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A Spectrum of Neurofibromatosis Type 1: A Case Series of 6 Patients with Diverse Radiological Manifestations

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ABSTRACT: Neurofibromatosis Type 1 (NF1) is a common autosomal dominant neurocutaneous disorder with a wide spectrum of clinical and radiological manifestations. This case series presents the clinical and magnetic resonance imaging (MRI) findings of six patients with varying presentations of NF1. Spectrum of MRI imaging findings include optic nerve glioma, focal areas of signal intensity (FAI), large lumbar neurofibroma, preseptal neurofibroma, extensive plexiform neurofibromas and intramedullary spinal cord signal changes, multiple neurofibromas and optic nerve involvement, intradural extramedullary neurofibroma at D12 vertebral level. This series illustrates the importance of MRI in diagnosing, characterizing, and monitoring the myriad of central and peripheral nervous system manifestations associated with NF1, ranging from benign FAI to potentially neoplastic lesions like optic gliomas and plexiform neurofibromas.

INTRODUCTION

Neurofibromatosis Type 1 (NF1), also known as von Recklinghausen's disease, is one of the most common autosomal dominant neurocutaneous disorders, with an estimated incidence of 1 in 2,500 to 3,000 live births (1). It is caused by a mutation in the *NF1* tumor suppressor gene on chromosome 17q11.2, leading to the dysregulation of the RAS/mitogen-activated protein kinase (MAPK) signalling pathway and a predisposition to benign and malignant tumor development (2). The clinical diagnosis is based on established National Institutes of Health (NIH) consensus criteria, which include café-au-lait spots, neurofibromas, axillary or inguinal freckling, optic pathway gliomas, Lisch nodules, and distinctive osseous lesions (3).

Magnetic resonance imaging (MRI) plays a pivotal role in the management of NF1 patients. It is crucial for detecting, characterizing, and monitoring the numerous intracranial, spinal, and peripheral manifestations of the disease. These include focal areas of signal intensity (FAI) (also known as unidentified bright objects or UBOs), optic pathway gliomas, neurofibromas (cutaneous, nodular, and plexiform), and malignant peripheral nerve sheath tumors (MPNSTs) (4, 5). This case series aims to illustrate the broad spectrum of MRI findings in seven patients with NF1, highlighting the importance of imaging in diagnosis and management.

CASE PRESENTATIONS:

Case 1: A 27-Year-Old Female with Optic Nerve Glioma

A 27-year-old female with complaints of headache and blurring of vision in right eye underwent an MRI brain. The study revealed few foci of T2/FLAIR hyperintensities in the bilateral globus pallidus, thalamus, and right cerebellar hemisphere,

which were consistent with focal areas of signal intensity (FA SI). The most significant finding was a fusiform dilatation of the posterior intraorbital portion, intracanalicular, and intracranial part of the right optic nerve, extending up to the level of the chiasm, suggestive of an optic nerve glioma (**Figure 1**). Multiple cutaneous neurofibromas were noted throughout the skull (**Figure 2**). A whole spine screening showed mild degenerative disc disease.

Case 2: A 70-Year-Old Female with a Large Lumbar Neurofibroma

A 70-year-old female presented with a large, well-defined, multiloculated, dumbbell-shaped, intradural extramedullary lesion in the right paravertebral region of the L1 vertebra (**Figure 3A, 3B**). The lesion is causing erosion and scalloping of the posterior elements. It is oriented along the right L1 exiting nerve root and is indenting the right kidney and crossing the midline to displace the cord to the left. The mass was hypointense on T1 and isointense on T2 with internal hyperintensities. Differential diagnoses of neurofibroma/sarcoma or metastasis were considered, with a recommendation for biopsy and PET-CT. Incidental findings included a right distal ureteric calculus with hydronephrosis.

Case 3: A 13-Year-Old Male with FAS I

A 13-year-old male with a known clinical history of neurofibromatosis type 1 (NF1) underwent an MRI brain. The study showed nearly symmetrical T2/FLAIR hyperintensities in the bilateral thalami and the right cerebellar peduncle, consistent with characteristic FAS I (**Figure 4**). No other significant intracranial or spinal abnormalities were detected.

Case 4: A 4-Year-Old Male with FAS I and Preseptal Neurofibroma

A 4-year-old male with a known history of café-au-lait spots on the trunk was examined. MRI brain with orbits revealed multiple T2/FLAIR hyperintensities involving the brain stem, bilateral basal ganglia, thalami, and cerebellar hemispheres, consistent with FAS I (**Figure 4A**). The largest lesion in the midbrain measured 7.5×8.9 mm. Additionally, preseptal soft tissue swelling with septations was noted involving the anterior aspect of the left lateral globe. This lesion showed no diffusion restriction and was diagnosed as a neurofibroma (**Figure 4B**), with a note of the mother having similar complaints.

Case 5: A 13-Year-Old Male with Extensive Plexiform Neurofibromas

A 13-year-old male presented with diffuse and extensive manifestations of NF1. MRI brain and whole spine revealed:

- **Intracranial:** Diffuse punctate T2 FLAIR hyperintensities in the deep white matter, dentate nucleus, right gangliocapsular region, and splenium of the corpus callosum (FAS I). Linear and nodular T2 FLAIR hyperintense lesions were seen in the subgaleal plane of the occipital region (**Figure 5**).
- **Cranial Nerves and Cervical Spine:** Enlargement of the 9th, 10th, and 11th cranial nerve root complexes with fusiform hyperintensities extending into the perivertebral regions. Multiple fusiform T2 hyperintensities along the cervical exiting nerve roots with foraminal widening and extension into pre-vertebral, para-vertebral, and posterior cervical spaces.
- **Spine:** Multiple fusiform T2 hyperintensities along the entire spine in the region of the neural foramen with extra-foraminal extension into the para-vertebral and pre-sacral regions. A long segment T2 intramedullary hyperintensity was seen occupying the cord across all spinal levels, raising concern for an underlying intrinsic cord lesion. These findings were all suggestive of widespread plexiform neurofibromas.

Case 6: A 62-Year-Old Female with Multiple Neurofibromas and Optic Nerve Involvement

A 62-year-old female underwent an MRI study that showed multiple soft tissue intensity lesions of varying sizes in the scalp and face, consistent with neurofibromas (**Figure 2**). A small intraconal T2 FLAIR hyperintense signal showing moderate enhancement on T1 post-contrast images was noted along the lateral aspect of the right optic nerve intraorbital segment, raising the possibility of a neurofibroma (**Figure 1**). Intracranial findings included mild prominence of sulcal spaces, few punctate T2 FLAIR hyperintensities in the frontal and parietal white matter and pons (suggestive of small vessel ischemic change), and a small T2 and GRE hypointense nodule in the right posteromedial thalamus (possibly an old granuloma or cavernoma).

DISCUSSION

This case series encapsulates the remarkable phenotypic and imaging heterogeneity of neurofibromatosis type 1. The patients range from a 4-year-old child with early signs of the disease to a 70-year-old adult with a large, complex spinal lesion, illustrating

the lifelong and progressive nature of NF1 (2). The findings across the cases can be broadly categorized into three groups: benign developmental lesions (FASI), benign neoplasms (optic gliomas, neurofibromas), and the potential for more aggressive disease.

Focal Areas of Signal Intensity (FASI) were a common finding in our paediatric and young adult patients (Cases 1, 3, 4, 5). FASI are regions of increased signal on T2-weighted and FLAIR sequences, typically located in the basal ganglia, thalamus, cerebellum, and brainstem. They are considered a hallmark of NF1 in children and adolescents, thought to represent areas of disordered myelination or hamartomatous change (6, 7). They are not true neoplasms and often decrease in size or disappear with age, as seen in the adult patients where they were less prominent or absent.

Optic Pathway Gliomas (OPGs) are another key feature of NF1, occurring in approximately 15-20% of affected children (8). Case 1 demonstrates a classic fusiform enlargement of the optic nerve, characteristic of an OPG. While OPGs can be asymptomatic, they can cause vision loss and require close surveillance. Interestingly, Case 6 also showed a potential intraconal optic nerve lesion in an adult, which may represent a new or longstanding stable lesion. This underscores that OPGs can be present in adults with NF1, though they are more common in the paediatric population (5).

The hallmark of NF1 is the development of neurofibromas, benign nerve sheath tumors. This series displays the full spectrum: from cutaneous neurofibromas (Case 1, 6) to nodular neurofibromas (Case 4) and the most challenging form, plexiform neurofibromas. Case 5 exemplifies the devastating potential of plexiform neurofibromas, which are pathognomonic for NF1. These lesions arise from multiple nerve fascicles, leading to a characteristic "bag of worms" appearance on MRI, with fusiform enlargement along the nerve distribution (9). In Case 5, the extensive involvement of cranial nerves, cervical and spinal nerve roots, and extension into the mediastinum and paravertebral spaces highlights their infiltrative nature. Importantly, plexiform neurofibromas carry a lifetime risk (8-13%) of transformation into malignant peripheral nerve sheath tumors (MPNSTs), a leading cause of mortality in NF1 patients (10). The long segment intramedullary T2 hyperintensity in this patient is a concerning finding that warrants further evaluation with contrast-enhanced MRI to rule out an intrinsic cord tumor, another possible complication.

Cases 2 presents with solitary, large spinal nerve sheath tumors. Case 2's dumbbell-shaped lesion, causing scalloping and neural foraminal widening, is highly suggestive of a neurofibroma or schwannoma. The "dumbbell" morphology occurs when the tumor extends through the neural foramen, creating an intra- and extraspinal component (4). The significant size and mass effect in this elderly patient highlight the potential for these benign tumors to cause considerable morbidity. The diagnosis in all cases was strongly suggested by MRI. The imaging features, in conjunction with clinical findings such as café-au-lait spots (Case 4), family history (Case 4), and known clinical diagnoses (Case 3), firmly place these patients within the NF1 spectrum. The recommendations for further evaluation, including contrast MRI (Cases 1, 4, 5), PET-CT (Case 2), and biopsy (Case 2), reflect the standard of care for characterizing indeterminate lesions and monitoring for potential malignancy.

CONCLUSION

This case series of seven patients demonstrates the broad and complex radiological spectrum of neurofibromatosis type 1. MRI is an indispensable tool for the diagnosis, characterization, and surveillance of the diverse manifestations of NF1, ranging from benign FASI and optic gliomas to the more complex and potentially aggressive plexiform neurofibromas. The cases underscore the need for a multidisciplinary approach to management, considering the patient's age, symptomatology, and the specific nature of the lesions identified. Early and accurate imaging diagnosis is critical for guiding treatment, monitoring for complications such as MPNST transformation, and improving long-term outcomes for patients with this multifaceted disorder.

FIGURE LEGENDS

Figure 1: Axial T2-weighted MRI of the orbits shows fusiform dilatation of the right optic nerve involving its intraorbital, intracanalicular, and intracranial segments, extending up to the level of the optic chiasm (arrow). This appearance is suggestive of an optic nerve glioma.

Figure 2: Sagittal FLAIR image of the scalp demonstrates multiple cutaneous neurofibromas (arrows) involving the subcutaneous tissues of the skull.

Figure 3: (A) sagittal and (B) axial T2 weighted images show a well-defined, multiloculated, dumbbell-shaped intradural extramedullary lesion in the right paravertebral region of the L1 vertebra (asterisk), causing erosion of the posterior elements with scalloping of neural foramina.

Figure 4: (A) Axial T2 image with fat suppression reveals multiple hyperintense foci involving the brain stem, bilateral thalamic regions, and bilateral cerebellar hemispheres (arrows), consistent with focal areas of signal intensity (FAST) in a patient with neurofibromatosis type 1. (B) sagittal T2-weighted image with fat suppression of the orbits shows a preseptal soft tissue swelling involving the anterior aspect of the left lateral globe (arrow). The lesion shows no restriction on diffusion-weighted imaging, consistent with a neurofibroma.

Figure 5: sagittal FLAIR image demonstrates linear and nodular hyperintense lesions in the subgaleal plane of the right occipital region (arrows).

REFERENCES

- [1] Friedman JM. Epidemiology of neurofibromatosis type 1. *Am J Med Genet.* 1999;89(1):1-6.
- [2] Gutmann DH, Ferner RE, Listernick RH, Korf BR, Wolters PL, Johnson KJ. Neurofibromatosis type 1. *Nat Rev Dis Primers.* 2017;3:17004.
- [3] Neurofibromatosis. Conference statement. National Institutes of Health Consensus Development Conference. *Arch Neurol.* 1988;45(5):575-8.
- [4] Ahlawat S, Fayad LM, Khan MS, et al. Current update on imaging of neurofibromatosis type 1. *Radiographics.* 2017;37(4):1173-1191.
- [5] Miller DT, Freedenberg D, Schorry E, et al. Health supervision for children with neurofibromatosis type 1. *Pediatrics.* 2019;143(5):e20190660.
- [6] Sevik RJ, Barkovich AJ, Edwards MS, et al. Evolution of white matter lesions in neurofibromatosis type 1: MR imaging findings. *Radiology.* 1992;182(2):511-6.
- [7] Griffiths PD, Blaser S, Boodram MB, et al. Focal areas of high signal intensity in children with neurofibromatosis type 1: a classification system and its relationship to clinical outcome. *AJNR Am J Neuroradiol.* 1998;19(5):967-73.
- [8] Listernick R, Charrow J, Greenwald MJ, Esterly NB. Optic gliomas in children with neurofibromatosis type 1. *J Pediatr.* 1989;114(5):788-92.
- [9] Poussaint TY, Jaramillo D, Chang Y, Korf B. MR imaging of the spine in neurofibromatosis type 1: a review. *AJR Am J Roentgenol.* 1996;166(5):1149-55.
- [10] Evans DG, Baser ME, McGaughan J, Sharif S, Howard E, Moran A. Malignant peripheral nerve sheath tumours in neurofibromatosis 1. *J Med Genet.* 2002;39(5):311-4.

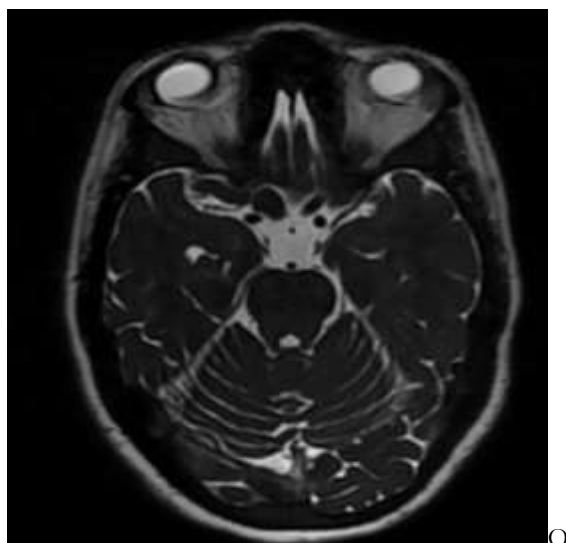


Figure 1

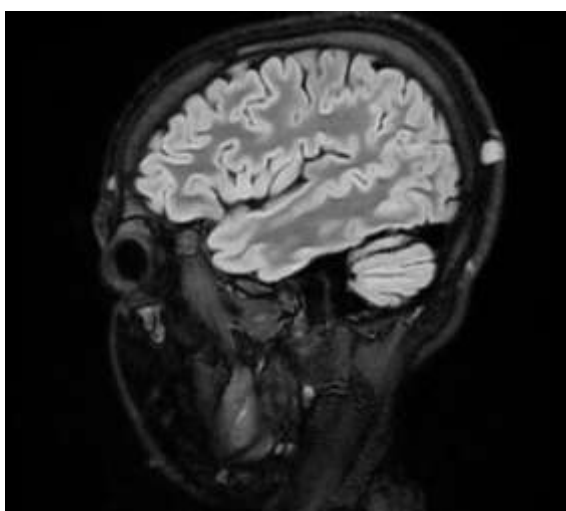


Figure 2

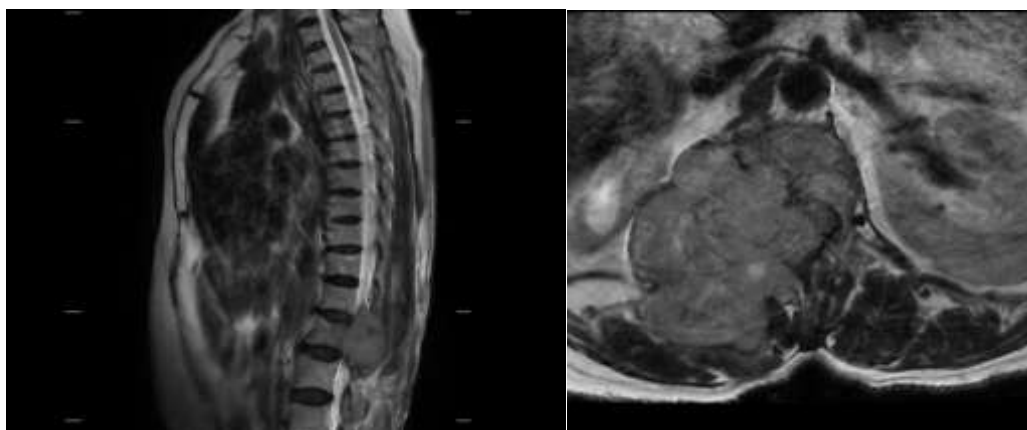


Figure 3A & 3B

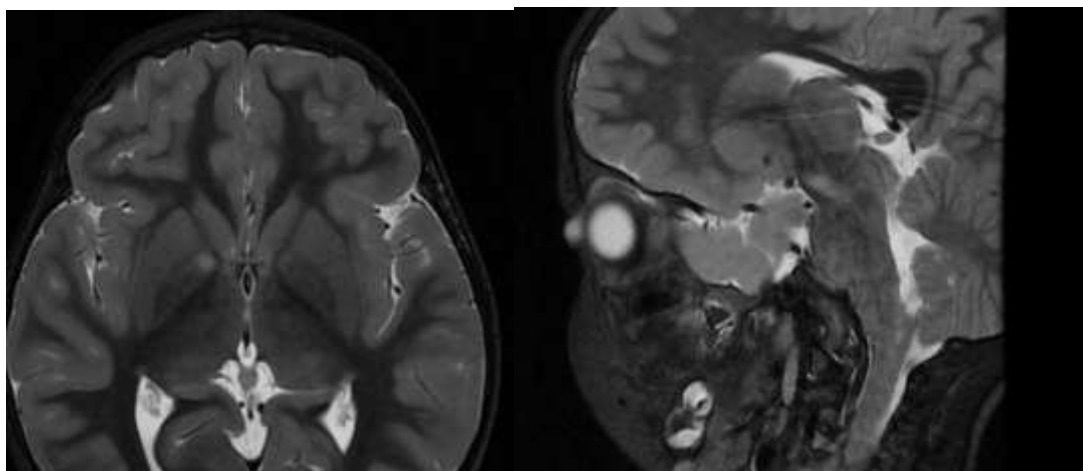


Figure 4



Figure 5