

Specimen No. (for NCNP use only)

Clinical Summary Form for *Adult 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: _____

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 gastrocnemius tongue other _____)

Muscle tone (normal decreased increased _____) Myotonia (N / Y _____)

Fluctuation of muscle power: (N / Y diurnal within a week within a month)

Myalgia: (N / Y site _____ time between exercise and onset of myalgia _____)

Limitation of neck movement (N / Y flexion extension rotation)

Deep tendon reflex: (normal decreased absent increased _____)

Sensory system: (normal / abnormal _____)

Autonomic system: (normal / abnormal _____)

Cerebellar sign: (N / Y _____); Involuntary movement: (N / Y _____)

Fundus examination _____ Others _____

Laboratory result:

CK at rest _____ (Normal range: _____); Aldolase _____ (Normal range: _____)

AST _____ ; ALT _____ ; CRP _____ ; ESR _____ ; ACE _____ ; M proteinemia (N / Y λ type κ type)

Serum lactate _____ (normal range: _____) CSF lactate _____

Serum pyruvate _____ (normal range: _____) CSF pyruvate _____

Autoantibodies _____

Infection HBV HCV HTLV-1 HIV Other _____

Endocrinological disease _____

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)

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) _____
