

【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 *Pediatric 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: _____

Family history:

Parental consanguinity (N / Y)

Neuromuscular disease (N / Y)

Perinatal period:

Maternal abnormality (N / Y)

Amniotic fluid (Normal, ↓ , ↑)

Fetal movement (normal, weak)

Family Tree**Neonatal history:**

Birth weight: _____ kg Body length: _____ cm GA: _____ weeks Perinatal asphyxia (N / Y)

Mode of delivery: normal delivery / forceps / vacuum extractor / cesarean section / other _____

Apgar score (1min/ 5min) _____ ☐ Hypoglycemia ☐ Abnormal electrolytes _____

Abnormal findings: ☐ Floppy ☐ Seizure ☐ Jaundice (mild moderate severe) ☐ Respirator use

☐ Tube feeding ☐ Poor suckling ☐ Weak crying ☐ Other _____

☐ Contractures (shoulder R / L, elbow R / L, wrist R / L, finger R / L, hip R / L, knee R / L, ankle R / L)

Developmental milestones (Age): Eye tracking () Head control ()

Sit without assistance () Crawl () Stand without assistance ()

Walk without assistance () Meaningful word () Two-word phrases ()

Physical examination: Date examined _____ [D/M/Y]

Body length/ height: _____ cm (SD) Body weight: _____ kg (SD)

Head circumference: _____ cm (SD) Chest circumference: _____ cm

Skin abnormality (N / Y Keloid Hyperextensibility Others _____)
 Spine deformity (N lordosis scoliosis) Chest deformity (N pigeon funnel) Hip dislocation (N / Y)
 Joint contracture (N / Y location _____)
 Joint hyperextensibility (N / Y location _____)
 Torticollis (N / Y) Limitation of neck movement (N / Y flexion extension other _____)
 High-arched palate (N / Y narrow wide) Cryptorchidism (N / Y) Minor anomaly (N / Y _____)
 Respiratory insufficiency (N / Y) Mechanical ventilation (N / Y since _____ NPPV or TPPV)
 Cranial nerve examination: Ptosis (N / Y) Ophthalmoplegia (N / Y) Facial weakness (N / Y)
 Dysarthria / dysphonia (N / Y _____) Dysphagia (N / Y)
 Motor system: Extremity weakness (N / Y proximal generalized distal asymmetrical)
 Neck weakness (N / Y) Muscle tone (normal ↓ ↑) Myotonia (N / Y _____)
 Muscle atrophy (N / Y _____) Winged scapula (N / Y _____)
 Muscle hypertrophy (N / Y GC tongue other _____)
 Deep tendon reflex (normal hypo- absent hyper- _____)
 Strength fluctuation (N / Y diurnal within a week within a month)
 Myalgia (N / Y site _____, time between exercise and onset of myalgia _____, after exercise)
 Maintaining sitting position (possible impossible)
 Difficulty standing up (N / Y Gowers' sign _____)
 Gait abnormality (N / Y _____ Non ambulant / wheelchair bound since age _____)
 Running (possible impossible)
 Cerebellar sign (N / Y _____) Involuntary movement: (N / Y _____)
 Sensory system (normal / abnormal _____)
 Intelligence (normal borderline low) IQ, DQ _____
 Seizure episode (N / Y _____) Stroke-like episode (N / Y _____)
 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 _____
 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)
 Fundus examination _____ 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 _____ / μ l Hb _____ g/dl Plt _____ / μ 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 γ 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) _____