# Atypical presentation of tuberous sclerosis and obsessive compulsive disorder in an adult male

**Arnab Bhattacharya, Shyamanta Das, Kamal Nath, Dipak Dutta, Sahoo Saddichha[1]**

Department of Psychiatry, Silchar Medical College and Hospital, Silchar, Assam, [1]University of British Columbia, Vancouver, Canada, National Institute of Mental Health and Neurosciences, Bangalore, India

**Abstract**

Tuberous Sclerosis (TSC) is clinically marked by a triad of adenoma sebaceum, epilepsy and mental retardation. It can however manifest as various neuropsychiatric disorders. We report a patient who presented with TSC and co-morbid Obsessive Compulsive Disorder.

**Key Words**

Epiloia, neuropsychiatry, tuberous sclerosis

**For correspondence:**

**Dr. Sahoo Saddichha,** National Institute of Mental Health and Neurosciences, Bangalore, India. E-mail: saddichha@gmail.com

*Ann Indian Acad Neurol 2012;15:161-2*

## Introduction

Tuberous sclerosis (TSC), or Bourneville Disease, is a congenital phakomatoses in which lesions occurring due to a limited hyperplasia of ectodermal and mesodermal cells are noted in the skin, nervous system, heart, kidney and various other tissues. It is clinically recognized by a triad comprising of adenoma sebaceum, epilepsy and mental retardation, with an autosomal-dominant inheritance,[1] affecting mainly two genes – hamartin (9q) and tuberin (16p). Subjects affected by TSC may manifest various neuropsychiatric disorders like psychosis,[2] hyperkinesis, aggression and autism in children,[3] and even mood disorders like mania.[4] In addition, several other neurpsychiatric comorbidities have been reported, which include schizophrenia, Capgras' syndrome, psychosis, auditory hallucinations, mania, anorexia nervosa and alcohol dependence.[5] We present the case of a patient with TSC who presented with an interesting array of neurocutaneopsychiatric manifestations.

## Case Report

Mr. R.N., a 28-year-old farmer hailing from a lower socioeconomic background, presented with a history of multiple skin lesions since the past 20 years and an acute onset of episodes of sudden loss of consciousness, tingling and numbness of the index and middle fingers of the left hand lasting for a minute and obsessive cleanliness for the past 8 months. There was no significant past or family history and, temperamentally, he has been a difficult child to manage. Physical examination with dermatological referral revealed multiple adenoma sebaceum on the face, with classical ash leaf hypopigmented macules on the trunk and Shagreen patch on the lower back. Blood investigations showed mild microcytic hypochromic anemia and eosinophilia. Electroencephalogram revealed occasional right temporal sharp slow waves suggesting focal epileptiform discharges and computed tomogram scan of the brain showed mild lateral ventricular asymmetry and multiple bilateral calcified subependymal nodules. IQ testing revealed a score of 70 (borderline range). Mental status examination revealed an obsessive fear of contamination accompanied by washing compulsions (YBOCS score of 32/48). Based on the above features, a diagnosis of TSC and comorbid obsessive-compulsive disorder (OCD) was made and the patient was started on phenytoin 300 mg/day and clobazam 10 mg/day, with which he is maintaining good seizure control and has an improved quality of life. He was started on behavior therapy for his OCD, for which he showed satisfactory response.

| Access this article online | |
|---|---|
| Quick Response Code: | Website: www.annalsofian.org |
| | DOI: 10.4103/0972-2327.95007 |

## Discussion

TSC is a dominantly inherited condition causing epilepsy, mental handicap and skin lesions. Our patient had developed skin lesions in the first decade of life itself but, being a male subject from a village background, this was probably not given

much importance. However, the problem came to light when the patient suffered from seizure attacks (both partial and generalized), which were much more obvious to him and his functioning had been hampered due to this.

This patient had an atypicality, in that the presentation as his onset of seizure was in the third decade of life. Usually, seizures are noted to start in the very first year of life itself.[6] Another aspect was the absence of a significant level of mental retardation in our subject. It has been said that mental subnormality is rare without seizures[7] and, perhaps, our patient escaped retardation due to late onset of seizures, by which time his cognitive capabilities were sufficiently developed so as to meet the requirements of his farming occupation.

His psychiatric comorbidity (OCD) is also an interesting facet of his case as this has been rarely reported. In fact, in one of the largest studies on TSC involving 241 patients,[5] only one was detected to have OCD. In our patient, this may have been an independent presentation or may have been attributable to either seizure disorder or TSC. To make a distinction is impossible; yet, the management of OCD remains similar. We chose behavior therapy as first-line treatment instead of pharmacotherapy due to the fact that many antiobsessional drugs (e.g., selective serotonin reuptake inhibitors and chlomipramine) have a propensity to lower the seizure threshold, which would have been extremely detrimental to our patient.

The above case study gives us an indication that the manifestations of TSC can be multiple and clinical features may appear at various times during the subject's life. Workers have noted variability in the expression of patients even within the same family.[8] Hence, a thorough family history becomes essential. It is therefore important that the clinician maintains a high degree of suspicion for the possibility of a hidden TSC even if few phenotypic features are observed and be open to the possibility of psychiatric comorbidities so that the patient is managed in a holistic manner.

## References

1. Gunther M, Penrose LS. The genetics of epiloia. J Genet 1935;31:413-30.
2. Sedky K, Hughes T, Yusufzie K, Lippmann S. Tuberous sclerosis with psychosis. Psychosom 2003;44:521-2.
3. Hunt A, Dennis J. Psychiatric disorder in children with tuberous sclerosis. Dev Med Child Neurol 1987;29:190-8.
4. Khanna R, Borde M. Mania in a five year old child with tuberous sclerosis. Br J Psychiatry 1989;155:117-9.
5. Muzykewicz DA, Newberry P, Danforth N, Halpern EF, Thiele EA. Psychiatric comorbid conditions in a clinic population of 241 patients with tuberous sclerosis complex. Epilepsy Behav 2007;11:506-13.
6. Osbourne JP. Diagnosis of tuberous sclerosis. Arch Dis Child 1988;63:1423-5.
7. Pampiglione G, Pugh E. Infantile spasms and subsequent appearance of tuberous sclerosis syndrome. Lancet 1975;2:1046.
8. Nevin NC, Pearce WG. Diagnostic and genetic aspects of tuberous sclerosis. J Med Genet 1968;5:273-80.

**How to cite this article:** Bhattacharya A, Das S, Nath K, Dutta D, Saddichha S. Atypical presentation of tuberous sclerosis and obsessive compulsive disorder in an adult male. Ann Indian Acad Neurol 2012;15:161-2.
**Received:** 21-11-11, **Revised:** 05-12-11, **Accepted:** 07-12-11

**Source of Support:** Nil, **Conflict of Interest:** Nil