Tuberous sclerosis complex (TSC) is an autosomal dominant genetic disorder resulting in the growth of benign tumours or hamartomas affecting any organ system. Up to 90% of patients present with seizures due to growth of benign tumours in the brain. Although significant morbidity is associated with cardiac, renal and pulmonary involvement, the neurological aspects and particularly seizures associated with tuberous sclerosis are extremely difficult to treat. The hallmark cutaneous manifestations include ash-leaf spots (figure 1), angiofibromas, ungual fibromas, subungual red comets and splinter haemorrhages (figure 2). Up to 80% of patients also have renal tumours known as angiomyolipomas (figure 3) which can result in spontaneous aneurysmal bleeds and haemorrhagic shock due to formation of abnormal vasculature.

TSC is caused by mutations in either the TSC1 or TSC2 genes. TSC1 and TSC2 form a complex responsible for the regulation of the mammalian target of rapamycin (mTOR) pathway. Drugs that inhibit mTORC1, such as sirolimus and everolimus, have demonstrated efficacy for the treatment of multiple aspects of TSC, including renal angiomyolipomata, refractory epilepsy associated with brain tumours, and lymphangioleiomyomatosis.
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REFERENCES
