

Self-assessment questions: Tuberous sclerosis: from gene to targeted therapy

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- 1 The following are part of the current diagnostic criteria for tuberous sclerosis:
 - (a) Seizures
 - (b) Intellectual disability
 - (c) Angiomyolipoma
 - (d) Shagreen patch
 - (e) Lymphangiioleiomyomatosis
- 2 Which statement(s) describe the genetics of tuberous sclerosis?
 - (a) Tuberous sclerosis is an autosomal dominant condition
 - (b) Most cases result from a new mutation with neither parent affected
 - (c) Pathogenic mutations in the genes encoding tuberous sclerosis 1 or 2 (*TSC1* or *TSC2*) lead to inhibition of mammalian target of rapamycin complex 1 (mTORC1) activity
 - (d) *TSC2* is a GTPase-activating protein
 - (e) *TSC1* is located on chromosome 11
- 3 Which statement(s) describe the clinical features of tuberous sclerosis?
 - (a) Infantile spasms occur in one-third of infants with tuberous sclerosis
 - (b) Renal angiomyolipomas (AMLs) occur in approximately 20% of adults with tuberous sclerosis
 - (c) Lymphangiioleiomyomatosis (LAM) almost exclusively affects females
 - (d) Hypomelanotic macules can be present at birth
 - (e) Cardiac rhabdomyomas usually regress spontaneously

Answers to these self-assessment questions can be found on page s92.