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Gene Section
Mini Review

TSC2 (tuberous sclerosis 2)

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Identity

HGNC (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 lymphangioleiomyomatosis (LAM) cells from women with sporadic LAM.

Implicated in

Tuberous Sclerosis Comples (TSC)

Disease

Autosomal dominant disease characterized by seizures, mental retardation, autism, benign tumors of the brain, heart, skin, kidney, and malignant kidney tumors.

Sporadic lymphangioleiomyomatosis (LAM)

Disease

Lung disease affecting almost exclusively women, characterized by diffuse bilateral proliferation of abnormal smooth muscle cells in the lungs.

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