TSC Global Awareness Day
May 15

UPDATED CLINICAL DIAGNOSTIC CRITERIA FOR TUBEROUS SCLEROSIS COMPLEX

GENETIC DIAGNOSTIC CRITERIA

The identification of either a *TSC1* or *TSC2* pathogenic mutation in DNA from normal tissue is sufficient to make a Definite Diagnosis of TSC. A pathogenic diagnosis is defined as a sequence variant that clearly prevents *TSC1* or *TSC2* protein production. Additionally, some mutations compatible with protein production (e.g. some missense changes) are well established as disease causing and also as sufficient to make a Definite Diagnosis of TSC. Other *TSC1* and *TSC2* variants may be consistent with a clinical diagnosis of TSC but are not considered to be diagnostic.

CLINICAL DIAGNOSTIC CRITERIA

MAJOR FEATURES	MINOR FEATURES
• Hypomelanotic macules (≥3)	• "Confetti" skin lesions
• Angiofibromas (≥3) or forehead plaque	• Dental enamel pits (>3)
• Ungual fibromas (≥ 2)	• Intraoral fibromas (≥2)
• Shagreen patch or multiple collagenomas	• Retinal achromic patch
• Multiple retinal hamartomas	• Multiple renal cysts
• Cortical dysplasias (≥3)*	• Nonrenal hamartomas
• Subependymal nodules (≥2)	
• Subependymal giant cell astrocytomas (≥2)	
• Cardiac rhabdomyoma	
• Lymphangioleiomyomatosis (LAM) **	
• Angiomyolipomas (AMLs; ≥ 2) **	

* Includes tubers and cerebral white matter radial migration lines
** A combination of the two Major clinical features LAM and AMLs without other features does not meet criteria for a Definite Diagnosis
Definite Diagnosis: 2 major features or 1 major feature with 2 minor features
Possible Diagnosis: Either 1 major feature, 1 major and 1 minor, or ≥ 2 minor features