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DYSTONIA

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What is dystonia

Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive, movements, postures, or both. Dystonic movements are typically patterned,twisting, and may be tremulous. Dystonia is often ini-tiated or worsened by voluntary action and associatedwith overflow muscle activation.

Dystonic tremor

A spontaneous oscillatory, rhythmical,although often inconstant, patterned movement produced by contractions of dystonic muscles often exacerbated by anattempt to maintain primary (normal) posture. The dystonic tremor may not berelieved by allowing the abnormal dystonic posture to fully develop without resistance(“null point”). Dystonic tremor may bedifficult to distinguish from essential-typetremor.

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An example of dystonic tremor

Overflow Motor

overflow commonly found in dystonia

is unintentional muscle contraction which accompanies, but is anatomically distinct from the primary dystonic movement.

It commonly occurs at the peak ofdystonic movements.

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Alleviating maneuvers (sensory tricks or gestes antagonistes)

Voluntary actions that specifically correct theabnormal posture or alleviate the dystonicmovements. These are usually simplemovements (“gestes”) involving, ordirected to, the body region affected by Dystonia but not consisting in a forcefulopposition to the phenomenology ofdystonia.

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Sensory trick

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Classification

dystonia syndromes are currently classified along 3 main axes: **etiology, age at onset and body distribution.**

Age at onset

- Infancy (birth to 2 years);
- Childhood (3–12 years);
- Adolescence (13–20 years);
- Early adulthood (21–40 years);
- Late adulthood (>40 years).

Body Distribution

- Focal
- Segmental
- Multifocal
- Generalized (with or without leg involvement)
- Hemidystonia

Classification by body region affected is clinically important because of implications for diagnosis and therapy.

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Focal:

Only one body region is affected. Typical examples of focal forms are blepharospasm, oro-mandibular dystonia, cervical dystonia, laryngeal dystonia, and writer’s cramp. Cervical dystonia, is considered a form of focal dystonia, although by convention the shoulder can be included as well as the neck.

Segmental. Two or more contiguous body regionsare affected. Typical examples of segmental formsare: cranial dystonia (blepharospasm with lower facial and jaw or tongue involvement) or bi-brachial dystonia.

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Multifocal. Two noncontiguous or more (contigu-ous or not) body regions are involved.

Generalized. The trunk and at least 2 other sites are involved. Generalized forms with leg involvement are distinguished from those without leg involvement.

Hemidystonia. More body regions restricted to one body side are involved. Typical examples of hemidystonia are due to acquired brain lesions in the contralateral hemisphere.

Temporal Pattern

1.Persistent. Dystonia that persists to approximately the same extent throughout the day.

2. Action-specific. Dystonia that occurs only during a particular activity or task.

3. Diurnal fluctuations. Dystonia fluctuates during the day, with recognizable circadian variations in occurrence, severity and phenomenology.

4. Paroxysmal. Sudden self-limited episodes of dystonia usually induced by a trigger with return to preexisting neurological stat

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Associated Features.

- Isolated dystonia. Dystonia is the only motor feature, with the exception of tremor.(pure,primery)
- Combined dystonia. Dystonia is combined with other movement disorders (such as myoclonus,parkinsonism, et(dystonia plus)

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Dystonic syndromes

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Early-Onset Generalized Isolated Dystonia

DYSTONIA beginning in childhood often progresses to generalized involvement, sometimes quite rapidly. These cases may be familial or sporadic, genetically defined or without known cause

DYT 1,

DYT6

Are examples

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Focal or Segmental Isolated Dystonia with Onset in Adulthood.

Cervical dystonia, blepharospasm, and writer's cramp are the most common forms of focal dystonia, usually with onset in the fifth decade.

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Blepharospasm

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Oromandibular dystonia

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Jaw closing oromandibular dystonia

Dystonia-Parkinsonim.

A number of disorders, many of which are inherited, combine dystonia and parkinsonian features, sometimes accompanied by pyramidal tract involvement or other neurological deficits. Non-motor features, including cognitive decline,

are not infrequent.

dopa responsive dystonia (DRD), Wilson’s disease, Parkin PINK1-, and DJ-1-associated parkinsonism (PARK2,6, and 7), X-linked dystonia-parkinsonism/Lubag(DYT3), rapid-onset dystonia-parkinsonism (DYT12), and neurodegeneration with brain iron accumulation (NBIA, including PANK2- and PLA2G6-associatedneurodegeneration,

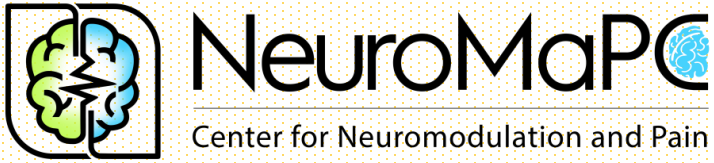
DRD

DRD or Segawa disease is mainly a pediatric-onset disorder due to defects in dopamine synthesis. The usual age of onset varies between 4 and 6 years. The ratio of females to males is 2.5:1. Prevalence has been reported to be 0.5 to 1 per million, but it is likely underestimated as the penetrance is low, and atypical cases are frequent. The onset is often insidious, with fatigability, clumsiness of gait, and dystonic postures often limited to one foot.

DRD

Progressive increase in severity of the dystonia during the day and a marked decrease, sometimes complete disappearance following sleep, are highly characteristic. The disorder is progressive and may lead to severe disability. The involvement of other parts of the body may occur quickly or take up to ten years. Cognitive functions are preserved.

Dystonia and/or parkinsonism usually show a rapid, marked and sustained response to low doses (3–5 mg/kg/ day) of L-dopa combined with an inhibitor of peripheral decarboxylation helping to determine the clinical diagnosis. The response is independent from the delay in initiating treatment.



DRD

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NDBIA

Neurodegeneration with brain iron accumulation (NBIA) encompasses a group of inherited disorders that share the clinical features of an extrapyramidal movement disorder accompanied by varying degrees of intellectual disability and abnormal iron deposition in the basal ganglia. The genetic basis of ten forms of NBIA is now known. The clinical features of NBIA range from rapid global neurodevelopmental regression in infancy to mild parkinsonism with minimal cognitive impairment in adulthood, with wide variation seen between and within the specific NBIA sub-types

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PANK dx (NDBIA)

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Myoclonus Dystonia

Rapid jerky movements mayoccur in dystonia patients.Particularly when affecting a limb, these can be mistaken for distinct myo-clonic jerks due to various causes. The term “myoclonic dystonia” is used to refer to this myo-clonic-like appearance of fast dystonic movements. Patients with “myoclonus dystonia” (DYT11) presenta combination of dystonia and myoclonus; this disorder is probably the same as “essential myoclonus”since many of these patients have subtle additional dystonia or some individuals have pure myoclonuswhile others in the same family have both myoclonus and dystonia40; in many cases, myoclonic jerks can bedistinguished from fast “jerky” dystonic movements based on clinical and electrophysiological features.41

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Dystonic jerk

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Essential myoclonus

Etiology

- Nervous system pathology
- Evidence of degeneration
- Evidence of structural (often static) lesions
- No evidence of degeneration or structural lesion

Inherited or acquired

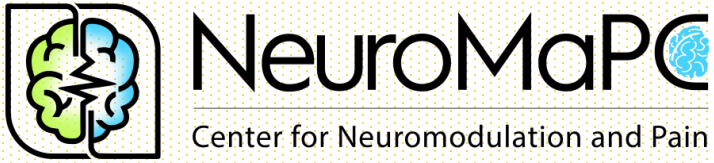
- Inherited
- Autosomal dominant
- Autosomal recessive
- X-linked recessive
- Mitochondria

Aquired

- Perinatal brain injury
- Infection
- Drug
- Toxic

- Vascular
- Neoplastic
- Brain injury

- Psychogenic



Aquired

Diagnosis

- The diagnosis of dystonia is clinical, the core being abnormal postures (with or without tremor) and the recognition of specific features, e.g. gestes antagonistes, overflow, mirror movements
- Appropriate investigations are required if the initial presentation or the course suggest heredodegenerative or secondary (symptomatic) dystonia

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Use of genetic test in diagnosis andcounselling

- DYT1 testing is recommended for patients with limb-onset, primary dystonia with onset before age 30 (level B), as well as in those with onset after age 30 if they have an affected relative with early onset dystonia (level B).
- In dystonia families, DYT1 testing is not recommended in asymptomatic individuals
- DYT6 testing is recommended in early-onset dystonia or familial dystonia with cranio-cervical predominance or after exclusion of DYT1 (good practice point).

Genetic testing

- A diagnostic levodopa trial is warranted in every patient with early onset dystonia without an alternative diagnosis (good practice point).
- Individuals with early-onset myoclonus affecting the arms or neck, particularly if positive for autosomal dominant inheritance and if triggered by action, should be tested for the DYT11 gene .
- Diagnostic testing for the PNKD gene (DYT8) is recommended in symptomatic individuals with PNKD
- Gene testing for mutation in GLUT1 is recommended in patients with paroxysmal exercised-induced dyskinesias, especially if involvement of GLUT1 is suggested by low CSF/serum glucose ratio, epileptic seizures or haemolytic anaemia .

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Use of brain imaging in the diagnosis of dystonia

- Structural brain imaging is not routinely required when there is a confident diagnosis of primary dystonia in adult patients, because a normal study is expected in primary dystonia .
- structural brain imaging (MRI) is necessary for screening of secondary forms of dystonia (good practice point). CT may be required to differentiate between calcium and iron accumulation.
- Presynaptic dopaminergic scan (DAT or 18F-DOPA) are useful to differentiate between dopa-responsive dystonia and juvenile Parkinson's disease presenting with dystonia (good practice point). This can also be useful to distinguish dystonic tremor from parkinsonian tremor

Treatment

- Botulinum toxin treatment continues to be the first choice treatment for most types of focal dystonia.
- BoNT/A (or type B if there is resistance to type A) can be regarded as first line treatment for primary cranial (excluding oromandibular) or cervical dystonia
- BoNT are safe and efficacious when repeated treatments are performed over many years (good practice point), but doctors and patients should be aware that excessive cumulative doses may be dangerous, particularly in children (good practice point).

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Other medical treatment

- Anticholinergic therapy
- Baclophen
- Clonazepam
- Dopaminergic therapy
- Anti Epiletic therapy(zonisomide myoclonus dystonia

Carbamazepin for PRRT

- Others :muscle relaxant,sodium oxybate,zolpidem,amantadine,caffein

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Palidal stimulation

Long-term electrical stimulation of the globus pallidus internus (GPi) is now established as an effective treatment for various types of dystonia, The use of deep brain stimulation(DBS) for dystonia currently addresses in particular primary generalized or segmental forms, complex cervical dystonia, and tardive dystonia in patients who do not achieve sufficient relief with conservative approaches .

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Thank You...

Any Questions?

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