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Genetic Testing for Robinow Syndrome

Information for Patients and Families

What do I need to know about testing my child for Robinow Syndrome?

Robinow syndrome (RS) is a condition caused by a change in someone's DNA. People with RS are small for their age, have some bone problems, look more like each other than their family members, and may have small genitalia. This blood test may prove that your child has RS. 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 RS and this testing. If you have more questions, please talk to a genetic counselor.

What is Robinow syndrome (RS)?

Robinow syndrome (RS) is a rare genetic condition. As with other syndromes, people with RS look alike. These children may have: small size and slow growth, short arm and leg bones, spine problems, short fingers and toes, heart or kidney problems, fused ribs, and abnormal nails. Typical facial features include a large head size (macrocephaly), broad forehead, eyes that are far apart (hypertelorism), small short nose, small chin, and low-set ears. Some people with RS have problems learning. Not everyone with RS has all of the findings or is affected to the same degree.

What causes RS?

RS can be caused by changes (mutations) in the *ROR2* gene. Everyone has two copies of the *ROR2* 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 both copies of the *ROR2* gene, he or she will not be able to make the normal protein and will have RS. People with a change in only one copy of the *ROR2* gene also have a normal copy of the gene. Thus, they do not have any signs of the disorder.

How does RS run in families?

RS can be inherited in an autosomal recessive or autosomal dominant pattern. Autosomal dominant RS (DRS) is the most common form, but there is currently no testing available for DRS. Individuals with mutations in *ROR2* have autosomal recessive RS (RRS). This means that both parents must be carriers to have a child with RRS. Carriers have one normal copy of the *ROR2* gene and one with a change. When both carriers pass the changed copy of the *ROR2* gene down to their child, that child develops RRS. When both parents are carriers, there is a 25% chance with each child that they will have RRS. RRS occurs more often in families in which the parents are related and those of Turkish and Omani origin.

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 RRS. Testing for mutations in *ROR2* 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 *ROR2* are found in the person with RRS, testing other family members, even during a pregnancy, is easy and fast.

Reasons for genetic testing for RRS:

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

What does it mean for my child if they find two mutations? What does it mean for our family?

Finding two changes in the *ROR2* gene confirms a diagnosis of RRS. When two changes in *ROR2* are found in a person with RRS, then other family members may have testing to see if they are carriers.

What does it mean if they find one mutation?

Finding only one change in a patient with possible RRS does not rule out or confirm the diagnosis. It is possible that have a second change in the *ROR2* gene that we cannot find with our test.

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

If someone does not have any changes in *ROR2*, then his/her RS is most likely not caused by problems in *ROR2*. This person may have DRS or they may have something else. Other possible diagnoses should be considered.

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 RRS or not. In this case, we may 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 you or your child has RRS, you should see a genetic specialist. Your doctor or hospital can help you set this up. This genetics 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 sequencing of *ROR2* is $1500. Cost for *ROR2* deletion/duplication analysis is $500. 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: 83891, 83898 x4, 83904 x6, 83912 for sequencing and 83891, 83900, 83901 x2, 83912 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 prenatal testing is $540. The CPT codes for these tests are 83891, 83898 x2, 83894, 83912 and 83891, 83898 x4, 83894, 83912 respectively.

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 RRS will increase what we know about this disorders and the genetic test.

Additional Resources:

Robinow Syndrome Foundation
Phone: 763-434-1152
Karla Kruger email: robinowfoundation@comcast.net
www.robinow.org

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