GENETIC RESEARCH ON THE CONGENITAL MYOPATHIES

Laboratory of Alan H. Beggs, Ph.D.

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

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Congenital Myopathies:
Information for Patients and Families

Introduction
In the Beggs laboratory at Boston Children's Hospital, we are studying the genetics of the congenital (present from birth) myopathies (muscle diseases). The congenital myopathies are rare genetic disorders that result in muscle weakness of variable severity. Diagnosis is usually made from a muscle biopsy. Some patients are affected from birth, while others develop symptoms in childhood or adulthood. These conditions often occur in individuals who have no prior family history of neuromuscular disease. In other cases, more than one member of a family may be affected.

In the Beggs Laboratory, we are trying to understand the genes and proteins involved in the congenital myopathies. Genes are pieces of inherited information that tell our bodies how to grow and develop. There are genes for hair color, eye color, and even how your muscles work. Sometimes individuals inherit a gene that does not work properly, causing them to develop health problems or disease. We hope that what we learn will be useful for improving diagnosis and treatment of these disorders. If you are the parent of a child with a congenital myopathy, or if you yourself are affected, you may be able to make an important difference in our research.

We are studying many types of congenital myopathies:

- Nemaline myopathy
- Central Core Disease
- Myotubular/centronuclear myopathy
- Congenital fiber type disproportion
- Multicore/minocore myopathy & Rigid Spine Muscular Dystrophy
- Congenital myopathies with non-specific muscle biopsy findings

Participation consists of:
Research Consent: Each family member who decides to participate will need to review and complete the consent form with our research study coordinator and/or a local clinician. 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 or saliva sample: We ask for a blood or saliva sample from all available and consenting family members. This sample will be used to isolate the genetic material (DNA). The DNA will be screened for mutations (changes) in genes that may be involved in the disease in your family. 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 laboratory.

Muscle tissue from an existing muscle biopsy: Studying muscle from a person who has a congenital 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

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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.

Potential Resources
There are several potential resources for individuals and families with congenital myopathies:

- The **Muscular Dystrophy Association** (www.mdausa.org) and **Muscular Dystrophy Campaign** (http://www.muscular-dystrophy.org) provide general information about various topics related to neuromuscular conditions.
- The **myopathy mail group** at (health.groups.yahoo.com/group/Myopathy) is an on-line listserv and support group composed of individuals with all different forms of neuromuscular conditions. It is important to realize that healthcare professionals do not generally oversee mail groups. Therefore, some of the information you receive may not pertain to your family.
- Our lab may also be a helpful resource. Our website provides general information about congenital myopathies and genetics (http://www.childrenshospital.org/research/beggs). We may also be able to connect your family with other individuals or families with congenital myopathies. If this possibility interests you, please contact Lindsay Swanson.

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

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We look forward to hearing from you!