

#MADEEASY



TUBEROUS SCLEROSIS
PART 2



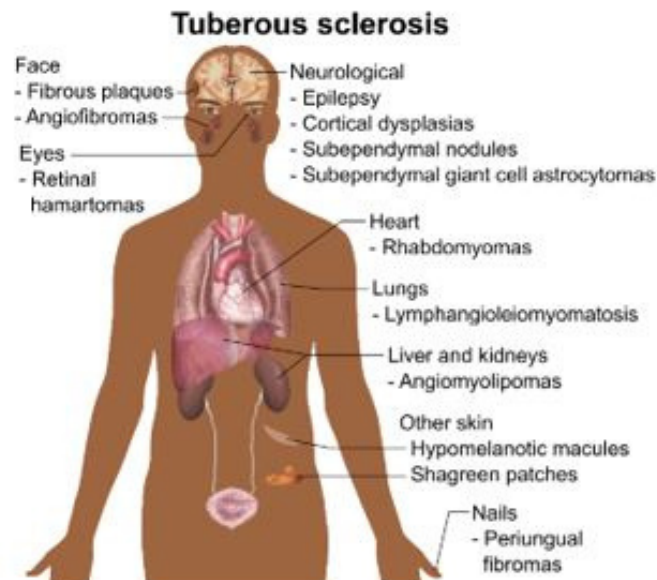


TUBEROUS SCLEROSIS PART 2

Histological Findings

- **Hypomelanotic Macules:**
- **Show a decreased number of melanocytes or melanin, resulting in the lighter appearance of the skin.**
- **Angiofibromas:**
- **Consist of fibrous tissue, blood vessels, dilated blood-filled spaces**
- **Shagreen Patch:**
- **Characterized by dermal collagen bundles arranged in a *whorled* pattern**
- **Ungual Fibromas:**
- **Composed of fibrous tissue with thin-walled blood vessels**

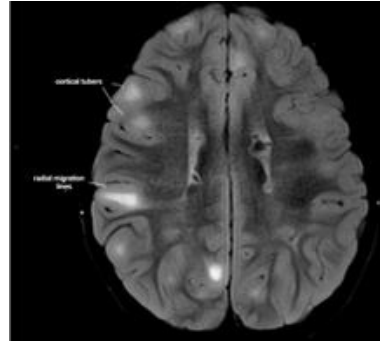
Other Clinical Features





OTHER CLINICAL FEATURES

- **Neurological:**
- **Cortical Tubers:** Hamartomas in the brain's cortex, leading to seizures (often the first sign in infancy)



- **Subependymal Nodules:** Calcified nodules along the ventricles, which can progress to subependymal giant cell astrocytomas
- **Intellectual Disability:** Common, varying from mild to severe.
- **Renal:**
- **Angiomyolipomas:** Benign tumours made up of blood vessels, muscle, fat, often found in the kidneys





OTHER CLINICAL FEATURES

- **Cysts:** Simple renal cysts can also occur
- **Cardiac:**
- **Rhabdomyomas:** Benign tumours of the heart muscle, often present at birth, can cause obstructive symptoms in severe cases.
- **Pulmonary:**
- **Lymphangiomyomatosis:** A lung condition seen primarily in women, characterized by cysts and smooth muscle growth leading to respiratory issues.

Diagnosis

- **Diagnosis of TSC is based on the presence of major and minor criteria.**
- **The presence of *two major features or one major and two minor features* confirms the diagnosis.**

Criteria	Major	Minor
Skin Lesions	Hypomelanotic macules, facial <u>angiofibromas</u> , shagreen patch, unguinal fibromas	Confetti skin lesions, dental enamel pits, intraoral fibromas
Neurological Lesions	Cortical tubers, subependymal nodules, SEGAs	White matter migration lines
Renal Lesions	<u>Angiomyolipomas</u>	Renal cysts
Cardiac Lesions	Rhabdomyomas	-
Pulmonary Lesions	<u>Lymphangiomyomatosis (LAM)</u>	-
Ophthalmic Lesions	Retinal hamartomas	Retinal achromic patches

- **Genetic Testing: Identification of mutations in TSC1 or TSC2 genes can confirm the diagnosis**





TREATMENT

- **Seizure Management:**
- **Antiepileptic drugs are commonly used**
- **Surgical intervention may be necessary for refractory seizures due to cortical tubers.**
- **Tumour Treatment:**
- **Everolimus (an mTOR inhibitor) is used for the treatment**
- **Surgical removal of symptomatic or large tumours**
- **Skin Lesion Treatment:**
- **Laser therapy**
- **Topical rapamycin for reducing the size and redness of facial angiofibromas**

- **Regular Monitoring:**
- **MRI scans for brain and renal tumours**
- **Regular eye exams to check for retinal hamartomas**
- **Cardiac evaluations for children with TSC**

