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Psychiatric Presentation in a Patient with Myotonic Dystrophy: A Case Report

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ABSTRACT

Myotonic Dystrophy type 1 (DM1) is a progressive life-threatening disorder that affects several systems in the human body. Besides physical involvements, previous studies reported various psychiatric and cognitive presentations in these patients. We presented a 65-year-old patient with adult-onset DM who suffered from multi-system involvement. She has also experienced a series of psychiatric symptoms including depressed mood, insomnia, fatigue, reference delusion, visual and auditory hallucinations besides impaired cognitive functions. With the diagnosis of major depressive disorder with psychotic features, she was treated with Sertraline and Haloperidol. The cognitive impairment was continued after improvement in mood, and donepezil 5 mg was prescribed. Whereas patients with DM1 and with psychiatric manifestations have significantly lower function than those without psychiatric symptoms, clinicians should be aware of the mental status examination and eventual psychiatric disorders in these patients. Our case presentation suggests a multidisciplinary approach to these patients to provide comprehensive health care.

Introduction

Myotonic dystrophy type 1 (DM1) is the most frequent adult-onset muscular dystrophy, with the autosomal-dominant transmission. It is a life-threatening chronic progressive multisystem disorder, which is clinically heterogeneous. There are limited pharmaceutical treatments for this disorder, and medication development for this condition is yet to be developed [1]. Besides muscular weakness and atrophy, the gastrointestinal, endocrine, cardiac and ocular systems are affected. The severity of DM symptoms ranges from infancy-onset lethal effects to late-onset mild symptoms [2].

Although individuals with DM may develop co-

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existing psychiatric and cognitive impairments, clinicians generally pay less attention to the psychiatric and cognitive evaluation of these patients. Brain hypoperfusion and hypoxia may deteriorate the cognitive and behavioral symptoms in these patients [3, 4]. A wide range of psychiatric symptoms have been reported; some cases of schizophrenia [5], thought disorders (delusional disorders, psychotic disorders not otherwise specified in juvenile DM1) [6], lack of initiative, inactivity and apathy [3], interpersonal difficulties, dysphoria and lack of interest [7].

Several reasons lead to these variations in neuropsychiatric and cognitive findings including the scarcity of studies, different study designs and the variety of disease severity. Bertrand and colleagues in their study on psychological features in a large group of patients with DM1 suggest that disease severity plays a crucial role in psychological effects in these patients [8]. The authors suggest that when evaluating patients with DM1, regardless of their disease severity, there is no difference between these patients and the healthy control group. However, more psychological symptoms are seen in patients with more severe phenotypes.

It is crucial that the clinicians attentively evaluate neuropsychological, psychiatric and cognitive function in these patients. The significance of this work is twofold: first, in the clinical setting, these neuropsychiatric and cognitive disabilities, notably, lack of interest and initiation may cause patients to be less active in their health-related programs, which in turn, influence both their physical and mental health, second, according to Sharma [3], family members, caregivers and people in close contact with patients with DM should be aware of this situation, because physical inactivity may be related to central nervous system (CNS) dysfunction rather than muscle weakness.

This study aims to highlight the psychiatric manifestations and cognitive dysfunction in a patient with myotonic dystrophy.

Case presentation

A 65-year-old woman suffering from myotonic dystrophy type 1 with a series of psychotic presentations was visited by a neurologist in the neurologic out-patient clinic at Roozbeh Hospital, a referral psychiatric hospital in Tehran, Iran, in February 2021. During the routine and periodic neurological follow-ups, the neurologist realized that the patient has recently developed some psychiatric symptoms. The patient was then referred to the psychiatrist for evaluation by taking history and examining the mental status closely.

She was a widow, with a primary school education level and mother to 4 children. She lived with two of her daughters, who also suffer from DM1. We observed depressed mood, reference delusion, auditory and visual hallucination in the mental status examination (MSE). The symptoms had started nearly two months earlier. Her family reported a major depressive disorder in the patient about 14 years earlier, following the loss of her daughter, who had been medically treated. There was no psychiatric hospitalization in the history.

Based on the patient's medical records, she suffered from multisystem involvement due to DM1. The first neurological symptoms and signs included marked muscle weakness and myotonia in the distal parts of both upper and lower limbs bilaterally, which became notable in adulthood. Muscle weakness was progressed over the last years; and the proximal of the limbs, neck and face muscles were involved too. She is currently wheelchair-bound and dependent on all of her daily living activities.

Due to a bilateral posterior subcapsular cataract, she had undergone bilateral phacoemulsification. Moreover, cardiac arrhythmia was present in her twenties. She had been CCU admitted and was diagnosed with Atrial Fibrillation (AF) and left bundle branch block (LBBB) in her 30th. In addition, she was affected by diabetes mellitus. Her latest medication list included Metformin, Metoprolol, Empagliflozin and Rivaroxaban.Totally, these clinical features suggested classic myotonic dystrophy. The diagnosis was established using molecular genetic testing which identified a heterozygous pathogenic variant in DMPK (DM1 Protein Kinase) gene. Furthermore, the electromyographic study showed myotonic discharges and myopathic-appearing motor units, predominantly in distal muscles. The patient had received no corticosteroid therapy. She did not have any experience of substance use.

Regarding the cognitive assessment, the patient's cognitive performance was significantly impaired. The profile of her cognitive assessment is presented in Table 1.

At the first psychiatric evaluation, based on the findings in the interview with the patient and her family, the diagnosis of major depressive disorder with psychotic features was made based on the Diagnostic and Statistical Manual of mental disorders (DSM-5) diagnostic system [9]. Therefore, treatment

Table 1. Cognitive evaluation scores

Cognitive test	Score/total
Persian Picture Naming Test	18/50
Rey Auditory Verbal Learning Test	
Immediate recall	5/15
Learning (trials 1-5)	23/75
Delayed recall	7/15
MOCA*	
Visuospatial	2/5
Naming	0/3
Memory and Delayed recall	0/5
Attention	0/6
Language	0/3
Abstraction	0/2
Orientation	4/6

*MOCA: Montreal Cognitive Assessment test

with Sertraline and Haloperidol was gradually titrated. At the second psychiatric visit, a relatively notable improvement in her mood and psychotic symptoms was reported by the patient and her family and observed by the psychiatrist. Her depressive and psychotic symptoms have been relatively controlled using 50 mg of Sertraline and 2.5 mg Haloperidol per day.

Based on the patient's cognitive impairment, we performed a volumetric brain MRI. We have observed global cortical atrophy mainly noted in bilateral posterior parietal regions (due to precuneus lobule atrophy and posterior cingulate dilatation). The medial temporal atrophy score on both sides was 3 [10]. The right hippocampus volume was 2.8cc, and the left hippocampus volume was 2.5 cc. In addition, there were white matter changes of Fazekas scores 2 on both sides (i.e., early confluent high signal foci) [11]. In conclusion, these findings were in favor of focal atrophy of bilateral temporal and posterior parietal lobes. In the follow-up, the cognitive impairment was continued after improvement in mood and donepezil 5 mg was prescribed.

Discussion

DM is the most frequent adult-onset muscular dystrophy; which according to recent studies affects 1 in every 2100 births [12]. It can cause early-onset bilateral cataracts, diabetes mellitus, skeletal muscle weakness, and myotonia (delayed muscle relaxation). In addition, previous studies reported a various range of psychiatric and cognitive symptoms in these patients. Because the psychiatric features of DM are not very frequently studied, there is variable information on the prevalence of these symptoms; for instance, Kalkman and colleagues observed that half of the patients with DM have a lifetime history of major depressive disorder [13]. Although another study showed no specific pattern of psychological features in DM1, the authors stated that 27% of patients with DM1 were at higher risk of developing a psychiatric disorder [8]. Regarding cognitive functions, Colombo [14] reported that up to 76% of patients with severe DM presented cognitive impairment.

In this study, we presented a patient with DM1 who developed cognitive and psychiatric symptoms. We observed depressed mood, insomnia, fatigue and psychotic symptoms which include reference delusion, visual and auditory hallucinations besides impaired cognitive functions, especially working memory and executive functions. Due to persistent cognitive impairment after improvement in mood, history of a chronic progressive cognitive decline, impaired activity of daily living along with brain MRI evidence, degenerative dementia should be considered.

Our case presentation recommends that clinicians should be aware of findings related to cognitive and psychiatric manifestations in DM1. Moreover, it has been suggested by Jacobs et al. [6], although their study population included mainly juvenile DM1. These impairments besides physical symptoms should be considered in other types of DM1 as well. Patients with psychiatric manifestations have significantly lower functions than those without psychiatric symptoms. This impaired function is more pronounced in patients with disorders of thought [6]. Therefore, evaluation of mental status examination should be considered as a part of comprehensive care. This will, in turn, improve the patient's quality of life and family support as there is a link between the disease severity and duration with cognitive performance.

Conclusion

MD as a multisystem disease affects various organs. A multidisciplinary approach is recommended to provide comprehensive health care.

Ethical Considerations

Both the patient and her main caregiver her daughter-provided informed consent to take part in this study. Identifying data were removed from the data and are not mentioned in the manuscript in order to respect the patient's right for privacy.

Compliance with ethical guidelines

There were no ethical considerations to be considered in this article.

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Conflict of Interests

The authors have no conflict of interest to declare.

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