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The Role of Imaging in the Diagnosis of Bourneville-Lhermitte-Dercum Disease (Tuberous Sclerosis) in Bamako: A Case Report with Literature Review

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ABSTRACT

Introduction: *Tuberous sclerosis complex (TSC) is a multisystem genetic disorder with autosomal dominant inheritance. The aim of our study was to evaluate the role of imaging in the diagnosis of this disease.*

Case Report: *We present the case of a 12-year-old boy referred to the radiology department of the "Les Etoiles" medical clinic for a brain CT scan due to febrile seizures. Clinical examination revealed angiofibromas on the face, symmetrically distributed around the nose, cheeks, forehead, and chin. Oral, dental, and ophthalmological examinations were normal. Brain CT scan, performed with and without contrast, revealed multiple calcified subependymal, periventricular (bilateral), and right cerebellar nodules. Abdominal ultrasound was normal. Based on these clinical and radiological findings, the diagnosis of tuberous sclerosis complex was established.*

Conclusion: *The diagnosis of tuberous sclerosis complex is based on the combination of suggestive clinical manifestations and characteristic radiological findings.*

Keywords

Tuberous sclerosis complex, Imaging, Bamako.

Introduction

Tuberous sclerosis complex (TSC) is part of the phakomatoses, a group of disorders characterized by the presence of congenital developmental abnormalities in the form of malformations, benign tumors, or hamartomas [1,2]. It is a multisystem genetic disease with autosomal dominant inheritance, first described in 1862 by Von Recklinghausen, and named "tuberous sclerosis" by Bourneville in 1880 [1,2]. The incidence reported in the literature is 1/6,000 to 1/10,000 births [2]. The first familial cases were reported in 1910; the mode of inheritance was clarified in 1935, but the responsible genes were not identified until 1987 and 1992 [3]. It is caused by mutations in two genes, Tuberous Sclerosis 1 or Tuberous Sclerosis 2 (TSC1 or TSC2). These genes are tumor suppressor genes and encode two proteins, hamartin (TSC1) and tuberin (TSC2), which interact to form the hamartin-tuberin complex. This complex plays a crucial role in inhibiting the mammalian target of rapamycin (mTOR) signaling pathway. The mTOR protein is a kinase that plays a central role in regulating cell proliferation and growth. The uninhibited mTOR signaling pathway is responsible for tumor formation in TSC. The classic triad of facial angiofibromas, epilepsy, and intellectual disability is present in less than one-third of cases. Clinical, radiological, and genetic criteria allow for a precise diagnosis [1,4]. Treatment is complex and requires multidisciplinary teams [1]. Imaging plays a central role and is an essential component of the multidisciplinary management of patients with tuberous sclerosis complex. The aim of our study was to investigate the value of imaging in the diagnosis of this disease.

Case Report

This was a 12-year-old male child referred to the radiology department of the "Les Etoiles" medical clinic for a brain CT scan in the context of recurrent febrile seizures that had started 6 years earlier. The medical history revealed a significant finding: the patient had experienced recurrent, but infrequent, seizures since the age of six. There was no family history of tuberous sclerosis complex (TSC). Clinical examination revealed angiofibromas, consisting of multiple papules of varying colors (chocolate to brownish) on the face, symmetrically distributed around the nose, cheeks, forehead, and chin (Figure 1). There was no intellectual disability. The oral and ophthalmological examinations were normal.

Figure 1: Photographic image showing the distribution of angiofibromas on the face.

The abdominal and pelvic ultrasound scan showed that the liver, spleen, pancreas, kidneys, bladder, and internal reproductive organs appeared normal (Figure 2).

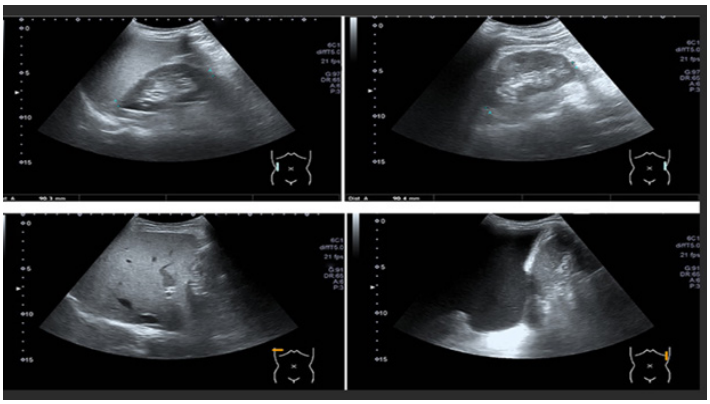

Figure 2: Ultrasound images showing a normal liver, kidneys, and spleen.

The brain CT scan, performed both with and without contrast agent, revealed multiple calcified nodules in the subependymal and periventricular regions bilaterally, as well as in the right cerebellum (Figure 3).

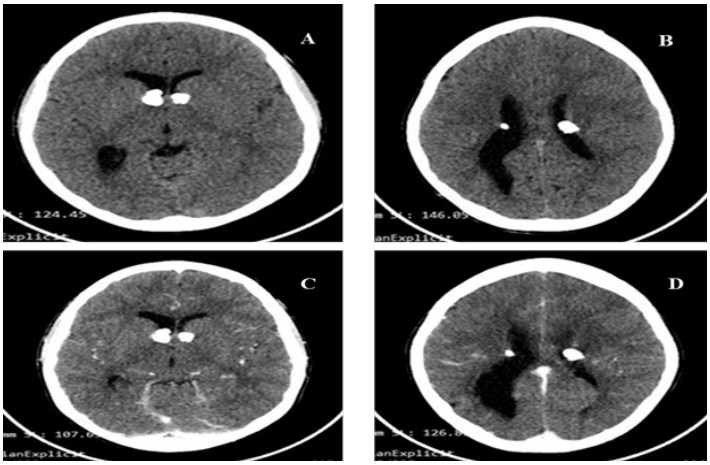

Figure 3: Axial CT scans of the head and brain, without contrast agent (A, B) and with contrast agent (C, D), showing calcified subependymal nodules in the periventricular region, suggestive of tuberous sclerosis.

Discussion

Tuberous sclerosis complex (TSC) is an autosomal dominant genetic disorder characterized by lesions affecting multiple organs and variable clinical manifestations [1,4]. Neurological and skin manifestations predominate: epilepsy occurs in 80-96% of cases, intellectual disability in 50-85% of cases, and facial angiofibromas in 85% of cases. Other features may include bilateral retinal hamartomas (50-87%), cardiac rhabdomyoma (30-50%), and benign tumors in the brain ventricles, kidneys, lungs, and bones [5]. Epilepsy and facial angiofibromas were present in our patient. Abdominal ultrasound may reveal renal cysts or non-renal hamartomas. In our patient, the ultrasound was normal. Brain CT scans may show cortical tubers (hypodense cortical lesions that do not enhance with contrast), subependymal nodules, which are often calcified and rarely enhance with contrast, and a giant cell subependymal astrocytoma, a rapidly growing lesion often located near the foramen of Monro, measuring 2-3 cm in diameter, heterogeneous, partially calcified, and enhancing with contrast, associated with hydrocephalus [4]. In our case, we observed the presence of completely calcified subependymal and cerebellar nodules. There were no signs of astrocytoma or giant cell tumor. The clinical and radiological diagnostic criteria for tuberous sclerosis complex (TSC) include 11 major and 6 minor features [4]. The diagnosis should be considered if there is a genetic criterion or 2 major clinical and/or radiological criteria, or 1 major clinical and/or radiological criterion combined with 2 minor (clinical and/or radiological) criteria [4]. These diagnostic criteria are as follows:

- **he genetic criterion:** a pathogenic mutation in the TSC1 or TSC2 gene.
- **Major clinical and radiological criteria:** Angiofibromas (≥3) or a cephalic fibrous plaque, hypomelanotic macules (≥3 and at least 5 mm in diameter), nail-bed fibromas (≥2), shagreen patch, retinal hamartoma,

cortical tubers, subependymal nodules, giant cell subependymal astrocytomas, cardiac rhabdomyoma, lymphangioleiomyomatosis, renal angiomyolipoma (≥2).

- **Minor clinical and radiological criteria:** enamel pits (≥3), oral fibromas (≥2), confetti-like skin lesions, non-renal hamartoma, multiple renal cysts, retinal hypopigmented macule.

The diagnosis of tuberous sclerosis complex (TSC) in our case was based on two major clinical and radiological criteria. In this particular case, there were no cognitive impairments. The absence of early seizures appears to be a key factor influencing the cognitive prognosis [6]. Oral, dental, ophthalmological, cardiac, renal, and pulmonary examinations were all normal in this patient. Tuberous sclerosis complex is a condition present in all populations, with no significant differences in prevalence among different ethnic groups. However, various cultural and socio-ethnic factors can influence the recognition, diagnosis, and management of this disorder.

Conclusion

Bourneville's tuberous sclerosis is a multisystem genetic disorder with a very wide spectrum of clinical manifestations, ranging from mild to severe. A multidisciplinary approach is essential for early diagnosis and appropriate, timely management. Imaging, particularly computed tomography (CT) of the brain, was crucial in establishing the diagnosis in our case, revealing characteristic lesions in addition to the skin manifestations. It remains an essential tool for diagnosis, monitoring, and the prevention of complications.

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