Evaluation and Management of Dystonia

Overview of Dystonia

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

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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- Bressman and Fahn
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!!!

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...

Dystonia Classification - NEW

- Age at onset:
  - Infantile (birth to 3 years)
  - Childhood (3-12 years)
  - Adolescent (13-17 years)
  - Adult (18-65 years)
  - Older adult (>65 years)
- Body distribution:
  - facial
  - Thoracic
  - Lateral
  - Generalized
  - Hemifacial
- Associated features:
  - Isolated dystonia or combined with another movement disorder
  - Febrile disorders
  - Generalized dystonia or systemic manifestations
  - Post-streptococcal toxins or systematic infections

Dystonia Classification - NEW

OLD
Blepharospasm

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

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

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)
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 well-accepted or not well-accepted) for the given condition in the specified population.
  - Results of at least 2 consistent Class I studies.
- Level B
  - Partially effective, ineffective, or harmful (or probably effective, probably ineffective, or probably harmful) for the given condition in the specified population.
  - Results of at least 1 Class II study or 2 consistent Class III studies.
- Level C
  - Possibly effective, ineffective, or harmful (or possibly effective, possibly ineffective, or possibly harmful) 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 bias, patient selection.
  - Studies not meeting criteria for Class I-III.

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

Dystonia Therapies – Anticholinergics

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

**Dystonia Therapies – Muscle Relaxants**

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

**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

**Dystonia Therapies – Intrathecal Baclofen**

- Level B
  - Benefit in generalized dystonia
- Level C
  - Benefit in segmental and generalized dystonia
Dystonia Therapies – Surgical denervation

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

Dystonia Therapies – Surgical Brain Lesions

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

Dystonia Therapies – DBS

• Level C
  • Bilateral GPI DBS – benefit for generalized
  • Unilateral GPI DBS – benefit for focal/cervical
  • Unilateral/bilateral STN DBS – mixed results
Dystonia Therapies – Motor Cortical Stimulation
- Level B
  - No benefit
- Level C
  - Unilateral epidural – benefit for focal upper extremity

Dystonia Therapies – Electroconvulsive Therapy
- Level C
  - Improvement in 3-6 sessions

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.
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

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!