Dear Physicians,

These forms will help us determine the clinical features of patients with phenotypic FSHD relative to patients with FSHD with the classical deletion at D4Z4. The questions are self-explanatory. Please email Dr. Tawil at Rabi_Tawil@urmc.rochester.edu with any questions regarding the content of these forms. Thank you for your assistance.

For University of Rochester Medical Center Use Only:

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<table>
<thead>
<tr>
<th>Form No:</th>
<th>Center #:</th>
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<th>Date: (mmddyyyy)</th>
<th>NMD#</th>
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<tr>
<td>8</td>
<td>4b</td>
<td>0 1</td>
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</tbody>
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Part I: Physician's Clinical Examination - to be completed by EXAMINING PHYSICIAN

A. Estimated age of onset of FSHD symptoms: [ ] (years of age)

- Enter 00 if from birth through first year
- Enter other ages to the closest multiple of 5 years (5, 10, 15 etc)
- Enter 99 if asymptomatic
- Enter XX for unknown value

B. Patient Reported Initial Symptoms/Signs

Ask the patient “What was (were) your first symptom(s) of FSHD?” Subject must provide answer without prompting from evaluator. **Check all that apply.**

1. Facial involvement
2. Proximal upper extremity involvement
3. Distal upper extremity involvement
4. Proximal lower extremity involvement
5. Distal lower extremity involvement
6. Someone other than patient noticed problem
7. Patient is without signs or symptoms

C. Family History of FSHD

1. No affected family members
2. Affected family members (specify)
3. Unknown

If Yes: [ ]

- Mother
- Father
- Siblings
- Other

Specify

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D. FSHD-Specific Physical Exam

1. (purposely left blank)

2. Facial Function:
   - Frontalis - hold forehead and have patient try to wrinkle forehead
   - Eyelid closure
   - Smile
   - Pucker - have patient mimic a kiss
   - Platysma - ask patient to contract the anterior neck muscles ("growl")

<table>
<thead>
<tr>
<th>Scoring for Items 2 through 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>U  N = Unaffected</td>
</tr>
<tr>
<td>A  F = Affected</td>
</tr>
<tr>
<td>U  C = Uncertain</td>
</tr>
</tbody>
</table>

3. Scapular Stabilizer Weakness
   Is there evidence of scapular winging?

4. Ankle Dorsiflexor Weakness
   Can they walk on their heels?

5. MMT: Manual Muscle Testing (MRC 0-5)

<table>
<thead>
<tr>
<th>Scoring for Items 2 through 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>Shoulder Abduction - Range of Motion (in °)</td>
</tr>
<tr>
<td>R  L = Affected</td>
</tr>
<tr>
<td>180 degrees</td>
</tr>
<tr>
<td>90 degrees</td>
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D. FSHD-Specific Physical Exam (continued)

6. Beevor's Sign (Movement of the umbilicus on asymmetric contraction of the abdominal muscles).
   - Place patient supine without a pillow.
   - Ask patient to lift head off the table.
   - Measure movement of umbilicus. Record "Absent" if none.
   - Present Absent

7. Ambulatory
   - Yes No
   - Ambulatory is defined as ability to walk independently. (Use of AFOs, walkers and canes is acceptable)

8. Clinical Severity Score (CSS) for FSHD

   Scoring Instructions: In FSHD, the typical sequence of disease progression over time is as follows: shoulders/upper arms - then foot dorsiflexion – then hip girdle and proximal leg muscles. However, degree of weakness does not necessarily reflect this time sequence. To obtain the Clinical Severity Score, score patients according to where they fall on that “time” sequence rather than on severity of regional involvement. For example: a patient with mild shoulder weakness and moderate pelvic girdle weakness would receive a score of 7 rather than 2.

   - No signs of muscle weakness.
   - Facial weakness only.
   - Mild scapular weakness. No limitations of abduction or elevation. Often asymptomatic.
   - Moderate scapular weakness. Arm abduction > 60 degrees and strength ≥ 3 in arm muscles. No lower limb involvement. Usually symptomatic.
   - Severe scapular weakness. Arm abduction < 60 degrees on at least one side. Strength <3 in at least one muscular district of the arms. No lower limb involvement. Abdominal muscles may be weak.
   - Foot extensor weakness. No pelvic girdle weakness.
   - Mild weakness of pelvic and proximal leg muscles (strength ≥ 4 in all muscles). Stands up from a chair without support.
   - Moderate pelvic girdle and proximal leg weakness (strength >3 in all muscles). Stands up from a chair with support of 1 arm.
   - Severe weakness of pelvic and proximal leg muscles (strength <3 in at least one of these muscles). Stands up with support of both arms. Cannot walk stairs. Walks unaided. Uses wheelchair for some outdoor activities.
   - Walking limited to a few steps with support. Needs wheelchair outdoors. Uses wheelchair indoors.
   - Completely wheelchair dependent.
D. FSHD-Specific Physical Exam (continued)

8. Clinical Severity Score (CSS) for FSHD (continued)

| Age adjusted Clinical Severity Score: (derived from Physician Evaluation Part I: Item D.8. - | Yes | No | Unknown |
| Age-Corrected CSS = [(CSS x 2)/age at examination] x 1,000 | Y | N | I/D * |

9. Does this patient have a history of hearing loss? Yes No Unknown

10. Does this patient have a history of retinal vascular problems (e.g., Coat's Disease)? Yes No Unknown

E. Clinical Certainty: To help us determine the clinical certainty of the diagnosis of FSHD, please provide the following based on medical record review/patient examination.

On clinical examination, do you see evidence of ptosis or weakness of extraocular muscles? (Excl c) Yes No Insufficient Data *

Has there been a muscle biopsy in this patient or an affected relative with features suggesting a diagnosis other than FSHD? (Excl d) Y N I/D *

Has there been an EMG in this patient or an affected relative showing myotonia or neurogenic changes? (Excl e) Y N I/D *

* "Insufficient Data" is defined as procedure not done or data not available.

Comments: __________________________________________________________

PRINTED Name of Examining Physician

__________________________________________________________

Signature of Examining Physician Date (mm-dd-yyyy)

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