

Missed tuberous sclerosis complex with multi-system complications in a single patient

ABSTRACT

Tuberous sclerosis complex (TSC) is an autosomal dominant disorder characterized by widespread clinical manifestations. Early diagnosis is usually possible when typical TSC related skin lesions and neurologic presentations are detected in young patients. Undiagnosed TSC patients are at increased risk of morbidity and mortality as disease progression will inevitably lead to complications. While case reports of single complications in pediatric patients have been documented, to the best of our knowledge, multi-system complications of TSC in adults have yet to be reported in the literature. We present a case of tuberous sclerosis diagnosed in adulthood with complications involving the central nervous, renal and respiratory systems. This case highlights the need for a multidisciplinary approach in the management of TSC as well as the role of imaging in both diagnosis and intervention.