

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 m²

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
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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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
CT

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 integrating signals from various growth factors and nutrients to control cell proliferation and differentiation.

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

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

4. 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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