Journal of Comprehensive Clinical Practice
An official Journal of **Vivekananda Polyclinic and Institute of Medical Sciences,** Lucknow, UP, India

CASE REPORT

Myoclonus Dystonia Syndrome among the Pediatric Population: A Case Report

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Abstract

Myoclonus-dystonia syndrome (MDS) is a rare movement disorder of the central nervous system. It is primarily characterized by myoclonus and may be associated with dystonia. MDS usually occurs in the early to 20s stage of life. Although the disease has a mild course, but can lead to functional disability in some minority of patients. In this paper, we present a probable case diagnosis of MDS based on the clinical characteristics. The patient presented significant improvement with topiramate and anti-dystonic treatment.

ARTICLE INFO

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Dates:
Published: 30-12-2023

Keywords:
Myoclonus dystonia syndrome, myoclonus, dystonia, movement disorder.

Conflict of Interest:
The authors have no conflict of interest

How to Cite:
Bansal K. Myoclonus Dystonia Syndrome among the Pediatric Population: A Case Report. Journal of Comprehensive Clinical Practice. 2023;17(2):53-55

Introduction

Myoclonus-dystonia syndrome (MDS) is a movement disorder manifesting symptoms of myoclonus, which may be associated with dystonia. It usually starts in the first two decades of life. MDS is an autosomal dominant hereditary disease and sometimes it may be sporadically encountered.[1] Myoclonic contractions commonly involve the upper extremities, especially the proximal parts. Alcohol intake suppresses myoclonic jerks.[1] These myoclonic jerks can be accompanied by dystonic episodes of mild-moderate severity.[1]

The case patient was diagnosed with a possible diagnosis of MDS based on clinical features, and the patient responded to topiramate therapy. To support the diagnosis genetic testing; next-generation sequencing (NGS) was done which showed a mutation in SGCE gene related to MDS.

Case Report

An 11-year-old boy presented with complaints of involuntary jerks of his right arm. The complaints started at 9 years of age and progressed over the period to the opposite side i.e., left arm interfering with his normal daily routine activities like writing. The patient was referred to us by a treating pediatrician. Birth history, development history, and family history were unremarkable. Neurological examination showed brief, focal dysrhythmic, sudden jerks of the left upper limb and intermittent twisting movement of distal parts of the upper extremity. Biochemical investigations couldn't detect any significant abnormality. MRI brain and EEG were normal. With these findings, a diagnosis of probable MDS was made.

The patient was started on levetiracetam followed by valproate and clonazepam. But no improvement was observed. Hence, the patient was started on topiramate along with trihexyphenidyl for dystonia. On follow up a marked decrease in his myoclonic and dystonic episodes was recorded.

To support our diagnosis, NGS was done which revealed a novel mutation in SGCE gene; c82G>A; Exon1; Heterozygous; Dystonia 11, myoclonic; DYT11:159900; autosomal dominant; VUS. This gene variant with heterozygous autosomal dominant inheritance is reported as a variant of uncertain significance given the limited literature availability.

Discussion

MDS is a rare movement disorder, which is characterized by predominantly myoclonus (rapid, and brief muscle contractions). This can be accompanied by sustained or repetitive twisting movements resulting in abnormal postures (dystonia). The onset of disease is most often seen initial decade of life or adolescence.[2]

The myoclonic jerks commonly affect the upper limbs, neck, and trunk. Dystonia (focal or segmental) can be seen in approximately 50% of affected individuals. In a few patients, dystonia can be seen as cervical dystonia and writer's cramp.[2]

The disease has a benign course and can remit spontaneously. But in some cases, the disease may progress to adulthood. It can spread to other unaffected body parts or worsen in terms of severity.[3]

Initially, another study[1] proposed five diagnostic criteria for MDS in 2009, based on available clinical data and a comprehensive literature review. Later, Roz *et al.*[3] proposed the **"Modified diagnostic criteria for myoclonus-dystonia** which says that the diagnosis of MDS requires four major criteria OR three major criteria, two minor criteria, and no exclusionary criteria. Further, Grunwald *et al.*[4] proposed classification criteria for MDS. As per this classification, our case is a probable case of MDS.

Genetics of MDS

SGCE is the main causative gene for MDS.[3] The disorder is inherited as an autosomal dominant disease. The penetrance of the gene is based on the paternal origin of the changed SGCE allele.

In the majority of cases, the disorder is inherited from a heterozygous parent, who might or might not have symptoms of myoclonic dystonia.

More than 90% of children who inherit an SCGE variant from some family member, will develop symptoms, while only about 5% of children who inherit an SCGE variant from their mother will develop symptoms.

Prenatal testing and pre-implantation genetic screening may be more feasible when the SGCE pathogenic variant has been found in the patient's family member.[2] SGCE-M-D is also compatible with an active life of normal duration.[2]

Anti-seizure drugs like valproate, levetiracetam, zonisamide, and benzodiazepines like clonazepam, and topiramate improve myoclonus in patients with myoclonus-dystonia. Dystonia may be relieved by the use of anticholinergics. Botulinum Toxin injection may be helpful in the case of Cervical Dystonia.

Deep brain stimulation targeting globus pallidus interna (Gpi) and ventral intermediate nucleus of the thalamus (VIM) are effective prolonged treatments, although GPi is generally selected for this purpose. Long-term GPi-DBS improves motor features in myoclonus-dystonia and upgrades social adjustment.[5]

Conclusion

MDS is rare movement disorder with the potential to cause serious disabilities. Symptoms may appear as early as infancy or preschool age, with myoclonic jerks or dystonia being the initial symptoms. SGCE gene variants can cause myoclonus dystonia syndrome. The SGCE gene is imprinted maternally by the mother and imprinted by father as well. Inherited variants of SGCE are originated by paternal gene. Symptomatic improvement and improved quality of life of patients can be achieved by antiepileptics and anti-dystonic treatment.

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