TSC2

Tuberous sclerosis complex is caused by a mutation of either of two genes, TSC1 and TSC2, which code for the proteins hamartin and tuberin respectively. These proteins act as tumor growth suppressors, agents that regulate cell proliferation and differentiation.

TSC2 is predicted to encode a 1784-amino acid tumor suppressor protein that may function, in part, as a GTPase-activating protein for Rap1. Given the high incidence of central nervous system abnormalities in individuals affected with tuberous sclerosis, the expression of TSC2 in developing and adult nervous system tissues was examined. Reverse transcription-PCR, Northern blot, and in situ hybridization analyses demonstrated high levels of expression of TSC2 in the adult brain and developing central nervous system. Abundant TSC2 expression was detected in the adult cerebellum, hippocampus, and olfactory bulb, with lower levels of expression observed in other tissues, including heart and kidney. This enrichment of TSC2 expression in neurons in the central nervous system suggests unique roles for this tumor suppressor gene product in the development and differentiation of nervous system tissues [1].

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Geist RT, Gutmann DH. The tuberous sclerosis 2 gene is expressed at high levels in the cerebellum and developing spinal cord. Cell Growth Differ. 1995 Nov;6(11):1477-83. PubMed PMID: 8562486.