Abstract

Objectives: To report the case of a patient diagnosed with tuberous sclerosis complex (TSC), describe its clinical features, diagnosis, and to attract attention on the fact that after 40 years of follow-up, the patient has presented practically all the manifestations described in the literature. Methods: A 42 year-old man diagnosed with TSC presented the emergency department due to left lumbar pain and self-limited gross hematuria. On clinical examination patient was haemodynamically stable, but with decrease in haemoglobin (6.8 g/dL). Abdominal CT scan showed a 20 cm diameter heterogeneous mass in the left kidney suggesting hemorrhage of angiomyolipoma. Results: Left radical nephrectomy was performed and the pathological study of the surgical specimen confirmed the diagnosis of angiomyolipoma. Immunohistochemical staining was positive with HMB-45. Conclusions: To recommend that patients with TSC be evaluated by a multidisciplinary group of clinicians, including urologists, neurologists and dermatologists. As patients with TSC survive into adulthood they will require more intervention by the urologist. CT scan is usually enough for the diagnosis of angiomyolipomas. Complete nephrectomy is appropriate when the whole kidney has been replaced by angiomyolipoma. The identification of molecular markers (HMB-45) facilitates histopathological diagnosis.

Keywords

Tuberous sclerosis complex, Angiomyolipoma, Kidney