also had progressive difficulty in walking due to spastic paraplegia of 6-month duration. He had neither cutaneous swellings or *café-u-lait* spots or other neurocutaneous markers suggestive of NF nor any first-degree relatives with NF. MRI scan and computed tomography (CT) scan of head and neck revealed a heterogeneous intensity mass lesion with contrast enhancement in the left infratemporal region, suggestive of trigeminal schwannoma, intraosseous schwannoma of the mandible, and multiple small tumoral masses in the neck arising from the cervical nerve roots [Figures 1 and 2]. MRI scan of spine showed an intradural schwannoma compressing the cord at thoracic level. He underwent surgical removal of the spinal lesion, mandibular lesion, and neck swellings. Histopathology and immunohistochemistry reports came as schwannoma [Figures 3 and 4]. Mutagen (Bleomycin)-induced chromosome sensitivity analysis to evaluate the DNA repair proficiency was done to look for any predisposition for malignancies. He was hyposensitive with a value of <0.8 and hence had no oncopathology predisposition.

Ultrasound scan of the abdomen and CT scan of the thorax were normal. Blood investigations were normal and urine test for vanillylmandelic acid was negative.

DISCUSSION

Schwannomatosis was recognized as a distinct clinical entity different from NF in 1996. It was initially reported in Japanese patients.^[4] Schwannomas are mainly benign tumors that commonly occur in individuals with NF2. Exact genetic cause of schwannomatosis is not clear. The candidate schwannomatosis gene SMARCB1, a tumor suppressor gene, was identified in 2007. Furthermore, recently a new gene LZTR1 was identified and was found to be positive in 100% of familial and 70% of sporadic cases.^[2] Both these genes are located on the short arm of chromosome 22. An inactivating germline mutation in exon 1 of the SMARCB 1 has been

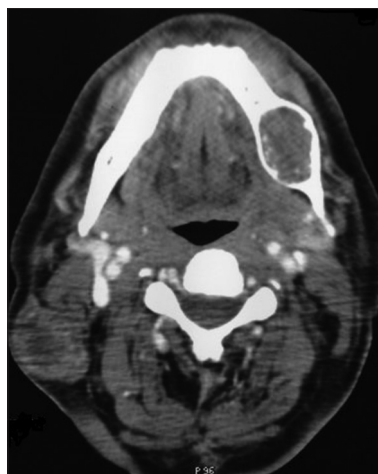


Figure 1: Computed tomography scan of the mandible – revealing intraosseous schwannoma of the mandible

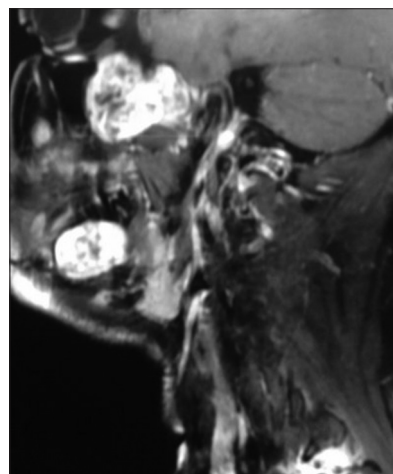


Figure 2: Magnetic resonance imaging scan – T1 weighted with gadolinium contrast, sagittal view revealing mandibular, and infratemporal schwannomas

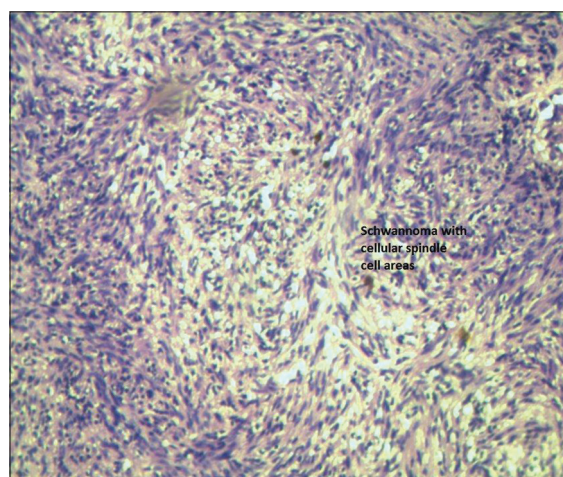


Figure 3: Photomicrograph – H and E, ×20 view of the lesion revealing highly cellular stroma with cells arranged in swirls and whorls

reported in some of the cases of schwannomatosis. It is a genetic disorder and familial occurrence is rare. Schwann

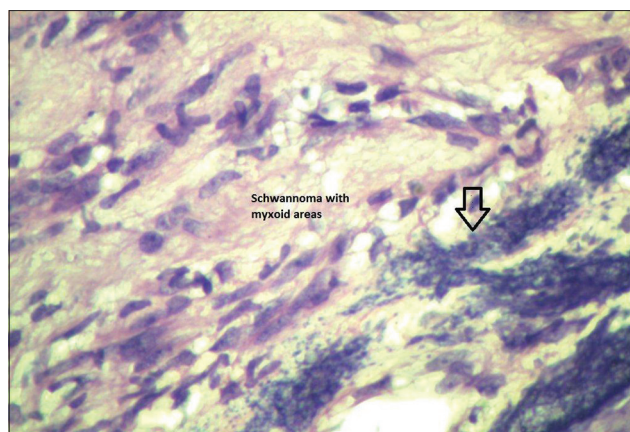


Figure 4: Photomicrograph – H and E, ×40 view showing myxoid areas in the tumor

cells are glial cells that myelinate the axons of neurons. Myelin is a lipid covering that increases the conduction velocity of action potentials. When Schwann cells proliferate out of control in an encapsulation, it is called schwannoma. Schwannomas are very homogeneous tumors consisting only of Schwann cells. They stay on the outside of nerve but may push it aside. Neurofibromas are very heterogeneous tumors which incorporate all sorts of cells and structural elements in addition to Schwann cells. They infiltrate the nerve and splay apart the individual nerve fibers. Neurofibromas are more likely to degenerate into cancer than schwannomas. Schwannomas mainly develop in the cranial, spinal, and peripheral nerves.^[3] Criteria for diagnosing schwannomatosis include two or more noncutaneous schwannomas, no evidence of vestibular tumors, and no known NF2 gene mutation or a pathologically confirmed nonvestibular schwannoma plus a first-degree relative with schwannomatosis.^[5] There is no distinct feature to distinguish between a schwannoma of schwannomatosis from that of NF. However, according to MacCollin *et al.*, intraneural/peritumoral edema, intratumoral myxoid changes, and intraneural growth may be more common in schwannomatosis.^[6] Our patient had myxoid changes within the tumor which were a positive sign indicative of schwannomatosis. There have been reports, confirming the expression of vascular endothelial growth factor in schwannomatosis. In such cases, alternative therapy with bevacizumab has also been successfully reported.^[5,7-9]

Intraosseous schwannoma is exceedingly rare. It accounts for <1% of primary bone tumors. Mandible is the most commonly affected site, probably due to long course of the inferior alveolar nerve through the bone. Intraosseous schwannoma may develop from nutrient canal, arise centrally from the bone, or erode through the bone cortex and become

intraosseous.^[10] Only a few cases of schwannomatosis been reported with intraosseous schwannoma of the mandible. Other sites of intraosseous schwannomas include the vertebrae, ribs, sacrum, radius, and ulna. Due to the benign nature of this lesion and the low potency for malignant transformation, a growing awareness regarding this disease is essential.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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