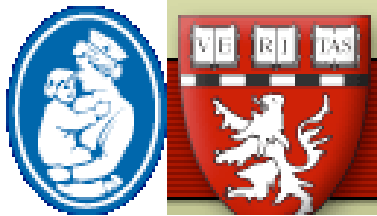


GENETIC RESEARCH ON THE CONGENITAL MYOPATHIES

Laboratory of Alan H. Beggs, Ph.D.

Children's Hospital Boston
The *first* place for children



HARVARD
MEDICAL SCHOOL

Centronuclear/Myotubular Myopathy: Information for Patients and Families

Introduction

We are a group of scientists and doctors studying myotubular (MTM) and centronuclear myopathy (CNM) at the Children's Hospital/Harvard Medical School in Boston, Massachusetts. We are trying to understand the changes that occur in the muscle of patients with MTM/CNM, with the hope that this will lead to improved treatment for these conditions. In order to carry out our research, we need the help of families. If you are the parent of a child with CNM/MTM, or if you yourself are affected, you may be able to make an important contribution toward scientific knowledge of these and other muscular conditions.

We currently know of at least three genes associated with CNM/MTM. The first gene to be discovered was the myotubularin gene (MTM1) on the X chromosome. This gene is associated with X-linked myotubular myopathy, which typically only affects boys. There are at least two other known genes, termed autosomal genes, responsible for causing CNM that affects both boys and girls equally. The dynamin 2 (DNM2) gene is associated with dominant CNM, and the BIN1 gene is associated with a small number of cases of recessive CNM. Our research is focused on trying to learn more about these genes and to locate other disease causing genes. We know that each of these genes carry special instructions to make a protein that is important for muscle function. When altered by a "typo," these instructions can result in a non-working product, causing MTM or CNM.

If you are the parent of a child with MTM or CNM, or if you yourself are affected, you may be able to make a meaningful difference by helping us understand the causes of these myopathies.

Participation consists of:

Informed Consent

Each family member who decides to participate will need to sign a consent form. If the participant is a minor, a parent/guardian will be the one who provides consent.

Medical information and family history

We will ask your permission to obtain relevant medical records, such as a muscle biopsy report, from your physician. We may also ask you some questions about your family medical history. This can be done through a brief telephone interview.

Blood sample

We ask for a blood sample from all available and consenting family members. This blood sample will be used to isolate genetic material (DNA). The DNA will be screened for mutation (changes) in genes that may be involved in myotubular and centronuclear myopathy. We can arrange the blood draw through either a participant's physician or a nearby medical facility. All costs for the blood draw are paid by our lab.

GENETIC RESEARCH ON THE CONGENITAL MYOPATHIES

Elizabeth Taylor DeChene, MS, CGC
Genetic Counselor

Alan H. Beggs, PhD
Lab Director

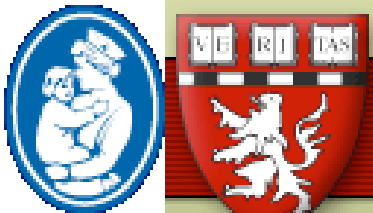
Division of Children's Hospital Boston
300 Longwood Avenue
Enders Building, 5th Floor
Boston, MA 02115
Phone: 617-919-2169 • Fax: 617-730-0786
Email: edechene@enders.tch.harvard.edu
Web Site:
www.childrenshospital.org/research/beggs/

GENETIC RESEARCH ON THE CONGENITAL MYOPATHIES

Laboratory of Alan H. Beggs, Ph.D.

Children's Hospital
Boston

The *first* place for children



HARVARD
MEDICAL SCHOOL

Muscle tissue from an existing muscle biopsy

Studying muscle from a person who has myotubular/centronuclear myopathy can tell us a lot about the genes and proteins involved in the disease. We can help find out if any frozen tissue is still available from an existing muscle biopsy and, with your permission, arrange to have it shipped to our laboratory. Alternatively, if you or your child is scheduled to undergo a surgical procedure in the near future, this may provide an opportunity to donate a muscle specimen. With some procedures, it is possible for the surgeon to remove a small piece of muscle without any additional risk or discomfort to the patient.

Cost and time commitment:

Participation in this study is free of charge. Travel to Boston is not required and individuals from anywhere in the world may participate. The telephone interview, blood draw, and paperwork should take no more than 2 hours to complete.

Reporting of Results:

It is possible that we may identify a gene mutation as the cause of the muscle disease in your family. In this case, and with your permission, we will be happy to make this information available to you via your healthcare provider. As a research laboratory, we are not authorized to release participant's results, but we can refer your physician to a clinical diagnostic laboratory that would confirm our findings and provide clinically useful results.

Contact us:

If you would like to enroll or if you have questions about this study, please contact:

Elizabeth Taylor DeChene, M.S. C.G.C., Genetic Counselor
Program in Genomics
Enders Building, floor 5
300 Longwood Avenue
Boston, MA 02115
Telephone: 617-919-2169
Fax: 617-730-0786
Email: edechene@enders.tch.harvard.edu

We look forward to hearing from you!