

Better Late Than Never... But Earlier Would Have Been Better: Adult Tuberous Sclerosis Poster

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Abstract

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David M. Stufflebeam ¹, Khadijah Mohiuddin ¹, Shivangi Patel ²

1. Internal Medicine, Mount Sinai Hospital, Chicago, USA 2. Family Medicine, Mount Sinai Hospital, Chicago, USA

Corresponding author: David M. Stufflebeam, davidstufflebeam@students.aucmed.edu

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Abstract

Introduction:

Tuberous sclerosis (TS) is an autosomal dominant neurocutaneous disorder caused by mutations in TSC1 or TSC2, leading to hyperactivation of the mTOR pathway and formation of hamartomas in multiple organs including the brain, skin, kidneys, heart, and lungs. TS is associated with epilepsy, cognitive impairment, and dermatologic lesions such as facial angiofibromas. Although prevalence is estimated at 1 in 20,000, variable presentation can lead to delayed recognition or underdiagnosis. This case highlights the importance of recognizing multisystem manifestations of TS.

Case Description:

A 63 year old male with history of prostate cancer treated with radiation therapy, pernicious anemia managed with vitamin B12 injections, hypertension, hyperlipidemia, neurocysticercosis, seizure disorder, and developmental delay presented for routine follow up. The patient reported feeling well and compliant with medications. A CT abdomen and pelvis from 2023 demonstrated multiple bilateral renal angiomyolipomas, including two large lesions in the left kidney meeting criteria for increased risk of spontaneous hemorrhage. Radiologic findings raised concern for tuberous sclerosis or von Hippel Lindau syndrome, and outpatient interventional radiology consultation was recommended, but not completed. Physical examination revealed facial angiofibromas, first documented in 2022, raising suspicion for TS.

Discussion:

TS has variable expression and may remain unrecognized until adulthood. Recognition of renal angiomyolipomas with seizure disorder, developmental delay, and facial angiofibromas should prompt evaluation for TS. Early identification allows surveillance for renal hemorrhage, neurologic complications, pulmonary disease, and other systemic involvement. Early diagnosis permits targeted therapy and guideline directed screening enabling physicians to prevent multi organ complications and improve long term outcomes.