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May is Cystic Fibrosis Awareness Month

What is Cystic Fibrosis (CF)?

Cystic Fibrosis (CF) is a genetic disease that affects different organs in the body and results in respiratory (breathing) and digestive problems. It is one of the most common genetic diseases among Caucasians and occurs less frequently in other ethnic groups. CF is a life-long illness, and the symptoms associated with this disease usually become worse over time. However, early diagnosis and starting specialized medical care early (such as aggressive drug treatments, therapies, and specialized nutrition) can improve the life expectancy and the quality of life for those with CF.

How is CF inherited?

CF is a genetic disease. The gene that causes CF is known as the CFTR gene. Each person has two copies of the CFTR gene: one copy is inherited from the mother and the other copy from the father. Changes called "mutations" or "pathogenic variants" in the gene cause the gene to not function properly. CF results when a child is born with two copies of nonfunctioning (abnormal) CFTR gene, with one copy inherited from each parent. Each parent has one normal copy and one abnormal copy of the gene and usually has no symptoms of CF; each parent is said to be a "carrier" of CF. When both parents are carriers of CF, there is a 25% (1 in 4) chance of having an affected child in each pregnancy. A person's chance of being a carrier of CF depends upon his/her ethnicity.

What is CF carrier screening?

CF carrier screening is a genetic test that tests for a select number of mutations in the CFTR gene known to lead to the CF disease. For the selected mutations, this test can tell whether a person has no copy, one copy, or two copies of the mutation. Although CF carrier screening can identify a majority of CF carriers, it is important to remember that not all carriers of CF will be detected by this test. The CF carrier screening's ability to detect carriers depends upon the person's ethnicity as well as how many mutations are being tested. The parents' ethnicity information is very important for this testing so that accurate ethnicity information can be given to the laboratory.

CF carrier screening at Nationwide Children's

The Institute for Genomic Medicine Clinical Laboratory at Nationwide Children's Hospital offers CF carrier screening that tests for 60 common mutations known to cause CF. This 60 mutation panel includes 23 mutations recommended by the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG). The laboratory offers a quick turnaround time, with most results completed less than 7 days from receipt of the sample by the laboratory.

What happens if both partners are both found to be CF carriers?

It is important to remember that this does not mean that the child will definitely have CF. However, when both partners are CF carriers, the couple has a 25% (1 in 4) chance in each pregnancy of having a child affected with CF. If both partners are found to be CF carriers, prenatal testing and genetic counseling will be offered to them.

Prenatal testing for CF

Prenatal testing refers to testing the baby (fetus) before birth. When both partners are known to be CF carriers, prenatal testing is offered. Amniocentesis is a common method of prenatal testing that is usually performed in the second trimester and involves sampling a small amount of amniotic fluid by inserting a needle through the mother's abdomen. The fluid contains cells from the fetus that can be tested to determine if the fetus is affected with CF.

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 should I do if my patient is positive for one of these mutations from the Newborn Screen for Cystic Fibrosis?

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. Collections can be performed at the following locations: Dublin CTH (Hospital Drive), East Broad CTH, Lima LSC, Mansfield CTH, Springfield CTH, Westerville CTH, Zanesville LSC, and the Outpatient (Orange) Lab.

If the Outpatient Lab on Main Campus is most convenient, the collections can be scheduled through Central Scheduling at 614-722-6200. Collections for all other locations should be scheduled directly with the site. Contact Laboratory Services Client Services at (800) 934-6575 for site-specific contact information.

Patients should be well hydrated and kept warm during sweat collection. The patient cannot leave the lab/facility during the sweat chloride collection. Child should be at least 48 hours old, as erroneous results can occur if newborn is less than 48 hours old.

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.

Reminders

Changes and Closures in Laboratory Service Center Hours

Our Grove City Laboratory Services location is temporarily closed due to staffing issues. We encourage providers and patients to access our lab services at one of our other laboratory service centers.

Medicaid Changes to NCH Network

As of Feb.1, Anthem Blue Cross Blue Shield of Ohio Medicaid plan is out-of-network for Nationwide Children's Hospital. Note that this only applies to children covered by Anthem's Medicaid plan. Nationwide Children's remains in-network for children who are covered by Anthem's commercial plan.

Test Updates

Reference Range Updates

Anti-Streptolysin O	IgA, Quantitative
C3 Complement	IgG, Quantitative
C4 Complement	IgM, Quantitative
Ceruloplasmin	PreAlbumin
Cystatin C	Rheumatoid Factor
Haptoglobin	Transferrin

See our website for detailed information: https://www.nationwidechildrens.org/specialties/ laboratory-services/for-providers/laboratoryservices-news-publications



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How can Nationwide Children's Laboratory Services help your practice?

If you would like to become a client or learn more information about Nationwide Children's Laboratory Services, contact us at (800) 934-6575 or visit our website at **NationwideChildrens.org/Lab.** Would you like to receive the Nationwide Children's Laboratory Services Newsletter electronically? Please e-mail us at **LaboratoryServices@NationwideChildrens.org** and let us know!

Lab Account Representatives are available via email to assist with any questions or concerns.

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