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Original Research Paper

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TUBEROUS SCLEROSIS: PRESENTATION OF A CASE AND REVIEW OF THE LITERATURE

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ABSTRACT Tuberous Sclerosis Complex (TSC) also known as Bournneville disease. TSC is a multisystemic genetic disorder with autosomal dominant inheritance, of variable expression, which is mainly characterized by the presence of benign tumors or hamartomas in the nervous system and skin, but which may also be present in the heart, kidney, lung and other organs. The most frequent symptom is epilepsy, affecting 80-90% of patients with TSC which manifests itself in childhood between 1 to 3 years of age. We present a case of sporadic onset tuberous sclerosis with epilepsy that had a causal link with TSC after admission to the emergency room in a convulsive status.

KEYWORDS : Tuberous Sclerosis, Neurocutaneous Syndromes, Bourneville Phakomatosis

INTRODUCTION

Tuberous Sclerosis Complex (TSC) also known as Bournneville disease, Pringle disease, Bourneville-Pringle disease and formerly as "Epiloia" which is an acronym for epilepsy, low IQ and sebaceous adenoma (1). The prevalence of TSC is 1/100,000. Its incidence is 1 in 6,000 - 10,000 births. At present it is estimated that in the world there are about 2,000,000 people affected by the disease. The disorder can affect both adults and children, people of both sexes, and any ethnic group; but women may have more marked signs (2,3). We present a case of sporadic onset tuberous sclerosis with late manifestations.

Case report

A 25-year-old male with a history of mild mental retardation and epilepsy managed with Carbamezepine. He was admitted to the emergency department in a convulsive state with an approximate duration of 30 minutes, a patient with generalized tonic-clonic seizures, gaze deviation but without sphincter relaxation, he was admitted to the emergency area for medical management.

The physical examination showed a patient was hemodynamically stable, tachycardic, and subsequently under sedation with a Ramsay 5 mental state. Angiofibromas were found in the left nasolabial surcon, 5 hypopigmented spots on the trunk, and retinal hamartoma was found in the fundus retinal.

A computerized axial tomography (CT) scan without contrast medium was found in axial, subependymal nodules in the right lateral ventricle (**Figure 1**). Non-contrast nuclear magnetic resonance imaging (NMR) showed poorly defined hypointense images in both frontal lobes in coronal T1 section, which were also observed in the FLAIR sequence (**Figure 2**).

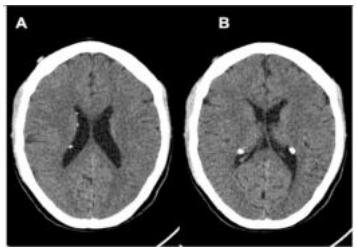

Figure 1. A-B. Cranial CT without contrast medium, axially, shows subependymal nodules in the right lateral ventricle.

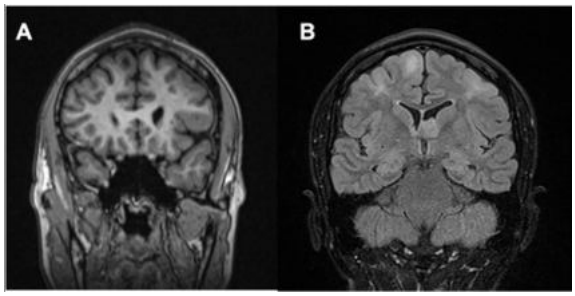

Figure 2. MRI shows cortical tubercles. **A.** Coronal T1 poorly defined hypointense images in the frontal lobes bilaterally. **B.** Coronal FLAIR poorly defined hyperintense images in the frontal lobes bilaterally.

The patient had a diagnosis of tuberous sclerosis due to the presentation of major and minor criteria, management was started with Vigabatrin, with a 2-year follow-up without new seizure episodes.

DISCUSSION

Tuberous sclerosis complex is a disease that is transmitted by autosomal dominant inheritance, it has a variable penetrance and expressiveness, which is the cause of the different phenotypes that the pathology manifests, even within the same family, which can make its recognition difficult (2). Most of the cases occur sporadically with a family history in about 30% of the cases, with the majority being due to de novo mutations, as happened in the present case (4,5).

About 96% of patients with TSC present manifestations in the skin and/or in the adnexa, these lesions evolve depending on the age of the patient, appearing in relative order, although this can be variable, they present as hypopigmented or hypomelanotic macules known as lesions in "Ash leaf", which are the most constant, but not pathognomonic of the disease, followed by facial angiofibromas, shagreen patches and subungual fibromas, such as those presented in the patient (1,2).

The most frequent neurological symptom is epilepsy, affecting 80-90% of patients with TSC which manifests in childhood between 1 and 3 years of age, as happened with our patient (6). In most infants, epilepsy begins with infantile spasms or focal seizures that can become generalized over time, however, at different ages, virtually all types of seizures have been documented in people with TSC (1). It is common to find in people with this condition refractory epilepsy to treatment in a higher proportion than the general population (3).

The diagnosis of TSC can be established through clinical signs and suggestive findings in the requested diagnostic images; of these criteria our patient met three major criteria (hypopigmented spots, subependymal nodules and facial angiofibromas) and a minor criterion (retinal hamartomas).

The initial treatment in TSC is anticonvulsants coupled with the ketogenic diet, which can be very effective. The first line for partial seizures and infantile spasms approved by the FDA in 2009 is vigabatrin (VGB), which irreversibly inhibits g-aminobutyric acid transaminase (GABA), which increases GABA levels, favoring anticonvulsant properties; one of the adverse effects associated with the administration of GBV is the irreversible loss of vision associated with toxicity at the level of the retina (6). Even in patients with a diagnosis of TSC who do not present symptomatic manifestations but alterations in the EEG, it is recommended to start early treatment with GBV.

To conclude, tuberous sclerosis is a phenotypic disease, it can present with mild presentations and patients have normal lifestyles, however, they have high morbidity and mortality associated with moderate and severe presentations.

Ethical standards and patient consent

We declare that the patient described in this study gave informed consent prior to inclusion in this study.

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