Understanding and treating gait abnormality in Dravet syndrome

Anne Stratton, MD, FAAP, FAAPMR

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Disclosures

- I have no financial or personal disclosures relevant to any information in this talk

Objectives

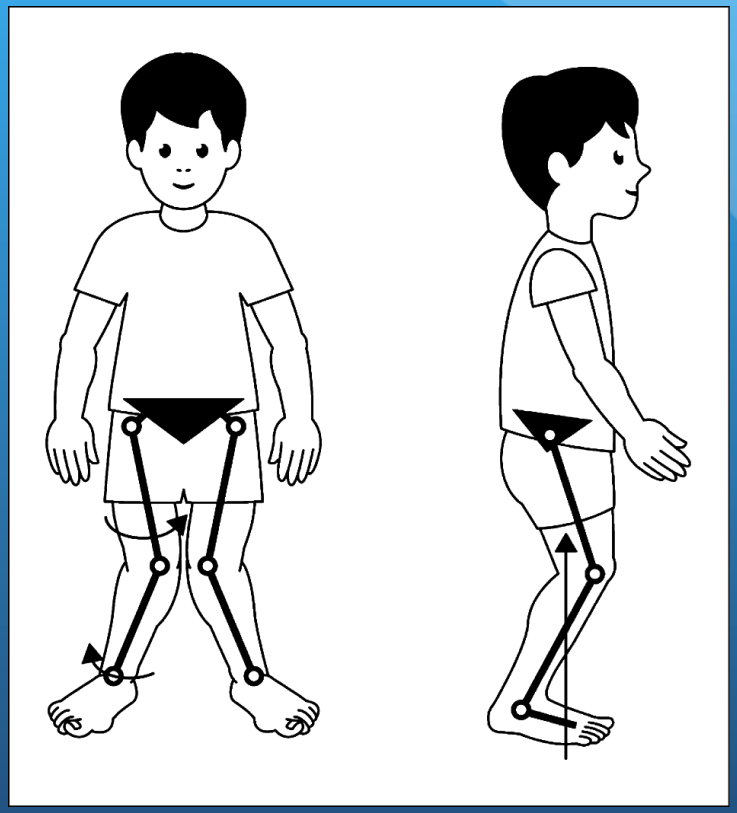
- Review the following factors associated with gait decline in Dravet syndrome
  - Characteristic gait abnormalities
  - Review physical changes
  - Review the timing of onset of gait changes
- Touch on the possible etiology of gait changes
- Discuss functional implications
- Discuss treatment options

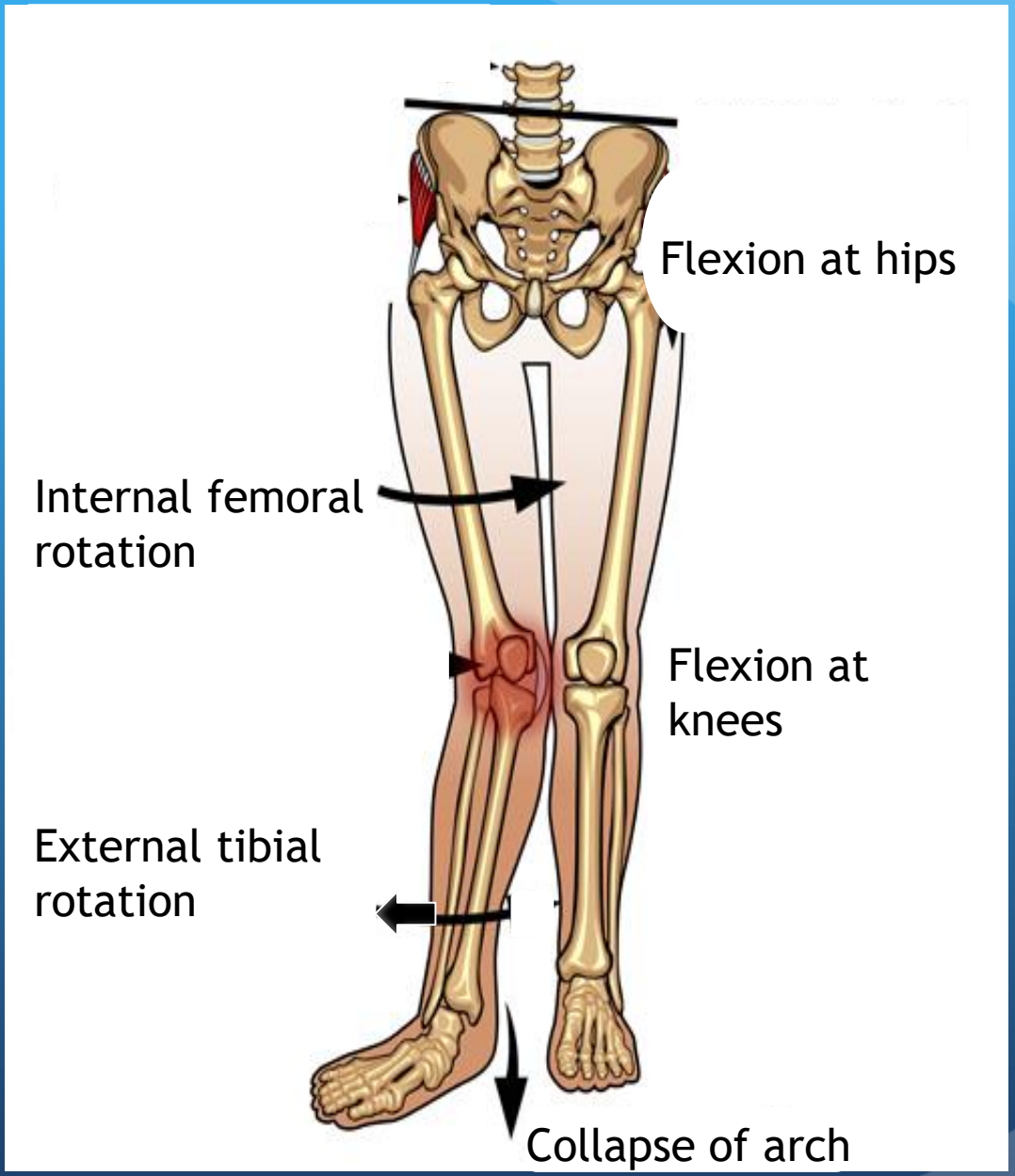
Characteristic gait abnormalities

- Ataxia
  - Impaired cerebellar function and joint proprioception
  - Impaired awareness of body position in space
  - Impaired balance: “drunken sailor”
- Crouch
  - Excessive hip, knee and ankle flexion
  - Inefficient pattern
  - “Sinking into the floor”
- Bradykinesia/ parkinsonism
  - Slowed movements
  - Decreased initiation
  - “Shuffling, freezing”
- Spasticity
  - Increased muscle tone, jerky, tight muscles

Physical changes

- Femoral anteversion
- Hip flexion
- Knee flexion
- Tibial lateral torsion
- Pes planovalgus
- Hip dysplasia
- Scoliosis




Flexion at hips
Internal femoral rotation
Flexion at knees
External tibial rotation
Collapse of arch

Timing of onset of changes

- 0-5 years:
  - Gait pattern: some variability, mostly within normal limits
  - Bony abnormalities: foot deformity develops, some hip internal rotation
- 6-12 years:
  - Gait pattern: some early crouch characteristics
  - Bony abnormalities: foot deformity, tibial torsion, scoliosis
- 13+ years:
  - Gait pattern: Crouch, possibly Parkinsonian
  - Bony abnormalities: foot deformity, tibial torsion, hip internal rotation/ femoral anteversion, scoliosis

Possible causes of gait decline

- Direct effects of SCN1A mutations
  - Sodium channel dysfunction
  - Anterior horn and peripheral motor nerve dysfunction
  - Cerebellar dysfunction
  - Basal ganglia dysfunction
- Secondary effects
  - Associated muscle weakness patterns
  - Orthopedic abnormalities change lever arms

Functional implications

- Crouch gait is inefficient
- Higher energy cost
- Stress on joints: knees
- Patients above 13 years have high use of assistance (walker/ wheelchair) for longer distance mobility

Treatment options

- Orthotics
- Physical therapy
- Spasticity treatments
- L-dopa
- Multi Level Orthopedic Surgery

Orthotics and Physical therapy

- Orthotics to support foot and ankle alignment
- May not be effective in correcting femoral alignment
- Can help compensate for weakness

- Physical therapy
- Strengthen extensors
- Work on balance
- Gait patterning
- Improve proprioception

Medication options

- Spasticity management
- Systemic: baclofen
- Focal: botulinum toxin injections

- Treatment for parkinsonian features
- Carbidopa-levodopa
- Trihexaphenadyl

Multilevel orthopedic surgery

- Psoas lengthening
- Hamstring lengthening
- Gastroc/soleus tendon shortening
- Femoral extension osteotomy
- Femoral rotational osteotomy
- Tibial rotation osteotomy
- Patellar advancement
- Correction of foot deformity

Summary

- Crouch gait pattern is common in adolescents and adults with Dravet syndrome
- Ataxia may be present but is not as prevalent
- Parkinsonism may also develop later in life
- Crouch may be due to a variety of factors including: nerve signaling abnormalities, weakness and boney malalignments
- Treatment options are mostly derived from CP literature and a combination approach is likely to be best.

References

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Thank you!

- Thank you to Dr. Kelly Knupp for helping put this conference together and inviting me to speak

Questions?