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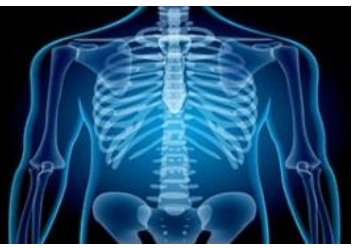

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A case of a silent heart tumor: Cardiac rhabdomyoma in a patient of Bourneville disease with West syndrome

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Abstract
Tuberous Sclerosis Complex (TSC), also known as Bourneville disease, is a genetic neurocutaneous disorder characterized by the formation of hamartomas. These benign growths arise from the embryonic ectoderm and can affect various organs, including the skin, nervous system, kidneys, lungs, and heart.
We present the case of a 1-year-old male diagnosed with Tuberous Sclerosis Complex (TSC), who was found to have a cardiac rhabdomyoma, as confirmed through 2D echocardiography and MRI. The importance of utilizing various imaging modalities in diagnosing and managing such cases is also emphasized.

Keywords: TSC, rhabdomyoma, tuberous sclerosis, cardiac tumour, echocardiography

Introduction
Tuberous Sclerosis Complex (TSC) is a neurocutaneous disorder that can impact virtually any organ system in the body. It is primarily characterized by skin lesions and neurological problems, with seizures being the most common manifestation. Clinically, Tuberous sclerosis complex typically presents with a characteristic triad of seizures, intellectual disability, and adenoma sebaceum. Patients with Tuberous Sclerosis Complex may present with vague symptoms, including chest pain, abdominal discomfort, or hematuria. In some cases, the condition is discovered incidentally during the evaluation of renal cysts, angiomyolipomas, oncocytomas, or renal cell carcinoma. Lung involvement in Tuberous sclerosis complex typically presents as well-defined cysts. Even in asymptomatic individuals with a significant family history, it is recommended to conduct screening through physical and clinical examinations.

Case report
A 14-month-old male patient presented with multiple episodes of seizures that began at 4 months of age. Initially controlled with medication, the seizures later progressed to epileptic spasms. On examination, the child displayed developmental delay for his age, with regressed motor and language milestones. Clinical examination revealed numerous depigmented patches with feathery borders on the face, bilateral upper limbs, trunk, and buttocks (Figure 1).
An electroencephalogram (EEG) was conducted, revealing multiple high-amplitude generalized waves. At 5 months of age, a non-contrast computed tomography (CT) scan of the brain showed multiple calcified ependymal nodules along the margins of the lateral ventricles, along with several hyperdense areas in the subcortical white matter (Figure 2).
Further imaging with Magnetic Resonance Imaging (MRI) revealed multiple hypointense foci on Gradient Recalled Echo (GRE) sequences, located in the periventricular region.Additionally, several ill-defined T2/FLAIR (Fluid-Attenuated Inversion Recovery) hyperintense lesions were observed in the cortical and subcortical white matter of both cerebral hemispheres. These lesions did not show restricted diffusion or blooming on GRE, suggesting the presence of cortical and subcortical tubers (Figure 3).

The child was later diagnosed with Tuberous Sclerosis Complex in association with West syndrome

Further screening with a 2D echocardiogram revealed a pedunculated hyperechoic mass measuring 9 x 7 mm, attached to the interventricular septum in the mid left ventricular (LV) cavity (Figure 4). A subsequent cardiac MRI showed a well-defined lesion isointense to the myocardium across all sequences, with early phase enhancement (1 minute after the perfusion scan) observed in the LV cavity, originating from the septum along the mid anteroseptal segment. This finding was most consistent with a diagnosis of rhabdomyoma (Figure 5).

Discussion

Cardiac rhabdomyomas are frequently observed in Tuberous Sclerosis Complex (TSC), an autosomal dominant genetic disorder caused by mutations in the TSC1 or TSC2 genes, which encode the proteins hamartin and tuberin, respectively (1). These two proteins form a complex that helps regulate cell growth and proliferation. This complex plays a crucial role in regulating cellular growth and preventing uncontrolled cell division. In conditions like Tuberous Sclerosis Complex (TSC), mutations in either the TSC1 or TSC2 genes impair the function of this complex, resulting in the development of benign tumors known as hamartomas. It is crucial to differentiate rhabdomyomas in children from other types of cardiac tumors, such as cardiac myxomas (typically found in the atria, attached to the interatrial septum), teratomas (located in the pericardium), as well as hemangiomas and fibromas [1].

Rhabdomyomas can be detected via prenatal ultrasound between 20 to 30 weeks of gestation [2]. Although most patients are asymptomatic, any symptoms that do occur are generally linked to the size and location of the tumours. If present, monitoring is advised with an electrocardiogram (ECG) annually or biannually to identify any hemodynamic issues or to look for signs of regression, along with yearly Holter monitoring to detect significant arrhythmias. However, in cases where they lead to obstruction, heart failure, or severe, refractory arrhythmias a surgical intervention might be necessary. Additionally, there complete removal can be challenging as they are often noted arising from the deep myocardium.

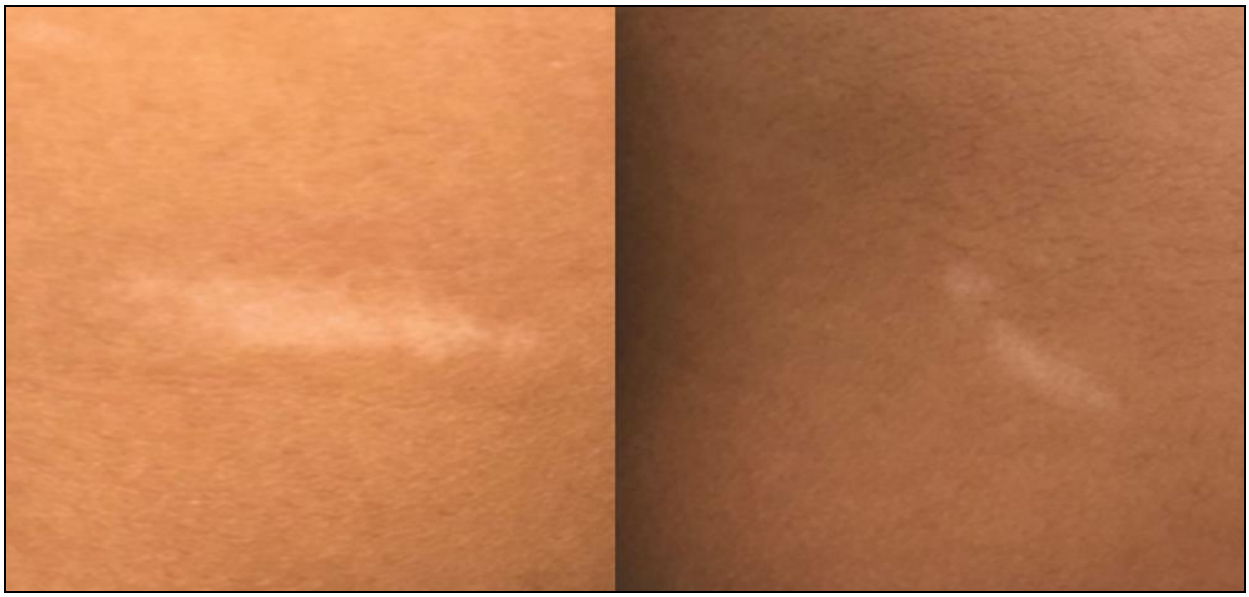

Fig 1: Numerous depigmented patches with feathery margins on the face, bilateral upper limbs, trunk and buttocks

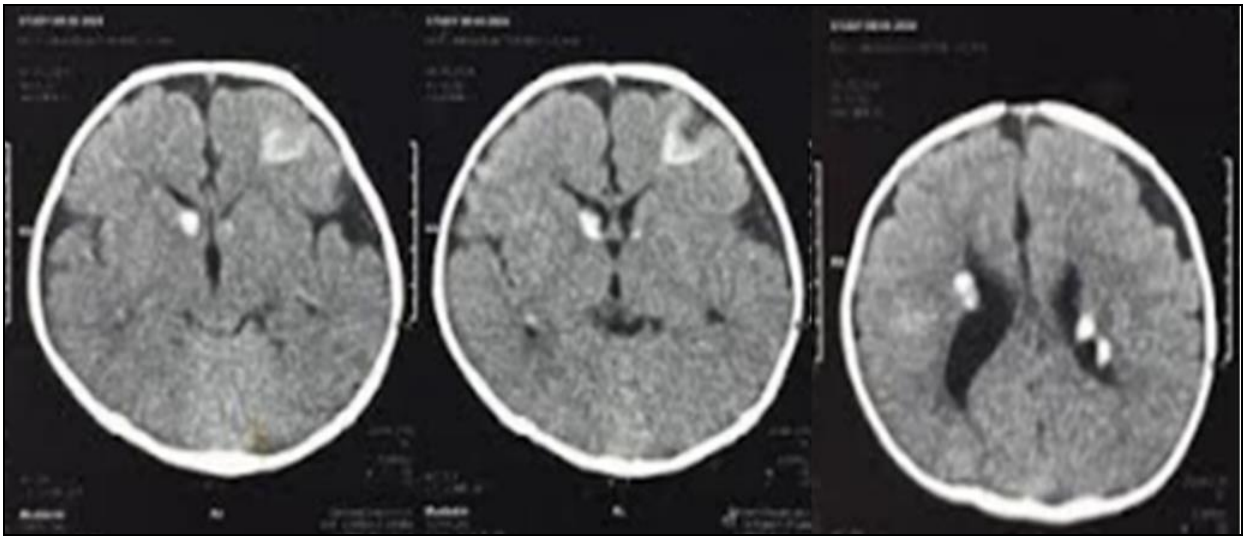

Fig 2: Multiple calcified ependymal nodules along the margins of lateral ventricle and few hyperdense cortical tubers in the subcortical white matter

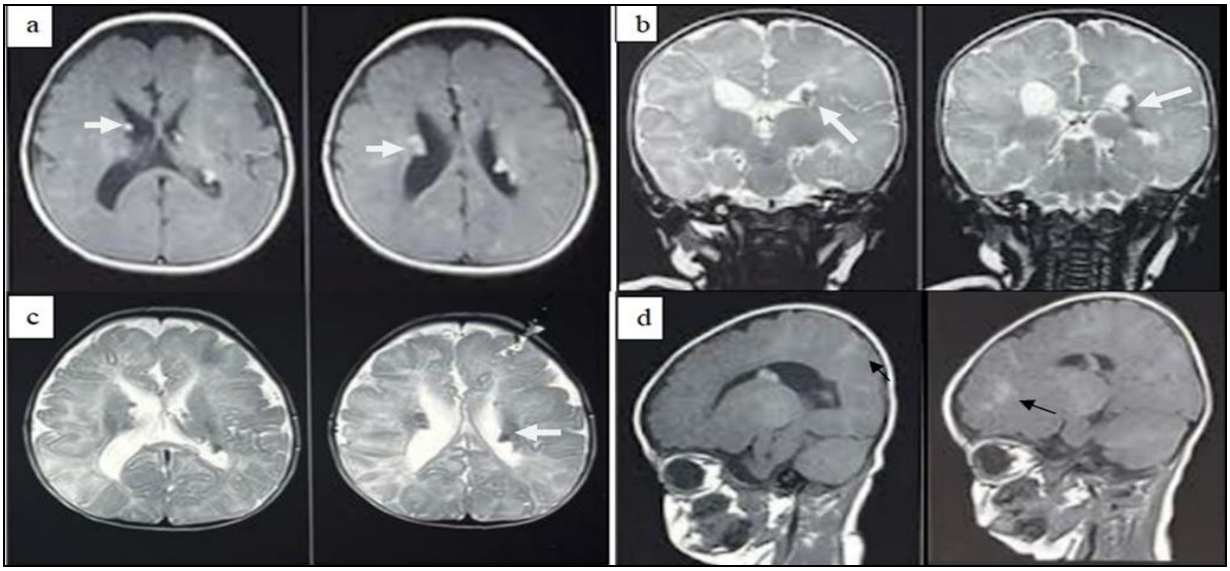

Fig 3: FLAIR axial (3a), T2 coronal (3b), T2 axial (3c) and T1 sagittal (3d) images showing multiple subependymal nodules (white arrows) in the periventricular location and few ill-defined cortical and subcortical tubers (black arrows) in cortical and subcortical white matter of the bilateral cerebral hemispheres.

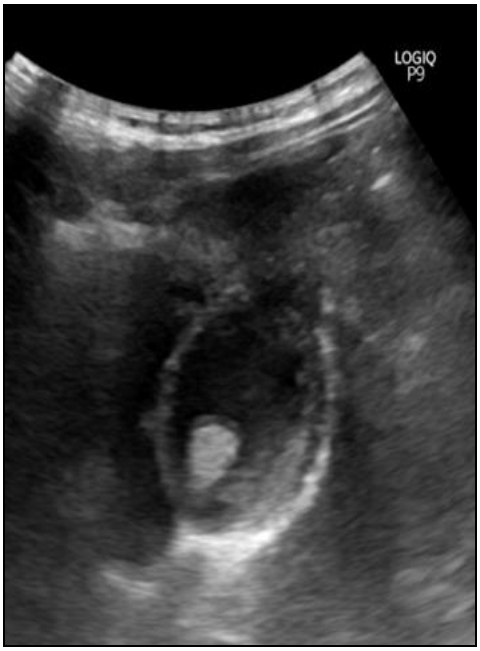

Fig 4: On ultrasound, an echogenic focus noted in the left ventricle along the interventricular septum in the mid segment.

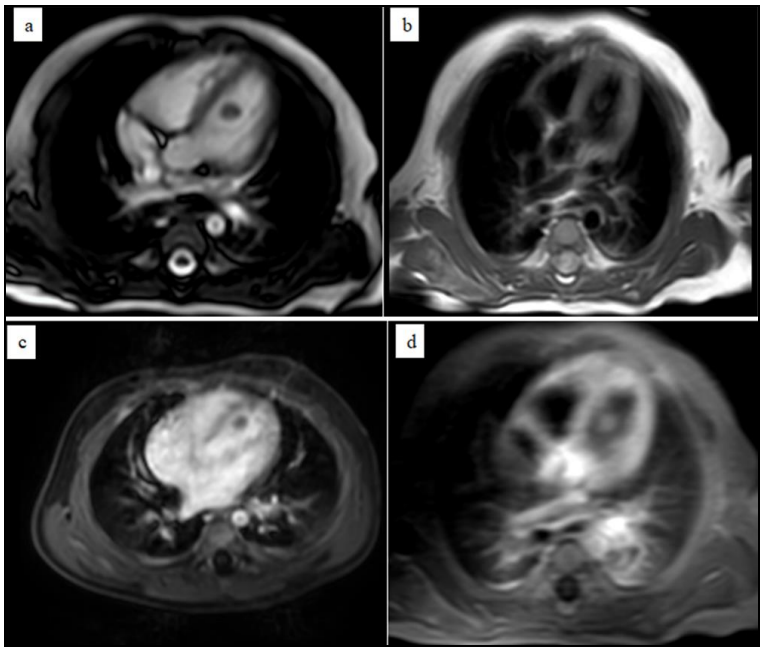

Fig 5: A well-defined lesion appearing isointense to the myocardium on (a) WB (white blood) and (b) BB (black blood) sequence in the LV cavity arising from the septum.On (c) T1 FS (fat saturated) WB image and (d) T1 FS PC (fat saturated post contrast) images showing early phase enhancement.

Conclusion

The spectrum of imaging findings in Tuberous Sclerosis Complex (TSC) is typically observed in older patients. Common manifestations include periventricular tubers and/or subependymal giant cell astrocytomas (SEGAs) in the central nervous system (CNS), renal angiomyolipomas (AML) in the kidneys, and lymphangioleiomyomatosis (LAM) in the lungs, with the latter often diagnosed in females of childbearing age. This case is unique due to the patient's age of only two years, presenting with imaging findings of cardiac rhabdomyoma and periventricular calcified tubers.

Conflict of Interest

Not available

Financial Support

Not available

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