Clinical findings of the phakomatoses

von Hippel-Lindau disease

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Figure 1. (A) A vascular nevus, often broad, purpuric, or even ecchymotic lesions, on the left knee of an adult with von Hippel-Lindau (VHL) disease. Unlike the other phakomatoses, skin lesions in VHL are relatively uncommon. (B) Hemangioblastoma (or angioma) of the retina from another affected adult. Note the single dilated feeding and single draining vessel (arrows). Hemangioblastomas affect just under 60% of subjects.² About half evolve to cause unilateral or bilateral blindness from subretinal edema, hemorrhage, or retinal detachment.² Between 38% and 58% of individuals with retinal hemangioblastomas have VHL. VHL may also be associated with benign cysts or cystic tumors of the kidney, lung, spleen, or other abdominal organs. Malignant hypernephroma, the third most common tumor in VHL, affects 25% of subjects and is the leading cause of death. Pheochromocytoma is a less common complication (about 3-20%).² Risk can be determined by mutation type.

von Hippel-Lindau (VHL) disease is an autosomally dominant, pleomorphic disease of multisystem tumors. Heterogeneous germline mutations of the *VHL* gene (3p25-26), usually arising de novo, are

found in 70% to 80% of individuals. Reviews of current genetic diagnosis, screening, and pathogenesis are available.¹ Physical findings are shown in figure 1 and neuroimaging findings in figure 2.

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Figure 2. Cerebellar hemangioblastoma in axial and coronal sections of a contrasted T1-weighted MRI. These are present in 60% of cases in well-studied kindreds.² About 20% of those with retinal hemangioblastoma will have a symptomatic cerebellar lesion. Cerebellar hemangioblastoma may result in devastating hemorrhage.

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