CASE REPORT OF TUBEROUS SCLEROSIS IN PATIENT PRESENTED WITH VASAMOL CONSUMPTION

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ABSTRACT :

Tuberous sclerosis complex (TSC) is a rare genetic neurocutaneous syndrome characterized by glial tumors in the brain and retina, fibromas in various organs, and a classic triad of epilepsy, intellectual disability, and adenoma sebaceum. However, nearly 50% of affected individuals have normal intelligence, and 15% do not experience seizures. Neurological symptoms typically appear in childhood, while skin manifestations often emerge later. We present the case of a 31-year-old woman diagnosed with TSC, who initially sought medical attention for vasamol consumption. The patient developed rhabdomyolysis, acute fulminant hepatitis, and acute kidney injury (AKI) following the ingestion. Her hepatitis was managed with N-acetylcysteine infusion, steroids, and glutathione. Due to worsening renal function, she underwent two dialysis sessions. On examination, multiple small, yellow papules consistent with adenoma sebaceum were noted on her face and neck. She also had hypopigmented ash-leaf macules, confetti lesions on her arms and legs, and shagreen patches on her back, which she had since childhood. The patient had no history of seizures or neurological symptoms, and a significant family history of TSC was noted. Brain MRI revealed cortical tubers, and ultrasonography showed a hyperechoic renal angiomyolipoma in her left kidney. Despite the absence of typical neurological symptoms, the clinical and radiological findings led to the diagnosis of TSC.

INTRODUCTION :

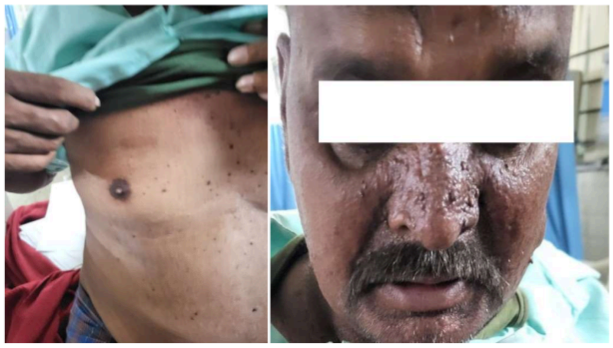
Tuberous sclerosis (TS), also known as epiloia, is a rare genetic disorder of autosomal-dominant inheritance with a prevalence ranging from one in 6,000 to one in 12,000. Both sexes and all ethnic groups can be affected. It is a multisystem disorder involving the brain, skin, heart, kidneys, eyes, lungs, and liver, and it typically manifests in late childhood. Von Recklinghausen and later Désirée-Magloire Bourneville described this disorder in the 19th century, and key pathological findings in some affected individuals were noted. It is known that mutations in either of the two genes, TSC1 and TSC2, which encode for the proteins hamartin and tuberin, respectively, are the causes of TS. These proteins are tumor growth suppressors, which regulate cell proliferation and differentiation. The classic triad of TS is seizures, intellectual disability, and angiofibromas, but this occurs in only 29% of patients with TS. It is important to note that skin involvement is crucial for suspecting the diagnosis of TS. We present the radiologic findings and skin lesions of a 31-year-old female who presented following vasamol consumption.

CASE REPORT :

A 31-year-old woman with no prior comorbidities presented with a history of vasamol consumption, reporting abdominal discomfort, severe myalgia, and throat discomfort. These symptoms were indicative of rhabdomyolysis, acute hepatitis, and acute kidney injury (AKI).

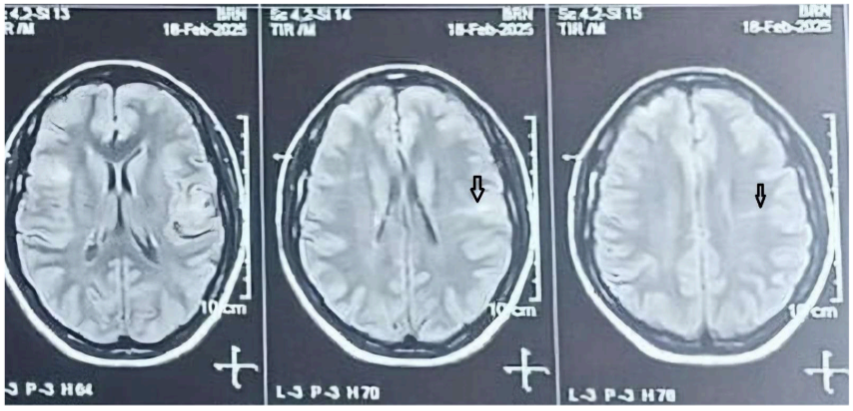
On examination, her vital signs and neurological assessment were normal, and her intelligence quotient was within the average range. Dermatological findings included multiple small, smooth, speckled yellow papules on her central face and neck, consistent with adenoma sebaceum. She also exhibited hypopigmented ash-leaf macules, confetti lesions on her arms and legs, and shagreen patches on her back, which she had noticed since childhood. She had no history of seizures or neurological symptoms. A significant family history of TSC was noted in her father, and both of her daughters displayed similar skin lesions.

ASH LEAF MACULES ADENOMA SEBACEUM CONFETTI LEISIONS

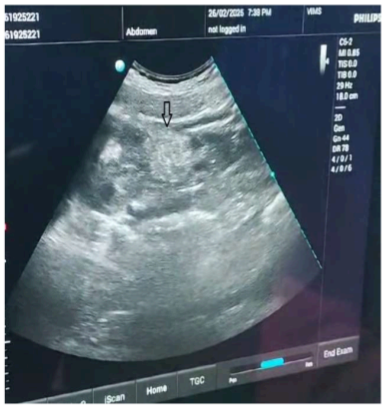


ASH LEAF MACULES(father) ADENOMA SEBACEUM (father)

Laboratory investigations revealed a creatinine level of 8.5 mg/dL at presentation, along with markedly elevated liver enzymes (SGOT: 3256 U/L, SGPT: 1567 U/L). Ultrasonography identified a hyperechoic renal angiomyolipoma in her left kidney, while MRI revealed cortical tubers. Retinal examination was unremarkable. Genetic testing was not performed due to financial constraints.



An ill-defined cortical lesion with long TR hyperintensity was observed in the frontoparietal region.



Ultrasonography of the left kidney revealed a renal angiomyolipoma

Her hepatitis was managed with N-acetylcysteine infusion, steroids, and glutathione. Due to worsening renal function, she required two sessions of dialysis for rising creatinine levels and oliguria. Upon discharge, her creatinine had improved to 2.4 mg/dL with adequate urine output, and her liver enzymes had returned to baseline.

DISCUSSION:

Tuberous sclerosis (TS) is a rare autosomal dominant neurocutaneous syndrome characterized by hamartomatous lesions in multiple organs, including the brain and kidneys. This genetic disorder has no predilection for sex or ethnicity, with an estimated prevalence ranging from one in 6,000 to one in 12,000. Approximately two-thirds of cases occur sporadically. TS typically presents with the classic Vogt triad: mental retardation, epilepsy, and facial angiofibromas. However, nearly half of affected individuals have normal intelligence, and about a quarter do not experience seizures. Given this variability, it is not surprising that our patient demonstrated normal cognitive function at this stage, placing them within the subset of individuals with preserved intelligence.

Mutations in the tumor-suppressor genes TSC1 (encoding hamartin) or, more commonly, TSC2 (encoding tuberin) underlie the pathogenesis of TS. These mutations lead to a loss of inhibition of the mammalian target of rapamycin (mTOR) pathway, resulting in the growth of hamartomas across various organs. In the brain, these include cortical tubers, subependymal nodules, and subependymal giant cell astrocytomas. In the kidneys, TS manifests as renal angiomyolipomas and renal cysts, while in the lungs, it may present as lymphangioleiomyomatosis. Other affected organs include the heart, where cardiac rhabdomyomas are commonly seen in fetuses and neonates before regressing in infancy, and the skin, where features such as angiofibromas, Shagreen patches, and hypomelanotic macules are observed. Our patient exhibited classic MRI findings of cortical tubers, and abdominal ultrasonography revealed renal angiomyolipomas and cysts, further supporting the diagnosis.

Neurological manifestations of TS include seizures, autism, and behavioral or psychiatric disorders. Seizures, which occur in approximately 80–90% of patients, typically begin in the first year of life and range from subtle focal seizures and infantile spasms to generalized seizures. However, during our interaction, the patient did not exhibit any signs of autism or psychiatric disturbances. Cutaneous features of TS include hypomelanotic macules (90%), facial angiofibromas (75%), and Shagreen patches (20–30%). Hypomelanotic macules are often present at birth, with most lesions becoming evident within the first two years of life. Facial angiofibromas usually develop during preschool years, appearing as small pink or red papules distributed in a characteristic “butterfly” pattern across the cheeks and nose while sparing the upper lip. This pattern was observed in our patient.

Given the combination of cutaneous manifestations and imaging findings, a definitive diagnosis of TS was established. This aligns with the Tuberous Sclerosis Complex Consensus, which now requires the presence of two or more distinct lesion types rather than multiple lesions of the same type within a single organ system.

CONCLUSIONS:

TS is a neurocutaneous syndrome and it is a rare genetic disorder. In addition to the clinical history of seizures and cutaneous lesions, imaging plays an important role in diagnosis. The moment this is suspected the individual's parents should be counselled and enrolled in multi-disciplinary treatment programs. It is true there is no cure but symptomatic treatment is available. Support groups should be formed in the country and individuals encouraged to join.

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