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

Germline
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 Complex (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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