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Genetic Testing for Bilateral Frontoparietal Polymicrogyria

Information for Patients and Families

What do I need to know about testing my child for *GPR56*-related bilateral frontoparietal polymicrogyria?

GPR56-related bilateral frontoparietal polymicrogyria (BFPP) is a brain finding caused by a change in someone's DNA. This change in the brain causes people to have developmental delay, seizures, and crossed eyes. This blood test may prove that your child has *GPR56*-related BFPP. It may also find something that we do not understand. We may need to test the child's parents to learn more. This sheet will provide more details about *GPR56*-related BFPP and this testing. If you have more questions, please talk to a genetic counselor.

What is bilateral frontoparietal polymicrogyria (BFPP)?

Bilateral frontoparietal polymicrogyria (BFPP) is a rare brain finding that can be seen by MRI. The brain normally has many folds on its surface. Patients with polymicrogyria (PMG) have many more folds that are smaller than usual and can be difficult to see. "Bilateral frontoparietal" simply refers to where in the brain the PMG is seen. Children with BFPP have developmental delay, seizures, and crossed eyes. Not everyone is affected to the same degree.

What causes *GPR56*-related BFPP?

GPR56-related BFPP is caused by a change (mutation) in the *GPR56* (G-protein coupled receptor 56) gene. Everyone has two copies of the *GPR56* 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 *GPR56* gene, he or she will not be able to make the normal protein and will have BFPP. People with a change in only one copy of the *GPR56* gene also have a normal copy of the gene. Thus, they do not have any signs of the disorder.

How does *GPR56*-related BFPP run in families?

GPR56-related BFPP is autosomal recessive. This means that both parents must be carriers to have an affected child. Carriers have one normal copy of the *GPR56* gene and one with a change. When both carriers pass the changed copy of the *GPR56* gene down to their child, that child develops BFPP. When both parents are carriers, there is a 25% chance with each child that they will have BFPP.

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 BFPP. Testing for mutations in *GPR56* is complex. It is like reading a very long 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 875 in the second paragraph). When two changes in *GPR56* are found in the person with BFPP, testing other family members, even during a pregnancy, is easy and fast.

Reasons for genetic testing for *GPR56*-related BFPP:

- confirm the diagnosis
- check if other family members are carriers
- provide information and resources for future pregnancies
- provide information during a pregnancy regarding possible BFPP 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 *GPR56* gene confirms a diagnosis of *GPR56*-related BFPP. When two changes in *GPR56* are found in a person with BFPP, then other family members may have testing to see if they are carriers.

What does it mean for my child if they find one mutation?

Finding only one change in the *GPR56* gene does not rule out or confirm a diagnosis of *GPR56*-related BFPP. It is possible that your child has a second change in the *GPR56* gene that we cannot find with our test. Dr. Dobyns is conducting research on BFPP and similar conditions. Please see his information below.

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

If your child does not have any changes in *GPR56*, then his/her BFPP is not caused by problems in *GPR56*. We cannot tell you the reason for your child's BFPP. Dr. Dobyns is conducting research on BFPP and similar conditions. Please see his information below.

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 *GPR56*-related BFPP 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 geneticist or genetic counselor help you order the test for your child. If you think that your child has *GPR56*-related BFPP, you should see a genetic specialist. Your doctor or hospital can help you set this up. This genetics specialist can order the testing for *GPR56*-related BFPP. 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 testing is \$1500. 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 x 4, 83904 x 6, 83912. 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 \$590. The CPT codes for these tests are 83891, 83898 x 2, 83894, 83912, and 83891, 83898 x 2, 83894, 83912, 99051, 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 my child's test?

Your doctor will send a form about your child's symptoms with the blood sample. This 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. Information from children with *GPR56*-related BFPP will increase what we know about this disorder and the genetic test.

Can we still participate in research studies?

Yes, your child (family) can participate in research studies. Dr. Dobyns is conducting research on BFPP and similar conditions. Please see his information below.

Additional Resources:

The Lissencephaly Network www.lissencephaly.org
Phone: 260-432-4310 Email: lissnet@lissencephaly.org

The Brain Malformation Research Project at The University of Chicago
William B. Dobyns, Principal Investigator
Contact Mary King at 773-702-8247

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