



Fatta B. Nahab, M.D. | Associate Professor of Neurosciences

Evaluation and Management of Dystonia

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Overview of Dystonia

- Defining Dystonia
- Classifying Dystonia
- Clinical features
- A difficult diagnosis to make...
- Causes
- Treatments

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What is Dystonia?

“A syndrome of sustained muscle contraction, causing twisting and repetitive movements and/or abnormal postures due to co-contraction of agonist and antagonist muscles.”

- Bressman and Fahn

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Dystonia Classification - OLD

- The benefit of most classification systems is to provide a link between what is seen **clinically** and what is happening **biologically**.
- In dystonia, that has not been the case and multiple classification schemes have been developed...each to fix something while further **complicating** things.
 - Example classifications:
 - By cause (writer's cramp, runner's dystonia, etc.)
 - By age of onset (primary generalized vs. secondary focal)
 - By distribution (generalized, hemi, segmental, focal)
- VERY CONFUSING FOR EVERYONE!!!

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Dystonia Classification - NEW

- In 2013, the experts developed a new set of 'consensus' guidelines to diagnose dystonia
- Revised definition:

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 initiated or worsened by voluntary action and associated with overflow muscle activation.
- Better? More complicated...

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Dystonia Classification - NEW

<p>Axis I. Clinical characteristics</p> <p>Clinical characteristics of dystonia</p> <p>Age at onset</p> <ul style="list-style-type: none"> • Infancy (birth to 2 years) • Childhood (3-12 years) • Adolescence (13-20 years) • Early adulthood (21-40 years) • Late adulthood (>40 years) <p>Body distribution</p> <ul style="list-style-type: none"> • Focal • Segmental • Multifocal • Generalized (with or without leg involvement) <p>Hemidystonia</p> <p>Temporal pattern</p> <ul style="list-style-type: none"> • Disease course <ul style="list-style-type: none"> ○ Static ○ Progressive • Variability <ul style="list-style-type: none"> ○ Persistent ○ Action-specific ○ Diurnal ○ Paroxysmal <p>Associated features</p> <p>Isolated dystonia or combined with another movement disorder</p> <ul style="list-style-type: none"> • Isolated dystonia • Combined dystonia <p>Occurrence of other neurological or systemic manifestations</p> <ul style="list-style-type: none"> • List of co-occurring neurological manifestations 	<p>Axis II. Etiology</p> <p>Nervous system pathology</p> <p>Evidence of degeneration</p> <p>Evidence of structural (often static) lesions</p> <p>No evidence of degeneration or structural lesion</p> <p>Inherited or acquired</p> <p>Inherited</p> <ul style="list-style-type: none"> • Autosomal dominant • Autosomal recessive • X-linked recessive • Mitochondrial <p>Acquired</p> <ul style="list-style-type: none"> • Perinatal brain injury • Infection • Drug • Toxic • Vascular • Neoplastic • Brain injury • Psychogenic <p>Idiopathic</p> <ul style="list-style-type: none"> • Sporadic • Familial
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Albanese, et al. Mov Dis 2013.

Dystonia Classification - NEW

OLD	NEW
Blepharospasm	Adult-onset, focal, static, action-specific (eye-opening), isolated dystonia without evidence of neurodegeneration

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Dystonia Classification

- With this new classification system that is even more complex than the prior system, it's important patients and clinicians (non-researchers) remain focused on the practical clinical components of dystonia

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Clinical Features of Dystonia

- Stereotyped abnormal movements and postures
- Repeatedly involves same muscle groups
- Sustained
- Tremor or myoclonus (muscle jerks) may occur
- Rigidity is usually absent
- Often activated by voluntary movements
- Sensory tricks (geste antagoniste)

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Dystonia...a difficult diagnosis to make

- Lack of familiarity with dystonia by non-neurologists
- Variable presentations
- “Bizarre” and variable clinical features
- Few specific diagnostic tests
- Many causes



Understanding Dystonia at the level of the Brain

- Great strides have been made to understand what is happening in the Dystonia brain.
- Three themes show up consistently:
 1. Loss of inhibition and Excess of Movement
 2. Sensory Dysfunction
 3. Dysfunction in brain plasticity



Dystonia Therapeutic Challenges

- Since the movement patterns and suspected brain regions are similar across dystonia, therapeutic strategies across subtypes are remarkably similar
- Dystonia is a ‘rare disorder’ with no gold standard diagnostic test
- Treatment trials are difficult to perform
- Demonstrating efficacy with any single treatment is challenging
- A lack of therapeutic evidence can mean different things:
 - It’s never been studied
 - No [good] studies have been performed - no placebo
 - The studies did not show statistical efficacy - needed by FDA
 - It doesn’t work



Judging the Quality of Evidence

AAN Classification of Recommendations

Level A	• Established as effective, ineffective, or harmful (or established as useful/predictive or not useful/predictive) for the given condition in the specified population. • Requires at least 2 consistent Class I studies.
Level B	• Probably effective, ineffective, or harmful (or probably useful/predictive or not useful/predictive) for the given condition in the specified population. • Requires at least 1 Class I study or 2 consistent Class II studies.
Level C	• Possibly effective, ineffective, or harmful (or possibly useful/predictive or not useful/predictive) for the given condition in the specified population. • Requires at least 1 Class II study or 2 consistent Class III studies.
Level U	• Data inadequate or conflicting; given current knowledge, treatment (test, predictor) is unproven. • Studies not meeting criteria for Class I-III.

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Dystonia Therapies – Botulinum toxins

- Level A
 - Botulinum toxins (BoNT) for Cervical Dystonia
- Level B
 - BoNT for blepharospasm
- Level C
 - BoNT for face, vocal cords, jaw, shoulder, hand, back, leg, medication-induced

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AAN 2016 Practice Guidelines; van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Anticholinergics

- Level B
 - Trihexiphenidyl – no benefit for generalized
- Level C
 - Trihexiphenidyl - benefit for generalized/focal

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van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Muscle Relaxants

- Level C
 - Baclofen - benefit for generalized/focal
 - Diazepam (valium) - no effect



van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Dopamine altering

- Level B
 - Levodopa - no benefit
- Level C
 - Levodopa - benefit (case report)
 - Antipsychotics - mixed results in generalized, focal, multi-focal
 - Tetrabenazine - no benefit



van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Intrathecal Baclofen

- Level B
 - Benefit in generalized dystonia
- Level C
 - Benefit in segmental and generalized dystonia



van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Surgical denervation

- Level C
 - Variable improvement in focal, cervical, and segmental



van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Surgical Brain Lesions

- Level C
 - Thalamotomy – benefit for generalized and hemidystonia
 - Pallidotomy – benefit for generalized



van den Heuvel 2016 Mov Dis.

Dystonia Therapies – DBS

- Level C
 - Bilateral GPI DBS – benefit for generalized
 - Unilateral GPI DBS – benefit for focal/cervical
 - Unilateral/bilateral STN DBS – mixed results



van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Motor Cortical Stimulation

- Level B
 - No benefit
- Level C
 - Unilateral epidural – benefit for focal upper extremity

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van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Electroconvulsive Therapy

- Level C
 - Improvement in 3-6 sessions

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van den Heuvel 2016 Mov Dis.

Dystonia Therapies – Allied Health

- Level B
 - Benefit following Extracorporeal shock wave therapy
- Level C
 - Improvement for focal upper extremity and generalized

What is ESWT?

Extracorporeal shockwave therapy is a treatment used in physical therapy, orthopedics, urology and cardiology. The shock waves are abrupt, high amplitude pulses of mechanical energy, similar to soundwaves, generated by an electromagnetic coil or a spark in water.

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van den Heuvel 2016 Mov Dis.

Alternative Dystonia Therapies

- Generally a mix of options that have been inadequately studied, with some anecdotal evidence
- Biofeedback/Meditation/Hypnosis
- Acupuncture
- Yoga/Tai chi
- Cannabis

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Summary

- Features of Dystonia vary greatly based on the appearance and etiology
- Due to the complexity of dystonia as an entity, diagnosis and management can be challenging
- Some of the most exciting developments in understanding dystonia coming from research into the underlying brain mechanisms that DO seem to have have commonalities across the various subtypes.
- Treatment options depend on symptom severity
- Alternative therapies should be considered cautiously and require further study
- Dystonia science CANNOT move forward without your involvement!

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