

# 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

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

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 )

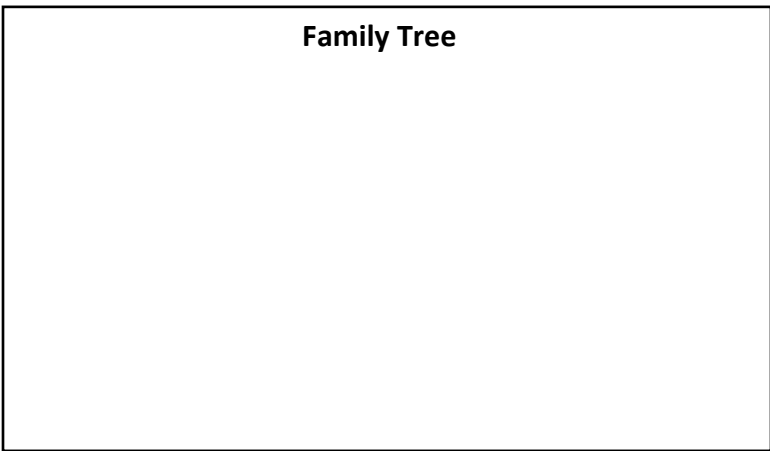
Neuromuscular disease ( N / Y )

## Perinatal period:

Maternal abnormality ( N / Y )

Amniotic fluid ( Normal, ↓ , ↑ )

Fetal movement ( normal, weak )



## 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 hyper- hypo - )  
Muscle atrophy ( N / Y \_\_\_\_\_ ); Winged scapula ( N / Y )  
Muscle hypertrophy ( N / Y gastrocnemius tongue other \_\_\_\_\_ )  
Fluctuation of muscle power: ( N / Y diurnal within a week within a month )  
Myalgia: ( N / Y site \_\_\_\_\_ time between exercise and onset of myalgia \_\_\_\_\_ )  
Maintaining sitting position ( possible impossible )  
Difficulty standing up ( N / Y Gowers' sign \_\_\_\_\_ )  
Gait abnormality ( N / Y \_\_\_\_\_ Non ambulant / wheelchair bound since age \_\_\_\_\_ )

**Deep tendon reflex:** ( normal hypo- absent hyper- \_\_\_\_\_ )

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

**Cerebellar sign:** ( N / Y \_\_\_\_\_ ); Involuntary movement: ( N / Y \_\_\_\_\_ )

**Intelligence:** ( normal borderline low ); IQ, DQ \_\_\_\_\_

**Seizure episode:** ( N / Y \_\_\_\_\_ ); **Stroke-like episode** ( N / Y \_\_\_\_\_ )

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

**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)

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### 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 )

### LABOLATORY FINDINGS

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

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

**Antibody testing:** Anti ARS Ab ( N / Y / not tested )                      Anti Jo-1 Ab ( N / Y / not tested )  
Anti SRP Ab ( N / Y / not tested )                      Anti mitochondrial M2 Ab ( N / Y / not tested )  
Anti HMGCR Ab ( N / Y / not tested )                      Anti TIF1 $\gamma$  Ab ( N / Y / not tested )  
Anti MDA5 Ab ( N / Y / not tested )                      Anti Mi-2 Ab ( N / Y / not tested )  
Anti NXP-2 Ab ( N / Y / not tested )                      Anti SAE Ab ( N / Y / not tested )  
Anti SS-A Ab ( N / Y / not tested )                      Anti SS-B Ab ( N / Y / not tested )  
Anti RNP Ab ( N / Y / not tested )                      Anti Scl-70 Ab ( N / Y / not tested )  
Anti DNA Ab ( N / Y / not tested )                      Anti nuclear Ab titer \_\_\_\_\_ / 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 \_\_\_\_/\_\_\_\_/\_\_\_\_)

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)

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**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 )      Beevor 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 ( N / Y )      Anti SRP Ab ( N / Y )      Others ( N / Y \_\_\_\_\_ )

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

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