

French National Registry on FSHD

CLINICAL EVALUATION FORM "FSHD with no D4Z4 contraction"

- Last Name (initial) __ First Name (initial) __ - Birth date (month/year): ____/____ - Gender: F <input type="checkbox"/> M <input type="checkbox"/> - Primary residence: _____	Date of consult: ____/____/_____ <input type="checkbox"/> Form #1 (INCLUSION) <input type="checkbox"/> Follow-up form # __ → PIN: _____
Name of the physician specialized in neuromuscular disorders recruiting the patient: _____ Center: _____	Signature _____ 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. Mutation in SMCHD1 gene: Yes No
 Not done
 - e. Methylation percentage: "FseI % methylation" _____ % Not done

B. DIAGNOSIS COMPLEMENT *To be completed at inclusion AND follow-up*

Data unchanged since last form

1. Other genetic tests: check the genetic tests that have been performed on your patient (several choices are possible):
2.

<input type="checkbox"/> Calpain-3	→	Results: ○ negative	○ pending	○ not done
<input type="checkbox"/> Dysferlin	→	Results: ○ negative	○ pending	○ not done
<input type="checkbox"/> VCP	→	Results: ○ negative	○ pending	○ not done
<input type="checkbox"/> FHL1	→	Results: ○ negative	○ pending	○ not done
<input type="checkbox"/> Others → Specify: _____	→	Results: ○ negative	○ pending	○ not done
_____	→	Results: ○ negative	○ pending	○ not done
_____	→	Results: ○ negative	○ pending	○ not done

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3. Other blood tests: check blood tests that have been performed on your patient (several choices are possible):

4. CPT-II in leukocytes → Results: negative pending not done
 Dried blood spot GAA testing (*Pompe disease*) → Results: negative pending not done
 GAA in lymphocytes (*Pompe disease*) → Results: negative pending not done
 Lowest CPK values: Year: _____ x normal value

5. **EMG** not done **done** → Result: Normal Myogenic Neurogenic

6. **Muscle biopsy:** not done **done** → Date of the muscle biopsy: ____/____/____

Result: Normal (go to question #b) or Abnormal, specify:

a. Histology / Histochemistry: if the biopsy is abnormal, specify (several choices are possible):

- Dystrophy → no yes
- Necrosis → no yes
- Inflammation → no yes
- Selective fiber atrophy → no yes
- Fiber-type grouping → no yes
- Vacuoles → no yes
- Inclusions → no yes
- Ragged red fibers or COX-neg. fibers → no yes, specify number: _____
- Others → Specify: _____

b. Immunohistochemistry: check the immunohistochemical analyzes done on the muscle biopsy (several choices are possible):

- DYS 1,2,3 Sarcoglycans Caveolins Dysferlin α -/ β dystroglycans Merosin
 Other, specify: _____

c. Western blot: check all western blots done on the muscle biopsy (several choices are possible):

- DYS 1,2,3 Sarcoglycans Calpain-3 Dysferlin α -/ β dystroglycans
 Other, specify: _____

d. Additional tests: check all additional tests done on the muscle biopsy (several choices are possible):

- Calpain-3 autocatalytic activity
 Dosage of mitochondrial respiratory chain enzymes
 Activity of glycolysis and glycogenesis enzymes
 Other, specify: _____

5. Muscle MRI

- not done **done, specify:** Upper limbs Lower limbs Full body

C. 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 Specialized
(support personnel, adjusted schedules)

3. Biometric data

Height _____ cm / Weight _____ kg

4. Handedness:

- Right-handed Left-handed Ambidextrous

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D. 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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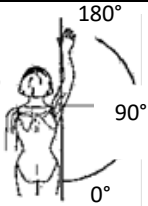
D. MUSCULAR AND FUNCTIONAL EVALUATIONS

(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: _____ <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: _____ <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: _____

E. 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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F. CARDIAC 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:
 Palpitations
 Syncopes, fainting
 Vertigo, dizziness
 Retrosternal pain
 No symptom but but abnormal exam
 Other, specify: _____
3. Cardiac exams (if done, specify for each exam **the month and the year**):

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

4. Diagnosis retained: _____

G. RESPIRATORY 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:
 Dyspnea on exertion
 Dyspnea at rest
 Orthopnea
 Daytime sleepiness
 Morning headaches
 Others, specify: _____
3. Concomitant skeletal anomalies:

 Spine deformation
 Pectus excavatum
 Other, specify: _____
 None
4. Exams (if done, specify for each exam **the month and the year**):

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

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

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H. 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: -----

I. 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

J. 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

K. 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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L. 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: ____

M. TYPICAL AND ATYPICAL CHARACTERISTICS OF FSHD

To be completed at inclusion AND follow-up

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

Check matching criteria

CRITERIA IN FAVOR OF FSHD1	
<input type="checkbox"/>	Asymetry
<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"/>	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: _____

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 ≥ 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 ≥ 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 ≥ 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.