What You Need to Know About Newborn Screening for Cystic Fibrosis
All babies born in Ohio have a blood test at birth called the Newborn Screening Test, which screens for 36 different treatable conditions. Each state has a Newborn Screening Test, but each one tests for a different number of conditions. One of the conditions that all states now screen for is cystic fibrosis.

**What tests are done by the state lab?**

In Ohio, the Newborn Screening Test for cystic fibrosis is done in two steps. The first step is measuring a chemical in the body called immunoreactive trypsinogen (IRT). If the IRT is high, the second test is done. The second test is a genetic test for the most common mutations (gene changes) seen in cystic fibrosis. If at least one mutation is found, this is a “screen positive” result and follow-up testing is needed.

The following cystic fibrosis transmembrane conductance regulator (CFTR) mutations are included in the Ohio newborn screening (as of April 1, 2016):

<table>
<thead>
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<th>Mutation</th>
<th>1078delT</th>
<th>1717-1G&gt;A</th>
<th>1898+1G&gt;A</th>
<th>1898+5G&gt;T</th>
<th>2183AA&gt;G</th>
<th>2184delA</th>
<th>2307InsA</th>
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<td>3120+1G&gt;A</td>
<td>3659delC</td>
<td>3849+10kbC&gt;T</td>
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<td>621+1G&gt;T</td>
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<td>A455E</td>
<td>A559T</td>
<td>delta F508</td>
<td>delta I507</td>
<td>G542X</td>
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<td>G85E</td>
<td>M1101K</td>
<td>N1303K</td>
<td>R1162X</td>
<td>R117H</td>
<td>IVS8 5T/7T/9T</td>
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<td>R334W</td>
<td>R347H</td>
<td>R347P</td>
<td>R553X</td>
<td>R560T</td>
<td>S1255X</td>
<td>S549N</td>
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<td>S549R</td>
<td>V520F</td>
<td>W1282X</td>
<td>Y1092X</td>
<td>Y122X</td>
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</tbody>
</table>

The newborn screen is just a screen for cystic fibrosis. Those with an abnormal (screen positive) newborn screen do not necessarily have cystic fibrosis. Most times (approximately 90%), it is a false positive, meaning the screen was abnormal but the child does NOT have cystic fibrosis. Instead, the child is a cystic fibrosis carrier. Carriers do not have the condition themselves but are at increased risk to have a child with cystic fibrosis when they have children of their own.

**What should I do if my patient is positive for one of these mutations?**

Children with an abnormal newborn screen should have a sweat chloride test done. This is the best test we have to figure out if a child has cystic fibrosis or not. For the sweat chloride test to be accurate, it must be done when a child is at least four weeks old. The child must also weigh enough to be able to sweat enough for the test.

For children in our region, the sweat chloride test is usually done on Wednesday mornings at Nationwide Children's Hospital. This is during the Infant CF Clinic day, which is designed to reduce the exposure of infants with CF to pathogens. The family meets with a genetic counselor and/or a pulmonologist later that day to discuss the results, concerns or any needs for additional testing. We discuss the results, what it means for the baby and what it means for other family members. Please do not schedule the sweat test from your office.
Why is it important to do the sweat chloride on a day where only infants and toddlers with cystic fibrosis are seen and evaluated?

People with cystic fibrosis often have various organisms that are present all around us growing in their respiratory tract. These organisms are mostly harmless and benign. But they may become a concern for the person with cystic fibrosis when they become ill and need an antibiotic. A small number of bacteria are of greater concern to persons with cystic fibrosis and not to anyone else. Children seen in the infant CF clinic do not grow any of the concerning organisms. If the infant coming in for the sweat-chloride test has CF, regardless of severity, he or she could be exposed unnecessarily to someone who may inadvertently transfer an organism that could cause a negative impact on his/her health. When the Nationwide Children’s pulmonary medicine team schedules the sweat chloride test, they assure that only infants are sent to the lab during that time.

What should you tell the family when you refer them for the sweat chloride test?

At Nationwide Children’s, counseling and support will be offered for every family, for every outcome. While a full genetic counseling session will be available, it might help for parents to have some background before they come to the appointment.

Cystic fibrosis is a genetic condition, which is inherited in an autosomal recessive manner. It affects both males and females. Cystic fibrosis occurs when a person inherits a particular pair of genes that do not work correctly. The affected person inherits one of these non-working genes from each parent. Usually the parents do not have cystic fibrosis themselves, but they are carriers of the disease. A carrier is a person who has one working gene and one non-working gene. One in 25 (4%) Caucasians is a carrier of cystic fibrosis. While it is less common, cystic fibrosis still occurs in other ethnic backgrounds. Generally, a carrier has no symptoms of cystic fibrosis because the working gene compensates for the non-working gene. To have a child with cystic fibrosis, both parents must be at least cystic fibrosis carriers.

When both parents are carriers, with each pregnancy, they have a 1 in 4 (25%) chance of having a child affected with cystic fibrosis, a 1 in 2 (50%) chance of having a child who is a carrier, and a 1 in 4 (25%) chance of having a child who does not carry the cystic fibrosis gene at all. When only one parent is a carrier, with each pregnancy, they have a 1 in 2 (50%) chance of having a child who is a carrier, and a 1 in 2 (50%) chance of having a child who does not carry the cystic fibrosis gene at all.

The Cystic Fibrosis Program at Nationwide Children’s

The Cystic Fibrosis Center at Nationwide Children’s is devoted to the diagnosis and management of patients with cystic fibrosis. Clinician-scientists in the center are also involved in research to help better the lives of patients.

Accredited by the Cystic Fibrosis Foundation, the center currently follows about 450 patients who come from Ohio and surrounding states. Multidisciplinary care from a team including physicians, nurses, dieticians, social workers, pharmacists and respiratory therapists ensures comprehensive, patient-centered care for every patient.

For more information about the Cystic Fibrosis Center at Nationwide Children’s, visit NationwideChildrens.org/cystic-fibrosis.

To refer a patient or ask a question of our expert team, call (614) 722-4766.
Referrals and Consultations
Online: NationwideChildrens.org/Pulmonary-Medicine
Phone: (614) 722-4766
Physician Direct Connect Line for 24-hour urgent physician consultations: (614) 355-0221 or (877) 355-0221.

Laboratory Testing and Pathology Consultations
Online: NationwideChildrens.org/Lab
Phone: (614) 722-5477 or (877) 934-6575.