

2012年International Tuberous Sclerosis Complex Consensus Conference  
TSC

**TSC**

**International Tuberous Sclerosis Complex Consensus Group**  
TSC

TSC





Westerveld M, et al. 2011, 2012 and 2013, Dufner Almeida LG, et al. 2020

- TSC1/TSC2
- ACMG America College of Medical Genetics Richards S, et al. 2015
- 10-15 TSC1/TSC2
- NGS High-read-depth NGS Tyburczy ME, et al. 2015 Giannikou K, et al. 2019 Peron A, et al. 2018

2022 4

B.

(3-5mm)	(Confetti)
(3)	(3)
(2)	(2)
( )	
2	
(LAM)*	
(2)*	

definite TSC 2 1 2 possible TSC 1 2 TSC1 TSC2 TSC TSC1 TSC2 TSC

LAM 2 TSC

Northrup H, et al. Pediatr Neurol 2021; 123: 50-66, 52-53.

NCCN National Comprehensive Cancer Network.





MRI SEN SEGA MRI CT MRI

EEG EEG 8 24 EEG

MRI CT

TAND

TAND





MRIT  
MRI

CT  
GFR > 60 mL/min/1.73 m<sup>2</sup>

MRI MRI  
MRI

GFR

MRI CT GFR

















SEGA

SEGA mTOR  
SEGA mTOR  
SEGA mTOR  
SEGA SEGA

mTOR  
mTOR 3

9.

9.7

9.7.1

SEGA 25 1 3 MRI  
1 3 SEGA SEGA

mTOR MRI

EEG 12 6 24  
3 EEG

2  
ACTH

EEG 24 EEG

mTOR

3

mTOR

TAND

TAND <https://tandconsortium.org/checklists/> 1  
TAND  
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TAND

SEGA

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3cm mTOR EXIST-2  
15mg 30 EXIST-2  
GFR  
EXIST-2

1

mTOR

mTOR

- 8.
- 8.3 BUN
- 11.
- 11.1.3 0.9%

MRI  
MRI CT 1 3 3cm  
1 1cm  
mTOR

HMB-45

mTOR GFR MRI CT

CT LAM LAM CT  
5 7 CT

CT LAM CT  
mTOR CT

CT LAM 6 1

mTOR LAM CT  
VEGF-D 1

VEGF-D 1

LAM 2

LAM

LAM

CT mTOR VEGF-D -D



1. mTOR signaling pathway is involved in the regulation of cell growth and metabolism. It is a central hub for signaling molecules that control cell growth and metabolism. The pathway is activated by growth factors and nutrients, leading to the phosphorylation of downstream targets such as S6 and 4E-BP1, which promotes protein synthesis and cell growth.

mTOR signaling pathway is involved in the regulation of cell growth and metabolism.

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□





VEGF mTOR

VEGF

FDA 3

mTOR



3) Krueger D. A, et al. Pediatr Neurol 2013; 49: 255-265.

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