How is my child tested for cystic fibrosis?

- Your care team notifies the Michigan Newborn Screen office of you or your partner's pregnancy.
- The blood sample from a heel poke done at birth is tested for an enzyme called immunoreactive trypsinogen (IRT).
- Normally, if IRT is high, then DNA analysis is done to look for 60 common CF gene mutations.
- If IRT is normal, no DNA analysis is done and the test is marked "negative" for CF.
- In an infant of a parent with CF, the blood is sent for DNA analysis of CF mutations, even if IRT is normal.

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MICHIGAN MEDICINE
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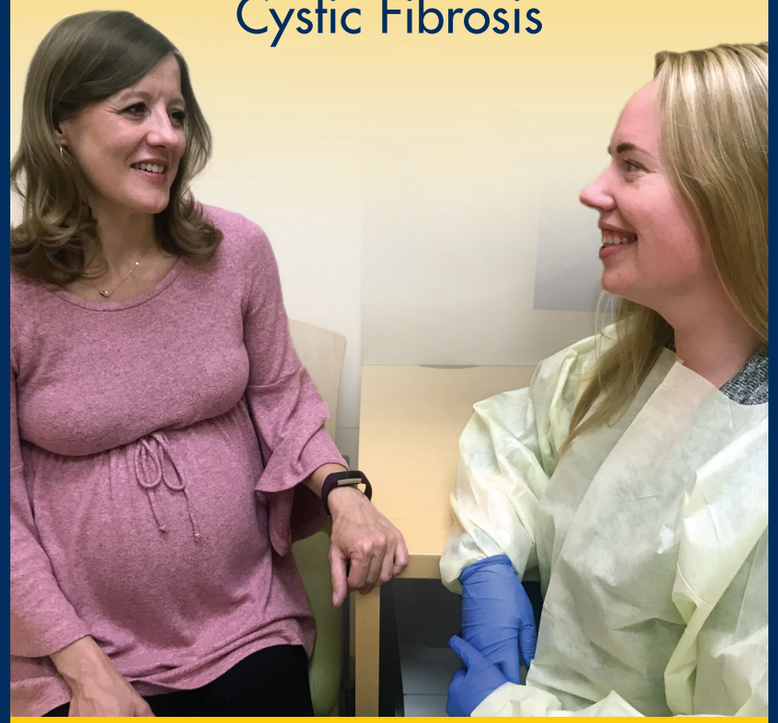
MICHIGAN
Newborn Screening

FOR PARENTS WITH
Cystic Fibrosis



A newborn screening tests all newborns for 55 disorders. This helps to provide early treatment to babies who might have a disease. Cystic fibrosis is one of the 55 disorders tested.

I have cystic fibrosis. Will my child test positive on a newborn screen?

Not necessarily. Since you, the parent with CF, have two mutations for CF, one mutation will be passed on to your baby. If the other parent does not have any CF mutations, the baby will have only one CF mutation, and will be a CF carrier. Babies that are CF carriers may have newborn screen tests come back either negative (low IRT) or positive (high IRT, with one CF mutation found).

- If the newborn screen test is negative, the blood sample will still be sent for the DNA analysis of CF mutations. This is why it is important to notify the Michigan Newborn Screen Office office about the baby's information.
- If the newborn screen test is positive, this does not necessarily mean your child has cystic fibrosis, but instead means a sweat test is needed.

When will I receive the results of the newborn screen?

Results are sent to your infant's pediatrician within a few weeks. After review, your infant's pediatrician will contact you.

What is the cost of newborn screening?

Newborn screening is a free, state-required program. All costs are covered by the state of Michigan including travel to CF centers for testing, if needed.

My partner has been tested for CF and the test was negative. Why does my infant need testing?

- Blood tests for CF mutations are screening tools and false negative results are possible.
- Blood tests for CF mutations do not usually look for all CF mutations. They are often limited to only 23 of the most common CF gene mutations.
- Sweat testing is the "gold standard" for CF diagnosis.

What is a sweat test, and how is it collected?

A sweat test is a measure of the amount of chloride (component of salt) in the sweat. High levels of sweat chloride are found in people with cystic fibrosis.

Testing takes about 60 minutes.

1. First, an electrode applies a very low current to the skin on the arm to stimulate it for 5 minutes. This can feel like a small tingling, like when a limb "falls asleep," but is not painful, and is not harmful.
2. Then, a collection device is wrapped on the skin and left to collect sweat for 30 minutes.
3. Finally, the chloride levels are measured in the collected sweat, and the results of the test are available the same day as testing.

What is the cost of sweat testing?

Sweat testing is usually covered by insurance. If the test is not covered by insurance, the state of Michigan will cover the costs of the sweat testing in a CF Center.

Without insurance, cost is $617

- Sweat collection - $258
- Sweat analysis - $359

If you are unable to pay and have a high deductible or no insurance, please contact your social worker:
Katie Hall 734-998-6067
Mari Pitcher 734-763-6670

I want my partner tested for CF genes. What testing should my partner have done?

- CFTR gene sequencing (full panel with reflex to all tiers, deletions and duplication) is the most accurate way to know if your partner is a carrier or not. Unfortunately, insurance doesn't usually cover this test.
- Testing for a more limited panel of CF gene mutations will identify the most common mutations, and can be useful if the CFTR gene sequencing is not available.