Hereditary spastic paraplegia vs spastic diplegia

Full body gait analysis may improve diagnostic discrimination between hereditary spastic paraplegia and spastic diplegia: A preliminary study
Background:

Who has seen a patient with spastic diplegia (SD)?
Cerebral Palsy
- Spastic Diplegia

- Most common motor disability in children
- Lesion of the central nervous system in the developing brain
- About 1 in every 323 children is identified with CP
- 77.4% of children identified with CP have spastic CP
Background:

Who has seen a patient with hereditary spastic paraplegia?
HSP
- Hereditary Spastic Paraplegia

- About 1 in every 10 000 children is identified with HSP
- Many go undiagnosed or misdiagnosed
- A diverse and heterogeneous group of inherited neurodegenerative disorders
HSP
-Hereditary Spastic Paraplegia

– Primary symptom is slowly progressive weakness and spasticity of the muscles of the legs.
– HSP can begin at any age.
– Usually progressive, without remission.
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<th>Review:</th>
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<td><strong>HSP</strong></td>
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<td>– Gait?</td>
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Gait Comparison Summary from Journal Club

**Hereditary Spastic Paraplegia**
- increased trunk compensations (trendelenburg/lateral sway)
- mixed weakness with spasticity
- knee hyperextension
- foot drop
- internally rotated and flexed at knees
- tight adductors/spastic

**Spastic Diplegia**
- hip flexion and adduction
- crouch gait, scissoring, in toeing
- lack of heel strike/forefoot contact
- high guard
- ++spasticity
- upper extremity involvement
- increased lateral trunk flexion during gait
- increase lumbar lordosis/ant pelvic tilt
- increase pronation of feet
Previous Studies
Previous studies

- 3 previous studies looked at gait patterns of patients with SD and HSP
  - Cimolin et al 2007
  - Piccinini et al 2010
  - Wolf et al 2011
Previous studies results:

– Both demonstrated a cautious abnormal gait to attempt to achieve equilibrium and stability.
– HSP group showed greater knee flexion at initial contact and long lasting knee hyperextension in mid-stance compared to SD group.
– In SD group knee hyperextension is linked to an increase of PF/knee extension couple.
– In HSP patients knee hyper extension is not connected to ankle PF and could be due to a compensatory knee stabilisation strategy.
– In the HSP group knee hyperextension may be due to rectus femoris weakness/hypo-activation, as demonstrated by EMG pattern.
- Have you seen knee hyperextension in children with HSP?
- Do you think this could be as a compensatory strategy to provide more stability?
- Do you think it’s spasticity?
- Do you think it’s weakness?
- Do you think it’s important to understand in order to provide appropriate treatment?
Purpose and Aim of this study
CGA
-Clinical Gait Analysis

– Using CGA observe and compare **full body** movements
– Focus on trunk and upper extremity as these can represent compensatory strategies for the gait
Similarities

– Both walk with internal rotation, crouch and limited sagittal plane motion.
Differences

**HSP**
- Increased range angle and higher peak angular velocity of the spine in the sagittal plane during the swing phase for the HSP group.
- HSP group compensates for distal movement disorders by a significant and rapid spine tilt.

**SD**
- Less able to compensate with pelvis and thorax movement.
- SD showed a significantly greater peak elevation of the shoulders with a significant increase in elbow flexion compared to HSP patients. SD kept their arms flexed and outward to gain stability and balance during locomotion.
- Use movements of the upper limb to compensate for gait deviations and to fine tune balance control. Positioning arms in a high guard position.
Small Group Discussions
Why is it important to distinguish between HSP and SD?

– Its progressive nature
– Family planning and genetic counselling
– Predictability of orthopedic surgery
How have changes in gait affected the child’s function?

- Limits UE function
- Decreased gait speed
- Affects participation with peers
- Difficulty with rugged terrain and stairs
- Increased risk of falling
- Delayed milestones
What mobility aids have been successful?

- Posterior walkers
- 4WW
- Cane/forearm crutch
How have patients with HSP and SD responded to Botox? Baclofen? Surgery?

- Recovery process following surgery is very long & risk of deconditioning (less willing to try bony surgeries in patients with HSP).
- Botox has sometimes worked. Others said its beneficial at first however eventually it may not be enough to overcome the spasticity.
- The effect of BOTOX on muscle relaxation is temporary. It provides a better condition to strengthen the muscle and to obtain a better alignment, therefore is a very good base for rehabilitation.
- Baclofen use in Canada vs United States: In Canada it’s not commonly used in ambulatory children.
How have patients with HSP and SD responded to AFOs?

– Good as long as gait dictates a need
– Referral for gait analysis
– Devin’s story about her patient with HSP and how orthotics helped
Do you see progressive decline in mobility and gross motor skills?

- Yes
- Due to increase spasticity? Weakness?
- Sometimes due to increase age and weight
- Discussed progressive decline due to sedentary lifestyle (Beth’s story)
Summary

- Increased awareness of HSP
- Importance of recognizing the reason for the gait deviation in order to provide appropriate treatment
- Questions for future research?
  - In patients with HSP is hip monitoring important?
  - How can a patient’s symptoms be so different from others in their family that also have HSP?
In patients with HSP is hip monitoring important?
How can a patient’s symptoms be so different from others in their family that also have HSP?

– As noted above, the severity of symptoms and age of onset can vary widely, even within the same family. One reason is that HSP is a group of genetically different disorders, not a single disorder. Some differences may be due to genetic mutations. A child may show symptoms before a parent and it’s possible for some family members to have very mild symptoms while others have more severe symptoms. This may be due to other genes, environment, nutrition, general health, or factors not yet understood. In some families, symptoms tend to start at younger ages with each generation.

http://sp-foundation.org/understanding-pls-hsp/hsp.html
References:

- https://rarediseases.org/rare-diseases/hereditary-spastic-paraplegia/
- http://emedicine.medscape.com/article/306713-overview?pa=Ox%2Bzo2hNWLQ0qyWte3FuJxeCNqj%2FQioUdf1Ozzw6pmmGFsGLx22FFGeliI97lUgz9olhdAwN0Co%2BnEbbU6zovEiL5fM42L%2B9xlMlua7G1g%3D