Tuberous Sclerosis Complex

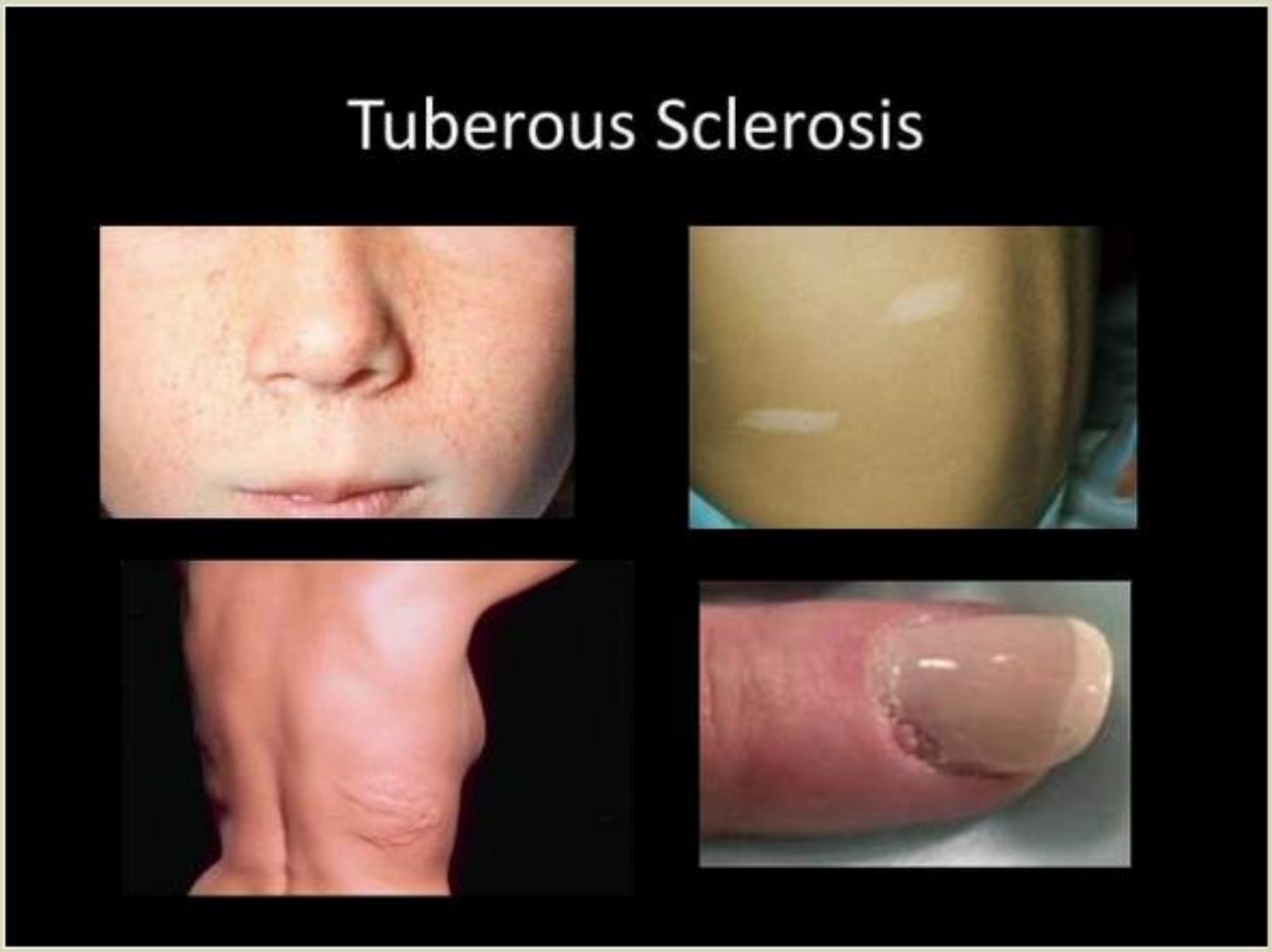
- Synonym: Bourneville-Pringle syndrome
- Inherited tumor disorder with multi-organ hamartomas.
- Abnormal differentiation/proliferation of germinal matrix cells
- Mutations in TSC tumor suppressor genes cause abnormal cellular differentiation, proliferation.
- ~ 50% of TSC cases inherited

Imaging

- Calcified subependymal nodules (SEN) (hamartomas)
- Subependymal giant cell astrocytoma (SEGA) (15%); most located at foramen of Monro
- Cortical/subcortical tubers (95%)
- White matter radial migration lines
- Cyst-like white matter lesions (cystoid brain degeneration)
- Cortical/subcortical tubers: Early T1 ↑ but variable after myelin maturation
- SEN enhancement more visible on MR than on CT
- AMT-PET distinguishes epileptogenic from nonepileptogenic tubers

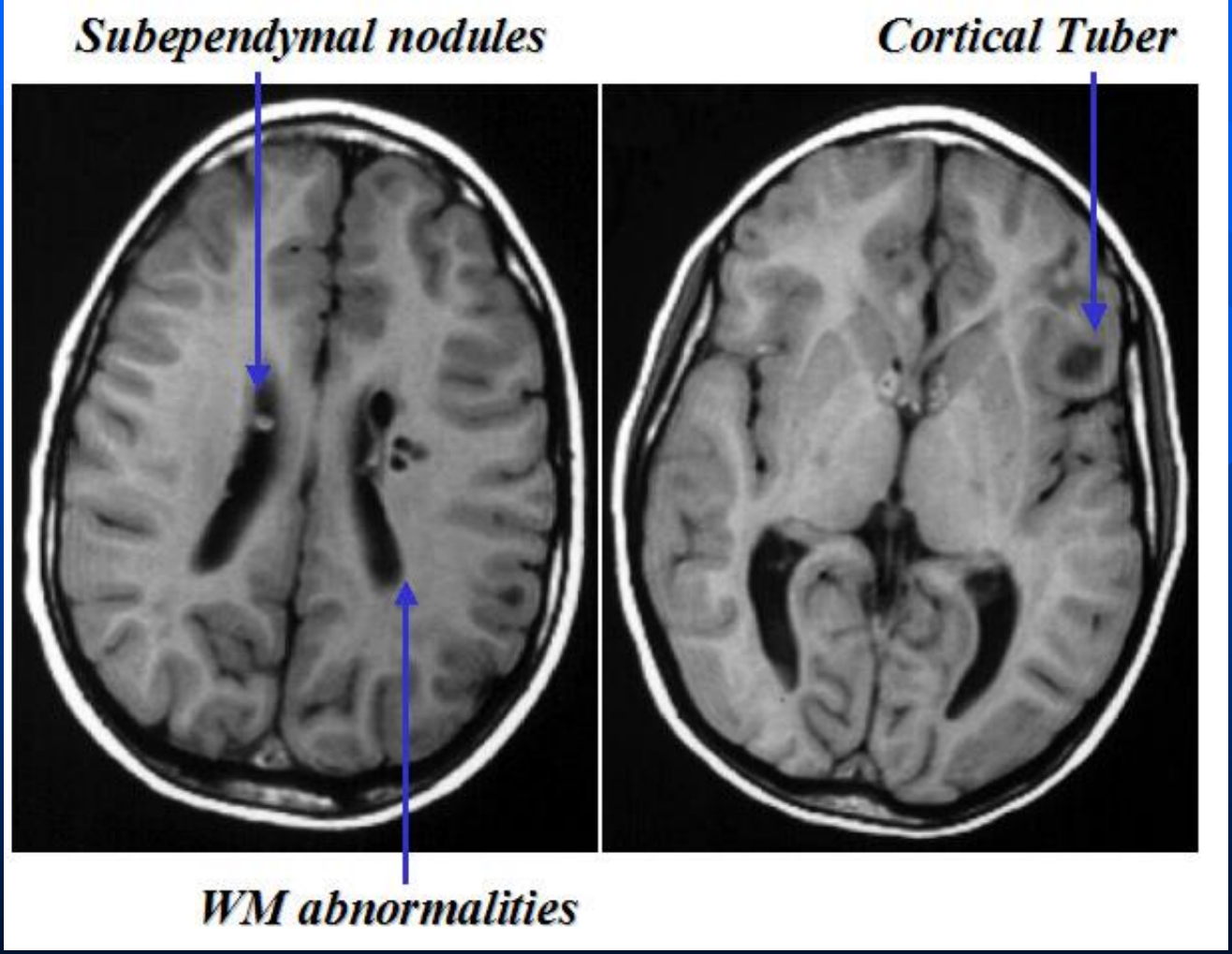
Associated abnormalities

- Renal: Angiomyolipoma and cysts (40-80%)
- Cardiac: Rhabdomyomas (50-65%); majority involute over time
- Lung: Cystic lymphangiomyomatosis/fibrosis
- Solid organs: Adenomas, leiomyomas
- Skin: Ash-leaf spots (majority), including scalp/hair; facial angiofibromas; shagreen patches
- Extremities: Subungual fibromas (15-20%), cystic bone lesions, undulating periosteal new bone formation
- Ocular: "Giant drusen" (50%), retinal astrocytomas (which may regress)
- Dental pitting of permanent teeth in most adults



Genetically determined syndrome of epilepsy associated with: Red patches on the face containing many blood vessels (adenoma sebaceum) . Areas of the skin that are white (due to decreased pigment) and have either an ash leaf or confetti appearance . Raised patches of skin with an orange-peel texture (shagreen spots), often on the back . Small tumor like lesions at the nail folds (subunguual fibromas) .

Tuberous Sclerosis



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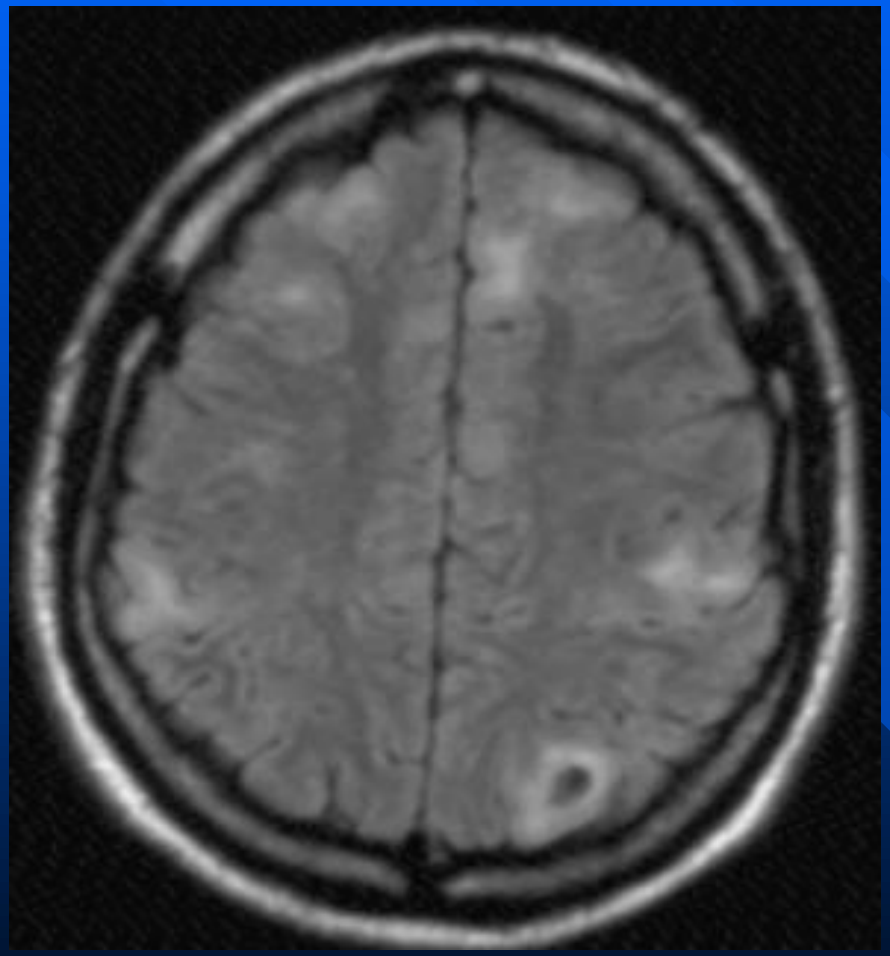
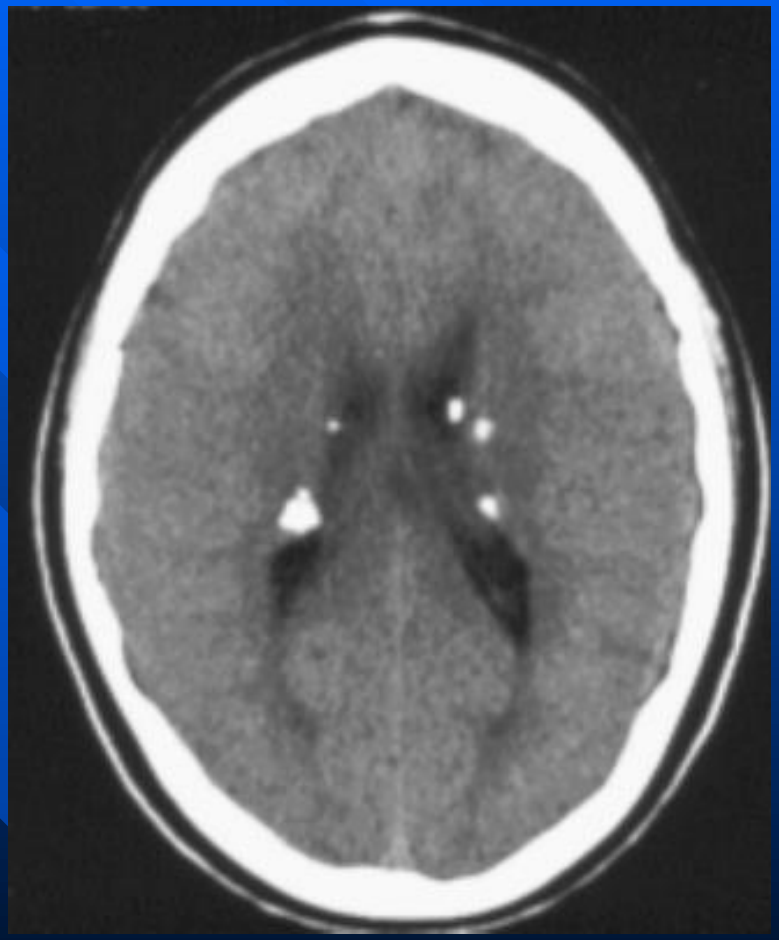
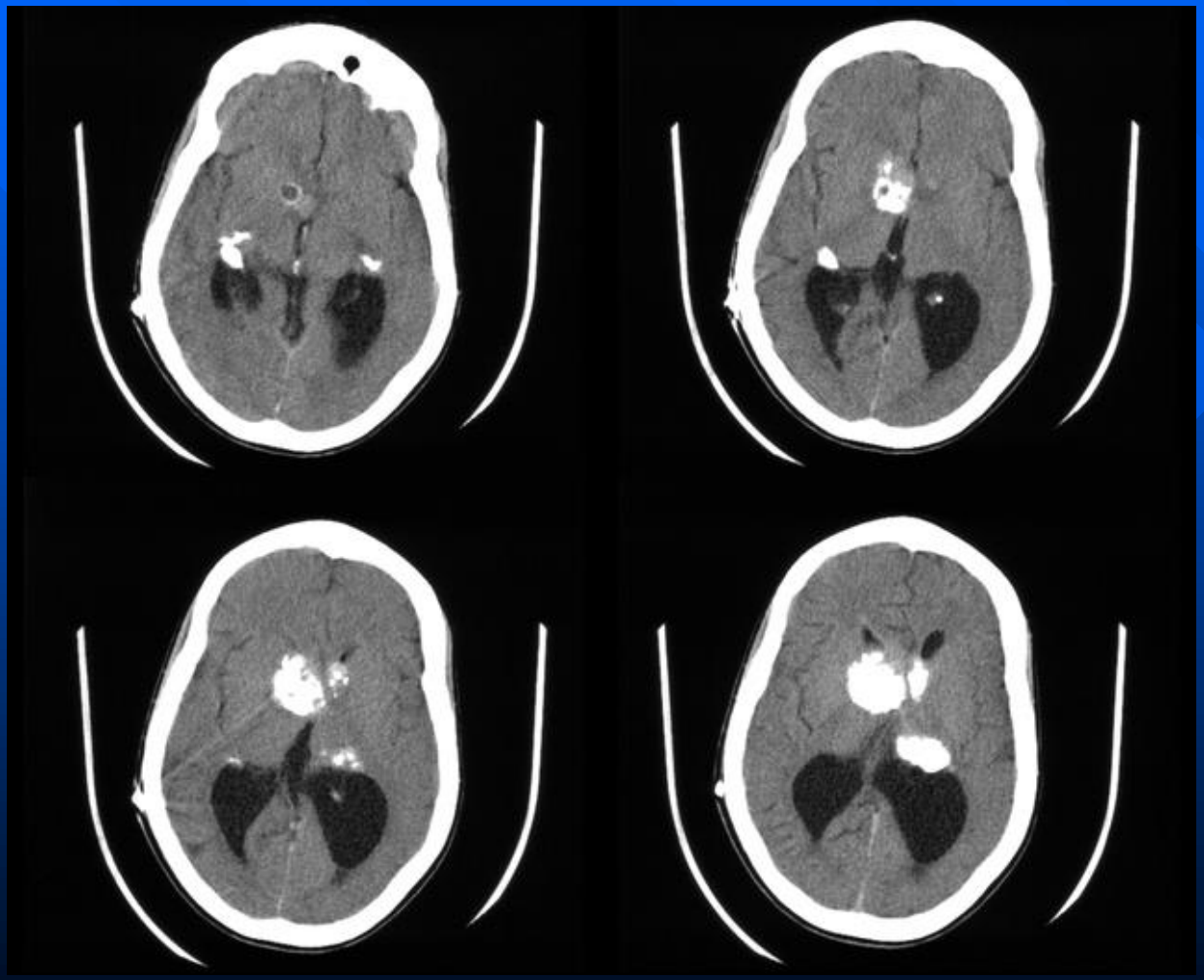




Tuberous sclerosis, giant astrocytoma



Giant cell astrocytoma in a patient with tuberous sclerosis