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. In Ohio and northern Kentucky, your state health department will fax newborn screen results to both you and the Cincinnati Children's CF Center.

**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” 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 the likelihood the baby will produce enough sweat for an accurate test
- Encourage them to visit the Cincinnati Children’s CF Center website and watch “What to Expect” video

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 and click on the Healthcare Professionals button.
Cystic Fibrosis Newborn Screening
Provider process following a positive newborn screen in Ohio
Communication of newborn screen results varies by state.

1 MUTATION

- Baby has f/up sweat test performed
  - Genetic Counselor meets with family to (1) discuss: newborn screen, autosomal recessive inheritance, and recurrence risks and (2) to obtain family history and options for further genetic testing
  - At the visit or within a few hours, Genetic Counselor gives family results and next steps, and contacts PCP with sweat test results

2 MUTATIONS

- Baby has f/up sweat test performed
  - Social worker or RN from CF meets with family to answer FAQs; selective mutation panel testing is ordered and performed by CF team
  - RN contacts PCP with results and together determine who will inform the family

- Does baby have CF?
  - Yes
    - RN arranges first visit with CF team
  - No
    - CF team informs family of negative results

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