CASE REPORT | OLGU SUNUMU

OPEN ACCESS

Tetrabenazine-responsive SGCE myoclonus-dystonia with delayed diagnosis: A case report

Gecikmiş tanılı tetrabenazin-yanıtlı SGCE miyoklonus-distonisi: Olgu sunumu

Cansu Kostakoğlu Duman[1], Özlem Polen Erdoğan[2], Yeşim Sücüllü Karadağ[2]

[1]Department of Neurology, Gaziantep Abdülkadir Yüksel State Hospital, Gaziantep, Türkiye
[2]Department of Neurology, University of Health Sciences, Ankara Bilkent City Hospital, Ankara, Türkiye

ABSTRACT

Myoclonus-dystonia syndrome (MDS) is a rare genetic movement disorder characterized by the combination of myoclonus and dystonia. It typically manifests during the first two decades of life, with mutations in the epsilon-sarcoglycan (SGCE) gene being the most commonly implicated cause. We present a case of SGCE-related MDS in a 19-year-old female who exhibited prominent upper-body myoclonus and mild cervical dystonia. This case supports the potential effectiveness of tetrabenazine in SGCE-related MDS and contributes to the limited but growing body of literature on its use in hyperkinetic movement disorders.

Keywords: Cervical dystonia, epsilon-sarcoglycan, myoclonus-dystonia syndrome, SGCE gene, tetrabenazine.

ÖZ

Miyoklonus-distoni sendromu (MDS), miyoklonus ve distoninin birlikteliği ile karakterize, nadir görülen genetik bir hareket bozukluğudur. Hastalık tipik olarak yaşamın ilk yirmi yılında ortaya çıkar ve epsilon-sarkoglikan (SGCE) genindeki mutasyonlar en sık saptanan nedendir. Bu yazıda, belirgin üst gövde miyoklonusu ve hafif servikal distonisi olan, SGCE ilişkili MDS tanılı 19 yaşında bir kadın olgu sunuldu. Bu olgu, tetrabenazinin SGCE ilişkili MDS'de potansiyel etkinliğini desteklemekte ve hiperkinetik hareket bozukluklarındaki kullanımına dair sınırlı ancak giderek artan literatüre katkı sağlamaktadır.

Anahtar sözcükler: Servikal distoni, epsilon-sarkoglikan, miyoklonus-distoni sendromu, SGCE geni, tetrabenazin.

Myoclonus-dystonia syndrome (MDS) is a rare, genetically determined movement disorder characterized by varying combinations of myoclonus and dystonia, typically emerging in the first two decades of life.[1,2] Mutations in the epsilon-sarcoglycan gene (SGCE, DYT11) on chromosome 7q21-q31 are identified in 50-70% of cases.[1-3] The condition is estimated to affect two per 1,000,000 individuals in Europe. Clinically, MDS manifests as lightning-like myoclonic jerks, predominantly involving the neck and upper extremities, and is often accompanied by focal dystonia, most commonly cervical or writer's cramp.[2-4] Psychiatric comorbidities such as anxiety, phobias, and obsessive-compulsive traits are frequently reported, further reducing quality of life.[3] Treatment options include benzodiazepines, valproate, levetiracetam, gabapentin, and anticholinergics, although these yield limited benefits.[5] Newer approaches such as tetrabenazine, zonisamide, and perampanel have shown promise in individual cases.[5-7] Botulinum toxin is effective for focal dystonia, and deep-brain stimulation (DBS) can provide significant benefit in severe cases.[2,5] Here, we present a patient with SGCE-related MDS who experienced substantial improvement with tetrabenazine after a decade-long diagnostic delay.

CASE REPORT

A 19-year-old woman presented with involuntary jerks of the head, neck, and arms beginning at age nine. Over time, the jerks intensified, spreading to the legs and worsening with stress, excitement, or fear, causing marked social withdrawal and academic

Corresponding author: Cansu Kostakoğlu Duman, MD. Gaziantep Abdülkadir Yüksel Devlet Hastanesi, Nöroloji Kliniği, 27100 Şahinbey, Gaziantep, Türkiye.
E-mail: cansu.ash@gmail.com | Doi: 10.5606/phhb.dergisi.2026.53

Received: October 31, 2025 | **Accepted:** April 10, 2026 | **Published online:** May 13, 2026

Cite this article as: Kostakoğlu Duman C, Polen Erdoğan Ö, Sücüllü Karadağ Y. Tetrabenazine-responsive SGCE myoclonus-dystonia with delayed diagnosis: A case report. Parkinson Hast Harek Boz Derg 2026;29(1):26-28. doi: 10.5606/phhb.dergisi.2026.53.

decline. Magnetic resonance imaging (MRI) of the brain was normal, and prior propranolol therapy was ineffective.

Her medical and birth history were unremarkable, but there was parental consanguinity. One cousin reported similar jerks, while two paternal relatives had balance and speech problems without a definitive diagnosis. Neurological examination revealed prominent myoclonic jerks in the head, neck, and proximal upper extremities, extending to the legs during walking. Mild right-sided cervical dystonia with shoulder elevation was observed. Laboratory and imaging evaluations, including copper studies and brain-spine MRI, were normal except for thoracic rotoscoliosis. Clonazepam (2 mg/day) provided partial relief. Ophthalmologic and abdominal imaging were normal, excluding paraneoplastic or systemic causes. Electroencephalography, nerve conduction studies, and somatosensory evoked potentials were normal. Electromyography revealed cortical or subcortical myoclonus. Whole exome sequencing identified a heterozygous SGCE c.691A>T (p.Arg372Ter) mutation. Following the establishment of the diagnosis of MDS, tetrabenazine was initiated as targeted therapy and gradually titrated to a dose of 75 mg/day, resulting in marked clinical improvement. The treatment was well tolerated, with no clinically significant adverse effects observed. Mood and QTc interval were routinely monitored during dose titration, and no abnormalities were detected. Clonazepam was subsequently discontinued, and botulinum toxin injections were administered to address cervical dystonia. Escitalopram (10 mg/day) was introduced for the management of anxiety and depressive symptoms. After six months, although formal rating scales were not applied, both the patient and her family reported a marked reduction in the frequency and amplitude of myoclonic jerks, accompanied by significant improvement in daily functioning. Anxiety and depressive symptoms improved in parallel with motor improvement following tetrabenazine initiation, alongside escitalopram treatment. Written informed consent was obtained from the patient.

DISCUSSION

Myoclonus-dystonia syndrome is an autosomal dominant disorder with reduced penetrance due to maternal imprinting, typically inherited from the paternal allele but occasionally sporadic.[1,2,8]

The SGCE gene mutation was confirmed in our patient, consistent with familial aggregation. Myoclonus-dystonia syndrome manifests as brief, shock-like myoclonus and variable dystonia, usually mild and sparing facial or truncal muscles.[9] Myoclonus predominantly affects the upper body and worsens with action or emotional stimuli, as in this case.

Psychiatric manifestations such as anxiety, phobias, and obsessive-compulsive symptoms are now recognized as core features of MDS, likely reflecting SGCE-related neurobiological dysfunction rather than secondary reactions to physical disability.[2] Our patient's anxiety likely compounded her motor symptoms, underscoring the need for integrated psychiatric care. Delayed diagnosis is frequent; in our patient, the lag was 10 years. Reports exist of delays exceeding 40 years, often when psychiatric symptoms precede motor findings.[10] Misdiagnoses such as Tourette syndrome or cerebral palsy are common.[11] Neuroimaging is typically normal, and neurophysiologic studies support a subcortical origin.[1,2,6] Pathophysiology may involve Purkinje cell and basal ganglia dysfunction, along with dopaminergic and serotonergic imbalance.[2,5]

No curative treatment exists for MDS. Zonisamide has class I evidence for improving both myoclonus and dystonia; however, its use may be limited by psychiatric adverse effects, particularly in vulnerable patients.[12] Benzodiazepines and antiseizure drugs like valproate and levetiracetam may alleviate myoclonus. In our patient, clonazepam produced a partial benefit. Anticholinergics or botulinum toxin are effective for focal dystonia.[9] Tetrabenazine was selected for our patient due to the predominance of disabling myoclonus, partial response to clonazepam, and emerging evidence supporting vesicular monoamine transporter 2 inhibition in SGCE-related MDS.[5] The mechanism likely relates to dopaminergic modulation, supported by studies demonstrating striatal hyperdopaminergia in SGCE knockout models.[5,12] Our patient demonstrated significant motor improvement with tetrabenazine, consistent with prior reports.[5,8] Perampanel, an AMPA receptor antagonist, has been reported to be beneficial mainly in refractory cases, based on limited case-level evidence.[7] In refractory or severe cases, DBS targeting the globus pallidus internus or ventral intermediate nucleus has demonstrated robust and sustained benefit. Deep-brain stimulation is generally considered for patients with significant disabilities who fail medical therapy, particularly at younger ages with shorter

disease duration.[9,12-15] Botulinum toxin injections, physical therapy, and psychotherapy complement pharmacologic management.[2] This case adds to the literature supporting tetrabenazine's efficacy in SGCE-related MDS and emphasizes the importance of early recognition. Genetic testing should be considered in young patients with myoclonus and dystonia, especially when conventional treatments fail.

In conclusion, SGCE-related MDS should be suspected in early-onset cases presenting with myoclonus and focal dystonia unresponsive to standard therapies. Tetrabenazine can provide meaningful symptomatic relief and functional improvement. Early diagnosis through genetic testing enables the timely initiation of targeted therapy, improving quality of life and preventing unnecessary diagnostic delays.

Author Contributions

C.K.D.: Idea/concept, Design, data collection and/or processing, analysis and/or interpretation, literature review, writing the article; Ö.P.E.: References and fundings, materials; Y.S.K.: Control/supervision, critical review.

Conflict of Interest

The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

Funding

The authors received no financial support for the research and/or authorship of this article.

Data Sharing Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.

AI Disclosure

The authors declare that artificial intelligence (AI) tools were not used, or were used solely for language editing, and had no role in data analysis, interpretation, or the formulation of conclusions. All scientific content, data interpretation, and conclusions are the sole responsibility of the authors. The authors further confirm that AI tools were not used to generate, fabricate, or 'hallucinate' references, and that all references have been carefully verified for accuracy.

REFERENCES

1. Rachad L, El Kadmiri N, Slassi I, El Otmani H, Nadifi S. Genetic aspects of Myoclonus-Dystonia Syndrome (MDS). Mol Neurobiol 2017;54:939-42. doi: 10.1007/s12035-016-9712-x.
2. Roze E, Lang AE, Vidailhet M. Myoclonus-dystonia: classification, phenomenology, pathogenesis, and treatment. Curr Opin Neurol 2018;31:484-90. doi: 10.1097/WCO.0000000000000577.
3. Vanegas MI, Marcé-Grau A, Cazurro-Gutiérrez A, Hamami F, Timmers ER, Weißbach A, et al. Approach to myoclonus dystonia syndrome: A European reference network survey. Mov Disord Clin Pract 2025;12:957-68. doi: 10.1002/mdc3.70028.
4. Correa-Vela M, Carvalho J, Ferrero-Turrion J, Cazurro-Gutiérrez A, Vanegas M, Gonzalez V, et al. Early recognition of SGCE-myoclonus-dystonia in children. Dev Med Child Neurol 2023;65:207-14. doi: 10.1111/dmcn.15298.
5. Luciano AY, Jinnah HA, Pfeiffer RF, Truong DD, Nance MA, LeDoux MS. Treatment of myoclonus-dystonia syndrome with tetrabenazine. Parkinsonism Relat Disord 2014;20:1423-6. doi: 10.1016/j.parkreldis.2014.09.029.
6. Salamon A, Zádori D, Horváth E, Vécsei L, Klivényi P. Zonisamide treatment in myoclonus-dystonia. Orv Hetil 2019;160:1353-7. doi: 10.1556/650.2019.31472.
7. Belli E, Del Prete E, Unti E, Mazzucchi S, Palermo G, Ceravolo R. Perampanel as a novel treatment for subcortical myoclonus in myoclonus-dystonia syndrome. Neurol Sci 2023;44:2943-5. doi: 10.1007/s10072-023-06803-y.
8. Bhattacharyya KB, Roy A, Biswas A, Pal A. Sporadic and familial myoclonic dystonia: Report of three cases and review of literature. Ann Indian Acad Neurol 2016;19:258-60. doi: 10.4103/0972-2327.168625.
9. Raymond D, Saunders-Pullman R, Ozelius L. SGCE myoclonus-dystonia. [Internet]. 2020 Jun 4 [cited 22.06.2025]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1414/
10. Kübler D, Borngräber F, Lohmann K, Kühn AA. Novel SGCE mutation in a patient with myoclonus-dystonia syndrome - Diagnostic delay of more than 40 years. J Clin Neurosci 2018;50:131-2. doi: 10.1016/j.jocn.2018.01.055.
11. Varga MG, Nand NP, LeDoux MS. Delayed diagnoses of SGCE myoclonus-dystonia. Tremor Other Hyperkinet Mov (N Y) 2020;10:23. doi: 10.5334/tohm.334.
12. Fearon C, Peall KJ, Vidailhet M, Fasano A. Medical management of myoclonus-dystonia and implications for underlying pathophysiology. Parkinsonism Relat Disord 2020;77:48-56. doi: 10.1016/j.parkreldis.2020.06.016.
13. Alghamdi MA, Hamad MH, Alghamdi I, Alghamdi G, Al-Jelaify M, Alshimemeri S, et al. Sodium oxybate-treated familial myoclonus-dystonia syndrome due to novel SGCE variant. Am J Med Genet A 2025;197:e63964. doi: 10.1002/ajmg.a.63964.
14. Wang X, Yu X. Deep brain stimulation for myoclonus dystonia syndrome: A meta-analysis with individual patient data. Neurosurg Rev 2021;44:451-62. doi: 10.1007/s10143-019-01233-x.
15. Krause P, Koch K, Gruber D, Kupsch A, Gharabaghi A, Schneider GH, et al. Long-term effects of pallidal and thalamic deep brain stimulation in myoclonus dystonia. Eur J Neurol 2021;28:1566-73. doi: 10.1111/ene.14737.