# Case Report

# A Case of Obsessive–Compulsive Disorder Comorbid with Miyoshi Myopathy

**Arpit Parmar, Rohit Verma[1]**

ABSTRACT

Obsessive–compulsive disorder (OCD) is a common neuropsychiatric disorder, with predominant involvement of cortico-striato-thalamo-cortico circuitry. Although late-onset cases (>35 years) usually show an association with various neurological disorders involving basal ganglia and thalamus, it is not the case with the young-onset patients. There have been no reports of OCD comorbid with dysferlinopathy which is usually considered as a disease involving only muscles. However, recently, studies suggest involvement of brain in this disease. Here, we report a case of dysferlinopathy comorbid with OCD and discuss the related literature.

**Key words:** *Dysferlinopathy, Miyoshi myopathy, obsessive–compulsive disorder*

## INTRODUCTION

Obsessive–compulsive disorder (OCD), especially with late onset (>35 years), has been frequently found to be comorbid with other illnesses, especially neurological illnesses. Many such associations have been reported till date, which include Parkinson's disease, Huntington's disease, Tourette's syndrome, Sydenham's chorea, traumatic brain injuries, and many others. Involvement of basal ganglia and other subcortical structures appears to be specific in the pathophysiology of OCD comorbid with these illnesses. However, there are no reports of OCD comorbid with dysferlinopathy. Although dysferlinopathy is considered a muscle-specific disease, recent reports have reported a brain-specific dysferlin transcript.[1] Reports also suggest brain-specific changes in such patients.

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| **Website:** www.ijpm.info | Quick Response Code |
| **DOI:** 10.4103/IJPSYM.IJPSYM_1_17 | |

We report a case of dysferlinopathy and associated OCD in a male patient.

## CASE REPORT

A 28-year-old male presented to the psychiatry outpatient department with illness of 4 years duration. It started with repeated doubts that his hands were dirty even when he acknowledged having washed them thoroughly. Although infrequent initially, such doubts started coming almost 10–15 times a day leading to discomfort. To relieve this distress, he would wash the hands repeatedly until he would feel "almost clean." He considered such thoughts to be his own and irrational; however, he could not control them by



**How to cite this article:** Parmar A, Verma R. A case of obsessive–Compulsive disorder comorbid with miyoshi myopathy. Indian J Psychol Med 2018;40:86-8.

Departments of Psychiatry and NDDTC, [1]Psychiatry, All India Institute of Medical Sciences, New Delhi, India

**Address for correspondence:** Dr. Arpit Parmar
Department of Psychiatry and NDDTC, All India Institute of Medical Sciences, New Delhi, India. E-mail: dr.arpitparmar@yahoo.in

himself and would yield to the thoughts and wash hands repeatedly. He started spending almost 2–3 h a day in such thoughts and acts over next 1 year. He also started experiencing other doubts including checking the locks repeatedly even when they were locked properly and checking the gas stove repeatedly. He would also follow a specific pattern while bathing and spend almost 1 h or so. On a typical day, he would spend almost 4–6 h a day on such repetitive thoughts and acts. He also suffered from an episode of depression during the same period.

Around the same time (just after the onset of previous symptoms), the patient also reports experiencing weakness involving his leg muscles. He would be unable to stand on tiptoe initially which gradually progressed over a year. He also reported pain and enlargement of calf muscles. He started experiencing difficulty in climbing stairs, standing up from squatting position, and standing for long hours. As walking became increasingly difficult, he had to take support while walking. After about 2 years, he started experiencing difficulty in bearing weight. Symptomatic treatment was done initially by the local physicians, which did not improve his symptoms. Patient denied difficulty in breathing, dyspnea, or any symptoms, suggestive of cardiac involvement. His history and family history were unremarkable. General physical examination revealed tenderness and hypertrophy, mainly involving the calf muscles. Power of both proximal limb muscles was decreased symmetrically bilaterally.

Neurology consultation was sought and a provisional diagnosis of myopathy was kept. Routine investigations including complete blood count and renal and liver function tests were normal. Thyroid function test, Vitamin D, prothrombin time, platelet count, and serum lactate dehydrogenase were also within normal range. Serum creatine phosphokinase (CPK) levels were 1515 U/L. Electromyography (EMG) reported normal motor and sensory nerve conduction study and a normal F-wave. Motor unit action potentials were small and polyphasic with short duration, showing early and full recruitment suggestive of myopathy. Muscle biopsy suggested end-stage muscle disease with loss of dysferlin and normal calpain on immunoblot, suggestive of dysferlinopathy. Genetic testing was not done. Brain magnetic resonance imaging was normal, while single photon emission computed tomography (SPECT) scan revealed higher regional blood flow in the areas of anterior cingulate cortex, basal ganglia, and thalamus.

Based on the clinical history and investigations, a diagnosis of OCD with moderate depressive episode and dysferlinopathy (Miyoshi myopathy [MM]) was kept. Patient's Yale-Brown obsessive–compulsive scale score was 23 while HAM-D score was 17 at baseline. He was started on capsule fluoxetine 20 mg and was stabilized on 60 mg. Although the patient reported initial improvement in his obsessive and depressive symptoms, he lost to follow-up after about 2 months.

## DISCUSSION

This is the first case of OCD comorbid with dysferlinopathy reported in the literature. OCD has been found to be associated with various neurological disorders involving subcortical structures, for example, Huntington's chorea, Sydenham's chorea, Parkinson's disease, and Tourette's syndrome.[2] The involvement of frontal area, basal ganglia, and thalamus appears to be specific in pathophysiology of OCD and the associated neurological diseases. This is also evident in our case as suggested by SPECT scan finding of increased regional cerebral blood flow in the areas of basal ganglia and thalamus.

Dysferlinopathy is considered primarily a muscle disease. It includes a spectrum of muscle disorders characterized by mainly two subtypes: limb girdle muscular dystrophy Type 2B and MM. However, recently, many subtypes have been identified including scapuloperoneal syndrome and congenital muscular dystrophy. Our patient showed a typical presentation of MM, which includes a young male of age 15–30 years experiencing involvement of distal leg muscles initially, with gradual involvement of the proximal leg muscles and forearm muscles as disease progresses.[3] The findings of tenderness and hypertrophy of calf muscles associated with the absence of dysferlin, normal calpain, and preservation of sensory/motor conduction values on EMG and increased CPK also suggest a diagnosis of MM. Similar findings of MM have also been reported in a case series of 28 patients presenting at tertiary care hospital in India.[4]

Although dysferlinopathy is primarily a disease involving the muscles, reports suggest brain involvement in this disease. A brain-specific transcript of dysferlin has also been reported recently. More importantly, the analysis of multiple brain tissue northern blot has also suggested highest expression of this brain isoform in putamen, the area commonly reported to be involved in OCD etiopathogenesis.[5,6] There is also a report of dysferlinopathy associated chorea in a 53-year-old male with lesions in the thalamus and head of the caudate.[7]

Based on these findings, we speculate that OCD in this patient might be related to altered expression of dysferlin in the subcortical areas (mainly, thalamus and basal ganglia). SPECT finding also suggests possible involvement of these regions in the emergence of OCD symptoms in our patient. However, it is not possible to make a conclusion regarding a causal association

between these two conditions, and so, further studies are required to show a possible association between dysferlinopathy and OCD.

**Financial support and sponsorship**
Nil.

**Conflicts of interest**
There are no conflicts of interest.

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