Neurofibromatosis Type I: A Pictorial Review of the CNS Manifestations in Pediatric Patients

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Introduction

Neurofibromatosis are autosomal dominant inherited neurocutaneous syndromes, which belong to a larger group of phakomatoses. Neurofibromatosis (NF) is the most common of the phakomatoses yet most affected individuals go undiagnosed as children. At least eight different forms of neurofibromatosis have been described with neurofibromatosis type 1 (NF-1) and neurofibromatosis type 2 (NF-2) regarded as the most common and best understood. Both are autosomal dominant disorders with increased risk of malignancy. However, they are clinically distinct diseases. Although NF-1 may affect many organ systems, central nervous system involvement tends to be less frequent and often less the most severe. A thorough understanding of the common CNS manifestations of NF-1 in children is necessary for the diagnosis and management.

Brain Manifestations

Hyperintense lesions of the brain

T1 Hyperintense lesions of the brain

- T2 Hyperintense lesions of the brain

Bilateral optic nerve tumors is considered pathognomonic for NF-1. Pathologically, these tumors are low-grade pilocytic astrocytomas (WHO grade I) and are usually slow-growing and benign. They are however symptomatic causing visual disturbances.

Bilateral optic pathway gliomas

- Frontal lobe herniation(arrow) through the absent left greater sphenoid wing

- Plexiform neurofibromas may involve long segments of the spinal nerves and extend into the spinal cord. Although there is a potential for malignant degeneration, less than 5% of patients with NF-1 develop neurofibromas.

Spinal Manifestations

- Dural ectasia

- Moya Moya

- Plexiform neurofibromas

- Sural nerve plexiform neurofibromas

- Bony Lesions of Skull

- Spinal cord compression

- Sphenoid wing dysplasia

- Sutura defects

References


