Cystic fibrosis (CF) is a rare genetic disorder affecting the lungs and digestive system that causes production of thick, sticky mucus that impacts lung function and prevents the normal breakdown of food in the body. Babies diagnosed with CF through newborn screening benefit from earlier management of symptoms, which can improve nutritional status, slow progression of lung damage, and improve quality of life.

Due to the importance of early management, all 50 states now perform newborn screening to test for CF. A confirmatory sweat test must be performed on any baby with a positive newborn CF screen. For babies born in our service area in Ohio, the state health department will fax newborn screen results to both your office and the Cincinnati Children’s CF Center. If a baby is born in northern Kentucky or southeastern Indiana and needs to come to Cincinnati Children’s for follow up, please contact us.

**POSITIVE NEWBORN SCREEN RESULTS** *(See other side for complete process)*

If you receive a positive screen report on a patient, a Cincinnati Children’s CF team member will contact you as soon as possible to discuss the results and next steps. They will also discuss a plan for contacting your patient’s family.

**PRIMARY CARE PROVIDER ROLE**

You are in a unique position to guide parents through the diagnostic process in a manner that’s informative and anxiety-reducing. Families have told us they want accurate information, emotional support, and a clear understanding about what to expect next. You can visit the CF center website (shown below) for a helpful video on how to discuss with families. It is important that the first call comes from you, the child’s PCP, but a follow-up call will come from the CF team with more details.

As soon as possible, call the parents to:

- Share the newborn screen result and explain next steps *(see other side)*
- Explain that most babies with a positive newborn screen and only 1 CFTR variant do not actually have CF
- Whenever possible, don’t make this call on a Friday, as doing so could lead to unnecessary stress and anxiety for the family as they wait until Monday to schedule the test
- Let them know they can visit the Cincinnati Children’s CF Center website, the CF Foundation website, or contact the Newborn Screen Coordinator for additional information

Because many parents want to know the symptoms of CF, share these:

- Difficulty growing or a need to consume extra calories to grow—most common
- Steatorrhea (fatty stools) or frequent, large, foul-smelling stools
- Other symptoms may include:
  - Salty-tasting skin
  - Shortness of breath
  - Daily, wet cough
  - Severe constipation

Prepare parents for the sweat test by informing them of the following:

- Do not use any lotions or creams on the baby’s skin 24 hours before the sweat test
- Continue regular feeding to ensure their baby is well hydrated for the test
- Dress their baby in warm clothing to increase sweat production during the test
- Watch “The Sweat Test: What to Expect” video on Cincinnati Children’s website

If you would like additional copies of this tool, or would like more information, please contact the Physician Outreach and Engagement team at Cincinnati Children’s.

If you have clinical questions about the CF testing process, email CFPULM@cchmc.org.

To learn more detailed information about newborn screening, the sweat testing process and other provider reference material or to watch the video referenced above, use [www.cincinnatichildrens.org/cf](http://www.cincinnatichildrens.org/cf) and click on the Healthcare Professionals button.

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**FAST FACTS**

- ~100 positive newborn screens Cincinnati Children’s CF Center receives annually
- ~10 positive newborn screens that result in a CF diagnosis by our CF Center annually
- >50% of the CF population is 18 years old and above
- >30,000 people living with CF in the United States
Cystic Fibrosis Newborn Screening
Provider process following a positive newborn screen in TriState area
Communication of newborn screen results varies by state.

Initial Newborn Screen (NBS) (performed at state level)

Ohio Department of Health faxes results to PCP and CCHMC CF Center
For PCPs in Kentucky or Indiana that receive positive newborn screens from the health department, please contact the CCHMC NBS CF Center to arrange follow up

(ASAP) CF Genetic Counselor (GC)/RN contacts PCP to discuss follow up, PCP then contacts family to review NBS results

For urgent issues, call 513-636-6771 and ask to speak to the CF newborn screening coordinator.

COMMUNITY PRACTICE SUPPORT TOOL / August 2023

1 MUTATION

GC/RN calls family, discusses results, explains genetics, schedules sweat test and genetic counseling to be done at 4 weeks of age

Baby has sweat test performed

Genetic Counselor meets with the family to:
1) review the newborn screen
2) discuss possible results of the sweat test
3) obtain a family history
4) explain autosomal recessive inheritance
5) discuss further genetic testing options for the family

At the visit or within a few hours, Genetic Counselor gives family results of sweat test, discusses next steps and sends a letter

Negative
Baby is likely a CF carrier

Quantity not sufficient
Repeat test in 2–4 weeks to collect sufficient sample

Borderline
Repeat test in 2 months

Positive
Baby has CF

CF team arranges close follow up

2 MUTATIONS

(ASAP) RN calls family, discusses results, explains genetics, schedules sweat test and CF clinic visit

Baby has sweat test performed

Baby has sweat test performed and then is seen in CF clinic to meet the CF team, review CF education and send additional testing (blood and stool sample and throat culture)

Sweat test results

Positive
Pulmonologist determines follow up based on exam/lab results

Quantity not sufficient/Borderline

Negative