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: Alterated 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 _____

_____

_____

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

_____

_____

_____

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

_____

_____

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)

Part B

NEUROLOGICAL EXAMINATION

Age at Onset of motor impairment

Subjective age at onset (**when subject has noticed the appearance of motor impairment in his/her daily activities**):_____ years old

Site of muscle weakness reported by patient at onset

Muscle group:

Facial muscles:	○ Yes ○ No	Shoulder girdle muscles:	○ Yes ○ No
Abdominal muscles:	○ Yes ○ No	Distal lower limb muscles:	○ Yes ○ No
Pelvic girdle muscles:	○ Yes ○ No	Distal upper limb muscles:	○ Yes ○ No
Asymmetry at onset:	○ Yes ○ No	If yes, ○ Right ○ Left	
Triggering events	○ Yes ○ No	If yes, event: (1) _____	
		(2) _____	

Objective evaluation of age at onset by specific questions:

Have your relatives never noticed that you were sleeping with half-open eyes? ○ Yes ○ No

If yes, since age _____

Can you drink with a straw? ○ Yes ○ No

If no, since what age have you been unable to drink with a straw? _____

Can you to puff your cheeks? ○ Yes ○ No

If no, since what age have you been unable to puff your cheeks ?_____

Have you always been able to whistle? ○ Yes ○ No

If no, since age_____

Have you noticed the appearance of winged scapula? ○ Yes ○ No

If yes, since age _____

Have you ever noticed thinness of upper arms or a dropped shoulder? ○ Yes ○ No

If yes, since age _____

Have you ever noticed asymmetry of the mouth or smile when looking in a mirror or in past photographs from childhood? ○ Yes ○ No

If yes, since age _____

_____

Other observations:_____

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

_____

Face:

Presence of:

Widened palpebral fissures: ..□ Yes □ No

Purckered lips: □ Yes □ No

Horizontal smile: □ Yes □ No

Orbiculari oris hypokinesia during speech: □ Yes □ No

Dysartria: □ 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)

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

(** Muscles to be considered for FSHD 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 examinated muscles) (date: _ _/_ _ /_ _ _ _)

 Myopathic pattern (Proximal ; Distal)

 Neurogenic pattern (Proximal ; Distal)

 Mixed pattern (Proximal ; Distal)

Electroneurography of four limbs (detail the examinated 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: _____