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### REVIEW

### Focal dystonia in musicians

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### KEYWORDS

Focal dystonia; Musician; Pathophysiology; Treatment; Botulin toxin; Sensory training

### Abstract

*Introduction:* A special group of focal dystonia is that known as occupational, which include dystonic disorders triggered by repetitive motor activity, closely associated with the professional activity of a specific task that the affected person performs. In this sense, musicians are a population particularly vulnerable to this disorder, which is presented during the execution of highly trained movements.

*Objective:* This article reviews the pathophysiology of focal dystonia and its therapeutic implications.

*Development:* The pathophysiological basis of focal dystonia in the musician is still not well established. However, due to the contribution of neurophysiological studies and functional neuroimaging, there is growing evidence of anomalies in the processing of sensory information, sensorimotor integration, cortical and subcortical inhibitory processes, which underline this disease.

Clinically, it is characterised by the appearance of involuntary muscle contractions, and is associated with loss of motor control while practicing music. It is a gradual appearance and sometimes there may be a history of musculoskeletal injuries or non-physiological postures preceding the appearance of the symptoms. The neurological examination is usually normal, although subtle dystonic post ures can develop spontaneously or with movements that involve the affected segments. The dystonia remains focal and is not generalised.

*Conclusions:* Treatment is based on using multiple strategies for the management of the dystonia, with variable results. Although a specific therapy has not been defined, there are general principles that are combined in each situation looking for results. This includes, among others, pharmacological interventions, management with botulinum toxin, and sensory re-training techniques.

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#### PALABRAS CLAVE

Distonía focal; Músico; Fisiopatología; Tratamiento; Toxina botulínica; Entrenamiento sensorial

### Distonía focales en los músicos

#### Resumen

*Introducción:* Un grupo especial de distonía focal son las conocidas como ocupacionales, las que incluyen trastornos distónicos desencadenados por una actividad motora repetitiva, íntimamente relacionada con la actividad profesional o tarea específica que realiza el afectado. En este sentido los músicos son una población especialmente vulnerable a esta patología, que se presenta durante la ejecución de movimientos altamente entrenados.

*Objetivo:* En este manuscrito se revisa la fisiopatología y sus implicancias terapéuticas. *Desarrollo:* Las bases fisiopatológicas de la distonía focal del músico aún no se conocen completamente. Sin embargo, gracias al aporte de estudios neurofisiológicos y de neuroimágenes funcionales, existe creciente evidencia de alteraciones en el procesamiento de información sensorial, integración sensorio-motora, procesos corticales y subcorticales de inhibición, que subyacen a esta patología.

Clínicamente, se caracteriza por aparición de contracción muscular involuntaria y se asocia a pérdida del control motor durante la ejecución musical. Es de aparición gradual y ocasionalmente pueden existir antecedentes de lesiones musculoesqueléticas o posturas no fisiológicas que anteceden la aparición de los síntomas. El examen neurológico es usualmente normal, aunque pueden desarrollarse posturas distónicas sutiles espontáneamente o con movimientos que involucran los segmentos afectados. La distonía permanece focal y no se generaliza.

*Conclusiones:* El tratamiento se basa en la utilización de múltiples estrategias para el manejo de la distonía, con resultado variables. Si bien no se ha definido una terapia especifica, existen principios generales que se combinan en cada situación buscando obtener resultados. Esto incluye intervenciones desde una perspectiva farmacológica, manejo con toxina botulínica, técnicas de reentrenamiento sensorial, entre otras.

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### Introduction

Dystonias are defined as a sustained and involuntary contraction of agonistic and antagonistic muscles, which can give rise to torsion, repetitive involuntary movements, and/or abnormal positions. From the neurophysiological point of view, we find the following: a) continuous muscular activity (spasms), that is to say, contractions lasting for more than 100 milliseconds; b) recruitment of activity of other distant muscles not participating in the movement c) preserved finality of the movement.1-3 On the basis of the segments compromised, dystonias can be classified into: a) focal, in which one part of the body is affected (such as the hand in writer's cramp or the eyes in the case of blepharospasm), b) multifocal, c) segmental, when two or more contiguous regions are compromised, d) generalized, and e) hemidystonias, when half of the body is compromised. They can also be classified by aetiology as primary and secondary. 3,4

Occupational dystonias make up a special group. As previously mentioned, musicians are a particular population exposed to this type of dystonia, which has been correlated to the requirements typical of musical learning (prolonged practising of exercises involving fine motor control).<sup>5</sup>

### Epidemiology

Generalized dystonias are uncommon with a prevalence rate of 0.2 to 11 per 100,000 inhabitants according to studies in American and European populations.<sup>6</sup> Insofar as focal dystonias are concerned this figure varies between 3 and 29.5 per 100,000 inhabitants.<sup>7,8</sup> For musician's dystonia, a form of focal dystonia, prevalence is estimated to be 0.5 to 1% of all musicians.9,10 Nevertheless, this figure varies significantly depending on the instrument played and performance demands (in particular, soloists). In their series, Jabusch et al.<sup>9</sup> described 144 professional musicians with focal dystonia, 81% males and 19% females, with a mean age of onset of 33 years (17-63 years). The instrumentalists most commonly affected are pianists, guitarists, and violinists. However, if we consider the universe of musicians by instrument, the situation changes, with violin players being the ones proportionately least affected. In this regard, there may be a certain bias, since most violinists play as part of an orchestra, with less demanding performance skill being required in comparison to a soloist.

Several epidemiological studies have put forth the following as possible predisposing factors: being female and have a positive family history of dystonia, <sup>11,12</sup> in addition to

a history of muscle-skeletal injury or nerve entrapment, <sup>13, 14</sup> obsessive personality traits and injury due to overuse. <sup>15</sup>

### Pathophysiology

The pathophysiological bases for musician's focal dystonia have still not been entirely elucidated. Nonetheless, thanks to the contributions of neurophysiological and functional neuroimaging studies, there is a growing body of evidence for alterations in the processing of sensory information, sensorimotor integration, cortical and subcortical inhibitory processes, as well as the influence of sensitive excitability stimuli in cortical areas in association with this pathology.

### Alterations in sensory processing and aetiological models based on altered cortical neuroplasticity

An alteration in tactile discrimination has been seen to exist in dystonic musicians, with an increased discrimination threshold in comparison with subjects with generalized dystonias and healthy controls. This has been related to a superposition of the areas of representation of the fingers affected in musicians with focal dystonia according to primary somatosensory mapping studies (SI) using magnetoencephalography and somatosensory evoked potentials.<sup>16-23</sup> This has led to the hypothesisthat alterations in sensory processing might underlie the pathogenesis of musician's dystonia.

The acquisition of the fine motor skills typical of musical training in healthy musicians is associated with an increased cortical representation of the fingers in SI, as well as with a decreased area of their tactile receptive fields which translates into a decreased threshold of discrimination. In an animal model (primates) that reproduces the conditions of excessive training in sequences of high-speed, stereotyped movements, this activity has been associated with a degradation in the topographic structure with superimposing of the fields of digital tactile representation in SI, as well as an increase in receptive fields, to the point where a single finger spreads its field of reception to that of adjacent fingers; 24-27 all of the above is associated with the appearance of dystonia. According to the authors, this would suggest that the demands of sensory processing in ever-decreasing lapses of time foster the appearance of dystonia. This is consistent with the fact that musician's dystonia has a higher incidence rate in soloists, subjected to greater demands of repetitive, high-speed stimuli, for many hours of daily practice.9

In normal conditions, the stimuli reaching SI almost simultaneously are integrated as a single stimulus. In contrast, those that arrive with a longer time interval, generate separate and distinct cortical representations. In the opinion of some authors, the poor spatial and temporal resolution seen in musicians with dystonia might have to do with the above-mentioned degradation in the cortical representation of sensory stimuli.<sup>18,19,26,28-30</sup>

## Alteration of inhibitory spinal, trunk, and intracortical circuits

At the level of the spine, these circuits can be assessed by testing the excitability of the reciprocal H-reflex inhibitory circuits between the antagonistic muscles of the forearm, which has led to the possibility of a dysfunction in the descending regulatory pathways on the inhibitory spinal interneurons. The inhibitory trunk circuits can be evaluated by examining the amplitude and duration of the R2 component of the blink reflex curve.<sup>31</sup> Finally, cortical inhibition can be quantified, among other ways, by measuring the latency following a pulse of transcranial magnetic stimulation (TMS). In all three cases, less inhibition can be seen in patients with dystonia in general, in comparison with healthy controls.<sup>31,32</sup>

Intracortical inhibition studies have revealed that, under normal conditions, their activity would be diminished insofar as the muscles performing a task are concerned and increased in the adjacent muscles not involved in the movement.<sup>33</sup> Thus, a mechanism of motor activity focalization is set up but is altered in patients with dystonia.<sup>34,35</sup>

#### Alterations in sensorimotor integration

In normal conditions, activation of the motor cortex triggers an influx of sensory "inputs" from the areas in movement.<sup>36</sup> This starts even before the movement begins. Experiences with somatosensorial evoked potentials (SSEP) in healthy subjects and patients with writer's cramp, reveal a lower degree of decrease in SSEP than in healthy controls during the phase prior to initiating movement.<sup>37-39</sup> According to the authors, the decrease in SSEP in healthy controls would correspond to a necessary decrease in the sensitivity of SI to the sensory stimuli, in a preparatory phase prior to beginning the movement, a mechanism that in patients with dystonia, would be dysfunctional.

Other authors have studied sensorimotor integration and interaction in greater depth. Tamburin<sup>40</sup> studied the influence of electrical stimulation of the digits (D2, D5) on motor evoked potentials (MEP) in the muscles of the hand by means of TMS in healthy subjects, revealing the existence of a topographic inhibition of the MEP; that is, the magnitude of response in the muscles was proportional to the proximity to the site of stimulation. This suggested the existence of a mechanism of focalization of motor cortex activity, which would be facilitated in the muscles receiving sensitive stimuli and inhibited in the muscles that are not involved. Using a similar methodology,<sup>41</sup> Abbruzzese compared patients with dystonia of the hand with healthy controls and found that in the latter, there is a failure in the previously mentioned mechanism of focalization, indicating the existence of increased cortical excitability.

## Findings in the study of sensorimotor integration in musicians with dystonia and healthy musicians

Arecent study compared sensorimotor integration in healthy controls who were not musicians, in healthy musicians, and in individuals with musician's dystonia.<sup>42</sup> It included the use of vibratory stimulation in hand muscles and TMS to evaluate the amplitude of the MEP in 3 muscles located in the same area: the *abductor digiti minimi* (ADM), the *abductor pollicis brevis* (APB), and the first dorsal interosseous muscle (FDI), in addition to other parameters of intracortical inhibition. In healthy subjects who were not musicians, the stimulus produced an increase in the amplitude of the MEP (less excitability of intracortical inhibitory circuits in the motor area of the muscle stimulated), with the opposite effect in those that did not receive the stimulus. In healthy musicians, there is a similar response in the muscle stimulated, which, however spread to the closest muscles while the opposite effect was maintained on the rest. In the muscicans with dystonia, the stimulus of any of the three muscles facilitated a response (MEP) in all of them, suggesting a lower degree of excitability of the intracortical inhibitory circuits. This suggests a situation in which no distinction is made between stimuli entering from any muscle, which coincides, in turn, with the alterations in the receptive areas and the areas of cortical sensory representation, as mentioned above.

These findings are consistent with a model in which musical practice in healthy musicians is associated with adaptive changes in sensorimotor integration that benefit performance, consisting of decreasing inhibition on the muscles closest (in both anatomical as well as functional terms) to the muscle stimulated. In dystonic musicians, these modifications would translate into a pattern of non-focalization of this phenomenon, secondary to an alteration in inhibitory circuitry, leading to the appearance of dystonia.

Another approach to the study of intracortical inhibitory activity has been used in a study<sup>43</sup> that reveals (by means of multi-channel EEG) that in dystonic musicians there is a loss of the synchronization present in healthy musicians between the cortical oscillatory frequencies in pre-motor and sensorimotor areas during tasks for inhibiting learned motor sequences. This alteration at the level of cortical synchronization yields new evidence that converge toward dysfunctioninintracortical inhibitory circuits as aphenomenon underlying the pathophysiology of musicians' dystonia.

In short, multiple pathophysiological alterations have been reported in musician's dystonia at the level of both cortical processing and representation of sensory stimuli in the fingers, as well as alterations in the interaction of these stimuli with the facilitation or selective topographic inhibition of the motor activity of the hand muscles. This has been attributed to an alteration in the capacity of the inhibitory circuitry to focalize action on the muscles not involved in musical performance.

Now then, to what degree the alterations described are determined by environmental factors or by factors intrinsic to the subject is a matter of debate. Although it is true that practising implies an activity that is characterized by repetitive sensory stimuli and the appearance of dystonia is associated with more hours of practice,<sup>9</sup> most musicians do not develop dystonia or the alterations in cortical representation or inhibitory circuitry already described. In this regard, most authors agree that this pathology is associated with other risk factors that are particular to each subject, in addition to prolonged musical training, as will be pointed out below.

### Phenomenology

Musician's focal dystonia is characterized by the appearance of involuntary muscle contraction during performance with

Table I Fingers anected in order of frequency	Table 1	Fingers affected in order of frequency <sup>49</sup>
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Finger affected	Flexion	Extension
Middle Ring finger Little finger Index finger Thumb	54.4% 44.3% 22.8% 16.5% 11.4%	24.1% 24.3% 25.3% 27.8% 6.3%

loss of motor control. It's appearance is insidious.<sup>9,11,44,45</sup> In a fair number of cases, there may be a history of musculoskeletal injury prior to the appearance of symptoms.<sup>11,46</sup>

Although the classical presentation is characterized by unilateral or bilateral involvement of the hands, in the case of wind instruments, it may debut with compromise of the perioral musculature or of the muscles of the tongue, in what has come to be known as "embouchure dystonia".<sup>9,47</sup>

According to different reports<sup>9,12,48</sup> the average age of onset is in the fourth decade of life. In a retrospective study carried out in Germany<sup>9</sup> that included 144 musicians with dystonia, the average duration of symptoms was 5.1 years (0.1-28 years), affecting for the most part classical music soloists (51%), with a 3:1 predominance of males. Insofar as distribution by type of instrument is concerned, the piano corresponded to 28% of the total; 26% corresponded to wind instruments; 20% to the guitar, and 11% to brass (trumpet, trombone, and others). With respect to handedness of the dystonia, it coincides with the requirements of dexterity for the instrument, affecting the right hand more often in guitarists and pianists, and the left hand more often in string instruments. The spread of involvement to other activities of daily life (writing or pushing keys) is reported in 34 to 45% of the total.48

With respect to the form of presentation, according to a retrospective study performed in Spain that included 86 cases,<sup>49</sup> the first symptom reported by the musicians was described as:

- -Lack of control of hand movements while performing (40.7%).
- -Sowness of the fingers (37.2%).
- -Tension or stiffness of the hand or forearm (9.3%).
- -Weakness of the hand (7%).
- -Trembling of the fingers (2.3%).
- —Pain (2.3%).

The fingers affected vary in frequency as shown in table 1. In addition, compensatory movements have also been reported that may compromise segments adjacent to the ones affected by the dystonia, thus interfering with performance. For instance, a patient who has one finger with dystonia in flexion may present compensatory extension in the adjacent finger.

Figure 1 presents an example of one form of presentation in the right hand of a pianist, with dystonia in flexion in the ring finger and compensatory extension in the middle finger. **Figure 1** Dystonia in the right hand of a right-handed pianist, primary dystonia, flexion of the 4<sup>th</sup> and compensatory extension of the middle finger.

As pointed out, there is another type of dystonia known as embouchure dystonia that accounts for 14% of musicians' dystonias.<sup>9</sup> It affects musicians who play wind instruments and involves the perioral, lingual and facial musculature, which translates into unco-ordinated movements and defective control of the flow of air through the instrument's mouthpiece. Given that the control of the musical tone depends on the tension and size of the opening of the lips, this symptomatology is often triggered (83%) in a specific register of tones. This may consist of tremor or, less often, of lateral deviation or protrusion of one or both lips, unilateral elevation of the labial commissure, as well as tremor or protrusion of the jaw.<sup>47</sup>

In addition to identifying dystonic movements, a meticulous examination of the muscles and skeleton is recommended, with special emphasis on the head, spine, shoulder girdle, the arm, forearm, and hand. It is essential for this to be repeated with the instrument in order to identify positions that may be associated with tension, muscle weakness, or pain, which in many cases precedes the appearance of dystonia.<sup>50-52</sup>

Finally, there are several rating scales for this disease. We recommend the Tubiana 6-point scale<sup>14</sup> because of how

 
 Table 2
 Tubiana scale, for the functional evaluation of musicians' occupational dystonia<sup>14</sup>

- Stage 0: Incapable of playing
- Stage 1: Plays notes, but stops because of blockage or lack of facility
- Stage 2: Plays short sequences without rapidity and with unsteady fingering
- Stage 3: Plays easy pieces but is unable perform more technically challenging pieces
- Stage 4: Plays almost normally but difficult passages are avoided for fear or motor problems
- Stage 5: Returns to concert pieces

easy it is to use. However, it does have the disadvantage that it can be difficult to classify the more subtle degrees of dystonia and it is more accessible to therapists with an understanding of music (table 2).

### Treatment

### General considerations

Multiple strategies have been used to manage musicians' dystonias with highly variable outcomes. Although no specific form of therapy has been defined, there are general principles that are combined in each situation in pursuit of the best results. Generally speaking, we can define interventions from a pharmacological perspective, management with botulinum toxin, techniques of sensory or paedagogical re-training, among others.

#### **Drug intervention**

*Trihexyphenidyl*: the doses used vary from one series to another (ranges of 1 mg to 30 mg daily).<sup>9</sup> The results are rather discouraging, with improvement being reported in only 33% of patients. Other anticholinergics have been used, albeit without any better results.

### Botulinum toxin

Injection of botulinum toxin into the dystonic muscles reduces the abnormal activity of these muscles by blocking the release of acetylcholine in the neuromuscular plaque. Botulinum toxin should be injected using electromyography in the muscles selected for intervention, so as to observe movement while the patient plays his/ her instrument, clearly distinguishing dystonic activity from compensatory movements. Patients with patterns of extension or flexion in the fingers are injected preferentially in the forearm. Again, doses vary depending on the different series in the literature.<sup>9,53</sup>

The muscles most often injected are: the *flexor digitorum* superficial is and profundus, the *flexor carpi radialis*, *flexor pollicis longus*, the *digitorum extensor* and the *indicis extensor*, and the *palmaris interosseus*. The mean dose per muscle group was 112 units (range of 88-150) in the arm and shoulder muscles, 38 units (range of 5-85) in the forearm extensors, 65 units (between 10 and 175) in the flexors of the forearm, and 26 units (from 5 to 84) in the muscles of the hand. After the BT injection, 49% of patients reported an improvement and 57% received more than one injection.<sup>9</sup> Schuele reports similar figures, proposing appraisal of improvement on the basis of three parameters: absence of muscle weakness following injection of the level of musical repertoire and position within the orchestra.<sup>51</sup>

Most authors report a worsening of musical performance with the use of botulinum toxin in the case of embouchure dystonia.<sup>53</sup> This, although not specifically stated, may have to do with the difficulty involved in maintained a proper "seal" with the perioral musculature as previously mentioned. No prognostic factors have been clearly established with respect to treatment with botulinum toxin; nevertheless, proper selection of the muscles to be injected and the use of electromyography are fundamental aspects to be taken into consideration.

### Paedagogical re-training

Most authors agree ast o the advant ages of a multidisciplinary approach to the treatment of musicians' dystonias, including the participation of a neurologist and a physical therapist, in addition to a psychotherapeutic approach in many cases.  $^{9,14,54}$ 

Paedagogical re-training covers any number of treatment approaches with the common element of being supervised by an instructor and being aimed at limiting and correcting the appearance of dystonic and compensatory movements. Although, the characteristics of the re-training involved may vary from one team to the next, there are certain basic principles:<sup>5,9,19,21,26,54,55</sup>

- -The movements of the body area involved are limited to the threshold of strength and speed at which the dystonic movement is triggered.
- -Detection and management of muscle weakness in the shoulder girdle, arm, forearm, and hands.
- -Compensatory movements can be avoided (at least partially) through the use of splints.
- -Visual feedback is favoured through the use of a mirror, so that the patient will become capable of distinguishing the dystonia from the typical movements involved in playing. Moreover, this makes it possible to correct positions that hinder playing.
- -Use of techniques (for instance, Feldenkreis) that increase the perception of non-dystonic movements of both the fingers as well as the segments adjacent to the fingers affected.

The aforementioned can be associated in addition to or as an alternative to non-specific musical exercises; that is to say, that do not belong to any repertoire in particular, but rather are designed to improve the technique of playing the instrument (respecting the principles stated previously). Both approaches are associated with symptomatic improvement in 50% of cases<sup>9</sup>, although they demand both dedication and time, which may limit treatment adhesion.

It is worth adding<sup>14,54</sup> that starting therapy early in the course of the disease, less severe symptoms, early age, and the absence of concomitant psychological disorders are all predictors of a favourable outcome.

### **Ergonomic treatment**

Ergonomic treatment consists in limiting dystonic movements through the use of splints in the finger affected or avoiding dystonic movements by means of repositioning or redesigning a key in the case of the wind instruments, for instance.<sup>56</sup> In this type of treatment, it is worth mentioning all types of support (music stands, straps, etc.) that can be of use when the hand affected by the dystonia must hold the instrument while playing.<sup>9</sup>

### Physical rehabilitation

Modern-day concepts of rehabilitation seek to re-establish a physiological position that supports musicians' free gestures.

It includes a comprehensive analysis of shoulder, arm, wrist, and finger movements, as well as general posture while performing. Body awareness and adjusting positions to favour relaxation are the mainstays of this form of intervention.<sup>51</sup>

### Emotional and occupational support

This approach must contemplate the multiple factors associated with the presence of dystonia, including aspects of mental health. Musicians tend to suffer stress as a result of being separated from the professional community for prolonged treatment and of experiencing a decline in their performance that hinders their return.<sup>51</sup>

Being aware of the pursuit of perfection, restructuring the routines of a fulfilling life, and educating the teaching or occupational environment with respect to this health problem are part of the treatment. This should take place in parallel to the previously mentioned approach, contemplating the presence of an interdisciplinary team that includes neurology, physical therapy, occupational therapy, psychology, among others.

### Conclusions

Musician's dystonia has a tremendous impact on patients' quality of life and in many cases, puts an end to their careers. There is a host of factors associated with this condition and its pathophysiology, although it is still being studied, there are some principles that are useful for preventive management and treatment.

Treatment incorporates multiple strategies, although no one specific form of therapy has been defined. There are previously expressed general principles that, in combination, seek to improve symptoms. The focus is interdisciplinary, with the possible inclusion of pharmacological interventions, management with botulinum toxin, re-training techniques, emotional and occupational support, among others.

### Conflict of interest

The authors state that there is no conflict of interest.

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