

## Case 2117: Optic pathways glioma in neurofibromatosis type 1: MRI findings

Subspecialty: Neuroradiology

Date of Publication: 2004.01.13

Zuccoli G, Della Giustina E, Guidetti D, Ferrozzi F, Nicoli F  
Arcispedale Santa Maria Nuova  
42100 - Reggio Emilia / ITALY

### Patient:

Age: 5 year(s)

Sex: male

### Clinical Summary

A patient with neurofibromatosis type I underwent a screening MR scan of the brain

### Clinical History and Imaging Procedures

A patient with neurofibromatosis type 1 underwent a screening MR scan of the brain. This showed enlargement of the right and left optic nerve with contrast enhancement (Fig.4a and 5a). The lesions appeared to be slightly hyperintense on T2-weighted images. The chiasma was enlarged and enhanced on contrast administration. No associated hamartomatous lesions were seen.

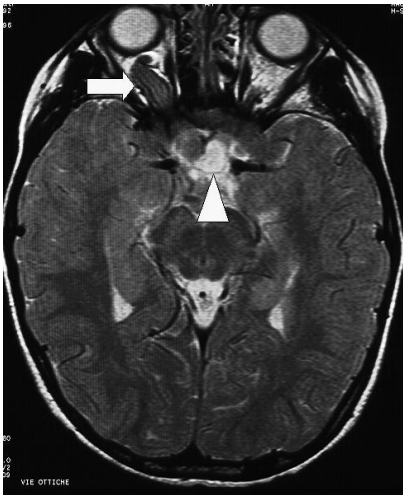
### Discussion

In 1987 the Consensus Development Conference at the National Institutes of Health (NIH) on Neurofibromatosis established the diagnostic criteria for NF1. Even though the neurofibromatosis type 1 (NF1) gene has been cloned and its protein product identified, the diagnosis of NF1 is still based on the NIH clinical criteria. The presence of an optic pathway tumour (OPT) is included among the NF1 diagnostic criteria. OPTs occur both in male and females with a slight prevalence for female patients. Children with NF1 and optic pathway gliomas frequently have visual abnormalities such as abnormal pupillary function, decreased visual acuity, decreased colour vision or optic atrophy at the time of OPT diagnosis. A significant association between the presence of an optic tract tumour and a reduction in the visual field has been recently shown, suggesting that a reduction in the visual field may indicate the presence of a post-chiasma optic pathway tumour. The Clinical Care Advisory Board of the National Neurofibromatosis Foundation and an NF1 Optic Pathway Glioma Task Force recommend an optic pathway MRI study of all children with any evidence of optic nerve disorders. However, screening with MRI has not been shown to improve the clinical outcome of these patients. MR imaging is the diagnostic modality of choice for OPT. These tumours show typical patterns on MR imaging. The tumour develops along the optic pathways, more often bilaterally, enlarging the optic nerves and the chiasma. Contrast enhancement is generally present. On T2-weighted images the tumour usually shows focal areas of high signal intensity with respect to normal brain parenchyma. Therapy tends to be conservative; in fact a spontaneous regression of OPT has been described by some authors. Changes in the signal intensity and a reduction in contrast enhancement of OPTs have been considered to be an index of their biological activity.

### Final Diagnosis

Optic pathways glioma in neurofibromatosis type 1 patient

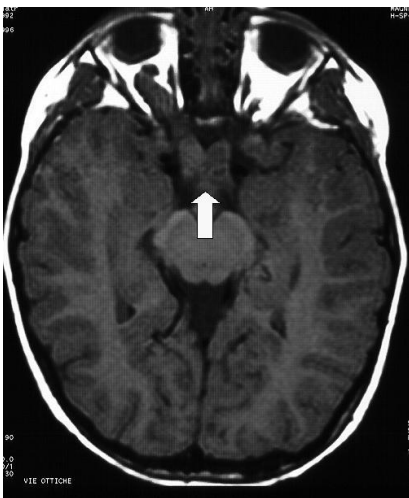
**Figure 1:** T2-weighted image



**Figure 1a**

Axial T2-weighted image shows the presence of an optic pathways tumour involving the chiasma (arrowhead) and the right optic nerve, which appears to be tortuous (arrow).

**Figure 2:** Axial T1-weighted image



**Figure 2a**

Axial pre-contrast T1-weighted image confirms morphological changes along the anterior optic pathways. Note the chiasma enlargement (arrow).

**Figure 3:** Axial proton density image

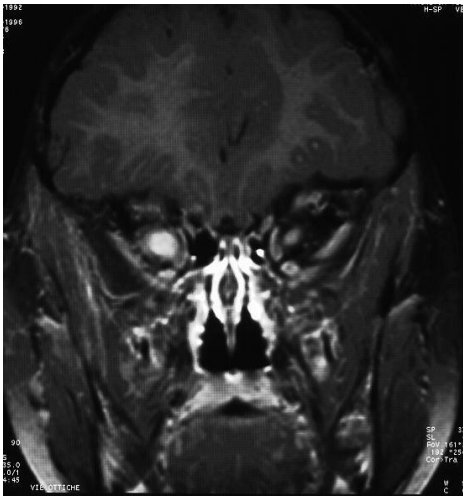


**Figure 3a**

Axial proton density (PD) image shows enlargement of the chiasma and intra-cranial optic nerves (arrow).

**Figure 4:** Contrast-enhanced T1-weighted image

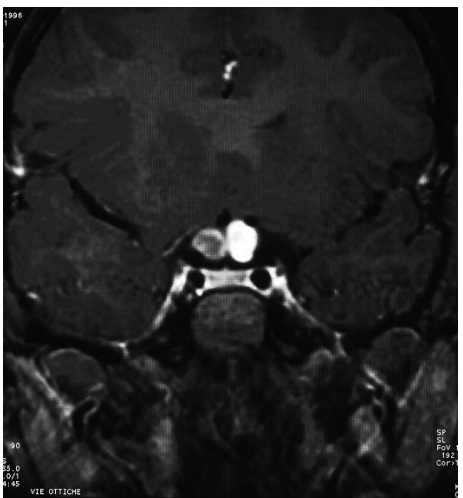
**Figure 4:** Contrast-enhanced T1-weighted image



**Figure 4a**

Coronal T1-weighted image shows the homogeneous contrast enhancement of the right extra-cranial optic nerve.

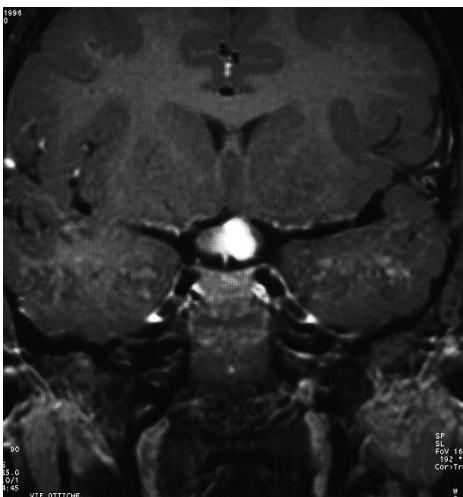
**Figure 5:** Contrast-enhanced T1-weighted image



**Figure 5a**

Coronal T1-weighted image shows the asymmetric contrast enhancement of both optic nerves at the pre-chiasma level.

**Figure 6:** Contrast-enhanced T1-weighted image



**Figure 6a**

The chiasma is enlarged and contrast enhanced.

**MeSH:**

[C04.557.580.600.580.590.650] Neurofibromatosis 1

An autosomal dominant inherited disorder (with a high frequency of spontaneous mutations) that features developmental changes in the nervous system, muscles, bones, and skin, most notably in tissue derived from the embryonic NEURAL CREST. Multiple hyperpigmented skin lesions and subcutaneous tumors are the hallmark of this disease. Peripheral and central nervous system neoplasms occur frequently, especially OPTIC NERVE GLIOMA and NEUROFIBROSARCOMA. NF1 is caused by mutations which inactivate the NF1 gene (GENES, NEUROFIBROMATOSIS 1) on chromosome 17q. The incidence of learning disabilities is also elevated in this condition. (From Adams et al., Principles of Neurology, 6th

ed, pp1014-18) There is overlap of clinical features with NOONAN SYNDROME in a syndrome called neurofibromatosis-Noonan syndrome. Both the PTPN11 and NF1 (GENES, NF1) gene products are involved in the SIGNAL TRANSDUCTION pathway of Ras (RAS PROTEINS).

[C04.557.465.625.600.380.795] Optic Nerve Glioma

Glial cell derived tumors arising from the optic nerve, usually presenting in childhood. Roughly 50% are associated with NEUROFIBROMATOSIS 1. Clinical manifestations include decreased visual acuity; EXOPHTHALMOS; NYSTAGMUS; STRABISMUS; pallor or swelling of the optic disc; and INTRACRANIAL HYPERTENSION. The tumor may extend into the optic chiasm and hypothalamus. (Adams et al., Principles of Neurology, 6th ed, p681)

[C04.557.465.625.600.380.080.667] Optic Nerve Glioma

Glial cell derived tumors arising from the optic nerve, usually presenting in childhood. Roughly 50% are associated with NEUROFIBROMATOSIS 1. Clinical manifestations include decreased visual acuity; EXOPHTHALMOS; NYSTAGMUS; STRABISMUS; pallor or swelling of the optic disc; and INTRACRANIAL HYPERTENSION. The tumor may extend into the optic chiasm and hypothalamus. (Adams et al., Principles of Neurology, 6th ed, p681)

#### References:

- [1] Gutmann DH, Aylsworth A, Carey JC, Korf B, Marks J, Pyeritz RE, Rubenstein A, Viskochil D. The diagnostic evaluation and multidisciplinary management of neurofibromatosis 1 and neurofibromatosis 2. JAMA. 1997 Jul 2;278(1):51-7.
- [2] Rasmussen SA, Friedman JM. NF1 gene and neurofibromatosis 1. Am J Epidemiol. 2000 Jan 1;151(1):33-40.
- [3] Friedman JM. Epidemiology of neurofibromatosis type 1. Am J Med Genet. 1999 Mar 26;89(1):1-6.
- [4] Sigorini M, Zuccoli G, Ferrozzi F, Bacchini E, Street ME, Piazza P, Rossi M, Virdis R. Magnetic resonance findings and ophthalmologic abnormalities are correlated in patients with neurofibromatosis type 1 (NF1). Am J Med Genet. 2000 Aug 14;93(4):269-72.
- [5] Zuccoli G, Ferrozzi F, Sigorini M, Virdis R, Bassi P, Bellomi M. Early spontaneous regression of a hypothalamic/chiasmatic mass in neurofibromatosis type 1: MR findings. Eur Radiol. 2000;10(7):1076-8.

#### Citation:

**Zuccoli G, Della Giustina E, Guidetti D, Ferrozzi F, Nicoli F (2004.01.13)**

Optic pathways glioma in neurofibromatosis type 1: MRI findings, {Online}

URL: <http://www.eurorad.org/case.php?id=2117>