

## TSC1 (Tuberous Sclerosis 1)

### Identity

Note	tumor-suppressor
Other names	<b>KIAA0234</b>
Hugo	<a href="#">TSC1</a>
Location	9q34 between D9S1199 and D9S1830

### DNA/RNA

Description	23 exons, of which 21 are coding; start codon is in exon 3. Different splice variants in the 5'UTR
Transcription	1 major detected transcript at 8.6 kb, 2 additional transcripts at 4 and 2.5 kb respectively; open reading frame : 3492 bp

### Protein

Note	called hamartin
Description	1164 amino acids, 130 kDa; C-terminal coiled-coil domain (amino acids 730 to 965)
Expression	housekeeping
Localisation	cytoplasmic localisation
Function	binds to ezrin, member of the ERM (ezrin-moesin-radixin) actin binding proteins
Homology	no strong matches with vertebrate proteins

### Mutations

Germinal	germline mutations cause tuberous sclerosis, known as a multiple hamartoma syndrome, autosomal dominant pattern of inheritance. Mutations are inactivating by protein truncation, no missense mutations documented. Some cases of germline mosaicism
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### Implicated in

Entity	<a href="#">Tuberous Sclerosis</a>
Note	Classified as one of the phakomatosis, a group of disorders which also includes <a href="#">neurofibromatosis type 1</a> and <a href="#">neurofibromatosis type 2</a> , <a href="#">von Hippel-Lindau disease</a> and Sturge-Weber syndrome
Disease	Growth of a variety of benign tumors (hamartomas) in multiple organs. Disease is clinically variable. Organs most frequently involved are the

heart, skin, brain, lung and kidneys.

**Prognosis** Lesions in the brain are associated with severe manifestations of TSC. Seizures occur in about 85% of the patients and they often start in the first year of life. About 50% of the children with seizures develop mental retardation. In the second and third decade of life, renal problems are found in the majority of TSC patients. Most characteristic renal abnormalities are cysts and angiomyolipomas. [PROGNOSIS](#)  
[Renal cell carcinoma](#) develops occasionally in TSC patients.  
Life expectancy largely depends on complications caused by the lesions in the brain and the kidneys.

## External links

### Nomenclature

[Hugo](#) [TSC1](#)  
[GDB](#) [TSC1](#)  
[Entrez\\_Gene](#) [TSC1\\_7248](#) tuberous sclerosis 1

### Cards

[Atlas](#) [TSC1ID183](#)  
[GeneCards](#) [TSC1](#)  
[Ensembl](#) [TSC1](#)  
[CancerGene](#) [TSC1](#)  
[Genatlas](#) [TSC1](#)  
[GeneLynx](#) [TSC1](#)  
[eGenome](#) [TSC1](#)  
[euGene](#) [7248](#)

### Genomic and cartography

[GoldenPath](#) [TSC1 - 9q34](#) [chr9:132796290-132849574 - 9q34.13](#) (hg17-May\_2004)  
[Ensembl](#) [TSC1 - 9q34.13 \[CytoView\]](#)  
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]  
[OMIM](#) [Disease map \[OMIM\]](#)  
[HomoloGene](#) [TSC1](#)

### Gene and transcription

[Genbank](#) [AF234185](#) [SRS] [AF234185](#) [ENTREZ]  
[Genbank](#) [AF274228](#) [SRS] [AF274228](#) [ENTREZ]  
[Genbank](#) [AF274229](#) [SRS] [AF274229](#) [ENTREZ]  
[Genbank](#) [AL445645](#) [SRS] [AL445645](#) [ENTREZ]  
[Genbank](#) [AF013168](#) [SRS] [AF013168](#) [ENTREZ]  
[RefSeq](#) [NM\\_000368](#) [SRS] [NM\\_000368](#) [ENTREZ]  
[RefSeq](#) [NM\\_001008567](#) [SRS] [NM\\_001008567](#) [ENTREZ]

<a href="#">RefSeq</a>	<a href="#">NT_086756</a> [SRS] <a href="#">NT_086756</a> [ENTREZ]
<a href="#">AceView</a>	<a href="#">TSC1</a> AceView - NCBI
<a href="#">TRASER</a>	<a href="#">TSC1</a> Traser - Stanford
<a href="#">Unigene</a>	<a href="#">Hs.370854</a> [SRS] <a href="#">Hs.370854</a> [NCBI] <a href="#">HS370854</a> [spliceNest]
<b>Protein : pattern, domain, 3D structure</b>	
<a href="#">SwissProt</a>	<a href="#">Q92574</a> [SRS] <a href="#">Q92574</a> [EXPASY] <a href="#">Q92574</a> [INTERPRO]
<a href="#">Interpro</a>	<a href="#">IPR008938 ARM</a> [SRS] <a href="#">IPR008938 ARM</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR007483 Hamartin</a> [SRS] <a href="#">IPR007483 Hamartin</a> [EBI]
<a href="#">CluSTr</a>	<a href="#">Q92574</a>
<a href="#">Pfam</a>	<a href="#">PF04388 Hamartin</a> [SRS] <a href="#">PF04388 Hamartin</a> [Sanger] <a href="#">pfam04388</a> [NCBI-CDD]
<a href="#">Blocks</a>	<a href="#">Q92574</a>
<b>Polymorphism : SNP, mutations, diseases</b>	
<a href="#">OMIM</a>	<a href="#">605284</a> [ <a href="#">map</a> ]
<a href="#">GENECLINICS</a>	<a href="#">605284</a>
<a href="#">SNP</a>	<a href="#">TSC1</a> [dbSNP-NCBI]
<a href="#">SNP</a>	<a href="#">NM_000368</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">NM_001008567</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">TSC1</a> [GeneSNPs - Utah] <a href="#">TSC1</a> [SNP - CSHL] <a href="#">TSC1</a> [HGBASE - SRS]
<b>General knowledge</b>	
<a href="#">Family Browser</a>	<a href="#">TSC1</a> [UCSC Family Browser]
<a href="#">SOURCE</a>	<a href="#">NM_000368</a>
<a href="#">SOURCE</a>	<a href="#">NM_001008567</a>
<a href="#">SMD</a>	<a href="#">Hs.370854</a>
<a href="#">SAGE</a>	<a href="#">Hs.370854</a>
<a href="#">Amigo</a>	<a href="#">process Rho protein signal transduction</a>
<a href="#">Amigo</a>	<a href="#">process cell adhesion</a>
<a href="#">Amigo</a>	<a href="#">process negative regulation of cell cycle</a>
<a href="#">BIOCARTA</a>	<a href="#">mTOR Signaling Pathway</a>
<a href="#">PubGene</a>	<a href="#">TSC1</a>
<b>Other databases</b>	
<b>Probes</b>	
<a href="#">Probe</a>	<a href="#">TSC1 Related clones (RZPD - Berlin)</a>
<b>PubMed</b>	
<a href="#">PubMed</a>	<a href="#">27 Pubmed reference(s) in LocusLink</a>

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