Dystonia and Treatment

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MOVEMENT DISORDERS
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**
Classification

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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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- Genetics
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.
• 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

<table>
<thead>
<tr>
<th>Favorable</th>
<th>Less Favorable</th>
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<tbody>
<tr>
<td>Reducible dystonia</td>
<td>Fixed postures or contractures</td>
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<tr>
<td>Younger age</td>
<td>Hemidystonia or secondary dystonia</td>
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<tr>
<td>Shorter disease duration</td>
<td>Tendency of axial symptoms to respond less</td>
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<tr>
<td>Primary generalized dystonia</td>
<td>Significant white matter involvement on MRI</td>
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<tr>
<td>DYT1-positive gene testing</td>
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<tr>
<td>Cervical dystonia</td>
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<td>Tardive dystonia</td>
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<td>Camptocormia – bending at waist when standing and walking, resolves when lying down</td>
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PHYSICAL THERAPY

- Posture and alignment
- Stretching and strengthening, flexibility