



## The University of Chicago Genetic Services Laboratories

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### Genetic Testing for Microcephaly

#### *Information for Patients and Families*

#### **What do I need to know about *ASPM* testing for my child?**

Some forms of microcephaly (small head size) are caused by a change in someone's DNA. People with autosomal recessive primary microcephaly (MCPH) have microcephaly at birth and throughout life. They have learning problems and possibly seizures, but no other neurological findings. Parents of a child with MCPH have a 25% chance of having another child with MCPH with each pregnancy. *ASPM* is one gene that causes MCPH. This blood test may prove that your child has MCPH. There is also a chance that the test will find something that we do not understand. This information sheet will provide more details about MCPH and this testing. Please talk to a genetic counselor, if you have more questions about testing.

#### **What is MCPH?**

People with MCPH have microcephaly at birth and throughout life. They have learning problems and possibly seizures, but no other neurological findings. They may be shorter than other individuals, but usually have normal weight and appearance.

#### **What causes MCPH?**

Some forms of microcephaly (small head size) are caused by a change in someone's DNA. MCPH can be caused by two changes (mutations) in the *ASPM* (abnormal spindle-like, microcephaly-associated) gene. We each have two copies of the *ASPM* gene. Some children with MCPH have a change in each copy of the *ASPM* gene that we can find by sequencing (reading the gene). Genes are written instructions to make proteins. When there is a change in the instructions, the protein may not be made or may not work properly. Thus, both copies of the *ASPM* protein are not working properly in these kids.

#### **Will we have another child with MCPH?**

If a child is found to have two changes in the *ASPM* gene, that child's parents are most likely carriers. This means that they have one normal copy of the *ASPM* gene and one copy with a change. They do not have any problems because one normal copy of *ASPM* is enough. However, they can pass the changed copy down in each pregnancy. If both parents pass their changed copy to their child, the child will have MCPH. This is a process that we cannot control. Parents of a child with MCPH have a 25% chance of having another child with MCPH with each pregnancy.

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

The first person to be tested in any family would be the individual with MCPH. Testing for mutations in *ASPM* is complex. It is like reading a very long book and looking for two spelling mistakes. You may read the whole book and miss one or both "typos," however when you do find them, then it is easy to test other family members (i.e. you know that a change is on page 875 in the second paragraph). Once the changes are found in the person with MCPH, testing other family members, even during a pregnancy, is easy and fast because we know where to look. Testing is now available at The University of Chicago Genetics Services Laboratory.

#### **Reasons for genetic testing for MCPH:**

- confirm the diagnosis
- provide information and resources for future pregnancies
- provide information during a pregnancy regarding possible MCPH in the baby

#### **What does it mean for my child if they find two changes? What does it mean for our family?**

Finding two changes in *ASPM* means that this is the cause of your child's microcephaly. You have a 25% chance of having another child with MCPH with each pregnancy. Other family members may be tested, if they choose to.

### **What does it mean if they find one change?**

Finding only one change in a patient with microcephaly means that *ASPM* is the likely cause of your child's microcephaly. It is likely that your child has a second change in the *ASPM* gene that we cannot find with our test. You probably have a 25% chance of having another child with MCPH with each pregnancy, however future testing is difficult because we don't know both changes.

### **What does it mean for my child if they don't find a change?**

Not finding a change means that *ASPM* is not causing your child's microcephaly. Another gene or other factors may be the cause. Without finding the exact cause, we cannot estimate the risk to future pregnancies.

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

Some patients will be found to have a change in the gene, but we are not sure what that change means.

### **How do I get my child tested?**

We recommend that a genetic doctor or genetic counselor help you order the test for your child. If you think your child may have MCPH, you should make an appointment with someone that works in genetics. This can be made through your local physician or hospital. They can order the testing. If there are any questions about ordering the testing, please ask them 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 *ASPM* is \$2400. All insurance companies are different, but most of them should cover at least part of the cost of testing. You can 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: 83891, 83898 x 9, 83904 x 9, 83912. Insurance companies use these codes to define the method of testing. In most cases, The University of Chicago will bill your hospital or lab, which will then 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 hospital or lab. In this case, we will need payment from you by check or credit card before testing. You will need to get repaid from your insurance company. The University of Chicago is not responsible for this.

If a change or variant is found in your child, cost for testing other family members is \$390. Prenatal testing is \$590. The CPT codes are 83891, 83898 x 2, 83894, 83912, for other family members and 83891, 83898 x 2, 83894, 83912, 99051 for prenatal testing.

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

Testing takes approximately 10 weeks for *ASPM* sequencing. Your physician will be informed of the results as soon as it is complete. Results will be faxed and mailed to your physician.

### **Additional Resources:**

#### **Foundation for Children with Microcephaly**

Phone: 602-487-6445

email: [jenni@childrenwithmicro.org](mailto:jenni@childrenwithmicro.org)

[www.childrenwithmicro.org](http://www.childrenwithmicro.org)

### **Laboratory Faculty and Staff:**

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