Dystonia/PGS/diagnosis

Dystonic dyskinesias are repetitive, relatively stereotyped, involuntary, purposeless contractions of one or more muscles. At the beginning of the development of dystonic syndrome, free movement can suppress involuntary muscle contractions. In some patients, a specific somatosensory stimulus (e.g. touching a certain part of the body) can temporarily suppress excessive muscle activity (gesture antagonist).

Classification of dystonia by age of onset

Dystonia beginning before the age of 12 usually begins in the lower extremities and quickly generalizes. Dystonia with onset between 13–20 usually starts on the upper limbs and tends to gradually generalize. Dystonias starting after the age of 20 are mainly focal.

Classification of dystonia according to distribution

The most common are **focal dystonias** in the form of cervical dystonia (torticollis) and blepharospasm (bilateral dystonic contractions of the orbicularis oculi muscle and adjacent muscles), less often graphospasm, oromandibular dystonia and laryngeal dystonia. **Segmental dystonia** affects 2 or more adjacent parts of the body (e.g. Meige syndrome with head muscle involvement and craniocervical dystonia). **Multifocal dystonia** is characterized by the involvement of 2 or more non-adjacent muscles or muscle groups. **Generalized dystonia** affects at least one limb and another part of the body, a variant of which is also **hemidystonia**.

Classification according to etiology

Dystonia is a syndrome that can have a number of causes (see table). According to the etiology, dystonia is divided into **the primary (idiopathic) sporadic or familial occurrence**, in which, in addition to dystonia, tremors may also appear, but the other clinical findings are normal. The group of primary dystonias also includes the **so-called dystonia plus syndromes** most often in combination with; parkinsonism or myoclonus. **Heredodegenerative dystonic syndromes** are genetically determined, dystonia usually dominates them and is associated with the occurrence of other neurological (additional extrapyramidal, pyramidal, cerebellar, peripheral motoneuron lesions and myopathy) and other symptoms (behavioural disorders, cognitive deficit, hepatopathy, splenomegaly, etc.) like **secondary dystonia**, which, however, is not genetically determined.

In primary dystonias, abnormal muscle activity appears initially only during certain movements (task-specific) and only later occurs even at rest. Conversely, in secondary dystonic syndromes, dystonia at rest occurs from the beginning. Hemidystonia is almost always secondary in origin. Also, early or more pronounced bulbar musculature involvement compared to limb involvement or limb involvement suggests a secondary etiology.

In electromyography, dystonia is characterized by permanent involuntary muscle activity, usually of an irregular character. In focal and segmental dystonias, it is necessary to rule out an epileptic origin by electroencephalography.

In the differential diagnosis of dystonias affecting the cervicocranial region, it is necessary to distinguish **facial hemispasm**, which is not assigned to dystonic syndromes, as well as chorea, tics and psychogenic origin. Hemispasm is clinically manifested by twitching of the periorbital and often perioral muscles, including the platysma muscle. Twitches can be irregular and even in bursts. Very rarely occurs bilaterally. Hemispasm occurs when the n. facialis is focally affected (e.g. contact with the posterior inferior cerebellar artery) or after its peripheral paresis.

Etiology of dystonic syndromes

Primary sporadic dystonia

- cervical dystonia (torticollis)
- blepharospasm
- graphospasm
- oromandibular dystonia
- laryngeal dystonia

Primary hereditary dystonia (dystonia plus syndrome)

- progressive torsion dystonia (Oppenheim)
- do-responsive dystonia (Segawa)
- myoclonic dystonia

Dystonia in herododegenerative diseases

- Wilson's disease
- Parkinson's disease
- Huntington's disease
- progressive supranuclear palsy
- multiple system atrophy
- corticobasilar ganglionic degeneration
- pantothenate kinase deficiency
- spinocerebellar ataxia
- juvenile parkinsonism
- mitochondrial disorders

Secondary dystonia

- perinatal trauma
- medication (dopaminergic medication, antipsychotics, anticonvulsants)
- encephalitis
- craniocerebral trauma
- hypoxia
- focal lesions of the spinal cord and brain
- peripheral lesions
- electric shock
- intoxication (methanol, carbon monoxide, manganese)
- hypoparathyroidism
- psychogenic

Therapy

Botulinum toxin is the drug of choice in the treatment of focal dystonias. Oral benzodiazepines (clonazepam, diazepam, tetrazepam)can be tried orally for generalized and torpid dystonic syndromes, tetrazepam), anticholinergics (procyclidine from 3×2.5 mg up to 60 mg daily, biperiden from 2×1 mg up to 16 mg daily in three doses), carbamazepine and tetrabenazine (from 2×12.5 mg up to 200 mg daily in 3 doses), which was not registered in the Czech Republic at the time. Baclofen can be administered orally (40–180 mg daily) or continuously intrathecally. Deep brain stimulation or selective lesion (rhizotomy, pallidotomy, thalamotomy) can be effective. For dystonia with onset in childhood, and adolescence, but also before the age of 45, it is necessary to test the response to L-DOPA (150-500 mg at least a month) and rule out Wilson's disease.