Movement Disorders in Pediatrics

Elizabeth Shaffer, MSN, APRN, FNP-C, CPN

Disclosures

- I have nothing to disclose with this presentation.

Objectives

- Define dystonia as a movement disorder
- Define spasticity as a disorder of tone
- Identify causes for both spasticity and dystonia
- Identify treatment options for both spasticity and dystonia

What is dystonia?

- Dystonia is a movement disorder
- Due to sustained or intermittent muscle contractions causing abnormal movements
- Results in movements that can are often disfiguring
- Dynamic in nature
- Can be painful
- Can lead to other orthopaedic complications and be associated with constipation and reflux.

Brain Involvement

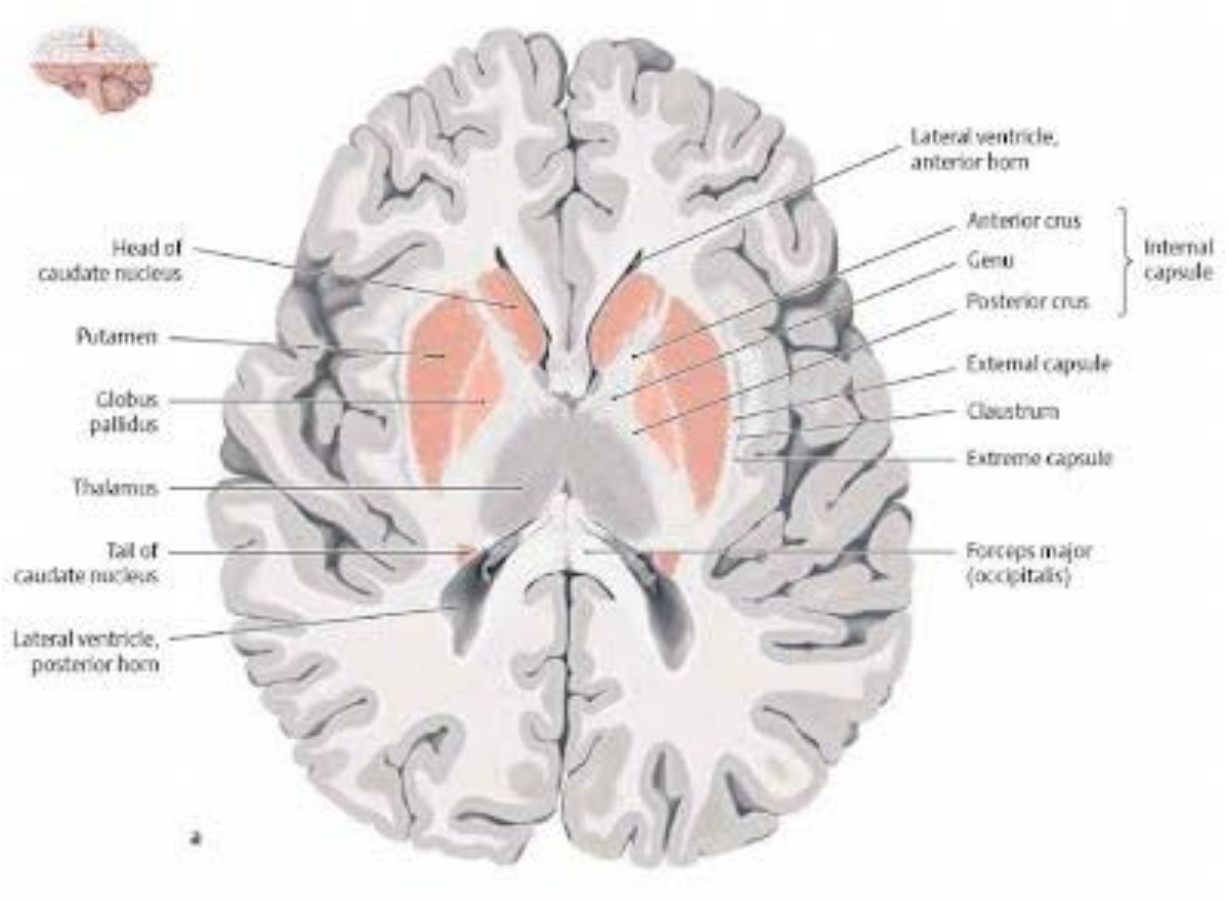


Image from University of Wisconsin (2011).

Causes of dystonia

- Inherited
- Acquired
- Idiopathic

Characteristics of Dystonia

- Age of onset
- Body Distribution
- Temporal Patterns
- Other symptoms with the dystonia

Dystonic Gait

- Dynamic
- Inversion or eversion of foot/feet
- Dragging of the foot/feet
- Posturing in the arms or hands
- Asymmetry in the shoulders
- Gait Dystonia - Case Study 24 Update - YouTube

Dopa-responsive Dystonia (DRD)

- Umbrella term to describe a group of clinically and genetically diverse disorders that respond to levodopa treatment
- Usually presents between early childhood and early adulthood
- Can begin simply with tripping and falling or leg cramps
- May progress to more generalized presentation
- Diagnosed through genetic testing, CSF neurotransmitters, or response to levodopa trial

Status Dystonicus

- Medical emergency
- Defined as “increasingly frequent and severe episodes of generalized dystonia” (Iodice & Pisani, 2019).
- Several different triggers
  - Fever, pain, and dehydration most common
- Managed with benzodiazepines
- Emergency department -> Hospital admission

Spasticity

- “Abnormal increase in muscle tone or stiffness of muscle” (NINDS, 2019).
- Velocity dependent and unidirectional
- Flexors in the arms and extensors in the legs
- Affects movement but is not a movement disorder
- Can be painful
- May be associated with deformity or disability
- Static

Characteristics seen with spasticity

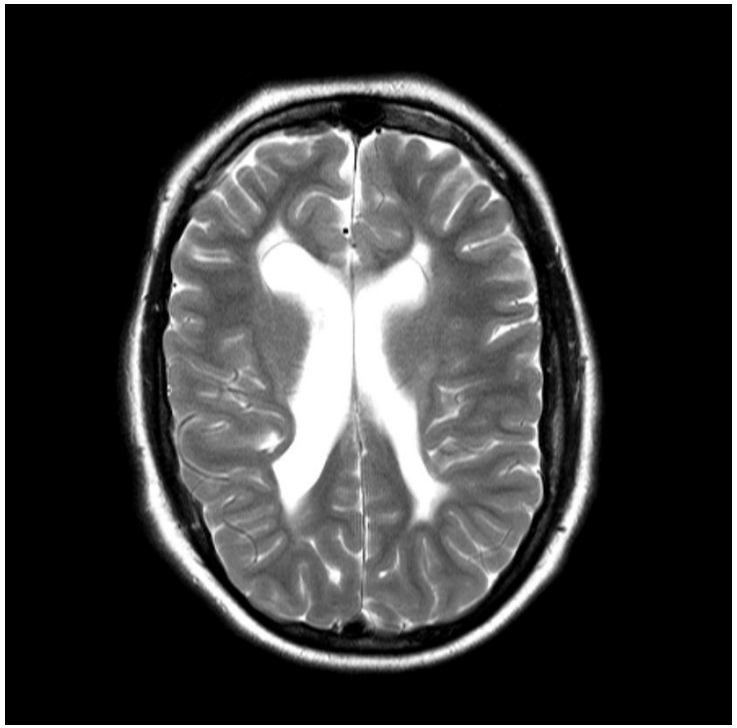
- Positive
  - Clonus
  - + Babinski
  - Hyperreflexia
- Negative
  - Weakness
  - Poor coordination
  - Loss of control

Singer et al, 2016

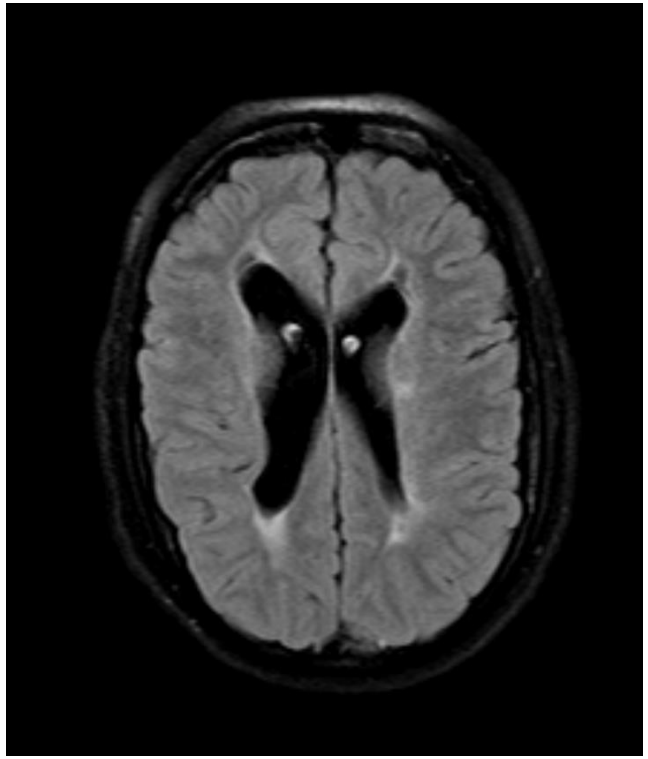
Causes of Spasticity

- Brain Malformations
- Injury to brain tissue
- Metabolic disorders
- Demyelinating disorders
- Other neurogenetic disorders

Periventricular Leukomalacia (PVL)



Axial T2



Axial FLAIR

Case courtesy of Assoc Prof Frank Gaillard, Radiopaedia.org, rID: 48619

Hereditary Spastic Paraplegia

- Most commonly presents in childhood
- Many different genes that have been identified to cause HSP
- Neurodegenerative in nature
- Weakness and spasticity in the legs
  - May also present with dystonia or rigidity
  - Reduced vibratory sense of proprioception in the legs
  - Hypertonic urinary bladder
- Pure/uncomplicated versus complex

Spastic Gait

- Walking pattern changes to overcome spasticity and weakness.
- Typically presents with shorter step length, wider step width, and slower step velocity (Singer et al, 2016).
- Assessment - Gait - Diplegic Gait - YouTube

Work-Up

- MRI Brain
- MRI Spine
- Laboratories
- Motion Analysis
- EMG

Treatment

- Oral medications
- Botulinum Toxin
- Surgical interventions
- Therapy (physical and occupational)

Carbidopa-levodopa (Sinemet)

- Levodopa -> dopamine in the central nervous system
- Trial levodopa as primary option for onset of dystonia over age 5
- Side effects
  - GI upset (nausea, vomiting, diarrhea), orthostatic hypotension, fatigue, insomnia
- Avoid abrupt withdrawal.
- Trial at least 3 months
- Start at 1mg/kg/day and increase to a goal of 4-5 mg/kg/day divided TID

Trihexyphenidyl (Artane)

- Anti-cholinergic
- Considered to be very effective in treating primary dystonia
- Data limited on use in treating dystonia related to cerebral palsy
- Side effects:
  - Sedation, dry mouth, constipation, urinary retention, blurred vision, irritability, decreased concentration and memory
- Dosing starts at 0.05-0.1mg/kg/day divided TID
- Avoid abrupt withdrawal

Baclofen

- Presynaptic GABA receptor agonist
- Helpful in treating spasticity and decreasing pain from dystonia
- Side effects:
  - **Sedation**, nausea, vomiting, dizziness, and headache
  - Withdrawal can occur if stopped abruptly – may present as itchiness, twitchiness, and severe irritability
  - Withdrawal can also precipitate psychosis and seizures
- Titrate up to desired dose – aiming for benefit without side effects
- Given 3-4 times per day

Other Medications

- Benzodiazepines
  - Clonazepam most common but others can be helpful
  - Can be helpful in scheduled dosing or used as a rescue medication for status dystonicus.
- Tizanidine
- Amantadine
- Gabapentin

Botulinum Toxin

- Effective for focal and segmental dystonia
- Also helpful in reducing spasticity
- Repeat treatment
- Effects wear off over time
- Not a great option for long-term management
- Side effects:
  - Soreness at the injection site, muscle weakness, fatigue, skin rash

Surgical Interventions

- Deep Brain Stimulation (DBS)
  - Surgical implantation of electrodes
  - Stimulates brain via programming to override abnormal signals
  - Specific inclusion criteria

Therapy

- Physical and/or Occupational therapy
  - Focus on mobilizing frozen joints
  - Limits contractures
  - Establish exercise programs
  - Implement usage of assistive devices
- Land or aquatic
- In the home, out of the home, and/or at school
- May help improve overall outcome (Hadders-Algra, 2014).

Case Study 1

- 3-year-old little girl
- 27-week twin
- 3-month NICU stay
- Initially presented to neurology for concerns of intermittent stiffening in her arms and legs R>L
- MRI showed PVL with resultant white matter volume loss and thinning of the corpus callosum

Case Study 1 - Video

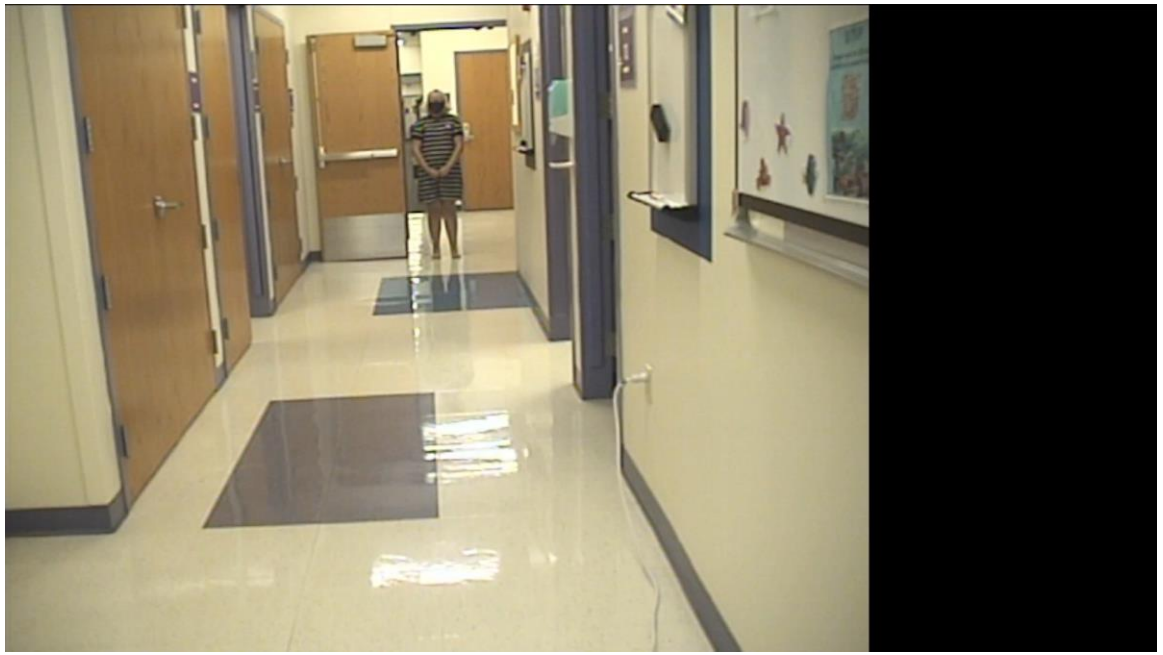
- How would you describe this gait pattern?
- Please type in the chat box words you might use when charting her gait.

Case Study 2

- 13-year-old young lady
- Congenital mirroring dystonia from mutation in *RAD51*
- Normal MRI of the brain
- Back pain and ankle pain
- Also reports pain with fine finger movements on exam
- Taking carbidopa/levodopa 1 tablet TID

Case Study 2 - Video

- How would you describe this gait pattern?
- Please type in the chat box words you might use when charting his gait.



Conclusion

- Spasticity
  - Stiff
  - Static
  - Not a movement disorder, but a disorder of tone
- Dystonia
  - Dynamic
  - Disfiguring
  - Movement disorder that impacts tone

References

Albanese, A., Bhatia, K., Bressman, S. B., Delong, M. R., Fahn, S., Fung, V. S., Hallett, M., Jankovic, J., Jinnah, H. A., Klein, C., Lang, A. E., Mink, J. W., & Teller, J. K. (2013). Phenomenology and classification of dystonia: a consensus update. *Movement disorders : official journal of the Movement Disorder Society*, *28*(7), 863–873. https://doi.org/10.1002/mds.25475

Anandan, C., & Jankovic, J. (2021). Botulinum Toxin in Movement Disorders: An Update. *Toxins*, *13*(1), 42. https://doi.org/10.3390/toxins13010042

Cloud, L. J., & Jinnah, H. A. (2010). Treatment strategies for dystonia. *Expert opinion on pharmacotherapy*, *11*(1), 5–15. https://doi.org/10.1517/14656560903426171

Di Muzio, B., Saber, M. (2021, May 30). *Periventricular leukomalacia.* Radiopedia. https://radiopaedia.org/articles/periventricular-leukomalacia?lang=us

Dystonia Medical Research Foundation. (2021). *Dopa responsive dystonia.* https://dystonia-foundation.org/what-is-dystonia/types-dystonia/dopa-responsive/#:~:text=Dopa-responsive%20dystonia%20%7C%20Dystonia%20Medical%20Research%20Foundation%20Dopa-responsive,synthetic%20form%20of%20a%20brain%20chemical%20called%20dopamine.

Fehlings, D., Brown, L., Harvey, A., Himmelmann, K., Lin, J.P., Mackintosh, A., Mink, J.W., Monbaliu, E., Rice, J., Silver, J., Switzer, L., & Walters, I. (2018). Pharmacological and neurosurgical interventions for managing dystonia in cerebral palsy: a systemic review. *Developmental Medicine & Child Neurology, 60*(4), 356-366. https://doi.org/10.1111/dmcn.12652

Hadders-Algra M. (2014). Early diagnosis and early intervention in cerebral palsy. *Frontiers in neurology*, *5*, 185. https://doi.org/10.3389/fneur.2014.00185

Iodice, A., & Pisani, F. (2019). Status dystonicus: management and prevention in children at high risk. *Acta bio-medica : Atenei Parmensis*, *90*(3), 207–212. https://doi.org/10.23750/abm.v90i3.7207

Medtronic. (2021, May). *Percept pc neurostimulator: Neurostimulator for deep brain stimulation therapy.* https://www.medtronic.com/us-en/healthcare-professionals/products/neurological/deep-brain-stimulation-systems/percept-pc.html

References

Michael, A.P. (2021). *Dystonia.* American Association of Neurological Surgeons. https://www.aans.org/en/Patients/Neurosurgical-Conditions-and-Treatments/Dystonia

Mission Health. (2019, June 6). *Gait dystonia – case study 24 update.* https://www.youtube.com/watch?v=rWKYDfklSkE

National Institute of Neurological Disorders and Stroke (NINDS). (2021, August 16). *Dystonias fact sheet*. https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Dystonias-Fact-Sheet.

National Institute of Neurological Disorders and Stroke (NINDS). (2019, March 27). *Spasticity information page.* https://www.ninds.nih.gov/Disorders/All-Disorders/Spasticity-Information-Page

Papadimitriou, I., Dalivigka, Z., Outsika, C., Scarmeas, N., & Pons, R. (2021). Dystonia assessment in children with cerebral palsy and periventricular leukomalacia. *European Journal of Paediatric Neurology, 32,* 8-15. https://doi.org/10.1016/j.ejpn.2021.03.005

ProHealth Continuing Education. (2013, Aug. 26). *Assessment – Gait – Diplegic Gait*. https://www.youtube.com/watch?v=Gwxjj0hZAMQ

Singer, H.S., Mink, J.W., Gilbert, D.L., & Jankovic, J. (2016). *Movement Disorders in Childhood: 2nd Edition*. Academic Press.

University of Wisconsin Department of Radiology. (2011). *Basal ganglia and thalamus.* https://sites.google.com/site/postgraduatetraining/anatomy/under-spin/slide-2?form=MY01SV&OCID=MY01SV

Questions?