Condition: Tuberous sclerosis complex

Inheritance:

Autosomal Dominant**.**

Genetic etiology:

Mutation of either *TSC1* or *TSC2* genes on chromosomes 9 and 16, respectively. Both function as tumor suppressor genes.

Frequency:

Approximately 1/6,000.

Clinical features:

TSC is a multisystem hamartomatous disorder. Dysplastic foci in the central nervous system are associated with seizures and developmental delay. Cardiac rhabdomyomas may occur in infancy, but regress spontaneously. Retinal hamartomas usually are asymptomatic. Renal cysts may occur, especially in patients with a contiguous deletion of *TSC2* and *PKD1* on chromosome 16; renal angiomyolipomas are common and may case bleeding or renal impairment. Women are at risk for life-threatening lymphangiomyomatsis of the lung. Skin lesions include hypomelanotic macules and collagenous plaques.

Management:

Symptomatic management and monitoring for complications; mTOR inhibitor everolimus may be used to treat symptomatic subependymal giant cell astrocytomas or renal angiomyolipomas.

Genetic counseling:

Affected individuals at 50% risk of transmitting a gene mutation; both familial and de novo cases occur. Genetic testing is available, though not all pathogenic mutations can be identified. Penetrance is complete.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.