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Gene Section
Mini Review

TSC1 (tuberous sclerosis 1)

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Identity

Other names: KIAA0234

HGNC (Hugo): TSC1

Location: 9q34

Local order: Between D9S1199 and D9S1830.

Note: Tumor-suppressor.

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.

Implicated in

Tuberous Sclerosis

Note
Classified as one of the phakomatosis, a group of disorders which also includes neurofibromatosis type 1 and neurofibromatosis type 2, von Hippel-Lindau disease 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.
Renal cell carcinoma develops occasionally in TSC patients.
Life expectancy largely depends on complications caused by the lesions in the brain and the kidneys.

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