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Multiple Neurofibromas Presenting With Quadriparaesis: A Rare Entity

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Abstarct: ***We are presenting a case report of a 40 years old male patient who presented to us in medicine OPD with complaints of weakness in all 4 limbs since few months.On detailed examination and thorough evaluation, he was diagnosed to have multiple dumbell shaped neurofibroma's, compressing cervical cord leading to quadriparaesis.***

Keywords: ***Neurofibroma, Dumbell shaped, Quadriparaesis, Cervical.***

Introduction:

Neurofibromatosis 1 (NF1), an autosomal dominant condition, is caused predominantly by mutations in the NF1 gene on chromosome 17.The gene responsible for NF1 encodes neurofibromin, a protein which acts as a tumor suppressor, the loss of which leads to an increased risk of developing tumors. Affected individuals develop both benign and malignant tumors.
They develop as discrete focal cutaneous or subcutaneous tumors or more diffuse plexiform neurofibromas that grow along the length of nerves frequently involving multiple nerve fascicles, branches and plexuses.
Neurofibromatosis, being found as one incident in 2500-3000 births, is a genetic disease that may involve either skin, nervous system or both systems simultaneously leading to cosmetic and functional disorders.
Though, cervical cord compression is infrequently found in nervous system compression, nevertheless, it plays a very important role as it may lead to severe neurological deficits and it may be treated by combination of surgical intervention with physical medicine and rehabilitation in early stage.
The treatment options in the advanced cases are limited.

Sir,

A 40-years-old male presented to us in medicine, with a history of weakness in all 4 limbs since 6 months. Patient was alright 6 months back and the complaint started with weakness which was insidious onset and gradually progressive in nature. General examination revealed normal vitals. There were multiple swellings present over the trunk and extremities. The right calf was hypertrophied (Fig's: 1,2,3)

In CNS examination higher functions and cranial nerve examination were normal. Tone in all the four limbs was normal. Bilateral biceps, triceps, supinator, knee and ankle reflexes were brisk. Plantars were bilateral extensors. There was

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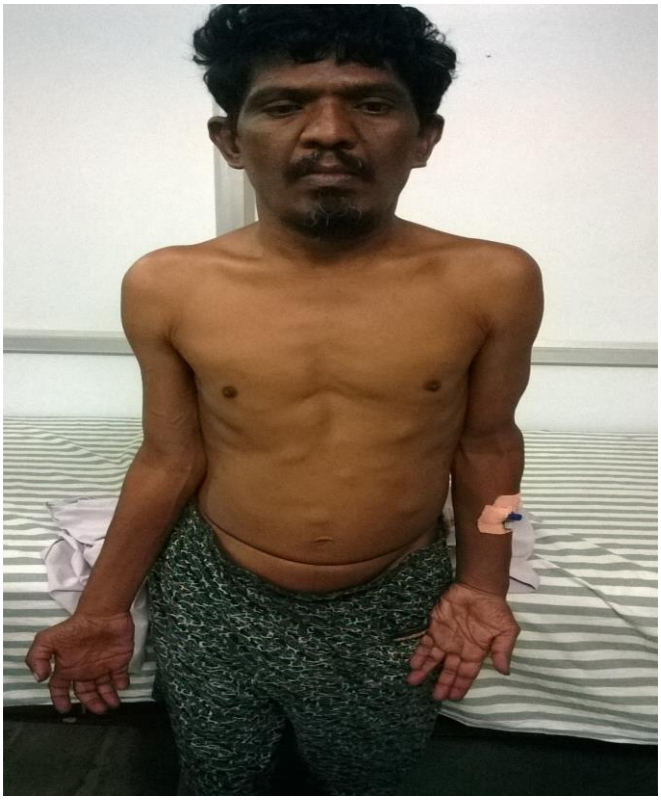
muscle wasting in both the C5 root distribution. There was no sensory deficit.

Investigations:

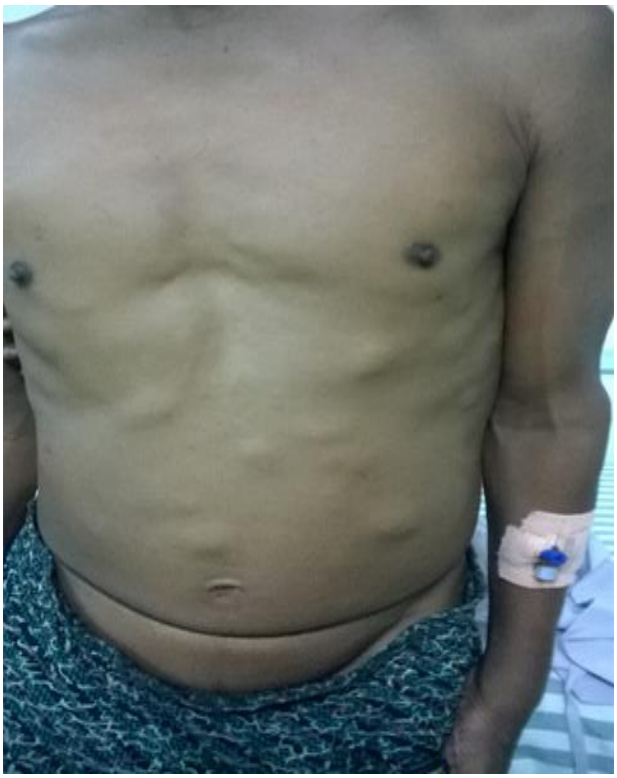
Magnetic Resonance Imaging (MRI) of the cervical spine and whole spine revealed multiple dumbbell shaped tumors at C1-C2 level (Fig's.4 and 5) showing extraforaminal component at C1-C2 level with multiple small neurofibromas at levels C6-C7, C7-T1, T1-T2 on right side and C6-C7, C7-T1, T1-T2 on left side with these features suggestive of neurofibromatosis.

USG right lower limb revealed multiple rounded hypoechoeic lesions scattered throughout subacute plane in thigh, calf and paraspinal regions suggestive of plexiform neurofibromas.

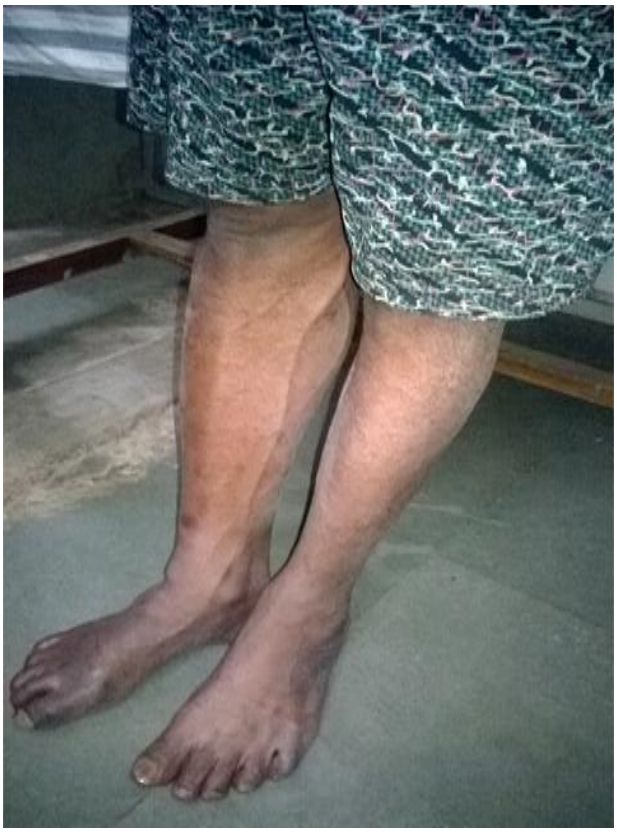
The patient was diagnosed as neurofibromatosis with extramedullary intradural cord compression. The patient underwent laminectomy with decompression and tumor removal was done (Fig.7).



(Fig.1) Multiple swellings over trunk and extremities.



(Fig.2) Multiple swellings over trunk.



(Fig.3) Right calf hypertrophy.

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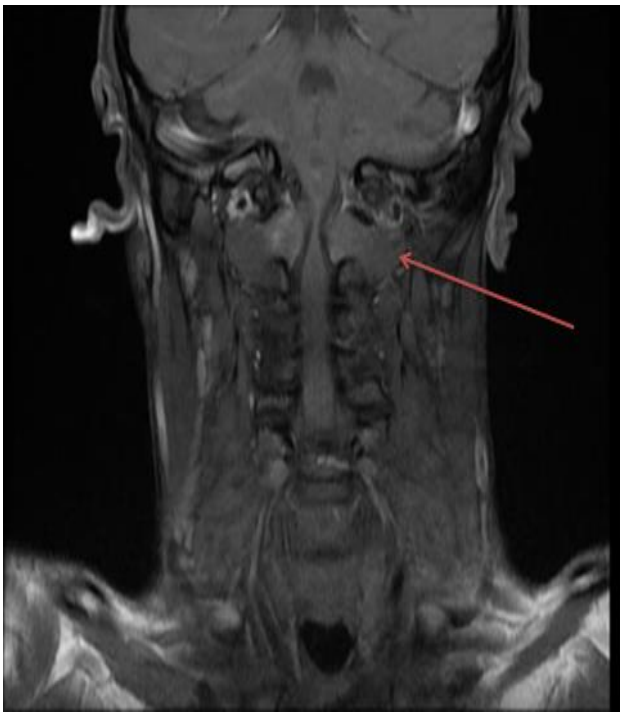
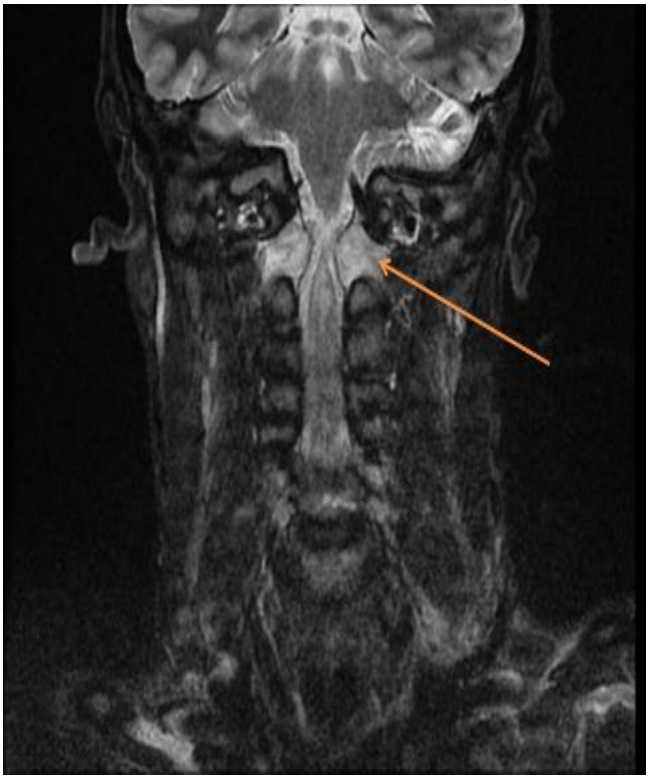
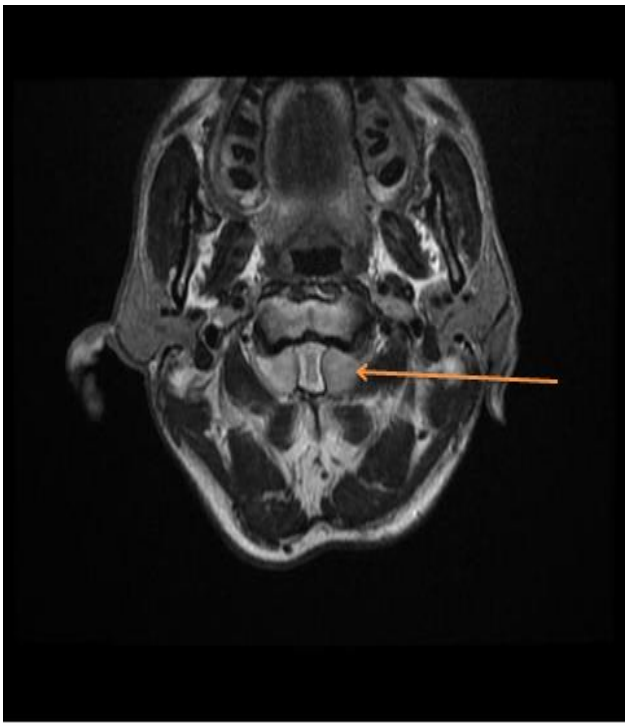


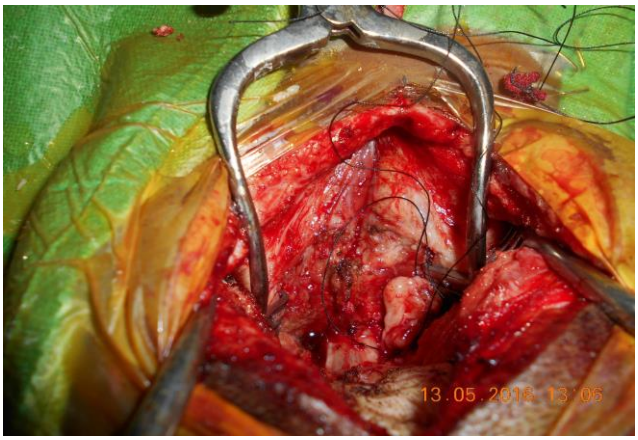
Fig.4: MRI saggital section showing bilateral dumbbell shaped neurofibromas at C1-C2 level.

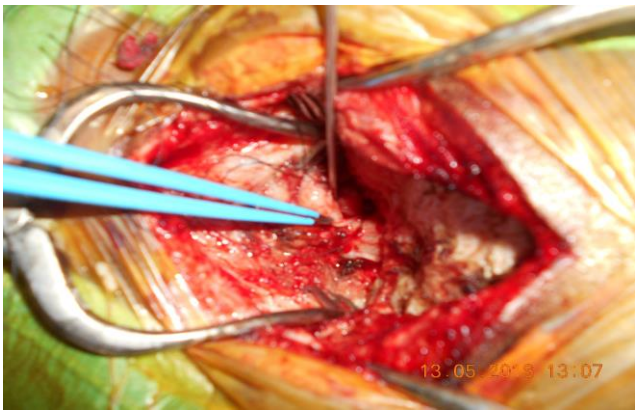


(Fig.5) MRI saggital section showing bilateral dumbbell shaped neurofibromas at C1-C2 level with contrast.



(Fig.6): MRI coronal section showing dumbbell shaped neurofibromas.





(Fig.7) Intraoperative images showing bilobed dumbbell shaped neurofibroma's at C1-C2 level.

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Discussion:

Leonard et al. (2007) [1] have reviewed approximately 1500 patients with NF1 followed-up by two major centers between the years 1996 to 2006 in their retrospective study. The cervical cord compression was detected in 13 patients aged between 9 and 61 years. The cervical cord compression was at the levels of C2 and C3 in majority of the patients. The 7 patients had progressive quadriparesis while totally 3 patients had paraparesis, found at lower extremity in 2 patients and at upper extremity in 1 patient, also incontinence was detected in 1 patient while 3 patients without kyphotic deformity had complaints of cervical pain. The 11 of 13 patients were had undergone a single and multiple-level cervical laminectomy and a subtotal resectomy for neurofibromas. A secondary operation was required in 2 patients by the advancing time.

Créange et al. (1999)[2] have reviewed approximately 158 patients with NF1 and they found spinal cord compression or cauda equine syndrome in five patients. Despite the surgery, two patients with cervical cord compression developed quadriplegia and one of them died. Quadriplegia and neurological sphincter dysfunction were developed in two patients because of cauda equina syndrome. Intraspinal neurofibromatosis did not show any progression in 1 patient for 7-year follow-up (Créange et al., 1999).
In patients with NF1, the cervical cord compression occurs due to compression of neurofibromas to the cervical nerve root in the patients diagnosed NF1. However, they have not been frequently reported

Mustafa GÜLER et al (2013)[3] in their study aimed to discuss treatment approaches by evaluating a patient with complex case of neurofibromatosis in whom whole spinal cord is full of neurofibromas accompanied by cervical cord compression[3].

The rarity in our case scenario is that a case of multiple neurofibroma's presenting with quadriparaesis due to cervical cord compression is an infrequent finding along with unilateral calf swelling due to neurofibroma invading the muscle.
There is limited data about the literature on the clinical features, treatment and prognosis of patients having NF1 with either cervical or lumbar cord compression from plexiform neurofibromas and the individual case reports about the same are also rare.

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