The University of Chicago Genetic Services Laboratories

5841 S. Maryland Ave., Rm. G701, MC 0077, Chicago, Illinois 60637
Toll Free: (888) UC GENES (888) 824 3637
Local: (773) 834 0555 FAX: (773) 702 9130
ucgslabs@genetics.uchicago.edu dnatesting.uchicago.edu
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Testing for Allan-Herndon-Dudley Syndrome

Information for Patients and Families

What do I need to know about testing my child for Allan-Herndon-Dudley syndrome?

Allan-Herndon-Dudley syndrome is caused by a change in someone's DNA. People with this disorder have developmental delay, poor head control, low muscle tone, and unclear or no speech. This blood test may prove that your child has Allan-Herndon-Dudley syndrome. There is also a chance that the test will find something that we do not understand. Thus, we may need to test the child's parents to learn more. This information sheet will provide more details about Allan-Herndon-Dudley syndrome and this testing. Please talk to a genetic counselor, if you have more questions about testing.

What is Allan-Herndon-Dudley syndrome?

Allan-Herndon-Dudley syndrome is a rare genetic condition. Common findings in these children include: developmental delay, poor head control, low muscle tone, and unclear or no speech. Not everyone with this disorder has all of the findings or is affected to the same degree. Patients (generally, males) with this condition also have a thyroid hormone problem that is less severe in their mothers.

What causes Allan-Herndon-Dudley syndrome?

Allan-Herndon-Dudley syndrome is caused by a change (mutation) in the *SLC16A2* (also known as *MCT8*) gene. Females have two copies of the *SLC16A2* gene, and males have one copy. 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 male has a change in his only copy of the *SLC16A2* gene, he will not be able to make the normal protein and will develop the findings of Allan-Herndon-Dudley syndrome. Females with a change in one copy of the *SLC16A2* gene will also have a normal copy of the gene. Thus, they will not have any signs of the disorder, but will have a 50% chance of passing it on to their sons.

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

The first person to be tested in any family is the individual thought to have Allan-Herndon-Dudley syndrome. Testing for this condition requires thyroid hormone testing as well as gene testing. Two tubes of blood are needed. One tube will be tested for the thyroid hormone problem. If this is abnormal, the other tube will be used for gene testing. This testing is like reading a long book and looking for a single spelling mistake. You may read the whole book and miss the "typo," however when you do find it, then it is easy to test other family members (i.e. you know that the change is on page 875 in the second paragraph). Once a change is found in the person with Allan-Herndon-Dudley syndrome, testing other family members, even during a pregnancy, is easy and fast. Both of these tests are now available at The University of Chicago Genetics Services Laboratory.

Reasons for genetic testing for Allan-Herndon-Dudley syndrome:

- confirm the diagnosis
- reassure that other family members are not affected
- provide information and resources for future pregnancies
- provide information during a pregnancy regarding possible Allan-Herndon-Dudley syndrome in the baby

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

Finding a mutation will confirm a diagnosis of Allan-Herndon-Dudley syndrome. Once a change has been found in an individual, then other family members may have testing, if they want to.

What does it mean for my child if they don't find a thyroid problem or a mutation?

If your child does not have thyroid abnormalities, then he does not have Allan-Herndon-Dudley syndrome. If he/she do have a thyroid hormone problem, the lab will do genetic testing. If the lab does not find a mutation,

your child may still have Allan-Herndon-Dudley syndrome. Dr. Refetoff is conducting research to improve early testing methods and look for other causes of Allan-Herndon-Dudley syndrome or similar conditions. Please contact him at (refetoff@uchicago.edu; 773-702-6939) for more information about these studies.

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 Allan-Herndon-Dudley syndrome or not. We, then, recommend testing parents to give us more information.

How do I get my child tested?

We recommend that a geneticist or genetic counselor help you order the test for your child. If you think that your child has Allan-Herndon-Dudley syndrome, you should see a genetics specialist. Your physician or hospital can help you set this up. This genetics specialist can order the testing for Allan-Herndon-Dudley syndrome. 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. Two tubes of blood are required for testing.

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

Cost for thyroid testing only (Tier 1) is $350 and *SLC16A2* sequencing (Tier 2) is $1200. Cost for deletion/duplication of *SLC16A2* 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: 84436, 84481, 84443, 84482 for thyroid testing (Tier 1), 81405 for sequencing (Tier 2) and 81403 for deletion/duplication analysis. 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 codes for these tests are 81403.

When/how will I get the results?

Thyroid testing takes about 2 weeks. If a thyroid abnormality is found, *SLC16A2* gene testing takes approximately 2-4 weeks. Results from each test will be faxed and mailed to your doctor as soon as they are complete.

What happens to the information from my child's test?

Your physician will send a form about your child with the blood sample. This information will help the lab understand your child's test result. Your child's findings and test results will be put into a public database after removing your child's name and all identifying information. This information from children with Allan-Herdon-Dudley syndrome will increase what we know about this disorder and the genetic test. You may call the lab if you have any questions about this.

Can we still participate in research studies?

Yes, your child (family) can participate in research studies. Now that we know some information about the cause of Allan-Herndon-Dudley syndrome, we can begin to learn more. The University of Chicago Labs will be working together to compare the results of testing and the features of the patients.

Additional Resources:

SLC16A2 (MCT8) Organization
www.mct8organization.org