Current Problems in Diagnostic Radiology ▮ (2017) ▮▮▮–▮▮▮

Current Problems in Diagnostic Radiology

journal homepage: www.cpdrjournal.com

Orbital Imaging Manifestations of Neurocutaneous Syndromes Revisited

Abanti Das, MBBS, MD[a], Sanjay Sharma, MBBS, MD[b,*]

[a] Department of Radiodiagnosis, All India Institute of Medical Sciences, New Delhi, India
[b] Department of Radiodiagnosis, Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India

A B S T R A C T

Neurocutaneous syndromes or phakomatoses represent a heterogeneous group of multisystemic disorders involving structures of ectodermal origin. Characteristic ocular manifestations are described for individual entities that are often the first clues to the underlying diagnosis. However, opaque ocular media or involvement of retrobulbar orbit limits adequate clinical evaluation. This underlines the role of imaging, especially cross-sectional imaging modalities, such as computed tomography and magnetic resonance imaging, which offer a comprehensive evaluation of orbit and its contents. This review aims to summarize the cross-sectional imaging features of orbital manifestations of common neurocutaneous syndromes encountered in clinical practice.

Neurocutaneous syndromes or phakomatoses are a group of heterogeneous multisystemic disorders characterized by involvement of structures that arise from the embryologic ectoderm. Hence, commonly affected sites include central nervous system, eye, and skin. Ocular manifestations are often the first that point to the underlying disorder. Although ophthalmologic examination can confidently diagnose the characteristic lesions described for individual phakomatoses, clinical evaluation often becomes unreliable in the presence of opaque ocular media because of cataractous lens or vitreous hemorrhage, which frequently accompanies these syndromes. Further, the retrobulbar region is also not easily amenable to adequate clinical evaluation. Imaging offers a noninvasive and comprehensive evaluation of globe and retrobulbar orbit and brain, detecting crucial ancillary findings that can provide a clue to the underlying diagnosis.[1] Apart from initial diagnosis and mapping out the extent of involvement, imaging also allows monitoring response to treatment.

Among the available imaging modalities, computed tomography (CT) and magnetic resonance imaging (MRI) remain the investigation of choice for evaluating orbital pathologies.[1] Multiparametric MR added further value to conventional MRI by allowing functional assessment of lesions. This has allowed predicting biological behavior of pathologies and prognostication.

This review aims to provide a brief overview of the imaging manifestations of common neurocutaneous syndromes, highlighting the key imaging features of their ocular and extraocular orbital manifestations on CT and MRI and distinguishing features from common imaging mimickers.

Neurofibromatosis

Neurofibromatosis (NF) is the most common neurocutaneous syndrome with an autosomal dominant inheritance and high penetrance. It is further subdivided into the following 2 forms: NF-1 and NF-2. Although the former occurs with a prevalence of approximately 1:3000, the latter is relatively rare with a prevalence of 1: 60,000.[2,3] Although familial, sporadic cases have also been frequently reported representing spontaneous mutations.[2] The responsible gene has been mapped to chromosome 17 for NF-1 and chromosome 22 for NF-2.

NF Type 1 (NF-1) (von Recklinghausen Disease)

The most common ocular manifestation of NF-1 is a Lisch nodule that is melanocytic iris hamartoma. These lesions develop in more than 90% of individuals aged 6 years and older and in more than 95% of individuals above 16 years.[4,5] This forms one of the diagnostic criteria of NF-1and is easily evaluated on clinical examination alone.

* Reprint requests: Sanjay Sharma, MBBS, MD, Department of Radiodiagnosis, Dr. Rajendra Prasad Centre for Ophthalmic Sciences, Department of Radiodiagnosis, All India Institute of Medical Sciences, Ansari Nagar, New Delhi 110029, India.
E-mail address: drssharma@hotmail.com (S. Sharma).

http://dx.doi.org/10.1067/j.cpradiol.2016.12.011
0363-0188/

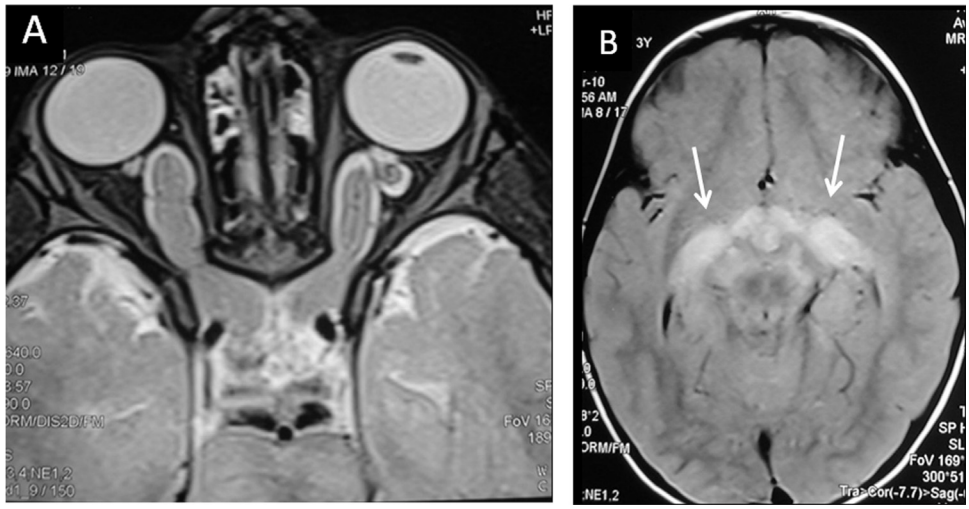

Fig. 1. A 3-year-old boy with neurofibromatosis type 1 and bilateral optic pathway glioma. (A) Axial T2W fat-saturated image shows bilateral thickened and tortuous optic nerves that are isointense to mildly hyperintense on T2W images with chiasmal extension. Note the homogenous appearance of the tumor with preservation of normal shape of optic pathway. No cystic components. (B) Axial fluid attenuated inversion recovery sequence (FLAIR) image indicate posterior extension of glioma along bilateral optic tracts (arrows).

Optic Pathway Glioma

Imaging plays an important role in the diagnosis of optic pathway tumors that also constitute one of the diagnostic criteria of NF-1. These occur in approximately 10%-15% of patients with NF-1 while 70% of optic pathway tumors are seen in NF-1.[2]

Optic pathway gliomas (OPGs) are low-grade pilocytic astrocytomas that involve the precortical visual pathway that includes optic nerves, optic chiasma, tracts, and radiation (Figs 1 and 2). They are either sporadic or syndromic. When associated with NF-1, they tend to be multifocal and bilateral with predominant involvement of prechiasmatic visual pathway, although any part of the optic pathway can be involved.[6] These tend to have a better prognosis compared to the sporadic forms and can even show spontaneous regression.[6] Young children and adolescents are mainly affected.[7]

Most patients present with diminished vision, proptosis, or precocious puberty.[6,7] On the contrary, many OPGs are incidental findings in NF-1. The goal of imaging is to confirm the presence of lesion, delineate its extent, and monitor response to treatment.

OPGs present as diffuse, fusiform, or bulbous enlargement of optic nerve that can extend to involve optic chiasma, optic tracts, and radiation (Fig 1). On plain radiographs, used earlier, classical findings were that of enlarged optic canals and a J-shaped sella turcica.[8] On CT, the lesion presents as uniform enlargement and tortuosity of optic nerves.[9] Some authors have described a downward kink of intraorbital segment of involved optic nerve[9] (Fig 2).

MRI is the preferred imaging modality for diagnosis in view of its better contrast resolution and the ability to better visualize lesions close to skull base and sella turcica, which often gets obscured by beam-hardening artifacts on CT. On MRI, lesions are hypointense to isointense on T1-weighted images and hyperintense on T2-weighted images with variable enhancement on postcontrast images (Fig 1). Gadolinium enhancement and cystic components are less commonly seen in lesions associated with NF-1.[10] A characteristic "pseudo-CSF sign" has been described in lesions of NF-1. It is hypothesized that in NF-1, apart from enlargement of the optic nerve, there may be abnormal extra-axial intradural tissue that could either represent arachnoid hyperplasia or tumor extension through the pia-arachnoid matter. This tissue mimics the signal intensity of cerebrospinal fluid (CSF) on T2-weighted image leading to the "pseudo-CSF" sign.[11]

A rare cause of enlargement of optic nerve sheath is dural ectasia or optic nerve sheath meningocele that occurs because of accumulation of CSF in perioptic dural sheath.[12] The most common association of this entity is NF-1,[13,14] and hence it presents as a differential diagnosis to OPG. CT can demonstrate enlargement of intraorbital segment of optic nerve, which is nonprogressive on serial imaging[13]; however, MRI is the investigation of choice to exclude other possible diagnoses, especially cystic subtypes of OPG and optic meningiomas.[13,15] MRI shows focal or segmental enlargement of meningeal space around the optic nerve, which follows the signal intensity of CSF on all pulse sequences.[13,15] Underlying optic nerve may be normal or thickened.

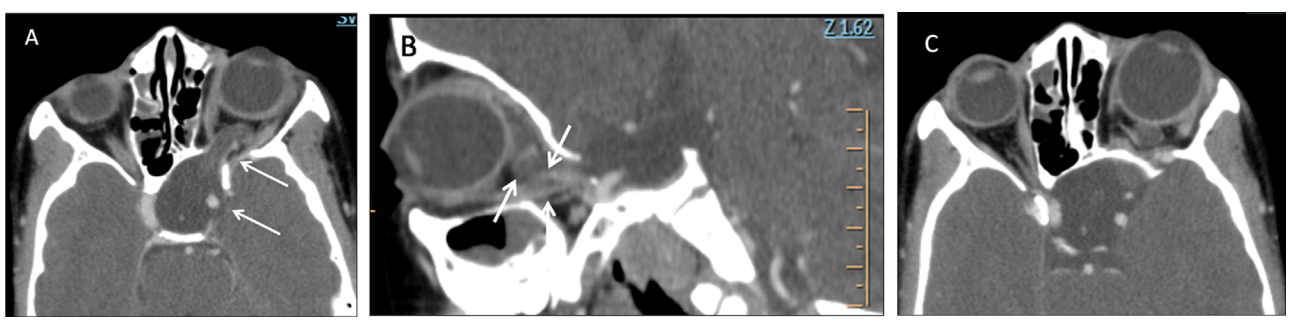

Fig. 2. A 10-year-old girl, a diagnosed case of neurofibromatosis type 1. (A) Axial contrast-enhanced CT (CECT) shows enlarged and tortuous left optic nerve extending till optic chiasma (arrows). Right optic nerve was normal. (B) Sagittal image shows characteristic downward kink of intraorbital segment of left optic nerve (arrows). (C) Note made of left megalopthalmos. (Color version of figure is available online.).