Clinical findings of the phakomatoses  
von Hippel-Lindau disease

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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.

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. About 20% of those with retinal hemangioblastoma will have a symptomatic cerebellar lesion. Cerebellar hemangioblastoma may result in devastating hemorrhage.
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