

Focal dystonia: diagnostic, therapy, rehabilitation

Eckart O. Altenmüller and Hans-Christian Jabusch

Definition, classification, symptoms and diagnostic

Definition

The general term dystonia is used to describe a syndrome characterised by involuntary sustained muscle contractions, frequently causing twisting and repetitive movements, or abnormal postures [1, 2]. If these symptoms are restricted to one body part, the syndrome is termed 'focal dystonia'. In task specific focal dystonia the most prominent characteristic is the degradation and loss of voluntary control of highly overlearned complex and skilled movement patterns in a specific sensory-motor task.

Classification

Dystonia can be classified by age of onset, cause, or by distribution of the body parts affected. Dystonia localised to a single body part such as the hand or neck is referred to as focal. *Focal dystonia* is by far the most frequent form of dystonia, accounting to about 90% of all dystonia syndromes. Dystonia localised to two contiguous body parts is referred to as *segmental dystonia*. Dystonia affecting body parts that are not next to each other is referred to as *multifocal dystonia*. Dystonia affecting one segment and another body part is classified as *generalised dystonia*. If it affects only one half of the body it is called *hemidystonia*.

Focal dystonia may be classified according to four criteria: age of onset, aetiology, affected body

region and severity of symptoms [1]. According to Fahn, classification by age of onset represents the best prognostic indicator as to whether there will be a spread of dystonic symptoms to other body parts. While it is acknowledged that an age-criterion is rather arbitrary, onset before 28 (median of 9 years) is classified as early-onset primary dystonia and thereafter as late-onset dystonia (median of 45 years).

Second, current classification for aetiology divides focal dystonia into just two major categories, idiopathic or primary (including familial and sporadic forms), and symptomatic or secondary [1]. Secondary focal dystonia can be caused by structural abnormalities of the brain, or by metabolic disorders. For example, focal dystonia may occur as an early sign of Wilson's disease, a defect of copper metabolism that causes abnormal liver function and central nervous system symptoms such as tremor, and dystonia. Patients taking medications for psychiatric diseases such as schizophrenia or psychosis may develop dystonia as a drug reaction, e.g., after medication with Haloperidol. Rarely, focal dystonia may occur as a psychogenic dystonia in psychiatric disorders. Dystonia may also be associated with other neurological disorders. These are classified as dystonia-plus syndromes. Dystonia may be associated with Parkinson's disease or with myoclonus, another movement disorder which is characterised by involuntary muscle jerking. Focal dystonia may be part of other neurodegenerative disorders, for example Huntington's disease. For an in-depth presentation of the subdivisions of focal dystonia by etiologic classification, see [2].

Third, focal dystonia may be classified according to the affected region or the task

involved in the dystonic movement: spasmodic torticollis (cervical dystonia), blepharospasm (eyelids), oromandibular dystonia, spastic dysphonia (vocal folds), or writer's or musician's cramp. For writer's or musician's cramp, the dystonic symptoms may affect a single finger up to an entire arm. With respect to the task specifically involved, embouchure dystonia may affect coordination of lips, tongues and breathing in brass and wind players, whereas pianist's cramp and violinist's cramp affect the control of hand movements in musicians when playing these instruments. Other activities producing dystonic movements involve playing golf (the 'yips') or dart ('dartism'), and – less frequently – playing tennis. Rarely, task specific dystonia occurs in other highly skilled hand movements, for example in watchmakers, dentists or surgeons.

Fourth, classification according to severity of symptoms distinguishes four stages. These stages are reflected in the most common clinical scaling instrument, the dystonia-severity scale [3].

- a) *Simple focal dystonia*: Dystonic movements are only present during a specific task, e.g., exclusively during writing
- b) *Complex focal dystonia*: Symptoms occur in multiple tasks, e.g., also when using utensils for eating, or when buttoning shirts
- c) *Dystonic cramps*: Sustained muscle contractions, abnormal movements and postures of the body part occur spontaneously and are more or less continuously observable.
- d) *Progressive dystonic cramps*: Sustained muscle contractions, abnormal movements and postures tend to spread to adjacent body parts

Some authors consider *focal tremor* as a special form of focal dystonia. It may occur as a predominant symptom as cervical tremor. Task specific focal tremor is occasionally seen as embouchure tremor or as task specific tremor of the bowing arm in string-players. Rosenbaum and Jankovic [4] found task-specific tremors in 10 (36%) out of 28 patients, eight of whom had task-specific focal dystonia. Commonly, focal tremor is a sustained

postural and kinetic tremor at around 7 Hz frequencies with mostly irregular tremor amplitude. There is no tremor during rest [5].

Symptoms

The most prominent symptom of focal dystonia is loss of voluntary control and muscular coordination of movements, be it control of the head position in cervical dystonia, of eye-blinks in blepharospasm, or of skilled hand movements in focal hand dystonia [1, 2]. Typically, focal dystonia occurs without pain, although muscle aching can present after prolonged spasms. Lack of pain distinguishes it from repetitive strain injury or occupational fatigue syndrome. It is important to make this distinction bearing in mind that on the other hand prolonged pain syndromes may lead to symptomatic dystonia, possibly due to the degradation of sensory receptive fields in the somatosensory cortex (see below). The loss of muscular coordination is frequently accompanied by a co-contraction of antagonist muscle groups. For example in writer's cramp, the co-activation of wrist flexor and wrist extensor muscles is regularly observed. Dystonia can be worsened by stress and anxiety, whereas it may be relieved with relaxation and sleep. In Figure 1, typical postures of dystonic movements in musicians are shown.

In musicians with hand dystonia, an association exists between the instrument group and the localisation of focal dystonia. In instruments with different workload, different complexity of movements or different temporospatial precision for both hands, focal dystonia appears more often in the more heavily used hand. Keyboard musicians (piano, organ, harpsichord) and those with plucked instruments (guitar, e-bass) are primarily affected in the right hand. All these instruments are characterised by a higher workload in the right hand. Additionally, guitar playing requires higher temporospatial precision in the right hand compared to the left hand. Bowed string players who have a higher workload and complexity of movements in the left hand are predominantly affected in the left hand [6].