Focal Hand Dystonia Affecting Musicians. Part I: An Overview Of Epidemiology, PathoPhysiology And Medical Treatments

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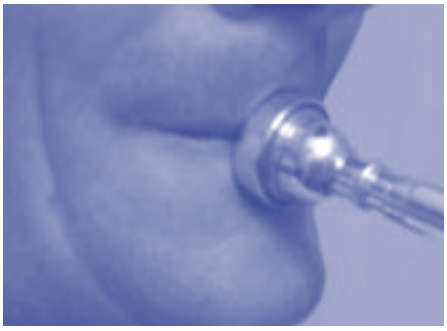
In 1911, Oppenheim coined the term 'dystonia' to describe disordered motor control, characterised by an association of hypotonia and tonic muscle spasm. Focal hand dystonia is one form of this disorder, in which symptoms are often task-specific and occur during skilled movements such as writing (writer's cramp) or playing a musical instrument (musician's cramp). Much research has been conducted on the pathophysiology of dystonia, but the underlying mechanisms still remain unclear. Hypotheseses about functional central nervous system alterations continue to gain more support. Scientific treatment-based publications on focal dystonia are sparse, and progress in evidence-based treatment options are necessary in order to assist this patient group. This paper will review the literature, documenting dystonia classification criteria, manifestations, pathophysiology and medical treatment techniques for musicians affected by focal hand dystonia.

INTRODUCTION

Dystonia is a syndrome characterised by involuntary, prolonged muscle contractions that can lead to sustained twisting postures (*Fahn 1998, Fahn et al 1998, Fahn et al 1987*). Three criteria can be utilised to assist in classifying this syndrome: age at onset, aetiology and distribution of symptoms (*Fahn et al 1987 and 1998*). An onset before 28 years of age is classified as early onset dystonia, and after this age as late-onset dystonia. Aetiology can be divided into either idiopathic (no obvious effects on the brain) or symptomatic (often the basal ganglia are affected, resulting in more generalised symptoms). Some literature uses the term primary or secondary for the same classification (*Fahn 1998*). Thirdly, dystonia can be classified according to its distribution of symptom manifestation: in generalised dystonia, symptoms may manifest in all extremities including the trunk; in hemidystonia, the symptoms are focused on one side of the body; segmental dystonia affects a segment of the body or adjacent body parts; and focal dystonia affects a single body part.

Any part of the body can be affected by focal dystonia, including the arms, legs, trunk, neck, face, eyelids or vocal cords (*Berardelli et al 1998; Deuschl and Hallett 1998*). Focal dystonia tends to be named according to the affected region: such as blepharospasm (eyelids), spastic torticollis (cervical dystonia), lingual dystonia, spastic dystonia and oromandibular dystonia. This paper will focus on focal hand dystonia, a late-onset primary dystonia that is often task-specific and includes writer's or musician's cramp. The symptoms may affect a single finger up to the hand/wrist; they do not tend to generalise and remain fairly task-specific (*Bressman et al 1998*) (Figures 1a and 1b).



Figures 1a and 1b: Task-specific, action-induced Focal Hand Dystonia has different forms, including writer's cramp and musician's dystonia (hand and embouchure)

FOCAL HAND DYSTONIA IN MUSICIANS

Musicians with focal hand dystonia display varying symptoms which tend to be specific to that individual person and can include incoordination, cramping and tremor (*Jankovic and Shale 1989*). The symptoms musicians display depend upon the instrument played, rather than hand dominance. Dystonia in pianists often manifests as a curling in of the fourth and fifth fingers of the right hand, whereas guitarists often show a curling in of the third finger of the right hand. Flautists tend to be affected in the left hand, whereas violinists and clarinettists can have involuntary contractions in either hand (*Altenmüller 1998; Brandfonbrener 1995; Hochberg et al 1983*). Although focal dystonia manifests as a motor problem, interestingly this can be influenced by a 'sensory trick' (eg playing the musical instrument while wearing a latex glove). Many entities appear to 'trigger' the manifestation of focal hand dystonia in musicians, such as a sudden increase in playing or practice time; a dramatic change in technique; a return to studies after a long break from the instrument; a trauma (not necessarily recent); current or a history of nerve

entrapment; psychological trauma or a change of instrument (*Brandfonbrener 1995*).

EPIDEMIOLOGY

Dystonia in musicians might be considered as a more common problem than is often realised. The prevelance among professional musicians can be estimated as 2-10% (*Jabusch 2006; Branfonbrener 1995; Lim et al 2001*), which is higher than that of writer's cramp (0.1%) in the general population (*Nutt et al 1988*). This high number certainly reflects the specific demands made upon musicians. More male than female musicians are affected with ratios ranging from 2:1 to 6:1 (*Lederman 1991; Brandfonbrener 1995; Lim et al 2001*), with only 6% reporting a history of either writer's cramp or musician's dystonia in their families.
A recent survey of 116 patients with focal hand dystonia, the largest series to be published, showed that the mean age at onset of symptoms is about 33 years (range 17-63 years) (*Jabusch 2006*). About half the patients were in professional soloist positions, 17% were tutti players in orchestras, 17% held teaching positions and 15% were students. These results showed that classical musicians are predominately affected by musician's focal hand dystonia (95%) with only a minor number of jazz and pop musicians affected. In this series, 28% were keyboard instrumentalists, 26% were woodwind players, 20% played plucked instruments, 15% were bowed string players, and 11% were brass players (*Jabusch 2006*).
There are reports of predisposing medical conditions, which may include: ulnar neuropathy (*Charness et al 1996; Ross et al 1995*), local trauma or peripheral nerve injury (*Brandfonbrener 1995*) and biomechanical limitations (*Leijnse 1997a and b; Leijnse et al 1992 and 1993; Wilson et al 1993*). However, these studies do not focus specifically on musician's cramp but also include other types of focal hand dystonia, which may influence the findings.

FOCAL HAND DYSTONIA IN MUSICIANS – PATHOPHYSIOLOGY

It is thought that focal task-specific hand dystonia develops due to a functional disturbance at several levels of the central nervous system. This is mainly expressed as an imbalance between inhibition and excitation of neurons. There is evidence for alteration in sensory and motor cortical representation, which may result in altered integration of sensory information from the periphery into adjacent and required motor programmes.
There follow some of the theories postulated for the development of this functional disturbance. Due to overlapping, it is difficult to identify if focal hand dystonia is due to a sensory, central or motor disturbance. However, the points will be outlined following the sensory motor loop to allow a clearer overview.

SENSORY ALTERATION

Animal studies showed that repetitive performance of a hand gripping movement can actively degrade cortical representation in the primary sensory cortex (*Byl et al 1996a and b; Wang et al 1995*). The changes are not only at the cortical level, but also involve other brain regions, which are also involved in sensory processing (*Lenz and Byl 1999; Sanger and Merzenich 2000*). Similar alterations have been found with

Figures 2a and 2b: Focal Hand Dystonia in musicians showing motor inco-ordination or loss of voluntary control in pianist's and cellist's right small and ring fingers.

increasing severity in healthy musicians and musicians with dystonia. An increased cortical representation of the left hand in healthy string players (*Elbert et al 1995*), and a blurring/overlapping of cortical representation in musicians with focal hand dystonia (*Elbert et al 1998*), echo the animal study findings.

DISTURBANCE OF SENSORIMOTOR INTEGRATION

Animal studies have shown that cortical abnormalities lead to deficient motor preparation (*Feve et al 1994; Van der Kamp et al 1995; Yazawa et al 1999*). The same phenomenon was displayed in a study of affected musicians. Due to disturbed sensory representations, the motor cortex receives altered information (*Rosenkranz et al 2000*), and deficiencies in sensorimotor integration can lead to loss of motor control (*Abbruzzese et al 2001; Siebner et al 1999*). Both functional and structural changes occur in any musician's brain, as they adapt to the demands of their activity. These plastic changes are necessary in order to achieve a high performance level; however it is argued that they may render the musician susceptible to development of dysfunctional motor control syndromes such as focal hand dystonia (*Pascual-Leone 2001*).

MOTOR DISTURBANCE

Studies with transcranial magnetic stimulation of the motor cortex have shown that cortical output again seems to be higher in patients with dystonia, which may contribute to the excessive motor output that occurs during voluntary movement (*Mavroudakis et al 1995; Ikoma et al 1996*). Mapping of cortical sites, from which specific muscle responses can be elicited by brain stimulation in patients with writer's cramp, also showed an alteration in size and location (*Thompson et al 1996*), suggesting some reorganisation of cortical excitability in dystonia. Ridding et al (*1995*) found that there was less intracortical inhibition in patients with focal, task-specific primary dystonia when tested at rest. They proposed that, under normal circumstances, one role of the inhibition was to 'focus' the motor command within the cortex so that the correct muscles were activated by the right amount in any task. A deficiency in this cortical inhibitory system could therefore contribute to the overflow of activity in dystonia. Indeed, reduced excitability of this inhibitory system could also account for the increased cortical output again noted from above.

In conclusion, it is not clear if sensory changes drive motor alteration, or whether repetitive movements drive sensory changes that lead to motor remapping. Due to the close interlinks within the loop, it is difficult to differentiate where the alteration has occurred. Functional disturbances of the basal ganglia have been found in primary dystonia (*Ceballos-Baumann and Brooks 1997 and 1998; Berg et al 2000; Naumann et al 1998 and 1996*), however, no studies were found that specifically related to musicians in this regard. Musicians appear to be a special group of people as the tasks they perform are extremely specific. Therefore, it is likely that, in musicians with focal hand dystonia, higher order motor areas are involved. For an in-depth presentation of the pathophysiology of focal dystonia, see Lim et al (*2001*).

GENETICS

There appears to be conflicting evidence to support a genetic contribution to development of focal hand dystonia in musicians. Early onset dystonia, which can manifest itself as focal or generalised dystonia, is commonly attributed to the gene DYT1 (*Bressman 1998; Bressman et al 1998*). A family history of movement disorders or writer's cramp has been described in about 10% of musician patients (*Altenmüller 1998; Hochberg et al 1990*). Brandfonbrener (*1995*) found no positive family history. There is no specific genetical study on larger samples of musicians with focal hand dystonia, but one study does analyse ten Ashkenazi Jewish patients with focal hand dystonia (eight with musician's cramp, two with writer's cramp) and no common mutation could be found, thus arguing against a genetic role in the etiology of musician's cramp (*Gasser et al 1996*).

PSYCHOLOGICAL FACTORS

Once established, the symptoms of musician's cramp can be aggravated by psychological stress, such as performance situations. However, psychological factors are not thought to significantly contribute to the development of dystonia (*Sheehy and Marsden 1982*). There is evidence that blepharospasm may have an association with obsessive-compulsive disorder (*Bihari et al 1992a and b*), and thus psychological status is included as a possible risk factor for development of focal dystonia. It could be argued that to be a musician of outstanding ability requires a certain personality in order to achieve the standard.
It is thus understandable that musicians with focal dystonia often despair when they cannot control their hands to perform tasks that they have practised for hours everyday. This can, in turn, cause development of secondary psychological difficulties.

DYSTONIA DIFFERENTIAL DIAGNOSIS

Dystonia can be distinguished from most common painful conditions, in which muscle pain usually continues after task completion, as pain is usually not evident with focal dystonia. If a patient is trying to override contractions by abnormally positioning joints and soft tissues, pain may result (Kember 1997). Muscle ache may occur after a prolonged spasm. When making a diagnosis of focal hand dystonia, other pathologies such as ganglions, Dupuytren's disease, trigger finger, meningiomas (Wynn Parry 1998), compression neuropathies and compartment syndrome affecting the intrinsic muscles of the hand must be excluded (Amadio and Russotti 1990).

TREATMENTS

Current literature that focuses on medical-based treatments for focal hand dystonia will be reviewed. Dystonia is difficult to treat (*Fahn et al 1987*) and is recalcitrant to intervention (*Byl and Topp 1998*). At present there appears to be no one cure for dystonia and many of the treatment modalities have significant limitations. It is therefore important that patients are educated regarding the limitations of treatment (*Lim et al 2001*). Many years ago, Gowers (*1893*) observed that patients with writer's cramp could benefit from a freer approach to writing, and if possible should be encouraged to learn to use the other hand. However, he comments that in about half of the cases the other hand can become similarly affected. The option of swapping hands is not as convenient for musicians, as they are often required to use two hands to play their instrument. Many articles identify a need for investigation into treatment approaches. Progress in scientifically analysed and reported treatment options for this condition are necessary, in order to offer affected patients evidence-based options.

Current treatments include:

- oral medications
- Botulinum toxin (BTX) injections
- surgery
- rehabilitative therapies and
- supportive approaches.

Medical-based treatment options will be discussed in this article. Rehabilitative therapies and supportive approaches are the focus of Part II.

ORAL MEDICATIONS

Oral medications do not cure focal hand dystonia, but can be used as palliative treatments. Anticholinergic drugs influence neurotransmission in the basal ganglia, and can thus be useful for treating patients with focal dystonia (*Altenmüller 1998*). Trihexiphenidyl is, at present, the most effective oral medication; however the side effects, even when using small doses, limit its long-term use (*Altenmüller 1998*). The side effects can include a dry mouth, fatigue and slight memory impairment. Dopaminergic medication appears to be less effective in treating focal dystonia than trihexiphenidyl (*Muller et al 1996*).

BOTULINUM TOXIN (BTX)

BTX type A is the most widely studied and utilised treatment of focal hand dystonia. BTX injections are seen as the treatment of choice for cervical dystonia and blepharospasm, and good results have been reported for writer's cramp, oromandibular and spasmodic dystonia (*Sojer et al 2001*). Intramuscular BTX injections weaken the muscle by reducing the release of acetylcholine (a neurotransmitter) at the neuromuscular junction (*Kedlaya et al 1999; Singer and Weiner 1995; Coffield at al 1994*). BTX injections must be repeated regularly (*Marion 1999*), as the effects are reversible and may only last one to three months. It is important to administer enough, but not too much BTX, otherwise muscle weakness and impairment can occur (*Altenmüller 2001; Cole et al 1991 and 1995; Ross et al 1997*).
Musicians who are affected by focal dystonia in a single digit are most effectively treated by administration of BTX (*Altenmüller 1998*). However, in most musicians, rather more complex movement patterns than just single fingers are impaired. As BTX injections are limited due to the associated weakness of nondystonic muscles (*Priori et al 2001*), and the as yet unknown long-term effects of these injections on the sensorimotor system, there is a need to develop new treatments for musicians with focal dystonia. Furthermore, these injections only blur the obvious symptoms without tackling the origin (*Cole et al 1991*).

SURGERY

There is controversy surrounding the use of surgery as a treatment technique for patients with focal dystonia. Winspur (*1998*) believes that any surgery (eg tendon transfers and nerve transpositions) is contradicted when treating such patients as it only causes further scrambling of disturbed motor programmes. He believes that attention to technique, change in instrument, resolving mechanical problems, and instrument adjustments should all be considered before surgery.

Lozano and Linazasoro (*2000*) comment that peripheral surgical techniques can alleviate focal dystonic symptoms for many patients. Others report no improvement in focal dystonic symptoms, even when clearly entrapped nerves are released (*Charness et al 1996*). Marion (*1999*) states that surgery is only indicated in very severe cases and requires thorough planning and discussion of indications, while Singer and Weiner (*1995*) state that only a surgeon with extensive training and experience in operating on patients with focal dystonia should do so. Objective scientific studies need to be completed to state if surgery would benefit this patient group.

CONCLUSIONS

Musicians with focal hand dystonia can display varying symptoms that are very specific to each individual. This condition appears to be more common in the musician than in the general population, probably due to the repetitive specific and detailed hand use required to play a musical instrument. Focal hand dystonia is thought to develop due to functional disturbance at several levels of the central nervous system. It is unclear if sensory changes drive motor alteration, or whether repetitive movements drive sensory changes that lead to motor remapping. The tasks musicians perform are extremely specific, and therefore it is probable that musicians with focal hand dystonia have involvement of the higher motor area. Dystonia is very difficult to treat and medical-based treatments are quite limited in effectiveness. Oral medications have numerous side effects and are purely palliative. Botulinum toxin injections frequently require re-administration and only treat the symptoms without tackling the origin of the problem. Surgery can be viewed as a contraindication and should only ever be entered into extremely cautiously by a senior consultant with experience in operating on musicians with dystonia. Thus, rehabilitative approaches are seen as being the treatment options that should be trialled first with this patient group, and these will be covered in the next paper.

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