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Adult-Onset Walking-Upstairs Dystonia

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Dear Editor,

Dystonia has been defined as a "movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive, movements, postures, or both."[1] Dystonia can be classified according to its anatomical distribution into focal, segmental, multifocal, hemidystonia, and generalized dystonia. Focal task-specific dystonia of the upper limbs and craniocervical regions is relatively common, whereas lower extremity dystonia rarely occurs in adults and it is often associated with trauma, parkinsonism, or psychogenic behaviors. However, in recent years there have been several reports on patients with adult-onset lower extremity dystonia triggered by standing, normal walking, or walking downstairs.[2-5]

A 55-year-old woman presented with a 2-year history of difficulty walking upstairs. Supplementary Video 1 (in the online-only Data Supplement) shows that she exhibited a lack of coordination of the right lower limb muscles during the swing phase when walking upstairs. Although we were not able to perform a stair-cycle analysis, her entire stance phase was normal (from weight acceptance to pulling up and forward continuance). In contrast, the early part of the foot-clearance to foot-placement parts of the swing phase was abnormal due to the presence of a transient block soon after beginning the hip and knee joint flexion, with a consequent overflexion of the hip and knee joint due to abnormal activation of the gluteus medius and the hamstring muscles. Her ankle joint movements were within the normal range. This pattern persisted also during distractibility maneuvers. She could walk downstairs normally (Supplementary Video 1 in the online-only Data Supplement), and also walk normally up a slope without steps and upstairs backwards.

The family and personal history of the patient, including her occupational background, were unremarkable. Neurological and neuropsychological examinations produced normal findings, with no symptoms or signs of conversion. The diagnostic workup ruled out other causes of movement disorders involving the lower limbs, such as contractures, spasticity, myotonia/neuromyotonia, tonic spasms, stiff-person syndrome, abnormal posture due to paresis or atrophy, sensory ataxia and/or pseudoathetosis, seizures, or epilepsia partialis continua.[6] EEG and EMG findings were within the normal ranges. MRI findings for the pelvis, brain, and spinal cord were also normal. A baropodometric assessment and gait analysis revealed symmetric normal balance and walking pattern, respectively. Genetic testing was negative for *DYT1* mutations. Another movement disorder that was mandatory to rule out was tics, which are stereotypic recurrent movements that can usually be markedly suppressed for short periods with effort; however, this was not the case for our patient. Moreover, tics usually predominate in the face, upper arms, and neck, whereas in our patient they presented as a movement disorder only in the right lower limb. In addition, a well-known diagnostic pitfall is that tics are often less prominent or even absent in a clinical examination, whereas in our case the movement disorder was always present, and not associated with anxiety or other neuropsychological abnormalities. These observations meant that psychogenic dystonia[7] was also

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excluded.

A motor tic usually has an abrupt onset, but the subsequent movement or posture might be slow or prolonged. Such a phenomenon, namely a dystonic tic, is characterized by the movements being suppressible and stereotypic, whereas our patient was not able to suppress the abnormal movements.[6,8] Instead, our patient presented with an abnormal pattern of repeated movements involving the same muscle groups of the right lower limb as in the definition of dystonia.[9] Nevertheless, dystonia is commonly induced by an action or activity, namely task-specific dystonia, which implies the presence of movements or postures that are predominantly or even exclusively present under specific circumstances.[6] Moreover, lower limb dystonia appearing in adulthood requires a prompt investigation for trauma, stroke, *DYT1* mutations, spinocerebellar ataxia, Wilson's disease, and a reversible dysmetabolic etiology, even though such task-specific dystonias affect selective motor activities that often involve highly skilled, overlearned tasks (e.g., writing, typing, or playing a musical instrument), and occur almost exclusively when performing such activities. Indeed, walking upstairs (as well as downstairs) involves complex and stereotypic movements that require the careful integration of multiple types of sensory information to guarantee the appropriate planning and execution of motor responses.

To the best of our knowledge, this is the first report on lower limb, task-specific dystonia, which in this case occurred when walking upstairs. Such dystonias have been rarely reported, and mainly during childhood: 1) foot dystonia only when walking forward in children with DYT1 dystonia, 2) walking-induced equinovarus deformity only when the leg is at the end of the swing phase, and 3) dystonia of the lower extremities only when walking downstairs. Indeed, the dystonia reported here was probably primary, given that the main causes of secondary (as well as genetic) dystonia were ruled out.

The pathophysiology of dystonia is mainly characterized by the failure of inhibitory phenomena at different levels of the central nervous system,[10] as indicated by reduced intracortical inhibition and spinal reciprocal inhibition,[11] which lead to the characteristic co-contraction of muscles, difficulty in voluntary muscle activation, and muscle activation overflow. There is also a contribution from deterioration of a sensorimotor integration mechanism, which adds to the generation of abnormal and irregularly tuned motor drive, with a consequential presence of undesired or parasite muscle activation during movements, including task-specific dystonias.[12] For this reason, dystonia is recognized also as a sensorimotor integration disorder.[13,14] Lastly, there is a disruption of homeostatic plasticity, with a prevailing facilitation of synaptic potentiation and loss of synaptic inhibition.[15] Beyond these widely accepted mechanisms, changes in higher-order motor control at the network and system levels have also been proposed.

Both psychogenic and organic dystonias share similar failures of inhibitory phenomena within the central nervous system. This suggests that the abnormal corticospinal excitability may in part be a consequence rather than a cause of dystonia or, alternatively, may represent a primary trait of the disease predisposing to both the types of dystonia.[11] In contrast, psychogenic dystonia does not usually show sensorimotor[16-18] and synaptic[13] plasticity abnormalities. Regarding sensorimotor integration, transcranial magnetic stimulation (TMS) was applied to our patient to probe short-latency intracortical inhibition (SICI) and short-latency afferent inhibition (SAI), which are measures of intracortical paired-pulse excitability and sensorimotor intracortical inhibition, respectively.

TMS was applied over the left M1 area using a double-cone coil wired to Magstim 200^2 and Bistim^2 devices (Magstim; Whitland, UK). The motor hot spot [i.e., the optimal location for eliciting motor evoked potentials (MEPs), with the amplitude at a given intensity being largest in the tibialis anterior muscle of the right leg] was determined by moving the coil systematically in steps of 0.5 cm over the M1 area. We first determined the resting motor threshold (RMT), and then recorded 15 MEPs using a stimulation intensity of 120% RMT, which were randomly intermingled with 15 paired-pulse stimuli to assess SICI; that is, a conditioning stimulus at 80% RMT was applied 2.5 ms before the test stimulus at 120% RMT.[19]

We assessed SAI in a separate section of the muscle.[20] We first determined the active motor threshold (AMT) of the tibialis anterior muscle of the right leg and its central motor conduction time (CMCT). We then recorded 15 MEPs using a stimulation intensity of 120% AMT, which were randomly intermingled with 15 paired-pulse stimuli to assess SAI (i.e., a conditioning electrical stimulus applied to the common peroneal nerve at the knee with an amplitude of 120% AMT preceded the magnetic test stimulus at 120% AMT with an interstimulus interval that was 8 ms shorter than the CMCT). The electrical stimulation was generated using a constant-current stimulator (Model DS7A, Digitimer, Welwyn Garden, UK) and applied via two Ag-AgCl disk electrodes, with the cathode placed just distal and anterior to the fibula head, and the anode placed over the ipsilateral patella. The MEP was quantified as the peak-to-peak amplitude, while SICI and SAI were expressed as percentage values using the following formula:

$$100-\left(\frac{\text{conditioned MEP}}{\text{test MEP}}\times 100\right)$$

Both SICI and SAI were clearly inhibited, by 12% and 15%, respectively. It is therefore likely that the patient suffered from organic rather than psychogenic dystonia, since SAI has been

shown to be significantly reduced in the former compared to the latter type of dystonia.[16-18] On the other hand, SICI has been described as abnormal in both organic and psychogenic dystonias.[16-18] Moreover, SAI may be affected by the type of sensory stimulation paradigm employed (i.e., cutaneous vs. mixed-nerve stimulation).[17] The present TMS findings therefore only partially help to differentiate psychogenic from organic dystonia, and so further studies are necessary to better outline the clinical correlate of TMS abnormalities in dystonia, since this is not yet obvious.[17] Nonetheless, the sensorimotor alterations together contribute to convert physiological movement sequences into abnormal and aimless ones, as is the case in task-specific dystonias.[21]

We can conclude that it is important to better assess the pathophysiology underling task-specific lower limb dystonia. Future studies are needed to confirm whether such a focal dystonia is a sensorimotor disorder. This information may be important for patient management and rehabilitation, and facilitate the adoption of strategies that can improve proprioception.

Supplementary Video Legend

Video 1. The patient exhibited an impairment of the swing phase at the right lower limb muscles when walking up-stairs, with particular regard to the early part of the foot-clearance, up to foot-placement. Such an impairment was due to a transient movement block soon after the beginning of hip and knee joint flexion. This led to an overflexion of these joints.

Supplementary Materials

The online-only Data Supplement is available with this article at https://doi.org/10.3988/jcn.2019.15.1.122.

Conflicts of Interest

The authors have no financial conflicts of interest.

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