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:yrsmos)
Clinical diagnosis :	
	(Performed on:[D/M/Y]
Hospital*:	Department*:
Address*:	
*Use Japanese for Japanese hospitals	
	FAX:
	E-mail:
Present lliness:	
Family history:	Family Tree
Parental consanguinity (N / Y)	
Neuromuscular disease (N / Y)	
Perinatal period:	
Maternal abnormality (N / Y)	
Amniotic fluid (Normal, ψ , \uparrow)	
Fetal movement (normal, weak)	
Neonatal history:	
Birth weight: kg Body lengt	h: cm GA: weeks Perinatal asphyxia (N / Y
Mode of delivery: normal delivery / f	Forceps / vacuum extractor / cesarean section / other
Apgar score (1min/ 5min)	☐ Hypoglycemia ☐ Abnormal electrolytes
Abnormal findings: ☐ Floppy ☐ Sei	zure Jaundice (mild moderate severe) Respirator use
\square Tube feeding \square Poor suckling	□ Weak crying □ Other
☐ Contractures (shoulder R / L, elbo	ow 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 () C	rawl () Stand without assistance ()
Walk without assistance () N	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 Hyp	erextensibility Others

Joint contracture (N / Y location	,	, , , , , , , , , , , , , , , , , , , ,
Joint hyperextensibility (N / Y location		
Torticolis (N / Y) Limitation of neck movemen		
High-arched palate (N / Y narrow wide) Cry	ptorchidism (N / Y) N	/linor anomaly (N / Y)
Respiratory insufficiency (N / Y) Mechanical vo	entilation (N / Y since	NPPV or TPPV)
Cranial nerve examination: Ptosis (N/Y) Oph	nthalmoplegia (N / Y)	Facial weakness (N / Y)
Dysarthria / dyspho	nia (N / Y) Dysphagia (N / Y)
Motor system: Extremity weakness (N / Y pro	kimal generalized di	stal asymmetrical)
Neck weakness (N / Y) Muscl	e tone (normal $\downarrow \uparrow$	·) Myotonia (N / Y)
Muscle atrophy (N / Y) W	/inged scapula (N / Y)
Muscle hypertrophy (N/Y GC	tongue other)
		·)
Strength fluctuation (N / Y diu	rnal within a week w	vithin a month)
Myalgia (N / Y sitetim	ne between exercise a	nd onset of myalgia)
Maintaining sitting position (po		, ,
	•)
		eelchair bound since age)
Running (possible impossible)		°,
Cerebellar sign (N / Y)		t: (N / Y
Sensory system (normal / abnormal		
Intelligence (normal borderline low) IQ, D		
Seizure episode (N / Y		
Others		,
Laboratory result:		
CK at rest (Normal range:) Aldolase	(Normal range:
AST ALT		
Serum lactate (normal range: _		
Serum pyruvate (normal range: _		
Autoantibodies		
Forearm exercise test		
EMG		
Imaging findings (Send imaging data if possible)		
Muscle imaging (CT MRI ultrasound)		
Brain/ spine imaging (CT MRI)		
EKG/ Echocardiogram		
Respiratory function VC L % VC		
Fundus examination 2 70 ve		
. ddds Califfication		
Record by:		Date :[D/M/Y
necolu by.		[D/ N// I

Additional clinical information for *Myositis*

(When myositis is suspected, also fill this form)

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CLINICAL FINDING	GS				
Skin lesion: Gottr	ron sign (N / Y)	Mechan	nic's hands (N / Y)	Heliotrope rash (N / Y)
Other (N / Y) S	skin biopsy (N / Y)
Motor system: M	1yalgia (N/Y)	Dysphag	gia (N/Y)		
LABOLATORY FIN	IDINGS				
CBC: WBC	/μΙ	Hb	g/dl	Plt	/μΙ
Urinalysis: Prote	in (N / Y)				
Antibody testing	: Anti ARS Ab	(N/Y/not tes	ted) An	nti Jo-1 Ab	(N / Y / not tested)
	Anti SRP Ab	(N/Y/not tes	ted) An	nti mitochondrial N	12 Ab (N/Y/not tested)
	Anti HMGCR Ab	(N / Y/ not test	ted) An	nti TIF1γ Ab	(N / Y / not tested)
	Anti MDA5 Ab	(N/Y/not tes	sted) An	nti Mi-2 Ab	(N / Y / not tested)
	Anti NXP-2 Ab	(N/Y/not tes	ted) An	iti SAE Ab	(N / Y / not tested)
	Anti SS-A Ab	(N/Y/not tes	ted) An	nti SS-B Ab	(N / Y / not tested)
	Anti RNP Ab	(N/Y/not tes	ted) An	nti Scl-70 Ab	(N / Y / not tested)
	Anti DNA Ab	(N / Y / not tes	ted) An	nti nuclear Ab ti	ter/ not tested
EXTRAMUSCULA	R MANIFESTATION	IS			
Interstitial pneu	ımonia (N / Y)	KL-6	Cardiac ab	onormality (N / Y_)
Chest X-ray					
Chest CT					
)
History of malig	gnancy (N / Y Diagi	nosis	Time	e of onset before n	nyositis)
Other connectiv	ve tissue disease (N / Y symptoms a	and findings		
Statin use (N /	Y : currently , in th	e past), provide	the name a	nd period of use _	-
(when the bloo	d sample is submit	ted, please also p	provide the	date of drawing bl	ood/)

Current use of steroid / immunomodulatory drugs (N / Y ______)

Other active inflammation (including flu, allergy, and trauma)_____

Additional	clinical	information	for	ECHD
Addillonai	CIINICAI	iniormation	TOT.	ranij

(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	f open (N / Y) since age yrs	Difficulty closing eyes	(N / Y) since age yrs
Difficulty whistling	(N / Y) since age yrs	Difficulty sucking with	n straw(N/Y)since ageyrs
Difficulty closing mo	outh (N/Y) since ageyrs	Difficulty raising arms	(N / Y) since age yrs
Difficulty placing thi	ings on overhead shelves (N/Y	') since age yrs	
Difficulty doing pull	over on gymnastic bar (N / Y	') since age yrs	
Able to stand up fro	m a chair without any difficulty	(N / Y)	
Able to stand up fro	om a chair using hands to support	(N/Y)	
Able to walk but un	able to stand up from a chair	(N/Y)	
Unable to walk		(N/Y)	
Physical findings:			
Wingod scanula / N	/Y) Scapula elevation or	a ala a lala cala di alta a / Ni	I/V) Bower sign (N/V)
	•	·	nest (N/Y) Lumber lordosis (N/
Ciliary sign (N/Y)	Tongue atrophy (N/Y) Dysph	·	
Ciliary sign (N/Y)	Tongue atrophy (N/Y) Dysph	·	
Ciliary sign (N/Y) <muscle <="" td="" weakness=""><td>Tongue atrophy (N/Y) Dysphatrophy></td><td>nagia (N/Y) Funnel ch</td><td>nest(N/Y)Lumber lordosis(N /</td></muscle>	Tongue atrophy (N/Y) Dysphatrophy>	nagia (N/Y) Funnel ch	nest(N/Y)Lumber lordosis(N /
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