

Specimen No. (for NCNP use only)

# Clinical Summary Form for *Pediatric Patient*

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 )

## 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 \_\_\_\_\_ )  
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 \_\_\_\_\_  
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 **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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