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Repetitive exercise dystonia: A difficult to treat hazard of runner and non-runner athletes

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ABSTRACT

Introduction: Runner's dystonia has previously been described in small series or case reports as a lower limb, task-specific dystonia. We have occasionally encountered this disorder and recognized the same phenomenon in non-runners regularly engaging in lower limb exercise. We wished to characterize the syndrome further, including outcomes, treatment, and the diagnostic usefulness of electrophysiology.
Methods: We conducted a retrospective review and follow-up survey of adults seen at Mayo Clinic (1996–2015) with task-specific dystonia arising after prolonged repetitive lower limb exercise. The findings were compared to all 21 previously reported cases of runner's dystonia.
Results: We identified 20 patients with this condition, 13 runners and seven non-runner athletes. Median age at dystonia onset was in mid-adulthood. Correct diagnosis was delayed by a median of 3.5 years in runners and 1.6 years in non-runners, by which time more than one-third of patients had undergone unsuccessful invasive procedures. Most patients had dystonia onset in the distal lower limb. Dystonia was task-specific with exercise at onset but progressed to affect walking in most. Sensory tricks were reported in some. Surface EMG was consistent with task-specific dystonia in nine patients. Botulinum toxin, levodopa, clonazepam, trihexyphenidyl, and physical therapy provided modest benefit to some, but all patients remained substantially symptomatic at last follow up.
Conclusions: Repetitive exercise dystonia is task-specific, confined to the lower limb and occasionally trunk musculature. It tends to be treatment-refractory and limits ability to exercise. Diagnosis is typically delayed, and unnecessary surgical procedures are common. Surface EMG may aid the diagnosis.

1. Introduction

Dystonia is characterized by involuntary, sustained or intermittently sustained muscle contractions causing abnormal postures or movements, which are typically repetitive, patterned, twisting, and sometimes tremulous. Dystonia may be initiated or exacerbated by voluntary action and associated with overflow muscle activation [1]. Focal dystonia may be triggered by specific tasks, usually in the upper limbs or craniocervical segments in adults.

A newly recognized type of focal task-specific dystonia, uniquely affecting the lower limbs or trunk, was recently described among long-distance runners [2]. We have increasingly encountered this condition in our movement clinic, and not only among runners, but also affecting those regularly engaging in other lower limb exercises such as cycling or elliptical training. Usually, the diagnosis has been elusive despite numerous tests and consultations.

We wished to better characterize this condition, extend to athletes engaging in other lower limb exercises, and assess the utility of electrophysiology in supporting the diagnosis. From our Mayo Clinic database, we reviewed the common clinical features of patients with this disorder, treatment strategies, and outcomes. A preliminary report of most of these patients has previously been published [3].

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2. Methods

2.1. Patient selection

Patients were identified through electronic searches of the Mayo Medical Records Linkage System, Movement Disorders Laboratory database, and the senior author's clinical database at Mayo Clinic, whose Institutional Review Board approved the study (IRB #14-005295). Adult patients (over age 18 years) who were evaluated at Mayo Clinic between January 1996 and June 2015 were eligible for inclusion if they had dystonia that initially occurred only during lower limb exercise. We stipulated that this must have developed while engaging in regular exercise for a minimum of two years, with "exercise" referring to physical activity more intense than casual walking. Search terms included "dystonia," "exercise/running/jogging," and various lower limb anatomic references. This yielded 413 cases for review. The diagnosis of dystonia was established by a movement disorders expert (all but three cases) or senior general neurologist familiar with task-specific dystonias. Patients with parkinsonism, functional, or other neurologic disorders to explain their dystonia were excluded, as were those whose diagnoses were no longer consistent with isolated dystonia during follow up. Written informed consent was obtained prior to patients' completing a follow-up questionnaire regarding their disease course, treatment trials, and current activity level. The patient in the video gave consent to be videoed for written or online publication.

Supplementary video related to this article can be found at http://dx.doi.org/10.1016/j.parkreldis.2016.03.013.

2.2. Data collection

Demographic information; personal and family medical history; dystonia characteristics; and results of laboratory, radiographic, and electrophysiologic tests were recorded. Surface EMG tracings were reviewed where available. Because of the tertiary referral nature of our practice, we gathered long-term follow-up data regarding treatment trials and functional outcome via postal questionnaire or telephone interview for patients not seen within six months of data collection. Similar data were abstracted from the original descriptions of all 21 previously reported cases of task-specific lower limb dystonia in athletes [2,4–10] and used as a comparison cohort. Standard descriptive statistics were utilized.

3. Results

3.1. Clinical features

Twenty patients met our definition of repetitive exercise dystonia, with equal numbers of men and women. The inclusion criteria stipulated that all were regular participants in intense, repetitive, ongoing lower limb exercise and that dystonia initially occurred only during exercise; this included 13 runners and seven non-runners (Fig. 1). The non-runner group included four cyclists and three whose preferred exercises were race walking, elliptical training, and modern dance. One patient was a triathlete whose dystonia initially occurred only during cycling and later also affected ambulation. No patient reported a family history of dystonia, though four had a family member with a different movement disorder. Individual patient characteristics are shown in Table 1.

Summary statistics for our groups of runners and non-runners, as well as for all previously reported cases of runner's dystonia, are shown in Table 2. The only statistically-significant difference by chi-squared analysis between Mayo cases (combined runner and non-runner groups) and previous reports was a history of prior limb injury or surgery, which was more likely in the Mayo group ($p = 0.0001$; other p values not shown). None of the features differed significantly between Mayo runner and non-runner groups.

Dystonic foot inversion and plantar flexion were the most common manifestations, but there was considerable phenotypic heterogeneity. Involvement of thoraco-abdominal and paraspinal muscles, with forward, backward, or lateral flexion of the trunk that also obeyed task specificity, was seen in five cases. All patients reported that dystonia disappeared within seconds of halting exercise.

Correct diagnosis was delayed by a median of 3.5 years in runners and 1.6 years in non-runners, with dystonia often being mistaken for an orthopedic condition or peripheral neuromuscular disorder. Unsuccessful invasive procedures were performed or recommended in nine cases. By the time of initial Mayo Clinic evaluation, most patients in both the runner and non-runner groups demonstrated progression of dystonia to also affect walking (i.e., they experienced the same involuntary dystonic posturing while walking that had previously occurred only during running or their other preferred lower limb exercise).

Most patients in both the runner and non-runner groups recalled suffering an injury (often minor such as ankle sprain) or undergoing surgery to the limb later involved by dystonia. The time from limb trauma to dystonia onset was highly variable, ranging from a few months to several years in each group of patients. Despite the history of prior limb injury or surgery, pain was generally not a feature of repetitive exercise dystonia and was reported by only two patients.

3.2. Diagnostic tests

Diagnostic tests were generally unrevealing apart from dynamic multichannel surface EMG (sEMG), which was performed in nine cases (six runners and three non-runners; Table 3). Our movement laboratory protocol includes the lumbar paraspinal, quadriceps, hamstring, anterior tibialis, gastrocnemius, and any other trunk or lower limb muscles visibly contracting or symptomatic to the patient. Recordings are made while sitting at rest, with isometric contraction, while standing still, and while walking. In all patients in this series who underwent sEMG, phasic or tonic co-contraction of agonist and antagonist lower limb and/or trunk muscles was seen only during ambulation and not while sitting at rest, consistent with (but not diagnostic of) task-specific dystonia. In seven of nine patients studied electromyographically, there was bilateral dystonic muscle activity despite unilateral symptoms (Table 3).

Nerve conduction studies and needle EMG were performed in 15 patients but did not document findings that we interpreted as diagnostically useful. All were normal except one with a chronic L5 radiculopathy on the dystonic side, but without evidence of active denervation. Another had diffuse, apparently benign fasciculations. MRI of the brain, cervical, thoracic, and lumbar spine was performed in 15, 12, 13, and 13 patients, respectively, never revealing a structural lesion to explain dystonia. Dopamine transporter imaging (^{123}I-ioflupane- 2-beta-carbomethoxy-3-beta(4-iodophenyl) nortropane, DaTscan™) was only performed in one patient and was normal.

Besides routine bloodwork, ceruloplasmin was checked in 11 patients; copper, six; and GAD65 antibodies, five, and all were normal. Genetic testing was uncommonly performed. *TOR1A* gene mutations (DYT1 dystonia) were absent in the two patients for whom they were checked. One patient was heterozygous for a novel variant of unknown significance in the *GCH1* gene (dopa-responsive dystonia) but reported only modest, very short-lived benefit from levodopa. *THAP1* (DYT6), *PRRT2* (paroxysmal