

**MEDICAL GENOMICS LABORATORY: TSC1/TSC2 PHENOTYPIC CHECKLIST FORM**



Patient Name: \_\_\_\_\_  Male  Female Date of Birth \_\_\_/\_\_\_/\_\_\_  
Referring Physician: \_\_\_\_\_ Date of Exam \_\_\_/\_\_\_/\_\_\_

*Thank you for completing this form; phenotypic information may improve our ability to interpret your results.*

**DEMOGRAPHIC INFORMATION**

Ethnicity:  White  Black  Native American  Hispanic  Asian  Other:

**DIAGNOSIS**

2012 International TSC Consensus Conference Clinical Criteria: (Northrup et al. Pediatric Neuro. 2013 Oct;49(4):243-54.)

**(Criteria are listed below with major criteria marked by # and minor criteria marked by \*.)**

- Definite TSC (2 major or 1 major plus 2 minor features)
- Possible TSC (1 major or 2 or more minor features)
- Does not meet TSC Criteria

Clinical Concern for Mosaicism:  No  Yes  
Family history:  Sporadic  Familial  Unknown

Known Familial Mutation:  No  Yes (provide information: \_\_\_\_\_)

**Familial cases:** Please provide pedigree and details on the affection status of family members on a separate page. Attach prior mutational analysis if available.

**SIGNS AND SYMPTOMS** (Major criteria marked by # and minor criteria marked by \*.)

**Dermatological**

- 1) Hypomelanotic Macules:  0  1-2  ≥3#  Unknown
- 2) "Confetti" skin lesions:  None  Present\*  Unknown
- 3) Facial Angiofibromas:  None  1-3  >3#  Unknown
- 4) Shagreen Patch:  None  Present#  Unknown
- 5) Cephalic Fibrotic Plaque:  None  Present#  Unknown
- 6) Ungal/Periungal fibromas:  None  1-2  >2#  Unknown
- 7) Hyperpigmented Macules:  0  1-2  ≥3  Unknown

Please provide detail on size and location of the dermatological findings on page 3.  
A digital picture of the skin findings would be very helpful.

**Neurological (Imaging)**

- 8) Cortical Dysplasia:
  - Cortical Tubers:  None  Present#  Unknown
  - Cerebral White Matter Radial Migration Lines:  None  Present#  Unknown
- 9) Subependymal nodule (SEN):  None  Present#  Unknown
- 10) Subependymal Giant Cell Astrocytoma (SEGA):  None  Present#  Unknown
  - Histopathologically Confirmed?  No  Yes

**Neurological/Psychiatric (Clinical)**

- 11) Seizures:  None  Present (Describe type, if known: \_\_\_\_\_)  Unknown
- 12) Developmental/Intellectual Disabilities:  None  Present  Unknown
- 13) Behavioral/Psychiatric Diagnoses:  None  ADHD  Isolated hyperactivity  Aggression  
 Autism  unknown  Other (\_\_\_\_\_)

**Renal**

- 14) Angiomyolipomas:  None  1-2  >2#  Unknown
  - Histopathologically Confirmed?  No  Yes
  - Is a malignant angiomyolipoma present?  No  Yes
- 15) Renal Cell Carcinoma:  None  Present  Unknown
- 16) Renal Epithelial Cysts:  None  1-2  >2\*  Unknown
- 17) Polycystic Kidney Disease Features:  None  Present  Unknown

Note: If present, there may be increased concern for TSC-PKD contiguous deletion.

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**Pulmonary**

18) Lymphangioleiomyomatosis (LAM):  None  Present#  Unknown  
 Histopathologically Confirmed?  No  Yes

**Cardiac**

19) Rhabdomyomas:  None  Present#  Formerly/Prenatally present, but regressed  Unknown

**Dental**

20) Dental Enamel Pits:  None  1-3  >3\*  Unknown  
 21) Intraoral Fibromas:  None  1-2  >2\* (Location: \_\_\_\_\_)  Unknown

**Ophthalmological**

22) Retinal Hamartomas:  None  Single  Multiple#  Unknown  
 23) Retinal Achromic Patch:  None  Present\*  Unknown  
 24) Retinal Astrocytic Hamartomas:  None  Present  Unknown

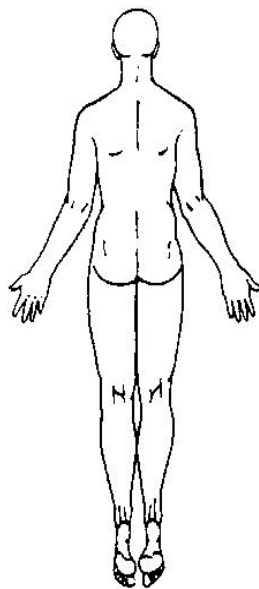
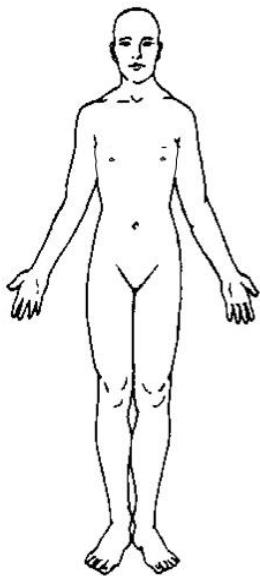
**Neuroendocrine**

25) Neuroendocrine Tumors:  None  Unknown  Yes (Specify type: \_\_\_\_\_)

**Other**

26) Nonrenal Hamartomas:  None  Present\*  Unknown  
 27) Additional Phenotypic Information:

**Indicate location/size of hypomelanotic macules or other dermatological lesions ↓**



<input type="checkbox"/>	Hypopigmented lesions
<input type="checkbox"/>	Hyperpigmented lesions
<input type="checkbox"/>	Shagreen patch
<input type="checkbox"/>	"Confetti" lesions

