Imaging considerations of central nervous system manifestations in pediatric patients with neurofibromatosis type 1

F. Menor, L. Marti-Bonmati, F. Mulas, H. Cortina and R. Olagüe
Departments of Diagnostic Radiology, La Fe Children’s Hospital, Valencia, Spain

Abstract. CT and MRI were used in a prospective study of the central nervous system (CNS) manifestations in 41 consecutive children with neurofibromatosis type 1 (NF-1). Gadolinium-DTPA was used in 15 patients. MRI was more effective than CT in delimiting the extension of the optic pathway glioma and in evaluating associated cerebral malformations. MRI visualized lesions generally undetected by CT, in the form of iso- or hyperintense loci with respect to the cerebral cortex in T2-weighted sequences. Well-delimited lesions of high signal intensity were observed in the globus pallidus (22 cases), the internal capsule (6 cases), corpus callosum (2 cases), anterior commissure (1 case) and semioval center (2 cases). Poorly defined hyper- or isointense areas were also observed affecting the cerebellar white matter (21 cases) and brain stem (17 cases). None of these lesions showed Gadolinium-DTPA enhancement, and were of no clinical significance. MRI has displaced CT in the initial diagnosis of patients with NF-1. Periodic annual MRI controls are only justified in patients with MRI changes to evaluate the progression or stabilization of the lesions.

Material and method
A prospective study was made of 41 consecutive children with NF-1 (24 males, 17 females) aged 2–13 years (mean, 8 years). All patients presented a suspected diagnosis of NF-1, based on accepted criteria [2]. In all cases an evaluation was made of the clinical alterations, with special emphasis on the school level of the children; in 11 cases the intelligence quotient (IQ) was established by the Terman-Merrill intelligence test or Weschler Scale. Those children with a school level in agreement with age and/or an IQ of over 90 were considered normal; those requiring individual support and/or presenting an IQ of 70–90 were considered to be borderline cases, whereas children with an IQ of under 70 were considered to be retarded.

All patients were subjected to an orbital and cranial (5 and 10 mm slice thickness) CT examination before and after the administration of iodine contrast. A MRI examination of the head was carried out in all cases, with a 0.5 T superconductive magnet. Images were obtained in the sagittal plane with a T1-weighted spin-echo sequence (T1W-SE, 550/30), and in the transverse plane with a proton density (PD) and T2-weighted double echo spin-echo sequence (DPW-SE and T2W-SE, 2200–2500/50,100). In addition, 20 patients had a STIR sequence (1500/44/100) in the coronal plane. Fifteen patients were studied following gadolinium-DTPA (Gd-DTPA) administration with T1-weighted gradient-echo sequence (T1W-GE, 560/16/190°) in the transverse plane. In all sequences the slice thickness was 7 mm with an acquisition matrix of 256 x 256.

Results
The disease was transmitted by autosomal dominant inheritance in 21 cases (51.2%). Café-au-lait macules were the fundamental cutaneous manifestation. However, in 2 cases an isolated macula was present and in another...
2 cases they were absent. The macules bore no relation to the alterations detected in the CNS by CT and/or MRI. Table 1 shows the incidence of CNS manifestations ordered by frequency.

The typical optic nerve glioma was unilateral in 7 cases and bilateral in 5, the growth extending to the chiasm in 8 patients – 4 with unilateral involvement and 4 with bilateral involvement of the optic nerve. The optic radiations were only affected in 1 case, whereas in another patient the optic glioma was found to grow rapidly over one year, followed by a spreading of the tumor. Contrast enhancement was absent or minimal in most cases with optic-chiasmatic involvement (Fig. 1); however, uptake was marked in cases of extension to the posterior optic pathway (Fig. 2). In 3 cases of unilateral optic nerve glioma an accompanying arachnoid hyperplasia was present in the same nerve. CT failed to demonstrate chiasmatic involvement in 1 patient, and of the posterior optic pathway in another. Surgery was performed in 2 cases, revealing a low-grade glioma in both; 1 patient was subjected to

![Fig. 1. a Sagittal T1W-SE image shows optic glioma involving the chiasm. b Same patient, axial T1W-GE image after administration of Gd-DTPA shows no enhancement of bilateral nerve optic glioma. Note a left temporal arachnoid cyst](image1)

![Fig. 2. a Axial PDW-SE reveals increased signal intensity in the chiasm and optic tracts. b Axial T1W-GE after Gd-DTPA administration shows optic glioma enhancement](image2)

![Fig. 3. a T2W-SE showing absence of flow-void artefact through the aqueduct and hyperintensity around it (white arrow). Note also a left temporal hyperintense, round lesion (black curved arrow). b T1W-GE after administration of Gd-DTPA shows marked enhancement of the temporal lesion (curved arrow), suggesting a neoplasm; peri-aqueductal gliosis showing lack of enhancement (straight arrow)](image3)