

TSC2

Identity

Hugo [TSC2](#)
Location 16p13.3

DNA/RNA

Description 41 exons; spans 41kb
Transcription At least 3 alternate splicings; 5.5kb mRNA complete cds; coding sequence: CDS 19-5442

Protein

Description Tuberin; 1807 amino acids; 190 kDaltons
Expression Expressed in most embryonic and adult tissues
Localisation Cytoplasmic
Function Potential GTPase activating protein (GAP) for [Rap1a](#) and/or Rab5; Interacts with hamartin ([TSC1](#) gene product) and Rabaptin-5
Homology 188 residues at the COOH terminus have homology to Rap/Ran GAP

Mutations

Germinal Large genomic deletions in <10% of cases; point mutations widely dispersed, with no cluster; truncating effect in 2/3 of cases
Somatic loss-of-heterozygosity in 2/3 of renal angiomyolipomas; Somatic mutations in angiomyolipomas and pulmonary lymphangiomyomatosis (LAM) cells from women with sporadic LAM

Implicated in

Entity [Tuberous Sclerosis](#) Complex (TSC)
Disease Autosomal dominant disease characterized by seizures, mental retardation, autism, benign tumors of the brain, heart, skin, kidney, and [malignant kidney tumors](#).

Entity Sporadic lymphangiomyomatosis (LAM)
Disease Lung disease affecting almost exclusively women, characterized by diffuse bilateral proliferation of abnormal smooth muscle cells in the lungs

External links

Nomenclature

Hugo	TSC2
GDB	TSC2
Entrez Gene	TSC2 7249 tuberous sclerosis 2

Cards

Atlas	TSC2ID184
GeneCards	TSC2
Ensembl	TSC2
CancerGene	TSC2
Genatlas	TSC2
GeneLynx	TSC2
eGenome	TSC2
euGene	7249

Genomic and cartography

GoldenPath	TSC2 - 16p13.3 chr16:2038600-2078713 + 16p13.3 (hg17-May_2004)
Ensembl	TSC2 - 16p13.3 [CytoView]
NCBI	Genes Cyto Gene Seg [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	TSC2

Gene and transcription

Genbank	AB014460 [SRS] AB014460 [ENTREZ]
Genbank	AC005600 [SRS] AC005600 [ENTREZ]
Genbank	L48522 [SRS] L48522 [ENTREZ]
Genbank	L48531 [SRS] L48531 [ENTREZ]
Genbank	L48537 [SRS] L48537 [ENTREZ]
RefSeq	NM_000548 [SRS] NM_000548 [ENTREZ]
RefSeq	NM_021055 [SRS] NM_021055 [ENTREZ]
RefSeq	NM_021056 [SRS] NM_021056 [ENTREZ]
RefSeq	NT_086834 [SRS] NT_086834 [ENTREZ]
AceView	TSC2 AceView - NCBI
TRASER	TSC2 Traser - Stanford
Unigene	Hs.90303 [SRS] Hs.90303 [NCBI] HS90303 [spliceNest]

Protein : pattern, domain, 3D structure

SwissProt	P49815 [SRS] P49815 [EXPASY] P49815 [INTERPRO]
Prosite	PS50085 RAPGAP [SRS] PS50085 RAPGAP [Expasy]
Interpro	IPR008938 ARM [SRS] IPR008938 ARM [EBI]

[Interpro](#) [IPR000331 Rap_GAP](#) [SRS] [IPR000331 Rap_GAP](#) [EBI]
[Interpro](#) [IPR003913 Tuberin](#) [SRS] [IPR003913 Tuberin](#) [EBI]
[CluSTr](#) [P49815](#)
[Pfam](#) [PF02145 Rap_GAP](#) [SRS] [PF02145 Rap_GAP](#) [Sanger] [pfam02145](#) [NCBI-CDD]
[Pfam](#) [PF03542 Tuberin](#) [SRS] [PF03542 Tuberin](#) [Sanger] [pfam03542](#) [NCBI-CDD]
[Blocks](#) [P49815](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [191092](#) [[map](#)]
[GENECLINICS](#) [191092](#)
[SNP](#) [TSC2](#) [dbSNP-NCBI]
[SNP](#) [NM_000548](#) [SNP-NCI]
[SNP](#) [NM_021055](#) [SNP-NCI]
[SNP](#) [NM_021056](#) [SNP-NCI]
[SNP](#) [TSC2](#) [GeneSNPs - Utah] [TSC2](#) [SNP - CSHL] [TSC2](#) [HGBASE - SRS]

General knowledge

[Family Browser](#) [TSC2](#) [UCSC Family Browser]
[SOURCE](#) [NM_000548](#)
[SOURCE](#) [NM_021055](#)
[SOURCE](#) [NM_021056](#)
[SMD](#) [Hs.90303](#)
[SAGE](#) [Hs.90303](#)
[Amigo](#) [function|GTPase activator activity](#)
[Amigo](#) [component|cytosol](#)
[Amigo](#) [process|endocytosis](#)
[Amigo](#) [component|membrane fraction](#)
[Amigo](#) [process|negative regulation of cell cycle](#)
[Amigo](#) [component|plasma membrane](#)
[Amigo](#) [process|protein folding](#)
[Amigo](#) [function|unfolded protein binding](#)
[BIOCARTA](#) [mTOR Signaling Pathway](#)
[BIOCARTA](#) [Control of Gene Expression by Vitamin D Receptor](#)
[PubGene](#) [TSC2](#)

Other databases

Other database <http://zk.bwh.harvard.edu/ts/TSC2-19.htm>

Probes

[Probe](#) [TSC2 Related clones \(RZPD - Berlin\)](#)

[PubMed](#)

[PubMed](#) [53 Pubmed reference\(s\) in LocusLink](#)

Bibliography

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The European Chromosome 16 Tuberous Sclerosis Consortium.

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Medline [8269512](#)

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Hum Mol Genet. 1994;3(10):1829-32.

Medline [7849708](#)

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Green AJ, Smith M, Yates JR.

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Medline [8162074](#)

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Henske EP, Neumann HP, Scheithauer BW, Herbst EW, Short MP, Kwiatkowski DJ.

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J Biol Chem. 1995;270(27):16409-14.

Medline [7608212](#)

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J Biol Chem. 1997;272(46):29301-8.

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Medline [9045618](#)

Tuberous sclerosis gene 2 product modulates transcription mediated by steroid hormone receptor family members.

Henry KW, Yuan X, Koszewski NJ, Onda H, Kwiatkowski DJ, Noonan DJ.

J Biol Chem. 1998;273(32):20535-9.

Medline [9685410](#)

Hamartin, the product of the tuberous sclerosis 1 (TSC1) gene, interacts with tuberin and appears to be localized to cytoplasmic vesicles.

Plank TL, Yeung RS, Henske EP.

Cancer Res. 1998;58(21):4766-70.

Medline [9809973](#)

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Medline [9580671](#)

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Medline [10052455](#)

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Cancer Res. 1999;59(6):1206-11.

Medline [10096549](#)

Tsc2(+/-) mice develop tumors in multiple sites that express gelsolin and are influenced by genetic background.

Onda H, Lueck A, Marks PW, Warren HB, Kwiatkowski DJ.

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Medline [10491404](#)

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Medline [10823953](#)

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Kobayashi T, Minowa O, Sugitani Y, Takai S, Mitani H, Kobayashi E, Noda T, Hino O.

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Medline [11438694](#)

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URL : <http://www.infobiogen.fr/services/chromcancer/Genes/TSC2ID184.html>

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