# TUBEROUS SCLEROSIS COMPLEX (TSC): RENAL AND CARDIOLOGICAL MANIFESTATIONS REQUIRE MULTIDISCIPLINARY APPROACH AND CAN BE PART OF THE UNDERSTANDING OF OTHER PREVALENT CONDITIONS

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INTRODUCTION: Tuberous sclerosis complex (TSC) arises from the onset of benign hamartomatous tumors related to early changes in cell differentiation, proliferation, and migration (associated with some mutations in genes such as TSC 1, 2 and PKD). Facial angiofibromas, although easier to recognize, often appear only in late childhood and adolescence. Neurological findings (subependymal hyperdense nodules and cortical tufts, the formation of subependymal giant astrocytomas -SEGA, change in CSF drainage, and frequent seizures) usually predominate. However, renal and cardiological manifestations have an important impact on this complex. We present the multicentric experience accumulated in the diagnostic and therapeutic management in different clinical settings of patients affected by TSC.

METHODS: A cross-sectional study involving patients with TSC in follow-up at Specialized Centers in Nephrology, Genetics, and Neurology in Brazil and Italy.

RESULTS: The clinical-radiological findings of 42 patients from 5 centers (3 hospitals (n = 24) and 2 neuroradiology satellite services (n = 18) were evaluated. Some demographic data is presented in Fig. 1. Mean creatinine was 0.76 ± 0.39 mg / dL. All patients had involvement of the tuberous sclerosis complex in more than one system. Almost all had neurological manifestations (clinical-radiological, in 95 of the children). About cardiological manifestations, they were more common among pediatric patients (35 of adults). Presence of cardiac hamartomas can be identified in echocardiography, even during the intrauterine life (prematurity may occur in TSC). Hypertension was also reported.

Nephrologic manifestations were present in 43 of adult patients in this cohort. See Fig. 2. The investigation included echography (92) and MRI (11 of nephrologic patients. AML was identified in 22 presented cysts (mean 2.3 cm, from 0.4-6.0 cm) and 33 of the patients (4, mainly for neurological symptoms, but recent evidence shows a significant response in the stabilization of angiomyolipoma dimensions.

CONCLUSION: Successful management of this degenerative disease requires a cross-disciplinary approach. A thorough knowledge of this condition may also pave the way for a better understanding of other kidney diseases that share genetic alterations with TSC (such as polycystic kidney disease).