Motion Analysis in Understanding Gait Pathology in Charcot-Marie-Tooth Disease

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Disclosure of Relevant Financial Relationships
We have no financial relationships to disclose.

Disclosure of Off-Label and/or investigative uses:
We will not discuss off label use and/or investigational use in this presentation

Objectives

• Define Charcot-Marie-Tooth (CMT) disease – impairments and associated gait issues
• Describe gait findings for 3 common presentations of ankle function in CMT
• What we have learned
• New Frontiers - Impedance
Sources

- Literature
- Our experience in the assessment of 68 patients with CMT using comprehensive motion measurement techniques
- Őunpuu et al., Gait and Posture, 2013.

Charcot-Marie-Tooth (CMT)

- Most commonly inherited neurological disorder = de-myelination of large peripheral nerves
- Subtypes affect myelin or axon
- AKA: hereditary sensory and motor neuropathy
- Characterized by:
  - distal muscle weakness and imbalance
  - foot and ankle deformities
  - associated gait implications
  - impairment progression at varying rates

Textbook gait description:

- foot drop (excessive equinus) in swing phase
- steppage (hyperflexion of knee and hip in swing)
- circumduction and pelvic hiking in swing

(Fenton, JOPA 1994)
(Morrisy, Pediatric Orthopedics)
(Vinci, Archives of Physical Medicine & Rehab 2002)
Textbook clinical description:

- Forefoot equinus and adductus
- Hindfoot varus
- Pes cavus
- Toe deformities – claw toes

(Guyton; Foot and Ankle 2000)

Clinical experience shows variations in presentation...

Clinical experience:
- Not all persons with CMT have similar clinical presentations
- A variety of gait patterns and deformity

Our Questions:
- What are the typical gait patterns?
- How do they relate to clinical findings (impairments)?
Our Patient Population

- Population (N=33)
  - 16 males/17 females
  - Age, 12 ± 4 years
  - Height, 1486 ± 145 mm
  - Mass, 40.9 ± 10.4 kg

Patient Demographics

- Onset of walking
  - 24 of the 33 patients were within the typical limits (<15 months)
  - 8 patients walked within 24 months
  - 1 patient was unknown
- Mean age of diagnosis was 8 ± 5 years (range 1 to 16 years)

Prerequisites of Typical Gait

- Stance phase stability
- Swing phase clearance
- Appropriate prepositioning at initial contact
- Adequate step length
- Energy conservation

• Stance phase stability
• Appropriate prepositioning at initial contact
• Swing phase clearance

• Stance phase stability
• Appropriate prepositioning at initial contact
• Pain

• Stance phase stability
• Appropriate prepositioning at initial contact
• Swing phase clearance
Clinical Examination

• The most common impairments at the ankle
  – reduced passive dorsiflexion ROM
    • mean 1 ± 7 deg (knee extended)
    • mean 7 ± 9 deg (knee flexed)
  – plantar flexor weakness
    • median 3, minimum 2 and maximum 5
  – dorsiflexor, forefoot invertors and evertors weakness
    • median 5, minimum 0 and maximum 5
  – cavus 54/66 sides

• Weak plantar flexors were consistent with the functional findings reported by many patients
  – difficulty in running
  – inability to toe walk
  – likely to result in the first functional complaint by a child with onset of CMT functional findings
• Weak plantar flexors = delayed heel rise
• Delayed peak dorsiflexion may be the first kinematic “walking gait sign” of CMT

Variation in Impairments

• Not all patients have the same impairments
• Therefore they walk differently
• Experience shows very distinct patterns of pathology that cannot be fully explained by disease progression or penetration alone
Three Subgroups

- Based on peak ankle dorsiflexion in terminal stance in reference to typically developing data:
  - $<\text{TPDF}$ n=13 sides – Group 1 (toe walkers)
  - $=\text{TPDF}$ n=30 sides – Group 2 (cavo-varus foot)
  - $>\text{TPDF}$ n=23 sides – Group 3 (flail foot)
- Asymmetry in presentation, 13 patients sides in different groups

Focus on Ankle Impairments

- Three patterns of ankle/foot function in CMT:
  - Toe walker with cavus (Group 1: n=13)
  - Combination of cavus and +/- plantar flexor tightness results in limited passive dorsiflexion range of motion
  - Weakness plantar flexors

Group 1 – toe walkers

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Typical Mean(SD)</th>
<th>CMT Grouped by Peak DF in Stance Mean(SD)</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Passive Dorsiflexion Range of Motion (°)</td>
<td>20 (2)</td>
<td>16 (3)</td>
<td>13 (1)</td>
</tr>
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<td>20 (3)</td>
<td>16 (2)</td>
<td>13 (1)</td>
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<tr>
<td>Plantar Flexion Strength</td>
<td>5/5/5</td>
<td>4/5/5</td>
<td>2/5/2</td>
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<tr>
<td>Dorsiflexion Strength (Knee @ 0°)</td>
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<td>4/5/2</td>
</tr>
<tr>
<td>Walking Speed (cm/sec)</td>
<td>126.9(10.5)</td>
<td>117.1(20.5)</td>
<td>111.1(15.8)</td>
</tr>
<tr>
<td>Peak Ankle Dorsiflexion in Stance (°)</td>
<td>13(3)</td>
<td>6(5)</td>
<td>14(2)</td>
</tr>
<tr>
<td>Peak Ankle Dorsiflexion in Middle Third of Swing (°)</td>
<td>3(4)</td>
<td>-8(6)</td>
<td>-1(5)</td>
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<tr>
<td>Peak Ankle Power Generation in Stance (W/kg)</td>
<td>3.67(0.99)</td>
<td>2.00(0.56)</td>
<td>2.20(0.72)</td>
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<tr>
<td>Ankle Angle at Initial Contact (°)</td>
<td>-2(5)</td>
<td>-11(6)</td>
<td>-5(7)</td>
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</table>
Focus on Ankle Impairments

- Three patterns of ankle/foot function in CMT
  - Equino-varus (Group 2: n=30)
  - Cavus, adductus and varus hindfoot, limited passive ankle dorsiflexion
  - Weakness
  - Pain

Group 2 – cavovarus foot

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Typical mean (n=42)</th>
<th>CMT grouped by peak dorsiflexion</th>
<th>Group 2 mean (n=13)</th>
<th>Typical mean (n=30)</th>
<th>Excessive mean (n=23)</th>
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<tr>
<td>Forefoot Eversion Strength</td>
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<td>5/5/3</td>
<td>4/5/2</td>
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<td>Walking Speed</td>
<td>126.9(10.5)</td>
<td>117.1(20.5)</td>
<td>111.1(15.8)</td>
<td>103.2(21.1)</td>
<td>&lt;0.001</td>
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<td>Peak Ankle Dorsiflexion in Stance</td>
<td>13(3)</td>
<td>6(5)</td>
<td>14(2)</td>
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<td>Peak Ankle Dorsiflexion in Middle Third of Swing</td>
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<td>Ankle Angle at Initial Contact</td>
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<td>-5(7)</td>
<td>-6(9)</td>
<td>&lt;0.001</td>
<td></td>
</tr>
</tbody>
</table>

Focus on Ankle Impairments

- Three patterns of ankle/foot function in CMT
  - Flail foot and ankle (Group 3: n=23)
  - Cavus which can lead to limited passive ankle dorsiflexion
  - Weakness substantial
  - Pain
<table>
<thead>
<tr>
<th>Group 3 – flail foot</th>
<th>Typical Mean(SD)</th>
<th>Reduced Mean(SD)</th>
<th>p</th>
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<td>5 (5/4)</td>
<td>0.03</td>
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</table>

| Walking Speed (cm/sec) | 126.9 (10.5) | 117.1 (20.5) | 0.001 |
| Walking Speed (cm/sec) | 111.1 (15.8) | 103.2 (21.1) | 0.001 |

| Peak Ankle Dorsiflexion in Stance | 13 (3) | 6 (5) | 0.01 |
| Peak Ankle Dorsiflexion in Stance | 13 (3) | 6 (5) | 0.01 |
| Peak Ankle Dorsiflexion in Stance | 14 (3) | 6 (5) | 0.01 |

**Impairment vs. Gait**

- Plantar flexor weakness
  - Common problem
  - Delayed heel rise – first sign
  - Increased peak ankle dorsiflexion in terminal stance – as problem gets more significant

- Limited passive dorsiflexion range of motion (standard clinical exam measurement)
  - Common problem
  - Masks ankle plantar flexor weakness
  - Is impacted by the severity of cavus
Impairment vs. Gait

- Weakness of plantar flexors
- Cavus
- Limited passive ankle dorsiflexion
- ALL of varying severity

Impact:
- Stance phase stability
- Swing phase clearance
- Appropriate prepositioning for initial contact

Associated Kinematic/Kinetic Findings

- Delayed peak dorsiflexion in terminal stance (59/66 sides)
- Increased peak dorsiflexion in terminal stance (23/66 sides)
- Reduced peak ankle plantar flexor moments and powers in terminal stance in all three groups as compared to the control group
Associated Kinematic/Kinetic Findings

- delayed peak dorsiflexion in terminal stance (59/66 sides)
- Increased peak dorsiflexion in terminal stance (23/66 sides)
- Reduced peak ankle plantar flexor moments and powers in terminal stance in all three groups as compared to the control group

Impairment vs. Gait

- Dorsiflexor weakness
  - Less common problem
  - Increased plantar flexion in swing and initial contact
  - Impacted by limited passive ankle dorsiflexion range
  - Compensatory increased hip flexion in swing
Impairment vs. Gait

- Dorsiflexor weakness
  - Impact:
    - Swing phase clearance
    - Appropriate prepositioning at initial contact

Associated Kinematic/Kinetic Findings

- Increased plantar flexion in swing and initial contact
- Absence of dorsiflexor moment in loading response

Group 1 – dashed/dot
Group 2 – dashed
Group 3 - solid
Knee-Ankle Sagittal Plane Kinematics and Kinetics

- less than typical (dash-dot)
- typical (large dash)
- greater than typical (solid)

Increased Equinus in Swing

- patients with excessive equinus in swing showed increased peak hip flexion in swing to aid in clearance

Conclusions

- Patients with CMT present differently from typically developing and within the diagnosis
- Therefore, TREATMENT needs to be SPECIFIC to the INDIVIDUAL patient
- Motion analysis is a means of better understanding ankle function for treatment decision-making
Important Measurements

- Plantar flexor and dorsiflexor strength
  - Muscle strength testing
  - Ability to toe and heel walk
  - Ankle sagittal plane kinematics (peak dorsiflexion degree and timing in terminal stance)
  - Ankle kinetics (peak moment and power in terminal stance)

Important Measurements

- Plantar flexor range of motion
  - Passive range of motion
  - Peak dorsiflexion in terminal stance (net outcome)
- Degree of cavus
  - Visual/clinical assessment
  - X-ray
  - Assessment of plantar fascia tightness

Other

- Family history
- Symptoms first noted by patient and decline
- Pain/injury – foot and ankle
- Asymmetry between lower extremity function
- Sensation and vibration limitations
- Hand coordination and strength
- Balance
- Hip dysplasia
Case Studies

- Sibling comparison
- Parent vs. child comparison
- Bracing outcomes
- Surgical outcomes

Mother - Daughter

- Background
  - Mother and daughter CMT type IIA
  - Mother was 14.5 years and daughter was 6.3 years at time motion analysis
  - Both walked at 12 months with no reported gait problems
- Referral for motion analysis
  - Mother for surgical decision making to correct bilateral toe walking
  - Daughter for surgical decision-making to correct foot and ankle deformity for successful bracing

Clinical Exam Measures

<table>
<thead>
<tr>
<th>Clinical Exam Measure</th>
<th>Mother</th>
<th>Daughter</th>
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<tbody>
<tr>
<td>Dorsiflexion knee 0°</td>
<td>-10</td>
<td>-10</td>
<td>10</td>
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<tr>
<td>Dorsiflexion knee 90°</td>
<td>-10</td>
<td>-10</td>
<td>15</td>
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<tr>
<td>Plantar flexor strength</td>
<td>4/5</td>
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<td>2/5</td>
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<td>Dorsiflexor strength</td>
<td>2/5</td>
<td>2/5</td>
<td>3/5</td>
</tr>
<tr>
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<td>4/5</td>
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<tr>
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<td>2/5</td>
<td>2/5</td>
<td>3/5</td>
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<tr>
<td>Heel bisector</td>
<td>4th toe</td>
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<tr>
<td>Plantar fascia</td>
<td>tight</td>
<td>tight</td>
<td>tight</td>
</tr>
</tbody>
</table>
Mother vs. Daughter - ankle

- **Mother:**
  - Toe walker
  - Increased kinematic ankle plantar flexion in stance and swing

- **Daughter:**
  - Flail foot
  - Increased peak ankle dorsiflexion in stance and plantar flexion in swing

Mother vs. Daughter - knee

- **Mother:**
  - Increased plantar flexion knee extension couple with knee flexor moment pattern

- **Daughter:**
  - Increased flexion in loading with associated knee extensor moment
Summary

- Gait analysis provides a unique opportunity to compare gait patterns between a parent and child completed during different decades
- There can be substantial variation in the presentation and function over two generations
- Predicting prognosis for a child’s deformity based upon a parent’s presentation is not possible

Next Step

- Objective evaluation of treatment outcomes
- Long-term assessments of both parent and child would provide an opportunity to better understand differences in rate of disease progression in terms of gait deterioration and long-term prognosis for gait function

Surgical Outcomes – Group 1

- Background
  - 14.5 year old female with CMT type IIA
  - Walked at 12 months with no reported gait issues
  - Slow and progressive onset of toe walking was reported
  - No previous surgical intervention
- Referral
  - Surgical decision making to correct bilateral toe walking
Pre - Surgery

Stance stability. Prepositioning at initial contact. Swing clearance.

Clinical Exam – pre

<table>
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<tr>
<th></th>
<th>2002</th>
<th>2014</th>
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<tbody>
<tr>
<td></td>
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</tbody>
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Pre Op – Knee and Ankle

- Increased plantar flexion in stance and swing
- Associated increased plantar flexion-knee extension couple
- Reduced ankle power generation
Surgical Decision

• Bilateral plantar fascia releases/short leg casting - indications:
  – Severe plantar fascia tightness
  – Plantar flexor contracture [plantar aspect of the foot (not calcaneus) versus shank]
  – Excessive equinus throughout the gait cycle (indicating reduced dorsiflexion range available)

• Avoid gastrocnemius lengthening due to:
  – Less than typical ankle power generation in terminal stance (indicating plantar flexor weakness)
  – Progressive disease and risk of increasing ankle plantar flexor weakness over time

Surgical Outcomes (long-term)

Pre - 2002               Post - 2014
Relaxed Standing
Clinical Exam – pre vs. post

<table>
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<tr>
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<th>2002</th>
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<tr>
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</tbody>
</table>

Pre vs. Post - ankle

- Pre vs. post:
  - Decreased excessive plantar flexion in stance and swing
  - Attains heel contact with associated dorsiflexor moment in stance
  - Peak ankle power generation in terminal stance not changed

Pre vs. Post - knee

- Pre vs. post:
  - Reduced excessive plantar flexion knee extension couple
  - Reduced knee extension and flexor moment in stance
Summary

• Gait analysis provided opportunity to evaluate long-term surgical outcome objectively
• Plantar fascia release is adequate to reduce increased plantar flexion in stance/swing with associated benefits in prerequisites of gait
• There was no reduction in ankle power generation over time although clinical exam suggested reduced ankle plantar flexor strength

Surgical Outcomes – Group 2

• Surgical decision making specifics for the cavo-varus foot using the traditional foot model is limited
• Still valuable information for understanding initial conditions and outcomes for:
  – Peak ankle dorsiflexion in stance
  – Peak ankle kinetics
  – Foot pressure distribution

Surgical Outcomes – Group 2

• Background:
  – 13 year old male with progressive weakness of ankle musculature
  – Significant cavovarus with supination position of the foot in stance, issues with brace wear
• Surgical intervention:
  – posterior tibialis tendon lengthening
  – closing wedge osteotomies of the midfoot including the 3 cuneiforms and cuboid
Surgical Outcomes – Group 2

- Foot pressure outcomes 1 year post surgery
- Reduced pressure under 5th meta head

Sx Outcomes – Group 2

- One year post surgery evaluation
- No change in sagittal ankle kinematics and kinetics

Surgical Outcomes – Group 2

- Background
  - 7-10 year old female
  - Walking at one year of age
  - Diagnosed at age 7
  - Issues with tripping and falling
- Referral
  - Baseline evaluation – document change over time
• Clinical findings:
  • Bilateral plantar flexor weakness (2/5)
  • Bilateral claw toes
  • Bilateral cavus
  • Bilateral normal passive ROM

• Surgical Intervention:
  – Bilateral anterior tibialis tendon transfers
  – Bilateral posterior tibialis tendon lengthenings
  – Bilateral calaneal osteotomies
  • Bilateral solid AFO’s

Post Surgery and 4 Years
Summary

• Gait analysis outcomes can help understand the impact of surgery and time on a progressive disease

Treatment Outcomes – Group 3

• Goals:
  – Provide ankle/foot support with bracing
  – May need surgical intervention to allow bracing

• Brace type:
  – Support for ankle plantar flexor and ankle dorsiflexor weakness as well as medial and lateral stability is generally needed
  – Patient weight and strength are factors in how a brace functions
Barefoot vs. Hinged AFO

- Hinged AFO:
  - Provides support for ankle dorsiflexor weakness
  - Allows continued excessive/delayed ankle dorsiflexion in terminal stance
  - No impact on peak ankle plantar flexor moment

Note: hinged AFO has plantar flexion stop.

Barefoot vs. PLS AFO

- PLS posterior leaf spring
  - Provides support for ankle dorsiflexor weakness
  - Allows continued excessive/delayed ankle dorsiflexion in terminal stance
  - No increase in peak ankle plantar flexor moment
  - Allows ankle power generation

Barefoot vs. Solid AFO

- Solid AFO:
  - Provides support for ankle dorsiflexor weakness
  - Allows continued excessive/delayed ankle dorsiflexion in terminal stance
  - Allows minimal increase in peak ankle plantar flexor moment
  - Allows ankle power generation

GCMAS at AACPDM, October 21, 2015  Lecture #2: Motion Analysis and CMT
Future?

• Diagnosis for CMT often delayed
• Multiple reasons
  – Confused with other diagnoses
  – No family history
  – Denial
  – Slow onset
• Easy diagnostic biomarker would be helpful

Electrical Impedance Myography (EIM)

• Painless and non-invasive procedure
• Measures the body’s opposition to an electric current
• Has been shown to detect non-mass-dependent muscle maturation in healthy children and no change in patients with spinal muscular atrophy over time (Rutkove, et al., 2012)

• Due to its high water content, muscle tissue tends to have greater conductivity than other types of tissue.
• Hypothesis:
  – patients with less muscle mass through atrophy or degeneration would have a higher impedance measurement, or more resistance, over that muscle bulk
Methods

• Three impedance (Z) measurements taken at one minute intervals

Preliminary Results


• EIM, a painless and non-invasive procedure, may be a candidate biomarker for the preliminary identification of CMT.
• Further data collection and analysis is required!
  – CMT
  – Other dx groups
  – Controls
Conclusions

- CMT is a complex progressive disease that presents with variations in impairment and therefore a variety of gait issues
- Therefore, treatment needs to be patient specific
- Motion measurement techniques have helped us better understand the pathomechanics of this disease and allowed us to critically evaluate treatment outcomes

Thank You