

Dystonia and Treatment



IRENE OH, MD
THE NEUROLOGY CENTER, ENCINITAS
MOVEMENT DISORDERS

Part 1



GENERAL INFORMATION

Phenomenology



- Involuntary, sustained contractions of muscles, causing repetitive or twisting movements of the affected body part.
- Typically worsen with action.
- Can be task-specific, such as with writer's cramp.
- *Geste antagoniste* - a specific touch to an affected body part can help improve the dystonia.

Classification



- **Age of onset**

- Early-onset = <26 years
- Late-onset = >26 years

- **Distribution**

- Focal
- Segmental
- Hemibody
- Multifocal
- Generalized

- **Etiology**

- Primary dystonia
- Secondary dystonia

- **Genetics**

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Age of Onset



- Early-onset – Average age of onset = 9 yrs. Usually begins in a limb, most commonly a leg, at first with activities and then more at rest. Generalizes within 5 yrs.
- Late-onset – Average age of onset = 30's-40's. Usually begins as a focal dystonia in the upper body, affecting the arms, neck, or cranial muscles. May worsen to become segmental, but rarely generalizes.

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Distribution



- Focal – single body region
- Segmental – contiguous body regions
- Hemibody – arm and leg on the same side
- Multifocal - >2 noncontiguous body parts
- Generalized – entire body

Examples of Focal Dystonias



- Blepharospasm – affects the eyelid
- Cervical dystonia – affects the neck
 - Most common adult-onset focal dystonia.
- Oromandibular dystonia – affects the mouth/jaw
- Limb Dystonia – affects the arm or leg
 - Writer's cramp

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Etiology



Primary Torsion Dystonia

- Dystonia is the sole abnormality.
- No acquired or environmental cause identified.
 - Labs and imaging negative.
- No dramatic response to levodopa.

Secondary Dystonia

- Often associated with other neurological symptoms.
- Tends to occur more commonly at rest.
- Associated with known environmental or acquired causes.
 - Stroke, trauma.
 - Tardive dystonia from meds.

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Genetics



- Genetic testing commercially available for DYT1.
- DYT1: DYT1 testing should be performed in patient with generalized dystonia with age of onset <26 yrs.
- DYT5 (=dopa-responsive dystonia): Diurnal fluctuations, worse later in the day. Extremely sensitive to treatment with levodopa.

Part 2



TREATMENT

PHARMACOLOGICAL - ORAL



- Trial of levodopa
- Anticholinergics – trihexiphenidyl (Artane)
- Baclofen
- Benzodiazepines – clonazepam (Klonopin)

- Other: muscle relaxants – tizanidine, cyclobenzaprine; anticonvulsants – carbamazepine, gabapentin; dopamine-depleting agents – tetrabenazine; dopamine antagonists

Anticholinergics



- Trihexyphenidyl is the best-studied medication for use in dystonia.
- Patients with young-onset generalized dystonia appear to have the most benefit.
- Side effects: dry mouth, blurred vision, urinary retention, memory problems, sedation, confusion.

Baclofen



- Typically less potent than anticholinergics, but better tolerated.
- Side effects: nausea, sedation

Benzodiazepines



- Usually used as a supplementary medication.
- Side effects: sedation, depression, confusion, dependence

PHARMACOLOGICAL - INJECTION



- For focal dystonias, botulinum toxin injections.
- For blepharospasm and cervical dystonia, botulinum toxin injections are considered first-line treatment.

SURGICAL



- Deep brain stimulation
 - Indicated in the treatment of medical refractory primary generalized dystonia.

Deep Brain Stimulation



- FDA-approved for the treatment of Parkinson's disease and essential tremor. Received the Humanitarian Device Exemption from the FDA for primary dystonia in 2003.
- Involves chronic electrical stimulation to specific brain nuclei through electrodes implanted deep in the brain.

DBS Outcomes



Favorable

- Reducible dystonia
- Younger age
- Shorter disease duration
- Primary generalized dystonia
- DYT1-positive gene testing
- Cervical dystonia
- Tardive dystonia
- Camptocormia – bending at waist when standing and walking, resolves when lying down

Less Favorable

- Fixed postures or contractures
- Hemidystonia or secondary dystonia
- Tendency of axial symptoms to respond less
- Significant white matter involvement on MRI

PHYSICAL THERAPY



- Posture and alignment
- Stretching and strengthening, flexibility