

Original Research Article

Imaging in neurofibromatosis type 1: An original research article with focus on spinal lesions

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Abstract

Background: Neurocutaneous syndromes encompass a group of disorders that affect the embryonic ectodermal plate, which includes the central and peripheral nervous systems, as well as the overlying skin.

Materials and methods: All patients were known case of neurofibromatosis type 1. All patients for this study had undergone MRI whole spine with brain screening. Conventional radiographs were also taken when and where needed after taking informed consent.

Results: In our study we found that 15 (60%) patients having lumbar neurofibromas, 7 (28%) patients having cervical neurofibromas, 2 (8%) patients having thoracic neurofibromas and 1 (4%) patients having sacral neurofibromas.

Conclusion: MRI allows non-invasive detection of spinal neurofibromas and helps determine intradural or extradural origin and their relationship to the neural foramina, spinal canal, and thecal sac and spinal cord. The target sign (bright on T2, with dark collagen centrally) is highly suggestive of a peripheral neurofibroma on MRI.

Key words

MRI, Neurofibromatosis type 1, Spinal lesions.

Introduction

Neurocutaneous syndromes encompass a group of disorders that affect the embryonic ectodermal plate, which includes the central and peripheral nervous systems, as well as the overlying skin. Mesodermal and endodermal structures may be involved, depending on the type and severity of the specific neurocutaneous syndrome. Common developmental abnormalities include dysplasia and often an increased incidence of neoplasms.

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is the most common of the neurocutaneous syndromes with an incidence of approximately 1 in 2,600 to 1 in 3,500 live births [1-3].

Approximately half of all cases result from spontaneous mutations of the NF1 gene. The genetic defect affects chromosome 17q12 and results in decreased production of neurofibromin, which acts as a tumor suppressor [4, 5].

The disease affects the brain, skull, orbits, spine, musculoskeletal system, and skin/integumentary system, although there is significant variability in the type and severity of clinical manifestations. Diagnostic criteria for NF1 include the presence of 2 or more of the following: 1st degree relative with NF1, 6 or more café-au-lait spots, 2 or more neurofibromas (NFs) or 1 plexiform neurofibroma (PNF), optic pathway glioma, bony dysplasia, axillary or inguinal freckling, and 2 or more Lisch nodules.

Central nervous system (CNS) abnormalities occur in approximately 15%-20% of NF1 patients. Spinal manifestations of NF1 include multiple bilateral intraspinal and para-spinal neurofibromas. Spinal lesions may be intradural, intradural and extradural, or purely extradural.

On imaging, smaller lesions are often T2 hyperintense with prominent enhancement, similar to schwannomas. Larger lesions are more heterogeneous in signal intensity and enhancement patterns. As with NFs elsewhere

and as discussed above, neurofibromas are more likely than schwannomas to demonstrate the “target sign,” which refers to peripheral increased and central decreased T2 signal intensity.

Intra- and extradural lesions extend through and expand the bony neural foramina with benign bony remodeling. Although the majority of cases of malignant peripheral nerve sheath tumors occur in the setting of neurofibromatosis, the estimated incidence of malignant peripheral nerve sheath tumors in NF1 patients is approximately 5%. Additional spinal manifestations include kyphoscoliosis, dural ectasia with posterior vertebral body scalloping, and lateral thoracic meningoceles [6, 7].

Aim and objectives

- To evaluate the radiological appearance and extent of the lesions of spine in NF1 patients.
- To detect additional lesions of neurofibromatosis type 1 in known patients of NF1.
- To detect complications of NF1.

Materials and methods

Study area

The study was carried out in the Department of Radiodiagnosis, S.B.K.S. Medical Institute and Research Centre, Waghodia, Vadodara.

Study design

- Type of the study: An Observational, Descriptive Hospital Based Study.
- Sample size: 21 patients.

Selection of subject

Inclusion criteria

- Only those patients who are willing to participate in study will be included.
- Patients with known case of Neurofibromatosis type 1 are included.
- Patients referred to the radiology department for evaluation of spinal cord

lesions found to have positive findings, will be included in this study.

- Patients coming for investigations for other diseases, and are accidentally found to have neurofibromas of spinal cord will be included in this study.

Exclusion criteria

- All patients unwilling were excluded from this study.

Study protocol

25 Patients with known case of neurofibromatosis 1 were evaluated, Where the Patients had presented with symptoms of spinal cord compression like pain and weakness of legs, urinary retention. Some patients were having additional symptoms including neurocutaneous lesions including café au lait spots. All patients were known case of neurofibromatosis type 1. All patients for this study had undergone MRI whole spine with brain screening. Conventional radiographs were also taken when and where needed after taking informed consent.

Results and Discussion

In our study we found spinal neurofibromas in including 7 children and 18 adult patients. In which 14(56%) patients were male and 11 (44%) patients were female (**Chart - 1**).

In our study we found that 15 (60%) patients having lumbar neurofibromas, 7 (28%) patients having cervical neurofibromas, 2 (8%) patients having thoracic neurofibromas and 1 (4%) patients having sacral neurofibromas (**Chart - 2**).

They appeared as nerve root-associated tumours, out of which, most were extradural, some lesions were intradural extramedullary. Out of intradural extramedullary lesion, some were completely in spinal canal and some were dumbbell shaped and cause widening of intervertebral foramina and extend outside spinal canal.

Some intradural extramedullary lesion cause spinal cord compression. They could appear at

different levels of the vertebral column either unilaterally or bilaterally and single or multiple. The size of the tumours varied from a diameter of a few millimeters to several centimeters, and the largest ones were located in the lumbar area.

Chart – 1: Showing sex distribution in study.

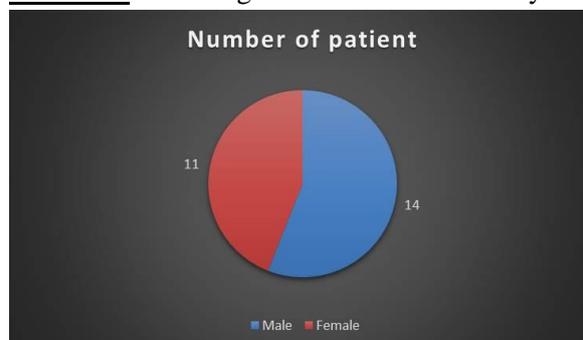
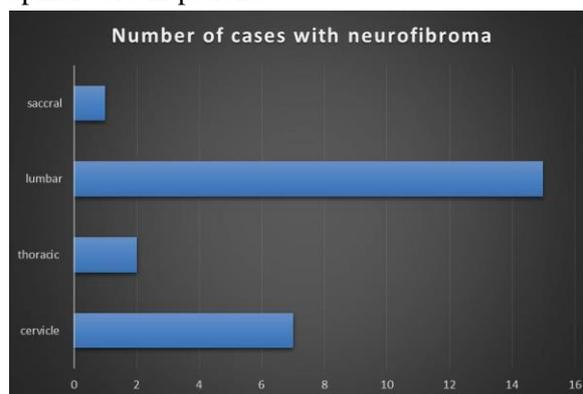


Chart – 2: Showing level of involvement of spinal cord in patients.



On T2-weighted MR images the tumours appeared hyperintense compared with the cord, while on T1-weighted images they were hypointense. Their contrast enhancement in MRI was striking. Compression of the cervical cord due to tumours was seen in seven patients.

On conventional radiograph we had found two cases with classical ribbon ribs appearance with bone remodeling and scoliosis of thoracic spine which are radiological features of neurofibromatosis type 1.

Conclusion

MRI allows noninvasive detection of spinal neurofibromas and helps determine intradural or extradural origin and their relationship to the neural

foramina, spinal canal, and thecal sac and spinal cord. **Image – 3:** T2 saggital showing heterogeneous cord. The target sign (bright on T2, with dark lobulated intra dural extra mudullary lesion collagen centrally) is highly suggestive of a compressing spinal cord. peripheral neurofibroma on MRI.

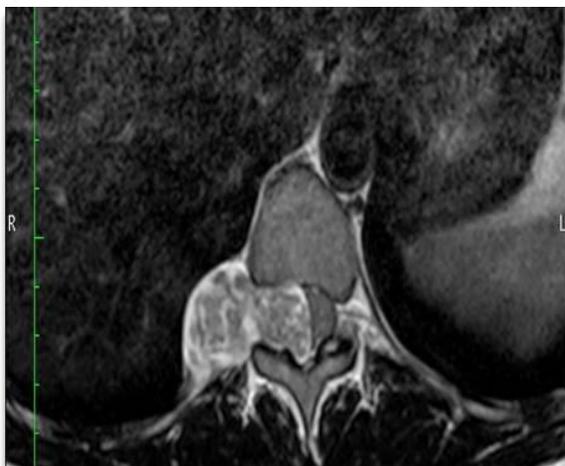
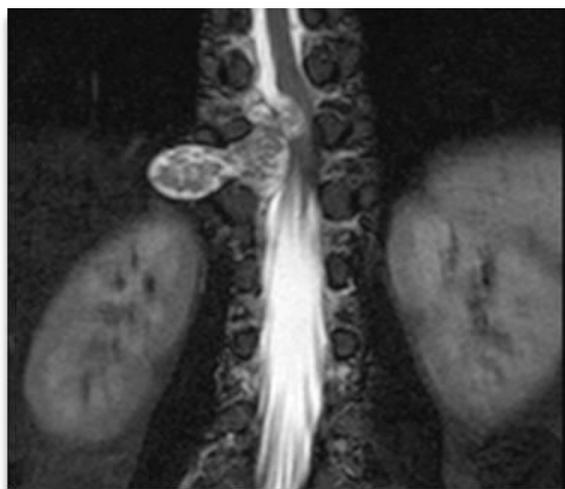
It may be seen with plexiform neurofibromas as well. Additional small neurofibromas can be well depicted with the help of mri. Complications of neurofibroma like spinal cord compression, cord edema, scoliosis, intralesional hemorrhage and necrosis can be well detected with MRI.

Conventional radiographs are also very helpful for diagnosing NF 1, classical ribbon ribs appearance with deformity of spinal column and pseudoarthrosis like features are easily diagnosed on conventional radiographs.



Case - 1: Neurofibroma involving thoracic spine.

Image – 1, 2: Dumbel shaped intra dural extra medullary lesion causing widening of neural foramen.



Case - 2: Neurofibroma involving para spinal region left side upper thorax.

Image – 4, 5, 6: Large heterogeneous para spinal mass, hyper intense on t2w and stir images with intra spinal extension.

Image - 4: T2 axial.

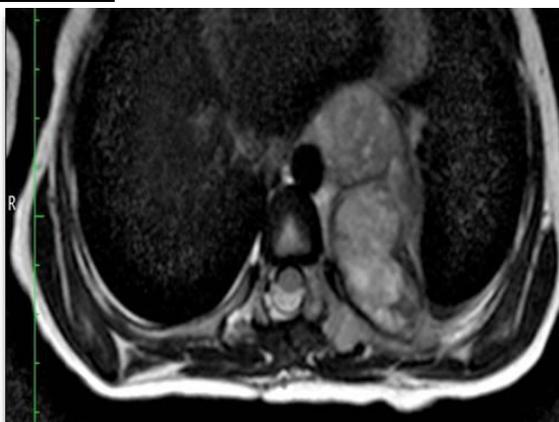


Image – 5: T2 saggital.

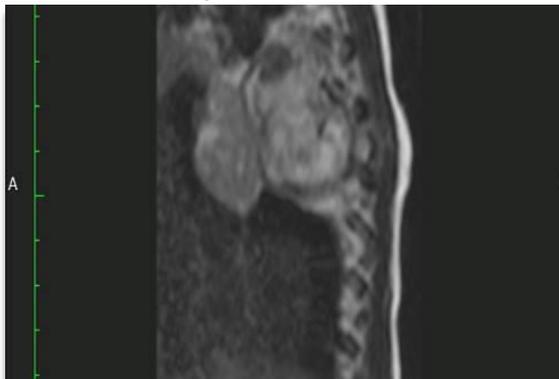


Image – 6: Stir coronal.

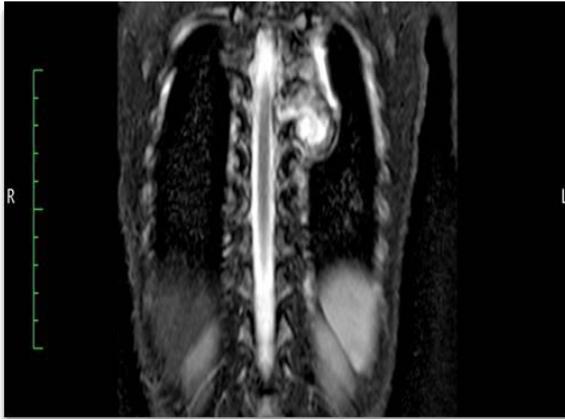


Image – 9: T1 post contrast axial.

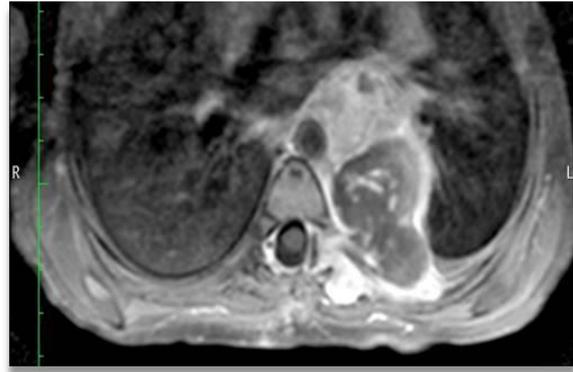


Image – 7, 8 9: Heterogeneous post contrast enhancement of lesion seen.

Image – 7: T1 post contrast coronal.

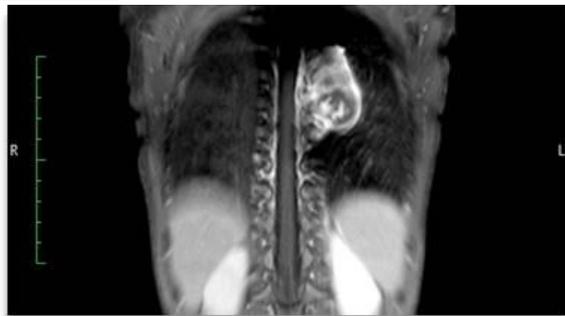
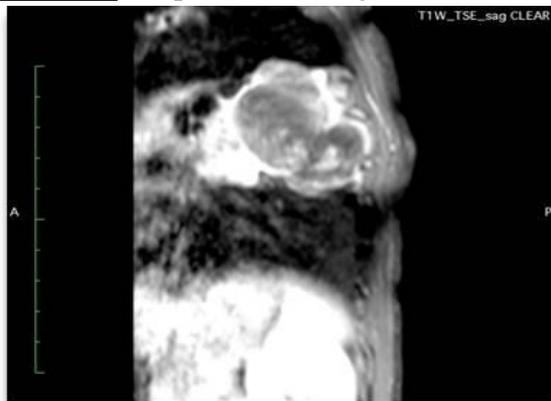


Image – 8: T1 post contrast sagittal.



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