

## 27. Neurofibromatosis and Von Hippel-Lindau Syndrome

Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is an autosomal dominant disorder with diagnostic criteria including two or more of the following: a first degree relative with NF1, axillary freckling, distinctive bone lesions, an optic pathway glioma, a plexiform neuroma, at least 6 café-au-lait spots, and more than one Lisch nodule or neurofibroma. Foci of abnormal signal intensity (FASI) are the most common NF1 findings on brain MRI. The FLAIR images in Figure 27.1 A demonstrate the typical high SI appearance of FASI in their characteristic location—the globus pallidus of the basal ganglia. Pallidal lesions are typically isointense to adjacent brain on T1WI, although they may demonstrate irregular mild high SI—a characteristic thought to be due to myelin clumping. Less commonly, FASI involve the pons (Fig. 27.1 B) or cerebellum (Fig. 27.1 C). Lesions often increase in prominence in middle childhood, fading by adolescence. Unlike a neoplasm, FASI do not demonstrate surrounding vasogenic edema or mass effect and will only rarely enhance. Optic gliomas—nearly always juvenile pilocytic astrocytomas (WHO Grade 1)—are the most common intracranial tumor of NF1. Bilateral lesions are nearly pathognomonic. NF1 gliomas appear most frequently as diffuse, isointense enlargements of the optic nerve, in distinction to the heterogenous, cystic appearance of non-NF1 lesions. Enhancement (best seen on fat-suppressed images) is variable but may aid in diagnosis, along with the high SI on T2WI seen in some lesions. Differential considerations include optic nerve sheath meningioma—characterized by a fusiform, tram-track appearance—along with sarcoidosis. Gliomas are rarely limited to the nerve itself. Involvement of the optic chiasm is frequent and is often visible as diffuse chiasmatic enlargement—as illustrated in the T1WI of Figure 27.2 A (white arrow). Hypothalamic gliomas are also common (Fig. 27.2 A, black arrow). Plexiform neuromas of the face and

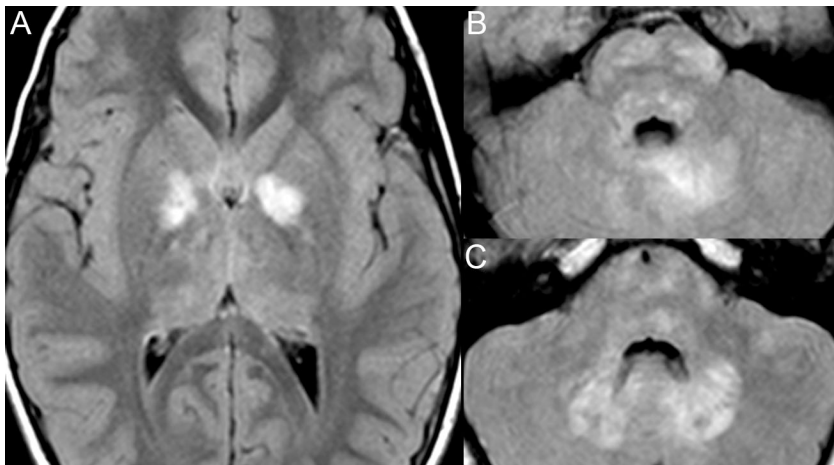


Fig. 27.1

cranial nerves are less common than optic gliomas but are nearly pathognomonic for NF1. Lesions demonstrate low to moderate SI on T1 and high SI on T2WI. Heterogeneous enhancement is common. Additional common findings include buphthalmos (enlargement of the globe) and sphenoid dysplasia (part of the NF1 diagnostic criteria).

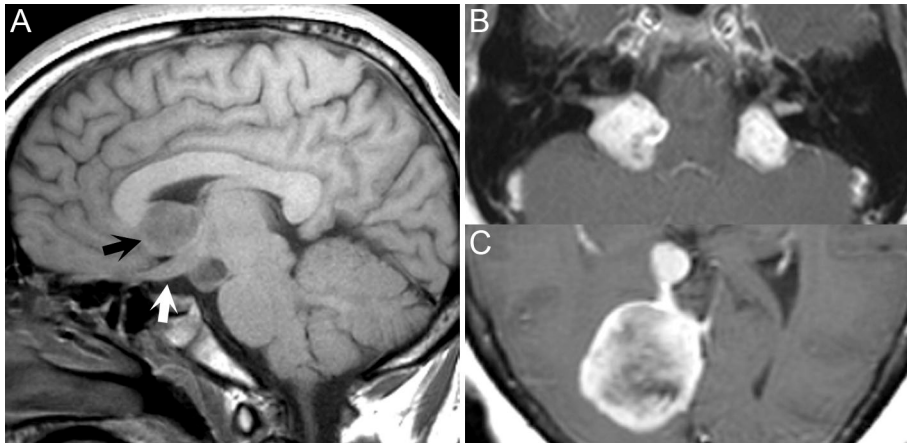


Fig. 27.2

Neurofibromatosis type 2 (NF2) is inherited in an autosomal dominant pattern with an incidence of about one-tenth that of NF1. No cutaneous neurofibromas or plexiform neuromas are present. Bilateral cerebellopontine angle masses (vestibular schwannomas)—the enhancing lesions in Figure 27.2 B—are diagnostic and require no further pathologic confirmation. Other criteria for diagnosing NF2 include a first degree relative with the condition plus either a unilateral cerebellopontine angle mass or two of the following: a glioma, schwannoma, neurofibroma, or juvenile posterior subcapsular cataract. Compared to spontaneously occurring vestibular schwannomas, those of NF2 are more commonly bilateral. They are otherwise similar in MRI appearance to non-NF2 lesions (see Chapter 5). The meningiomas of NF2 are also similar in appearance to their spontaneously occurring counterparts (see Chapter 4), although clinically they occur in younger patients. Figure 27.2 C demonstrates two bright, heterogeneously enhancing meningiomas similar in appearance to those previously illustrated in Figure 4.1 B. The suggestion of intracranial NF2 warrants screening for spinal lesions (see Chapter 36) with contrast-enhanced MRI. Von Hippel-Lindau (VHL) disease is an autosomal dominant condition consisting of pheochromocytomas, tumors of the kidney (clear cell carcinoma) and pancreas (cystadenoma), angiomas of the retina, and hemangioblastomas of the posterior fossa and spinal cord. Multiple or spinal hemangioblastomas necessitate a full imaging workup of VHL. Hemangioblastomas are classically described as cystic masses with a solid, subpial, mural nodule. On MRI the cystic portions demonstrate high SI on T2 and low SI on T1WI.

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The SI of the mural nodule may be mixed due in part to low SI flow voids within or just peripheral to it. Enhancement of only the nodule without rim enhancement of the cyst is characteristic. 60% of all posterior fossa hemangioblastomas have this appearance, cystic with a “mural” nodule, while 40% are solid. Cord hemangioblastomas are less prevalent but more specific for VHL than cerebellar lesions and are commonly solid.