Understanding and treating gait abnormality in Dravet syndrome

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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


Thank you!

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Questions?