

# French National Registry on FSHD

## CLINICAL EVALUATION FORM "Type 1 FSHD"

|  |   |
|--|---|
| - Last Name (initial)  __       First Name (initial)  __ <br>- Birth date (month/year): ____/____      - Gender: F <input type="checkbox"/> M <input type="checkbox"/><br>- Primary residence: _____ | Date of consult: ____/____/_____<br><input type="checkbox"/> Form #1 (INCLUSION)<br><input type="checkbox"/> Follow-up form #  __  → PIN: _____ |
| Name of the physician specialized in neuromuscular disorders recruiting the patient: _____<br>Center: _____  | Signature _____<br>Office stamps of physician (including name) _____  |

### A. CIRCUMSTANCES AND ELEMENTS OF DIAGNOSIS

*To be completed at inclusion only*

1. Year of clinical diagnosis \_\_\_\_\_      2. Age at first symptoms \_\_\_\_\_ years old

3. First symptom according to patient

|   |  |
|---|--|
| <input type="checkbox"/> Asymptomatic patient             | <input type="checkbox"/> Facial involvement              |
| <input type="checkbox"/> Proximal upper limb involvement  | <input type="checkbox"/> Proximal lower limb involvement |
| <input type="checkbox"/> Distal upper limb involvement    | <input type="checkbox"/> Distal lower limb involvement   |
| <input type="checkbox"/> Other → if other, specify: _____ |  |

4. Family history: affected family members (several answers possible)

None    Mother    Father    Brothers    Sisters    Others: \_\_\_\_\_

→ GENEALOGICAL TREE: ○ = healthy woman;  
 = healthy man; ● = affected woman; ■ = affected man

5. Molecular diagnostic of FSHD1

a. Laboratory:    Paris    Marseille  
 Other: \_\_\_\_\_

b. Analysis year: \_\_\_\_\_

c. 4q35/ D4Z4: number of repeats: |\_\_|\_\_| or ECORI  
 fragment length: \_\_\_\_\_ kb (from 10 to 38)

d. Check if mosaic:

### B. FOLLOW-UP DATA *To be completed at inclusion AND follow-up*

Data unchanged since last form

1. Current civil status

Single    Separated    Domestic partnership    Civil union (PACS)    Married    Divorced    Widowed

2. Employment status

Employed    Unemployed    Retired    Never employed    Termination of employment, check if due to FSHD:

Pupil / Student

→ Education level: \_\_\_\_\_

→ Education environment:    Normal (no help)    Normal with help (support personnel, adjusted schedules)    Specialized

3. Biometric data

Height \_\_\_\_\_ cm / Weight \_\_\_\_\_ kg

4. Handedness:

Right-handed    Left-handed    Ambidextrous

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## C. MUSCULAR AND FUNCTIONAL EVALUATIONS *To be completed at inclusion AND follow-up*

Data unchanged since last form

| 1. Facial involvement      | Un-affected | Affected | Un-certain |
|----------------------------|-------------|----------|------------|
| Wrinkle forehead           |             |          |            |
| Close eyes                 |             |          |            |
| Smile                      |             |          |            |
| Pucker lips (mimic a kiss) |             |          |            |
| Show teeth (platysma)      |             |          |            |

2. Tongue atrophy/weakness:  No  Yes

3. Weakness of the scapula fixator muscles:

Scapular winging:  No  Yes

4. Axial involvement:  No  Yes

5. Ability to walk:  No  Yes

**6. Functional autonomy:**

- Getting out of bed:  without using arms  without using arms  with the help of someone
- Getting up from a chair:  without using arms  without using arms  with the help of someone

**7. Manual muscle testing: 0-5**

| FACIAL INVOLVEMENT   | R   | L  |
|--|---|----|
| Orbicularis oculi  | __  | __ |
| Orbicularis oris   | __  | __ |
| AXIAL INVOLVEMENT  |   |    |
| Neck flexor muscles  | __  |    |
| Neck extensor muscles  | __  |    |
| Trunk flexor muscles   | __  |    |
| Trunk extensor muscles   | __  |    |
| UPPER LIMBS  | R   | L  |
| Shoulder antepulsion   | __  | __ |
| Right scapular retropulsion (fixator)                                    | __  | __ |
| Shoulder Abduction: overall  | __  | __ |
| Shoulder abduction: deltoid muscle <i>(the scapula is held manually)</i> | __  | __ |
| Forearm flexion (biceps)   | __  | __ |
| Forearm extension (triceps)  | __  | __ |
| Wrist flexion  | __  | __ |
| Fingers flexion  | __  | __ |
| Fingers extension  | __  | __ |
| LOWER LIMBS  | R   | L  |
| Hip flexion (ilio-psoas)   | __  | __ |
| Leg extension (quadriceps)   | __  | __ |
| Leg flexion (hamstrings)   | __  | __ |
| Foot dorsiflexion (tibialis anterior)                                    | __  | __ |
| Plantar flexion (triceps sural)  | __  | __ |
| Comments: _____  | Asymmetry: <input type="checkbox"/> No <input type="checkbox"/> Yes |    |

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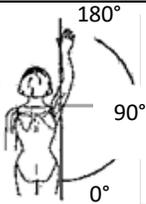
## C. MUSCULAR AND FUNCTIONAL EVALUATIONS

(continued)

**8. Beevor's sign:** Movement of the navel towards the neck on flexing the neck (weakness of the lower abdominal muscles)

Untested     Absent     Present: direction  $\uparrow$  \_\_\_\_\_ cm / direction  $\downarrow$  \_\_\_\_\_ cm

**9. Functional tests**

| A. UPPER LIMBS   |   |   |
|--|---|---|
| Active elevation of arms (degrees)                             | L _____° R _____° <input type="checkbox"/> Not done                       |                  |
| Brooke scale   | ____ / 6 (see appendix #1) <input type="checkbox"/> Not done              |   |
| B. LOWER LIMBS   |   |   |
| Timed 10-meter walk test                                       | _____ sec. <input type="checkbox"/> NA* <input type="checkbox"/> Not done | <input type="checkbox"/> With assistance → Specify: _____<br><input type="checkbox"/> No assistance |
| Four square step test  | _____ sec. <input type="checkbox"/> NA* <input type="checkbox"/> Not done | <input type="checkbox"/> With assistance → Specify: _____<br><input type="checkbox"/> No assistance |
| Vignos scale   | ____ / 10 (see appendix #1) <input type="checkbox"/> Not done             |   |
| C. CLINICAL SEVERITY SCORE                                     |   |   |
| _____ / 10 (see appendix #2) <input type="checkbox"/> Not done |   |   |

**10. Reduced joint flexibility:**  No     Yes → Specify:  Shoulders     Elbows     Wrists     Fingers     Hips  
 Knees     Ankles     Other, specify: \_\_\_\_\_

**11. Joint or muscle pain:**  No     Yes

*If yes, specify:*

a. Date of onset (month / year): \_\_\_\_\_ / \_\_\_\_\_

b. Visual Analog Scale (VAS) on the day of consult: place a thin line perpendicular to the VAS line (**done by the patient**)

No pain \_\_\_\_\_ Extremely painful

c. Pain management:  None     Stretching     Massage

Drugs → specify: \_\_\_\_\_  Others → specify: \_\_\_\_\_

## D. THERAPEUTIC CARE

Data unchanged since last form

**1. Non-drug treatment:**

None     Physical therapy → number of sessions / week: \_\_\_\_

Orthophony → number of sessions / week: \_\_\_\_     Scapular fixation surgery →  Right     Left

**2. Assistive device**

None     Cane →  simple     crutch     tripod

Walker

Manual wheelchair → frequency of use:  occasionally     regularly     exclusively

Electric wheelchair → frequency of use:  occasionally     regularly     exclusively

Other, specify: \_\_\_\_\_

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### E. CARDIAC INVOLVEMENT *To be completed at inclusion AND follow-up*

Data unchanged since last form   
  Not assessed   
  ABSENCE   
  PRESENCE \*  
\*please complete below

- Age at onset: \_\_\_\_ →  Patient interview data    or     Follow-up data
- Symptoms:   
  Palpitations   
  Syncopes, fainting   
  Vertigo, dizziness   
  Retrosternal pain  
 No symptom but but abnormal exam   
  Other, *specify* \_\_\_\_\_
- Cardiac exams (if done, specify for each exam **the month and the year**):

|                   |   |   |
|-------------------|---|---|
| ECG               | <input type="checkbox"/> Done ___ / ____<br><input type="checkbox"/> Not done | <input type="checkbox"/> No anomaly <input type="checkbox"/> Conduction block <input type="checkbox"/> Arrhythmia<br><input type="checkbox"/> Others, <i>specify</i> : _____  |
| Holter test       | <input type="checkbox"/> Done ___ / ____<br><input type="checkbox"/> Not done | <input type="checkbox"/> No anomaly <input type="checkbox"/> Conduction block <input type="checkbox"/> Arrhythmia<br><input type="checkbox"/> Others, <i>specify</i> : _____  |
| Echo-cardiography | <input type="checkbox"/> Done ___ / ____<br><input type="checkbox"/> Not done | - <b>Left Ventricular Ejection Fraction:</b> _____ %<br>- <b>Cardiomyopathy:</b><br><input type="checkbox"/> No <input type="checkbox"/> Yes → <input type="checkbox"/> Dilated <input type="checkbox"/> Hypertrophic <input type="checkbox"/> Other: _____<br>- <b>Other, specify:</b> _____ |

4. Diagnosis retained: \_\_\_\_\_

### F. RESPIRATORY INVOLVEMENT *To be completed at inclusion AND follow-up*

Data unchanged since last form   
  Not assessed   
  ABSENCE   
  PRESENCE \*  
\*please complete below

- Age at onset \_\_\_\_ →  Patient interview data    or     Follow-up data
- Symptoms:   
  Dyspnea on exertion   
  Dyspnea at rest   
  Orthopnea  
 Daytime sleepiness   
  Morning headaches   
  Others, *specify*: \_\_\_\_\_
- Concomitant skeletal anomalies:  
 Spine deformation   
  Pectus excavatum   
  Other, *specify*: \_\_\_\_\_   
  None
- Exams (if done, specify for each exam **the month and the year**):

|                     |   |   |
|---------------------|---|---|
| PFT                 | <input type="checkbox"/> Done ___ / ____<br><input type="checkbox"/> Not done | FVC sitting _____%    FVC supine _____%   |
| Blood gases at rest | <input type="checkbox"/> Done ___ / ____<br><input type="checkbox"/> Not Done | PO <sub>2</sub> _____ mmHg    PCO <sub>2</sub> _____ mmHg    Sat _____ %  |
| Polysomnography     | <input type="checkbox"/> Done ___ / ____<br><input type="checkbox"/> Not Done | <b>Type of apnea:</b> <input type="checkbox"/> Obstructive <input type="checkbox"/> Central <input type="checkbox"/> Mixed<br>Apnea index:   _____   per hour |

- Assisted ventilation:   
  No   
  Yes →  Invasive (tracheostomy)   
  Non-invasive, nasal mask  
 →  Permanent   
  Intermittent, as required   
  Intermittent, nocturnal

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### G. OCULAR INVOLVEMENT *To be completed at inclusion AND follow-up*

Data unchanged since last form     Not assessed     ABSENCE     PRESENCE \*

\*please complete below

1. Age at onset: \_\_\_\_ →  Patient interview data    or     Follow-up data
2. Diagnosis:  Vascular retinopathy     Retinal Detachment     Coats' disease     Corneal ulcer  
 Others, specify: \_\_\_\_\_
3. Surgery:  No     Yes → if yes, reason of surgery: \_\_\_\_\_

### H. AUDITORY INVOLVEMENT

*To be completed at inclusion AND follow-up*

Data unchanged since last form     Not assessed     ABSENCE     PRESENCE\*

\* please complete below

Year of last audiogram: \_\_\_\_\_  Not done

1. Age at onset: \_\_\_\_ →  Patient interview data    or     Follow-up data
2. Hearing aid:  No     Yes

### I. GASTRO-INTESTINAL INVOLVEMENT

*To be completed at inclusion AND follow-up*

Data unchanged since last form     Not assessed     ABSENCE     PRESENCE \*

\*please complete below

1. Age at onset: \_\_\_\_ →  Patient interview data    or     Follow-up data
2. Symptoms:  Dysphagia     Pulmonary aspiration → specify number of pulmonary aspirations: \_\_\_\_ / week  
 Other, specify: \_\_\_\_\_
3. Endoscopic evaluation of swallowing:  Not done     done: Dysphagia detected ? :  No     Yes

### J. METABOLIC INVOLVEMENT

*To be completed at inclusion AND follow-up*

Data unchanged since last form     Not assessed     ABSENCE     PRESENCE \*

\*please complete below

1. Age at onset: \_\_\_\_ →  Patient interview data    or     Follow-up data
2. Diagnosis:  Hypercholesterolemia     Hypertriglyceridemia     Other, specify: \_\_\_\_\_
3. Lipid-lowering treatment:  No     Yes → specify name(s) and length of treatment(s):  
\_\_\_\_\_

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### K. ENDOCRINE INVOLVEMENT

*To be completed at inclusion AND follow-up*

Data unchanged since last form

Not assessed

ABSENCE

PRESENCE \*

\*please complete below

1. **Age at onset:** \_\_\_\_ →  Patient interview data or  Follow-up data
2. **Type of disease:**  Thyroid disorder  Diabetes  Other, specify: \_\_\_\_\_
3. **Hormone therapy:**  No  Yes → specify name(s) and length of treatment(s):  
\_\_\_\_\_
4. **Sexual hormone therapy:**  No  Yes → specify name(s) and length of treatment(s):  
\_\_\_\_\_

### FOR ALL FEMALE SUBJECTS

Pre-puberty  Puberty  Pre-menopause  Menopause → Age at menopause: \_\_\_\_

- **Contraceptive treatment:**  None  Oral contraceptive  Other, specify: \_\_\_\_\_

- **Pregnancy:** Number of pregnancies (gestity): \_\_\_\_ Number of children: \_\_\_\_

### L. TYPICAL AND ATYPICAL CHARACTERISTICS OF FSHD

*To be completed at inclusion AND follow-up*

- |                          |   |
|--------------------------|---|
| <input type="checkbox"/> | <b>TYPICAL:</b> at least 2 criteria in favor of a FSHD1 diagnosis <u>AND</u> 0 atypia criterion |
| <input type="checkbox"/> | <b>WITH ATYPIA:</b> at least 1 atypia criterion   |

#### Check matching criteria

| CRITERIA IN FAVOR OF FSHD1 |   |
|----------------------------|---|
| <input type="checkbox"/>   | Evidence of dominant transmission   |
| <input type="checkbox"/>   | Facial involvement (orbicularis oculi or orbicularis oris)  |
| <input type="checkbox"/>   | Involvement of scapular fixator or tibialis anterior  |
| ATYPIA CRITERIA            |   |
| <input type="checkbox"/>   | Discordance between the degree of involvement and the number of D4Z4 RU<br><i>Example: patient with 9 RU in a wheelchair or patient with 2 UR able to walk.</i> |
| <input type="checkbox"/>   | Ptosis, ophthalmoplegia or diplopia   |
| <input type="checkbox"/>   | Cardiological or respiratory involvement in the foreground OR SNC involvement   |
| <input type="checkbox"/>   | Predominant involvement at the proximal lower limb or distal upper limb level   |
| +                          |   |
| <input type="checkbox"/>   | EMG suggesting a neurogenic motor neuron disease or myotonia  |
| <input type="checkbox"/>   | Muscle biopsy atypical or suggesting a muscle disorder  |
| <input type="checkbox"/>   | Genetic diagnosis of another neuromuscular disorder confirmed   |
| <input type="checkbox"/>   | Others: _____   |

## French National Registry on FSHD

### APPENDIX 1

#### BROOKE AND VIGNOS SCALES

##### UPPER LIMBS (Arms and shoulders)

- 1 – Starting with arms at the sides, the patient can abduct the arms (kept extended) in a full circle until they touch above the head.
- 2 – Can raise arms above head only with elbows flexed or by using accessory muscles.
- 3 - Can raise hands to the mouth but cannot raise an 8-oz glass of water to the mouth (using both hands if necessary).
- 4 – Can raise hands to the mouth but cannot raise an 8-oz glass of water to the mouth.
- 5 - Cannot raise hands to the mouth but can use hands to hold a pen or pick up pennies from the table.
- 6 – Cannot raise hands to the mouth and has no useful function of hands.

##### LOWER LIMBS

- 1 – Walks and climbs stairs without assistance.
- 2 - Walks and climbs stair with the aid of the railing.
- 3 – Walks and climbs stairs slowly with the aid of the railing (over 12 seconds to climb four stairs).
- 4 – Walks unassisted and rises from a chair but cannot climb stairs.
- 5 – Walks unassisted but cannot rise from a chair or climb stairs.
- 6 – Walks only with assistance or walks independently with long leg braces.
- 7 – Walks with long leg braces but requires assistance for balance.
- 8 – Stands with long leg braces but unable to walk even with assistance
- 9 – Wheelchair-bound.
- 10 – Bedridden.

### APPENDIX 2

#### CLINICAL SEVERITY SCORE

- 0 = No sign of muscle weakness.
- 1 = Facial weakness only.
- 2 = Mild scapular involvement without limitation of arm abduction; no awareness of disease symptoms is possible.
- 3 = Moderate involvement of scapular and arm muscles or both (arm abduction  $>60^\circ$  and strength  $\geq 3$  in arm muscles); no involvement of pelvic and leg muscles.
- 4 = Severe scapular involvement (arm abduction  $<60^\circ$  on at least one side); strength  $< 3$  in at least one muscular district of the arms; no involvement of pelvic and leg muscles.
- 5 = Tibioperoneal weakness; no weakness of pelvic and proximal leg muscles.
- 6 = Mild weakness of pelvic and proximal leg muscles or both (strength  $\geq 4$  in all these muscles); able to stand up from a chair without support.
- 7 = Moderate weakness of pelvic and proximal leg muscles or both (strength  $\geq 3$  in all these muscles); able to stand up from a chair with monolateral support.
- 8 = Severe weakness of pelvic and proximal leg muscles or both (strength  $< 3$  in at least one of these muscles); able to stand up from a chair with double support; unable to climb stairs; able to walk unaided.
- 9 = Unable to stand up from a chair; walking limited to several steps with support; may use wheelchair for most activities.
- 10 = Wheelchair-bound.