MTM1 CARRIER SURVEY

GOAL/OBJECT OF STUDY

The Beggs Lab invites any mother who has a child genetically diagnosed with congenital myopathy, including *MTM1* or other genes, to complete a questionnaire about their medical history and current abilities. Researchers are looking to survey *MTM1* carriers, women who are not *MTM1* carriers, but have a son with XLMTM, and women who have a child with congenital myopathy due to a change in a gene other than *MTM1*, such as *DNM2, RYR1, BIN1,* etc.

We hope to learn more about what symptoms, if any, mothers of children with congenital myopathies may have, to be better able to characterize the impact of MTM1 mutations on the body.

STEPS FOR PARTICIPATION

Women who choose to complete this questionnaire will be asked to answer questions about their past medical history and current health status during a telephone interview. This call will take between 30 to 45 minutes and can be scheduled at your convenience. We will send the questionnaire to you in advance if you would like to review the questions prior to our scheduled phone call.

GET INVOLVED

If you **are enrolled in the Beggs Lab** and would like to complete this questionnaire for *MTM1* carriers, please contact Casie Genetti, the genetic counselor for the Beggs Lab, to arrange a time for the telephone interview.

If you **are not previously enrolled in the Beggs Lab** and would like to participate in this questionnaire, please contact Casie to discuss how to get involved as a participant in the Beggs Lab.

If you have any questions about our research or myotubular myopathy in general, please contact:

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