

**Physical and Functional Examination
for Phenotypic Facioscapulohumeral Dystrophy (FSHD) Study
Created by Rabi Tawil, MD, for use by offsite physicians**

Ver 5, Pgs 1-3
& Addend Pg 4
12/01/2008

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:

Form No:	Center #:	Fields Center #:	Initials:	Date: (mmddyyyy)
8 4b	0 1	<input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>	<input type="text"/> <input type="text"/> <input type="text"/>	<input type="text"/> <input type="text"/> - <input type="text"/> <input type="text"/> - <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
		NMD#		
		<input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>		

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 | <input type="checkbox"/> |
| 2. Proximal upper extremity involvement | <input type="checkbox"/> |
| 3. Distal upper extremity involvement | <input type="checkbox"/> |
| 4. Proximal lower extremity involvement | <input type="checkbox"/> |
| 5. Distal lower extremity involvement | <input type="checkbox"/> |
| 6. Someone other than patient noticed problem | <input type="checkbox"/> |
| 7. Patient is without signs or symptoms | <input type="checkbox"/> |

C. Family History of FSHD

- | | | | | |
|--------------------------------------|--------------------------|----------------|-----------------------------------|---------------------------------|
| 1. No affected family members | <input type="checkbox"/> | | | |
| 2. Affected family members (specify) | <input type="checkbox"/> | If Yes: | Mother <input type="checkbox"/> | Father <input type="checkbox"/> |
| | | | Siblings <input type="checkbox"/> | Other <input type="checkbox"/> |
| 3. Unknown | <input type="checkbox"/> | | | Specify _____ |

For University of Rochester Medical Center Use Only:

Form No: 8 4b	Center #: 0 1	Fields Center #: [][][][][][]	Initials: [][][]	Date: (mmddyyyy) [][] - [][] - [][][][]
		NMD#: [][][][][]		

Missing Value Codes: D = Not Applicable X = Unknown

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")

[][]

Scoring for Items 2 through 4		
U	N	= Unaffected
A	F	= Affected
U	C	= Uncertain

3. Scapular Stabilizer Weakness

Is there evidence of scapular winging?

[][]

4. Ankle Dorsiflexor Weakness

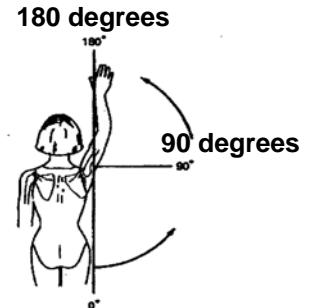
Can they walk on their heels?

[][]

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

	R	L
Shoulder Forward Flexion	[][]	[][]
Shoulder Adduction	[][]	[][]
Elbow Flexion	[][]	[][]
Elbow Extension	[][]	[][]
Wrist Extension	[][]	[][]
Hip Flexion	[][]	[][]
Knee Extension	[][]	[][]
Knee Flexion	[][]	[][]
Foot Dorsiflexion	[][]	[][]

Shoulder Abduction - Range of Motion (in °)	
R	[][][] °
L	[][][] °



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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).

Present
 Absent
 Not Done

Place patient supine without a pillow.

Ask patient to lift head off the table.

Measure movement of umbilicus. Record "Absent" if none.

If, Present, describe:	
Direction	↓ ↑
Distance (cm)	_____ .

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.

- 0** No signs of muscle weakness.
- 1** Facial weakness only.
- 2** Mild scapular weakness. No limitations of abduction or elevation. Often asymptomatic.
- 3** Moderate scapular weakness. Arm abduction > 60 degrees and strength ≥ 3 in arm muscles. No lower limb involvement. Usually symptomatic.
- 4** 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.
- 5** Foot extensor weakness. No pelvic girdle weakness.
- 6** Mild weakness of pelvic and proximal leg muscles (strength ≥ 4 in all muscles). Stands up from a chair without support.
- 7** Moderate pelvic girdle and proximal leg weakness (strength ≥ 3 in all muscles). Stands up from a chair with support of 1 arm.
- 8** 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.
- 9** Walking limited to a few steps with support. Needs wheelchair outdoors. Uses wheelchair indoors.
- 10** Completely wheelchair dependent.

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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. - [][][][] . Age-Corrected CSS = [(CSS x 2)/age at examination] x 1,000

9. Does this patient have a history of hearing loss?

Yes <input type="checkbox"/>	No <input type="checkbox"/>	Unknown <input type="checkbox"/>
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**10. Does this patient have a history of retinal vascular problems
(e.g., Coat's Disease)?**

Yes <input type="checkbox"/>	No <input type="checkbox"/>	Unknown <input type="checkbox"/>
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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 <input type="checkbox"/>	No <input type="checkbox"/>	Insufficient Data * <input type="checkbox"/>
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Has there been a muscle biopsy in this patient or an affected relative with features suggesting a diagnosis other than FSHD?
(Excl d)

Y <input type="checkbox"/>	N <input type="checkbox"/>	I/D * <input type="checkbox"/>
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Has there been an EMG in this patient or an affected relative showing myotonia or neurogenic changes?
(Excl e)

Y <input type="checkbox"/>	N <input type="checkbox"/>	I/D * <input type="checkbox"/>
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* "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)