Congenital Muscle Disease
Clinical Questionnaire
Please complete and return with sample

Name: _____ **DOB** _____ **Date** _____

☐ **Male** ☐ **Female** **Ethnicity** _____

Suspected Diagnosis:

- ☐ **Congenital Myopathy**
- ☐ **Congenital Muscular Dystrophy**
- ☐ **Limb Girdle Muscular Dystrophy**
- ☐ **Emery Dreifuss Muscular Dystrophy**
- ☐ **Other (please specify)** _____

Growth

Gestational Age	_____wks
Birth weight	_____gms
Birth length	_____ cm
OFC	_____ cm
Age at Exam	_____ yrs & mos
Height	_____ cm
Weight	_____ kg
OFC	_____ cm

Features Present:	**Yes**	**No**
Muscle Biopsy Performed	☐	☐
Central nuclei	☐	☐
Cores	☐	☐
Nemaline rods	☐	☐
Nemaline rods and cores	☐	☐
Type 1 fiber predominance	☐	☐
Inflammation	☐	☐
Nonspecific	☐	☐
Other:_____	☐	☐
If Dystrophy present:		
Regeneration/degeneration	☐	☐
Inflammation	☐	☐
Fibrosis	☐	☐
Immunostaining Results		
Reduced or absent IHC	☐	☐
(specify antibody)_____		
Cognitive Delay	☐	☐
Severity _____		

Family History
(please describe)

Features Present:	**Yes**	**No**
Neuromuscular Findings		
Weakness:	☐	☐
(specify: facial, neck, distal, proximal)		
_____		
Ptosis	☐	☐
Foot drop	☐	☐
Muscular hypertrophy	☐	☐
Muscular atrophy	☐	☐
Ophthalmoparesis	☐	☐
Other neuromuscular findings: describe _____		
Cardiac Findings		
Cardiomyopathy	☐	☐
Cardiac Arrhythmia	☐	☐
Other cardiac findings: describe_____	☐	☐
Additional Findings:		
Contractures	☐	☐
Scoliosis	☐	☐
Feeding support (Gtube/NGtube)	☐	☐
Respiratory support (non-invasive/vent)	☐	☐
Other:_____		

LAB USE ONLY (Do not write in this box)