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Tremor and Other Hyperkinetic Movements

Case Reports

Speech-activated Myoclonus Mimicking Stuttering in a Patient with Myoclonus–Dystonia Syndrome

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Abstract

Background: Acquired neurogenic stuttering has been considered a fairly uncommon clinical occurrence; speech-activated myoclonus is a rare entity that can mimic stuttering and is caused by a wide array of etiologies.

Case Report: Here we report a patient with myoclonus–dystonia syndrome (MDS), due to an identified disease-causing mutation, who displayed speech-activated myoclonus mimicking stuttering.

Discussion: In MDS, myoclonus has only infrequently been reported to affect speech. This case further expands the spectrum of conditions causing the rare clinical phenomenon of speech-activated myoclonus.

Keywords: Stuttering, action myoclonus, autosomal dominant familial dystonia, epsilon-sarcoglycan

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Introduction

Stuttering consists of frequent repetitions, prolongations, or interruptions of sounds or syllables that disrupt the prosody of speech.[1,2] Acquired neurogenic stuttering has been considered a fairly uncommon clinical occurrence, though Theys et al.[3] report a 5.3% incidence in their study of 319 stroke patients. The neural substrate of acquired neurogenic stuttering remains elusive, in large part because of the myriad brain structures and varied etiologies implicated.[1,4] Speech-activated myoclonus is a rare entity that can mimic stuttering and, like stuttering and other forms of myoclonus, is caused by a wide array of etiologies, most of which are acquired: neurodegenerative disease,[5] clozapine,[5,6] pharyngitis,[5] reading epilepsy,[7] reflex seizures in the context of juvenile myoclonic epilepsy,[7,8] localization-related epilepsy secondary to hypoxic brain injury,[9] Lance–Adams syndrome,[10] and as an unclear consequence of primary intestinal T-cell non-Hodgkin's lymphoma.[11] Here we report a patient with genetic dystonia, myoclonus–dystonia syndrome (MDS), with an identified disease-causing mutation who presented with speech-activated myoclonus mimicking stuttering.

Case report

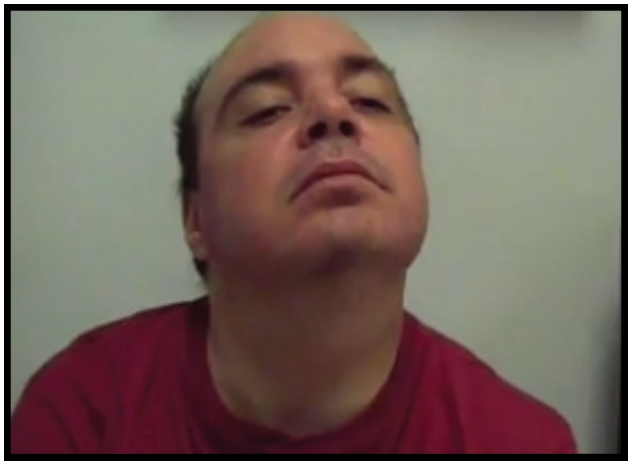
A 45-year-old Caucasian male with a history of dystonia presented to our movement disorders clinic for further evaluation and management of his condition. The patient reported that his symptoms started at age 15 after a diving accident in which he sustained a head injury that resulted in 10 minutes of unconsciousness. Following that accident, he suffered gradually progressive neck stiffness and retrocollis. Carbidopa–levodopa and botulinum toxin were ineffective treatments for his cervical dystonia. The patient drank alcohol occasionally, with resultant partial amelioration of his neck stiffness. In addition to his cervical dystonia, the patient noted some mild jerky movements in both of his arms. Around age 40, he developed onset of slowly progressive insuppressible facial twitching, which resulted in dysarthria and dysphagia.

The patient's prenatal history was unremarkable, and he met all milestones without any delay. However, in high school he was apparently labeled with a learning disability and was also thought to have attention deficit disorder, although he did not recall treatment with stimulants. He had no symptoms of obsessive-compulsive disorder. Of note, the patient did not have a history of significant exposure to heavy metals or chemicals.

The patient had a very strong family history of abnormal movements: myoclonus in his paternal grandmother and two sons (in their 20s), dystonia in his paternal grandmother's brother, and genetically proven MDS (previously known as DTY11) in his sister. His sister had a similar but more severe phenotype of myoclonus–dystonia; she did not exhibit any speech-activated myoclonus. Genetic testing identified a known disease-causing mutation in the epsilon-sarcoglygan (SGCE) gene: IVS1+1 G→A. This sequence change abolishes the splice donor site in the first intron and was previously reported in a large pedigree where this mutation cosegregated with MDS.[12] The patient's father was apparently unaffected. The patient had one nephew (son of his affected sister) in his early 20s described as having obsessive-compulsive disorder, though he did not display any abnormal movements.

Neurological examination (Video 1) revealed hypokinetic dysarthria during spontaneous speech. While reading, his speech was abruptly interrupted by synchronous myoclonic movements affecting his neck (causing slight lateral head rotation to the right), bilateral orbicularis oculi, and vocal musculature. The patient continued to exhibit stuttering-like speech while reading even when no visible myoclonus was evident, presumably as a result of myoclonus in his vocal musculature. His speech was most affected while reading aloud, with much less prominent speech disruption during spontaneous vocalization. The patient's facial expression was normal between myoclonic jerks. He demonstrated moderate–severe retrocollis and mild laterocollis to the left, with significant hypertrophy of the trapezius muscles bilaterally; even passive flexion of the head was quite painful and limited. He did not have dystonia affecting any other body segments. The patient demonstrated subtle myoclonic jerks in his distal arms bilaterally, both asynchronous and synchronous. He had some difficulties with tandem gait.

The patient was offered deep brain stimulation,[13] but he declined and was subsequently lost to follow-up. Electrophysiological investigations were unable to be obtained as a result.



Video 1. Neurologic Examination Demonstrating Speech-activated Myoclonus. The patient has moderate-severe retrocollis and mild laterocollis to the left, with irregular hyperkinetic head and neck movements during attempted head rotation, consistent with superimposed cervical action myoclonus. Myoclonus can also be seen affecting the face and arms. Spontaneous speech is relatively normal in this video segment. While reading, the patient's speech is frequently interrupted by synchronous myoclonus involving the neck, orbicularis oculi, and vocal musculature.

Discussion

MDS is a heterogeneous clinical entity characterized by bilateral, alcohol-sensitive myoclonic jerks primarily involving the neck, trunk, and upper limbs, frequently accompanied by cervical and/or brachial dystonia.[12,14] A mutation in SGCE accounts for 30–40% of typical presentations; it is inherited in an autosomal dominant fashion and maternally imprinted.[14,15] MDS is clinically heterogeneous, with some phenotypes of isolated dystonia[16–18] or isolated myoclonus.[17,18] The origin of the myoclonus in MDS remains ambiguous, though electrophysiologic and functional imaging investigations implicate a subcortical process.[14,18–20] Symptom onset in MDS is typically in childhood or early adolescence.[14] Psychiatric comorbidities, such as obsessive-compulsive disorder, are increasingly recognized.[14,16] Cognitive impairment is not a feature of the disease.[14]

In MDS, myoclonus has only infrequently been reported to affect speech.[21] In a large family with a deletion in exon 7 of SGCE, six of nine affected patients displayed laryngeal myoclonus affecting speech, described as "discrete, non-rhythmic jumps in the voice."[21] Another large kindred with MDS in the northwestern United States noted six of 10 affected family members exhibited facial myoclonus, though its impact upon their speech was not elucidated.[22] Our patient demonstrated speech-activated myoclonus that was most pronounced while reading aloud as opposed to speaking spontaneously. Speech-activated myoclonus is typically a function of the type of speech: spontaneous or rote.[5] In their case series, Slee et al.[5] demonstrated that oropharyngeal movements not involving the vocal cords (e.g. whistling, chewing, swallowing) failed to trigger myoclonus. The authors also noted that "internal speech" and writing did not activate myoclonus, concluding that in all three patients, the myoclonus was elicited by vocal speech production.[5]

In summary, we present a patient with a genetic type of dystonia, MDS, with speech-activated myoclonus mimicking stuttering, further expanding the spectrum of conditions causing this rare clinical phenomenon.

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