

Full Length Case Report

SHEER NEUROLOGICAL FORM OF TUBEROUS SCLEROSIS: ABOUT AN OBSERVATION AT FANN DEPARTMENT OF NEUROLOGY IN DAKAR (SENEGAL)

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ABSTRACT

Tuberous sclerosis complex (TSC) is an important neurocutaneous syndrome to diagnose because of its phenotypic variability and the often serious nature of its manifestations. The pure neurological form of STB is rare in clinical practice. We report here a recent case of tuberous sclerosis with pure neurological symptomatology in a Senegalese child. This was a 2-year-old child with no prior personal history except for a delayed psychomotor development. No family history had been reported. He was received in neuropediatric consultation for partial motor seizures secondarily generalized for 15 months. The neurological and neuropsychological examination showed a mild cognitive deficit and a bilateral pyramidal syndrome. Despite complet clinical evaluation, there was no dermatological lesion. The rest of the clinical examination was strictly normal. The diagnosis of tuberous sclerosis has been made on the basis of neurological manifestations (refractory epileptic seizures), age of the patient, and characteristics of brain imagery and absence of other cause that can justify his illness. The patient was stabilized by carbamazepine and clonazepam. The evolution after 4 months was favorable with a rarefaction of the crises and the decrease of their intensities. Tuberous sclerosis must be evoked before any child with seizures refractory to the usual treatment. Early diagnosis helps to improve management and to consider genetic counseling within the family.

Key words: Sheer Neurological, Tuberous Sclerosis.

INTRODUCTION

Tuberous sclerosis is a heterogeneous disease with a highly variable clinical presentation whose manifestations can differ widely in number and severity over a lifespan (Anne *et al.*, 2015). The pure neurological form of tuberous sclerosis is rare in clinical practice. We report here a recent case of tuberous sclerosis with pure neurological symptomatology in a Senegalese child.

Observation

This was a 2-year-old child with no prior personal history except for a delayed psychomotor development. No family history had been reported. He was received in neuropediatric consultation for partial motor seizures secondarily generalized for 15 months. The neurological and neuropsychological examination showed a mild cognitive deficit and a bilateral pyramidal syndrome made of a discreet motor deficit of the 4/5 proportional at the left half-body and a right brachial predominance with osteotendinous reflexes that are sharp bilateral and a Babinski's test positive on the left.

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Despite complet clinical evaluation, there was no dermatological lesion. The rest of the clinical examination was strictly normal. The cerebral tomodensitometry, performed initially, had revealed periventricular calcifications (Figure 1). The electroencephalogram had shown of bilateral irritative signs with a maximum in the temporal bifront regions. Biology had not shown any peculiarity. The diagnosis of Tuberous sclerosis was retained on the basis of cerebral magnetic resonance imaging (MRI), which showed bilateral cortical and subcortical tubal lesions associated with a left ependymal nodular lesion (Figure 2). As part of a review of the extension of the renal and cardiac ultrasounds were carried out without notable particularity. The balance sheet phosphocalcic and an eye fund had proved normal. The patient was given sodium valproate at the beginning of the seizure treatment unsuccessful, followed by carbamazepine and clonazepam. The evolution was marked by a notable decrease in the frequency of crises.

DISCUSSION

With multisystem involvement beyond the brain and the skin, tuberous sclerosis complex (TSC) is an important neurocutaneous syndrome to diagnose because of its phenotypic variability and the often serious nature of its

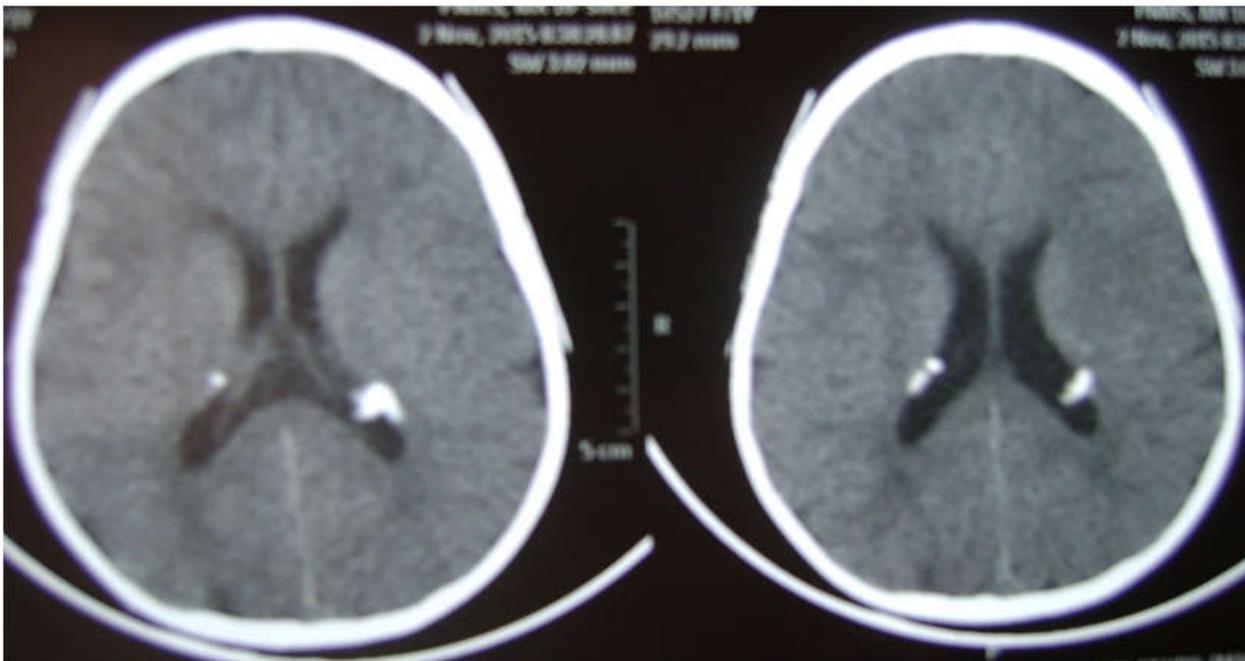


Figure 1: Cerebral CT aspects without contrast medium showing spontaneous hyperdensities compatible with periventricular calcifications

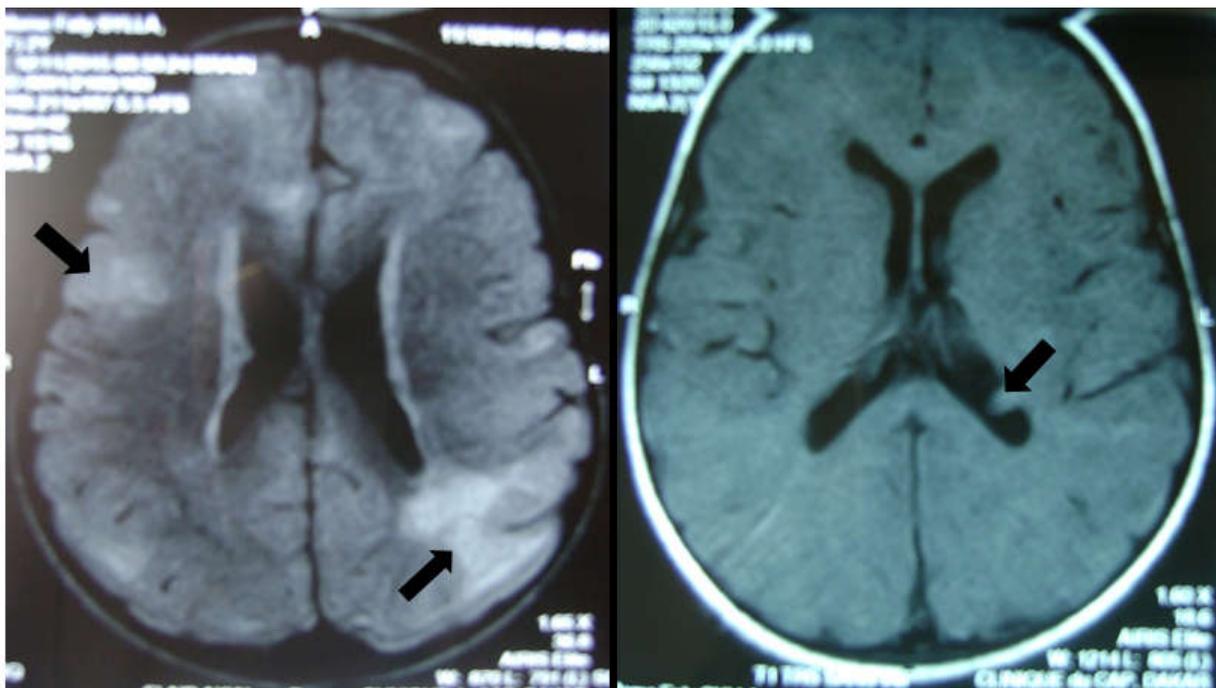


Figure 2. Cortico-subcortical, bilateral, asymmetrical, hyper-intense cortico-cortical anomalies associated with an ependymal nodular lesion in the temporal horn of the left ventricle compatible with a tuberous sclerosis

manifestations. Disordered cellular differentiation and proliferation, primarily in the form of hamartomas, affect the skin, brain, kidney, heart, lungs and eyes (Monica, 2015). Tuberous sclerosis complex (TSC) was initially described approximately 150 years ago by von Recklinghausen in 1862 (Von Recklinghausen, 1862). The frequency of STB is estimated from 1/6000 to 1/10,000 live births and a population prevalence of around 1 in 20,000 (O'Callaghan *et al.*, 1998; Sampson *et al.*, 1989). So TSC is a very rare disease and very few cases described in sub-Saharan Africa.

Our patient is a Senegalese child with isolated neuropsychiatric signs without other associated involvement. Epileptic seizures were the dominant signs. These structural brain abnormalities are likely responsible for the common neurologic manifestations of seizures, mental retardation, and behavioral abnormalities (Roach *et al.*, 1998; Christophe *et al.*, 2000). The term "tuberous sclerosis of the cerebral convolutions" was first used by the French physician Bourneville in 1880 to describe the potato like appearance of cerebral lesions seen during the autopsy of a girl with intellectual disability who died as a result of refractory seizures (Bourneville, 1880).

The central nervous system CNS is affected in more than 90% of individuals with tuberous sclerosis, with the presence of pathological lesions such as cortical or subcortical tubers, subependymal nodules, giant cell astrocytomas, and white matter migration lines (radiologically detectable lines of dysplastic white matter between the periventricular region and the cortical surface) (Northrup, 2013). However, it exists in the disease the clinical triad of adenoma sebaceum, seizures and mental retardation is found in less than half of the patients (Dharmendra Jain *et al.*, 2013). Therefore, before the neurological signs of our patient and the context, we carried out a radiological assessment based on cerebral scan (Figure 1) and then a brain MRI (for more precision when we had suspected this entity because of the periventricular radiological signs Correlated to the patient's clinical setting. Our patient does not present any damage to another organ even cutaneous. The 2012 International Tuberous Sclerosis Complex Consensus Group, comprising 79 specialists from 14 countries, was organized into 12 subcommittees, each led by a clinician with advanced expertise in tuberous sclerosis complex and the relevant medical subspecialty (Hope Northrup, 2013).

It was agreed in this consensus that Clinical features of tuberous sclerosis complex continue to be a principal means of diagnosis. Key changes compared with 1998 criteria are the new inclusion of genetic testing results and reducing diagnostic classes from three (possible, probable, and definite) to two (possible, definite). Additional minor changes to specific criterion were made for additional clarification and simplification (Hope Northrup, 2013). The diagnosis of tuberous sclerosis has been made on the basis of neurological manifestations (refractory epileptic seizures), age of the patient, and characteristics of brain imagery and absence of other cause that can justify his illness. We did not carry out a genetic test but our patient responded well to this diagnosis of tuberous sclerosis according to the last criteria (Hope Northrup, 2013) of the International Tuberous Sclerosis Complex Consensus Group. Regarding treatment, after a period refractory to sodium valproate, our patient was stabilized by carbamazepine and clonazepam. His mother also benefited from useful medical advice for the child's follow-up. The evolution after 4 months was favorable with an important rarefaction of the crises and the decrease of their intensities

Conclusion

Tuberous sclerosis must be evoked before any child with seizures refractory to the usual treatment. Early diagnosis helps to improve management and to consider genetic counseling within the family.

Conflict of interest: No

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