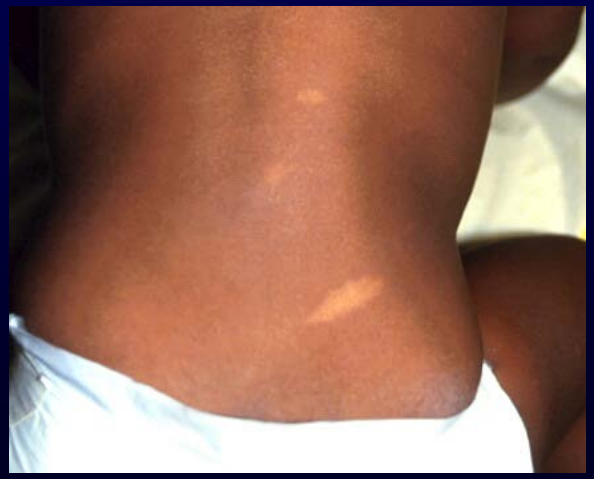
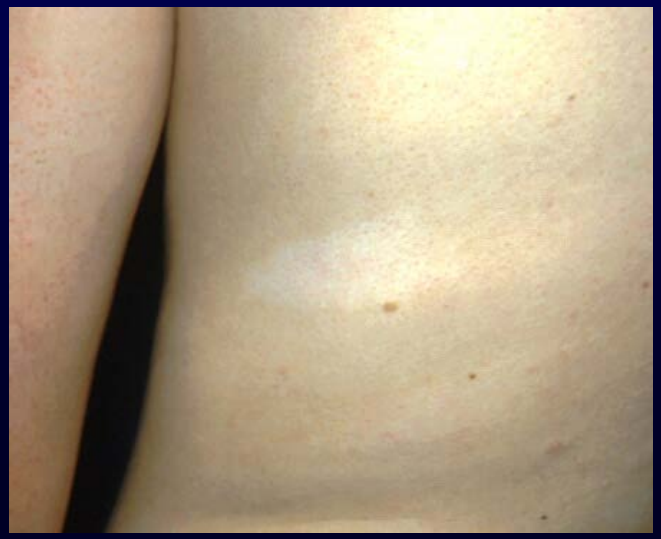
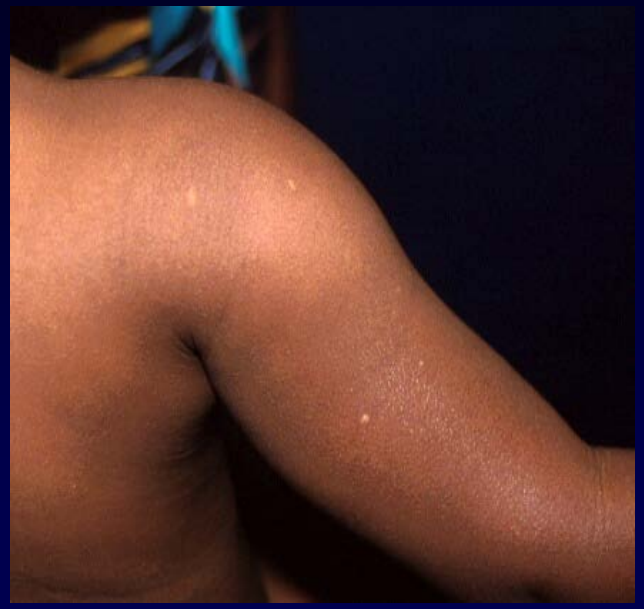
Tuberous Sclerosis

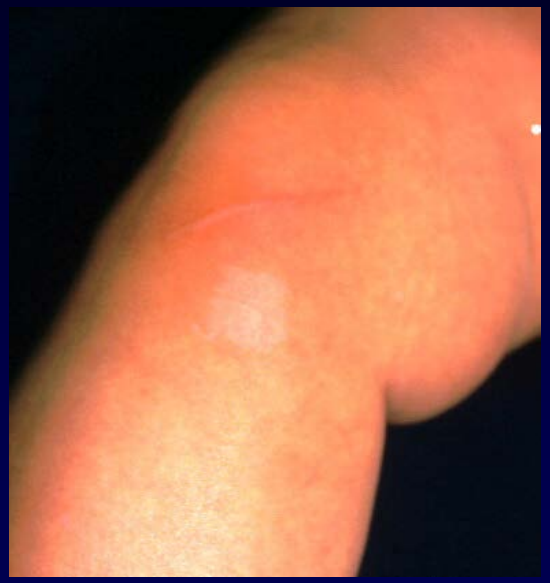
- Facial angiofibromas, ungual fibromas
- Hypomelanotic macules (ashleaf spots)
- Shagreen patch
- Seizures
- Subependymal nodule (± calcification), cortical tuber
- Cardiac rhabdomyoma, renal angiomyolipoma
- 80% occur by spontaneous mutations; 2 genes
  - TSC2: 16p13.3, tuberin, GTPase activator, regulates cell growth
  - TSC1: 9q43, hamartin, putative growth suppressor, transmembrane protein, interacts with tuberin

Hypopigmented Spots

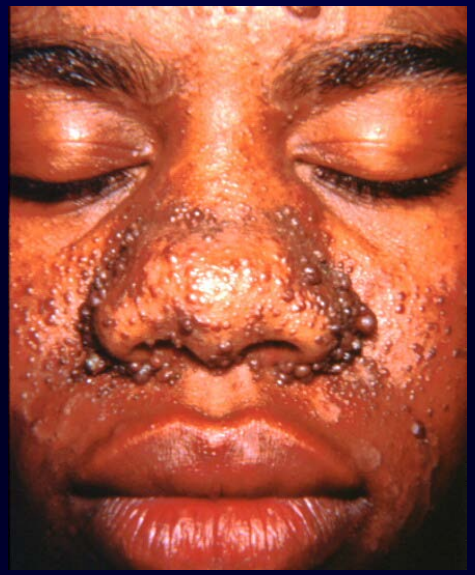


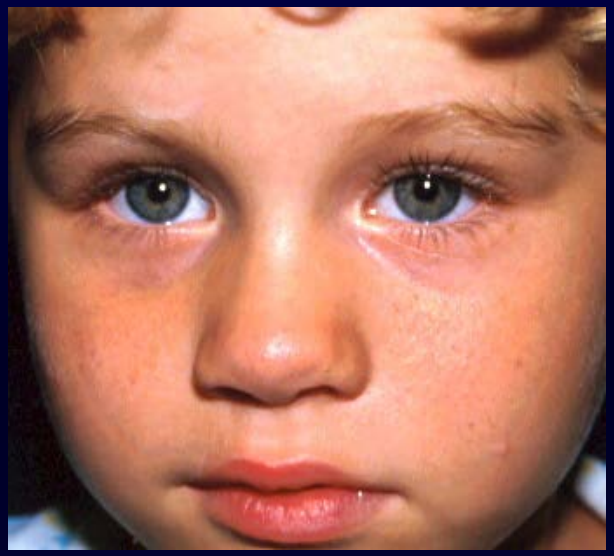


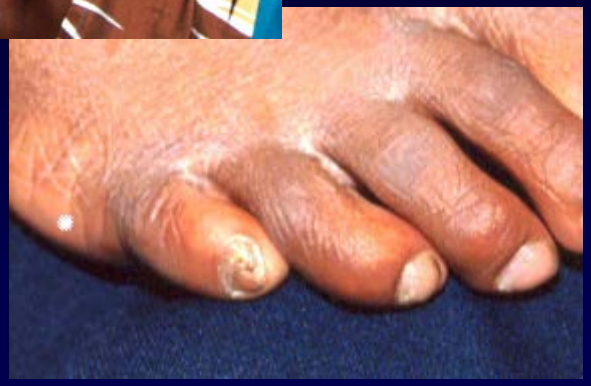


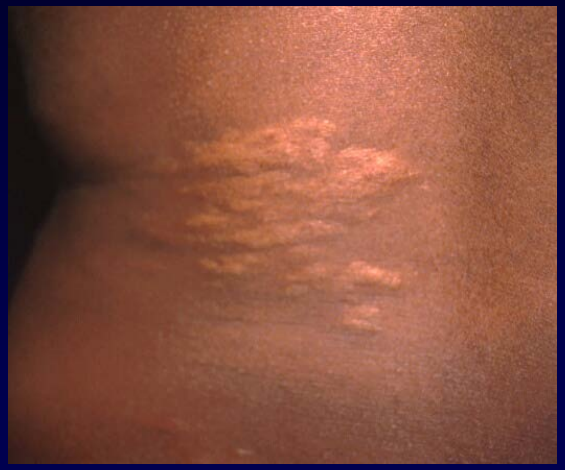


Fibroangiomas in TS









TS Diagnostic Criteria

2 major or 1 major, 2 minor

Major

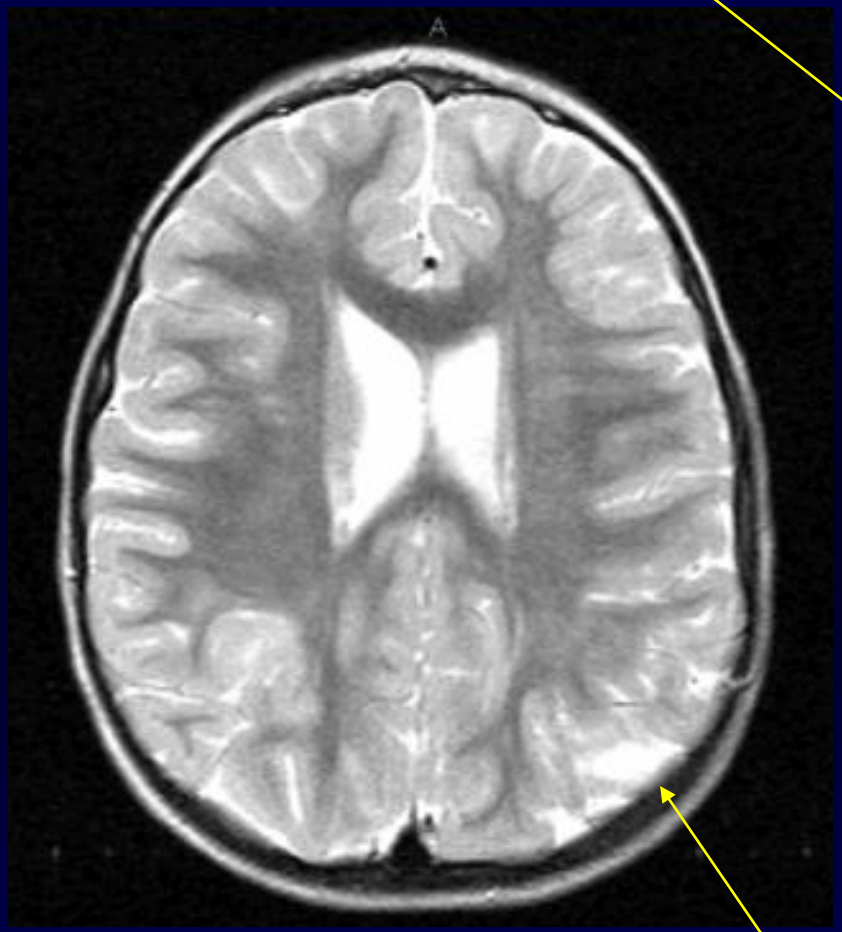
- facial angiofibromas
- ungual fibromas
- ≥ 3 hypomelanotic macules
- multiple retinal hamartomas
- shagreen patch
- cortical tuber
- subependymal nodule (± calcification)
- subependymal giant cell astrocytoma
- cardiac rhabdomyoma
- lymphangiomyomatosis or angiomyolipoma

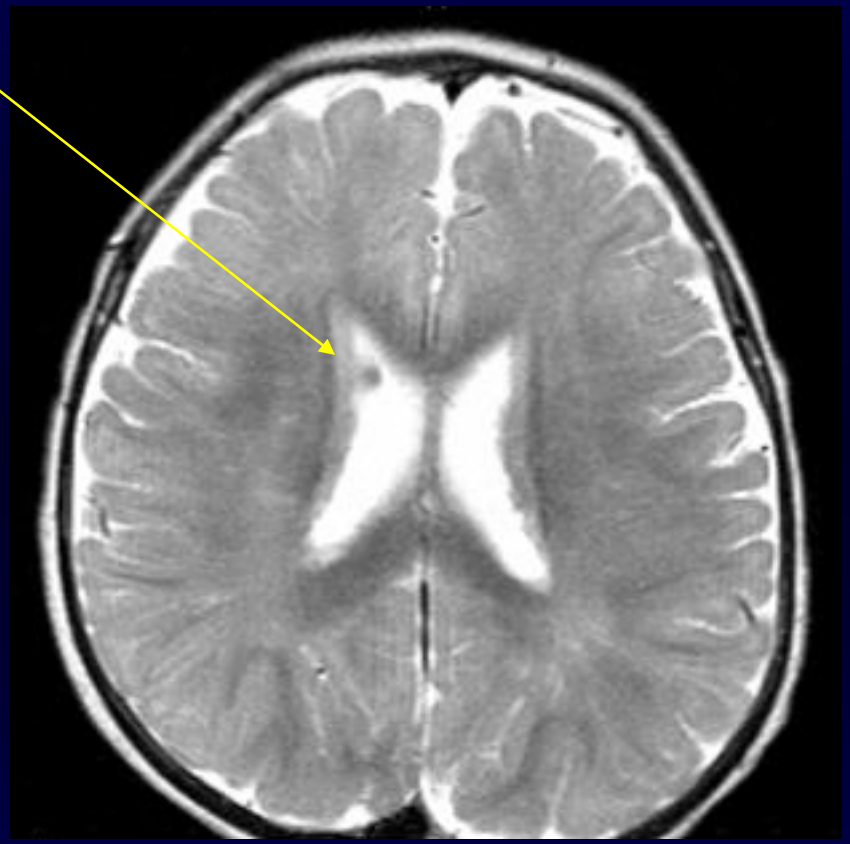
Minor

dental pits, hamartomatous rectal polyps, bone cysts, cerebral white matter radial migration lines, gingival fibromas, nonrenal hamartoma, retinal achromic patch, “confetti” skin lesions, multiple renal cysts

J Child Neurol 13, 1998

Subependymal nodule





Boy with focal seizures

Boy with infantile spasms

Cortical tubers