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Hamartomatous Spinal Cord Lesion in Neurofibromatosis

Neurofibromatosis is associated with a wide spectrum of CNS disease, including hamartomatous or heterotopic lesions as well as benign and malignant neoplasms [1–5]. Several recent reports describe MR findings of multifocal areas of increased signal intensity in the brain, which do not cause mass effect and which are found in asymptomatic patients with neurofibromatosis. These lesions are thought to represent hamartomas [4, 6]. We present a patient who has these hamartomatous lesions in both the brain and spinal cord.

Case Report

A 10-year-old girl with neurofibromatosis underwent a screening MR examination in March 1987. The patient was asymptomatic and neurologically intact. The diagnosis was based on multiple café au lait spots, a plexiform neurofibroma of the left arm, and family history (the patient's mother had neurofibromatosis). The brain MR showed multiple areas of increased signal intensity on T2-weighted images bilaterally in the basal ganglia, thalami, and cerebellar white matter (Figs. 1A and 1B). These areas were thought to represent hamartomas on the basis of the location of the multiple lesions and the lack of any mass effect.

In March 1988, the patient returned for a second MR to explain a mild thoracic scoliosis detected on physical examination. Again, no neurologic signs or symptoms were present. In addition to dural ectasia, areas of increased signal in the cord on T2-weighted and gradient-echo imaging were found at T12 and L1 without cord enlargement (Figs. 1C and 1D). These, too, were thought to represent glial hamartomas.

Discussion

Neurofibromatosis is the most common disorder among the neurocutaneous syndromes with an incidence of 1:3000 live births [1, 3, 4]. A vast array of CNS lesions occur in this disorder, including cranial nerve neuromas, meningiomas, gliomas, pilocytic astrocytomas, ependymomas, angiomas, and hamartomas [1–5]. Theories proposed to explain these diverse findings include dysplasia, hyperplasia, and neoplasia of neural crest derivatives in the central, peripheral, and autonomic nervous systems. It has been suggested that these aberrant neural crest derivatives are acted on by abnormal melanin and by a nerve-growth factor to produce this spectrum of abnormalities [1, 2]. The hamartomas of neurofibromatosis consist primarily of neural-crest derived neurons, glial cells, and Schwann cells in addition to fibroblasts, vascular endothelial cells, and mast cells [2], resulting

in atypical glial cell nests, subependymal glial nodules, and ependymal ectopias [4, 5].

With the emergence of MR, many of these hamartomatous lesions are now being detected. Most often, they have been found in the basal ganglia, thalamus, hypothalamus, cerebral peduncles, and cerebellar white matter. The lesions generally do not produce masses; they appear normal on T1-weighted MR but have increased signal on T2-weighted images. They are seen in a high percentage of asymptomatic patients with neurofibromatosis [2–4, 6]. Although inferences to spinal cord hamartomatous lesions have been made in the literature [3, 4], no report on the use of MR has shown the typical brain hamartomas and spinal lesions together in the same patient. Mayer et al. [3] did, however, present a patient with neurofibromatosis who had bilateral acoustic neuromas, no evidence of intracranial hamartomas, and increased MR signal in a normal-sized cervical cord. This patient was asymptomatic and thus was presumed to have a spinal hamartoma. The brain lesions (Figs. 1A and 1B) in conjunction with the spinal cord lesions (Figs. 1C and 1D) make our case unique. The lack of cord enlargement, the lack of any signs or symptoms referable to the spinal cord, the presence of the "typical" brain hamartomas and a normal spine T1-weighted MR image indicate heterotopias rather than a more aggressive lesion. Further investigation is warranted to determine the incidence of these spinal hamartomas in patients with neurofibromatosis.

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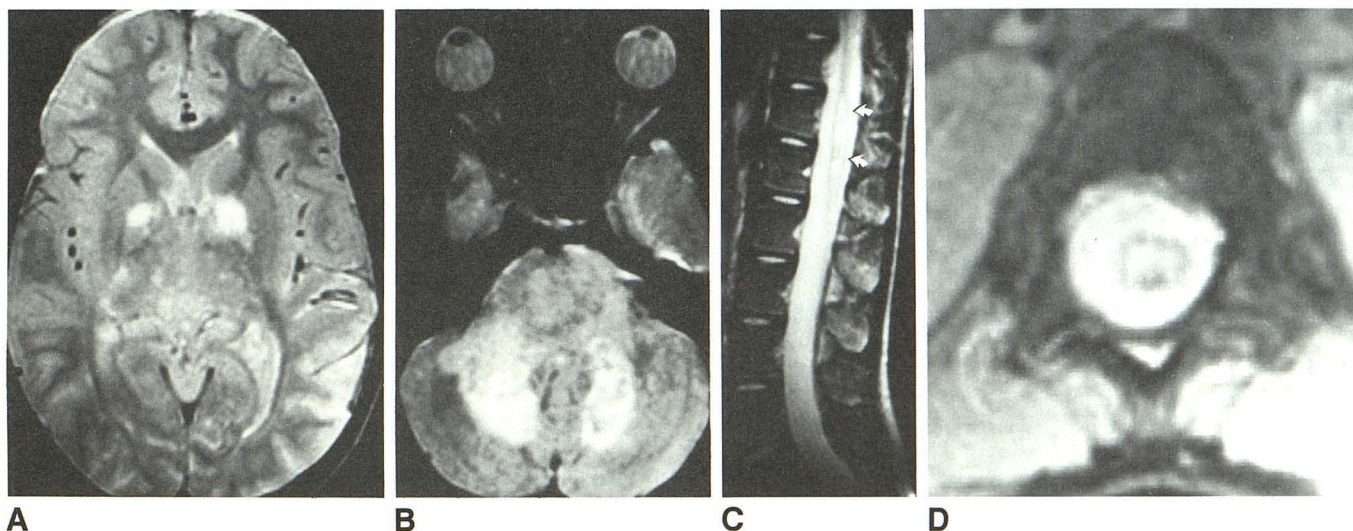


Fig. 1.—A and B, T2-weighted axial MR images (2100/60) show presumed hamartomatous/heterotopic lesions in the brain. In A, lesions are in basal ganglia and thalami. Similar lesions are shown in B in dentate nuclei and middle cerebellar peduncles. C and D, Presumed similar lesions in the distal cord. Both C, sagittal T2-weighted image (2000/80), and D, gradient-echo axial image through the T12 level (300/14; flip angle 10°), show increased intramedullary signal (arrows in C) without thoracic cord enlargement.