

**【Infectious disease screening】 \*Mandatory**

HBs-Ag ( - / + / not tested ) HCV-Ab ( - / + / not tested ) HTLV-1-Ab ( - / + / not tested ) HIV-Ag/Ab ( - / + / not tested )

**Clinical Summary Form for *Adult Patient***

Specimen No. (for NCNP use only)

**Patient name:** (in English) \_\_\_\_\_ **Medical record No.:** \_\_\_\_\_

(in Japanese/Chinese) \_\_\_\_\_

**Date of birth:** \_\_\_\_\_ [D/M/Y] (Age: \_\_\_\_\_ yrs \_\_\_\_\_ mos) **Sex:** M / F

**Ethnicity:** \_\_\_\_\_

**Clinical diagnosis :** \_\_\_\_\_

**Sample type :** Skeletal muscle / Peripheral nerve / Skin / Blood / Other **Sample for culture :** Yes / No

**Site of muscle biopsy:** (R L) \_\_\_\_\_ (Performed on: \_\_\_\_\_ [D/M/Y])

**Hospital\*:** \_\_\_\_\_ **Department\*:** \_\_\_\_\_

**Address\*:** \_\_\_\_\_

\*Use Japanese for Japanese hospitals

**TEL:** \_\_\_\_\_ **FAX:** \_\_\_\_\_

**Attending physician\*:** \_\_\_\_\_ **E-mail:** \_\_\_\_\_

**Chief complaint:** \_\_\_\_\_

**Present illness:** \_\_\_\_\_

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

**Past history:** \_\_\_\_\_

\_\_\_\_\_

**Underlying disease(s)** \_\_\_\_\_

**Medication (s)** (N / Y name and period used \_\_\_\_\_)

**Steroid** (N / Y name and period used \_\_\_\_\_)

**Developmental history** (Normal Abnormal \_\_\_\_\_)

**Family history:**

Parental consanguinity N / Y \_\_\_\_\_

Neuromuscular disease N / Y \_\_\_\_\_

\_\_\_\_\_

**Physical examination:**

Height: \_\_\_\_\_ cm Weight: \_\_\_\_\_ kg

Skin abnormality ( N / Y \_\_\_\_\_ )

Spine deformity ( N lordosis scoliosis )

Chest deformity ( N pigeon funnel )

Joint contracture ( N / Y site \_\_\_\_\_ )

High-arched palate ( N / Y narrow wide )

Minor anomaly ( N / Y \_\_\_\_\_ )

Respiratory insufficiency ( N / Y ) Mechanical ventilation ( N / Y since \_\_\_\_\_ , NPPV or TPPV )

**Family Tree**

**Neurological findings:**

Intelligence ( normal borderline low ) Cognitive function ( MMSE HDS-R\_\_\_\_\_ )

Cranial nerve: Ptosis ( N / Y ) Ophthalmoplegia ( N / Y \_\_\_\_\_ )

Facial muscle involvement ( N / Y )

Dysarthria / dysphonia ( N / Y \_\_\_\_\_ ) Dysphagia ( N / Y \_\_\_\_\_ )

Motor system: Grip power ( R \_\_\_\_\_ kg / L \_\_\_\_\_ kg)

Walking ( N / Y possible with assistance tool \_\_\_\_\_ )

MMT		Neck	Shoulder	Elbow	Wrist	Fingers	Hip	Knee	Ankle
Flexion	R		abduction						
	L		abduction						
Extension	R								
	L								

Muscle atrophy ( N / Y \_\_\_\_\_ ) Winged scapula ( N / Y \_\_\_\_\_ )

Muscle hypertrophy ( N / Y GC tongue other \_\_\_\_\_ ) Muscle tone ( normal ↓ ↑ )

Myotonia ( N / Y \_\_\_\_\_ ) Strength fluctuation ( N / Y diurnal within a week within a month )

Myalgia ( N / Y site \_\_\_\_\_, time between exercise and onset of myalgia \_\_\_\_\_, after exercise)

Neck movement limitation ( N / Y flexion extension rotation ) DTR ( normal ↓ absent ↑ \_\_\_\_\_ )

Involuntary movement ( N / Y \_\_\_\_\_ ) Cerebellar sign ( N / Y \_\_\_\_\_ )

Sensory system ( normal / abnormal \_\_\_\_\_ )

Autonomic system ( normal / abnormal \_\_\_\_\_ )

Fundus examination \_\_\_\_\_ Others \_\_\_\_\_

**Laboratory result:**

CK at rest \_\_\_\_\_ (Normal range: \_\_\_\_\_) Aldolase \_\_\_\_\_ (Normal range: \_\_\_\_\_)

AST \_\_\_\_\_ ALT \_\_\_\_\_ CRP \_\_\_\_\_ ESR \_\_\_\_\_ ACE \_\_\_\_\_

Serum lactate \_\_\_\_\_ (normal range: \_\_\_\_\_) CSF lactate \_\_\_\_\_

Serum pyruvate \_\_\_\_\_ (normal range: \_\_\_\_\_) CSF pyruvate \_\_\_\_\_

Autoantibodies \_\_\_\_\_

M proteinemia ( N / Y λ type κ type )

Infectious disease (other than the list in the beginning) \_\_\_\_\_

Endocrinological disease \_\_\_\_\_

Forearm exercise test \_\_\_\_\_

EMG \_\_\_\_\_

NCS/ RNS \_\_\_\_\_

Imaging findings (send imaging data if possible)

Muscle imaging ( CT MRI ultrasound ) \_\_\_\_\_

Brain/ spine imaging ( CT MRI ) \_\_\_\_\_

EKG \_\_\_\_\_ Echocardiogram \_\_\_\_\_

Respiratory function VC \_\_\_\_\_ L % VC \_\_\_\_\_ % ( Sitting Supine )

Others \_\_\_\_\_

Record by: \_\_\_\_\_ Date: \_\_\_\_\_ [D/M/Y]

## Additional clinical information for **Myositis**

(When myositis is suspected, also fill this form)

Specimen No. (for NCNP use only)

### CLINICAL FINDINGS

**Skin lesion:** Gottron sign ( N / Y )      Mechanic's hands ( N / Y )      Heliotrope rash ( N / Y )  
Other ( N / Y \_\_\_\_\_ ) Skin biopsy ( N / Y \_\_\_\_\_ )  
**Motor system:** Myalgia ( N / Y )      Dysphagia ( N / Y )

### LABORATORY FINDINGS

**CBC:** WBC \_\_\_\_\_ / $\mu$ l      Hb \_\_\_\_\_ g/dl      Plt \_\_\_\_\_ / $\mu$ l

**Urinalysis:** Protein ( N / Y )

#### Antibody testing:

Anti ARS Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti Jo-1 Ab ( -/+ : titer \_\_\_\_\_ / not tested )  
Anti SRP Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti mitochondrial M2 Ab ( -/+ : titer \_\_\_\_\_ / not tested )  
Anti HMGR Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti TIF1 $\gamma$  Ab ( -/+ : titer \_\_\_\_\_ / not tested )  
Anti MDA5 Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti Mi-2 Ab ( -/+ : titer \_\_\_\_\_ / not tested )  
Anti NXP-2 Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti SAE Ab ( -/+ : titer \_\_\_\_\_ / not tested )  
Anti SS-A Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti SS-B Ab ( -/+ : titer \_\_\_\_\_ / not tested )  
Anti RNP Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti Scl-70 Ab ( -/+ : titer \_\_\_\_\_ / not tested )  
Anti DNA Ab ( -/+ : titer \_\_\_\_\_ / not tested )      Anti nuclear Ab titer \_\_\_\_\_ , type \_\_\_\_\_ / not tested

### EXTRAMUSCULAR MANIFESTATIONS

Interstitial pneumonia ( N / Y )      KL-6 \_\_\_\_\_      Cardiac abnormality ( N / Y \_\_\_\_\_ )  
Chest X-ray \_\_\_\_\_  
Chest CT \_\_\_\_\_  
Concurrent malignancy ( N / Y Diagnosis \_\_\_\_\_ )  
History of malignancy ( N / Y Diagnosis \_\_\_\_\_ Time of onset before myositis \_\_\_\_\_ )  
Other connective tissue disease ( N / Y symptoms and findings \_\_\_\_\_ )  
Statin use ( N / Y : currently , in the past ), provide the name and period of use \_\_\_\_\_  
(when the blood sample is submitted, please also provide the date of drawing blood \_\_\_\_ / \_\_\_\_ / \_\_\_\_ )  
History of COVID-19 vaccine inoculation ( N / Y, Number of inoculations \_\_\_\_\_ )  
Date of last inoculation \_\_\_\_\_ Name of COVID-19 vaccine \_\_\_\_\_ )  
Current use of steroid / immunomodulatory drugs ( N / Y \_\_\_\_\_ )  
Other active inflammation (including flu, allergy, and trauma) \_\_\_\_\_  
\_\_\_\_\_

## Additional clinical information for **FSHD**

(When FSHD is suspected, also fill this form)

Specimen No. (for NCNP use only)

**Family History** ( N / Y ) If yes (Y), draw a pedigree in the clinical summary form

### Symptoms:

Sleep with eyes half open ( N / Y ) since age \_\_\_\_ yrs      Difficulty closing eyes ( N / Y ) since age \_\_\_\_ yrs  
Difficulty whistling ( N / Y ) since age \_\_\_\_ yrs      Difficulty sucking with straw ( N / Y ) since age \_\_\_\_ yrs  
Difficulty closing mouth ( N / Y ) since age \_\_\_\_ yrs      Difficulty raising arms ( N / Y ) since age \_\_\_\_ yrs  
Difficulty placing things on overhead shelves ( N / Y ) since age \_\_\_\_ yrs  
Difficulty doing pullover on gymnastic bar ( N / Y ) since age \_\_\_\_ yrs  
Able to stand up from a chair without any difficulty ( N / Y )  
Able to stand up from a chair using hands to support ( N / Y )  
Able to walk but unable to stand up from a chair ( N / Y )  
Unable to walk ( N / Y )

### Physical findings:

Winged scapula ( N / Y )      Scapula elevation on shoulder abduction ( N / Y )      Beever sign ( N / Y )  
Ciliary sign ( N / Y )      Tongue atrophy ( N / Y )      Dysphagia ( N / Y )      Funnel chest ( N / Y )      Lumbar lordosis ( N / Y )

### <Muscle weakness/atrophy>

Orbicularis oculi	( Normal / R>L / R<L / R=L )	Quadriceps femoris	( Normal / R>L / R<L / R=L )
Orbicularis oris	( Normal / R>L / R<L / R=L )	Hamstrings	( Normal / R>L / R<L / R=L )
Biceps brachii	( Normal / R>L / R<L / R=L )	Tibialis anterior	( Normal / R>L / R<L / R=L )
Deltoid	( Normal / R>L / R<L / R=L )	Gastrocnemius	( Normal / R>L / R<L / R=L )
Pectoralis major	( Normal / R>L / R<L / R=L )	R>L: Weakness predominantly in the right	

### <Extramuscular abnormalities>

Audiometry ( ND / Normal / Impaired R \_\_\_\_ dB L \_\_\_\_ dB )      Retinal vessel abnormality ( N / Y )  
Others \_\_\_\_\_

**Association with autoimmune disease** ( N / Y \_\_\_\_\_ )

Anti-AChR Ab ( - / + : titer \_\_\_\_ / not tested )      Anti SRP Ab ( - / + : titer \_\_\_\_ / not tested )      Others ( \_\_\_\_\_ )

Other findings (Describe any findings that led to the clinical diagnosis of FSHD) \_\_\_\_\_