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Case Report

Case Report: Bourneville Tuberous Sclerosis

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Abstract

The objectives of our work were as follows: To study the different clinical and paraclinical diagnostic means of BTS. To identify the different therapeutic options for BTS. Specify the evolutionary profile.

Finally, the importance of early screening using currently available methods, and of the resulting genetic counselling, should be stressed.

BTS is a generalised, inherited, autosomal dominant dyssembryoplasia with incomplete penetrance and variable expressivity. It is characterised by multisystem involvement. Cerebral and renal disorders are particularly serious, as they cause severe sequelae and can be life-threatening. The course of this disease seems difficult to predict. It is most often seen as a disabling condition requiring multidisciplinary management.

Study the different clinical and paraclinical diagnostic methods of TBS. Identify the different therapeutic possibilities for STB. Specify the evolving profile. The value of early detection using currently available means, and the resulting genetic counseling.

TBS is a generalized, hereditary dysembryoplasia, with autosomal dominant transmission, incomplete penetrance and variable expressivity. It is characterized by multisystem involvement. Cerebral and renal damage have a particularly serious clinical aspect, because they are responsible for serious sequelae and can be life-threatening. The course of this disease seems difficult to predict. It most often appears as a disabling pathology, requiring multidisciplinary management.

Keywords: RMI; STB; Epilepsy

Introduction

Tuberous sclerosis of Bourneville S.T.B, or systemic tuberous sclerosis S.T.S or also known as EPILOIA Epilepsy - low intelligence - Adenoma sebaceum, is a rare congenital condition which highlights the multiplicity of organs affected. Its prevalence is estimated at an average of 1/10,000 live births 1.

In its typical form, it combines epilepsy, mental retardation and angiofibromas of the face. However, some patients may be virtually asymptomatic, while others are severely disabled. Although the clinical picture of BTS is dominated by neurological and cutaneous involvement, it remains underestimated in our Moroccan context due to a lack of awareness of the early signs of the disease. We report a case of BTS in the intensive care unit of the Hassan II Hospital in Agadir.

In order to gain a better understanding of this pathology, we propose to analyse an observation, with a review of the literature, in order to gain a better understanding of its variable manifestations.

Patient and Observation

Patient Information

Patient aged 17 years with no particular pathological history/ orphan: no family, referred from TATA peripheral hospital for partial convulsive malaise with headaches in a febrile context.

Clinical Findings

Neurological: the patient was intubated, ventilated and sedated.

Glasgow score; difficult to assess

Symmetrical reactive pupils

Haemodynamic: stable with blood pressure 120mmhg/ 69mmhg/ heart rate 89bat/ min/ dextro: 1.16 g/L

Respiratory: Respiratory rate 14c/min

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Sao2 98%.

With ventilatory parameters

FiO2; 80%/ Vc:420Ml/ Fr: 14c/ Min

Infection: fever 38.9.

Timeline of Current Episode

Patient admitted to the intensive care unit at Agadir hospital for further treatment.

She underwent brain imaging, which revealed images suggestive of tuberous sclerosis; parenchymal and subepidymal micronodules plus passive biventricular dilatation.

Diagnostic Assessment

Lumbar puncture performed: normal

Appearance: rock water/ leukocytes: 3/ glycorrhaphy >0.5

Hence the importance of performing a cerebral MRI in order to make the diagnosis as soon as possible, together with a biological work-up. After 3 days on controlled ventilation, patient weaned from

Intubated, Glasgow score 15/15, respiratory and HMD stability.

Therapeutic Intervention

Regression of seizures under anticonvulsant treatment;

1/depakine 500mg°3

2/Kepra 250 mg 1cp°2

3/oedes 20mg 1cp/D

Follow-up and Outcomes

On clinical examination, the patient presented with café au lait spots on the anterior surface of the left haemothorax + pigmented macculopapular lesions.

On neurological examination; carried out in the first few days of hospitalisation

There was general hypotonia / ROT present but depressed especially in the mb / bilateral indifferent CRP / negative Hoffman sign / negative Babinski sign.

The rest of the neurological examination was difficult to assess given the state of consciousness.

Cardiovascular examination: B1, B2 well percussed, no additional noises or particular murmurs, an echocardiac probe was performed which did not reveal anything in particular. No signs of heart failure (LV not dilated / ejection fraction normal at 65%).

ICV of good calibre, no signs of overload.

No valvular disease.

The rest of the general examination was unremarkable.

Discussion

Tuberous sclerosis of Bourneville STB is a neurocutaneous syndrome whose multisystem involvement most often affects the skin, brain, kidneys, lungs and eyes. Which corresponds to our case?

Diagnosis is based on clinical features; skin lesions are present in 90% of patients and seizures are the initial presentation in 80% of patients. Patients with BTS often present with a high incidence of neuropsychiatric symptoms, including mental retardation, autism and learning difficulties. Epilepsy and brain and kidney tumours are aggressively managed with medical and surgical treatments. We report the case of a 17-year-old female patient referred for seizure disorder with seizures.

Clinical examination revealed café au lait spots on the anterior surface of the left haemothorax and pigmented macculopapular lesions. On re-interviewing the mother, these spots dated back to childhood, with the notion of convulsive seizures, which had disappeared by the age of 8, with good psychomotor development, and a notion of consanguinity of the parents, with no other family history, which presented sufficient major criteria to retain the diagnosis of Tuberous Sclerosis of Bournonville [1].

In the face of this clinical picture, the examination was completed by a cerebral MRI which revealed multiple bilateral subependymal and intraventricular nodules as well as signal anomalies in the subcortical white matter at the frontal and temporal levels.

These findings are consistent with those of Jawad El-Azhari [1].

Ophthalmological examination, electrocardiogram and cardiac ultrasound were normal 234 [1-3].

Symptomatic treatment was initially proposed; airway clearance, neurological protection, avoidance of ACSOS.

plus an antiepileptogen [4] with background treatment based on vascular laser for angiofibromas, and annual and paraclinical monitoring every 4 years [1].

Conclusion

BTS is a generalised, inherited, autosomal dominant dysembryoplasia with incomplete penetrance and variable expressivity. It is characterised by multisystem involvement. Cerebral and renal disorders are particularly serious, as they cause severe sequelae and can be life-threatening. The course of this disease seems difficult to predict. It is most often seen as a disabling condition requiring multidisciplinary management.

Author Statements

Competing of Interest

Authors declare no competing of interest

Authors Contribution

All the authors have read and agreed to the final manuscript.

Patient's Perspective

The patient was informed before starting the medical treatment for which he agreed.

Informed Consent

The patient expressed his consent to the publication of this case report with figures.

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