TSC1

Tuberous sclerosis 1 (TSC1), also known as Hamartin, is a protein that in humans is encoded by the TSC1 gene.

TSC1 forms a multi-protein complex with TSC2 and TBC1D7 known as the TSC complex. This complex negatively regulates mTORC1 signaling by functioning as a GTPase-activating protein (GAP) for the small GTPase Rheb, an essential activator of mTORC1. The TSC complex has been implicated as a tumor suppressor.

Defects in this gene can cause tuberous sclerosis, due to a functional impairment of the TSC complex.

Defects in TSC1 may also be a cause of focal cortical dysplasia. TSC1 may be involved in protecting brain neurons in the CA3 region of the hippocampus from the effects of stroke.

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2 tumor suppressor genes: TSC1 (on chromosome 9q34) codes for hamartin and TSC2 (on chromosome 16p13) encodes tuberin

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Interactions TSC1 has been shown to interact with:

AKT1, NEFL, PLK1, and TSC2.