IC 35: An Overview of Childhood Onset Dystonia: Evaluation, Diagnosis and Treatment

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Childhood Onset Dystonia: Objectives

• Learn about the spectrum of etiologies seen in childhood dystonia

• Establish a diagnostic algorithm

• Discuss the spectrum of treatments that can be offered

Movement Disorders: Definitions / Diagnosis

• A movement disorder is any condition that affects volitional well coordinated movements or causes excess involuntary movements.

• Can be Primary (genetic / idiopathic), or Secondary (toxins, psychogenic, stroke ...)

Movement Disorders: Definitions / Diagnosis

• Hyperkinetic
  – Dystonia
  – Chorea
  – Tics
  – Ballismus
  – Stereotypies
  – Abdominal dyskinesia
  – Akathic / Tardive movements
  – Asynergia/ataxia
  – Athetosis
  – Dysmetria
  – Hemifacial spasm
  – Hyperekplexia
  – Hypnogenic dyskinesia
  – Jumpy stumps
  – Moving fingers/toes
  – Myokymia
  – Myorhythmia
  – Paroxysmal dyskinesia
  – Restless legs

• Hypokinetic
  – Bradykinesia (Parkinsonism / Parkinson's)
  – Apraxia
  – Blocking tics
  – Cataplexy / drop attacks
  – Catatonia
  – Freezing phenomena
  – Hesitant gait
  – Hypothyroid slowness
  – Rigidity
  – Stiff muscles

Disclosure Information

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Speaker Name: Mark Gormley Jr. MD
Relevant Financial Relationships: Research support from Allergan and Ipsen
Disclosure of Off-Label and/or investigative uses:
I will discuss Botulinum toxin off label use and/or investigational use in my presentation.

Speaker Name: Peter D. Kim, MD, PhD
Relevant Financial Relationships: None
Disclosure of Off-Label and/or investigative uses:
I will discuss DBS under the humanitarian device exemption for dystonia

Speaker Name: Tim Feyma MD
Relevant Financial Relationships: None
Disclosure of Off-Label and/or investigative uses:
I will discuss off label uses of medicines because I treat kids.
Focusing on Dystonia…

Dystonia: Definition

In the course of the last five years I have repeatedly observed an affliction, whose meaning and classification caused great difficulties. When examining the first cases I was trying to decide between a diagnosis of hysteria, and idiosyncratic bilateral athetosis; but then I soon realized that neither of these diagnoses was appropriate, and that this was a new condition… I have selected the titles dysbasia lordotica progressiva and dystonia musculorum deformans and would prefer the latter.

Oppenheim H. / Neuro Centrabl. 30:1090-1107 1911

Dystonia: Definition

- A Hyperkinetic movement disorder
- Dystonia:
  - Movements sustained at the peak of movement
  - Usually twisting and oft repetitive
  - Can progress to sustained prolonged abnormal postures
  - Patterned to often involve the same muscle groups

Dystonia: Anatomy

- Dysfunction occurs when an imbalance in motor control arises:
Dystonia: Anatomy

- Dysfunction occurs when an imbalance in motor control arises:

\[ \text{Dystonia: Anatomy} \]

\[ \text{Dystonia: Anatomy} \]

Supreme Court Justice Potter Stewart about pornography in 1964

“I shall not today attempt further to define the kinds of material I understand to be embraced…, but I know it when I see it.”

Dystonia: Diagnosis

- Etiology workup is guided by the history + exam and can include:
  - Imaging
  - Blood for metabolic + genetic studies
  - Cerebrospinal fluid

Dystonia: Diagnosis

- Hyperkinetic: dystonia, focal, idiopathic

Dystonia: Diagnosis

- Hyperkinetic: hemi-dystonia, post stroke
Dystonia: Diagnosis

- Hyperkinetic: dystonia, focal, tardive

Dystonia: Diagnosis

- Hyperkinetic: dystonia, generalized, kernicterus

Dystonia: Diagnosis

- Hyperkinetic: dystonia, generalized, HIE

Dystonia: Workup

- Genetic:
  - Hereditary DAT deficiency
  - Tyrosine Hydroxylase deficiency
  - Dystonia genes (DYT1-21 - the primary dystonias include 1, 2, 3, 16, 17 - 21)
  - SCA 6, 17, 2, 3, 17
  - Childhood Huntington’s Disease
  - GLUT-1 deficiency
  - Prenatal disorders (Sepiapterin Reductase deficiency is 1 of 4 types described)
  - AADC deficiency
  - Axonal Intermittence
  - Gangliolodosis
  - Gliadin Abs
  - Homozygous
  - Juvenile Parkinson’s
  - Mitochondriol Lysosomal Pathology
  - Mitochondrial Leber’s
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- Non-Genetic:
  - Nutritional (Vitamin E, Copper)
  - Autoimmune
  - Medication induced + Tardive syndrome
  - Stroke
  - Trauma
  - Infection

Dystonia: Workup

- Imagen:

Dystonia: Quantification?

- Diagnosis is based upon observation, but rating scales do exist.
  - Unified Dystonia Rating Scale
  - Fahn-Marsden Scale
  - Hypertonia Assessment Tool
  - Barry-Albright Dystonia Scale
  - Movement Disorder-Childhood Rating Scales
Dystonia: Treatment

- Disease Specific
- Physical
- Medications
- Surgical

Goal of Treating Pediatric Dystonia

Improve function

Treatment Goals

- Dystonia impairs movement, swallowing, articulation, etc.
- Often a differential between motor function and cognition, i.e. cognition higher than motor skills
- Because of the difficulties of treating dystonia goals often basic, but still difficult to achieve
- Communication and independent mobility common goals

Treatment Options in Pediatric Dystonia

- Therapy
  - Physical, occupational, speech therapy
  - Assistive technologies
  - Oral medication
  - Bracing and casting
  - Neurolytic blocks
    - Botulinum toxin
    - Phenol blocks
  - Orthopaedic surgery
  - Neurosurgical treatments
    - Intrathecal baclofen pump
    - Selective dorsal rhizotomy
    - Deep brain stimulator

Combination therapy most typical

PT/OT/SP

- Typically children with dystonia receive ongoing PT/OT/SP focused on specific functional goals
- Contractures less of an issue in pure dystonia, however most patients with mixed tone and contractures still common
- Positioning particularly important since postural correction often easily altered
Bracing for dystonia
- Still commonly used, but less so than with spasticity
- Ambulatory patients often find braces uncomfortable because of posturing and not helpful
- Trial and error like much of dystonia treatment
- Assistive technology otherwise common

Dystonia treatment
Some dystonias improve with light touch on the involved part (geste antagoniste)

Tarsy and Simon, NEJM 2006

Communication
- In patients with severe dystonia and GMFCS IV-V dysarthria and dysphagia common
- Many patients and their families would be happy just with independent communication
- Improvement of articulation by improving dystonia often difficult
- Speech and occupational therapy a mainstay
- Use of augmentative systems common but often not successful
- Rehabilitation engineer and seating specialist key to success

Communication
- Often, unless the dystonia is controlled success with augmentative communication isn’t possible
- Oral Rx, focal tone management, bracing, neurosurgical procedures often used

Dysphagia in dystonia
- Dysphagia common in pediatric dystonia
- Even in patients with pure dystonia but GMFCS IV-V still can eat orally
- Usually a modified diet, i.e. pureed with thickened liquids
- Gastrostomy tubes still common, but usually some oral intake

Phenol/botulinum toxins
- Trials of phenol/botulinum toxins common
- Very goal specific, i.e. one upper extremity for joystick access, multiple injections in lower extremities for walking or cares
- Start low doses, multiple muscles, use EMG or U/S; E-stim may not work if posturing during injections
- Pain relief a very common indication
- Treatment intervals often shorter than spasticity
- Treatments less likely ongoing than in spasticity
Botulinum toxin to treat dystonia in CP

- Hemidystonic cerebral palsy
- GMFCS II
- Cognitively normal
- Bracing not helpful
- Oral Rx not helpful
- Poor orthopedic surgery candidate because of posturing
- Botulinum toxin R gastroc q 4 months

Orthopedic surgery in dystonia

- Less successful in pure dystonia than spasticity
- Posturing often continues limiting functional improvement
- If contractures present often return in a few years
- Hip dysplasia less common in pure dystonia

Hip Subluxation in CP

Hip Subluxation by GMFCS

Hip Subluxation and Spasticity

Hip subluxation in Dystonia
Hip dysplasia in Dystonia

Dystonia: Treatment

- Oral Pharmacotherapy:
  - Levodopa
  - Anticholinergics (trihexyphenidyl)
  - Baclofen
  - Tetrabenazine
  - Anticonvulsants (carbamazepine, pregabalin, levetiracetam, zonisamide)
  - Benzodiazepines
  - Tizandine
  - Anti-Psychotics

Dystonia: Treatment

- What dose the evidence look like?
Dystonia: Deep Brain Stimulation

- Technical challenges
  - Size
  - Anatomic placement
  - Complication avoidance
  - Stimulation settings
  - Ability to manipulate field shape in vivo
Dystonia: Deep Brain Stimulation

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Dystonia: Baclofen Pump

- GABA\textsubscript{B} agonist
- Intraventricular vs intrathecal with high cervical catheter
- More potency than oral
- Effective treatment for spasticity and dystonia

Dystonia: Rhizotomy

- Ventral Rhizotomy
  - Cut 80% of ventral (motor) roots intradurally
  - For dystonia
- Dorsal Rhizotomy
  - Cut 30% of dorsal roots intradurally
  - For spasticity

Dystonia: Selective Peripheral Neurectomy

- Simple
- Cheap
- Durable result
- Numbness, irreversible
Dystonia: Patient Cases
- Hyperkinetic: dystonia (focal), tardive

Autoimmune basal ganglia encephalopathy

Questions?