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TUBEROUS ESCLEROSIS COMPLEX AND PSYCHIATRIC COMORBIDITY: TWO CASE REPORTS

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INTRODUCTION

Tuberous Sclerosis Complex (TSC) is a genetic inherited disease characterized by hamartomatous growths in several organs as brain, skin, kidneys, hearth and eyes. The estimated incidence is approximately 1:6000 live births. The diagnosis is made clinically. Seizures are present in 87% of patients. Psychiatric comorbidity has been reported.

OBJECTIVES

We report the clinical course of two patients with previous diagnosis of TSC. Psychiatric symptoms start in the adulthood without seizures history and absence of Subependimal Giant Cells Tumor (SGCT). The evolution and clinical features are described.

METHODS

Patient 1

Married 33-years-old woman with two children affected with TSC. She was diagnosed after headache presentation in 2011. Initial MRI showed periventricular glioneuronal hamartomas. In January 2013 start with self-injurious (swallowing of objects) and autistic behaviours as well as several hospital urgency room visits. In addition, the patient presented with dull mood, emotional indifference and intellectual impairment, with no response to medication.

Patient 2

Married 43-years-old woman with a daughter affected with TSC. Diagnosis was made in 1999 and psychotic symptoms (delusional beliefs and auditory hallucinations) started in 2011 without previous psychiatric history. The MRI in 2013 shown subependymal nodules. Treatment with risperidone was effective.

RESULTS

Psychiatric symptoms are very often associated to the physical findings on TSC, even in adulthood diagnoses.

CONCLUSIONS

Psychiatric comorbidities are well described in literature. About 10-20% adult patients with TSC present clinically significant behavioral problems as self-injuries, frequently associated with SGCT. The European Expert Panel recommended regular assessment of cognitive development and behaviour and symptomatic treatment.

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