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Genetic Testing for Charcot Marie Tooth Disease

Information for Patients and Families

What do I need to know about testing myself or my child for Charcot Marie Tooth disease?

Charcot Marie Tooth disease (CMT) is a condition caused by a change in someone's DNA. People with CMT have a problem with their nerves that causes weakness of the distal muscles (those closest to their hands and feet) and decreased feeling. This blood test may prove that you or your child has CMT. It may also find something that we do not understand. We may need to test the individual's parents to learn more. This sheet will provide more details about CMT and this testing. If you have more questions, please talk to a genetic counselor.

What is Charcot Marie Tooth Disease?

Charcot Marie Tooth disease (CMT) is a rare genetic condition. People with CMT have a problem with their nerves that causes progressive weakness of the distal muscles (those closest to their hands and feet) and decreased feeling. They can also have high-arched feet and decreased reflexes.

What causes CMT?

CMT can be caused by a change (mutation) in a number of different genes, including the *DNM2* gene. Everyone has two copies of the *DNM2* gene. Genes are instructions to make proteins. When there is a change in the instructions, the protein may not be made or may not work properly. If a person has a change in one copy of the *DNM2* gene, he or she will not be able to make enough normal protein and will have CMT.

How does CMT run in families?

CMT caused by changes in the *DNM2* gene is autosomal dominant. This means that they have one normal copy of the *DNM2* gene and one with a change. Thus, people with this form of CMT have a 50% chance of passing it down to each child.

Can my child be tested? Can I be tested? Can my family members be tested?

The first person to be tested should be the person with CMT. Testing for mutations in *DNM2* is complex. It is like reading a book and looking for spelling mistakes. You may read the whole book and miss the "typos," however when you do find them, then it is easy to test other family members (i.e. you know that the change is on page 200 in the second paragraph). When changes in *DNM2* are found in the person with CMT, testing other family members, even during a pregnancy, is easy and fast.

Reasons for genetic testing for CMT:

- confirm the diagnosis
- check if other family members are carriers
- provide information and resources for future pregnancies
- provide information during a pregnancy regarding possible CMT in the baby

What does it mean for myself or my child if they find a mutation? What does it mean for our family?

Finding a change in the *DNM2* gene in a patient with possible CMT confirms the diagnosis of CMT and allows other family members to get testing.

What does it mean if they don't find a mutation?

If someone does not have any changes in *DNM2*, then his/her CMT is most likely not caused by problems in *DNM2*. This person may have CMT caused by a change in another gene.

What does it mean for my child if they find a variant of unknown significance?

A small number of patients will have a change in the gene, but we are not sure whether that change causes CMT or not. In this case, we recommend testing parents to give us more information.

How do I get my child tested?

We recommend that a neuromuscular or genetic specialist help you order the test for your child. If you think that you or your child has CMT, you should see a neuromuscular or genetic specialist. Your doctor or hospital can help you set this up. This neuromuscular or genetic specialist can order the testing. If there are any questions about ordering the testing, please ask the physician or genetic counselor to contact The University of Chicago Genetics Services Laboratory. A blood sample is required for testing.

How much does the testing cost and will my child's health insurance cover it?

Cost for *DNM2* sequencing is $1560, cost for *DNM2* deletion/duplication testing is $1000. All insurance companies are different, but most of them should cover at least part of the cost of testing. We recommend that you contact your insurance company to learn more about your coverage prior to testing. You should ask your insurance company what your coverage is for the following CPT (Current Procedural Terminology) codes: 81406 for sequencing, and 81405 for deletion/duplication testing. Insurance companies use these codes to define the method of testing. The University of Chicago or your hospital or lab will bill your child's insurance company. You may receive a bill for any amount not covered by your insurance company, i.e. copayment, deductible, etc. If you do not have medical insurance, you will need to pay by check or credit card before the lab will start testing.

If a mutation is found in your child, testing of other family members is $390 and testing during a pregnancy is $540. The CPT code for these tests is 81403.

When/how will I get the results?

Testing takes approximately 4-6 weeks. Results will be faxed and mailed to your doctor.

What happens to the information from this test?

Your doctor will send a form about you/your child's symptoms with the blood sample. This will help the lab understand the test result. The symptoms and test results will be put into a public database after removing the name and all identifying information. Information from people with CMT will increase what we know about this condition and the genetic test.

How can I meet or talk to other families?

Support groups are a great way to talk to other families that have gone through similar experiences. The CMT Yahoo Support Group (http://groups.yahoo.com/--search under charcot marie tooth) is an online group for families to chat about their lives with CMT. Please remember that all people with CMT are not the same, so their symptoms may be different from yours (your child's). Also note that the information given on this type of website is not written by a physician, so all information may not be 100% correct. It is, however, a great place to share ideas and feelings.

Additional Resources:

Charcot Marie Tooth Association
700 Chestnut Street
Chester PA 19013-4867
Phone: 800-606-2682; 610-499-9264
Email: info@charcot-marie-tooth.org
http://www.charcot-marie-tooth.org/

The Muscular Dystrophy Association
3300 E. Sunrise Drive
Tucson, AZ 85718
800-572-1717
http://www.mda.org/

European Charcot-Marie-Tooth Consortium
Department of Molecular Genetics
University of Antwerp
Belgium
Fax: 03 2651002
Email: gisele.smeyers@ua.ac.be

The Hereditary Neuropathy Foundation
1751 2nd Ave Suite 103
New York NY 10128
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www.hnf-cure.org

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