



A **rare autosomal dominant** genetic disorder that causes **non-cancerous tumors** to form in various organs, including the brain, skin, kidneys, and heart. Symptoms can include **seizures**, **developmental delays**, and **skin abnormalities**. Early diagnosis and comprehensive management are essential for **improving quality of life**.

AD
Autosomal dominant

CLINICAL FEATURES

MAJOR CRITERIA

> 3 **Angiofibroma** (facial) or adenoma sebaceum

> 3 **Ash-leaf spots**



Tubers (cortical, subcortical)



Mental disability (intellectual disability)

Subependymal giant cell astrocytoma (SEGA)

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Hamartomas (CNS, retinal, and skin)

Mitral regurgitation



Rhabdomyoma (cardiac)

autosomal dominant



Angiomyolipoma (kidney)

Seizures
Shagreen patches

MINOR CRITERIA

- | | |
|---|---|
| <ul style="list-style-type: none"> Confetti skin lesions Gingival fibromas Periungual fibromas Dental enamel hypoplasia | <ul style="list-style-type: none"> Hamartomatous rectal polyps Cerebral white matter radial migration lines Retinal achromatic patches Bone cysts |
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ASSOCIATED FEATURES

- Seizures** (80-90%; infantile spasms, GTC)
- Developmental delay** (10-15%)
- Autism spectrum disorder
- Cardiac failure (usually present in prenatal period or in infancy)
- Renal failure

DIAGNOSIS

- Clinical Diagnosis:** requires **2 major features** OR **1 major and 2 minor features**.
- Genetic Testing:** autosomal dominant mutations in **TSC1 (Chr. 9)** or **TSC2 (Chr. 16)** are associated with tumor suppressor genes.
- Positive genetic testing in **75-80%** of patients.

MANAGEMENT

- Seizure management:** antiepileptic medications and surgery for refractory cases.
- Behavioral support:** early intervention for developmental delays and autism therapies.
- Skin lesions:** treatments may include topical therapies and lasers.
- Kidney care:** regular imaging for tumors and possible surgery.
- Pulmonary monitoring:** screening for lymphangioleiomyomatosis.
- Cardiac:** serial echocardiograms with regular cardiology follow up for rhabdomyomas.
- Genetic counseling:** for affected individuals and families.



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