



## The University of Chicago Genetic Services Laboratories

5841 S. Maryland Ave., Rm. G702, 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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### Genetic Testing for Oral-Facial-Digital Syndrome, Type 1

#### **Information for Patients, Families, and Non-Genetic Professionals**

#### **What do I need to know about testing my child for oral-facial-digital syndrome, type 1?**

Oral-facial-digital syndrome, type 1 (OFD1) is caused by a change in someone's DNA. People with OFD1 have differences in their mouth, teeth, face, fingers and toes, and may have learning problems. This blood test may prove that your child has OFD1. However, some of the children with OFD1 will have a negative result, so this test will not completely rule out OFD1. 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 OFD1 and this testing. Please talk to a genetic counselor if you have more questions about testing.

#### **What is oral-facial-digital syndrome, type 1?**

Oral-facial-digital syndrome, type 1 (OFD1) is a rare genetic condition. As with other syndromes, individuals with OFD1 look somewhat alike. Common findings in these children include: a lobed tongue, bumps on their tongue, cleft palate, teeth abnormalities, cleft lip, small chin, short fingers/toes, extra or fused fingers/toes. Some patients have learning problems, usually mild, and some also have kidney disease. Not everyone with OFD1 has all of the findings or is affected to the same degree.

#### **What causes OFD1?**

OFD1 is caused by a change (mutation) in the *OFD1* gene. Females have two copies of the *OFD1* gene, and males have one copy. About 80% of females with OFD1 have a change in one copy of *OFD1*. 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, the smaller amount of good protein from the *OFD1* gene causes the features in OFD1. When males have a change in their only copy of *OFD1*, they usually do not survive pregnancy. This is why we see many more females with OFD1 than males.

#### **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 thought to have OFD1. Testing for mutations in *OFD1* 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 OFD1, 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 OFD1 :**

- 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 OFD1 in the baby

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

Finding a change will confirm that the child has OFD1. Once a change has been found in an individual, then other family members may be tested, if they choose to.

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

Not finding a change does not mean that your child does not have OFD1. If your doctor is not sure about the diagnosis, a negative result may lean against it. We are only able to find changes in about 80% of people with OFD1. Researchers are working to improve our testing.

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

A small number of patients will be found to have a change in the gene, but we are not sure whether that change causes OFD1 or not. In this situation, we recommend testing parents. If a parent is found to have the same change (and presumably does not have OFD1), then most likely this change is just a normal variant. If it is not found in a parent, it is more likely related to OFD1.

### **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 OFD1, 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 for OFD1. 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 *OFD1* is \$1700. Cost for deletion/duplication testing is \$1000. 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: 81406 for sequencing and 81405 for deletion/duplication testing. 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.

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

Testing takes approximately 4-6 weeks for *OFD1* 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:**

#### **AboutFace International**

Phone: 800-665-FACE

Email: [info@aboutfaceinternational.org](mailto:info@aboutfaceinternational.org)

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

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