CORIELL INSTITUTE
FOR MEDICAL RESEARCH

NIGMS HUMAN GENETIC CELL REPOSITORY
CMD Biobank Neurology Clinical Data Elements Form

This form is to be filled out by the neurologist upon your next visit. Please email the completed form to NIGMS@coriell.org or fax to an NIGMS Repository Project Manager at 856-966-5067. For questions, please call the NIGMS Repository Genetic Counselor at 856-757-4822.

Dear Neurologist,

Your patient has recently submitted a sample to the Congenital Muscle Disease (CMD) Biobank at the Coriell Institute for Medical Research. We would like to ask you several questions. We will link this de-identified information with the biobank sample. We have partnered with Cure CMD to link to the CMD International Registry (CMDIR) to make additional de-identified data available, including age of onset, maximal motor function, pulmonary capacity and muscle/skin/genetic disease confirmation, when available.

1. Does this patient have a clinical diagnosis of congenital muscle disease (CMD)?
    Please check one:
    ☑ Congenital myopathy ☐ Congenital muscular dystrophy
    ☐ Congenital myasthenic syndrome ☐ No history of congenital muscle disease ☐ I do not know

2. What is the patient's diagnosis? _____

3. How has the diagnosis been confirmed? *Please check all that apply:*
    ☑ Family history ☑ Creatine kinase level (blood test):_____ ☑ Muscle biopsy ☑ Skin biopsy
    ☑ Brain MRI ☑ Electrocardiogram (ECG) ☑ Electromyography (EMG)
    ☑ Muscle imaging (MRI or ultrasound)
    ☑ Genetic testing - gene(s) tested: _____
    Mutations identified (please attach copy of report): _____

4. Has the patient had an MRI or CT scan of the brain? ☐ Yes ☐ No ☐ I do not know
    If yes, what was the result of the MRI or CT scan of the brain?
    ☑ Normal ☑ Abnormal ☑ I do not know
    If abnormal, what was the finding? *Please check all that apply*:
    ☑ Lissencephaly ☑ Hydrocephalus ☑ Corpus callosum involvement ☑ Polymicrogyria
    ☑ Cerebellar or brainstem hypoplasia ☑ Cerebellar cysts ☑ White matter change
    ☑ Other, please specify: _____

Motor Function	Achieved?		Maintained?	
Holding head up without assistance	☑ Yes	☑ No	☑ Yes	☑ No
Sitting without assistance	☑ Yes	☑ No	☑ Yes	☑ No
Walking without assistance	☑ Yes	☑ No	☑ Yes	☑ No
Running	☑ Yes	☑ No	☑ Yes	☑ No