Facial Weakness and Impact on Communication and Swallowing in FSHD

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My Objective

• To evaluate a quantitative measure of facial weakness
• To determine the impact of FSHD on communication and swallowing difficulty.
• To determine how our quantitative measures of facial weakness compare to other aspects of FSHD (duration, severity, communication, swallowing difficulty, and physical function)
Background

- FSHD is one of the most common muscular dystrophies
- 1 in 8,333 to 1 in 20,000
  - About 21,000 cases in USA
- Progression: face/scapular muscles, upper arms, lower legs/abdominal

Facioscapulohumeral Muscular Dystrophy

- There is considerable variability between individuals but weakness is progressive over time
  - About 20% of individuals over the age of 50 will require a wheelchair
- Prior studies have shown a consistent loss of strength using manual muscle testing of quantitative myometry of about 3-5% per year
  - But there are no validated measures facial weakness
- Only limited studies regarding the impact of facial weakness on communication or swallowing

Methods

• Prospective, cross-sectional observational study
• Performed at two sites (KUMC/University of Utah)
• Inclusion criteria
  ◦ > 18 years
  ◦ Genetic confirmation (or immediate relative)
  ◦ Clinically affected
  ◦ Could complete study procedure
• Single-visit
Methods

• IOPI (Iowa Oral Performance Instrument)
  ◦ Quantifies lip, tongue, and cheek pressure

• SWAL-QOL
  ◦ Patient-reported swallowing difficulty

• CPIB (Communicative Participation Item Bank)
  ◦ Patient-reported communication questionnaire
Methods: IOPI

- Lip, tongue, and cheek strength
- Displacement of air pressure
- Three trials, maximum

(http://iopimedical.com/)
Methods: SWAL-QOL

• Questionnaire to determine quality of life as affected by swallowing difficulties

• 10 categories, scored as a percentage of normal

  ◦ (100% = healthy)

  ◦ Categories include: food selection; burden; mental health; social functioning; fear; eating duration; eating desire; communication; sleep; and fatigue

Methods: Communicative Participation
Item Bank – short version

• 10-item questionnaire
  ◦ Asks whether condition interferes with communication
  ◦ Question: “Does your condition interfere with talking with people you know?”
  ◦ Scored from 0=‘very affected’ to 3=‘not at all’

• Summary score out of 30

Patient Demographics

<table>
<thead>
<tr>
<th></th>
<th>Kansas</th>
<th>Utah</th>
<th>Total (Range)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Mean Age</strong></td>
<td>55.37</td>
<td>50.76</td>
<td>52.75 (23-83)</td>
</tr>
<tr>
<td><strong>Range</strong></td>
<td>23-83</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Gender (% female)</strong></td>
<td>47.37%</td>
<td>44.00%</td>
<td>45.45%</td>
</tr>
<tr>
<td><strong>Mean Disease Duration (symptom onset)</strong></td>
<td>26.89</td>
<td>32.31</td>
<td>29.92</td>
</tr>
<tr>
<td><strong>Range</strong></td>
<td>5-64</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Mean Clinical Severity Score (non age-adjusted)</strong></td>
<td>6.5</td>
<td>5.64</td>
<td>6</td>
</tr>
<tr>
<td><strong>Range</strong></td>
<td>2-9</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>FSHD Clinical Score</strong></td>
<td>9.05556</td>
<td>7.64</td>
<td>8.232558</td>
</tr>
<tr>
<td><strong>Range</strong></td>
<td>2-13</td>
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IOPI

Men IOPI Values

Women IOPI Values

Iowa Oral Pressure Instrument

**Lip Strength vs CSS**
- \( y = 0.0049x + 5.9303 \)
- \( R^2 = 0.0008 \)

**Buccal Strength vs CSS**
- \( y = -0.124x + 7.9237 \)
- \( R^2 = 0.1784 \)
Lip Strength vs Disease Duration

\[ y = -0.0437x + 16.214 \]

\[ R^2 = 0.0025 \]

Buccal Strength vs Disease Duration

\[ y = -0.2435x + 23.012 \]

\[ R^2 = 0.2444 \]
<table>
<thead>
<tr>
<th></th>
<th>Mean</th>
<th>STD</th>
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<tbody>
<tr>
<td>Burden</td>
<td>91%</td>
<td>14%</td>
</tr>
<tr>
<td>Eating Duration</td>
<td>86%</td>
<td>19%</td>
</tr>
<tr>
<td>Eating Desire</td>
<td>92%</td>
<td>12%</td>
</tr>
<tr>
<td>Food Selection</td>
<td>91%</td>
<td>15%</td>
</tr>
<tr>
<td>Communication</td>
<td>91%</td>
<td>13%</td>
</tr>
<tr>
<td>Fear</td>
<td>89%</td>
<td>14%</td>
</tr>
<tr>
<td>Mental Health</td>
<td>91%</td>
<td>17%</td>
</tr>
<tr>
<td>Social</td>
<td>93%</td>
<td>15%</td>
</tr>
<tr>
<td>Sleep</td>
<td>65%</td>
<td>22%</td>
</tr>
<tr>
<td>Fatigue</td>
<td>55%</td>
<td>20%</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>86%</td>
<td>12%</td>
</tr>
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- While the overall score is not that low, about 1/3 of people have a total SWAL-QOL score below 80%
SWAL-QOL

Lip Strength vs SWAL-QOL

\[ y = 0.0002x + 0.8564 \]

\[ R^2 = 0.0003 \]

Buccal Strength vs SWAL-QOL

\[ y = 0.0054x + 0.7743 \]

\[ R^2 = 0.113 \]
CPIB

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<tr>
<td><strong>Mean</strong></td>
<td>24.84</td>
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<tr>
<td><strong>Standard Deviation</strong></td>
<td>5.93</td>
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<tr>
<td><strong>N</strong></td>
<td>44</td>
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- Ceiling affect with 18 reporting score of 30
- But the range as low as 12
- There was a positive correlation between the SWAL-QOL and the CPIB, with a $r=0.61$
CPIB

**Lip Strength vs CPIB Score**

\[ y = 0.0154x + 24.725 \]

\[ R^2 = 0.0011 \]

**Buccal Strength vs CPIB Score**

\[ y = 0.3093x + 20.106 \]

\[ R^2 = 0.1433 \]
Conclusion

• The IOPI offers an easy to perform standard assessment face and tongue weakness for FSHD
  ◦ Includes standard protocols and normative values
  ◦ Both lip strength and buccal strength were reduced in FSHD

• Buccal strength as a better indicator of disease state
  ◦ With modest associations to overall severity and disease duration
  ◦ Modest associations with communication difficulties or swallowing
  ◦ May be good measure to follow over time

• While most FSHD patients score ~normal for swallow and communication; a smaller group, around 1/3 may have difficulties
Implications

• What does it mean?

• Focus on buccal strength and buccal movement
  ◦ We often think of measuring changes in orbicularis oris strength, and while this is affected, it may be static
  ◦ Buccal strength is combination of muscles (orbicularis oris, buccinator, etc) which together may show some progression over time

• Follow-up studies needed
  ◦ Other factors beside lip or buccal strength which may affect communication or swallow
  ◦ Other aspects of lip weakness not explored here: e.g. the social impact of not having facial expression understood
Thank You

• The research team at KU
• The research team at University of Utah
• Karlien Mul, a summer research fellow at KU working in FSHD
• The KU summer learning experience
• The patient and family members with FSHD who participated in our study
• CTSA multi-institute pilot funding