Eye Findings in Neurocutaneous Syndromes

Each phakomatosis has its own ocular "signature"

NF1 — **Neurofibromatosis type 1**
Lisch nodules (iris hamartomas) · café-au-lait spots, neurofibromas

NF2 — **Neurofibromatosis type 2**
juvenile posterior subcapsular cataract · optic nerve sheath meningioma · bilateral vestibular schwannomas

TSC — **Tuberous sclerosis**
retinal hamartomas · facial angiofibromas, hypopigmented macules

SW — **Sturge-Weber syndrome**
port-wine stain (trigeminal territory) and glaucoma

VHL — **Von Hippel-Lindau disease**
retinal (and cerebellar) hemangioblastoma · pheochromocytoma, renal carcinoma

Lisch nodules	**Port-wine stain**	**Retinal hemangioblastoma**
NF1	+ glaucoma: Sturge-Weber	Von Hippel-Lindau

Almost all are autosomal dominant, except Sturge-Weber, which is sporadic