Clinical Reports

Late onset epilepsy in undiagnosed tuberous sclerosis

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Summary: A 67 year old woman first developed left-sided sensorimotor seizures postoperatively in association with infection and mild hyponatraemia. She was found to have previously unrecognized tuberous sclerosis, with gross pathognomic features: periungual fibromata, facial angiofibromata, calcified subependymal nodules, cortical tubers and multiple renal angiomyolipomata. Her son who was epileptic was also found to have other mild features of tuberous sclerosis as a result. As far as we are aware our patient is the oldest for the first presentation of tuberous sclerosis.

Introduction

Tuberous sclerosis is transmitted by autosomal dominant inheritance and commonly affects the central nervous system, retina, skin, kidneys and/or heart. Its prevalence is approximately 1 per 10,000 according to pathological and epidemiological studies,¹ which is second only to neurofibromatosis amongst the neurocutaneous syndromes. The most frequent presenting symptom of tuberous sclerosis is seizures. Seizures usually begin in mentally retarded patients in the first months of life, whereas in patients of average intelligence the peak age of onset is between their second and fifth birthday.² Patients of normal intelligence can present with seizures in the late teens.³

We report the case of an elderly woman who presented with epilepsy secondary to previously undiagnosed tuberous sclerosis.

Case report

A 67 year old woman presented with confusion, pyrexia, lower abdominal pain and diarrhoea. A left iliac fossa abscess was drained and she received intravenous antibiotics. Her confusion settled after 4 days. Seven days postoperatively she had three sensorimotor seizures over 7 hours, involving the left arm and leg. In the past she had a solitary blackout after an explosion during the Second World War. She had been a pottery worker and was not mentally retarded.

On examination, there were prominent periungual fibromata (Figure 1) and facial angiofibromata, but there were no retinal phakomata or neurological signs. Computed tomographic (CT) head scan showed right occipital lobe calcified subependymal nodules. Biochemically she was hyponatraemic at 127 mmol/l (normal 135–145 mmol/l), and also hypoalbuminaemic. Other biochemistry, including glucose and ionized calcium, was normal.

A diagnosis of focal epilepsy due to tuberous sclerosis, triggered by infection and hyponatraemia was made. Her seizures were controlled by a combination of phenytoin and sodium valproate. After 11 days they ceased.

Two weeks later she became confused and pyrexial from a left iliac fossa inflammatory mass. A sigmoid colectomy was performed. Postop-

Figure 1 Right hand showing prominent periungual fibromata.

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eratively she developed cardiopulmonary failure and died. Histopathology of the removed sigmoid colon mass showed an adenocarcinoma. At post-mortem there were the characteristic cerebral changes of tuberous sclerosis localized to the right occipito-parietal area (cortical tubers and subependymal nodules). There were also multiple bilateral renal angiomyolipomata.

Examination of her son, who has epilepsy, confirmed a diagnosis of tuberous sclerosis. He has periungual fibromata and facial angiofibromata, but is not mentally retarded and CT head scan is normal.

Discussion

The main complications of tuberous sclerosis in the elderly are progressive renal and pulmonary disease as well as an increased susceptibility to global cognitive impairments.⁴ Our patient exhibited susceptibility to confusion caused by intra-abdominal infection.

This case is rare for two reasons. Firstly, despite gross pathognomic signs of tuberous sclerosis, this elderly lady had previously escaped diagnosis by clinicians and had led a normal life. As far as we are

aware this is the latest age of presentation of tuberous sclerosis. She had easily visible periungual fibromata and facial angiofibromata. Other pathognomic features of tuberous sclerosis were calcified subependymal nodules, cortical tubers and multiple renal angiomyolipomata.¹ Secondly, the onset of epilepsy was at the age of 67, despite cortical tubers being present. Epilepsy due to tuberous sclerosis usually starts in infancy or childhood, in 80–90% of cases.^{1,2,5,6} Some patients,⁵ a third of those with average intelligence,² do not develop epilepsy. It is exceptional for a patient with such gross features of tuberous sclerosis as in this instance, not to have developed epilepsy earlier.

The epilepsy consequent to her genetic predisposition was made evident by a stressing illness (with infection and hyponatraemia).⁷

This case also underlines the importance of making a clinical diagnosis of tuberous sclerosis; leading on to screening other family members for this autosomal dominant condition. The patient's son was found to have tuberous sclerosis; he has only mild dermatological features. It should be noted that he had previously been diagnosed as idiopathic epilepsy. He has now received genetic counselling for his autosomal dominant disorder.

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