

Genetic testing for TSC1 and TSC2

Genetic testing for TSC

Genetic testing for TSC1 and TSC2

Genetic testing for TSC1 and TSC2

Genetic testing for TSC1 and TSC2

Genetic testing for TSC1 and TSC2¹⁾ TSC1/TSC2 testing is recommended for patients with mTOR-inhibitor responsive tuberous sclerosis complex (TSC) or mammalian target of rapamycin (mTOR) inhibitor responsive TSC²⁾

Genetic testing for TSC1 and TSC2³⁾ TSC2 testing⁴⁾ is recommended for patients with TSC⁵⁾ and 1) TSC1/TSC2 testing is recommended for patients with TSC1 and TSC2 testing⁶⁾ and 1) TSC1 and TSC2 testing is recommended for patients with TSC1 and TSC2 testing⁷⁾

Genetic testing for TSC1 and TSC2⁶⁾ and 1) TSC1 and TSC2 testing is recommended for patients with TSC1 and TSC2 testing⁷⁾

Genetic testing for TSC1 and TSC2⁷⁾ and 1) TSC1 and TSC2 testing is recommended for patients with TSC1 and TSC2 testing⁸⁾

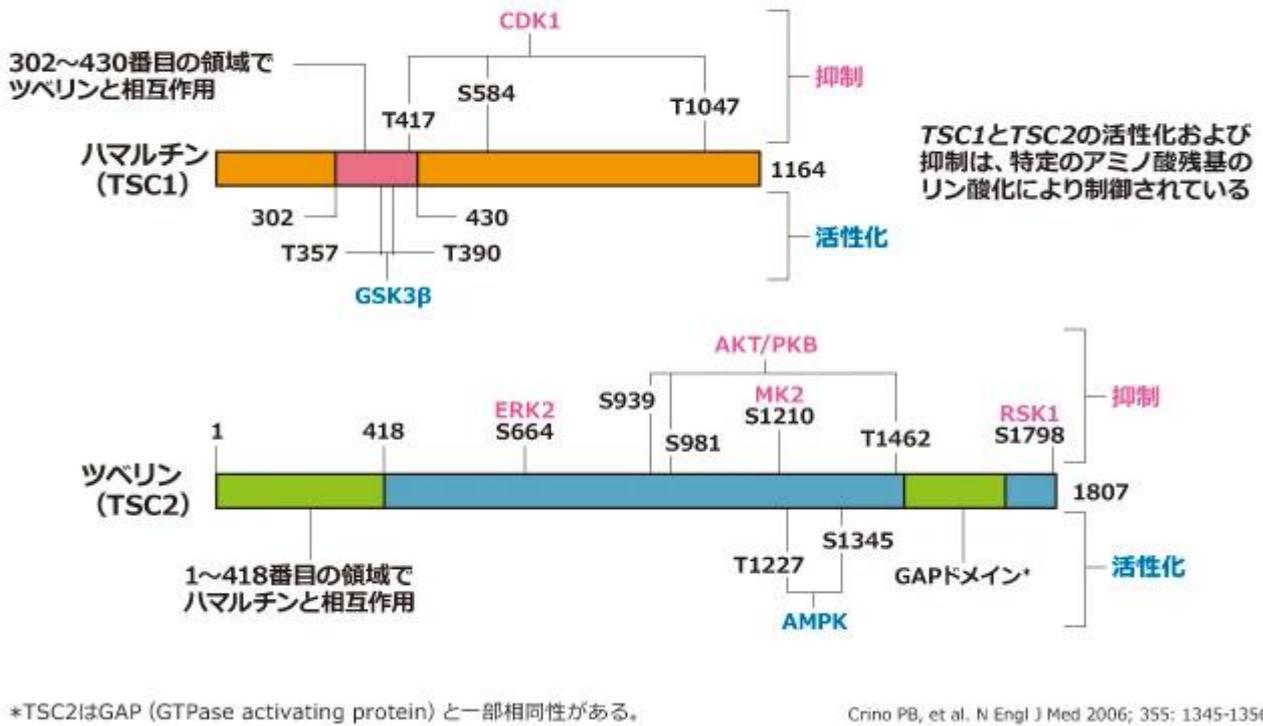
mTOR inhibitor responsive TSC1/TSC2 testing is recommended for patients with mTOR inhibitor responsive TSC⁷⁾

1) TSC1 and TSC2 testing

	TSC1	TSC2
Gene	9q34	16p13.3
Gene size	55kb	40kb
Exons	23	41
Gene size	8.6kb	5.5kb
Gene LOH	Yes	No
Gene	Hamartin	Tuberin
Gene size	1,164 amino acids 130kDa	1,807 amino acids 180kDa

Curatolo P, et al. Lancet 2008; 372: 657-668

図1 TSC1/TSC2遺伝子の構造



TSC1 / TSC2

TSC1とTSC2は、TSC1とTSC2の遺伝子変異が、TSC1は約10%、TSC2は約80%の患者で認められる。TSC1の変異は約10%、TSC2の変異は約60%、70%の患者で認められる。

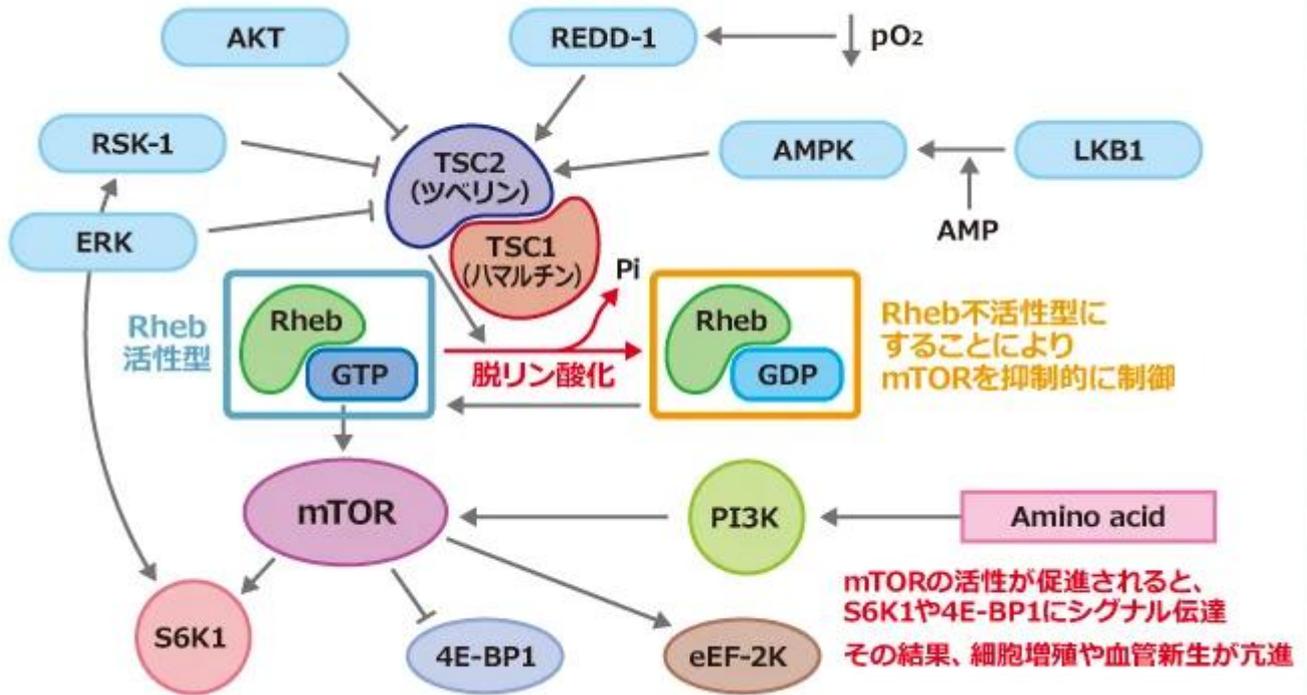
TSC1とTSC2は、TSC1とTSC2の遺伝子変異が、TSC1は約10%、TSC2は約80%の患者で認められる。²⁾

TSC1とTSC2は、TSC1とTSC2の遺伝子変異が、TSC1は約10%、TSC2は約80%の患者で認められる。PKD1(polycystic kidney disease type1)は、TSC1とTSC2の遺伝子変異が、TSC1は約10%、TSC2は約80%の患者で認められる。GAP(GTPase activating protein)は、TSC1とTSC2の遺伝子変異が、TSC1は約10%、TSC2は約80%の患者で認められる。

TSC1 / TSC2

	TSC1	TSC2
患者数	0	54
割合	0%	18%
患者数	26	26
割合	17%	9%
患者数	55	67
割合	36%	23%
患者数	60	55
割合	39%	19%
患者数	11	31
割合	7%	11%
患者数	2	59
割合	1%	20%
患者数	154	292

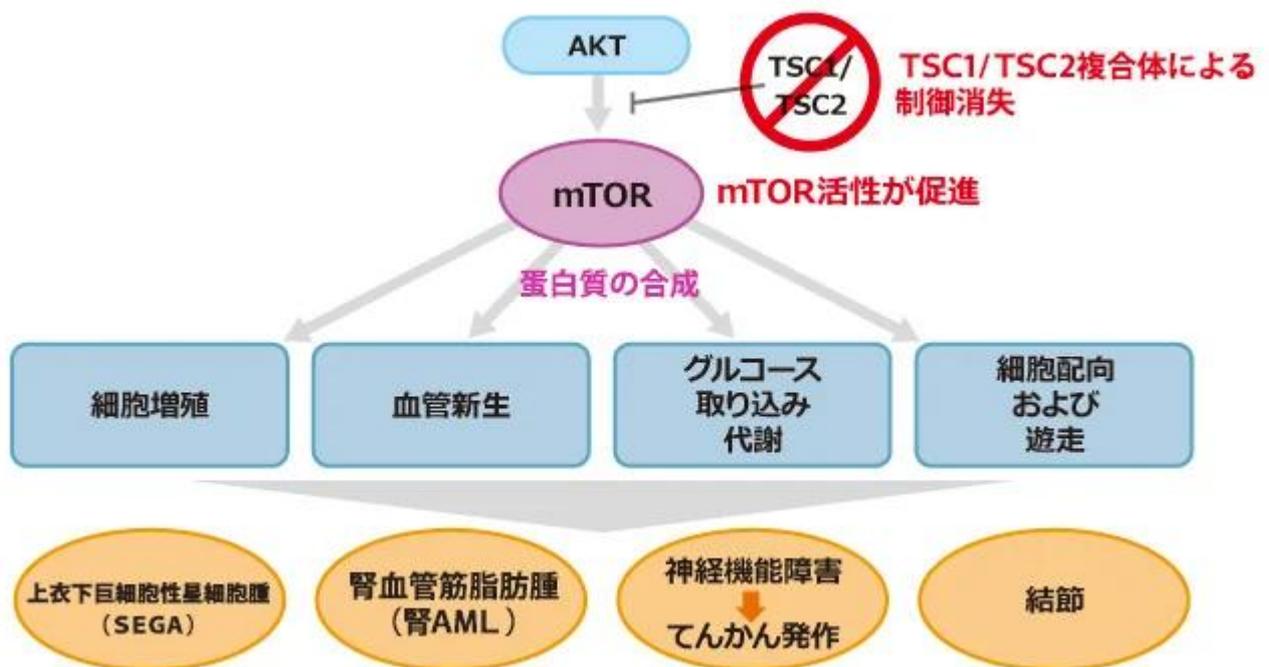
図2 TSC1/TSC2複合体によるmTOR活性の制御



Jozwiak J, et al. Lancet Oncol 2008; 9: 73-79

Image

図3 TSC1/TSC2遺伝子の変異によるmTOR活性の上昇



Crino PB, et al. N Engl J Med 2006; 355: 1345-1356, Moavero R, et al. Childs Nerv Syst 2010; 26: 1495-1504より作図

□□□□

- 1) Gunther M and Penrose LS. J Genet 1935; 31: 413-430
- 2) Cheadle JP, et al. Hum Genet 2000; 107: 97-114
- 3) Van Slegtenhorst M, et al. Science 1997; 277: 805-808
- 4) European chromosome 16 tuberous sclerosis consortium. Cell 1993; 75: 1305-1315

- 5) Curatolo P, et al. Lancet 2008; 372: 657-668
- 6) Crino PB, et al. N Engl J Med 2006; 355: 1345-1356
- 7) □□□□. □□□□ 2002; 106: 1556-1565
- 8) Verhoef S, et al. Am J Hum Genet 1999; 64: 1632-1637
- 9) Jozwiak J, et al. Lancet Oncol 2008; 9: 73-79
- 10) Moavero R, et al. Childs Nerv Syst. 2010; 26: 1495-1504

□□□□□□□□□□□□□□□□

□□□□□□

Source URL: https://www.pro.novartis.com/jp-ja/products/afinitor/tsc/overview_02