



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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| evidence of degeneration or structural lesion ited or acquired evited Autosomal dominant Autosomal recessive V-linked recessive |
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| Mitochondrial quired Perinatal brain injury Infection Drug Toxic |
| • Vascular • Neoplastic • Brain injury • Sychogenic opothic • Familial UC San Diego |
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| 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

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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:

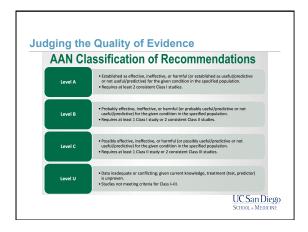
- Loss of inhibition and Excess of Movement
 Sensory Dysfunction
- 3. Dysfunction in brain plasticity

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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

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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

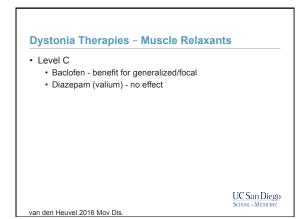
Level B

Trihexiphenidyl – no benefit for generalized
Level C

Trihexiphenidyl - benefit for generalized/focal

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van den Heuvel 2016 Mov Dis.



Dystonia Therapies – Dopamine altering

Level B

Levodopa – no benefit

· Level C

- Levodopa benefit (case report)
 Antipsychotics mixed results in generalized, focal, multifocal
- Tetrabenazine no benefit

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Dystonia Therapies – Intrathecal Baclofen

Level B

Benefit in generalized dystonia

Level C
Benefit in segmental and generalized dystonia

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Dystonia Therapies – Surgical Brain Lesions

Level C

Thalamotomy - benefit for generalized and hemidystonia
Pallidotomy - benefit for generalized

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| Level C | |
|---|--------------|
| Bilateral GPi DBS - benefit for generalized | |
| Unilateral GPi DBS – benefit for focal/cervical Unilateral/bilateral STN DBS – mixed results | |
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- Level B
- No benefit

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Level C
 Unilateral epidural – benefit for focal upper extremity

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| Level C | |
|---|-----------------------------------|
| Improvement in 3-6 sessions | |
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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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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! UC San Diego

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