

**Part A**

Referring Hospital: \_\_\_\_\_ Referring Physician: \_\_\_\_\_

Compilation Date: \_\_/\_\_/\_\_\_\_

Patient Initials: \_\_\_\_\_ Patient code: \_\_\_\_\_

Date of birth: \_\_/\_\_/\_\_\_\_ Sex:         M     F

Handedness:     Left     Right

Weight: \_\_\_\_\_ kg

Height: \_\_\_\_\_ cm

Family Ancestry - geographic origins:

Maternal: \_\_\_\_\_ Mother Surname: \_\_\_\_\_

Paternal: \_\_\_\_\_

Consanguinity:  Yes     No

Current profession: \_\_\_\_\_ Since year: \_\_\_\_

*If you were previously employed:*

Previous profession(s) :

\_\_\_\_\_ From year: \_\_\_\_ to year \_\_\_\_

\_\_\_\_\_ From year: \_\_\_\_ to year \_\_\_\_

\_\_\_\_\_ From year: \_\_\_\_ to year \_\_\_\_

Highest degree:     University degree     High-school diploma     Primary school diploma     None

(years of education: \_\_\_\_\_)

**Clinical history**

Previous evaluation in other center(s):  Yes  No If yes, centre: (1) \_\_\_\_\_  
(2) \_\_\_\_\_

FSHD score at last clinical examination: \_\_ \_ Date: \_\_ / \_\_ / \_\_ \_ \_

**Comorbidities:**

Diabetes mellitus:  Yes  No  Not evaluated

If Yes,  type I  type II Age at diagnosis: \_\_ \_

Therapy, Drugs :

\_\_\_\_\_ Dose: \_\_\_\_\_ unit \_\_\_\_\_ From year: \_\_\_\_\_ to year \_\_\_\_\_

\_\_\_\_\_  
\_\_\_\_\_

Thyroid hormones alterations:  Yes  No  Not evaluated

If Yes,  hypothyroidism  hyperthyroidism Age at diagnosis: \_\_ \_

Therapy, Drugs :

\_\_\_\_\_ Dose: \_\_\_\_\_ unit \_\_\_\_\_ From year: \_\_\_\_\_ to year \_\_\_\_\_

\_\_\_\_\_  
\_\_\_\_\_

Hepatitis:  Yes  No  Not evaluated

If Yes,  HBV  HCV  Toxic Age at diagnosis: \_\_ \_

Therapy, Drugs :

\_\_\_\_\_ Dose: \_\_\_\_\_ unit \_\_\_\_\_ From year: \_\_\_\_\_ to year \_\_\_\_\_

\_\_\_\_\_  
\_\_\_\_\_

Diagnosis of cancer:      Yes      No

If yes, specify: \_\_\_\_\_ Age at diagnosis: \_\_ \_\_

Therapy, Drugs :

\_\_\_\_\_ Dose: \_\_\_\_\_ unit \_\_\_\_\_ From year: \_\_\_\_\_ to year \_\_\_\_\_  
\_\_\_\_\_

Retinal vasculopathy:      Yes      No      Not evaluated

Sensorineural deafness:      Yes      No

Audiometry:      Altered      Normal      Not performed

Epilepsy:      Yes      No

Cognitive impairment:      Yes      No

**Other disease(s)**

Other diseases      Yes      No

If yes, specify: \_\_\_\_\_

\_\_\_\_\_  
\_\_\_\_\_

Previous trauma: joint, bone fractures:      Yes      No     If yes, specify site and age\_\_ \_\_

\_\_\_\_\_

**Drugs:**

Statins:             Yes     No    If yes, Type: \_\_\_\_\_

Dose: \_\_\_\_\_ unit \_\_\_\_\_    From year: \_\_\_\_\_ to year \_\_\_\_\_

Others chronic treatments:  Yes     No

If yes

Drug: \_\_\_\_\_ Dose: \_\_\_\_\_    From year: \_\_\_\_\_ to year \_\_\_\_\_

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**Reproductive History:**

Have you ever been pregnant?     Yes     No

Are you pregnant now?             Yes     No

How many times have you been pregnant:    \_\_ \_\_

Spontaneous abortion:             Yes     No    if yes, Number of spontaneous abortion: \_\_ \_\_

How many vaginal deliveries have you had? {Please count stillbirths as well as live births}: \_\_ \_\_

How many cesarean deliveries have you had? {Please count stillbirths as well as live births}: \_\_ \_\_

How many of the deliveries resulted in a live birth? : \_\_ \_\_

How old were you at the time of your first live birth? age \_\_ \_\_

How old were you at the time of your last live birth? age \_\_ \_\_

Prenatal diagnosis     Yes (N° \_\_\_\_\_)     No    If yes, result: \_\_\_\_\_

Modification of the disease after the pregnancy:     None     Worsening     Improvement

**Menopause:**             Yes     No    If yes, physiological menopause:     Yes     No    age \_\_ \_\_

Hormonal therapy:     Yes     No    Modification of the disease:  None     Worsening     Improvement

**Physical activity:** Have you ever regularly played a sport?  Yes  No

If yes, report the two most played sports:

Sport (1): \_\_\_\_\_  Professional  Amateur From age: \_\_ to age \_\_

Modification of the disease:  None  Worsening  Improvement

Sport (2): \_\_\_\_\_  Professional  Amateur From age: \_\_ to age \_\_

Modification of the disease:  None  Worsening  Improvement

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**Physiokinesitherapy (PKT):**  Yes  No

If yes, Duration of PKT treatment: From year: \_\_\_\_ to year \_\_\_\_

Modification of the disease:  None  Worsening  Improvement

**Surgery:**  Yes  No

If yes, operation (1): \_\_\_\_\_ year: \_\_\_\_

Anesthesia:  General  Local  Epidural

Modification of the disease:  None  Worsening  Improvement

If yes, operation (2): \_\_\_\_\_ year: \_\_\_\_

Anesthesia:  General  Local  Epidural

Modification of the disease:  None  Worsening  Improvement

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**Family history** (*information from at least three generations should be collected*):

“Was/is any of your relatives wheelchair bound?”

“Did/does any of your relatives have a posture like yours?”

“Was any of your relatives sleeping with half-open eyes?”

Other considerations \_\_\_\_\_

\_\_\_\_\_

**(pedigree attached)**



Duration (years) from onset\_\_\_\_\_

Recurrent/chronic pain:  Yes  No If yes, since age \_\_ \_\_

Specify location\_\_\_\_\_

Precocious muscle fatigue during the common daily activities,before the onset of muscle impairment:

Yes  No If yes, since age \_\_ \_\_

Specify location\_\_\_\_\_

Other observations  Yes  No

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**Face:**

Presence of:

Widened palpebral fissures: .. Yes  No

Puckered lips:  Yes  No

Horizontal smile:  Yes  No

Orbiculari oris hypokinesia during speech:  Yes  No

Dysarthria:  Yes  No

Orbicularis oculi evaluation:  Normal (able to close heavily eyes)

Partial (able to close eyes but incapable to close heavily eyes)

Unable (unable to completely close eyes)

Ability to protrude lips:  Normal  Partial  Unable

Ability to puff out cheeks (against no resistance):  Normal  Partial  Unable

Asymmetric involvement of facial muscle:  Yes  No

if yes, specify side\_\_\_\_\_

**Scapular girdle:**

Ability to abduct arms:  Whole (180°)

Complete but abnormal (patient can rise arms above head but only by flexing the elbow or using the accessory muscle)

Incomplete: >45° but <180° (specify if:  ≥90° or  <90°)

Incomplete: ≤45°

**Pelvic girdle:**

- Ability to climb 4 stairs:
- Without support
  - Without support but abnormally
  - With support (since age\_ \_ )
  - Unable (since age\_ \_ )

- Ability to walk:
- Without support
  - With support (since age\_ \_ )
  - Unable (since age\_ \_ )

- Gait:  Normal  Waddling  Hyperlordotic  Steppage

- Ability to stand up from a chair:
- Without support
  - With support ( since age \_ \_ )
  - Unable (since age \_ \_ )

- Ability to rise from the floor:
- Without support
  - With support ( since age \_ \_ )
  - Unable (since age \_ \_ )

- Use of wheelchair:  Not necessary  With manual control  With electric control  Bed bound

**Legs:**

- Ability to walk on tiptoes and/or heels:  Normal  On tiptoes only  On heels only  Unable

- Beevor's sign:**  Positive  Negative

**Part C**

*Medical Research Council (MRC) score:*

Scores range from 0 to 5, with .5 increments (e.g. 3, 3.5, 4, 4.5, etc)

<i><b>MUSCLE</b></i>	<i><b>RIGHT MRC score</b></i>	<i><b>LEFT MRC score</b></i>	<i><b>ATROPHY Yes (right or left) /no</b></i>
Extrarotator muscles of upper limb*			
Triceps*			
Biceps*			
Common finger extensors*			
Wrist extensors*			
Long fingers flexors*			
Wrist flexors*			
Gluteus maximus			
Iliopsoas			
Biceps femoris			
Quadriceps			
Triceps surae			
Tibialis anterior			

(\* Muscles to be considered for FSDH score “Upper limbs involvement”)

Strength of neck extensors muscles: MRC score \_\_\_\_\_

Weakness of pectoralis muscles:     Yes     No                      If yes,  Right     Left

Pectoralis muscles atrophy:             Yes     No                      If yes,  Right     Left

**PRESENCE OF FOLLOWING TYPICAL FEATURES:**

Scapular winging at rest:  Yes  No

(if yes, specify:  Symmetric winging, or  Asymmetric winging  > right;  > left)

Scapular winging on attempted shoulder abduction or forward flexion:  Yes  No

(if yes, specify:  Symmetric winging, or  Asymmetric winging  > right;  > left)

Horizontal clavicles:  Yes  No

Forward sloping of shoulders at rest:  Yes  No

Atrophy of pectoral muscles/ axillary creases:  Yes ( > right;  > left)  No

Sunken or flattened appearance of the chest:  Yes  No

“Poly-hill sign” with neck, shoulders, and arms observed from behind in

fullest possible abduction (70–90°), with external rotation of the shoulders :  Yes  No

Hyperlordosis:  Yes  No

Orbiculari oris hypokinesia during speech:  Yes  No

**PRESENCE OF UNCOMMON FEATURES:**

Myotonic phenomenon:  Yes  No

Rippling phenomenon:  Yes  No

Eyelid ptosis:  Yes  No

Extra-ocular weakness:  Yes  No

Pharyngeal and lingual muscle weakness (persistent dysphagia):  Yes  No

Bent syndrome:  Yes  No

Early contractures:  Yes  No

(If yes, specify site \_\_\_\_\_)

Dropped head:  Yes  No

Pes cavus:  Yes  No

Myoglobinuria:  Yes  No

Ogival palatus:  Yes  No

Others: \_\_\_\_\_

**Creatine phosphokinase (CPK)** (value of two blood assays separated by at least one month):

- Normal range
- < 4x normal value (<1000 U/L)
- > 4x normal value (>1000 U/L)

**Instrumental evaluation**

Cardiac involvement (ECG, echocardiogram):

Last ECG's report \_\_\_\_\_ (date: \_\_/\_\_/\_\_\_\_)

Last echocardiogram's report \_\_\_\_\_ (date: \_\_/\_\_/\_\_\_\_)

Electromyographic pattern of four limbs (detail the examined muscles) (date: \_\_/\_\_/\_\_\_\_)

- Myopathic pattern ( Proximal ;  Distal)
- Neurogenic pattern ( Proximal ;  Distal)
- Mixed pattern ( Proximal ;  Distal)

Electroneurography of four limbs (detail the examined nerves) (date: \_\_/\_\_/\_\_\_\_)

- Normal
- Abnormal

Report of last pulmonary function tests (FVC, MIP, MEP, Cough peak flow) (date \_\_/\_\_/\_\_\_\_):

\_\_\_\_\_  
\_\_\_\_\_

Report of muscle biopsy (if available; please specify date and biopsied muscle)\*: (date \_\_/\_\_/\_\_\_\_)

Biopsied muscle: \_\_\_\_\_

Report: \_\_\_\_\_  
\_\_\_\_\_

Other genetic test previously performed (if available): \_\_\_\_\_  
\_\_\_\_\_

*\*(please attach reports)*

Name of the Examiner: \_\_\_\_\_

**I - Facial weakness**

0 - no weakness

1 - moderate weakness;

partial ability to do at least one of the following tasks:

- to close eyes
- to protrude lips
- to puff out cheeks

2 - severe weakness;

unable to do at least one of the following tasks:

- to close eyes
- to protrude lips
- to puff out cheeks

**II - Scapular girdle involvement**

0 - no involvement

1 - mild involvement with no limitation of arm abduction

2 - arm abduction > 45°

3 - arm abduction < 45°

**III - Upper limbs involvement \***

0 - no involvement

1 - at least two muscles affected with MRC >3

2 - at least two muscles with MRC ≤ 3

*\*The following 4 muscles will be assessed on each side: 1. triceps; 2. biceps; 3. common finger extensors and wrist extensors; 4. long finger flexors and wrist extensors. Only the weaker muscles will be considered for evaluation.*

**IV - Legs involvement**

The ability to walk on tiptoes and heels will be assessed on each side:

0 - no involvement

1 - unable to walk on tiptoes or heels (only one task impaired)

2 - unable to walk on tiptoes and heels (two tasks impaired)

**V - Pelvic girdle involvement**

0 - no involvement

1 - able to walk and to climb stairs without support but abnormally/ because of posterior leg muscle hypotrophy

2 - able to walk unaided, to climb stairs or to stand up from a chair with support

3 - able to walk unaided but unable to stand up from a chair or to climb stairs without support/ more than 12 seconds

4 - able to walk with support

5 - wheelchair bound

**VI - Abdominal muscle involvement**

0 - no involvement

1 - presence of Beevor's sign

FSHD clinical score: \_\_\_\_\_

	TYPICAL FEATURES	UNCOMMON FEATURES
<b>1. ONSET OF MUSCLE WEAKNESS</b>	<input type="checkbox"/> Facial weakness of orbicularis oculi or oris <input type="checkbox"/> Scapular weakness with altered ability to abduct arms <input type="checkbox"/> Humeral muscles (biceps/triceps)	<input type="checkbox"/> Distal lower limbs onset with triceps surae weakness <input type="checkbox"/> Distal upper limbs onset <input type="checkbox"/> Pelvic girdle onset
<b>2. AXIAL MUSCLES INVOLVEMENT</b>	<input type="checkbox"/> Hyperlordosis <input type="checkbox"/> Beevor's sign	<input type="checkbox"/> Camptocormia <input type="checkbox"/> Dropped head
<b>3. FACIAL INVOLVEMENT</b>	<input type="checkbox"/> Weakness of Orbicularis oculi (facial score $\geq 1$ ) <input type="checkbox"/> Weakness of Orbiculari oris (facial score $\geq 1$ )	<input type="checkbox"/> Weakness of extra-ocular muscles <input type="checkbox"/> Weakness of masticatory muscles (persistent dysphagia)
<b>4. SCAPULAR GIRDLE INVOLVEMENT</b>	<input type="checkbox"/> Impairment of upper limb abduction with winged scapula or limitation of forward flexion (scapular FSHD score $\geq 1$ )	<input type="checkbox"/> Isolated distal upper limb muscle weakness <input type="checkbox"/> Impairment of arms abduction ( $<90^\circ$ ) without winged scapula at rest and/or on attempted shoulder abduction or forward flexion
<b>5. PELVIC GIRDLE INVOLVEMENT</b>	-----	<input type="checkbox"/> Isolated and/or prevailing pelvic girdle muscle weakness
<b>6. LOWER LIMBS INVOLVEMENT</b>	<input type="checkbox"/> Weakness of tibialis anterior muscles weakness	<input type="checkbox"/> Early gastrocnemius and/or soleus atrophy/weakness
<b>7. BLOOD CPK LEVEL</b> (at least two samples 1 month apart)	<input type="checkbox"/> Normal range <input type="checkbox"/> $< 4x$ normal value ( $<1000$ U/L)	<input type="checkbox"/> Value $> 4x$ normal value ( $>1000$ U/L)
<b>8. OTHER SIGNS</b>	<input type="checkbox"/> Shoulders winging on attempted shoulder abduction or forward flexion <input type="checkbox"/> Horizontal clavicles <input type="checkbox"/> Forward sloping of the shoulders at rest <input type="checkbox"/> Sunken or flattened appearance of the chest <input type="checkbox"/> Atrophy of pectoralis muscles <input type="checkbox"/> Orbiculari oris hypokinesia during speech	<input type="checkbox"/> Myotonic phenomenon <input type="checkbox"/> Rippling <input type="checkbox"/> Eyelid ptosis <input type="checkbox"/> Extra-ocular muscle weakness <input type="checkbox"/> Early muscle contractures <input type="checkbox"/> Cardiomyopathy <input type="checkbox"/> Early respiratory insufficiency (Non Invasive Ventilation, NIV; FSHD score $<12$ ) <input type="checkbox"/> Pes cavus <input type="checkbox"/> Myoglobinuria

**Importantly:** Indicate the presence of comorbidities / results of previous injuries / illnesses that can possibly affect the neurological examination: \_\_\_\_\_

**Extra-muscular involvement:**  hearing loss,  epilepsy,  retinal involvement,  cognitive impairment

## CATEGORY A

### Category A1

Severe facial weakness (unable **both** to close eyes **and** to protrude lips) + impairment of upper limb abduction with winged scapula (scapular FSHD score  $\geq 1$ ) + absence of uncommon features

### Category A2

Facial weakness (upper **and** lower facial weakness) + impairment of upper limb abduction with winged scapula (scapular FSHD score  $\geq 1$ ) + absence of uncommon features

### Category A3

Facial weakness (upper **or** lower facial weakness) + impairment of upper limb abduction with winged scapula (scapular FSHD score  $\geq 1$ ) + absence of uncommon features

## CATEGORY B

### Category B1

Impairment of upper limb abduction with winged scapula (scapular FSHD score  $\geq 1$ ), no facial weakness + absence of uncommon features

### Category B2

Facial weakness (facial FSHD score  $\geq 1$ ), no impairment of upper limb abduction + absence of uncommon features

## CATEGORY C

### Category C1

Subject with presence of at least one typical sign + FSHD score =0

### Category C2

Subject without signs of muscle weakness + FSHD score =0

## CATEGORY D

### Category D1

Subject fulfilling criteria of categories A1, A2, A3, B1, B2 + at least one uncommon feature

### Category D2

-Subject fulfilling criteria of categories C1 or C2 + at least one uncommon feature  
-Subject no fulfilling criteria of any of the above categories

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