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A rare case of schwannomatosis

Dr. Reeta Dhar[1], Dr. Shilpi Sahu[2], Dr. Kuldeep singh[3]
[1.] *Professor and HOD, Department of pathology. MGM Medical College, Navi Mumbai*
[2.] *Associate professor,Department of pathology. MGM Medical College,Navi Mumbai*
[3.] *Resident,Department of pathology. MGM Medical College, Navi Mumbai*

Abstract: *Schwannomas are benign,slowly growing, encapsulated peripheral nerve tumors. Schwannomas are homogenous tumors and consist of only Schwann cells. They develop outside of the nerve, but may push it aside or against adjacent structure causing damage to the nerve. Most schwannomas occur as solitary lesions. Presence of multiple schwannomas in a single patient suggeststumorogenesis and a possible association with one of the syndromes such as neurofibromatosis 2 or with Schwannomatosis. Schwannomatosis is diagnosed in the presence of multiple pathologically proven schwannomas in the absence of neurofibromatosis2 (NF2). We report a very rare case of multiple schwannomasin the absence of typical signs and symptoms of NF1 and NF2.*
Key word: *Multiple schwannomas, Schwannomatosis,Schwannoma, Neurofibromatosis*

I. Introduction:

Schwannomas also known as Neurilemmoma or Schwann cell tumor are benign nerve sheath tumor composed of Schwann cells. Their function is to form myelin sheath of nerves and facilitate transmission of nervous impulses. Theycan be found in various part of body with most common site being head. Unlike neurofibromas, schwannomas rarely metastasize and symptoms are because of mass effect of the tumor.Schwannomatosis is defined, therefore, as an extremely rare tumor syndrome characterized by the presence of multiple schwannomas in the absence of typical signs of NF1 and NF2 syndromes. Usually, patients with Schwannomatosis develop cranial, spinal or peripheral nerve schwannomas but do not develop vestibular lesion(VS), typical of NF2[1]**.**

II. Case report:

A 55 year old male patient came with two spinal swellings and one right hand middle finger tumor since 4 months. Patient was apparently well 4 months ago when he noticed swelling on the back and right middle finger. Swellings gradually increased in the size. Patient did not have any significant complaints except tingling in right leg. Finger mass was completely asymptomatic. Patient did not have any vestibular complaints and MRI did not reveal any lesion of eighth cranial nerve. MRI of the spine revealed two intradural extramedullary masses one measuring 2x1.5 cm. well defined mass at the level of D11-D12 vertebrae another lesion measuring 0.7x0.5 cm. mass at the level of L4 vertebrae.

Patient was operated and larger spinal mass was excised first which was at the level of D10-D11 vertebrae.

GROSS- We received single grey white, nodular tissue piece measuring 2.5x1x0.5 cm. cut section of mass showed grey white homogenous area.

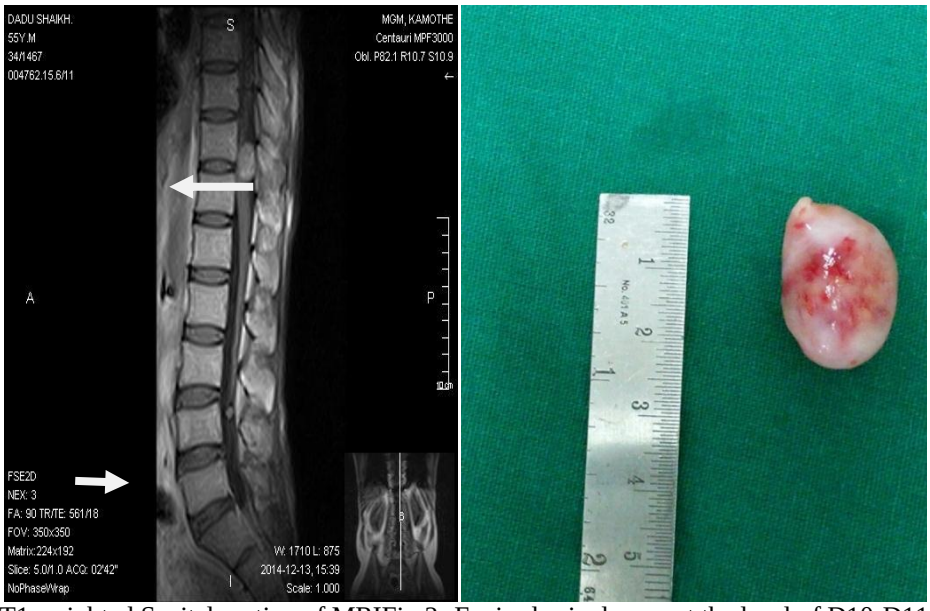

Figure1 –T1 weighted Sagital section of MRIFig 2- Excised spinal mass at the level of D10-D11 Showing lesions (arrows)

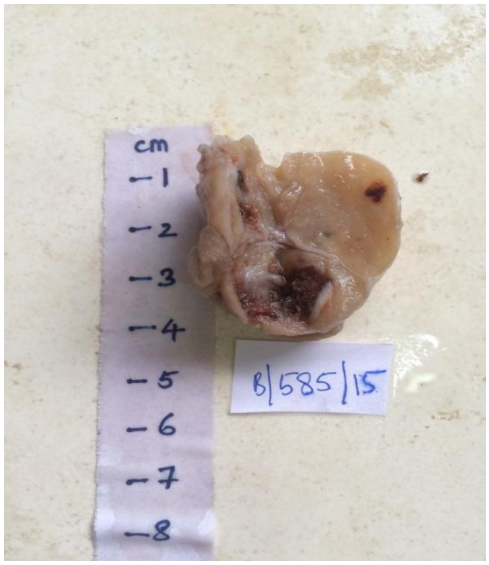

Fig3-Cut section of finger mass

After one month patient was again operated for spinal mass at L4 level and finger mass.

Gross-Specimen of right hand middle finger mass was single, globular,grey-white tissue measuring 4x2.5xx0.5 cm. Cut section showed grey-white areas along with areas of haemorrhage.

Specimen of spinal mass at L4 was single, grey brown tissue measuring 0.5 cm.

Microscopy-On microscopic examination all the three masses showed similar histomorphological features. H & E stained sections studied show a mixture of two growth patterns. In the antoniA pattern of growth, elongated cells with cytoplasmic processes were arranged in fascicles in areas of moderate to high cellularity and scant stromal matrix along with Verocay bodies which were nuclear free zone of processes that lie between the regions of nuclear palisading. In the Antoni B pattern of growth, the tumor was less densely cellular and consists of loose meshwork of cells and myxoidstroma.

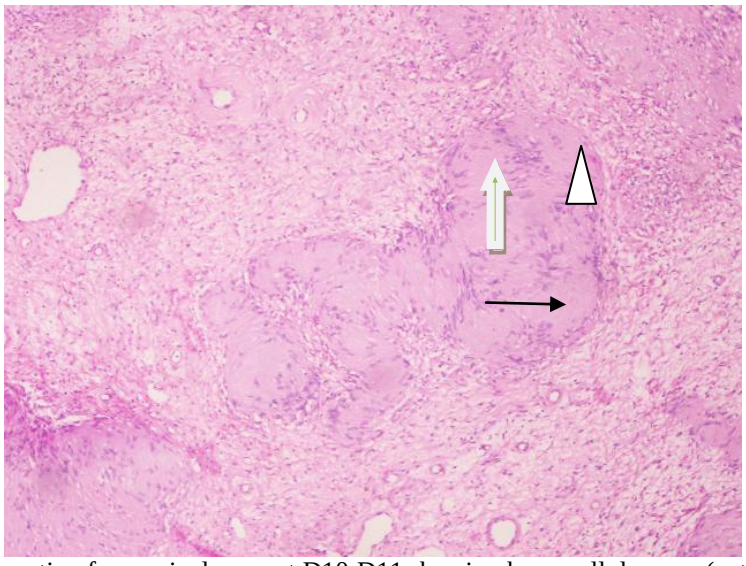

Fig 4- H&E stained section from spinal mass at D10-D11 showing hypercellular area (antoniA) {arrowhead} including verocay bodies {arrow} as well as looser myxoid region (antoni B) {thick arrow}. Sections from finger mass and spinal tumor at L4 showed similar histomorphological features.

III. Discussion:

Schwannomatosis was first reported in 1973 as neurofibromatosis type three [2]. Schwannomas are benign peripheral nerve sheath tumors that can occur in isolation or as part of neurofibromatosis type 2 (NF2), where they typically involve the vestibular nerves bilaterally. Cutaneous schwannomas may be easily confused with neurofibromatosis, which are a feature of neurofibromatosis type 1[3]. Schwannomatosis, a recently defined entity characterised by multiple cutaneous and spinal schwannomas,without vestibular involvement or other sign of NF1 or NF2.Although the precise incidence of Schwannomatosis is still unknown. Data suggest incidences may be similar to the incidences of NF2, accounting for as many as 2.4 to 5% cases of all resected schwannomas [7]. An important aspect of classification of Schwannomatosis is a differential diagnosis of neurofibromatosis. However making distinction between Schwannomatosis and neurofibromatosis type 2 on histology and clinical ground is sometimes difficult.A presumptive diagnosis can however, be given in the absence of radiological evidence of vestibular tumor below the age of 18 or absence of clinical evidence of VIII nerve dysfunction after the age of 30 years[4].
Presently the following diagnostic criteria used for Schwannomatosis[5, 6].

Definite:

Age more than 30 and two or more than two non-intradermal schwannomas, at least one with histologic confirmation and no evidence of vestibular tumor on MRI and no known NF mutation. Or
One nonvestibular schwannomas plus a first degree relative with Schwannomatosis.

Possible:

1) Age less than 30 and 2 or more than two nonintradermal schwannomas, at least one with histologic confirmation and no evidence of vestibular tumor on MRI scan and no known NF mutation. Or
2) Age more than 45 and two or more than two schwannomas, at least one with histologic confirmation and no symptoms of eight nerve dysfunction and no NF2.Or
3) Non vestibular schwannomas and first degree relative with Schwannomatosis.

Segmental:

Diagnosed as definite or possible but limited to one limb or less than or equal to five contiguous segment of spine.

Several authors have reported cases of patients with multiple schwannomas without vestibular lesions, suggesting the existence of a distinct syndrome from the neurofibromatosis.At this time there are no NIH criteria for Schwannomatosis. MRI examination has used has fundamental instrument to exclude the presence of vestibular involvement[1]. Several authors have studied the Schwannomatosis molecular genetics. But mutation analysis of the tumors from patient with Schwannomatosis is not routinely undertaken. Some patient can be shown to be somatic mosaic for NF2 mutations as demonstrated by the presence identical mutations in more than one tumor[3].Jacoby et al examined the NF2 locus in 20 unrelated Schwannomatosis patients and their

affected relatives. They found that the tumors showed typical truncating mutations of the NF2 gene and loss of heterogygosity of the surrounding region of chromosome 22. They found that unlike patient with NF2, no heterogygous NF2 gene changes were seen in normal tissue[6].

IV. Conclusion:

Schwannomatosis is a different syndrome from neurofibromatosis. In our case patient has three lesions which was diagnosed as schwannomas on histopathology but patient did not have any vestibular lesion as well there is no family history. Thus, this is a case of Schwannomatosis. The treatment consists in surgical removal of symptomatic lesions with preservation of the root as early as possible.

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References:

[1]. Landi A,Dugoni DE, Marotta N, Mancrella C, Delfini R. Spinal Schwannomatosis in the absence of neurofibromatosis: A very rare condition. Int j surg case report.2011;2(3): 36-39
[2]. Nimura M Neurofibromatosis. Rinsho Derma 1973;15:653-663
[3]. Murray AJ, Hughes TA, Neal JW, Howard E, Evans DGR, Harper PS. A case of multiple cutaneous schwannomas; Schwannomatosis or neurofibromatosis type 2?. J NeurolNeurosurg Psychiatry 2006:77:269-271
[4]. Hingway SR. Multiple schwannomas, including a plexiform variant, in a patient in the absence of neurofibromatosisI or II. J SciSoc 2015;42:37-41
[5]. Ferner RE, Huson SM, Evans DG. Neurofibromatosis in clinical practice. 1st ed. London: Springer-Verlag London Limited,2011:1-46
[6]. Jacoby LB, Jones D, Davis K, Kronn D, Short MP, Gussella J. Molecular analysis of the NF2 tumor-suppressor gene in Schwannomatosis. Am j Hum Genet. 1997;61:1293-1302
[7]. Perry Arie, Brat Daniel J.2010. Practical surgical Neuropathology:A Diagnostic Approach. Churchill Livingstone Elsevier