

Clinical Center Genomics Opportunity (CCGO)

The National Institutes of Health (NIH) has free genetic testing slots available for individuals with undiagnosed muscle conditions. This can include people with Titin variants of unknown significance.

Carsten Bönnemann, M.D., National Institute of Neurological Disorders and Stroke, will use exome sequencing to explore the diversity of gene mutations and phenotypes in a well-defined, pediatric, neuromuscular disorder cohort.

Cure CMD and the Congenital Muscle Disease International Registry will assist with recruitment in the CMD community for this opportunity.

Eligibility:

- Symptoms of a muscle condition that started in childhood as demonstrated by medical records
 - Previous genetic testing that has resulted in Negative or VOUS results **(1)**
 - **Not** previously evaluated through the NIH Muscle Research Program (Carsten Bönnemann, M.D.)
 - Be able to travel to the NIH in Bethesda, Maryland for a 2 day comprehensive evaluation; travel and lodging expenses will be covered by the NIH **(2)**
 - Eligibility must be determined and visit scheduled before September 30, 2016 (visit may occur after this date)
- (1) A comprehensive work-up as demonstrated by medical records, with multiple genes tested and returned with negative or VOUS findings. (VOUS-Variance of Unknown Significance).
- (2) Travel from within the United States and Canada is covered. If you reside in another country, you will be responsible for travel to a port of entry within the United States first, and then travel and lodging will be covered.

How to Enroll:

- Ensure you are registered with the Congenital Muscle Disease International Registry (CMDIR)
 - You can register at www.cmdir.org
 - If you have already registered, **please do not register again**. Request your login information by emailing counselor@cmdir.org
 - Submit prior genetic testing showing negative or VOUS findings as well as a signed medical release **(3)** to the CMDIR via any of the following:
 - Email: counselor@cmdir.org
 - Fax: +1-310-684-2023
 - Secure Upload: <https://www.dropbox.com/request/rvxGaCN1tVU2HmYOVLfR>
 - Mail: 19401 S. Vermont Ave, Suite J100
Torrance, CA 90502 USA
 - Eligibility will be determined based on medical records and responses to an intake survey. If you are eligible, records will be provided to you in electronic format to forward directly to the NIH.
- (3) To ensure a faster determination of eligibility, please provide additional medical records directly. These may include: muscle biopsy report, clinic notes, hospital discharge summaries, pulmonary function testing, sleep studies, lab tests (blood work), echocardiograms and EKG's. At minimum, you must provide genetic testing results to be considered for eligibility. If you live outside the United States, you will be required to provide all necessary medical records as we will be unable to do so on your behalf in a timely manner.