Neurofibromatosis type 1: a review of MRI findings

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Learning objectives

To illustrate and analyze radiological findings of neurofibromatosis type 1 involving central and periferical nervous system on MR.

Background

Nerofibromatosis type 1 (NF1), also known as Von Recklinghausen's disease, is the most common neurocutaneous disorder.

NF1 is inherited as an autosomal dominant disorder with variable penetrance, caused by defects in the NF1 gene on chromosome 17 responsible for encoding the protein neurofibromin. This protein is a tumor suppressor, acting as a negative regulator of the Ras family guanine triphosphates (GTPases). Patients with NF1 usually present during the first decade of life. NF1 is a complex disorder, affecting multiple cell types and multiple systems of the body, with a wide range of expression and unpredictable behavior.

The diagnosis is based on an individual demonstrating at least 2 of the following 7 clinical criteria: 6 or more café-au-lait spots (pubertal dependent size dimension); 2 or more neurofibroma, or a plexiform neurofibroma, axillary or inguinal freckling; distinctive osseous lesions; optic nerve glioma; more than 2 iris Lisch nodules; or a first degree relative with NF1. No neuroimaging findings are included in the diagnostic criteria for NF1.

Findings and procedure details

We have collected neurofibromatosis patients diagnosed database of a tertiary hospital and we selected those who had lesions characteristic of this disease. We have described its characteristics as well as the RM sequence we used in each case.

The main radiological manifestations of the neurofibromatosis type 1 are:

1. Tumors.

Astrocytomas are the main type of CNS tumor in children with NF1, and pilocytic astrocytoma (World Health Organization grade 1) is the main histologic type. Although
these tumors may originate in any part of the brain, the optic pathways and brainstem are most often affected (Fig. 1 on page 5).

1.1 Optic pathway gliomas (Fig. 2 on page 5) are the most common tumor of the CNS in NF1. These gliomas may involve any portion of the optic pathway, including one or both optic nerves, the chiasm, optic tracts, the lateral geniculate bodies, or the optic radiations.

Optic nerve gliomas manifest as enlargement of the optic nerve sheath complex with a configuration that can be tubular, fusiform, eccentric, or globular with kinking and tortuosity. These lesions are usually hypointense on T1 images and hyperintense on T2 images with homogenous enhancement, if present. Heterogeneous enhancement may be seen when the tumors are large.

1.2 Brainstem gliomas in NF1 can be detected in approximately 9% of patients. Most brainstem gliomas in NF1 have a relatively benign and indolent course and may stabilize or regress. These lesions are isointense on T1 images and hyperintense on T2 images. Enhancement may be variable.

Tectal glioma (Fig. 3 on page 6) can occur in NF1 patients presenting with hydrocephalus caused by obstruction at the aqueduct of Sylvius.

1.3 Cerebellar gliomas (Fig. 4 on page 6) are rare in association with NF1 and have been considered to be malignant in a high proportion of cases.

2. Areas of myelin vacuolization or neurofibromatosis spots (Fig. 5 on page 7).

These lesions are foci of hyperintense T2 signal intensity. NF spots are found in 43% to 93% of pediatric NF1 patients. These foci are typically located in the basal ganglia, internal capsula, brainstem, and cerebellum. The globus pallidus has often been reported as the commonest site, usually with bilateral involvement. These foci appear by age 3 years and increase in number and size until about 10 to 12 years of age. They then decrease in size and are uncommon after the age of 20. These lesions do not enlarge, do not enhance, do not cause mass effect or clinical symptoms, and do not require treatment.

3. Cerebrovascular abnormalities

Cerebrovascular abnormalities in association with NF1 are uncommon. The types of abnormalities that can be seen include stenoses, occlusions, moyamoya disease, aneurysms, internal carotid dolichoectasia (Fig. 6 on page 7), arteriovenous
malformations, and fistulae. In moyamoya, the internal carotid artery is the most commonly involved intracranial vessel with obstruction of the supraclinoid portion (Fig. 7 on page 8). Symptoms are caused by cerebral ischemia or infarction.

4. Bone dysplasia: sphenoid and orbit

Sphenoid bone dysplasia is a distinctive, although uncommon, manifestation of NF1. This abnormality is a defect in the greater sphenoid wing and enlargement of the middle cranial fossa and was well described radiologically long before CT was available. These patients may present with pulsatile exophthalmos related to herniation of the temporal lobe into the orbit. Sphenoid dysplasia can be seen isolated or associated with an underlying plexiform neurofibroma (PNF) (Fig. 8 on page 9). A modified concept of sphenoid dysplasia that emphasizes the interaction between neurofibromas and sphenoid bone during skull development has been proposed.

5. Macrocephaly

Macrocephaly is defined as the fronto-occipital circumference greater than or equal to 2 SD above the mean for sex and age. Macrocephaly unrelated to CNS pathology (eg, hydrocephalus) is observed in patients with NF1, and it has been demonstrated that NF1 is a true macrocephaly syndrome.

6. Neurofibromas

Neurofibromas are benign peripheral nerve sheath tumors comprised of a heterogeneous mixture of cellular elements. Neurofibromas can arise anywhere in the body along the nerve and can cause a significant degree of morbidity, mortality, and cosmetic disfigurement.

The classification of neurofibromas includes discrete cutaneous neurofibromas of the dermis or epidermis, discrete subcutaneous neurofibromas that lie deeper in the skin, deep nodular neurofibromas, and diffuse PNFs (Fig. 8 on page 9). PNFs are characterized by longitudinal growth along nerves, involving multiple fascicles and branches, and may cause disfigurement, overgrowth, nerve compression, and malignancy. On MR imaging, the PNFs are of low signal intensity on T1-weighted images and are hyperintense on T2 images. Enhancement after the administration of paramagnetic contrast is variable.

7. Spinal manifestations
Spinal manifestations of NF1 include scoliosis with or without kyphosis; meningeal defects, ranging from dural ectasia, causing vertebral scalloping, to meningoceles; congenital vertebral defects; and secondary vertebral involvement from paraspinal neurofibromas (Fig. 9 on page 9).

**Images for this section:**

![Fig. 1: Temporal lobe pilocytic astrocytoma in NF1. (A) Coronal T2 images demonstrates a cystic mass with a solid component mural. (B) Postgadolinium coronal T1 images shows marked enhancement of the solid component.](image-url)
**Fig. 2:** Bilateral optic pathway glioma. Axial T2, coronal T2 y sagital T1 with contrast MR images demonstrate fusiform enlargement of the right optic nerves that extends to the orbital apex.

**Fig. 3:** Glioma tectal with obstructive hydrocephalus. Sagital T1 MR image without contrast demonstrate expansion the tectum and there is marked enlargement of the lateral and third ventricles. The aqueduct and fourth ventricle are small. This patient shows changes after endoscopic third ventriculostomy.
Fig. 4: Cerebellar glioma in NF1. (A) Axial T2 MR image demonstrates a lesión hiperintense situated in the cerebellar vermis. (B) Sagital T1 RM image shows a area of low signal intensity. (C) Sagital T1 postgadolinium RM image shows that the area abnormality has enhanced.

Fig. 5: Myelin vacuolization or neurofibromatosis spots. (A) Axial T2 MR image demonstrates multiple NF spots in the cerebellum and pons. (B) Axial T2 MR image demonstrates foci of T2 prolongation in the globus pallidi. Occasionally these foci may be slightly hyperintense on T1 images (C), particularly in the basal ganglia.
Fig. 6: Cerebrovascular abnormalities. Sequence 3D TOF MIP axial and frontal view demonstrate right internal carotid dolichoectasia.
**Fig. 7:** Cerebrovacuolar abnormalities. Axial 3D TOF of demonstrates severe stenooclusive changes of the right supraclinoid and petrous internal carotid artery.

**Fig. 8:** Spheno-orbital wing dysplasia in NF1. Axial T2 MR image demonstrates a left spheno-orbital plexiform neurofibroma causing proptosis, widening, and deformity of the orbit and sphenoid wing.
Fig. 9: (A) Coronal shows multiple foraminal and paraspinal neurofibromas. (B) Axial T2 images demonstrates an extensive mass involving the pterygoid fossa and masticator space. The center of the lesion is often of low signal intensity and corresponds pathologically to a dense central core of collagen, referred to as the target sign (arrow).
Conclusion

Neuroradiological findings constitute an important basis in the diagnosis, influencing the clinical accuracy and the genetic counsel in these patients. Particularly, MR imaging plays an important role in the diagnosis and management of the patient with neurofibromatosis type 1.

Personal information

References


