Clinical Data

Record ID: _____

Physician Name: _____

Clinic Name: _____

Region Name: _____

Last Exam Date: _____

Weight (kg): _____

Height (cm): _____

Diagnosis

Does the patient have an LGMD associated genetic mutation?
- ◯ Yes
- ◯ No
- ◯ Unknown

If YES, please provide the sequence: _____

Please specify the limb girdle genetic type:
- ◯ 1A ◯ 1B ◯ 1C ◯ 1D ◯ 1E ◯ 1F
- ◯ 1G ◯ 1H ◯ 2A ◯ 2B ◯ 2C ◯ 2D
- ◯ 2E ◯ 2F ◯ 2G ◯ 2H ◯ 2I ◯ 2J
- ◯ 2K ◯ 2L ◯ 2M ◯ 2N ◯ 2O ◯ 2Q

Were variants of unknown significance (VUS) present?
- ◯ Yes
- ◯ No
- ◯ Unknown

VUS filename: _____

If NO above, was a DBS (dry blood spot) test positive?
- ◯ Yes
- ◯ No
- ◯ Not done

What is the presenting phenotype?
- ☐ Proximal
- ☐ Distal
- ☐ Proximodistal
- ☐ Pompe
- ☐ MDC1C
- ☐ LAMA2
- ☐ Myoshi
- ☐ DMAT
- ☐ MED/WWS
- ☐ LGMD not yet classified
- ☐ Congenital myopathy
- ☐ Congenital muscular dystrophy (other)
- ☐ Metabolic myopathy
- ☐ Chronic myopathy (not yet specified)
- ☐ Scapuloperoneal

Clinical History

Age at diagnosis: _____

Age at symptom onset: _____

What brought the patient to medical attention? (check all that apply)
- ☐ Weakness
- ☐ Cramps
- ☐ Stiffness
- ☐ Myalgia
- ☐ Respiratory symptoms
- ☐ Cardiac Symptoms
- ☐ Myoglobinuria/rhabdomyolysis
- ☐ Elevated creatinine kinase (CK)
- ☐ Affected relative
- ☐ Other (specify): _____

Location of weakness:
- ☐ Proximal lower extremity
- ☐ Distal lower extremity
- ☐ Proximal upper extremity
- ☐ Distal upper extremity
- ☐ Other weakness (specify): _____

Do member(s) of the patient's family have similar symptoms or a similar diagnosis?
◯ Yes
◯ No
◯ Unknown

If YES, please specify: _____

Presence of consanguinity?
◯ Yes
◯ No
◯ Unknown

Has the patient participated in a clinical trial?
◯ Yes
◯ Past
◯ Never
◯ Unknown

If YES or PAST, please specify trial(s): _____

Diagnosis Tests

Has an EMG been done?
◯ Yes
◯ No
◯ Unknown

If YES, please specify:
◯ Normal
◯ Abnormal

Has a muscle biopsy been done?
◯ Yes
◯ No
◯ Unknown

If YES, please specify:
◯ Normal
◯ Abnormal

Date of muscle biopsy: _____

Muscle biopsy, laboratory name: _____

Muscle(s) biopsied:
☐ Arm, Deltoid
☐ Arm, Biceps
☐ Arm, Other
☐ Leg, Quadriceps (vastus lateralis)
☐ Leg, Gastrocnemius

Has an MRI been done?
◯ Yes
◯ No
◯ Unknown

If YES, please specify:
☐ Muscle ◯ Normal ◯ Abnormal Date of MRI: _____
☐ Cardiac ◯ Normal ◯ Abnormal Date of MRI: _____

Creatinine kinase (CK)

Maximum CK value: _____

Maximum CK date: _____

Maximum CK, specify:
◯ Normal
◯ Abnormal

Most recent CK value: _____

Most recent CK date: _____

Clinical Characteristics

Motor Function

Current motor function (select all that apply):
☐ Walks independently
☐ Walks with assistive devices
☐ Stands independently
☐ Sits independently
☐ Unknown

Best lifetime motor function:
◯ Walked independently
◯ Sat independently
◯ Unknown

Date of best lifetime motor function: _____

Age at which patient first walked (unit): _____ ◯ Months ◯ Years

Does the patient experience muscle aches/pains? ◯ Yes ◯ No ◯ Unknown

If YES, specify when (check all that apply): ☐ At rest ☐ During exercise ☐ After exercise

Are mobility aids used? ◯ Yes ◯ No

If YES above, indicate the use parameters of any mobility aids that the patient uses (check all that apply):

	Duration (Full-time, Part-time, or Intermittent):	Specific Age:
☐ Wheelchair	_____	_____
☐ Motorized scooter	_____	_____
☐ Walker/Walking frame	_____	_____
☐ Cane/stick	_____	_____
☐ Leg braces	_____	_____
☐ Knee splints	_____	_____
☐ Other	_____	_____

Does the patient experience any of the following conditions?

	Yes	No	Unknown
Facial weakness	◯	◯	◯
Dysphagia	◯	◯	◯
Ptosis	◯	◯	◯
Ophthalmoplegia	◯	◯	◯
Winged scapula	◯	◯	◯
Ocular myotonia	◯	◯	◯
Percussion myotonia	◯	◯	◯
Grip myotonia	◯	◯	◯
Hearing loss	◯	◯	◯
Contractures	◯	◯	◯

If contractures are present, select location(s): ☐ Elbow ☐ Wrist ☐ Finger ☐ Knee ☐ Ankle ☐ Hip ☐ Spine ☐ Other

Does the patient have scoliosis? ◯ Yes ◯ No ◯ Unknown

If YES, what is the Cobb angle/degree of curvature? _____

If YES, has surgery been performed? ◯ Yes ◯ Recommended ◯ Not yet indicated ◯ Unknown

Is neuropathy present? ◯ Yes ◯ No ◯ Unknown

Has a bone mineral density test been done? ◯ Yes ◯ No ◯ Unknown

If YES, was the result? ◯ Normal ◯ Osteopenic ◯ Osteoporotic ◯ Unknown

Cardiac

Does the patient have a muscular dystrophy-related heart condition?
- ◯ Yes
- ◯ No
- ◯ Unknown

If YES, please specify: _____

If YES, does the patient take medication for a heart condition?
- ◯ Yes, for muscular dystrophy-related condition
- ◯ Yes, for unrelated condition
- ◯ No
- ◯ Not applicable

If YES, please specify: _____

Is date of last Holter monitor known?
- ◯ Yes
- ◯ No

If YES, specify date: _____

ECG diagnosis: _____

ECG Date: _____

Echocardiagram date of test: _____

LVEF(%): _____

Laboratory: _____

Respiratory

Date of last PFT: _____

Sitting FVC (%predicted): _____

Supine FVC (%predicted): _____

Has a sleep study been done?
- ◯ Yes
- ◯ No
- ◯ Unknown

If YES, were results:
- ◯ Normal
- ◯ Abnormal

Ventilation

Non-invasive ventilation:

- ◯ Pre-existing (other diagnosis)
- ◯ Recommended
- ◯ Recommended & used
- ◯ Recommended & not used
- ◯ Recommended & declined
- ◯ Recommended & not tolerated
- ◯ Not yet indicated
- ◯ Not available
- ◯ Unknown

If YES/RECOMMENDED, enter the date first used/recommended: _____

If YES, check ventilation type and duration:

	Ventilation type		
Duration	CPAP	BiPAP	Sip & puff
Full-time	☐	☐	☐
Part-time - nocturnal only	☐	☐	☐
Part-time - nocturnal and periodic daytime	☐	☐	☐

Invasive ventilation:
- ◯ Yes, part-time
- ◯ Yes, full-time
- ◯ No
- ◯ Unknown

If YES, enter date first used: _____

Digestive Endocrine

Is dysphagia present?
- ◯ Yes
- ◯ No
- ◯ Unknown

If YES, select severity of dysphagia:
- ◯ Mild
- ◯ Moderate
- ◯ Severe
- ◯ Unknown

If YES, how was the dysphagia evaluated?
- ◯ Patient report
- ◯ Occupational therapist/speech-language pathologist
- ◯ Modified barium swallow (MBS)
- ◯ Fibreoptic endoscopic evaluation of swallowing (FEES)

If YES, has the patient had myomectomy surgery?
- ◯ Yes
- ◯ No
- ◯ Unknown

Major nutritional route:
- ◯ Oral
- ◯ Enteral

Medications And Vaccinations

Has the patient ever been on immunosuppressive therapy?
- ◯ Yes
- ◯ No
- ◯ Unknown

Has the patient ever taken a statin?
- ◯ Yes
- ◯ No
- ◯ Unknown

Has the patient received an annual flu vaccine?
- ◯ Yes
- ◯ No
- ◯ Unknown

Has the patient recived a pneumococcal vaccine in the last 10 years?
- ◯ Yes
- ◯ No
- ◯ Unknown

Date of last vaccine? _____

Access To Services Devices

Select the services that the patient accesses (check all that apply):
- ☐ Physiotherapy
- ☐ Pediatrician
- ☐ Speech language pathology
- ☐ Social work
- ☐ Physiatry
- ☐ Homecare
- ☐ Occupational therapy
- ☐ Respite care

Notes

Notes: _____