NORTH CAROLINA NEWBORN SCREENING PROGRAM

Cystic Fibrosis (CF) Provider Guide

Immunoreactive Trypsinogen (IRT) ≥ 120 ng/mL and 1 CFTR Variant Identified	Elevated Immunoreactive Trypsinogen (IRT) >96% tile and 1 CFTR Variant Identified	Elevated Immunoreactive Trypsinogen (IRT) and 2 CFTR Variants Identified (Both CF disease-causing)
Immediate Next Steps • **Contact** family to notify them of the newborn screening result and assess symptoms. • **Evaluate** infant (poor weight gain, absent stooling, abdominal pain, voracious appetite); arrange immediate referral if symptomatic. • **Schedule** sweat testing at an accredited CF center. Call the designated contact person listed on the attached resource list for specific referral instructions. *If the infant has a positive family history of CF or is symptomatic, consult with the CF specialist for additional recommendations.* • **Goal** is for sweat testing to be accomplished before 4 weeks of age.	**Immediate Next Steps** • **Contact** family to notify them of the newborn screening result and assess symptoms. • **Evaluate** infant (poor weight gain, absent stooling, abdominal pain, voracious appetite); arrange immediate referral if symptomatic. • **Schedule** sweat testing at an accredited CF center. Call the designated contact person listed on the attached resource list for specific referral instructions. *If the infant has a positive family history of CF or is symptomatic, consult with the CF specialist for additional recommendations.* • **Goal** is for sweat testing to be accomplished before 4 weeks of age.	**This screening result is likely a true diagnosis of cystic fibrosis (CF). Medical intervention needs to start as soon as possible.** **Consult** with a CF specialist for immediate referral to an accredited CF center. Contact information for accredited CF centers can be found on the resource list provided. **Contact** family to notify them of the newborn screening result and assess symptoms. **Goal** is for evaluation ASAP.
Review with Family • Discuss this result with the family and share the follow-up plan. • Since newborn screening only tests for 139 specific CF variants, explain the importance of pursuing a sweat chloride test to confirm or rule out a diagnosis. • Educate family about signs, symptoms, and when urgent treatment may be needed.	**Review with Family** • Discuss this result with the family and share the follow-up plan. • Since newborn screening only tests for 139 specific CF variants, explain the importance of pursuing a sweat chloride test to confirm or rule out a diagnosis. • Educate family about signs, symptoms, and when urgent treatment may be needed.	**Review with Family** • Discuss this result with the family and explain that newborn screening has detected 2 CFTR variants and it is highly likely the infant has cystic fibrosis • Share the follow-up plan which includes having the infant evaluated by a CF specialist as soon as possible. The CF specialist will schedule a sweat test, provide them with more information and discuss recommended treatment.

Differential Diagnosis

An elevated IRT with at least one CFTR variant is primarily associated with:

- Cystic fibrosis carrier — about 1 in 25 Caucasians are carriers
- Cystic fibrosis — Incidence of 1 in 3,500
- CFTR-related metabolic syndrome (CRMS)

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False Positives

Most but not all infants with only one CFTR variant found on screening are unaffected carriers. Sweat testing at an accredited CF center is needed to determine if infant has or does not have CF.

Clinical Summary

- CF is an autosomal recessive disorder caused by specific cystic fibrosis transmembrane conductance regulator (CFTR) gene variants.
- Individuals with only one variant in the CFTR gene are considered carriers.
- A CF carrier is healthy and does not have cystic fibrosis.
- Because the North Carolina Newborn Screening Program only screens for a panel of the 139 most common CF variants, it is possible that a second CFTR variant exists that is not identifiable by the variant panel.

What treatment options are available?

Although CF cannot be cured, the symptoms can be treated. Possible treatments can include:

- Prescription enzymes to help absorb food better
- Healthy, high-calorie diet
- Vitamins
- Antibiotics to prevent and control infections
- Mucus thinners and airway clearance
- CFTR Modulator Therapies: designed to correct the malfunctioning protein made by the CFTR gene

Where do I go for more information?

Newborn screening for providers:
https://www.cincinnatichildrens.org/service/c/cystic-fibrosis/healthcare-professionals

Sweat testing:
https://www.cff.org/intro-cf/sweat-test

For 2 mutation results:
https://www.cff.org/intro-cf/about-cystic-fibrosis

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

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