Case report

We report 37-year-old patient with cortical and subependymal tubers suspicious for tuberous sclerosis. Tuberous sclerosis, also known as Bourneville’s disease, is a phacomatosis with an incidence 1:6000 – 1:10000. Classical findings include the triad of adenoma sebaceum, seizures, and mental retardation. The inheritance is autosomal dominant; however, spontaneous mutation is described up to 66 % [1-9].

Possible radiological findings in context with TS are multiple benign tumors, such as cardiac rhabdomyoma, renal angiomyolipoma, as well as neurological lesions: cortical and subependymal tubers accompanied by white matter changes (Figure 1). Furthermore, lymphangioleiomyomatosis, multifocal micronodular pneumocyte hyperplasia and renal cysts, as well as bone, liver and alimentary tract involvement may be found [2-5].

Moreover, the patient revealed multiple lesions in both kidneys, as found in abdominal ultrasound: Consecutive MR imaging confirmed the presence of multiple small lesions in both kidneys. These lesions showed high signal on T1-weighted in phase gradient-echo images and signal loss on opposed phase gradient-echo images (Figure 2), as found in fat containing benign tumors.

Renal angiomyolipomas are benign tumors with variable amounts of abnormal vessels, immature smooth-muscle and fat cells. Angiomyolipomas usually are asymptomatic, when they are detected. However, follow-up, especially in case of large tumors, is obligatory due to the risk of rupture with consequent bleeding [3,6]. In patients with tuberous sclerosis bilateral angiomyolipomas of the kidneys are found frequently (55-75%) [1]. Due to the MR appearance of these lesions, the suspected diagnosis was bilateral renal angiomyolipoma in tuberous sclerosis.

An adequate therapy using Votubia® (everolimus), which is recommended for the treatment of tuberous sclerosis patients with kidney tumors, was initiated [8]. Follow-up MR examinations showed size reduction in some of the lesions after therapy with Votubia® (everolimus).

Disclosure

No conflicts of interest. No grants or financial supports.

References


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Key words: tuberous sclerosis, bourneville’s disease, renal angiomyolipomas, magnetic resonance

Received: August 02, 2017; Accepted: August 10, 2017; Published: August 12, 2017


