Tuberous Sclerosis Complex (TSC; Bourneville Disease)

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

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Authors’ objectives
Tuberous sclerosis complex (TSC) is an inherited neurocutaneous disorder characterized by variable expressivity of benign tumors (hamartomas) in multiple organ systems. TSC is a highly penetrant, complex disease that causes seizures, cognitive impairment, skin lesions, and, infrequently, malignancy, in addition to hamartomas in different organ systems. Cardiac rhabdomyomas, seizures, and skin lesions are the most common symptoms reported at initial presentation. Consensus clinical diagnostic criteria have been developed for the clinical diagnosis of TSC. Although TSC is inherited in an autosomal dominant manner, it is estimated that two thirds of cases are sporadic or de novo. Two genes have been associated with TSC: tuberous sclerosis complex 1 (TSC1) on chromosome 9 at band q34, which produces the protein hamartin; and tuberous sclerosis complex 2 (TSC2) on chromosome 16 at band p13, which produces the protein tuberin.

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