

Instructions for biopsy sample collection and completing CMD BioBank donation paperwork:

1. **DO NOT FREEZE!!!** Please keep the biopsy sample at room temperature and ship at **room temperature** –ideally on the day that the biopsy is taken—via overnight delivery for receipt at Coriell Monday-Friday. We need a 3 or 4mm skin punch biopsy. It is okay to refrigerate the biopsy tube until it is shipped.
2. **Informed Consent Form** – Please complete all items on Page 4. The submitter information can be left blank, unless your or your child’s physician is a researcher. If he or she is a researcher, the physician may be listed as the submitter. This will entitle him/her to a free cell line or DNA sample in exchange for each sample submitted.
3. **Assent Form** – Children ages 7-17 may complete this form if intellectually capable.
4. **Submission Form** – Complete pages 1-2 of the Submission Form. Leave pages 3-4 blank, unless your/your child’s physician is a researcher. If he/she is a researcher, please have him/her complete pages 3-4.

Submitting Investigator – If your/your child’s physician is a researcher, you may enter their information here.

Diagnosis – Enter the diagnosis of the affected individual on all family members’ Submission Forms.

OMIM# – Leave blank.

Sample ID # – Provide donor’s name here. All identifying information is removed and replaced with a number.

Date of sample collection – Enter date of biopsy.

Age at sample collection – Enter donor’s current age.

Gender, Race, Ethnicity, and Ancestry of the donor – Complete all items.

Source of clinical information- We will be matching up samples to data recorded in the Congenital Muscle Disease International Registry. If you/your child are registered in the CMDIR, please mark “Other “and write “CMDIR”. The more clinical information we can get, the more valuable the samples are to scientists who will be accessing them. If you have copies of genetic test reports, please enclose them as this information is especially valuable-but not required. If you haven’t been tested, or if mutations haven’t been identified, we are still able to accept the samples.

Family History – If other family members have the same diagnosis as the affected individual, please explain who the other family members are and their relationship to the affected individual.

Have samples been submitted by other family members? Please mark “Yes” if more than one person from your family is donating a sample. In the box, indicate who else has submitted a sample in relation to the donor. For example: On your child’s Submission Form, indicate “mother and father” if both parents are also donating.

Literature References – If your/your child’s case has been described in the medical literature, please provide the journal name, year of publication and first author name if known.

5. **Neurology CDE Form** – Please have your/your child’s neurologist complete this form and fax it to: 856-964-0254, ATTN: NIGMS Project Manager

Please contact Tara Schmidlen, MS, CGC at 856-757-4822 or tschmidl@coriell.org with questions.

THANK YOU FOR DONATING A SAMPLE TO THE CMD BIOBANK!