



## The University of Chicago Genetic Services Laboratories

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CLIA #: 14D0917593 CAP #: 18827-49

### Ataxia Exome Panel

#### What do I need to know about testing for ataxia?

Ataxia can be caused by a change in your DNA. This test may provide the underlying hereditary (genetic) basis of you or your child's ataxia. This information sheet was sent to you because you have an upcoming appointment with Dr. Christopher Gomez at the University of Chicago and it has been determined that this testing may be appropriate for you to consider. This information sheet will provide more details about ataxia and this testing to prepare you to discuss this testing option at your upcoming appointment.

#### What is ataxia?

Ataxias are a group of neurological conditions that can affect individuals of all age groups. Commonly ataxia is characterized by incoordination of voluntary movements. This can result in difficulties or disturbances of stance, gait, eye movements, muscle tone, skilled movements and speech. Not everyone with ataxia has all of the findings or is affected to the same degree.

#### What causes ataxia?

Ataxia can be caused by hereditary (genetic) and non-hereditary (acquired) factors. The following test is not for individuals with a suspicion of a non-hereditary (acquired) form of ataxia. In order to explain the causes of hereditary (genetic) ataxia, it is important to review some genetic concepts. As you may be aware, our genes are what determine the way we grow and develop. Each person has approximately 20,000 genes. Hereditary ataxias can be caused by a change (mutation) in a gene. There are many different genes that are implicated in ataxia and many ways that it can be inherited. The UCGS Ataxia Exome Panel analyses 462 genes involved in ataxia that were assembled by research and clinical experts in the field. We recommend talking with your physician regarding the hereditary causes of ataxia prior to ordering this test.

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

The diagnosis of ataxia requires detailed family and medical history, physical examination, neuroimaging, and an increasing number of laboratory and immunological tests. Genetic testing for common genetic causes of ataxia may already have been performed on you or your child. Our Ataxia Exome Panel analyzes over 300 genes implicated in ataxia; however it is most appropriate to order this test after you've already had genetic testing for the very common causes of hereditary ataxia. Please consult with your physician regarding what genetic testing, if any, has already been performed.

The first person to be tested in any family would be the individual with ataxia. Testing for ataxia is complex. It is like reading a very 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 ataxia, testing other family members, even during a pregnancy, is easy and fast because we know where to look.

#### How do I get myself (or my child) tested?

Testing can be coordinated at your upcoming appointment with Dr. Christopher Gomez at the University of Chicago. If you are interested, we recommend that you discuss this with Dr. Gomez to determine if genetic testing is warranted and help you order the test. The test requires 3 – 5 ml (1/2 to 1 teaspoon) of blood which can be drawn at the time of your appointment.

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

Cost for the Ataxia Exome Panel is \$4800. All insurance companies are different. You should contact your insurance company to learn more about your coverage prior to testing. You will want to ask your insurance company what your coverage is for the following CPT (Current Procedural Terminology) codes: 81403, 81404, 81405, 81406, 81407, 81408. Insurance companies use these codes to define the method of testing. Your insurance company may also ask you for an ICD-9 code. In this case, it will typically be 334.2. It is important to determine whether your insurance company will cover the complete cost, some of the cost, or none of the cost of

the testing prior to your appointment with Dr. Gomez at the University of Chicago. In some cases, your insurance company will request a 'preauthorization' of testing or require additional information from your physician to determine whether they will cover testing.

In most cases, The University of Chicago will bill your insurance company. You may receive a bill for any amount not covered by your insurance company, i.e. co-payment, deductible, etc. In some cases, The University of Chicago cannot bill your insurance company. In this case, we will need payment from you by check or credit card before testing.

**What if I have Medicare? Will Medicare cover this test?**

This test is not covered by Medicare. If you have Medicare and are interested in having this test performed, we will need payment from you by check or credit card before testing.

**When/how will I get the results?**

Testing takes approximately 12-16 weeks. Your physician will be informed of the results as soon as it is complete. Results will be faxed to your physician.

**What does it mean if they find a change? What does it mean for our family?**

Finding a change will confirm the genetic cause of a person's ataxia. Once a change has been found in an individual, then other family members may be tested, if they choose to.

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

Not finding a change does not mean that you or your child does not have ataxia. We are only able to find changes in about a proportion of people with ataxia. Researchers are working to improve our testing.

**What does it mean if they find a variant of unknown significance?**

A number of patients will be found to have a change in a gene, but we are not sure whether that change causes ataxia or not. It is not uncommon for a patient to have a number of variants of unknown significance. As research into the genetics of ataxia improves, more information may become available about these variants in the future.

*Committed to CUSTOMIZED DIAGNOSTICS, TRANSLATIONAL RESEARCH & YOUR PATIENTS' NEEDS*