Galore International Journal of Health Sciences and Research
Vol.4; Issue: 3; July-Sept. 2019
Website: www.gijhsr.com
Case Report *P-ISSN: 2456-9321*

Recurrent Depressive Disorder in a Patient with Limb Girdle Muscular Dystrophy

Dr. Joylin Jovita Mascarenhas[1], Dr. Evelyn George[2]

Assistant Professor[1], Post Graduate Resident[2],
Dept of Psychiatry, Father Muller Medical College, Mangalore

Corresponding author: Dr. Joylin Jovita Mascarenhas

ABSTRACT

Limb Girdle Muscular Dystrophy (LGMD) is a genetically determined degenerative disease affecting voluntary musculature, affecting both males & females equally, beginning in the second or third decades. It is the most common muscular dystrophy in India and is associated with significant disability and poor quality of life. Patients with LGMD receive at least one psychiatric diagnosis in their life time, with depression and phobias being the most frequent. However, there is a dearth of information in the available literature on the same.

Keywords: Limb Girdle Muscular Dystrophy (LGMD), Recurrent depressive disorder

INTRODUCTION

The historical description of Limb Girdle Muscular Dystrophy (LGMD) dates back to the late eighteenth century when Erb and Leyden-Mobius described patients with weakness primarily involving the shoulder and the hip girdles respectively, with sparing of the facial muscles. They were more benign than the pseudo-hypertrophic form described by Duchenne[1] In 1954, Walton and Nattrass defined LGMD as a distinct nosological entity. Gardner Medwin retained the term limb girdle muscular dystrophy to designate a major category of dystrophy comprising four subtypes: (i)Autosomal recessive or sporadic (Erb, Leyden-Mobius) (ii) Myopathy limited to quadriceps (iii) Autosomal recessive muscular dystrophy in childhood and (iv) Late onset Autosomal dominant type.[1] The prevalence of LGMD ranges from 1 per 14500 to 1 per 123000 populations. Though one-fourth of all patients presenting in neuromuscular clinics in India have LGMD, its prevalence in the Indian setting is not known.[1] Autosomal recessive forms of LGMD outnumber the dominant forms and has been attributed to the consanguineous union, which is customary in some parts of the country.[1] LGMD is the most common adult onset muscular dystrophy encountered in India. Particular attention has been devoted to its phenotype (sarcoglycanopathy >dysferlinopathy) with interesting clinical signs being described.[1-3]

Recent studies have shown that psychiatric disorders such as autism, dysthymia, depression are common in these patients.[4] A specific profile characterized by low self-esteem with feelings of sadness and internalized culpability has also been noticed in these patients.[5] However, from a psychiatric point of view, particular attention has been given to cognitive functions and the role of dystrophy associated genes on the brain.[6] Data regarding depression and its effects on the disorder are scanty. Hence, we present a case with LGMD with Recurrent Depressive Disorder.

CASE ILLUSTRATION

A 36 year old male, single, educated upto Master's degree, living alone, unemployed and supported financially by family, presented to the Psychiatry Out-patient department on 15th June, 2016, with complaints of sleep disturbance for over two years. Patient was referred by a catholic priest, as patient's sleep disturbance persisted even after religious treatment in

the form of individual counselling and prayers. On General physical examination, patient had proximal muscle weakness in both lower and upper limbs (LL>UL), especially adduction of thighs & flexion of knees. He required help to get up from sitting position. He would bring his knees together and supporting the outer aspects of the thighs with hands, while getting up from squatting position. Hip -abduction sign was positive. Deep tendon reflexes-hyporeflexia & hypertrophy of Bilateral calf muscles were noticed. On reviewing his past records & history taking, we found that the patient first noticed difficulty in getting up from the bed at the age of 14 years, which was progressive in nature. However, they consulted multiple doctors, with no relief of symptoms. Finally, patient consulted in the neuromuscular clinic at NIMHANS in 1999(at the age of 19yrs). Serum CPK was high. Left biceps biopsy was taken and histopathology report of the same revealed local and mild muscular dystrophy. In conjunction with clinical features, patient was diagnosed with limb girdle muscular dystrophy. He was advised physiotherapy. On 21st of June, 2010 stem cell therapy was initiated at a speciality hospital in Bengaluru, using stem cells (derived from umbilical cord) to decrease the progression of the disease. Patient underwent five such sessions, but without any benefits. Hence, he stopped the treatment & continued physiotherapy.

Patient had no past or family history of psychiatric illness (including substance use/medication) or neurological /medical disorders. He was born out of a non-consanguineous union. His family support was good. Premorbidly, he was well adjusted. Despite the illness & treatment for the same, patient continued his studies and completed his post graduation in commerce. He worked as an assistant for a Chartered accountant & was able to support himself, until 2years ago.

On further evaluation, we found that patient had sleep disturbance and low mood for the past 2years, following break-up with his partner, in view of his neurological disorder and resultant disability. Patient was initially noticed to have difficulty in sleep initiation and stress regarding his lost relationship. The sleep disturbance at night and drowsiness during day time affected his work and he had frequent absenteeism. The religious treatment helped him to some extent in dealing with desolation. However, his sleep disturbance was persistent and in the past two months, it worsened with difficulty in termination of sleep. He would wake up three to four hours before his usual time. He also reported of feeling sad most times of the day, which was worse in the morning. He was unable to concentrate, felt tired easily & had disconnected himself from his friends and family members. Though, he acknowledged that his family was his pillar of strength, he felt that his disability and dependency on others was a burden to them & he saw no scope of improvement. He contemplated suicide but did not act on it due to his religious beliefs as a Catholic. On mental status examination, he had decreased psychomotor activity, affect was depressed and had ideas of hopelessness ,helplessness and worthlessness .The content of his talk was regarding his illness, lack of definitive treatment for the same and fear of being dependent(anxious preoccupation).There were no cognitive deficits upon examination. Investigations including hemogram, liver and renal function tests, thyroid, sugar and lipid profile, ECG and Neuroimaging were within normal limits. A diagnosis of Organic Depressive disorder (D/D: Double Depression; Current episode-Moderate Depressive Disorder without somatic syndrome & Longitudinal-Dysthymia) was made. Clinical rating scales were administered and the scores were suggestive of moderate depression (HAM-D=17, BDI=24). He was started on mirtazapine and a low dose of clonazepam. Cognitive behaviour therapy and stress management was also initiated subsequently.

The dosages of medications were titrated accordingly and patient gradually improved .Patient reached premorbid levels of functioning and started working from home. After remission of symptoms for a year, medications were tapered and stopped (May 2018).

However, patient presented to the OPD again in March 2019, with recurrence of symptoms. However, this time, the symptoms fulfilled the ICD-10 criteria of mild depressive episode without somatic syndrome .Patient was re-evaluated to rule out organic causes. Neurological examination showed no progression of the disorder (LGMD) since the first presentation. In view of these two episodes, the diagnosis was revised to recurrent depressive disorder. The same treatment was instituted & patient improved. Family members were advised regarding the maintenance treatment/prophylactic treatment.

DISCUSSION

This is the first case report on Recurrent Depressive disorder in a patient with Limb Girdle Muscular Dystrophy. Previous studies show that patients affected by LGMD receive at least one life time psychiatric disorder, with depression & phobias being the most diagnosed conditions.[6,7] Though the point prevalence of depression in normative collectives ranges between 2-5%, it can be much higher in patients with muscular dystrophy attending neuromuscular clinic.[8] The higher frequency in these patients could be because of usage of rating scales like Beck's Depression Inventory to diagnose depression. There are a lot of overlapping symptoms, especially on the somatic dimension of depression such as fatigability, sleep disturbance, decreased facial expression, decreased initiation & muscle weakness which can lead to the misconception of a major depressive disorder.[8]

In our case, the patient had several risk factors which predisposed him to develop depression, which included: being single, living alone, interpersonal relationship stressor, chronic illness with associated disability, progressive nature of the illness with no definitive treatment and failure to benefit from several sessions of high cost treatment procedures. The cause of depression in patients with LGMD is unclear. Depression can occur as a psychological adaptation to the disease or as a direct manifestation of genetic and /or CNS abnormalities.[8] In most of the cases, the various biological, social and psychological factors contribute to the illness.

Depression has been characterized by activation of the sympathetic nervous system and withdrawal of parasympathetic tone to the heart, increased resting HR and reduced Heart Rate Variability (HRV).[4] The physical & social hardships of the LGMD causes immense stress which activates the sympatho-adrenal and hypothalamic-pituitary axis. The Renin - Angiotensin System (RAS) is activated in muscular dystrophy which is not only implicated in the pathogenesis of the disorder but also responsible for the autonomic dysregulation (A hallmark of depression & other stress related disorder).The synergistic action of autonomic dysregulation and psychological stress leads to exacerbation of disease progression in muscular dystrophy.[4] Angiotensin II (Ang II) is also a well known stress hormone and ARBs(Angiotensin receptor Blockers) and ACEi (Angiotensin converting enzyme inhibitor) are found to be effective in treating depression, mood disorders and neurodegenerative and traumatic disorders of the brain.[9] Rodent studies have found that centrally acting AT2R (Ang II type 2 receptor) agonists may serve as potential anxiolytic agent.[4] Serotonin, a neurotransmitter also modulates stress responses by interacting with the HPA axis and sympathetic nervous system. Selective serotonin reuptake inhibitors (SSRI) and Serotonin norepinephrine reuptake inhibitors (SNRI)

reduce depressive symptoms, increase HRV, reduce Inflammatory markers, normalize urinary cortisol excretion, and reduce plasma catecholamine levels.[4] In our case, the noradrenergic & specific serotonergic antidepressant (NaSSA), Mirtazapine was started and marked improvement was seen. However, the underlying LGMD & other risk factors continued to be a precipitating as well as perpetuating factors in our patient. Hence, stress management and maintenance pharmacotherapy is imperative to prevent relapses, progression of illness and improve the quality of life.

CONCLUSION

There is an increased risk of developing depressive disorder with chronic medical conditions, neurological and neuromuscular disorder. Identification of depression in the face of existing neurological disorder & overlapping symptoms is crucial in improving the quality of life, progression of illness & recurrence of depression. Failure to treat depression increases the tendency to assume a chronic course, be recurrent and over time associated with increasing disability.

Declaration of Patient Consent:

The authors certify that they have obtained all required patient consent forms. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

REFERENCES

1. Khadilkar SV, Singh RK. Limb girdle muscular dystrophies in India. Neurol India 2008; 56:281-8.
2. Khadilkar SV, Singh RK. Hip abduction sign: A new clinical sign in sarcoglycanopathy. J Clin Neuromusc Dis Sep 2001; 3:13-5.
3. Pradhan S. Diamond on quadriceps: A frequent sign in dysferlinopathy. Neurology 2008; 70:322.
4. Sabharwal R, The link between stress disorders & autonomic dysfuction in muscular dystrophy, Frontiers in Physiology, 2014; 5(25):1-6.
5. Miladi N1, Bourguignon JP, Hentati F, Cognitive and psychological profile of a Tunisian population of limb girdle muscular dystrophy, NeuromusculDisord. 1999; 9(5):352-4.
6. Lovestone S, Other Disorders of the Nervous System, Chapter 14,Lishman's Organic Psychiatry: A textbook of Psychiatry,4th edition, edited by David AS, Fleminger S, Kopelman MD et al, 2009; Blackwell publishing, Chichester:845-905.
7. Melo M et al, Becker and limb-girdle muscular dystrophies: A psychiatric and intellectual level comparative study, American journal of Medical Genetics, 1995, 60(1):33-38.
8. Winblad et al., Depression in Myotonic Dystrophy type 1: clinical and neuronal correlates Behavioral and Brain Functions 2010; 6(25):1-7.
9. Dubo Yang, G., Xi, Z.X, Wan, Y., Wang, H., and Bi, G, Changes in circulating and tissue angiotensin II during acute and chronic stress, Biol. Signals,1993; 2:166–172.

How to cite this article: Mascarenhas JJ, George E. Recurrent depressive disorder in a patient with limb girdle muscular dystrophy. Galore International Journal of Health Sciences & Research. 2019; 4(3): 103-106.

******