

NEXT STEPS AFTER A SINGLE CYSTIC FIBROSIS VARIANT IS DETECTED

- **No further testing is required at this time.**
- Ensure screening results are included in the infant’s medical record.
- Genetic counseling should be provided to the family, ideally by a licensed genetic counselor.
- Assist parents with obtaining their own variant testing, if desired.
- Additional testing is at the discretion of the provider if signs or symptoms of cystic fibrosis (CF) are present. See the reverse side of this page for a list of contracted Newborn Screening (NBS) Program’s CF referral centers.

Contact the Florida NBS Program with questions or to obtain the supplemental sequencing report at 866-804-9166.

HOW NBS SCREENS FOR AND REPORTS CYSTIC FIBROSIS

CF screening involves up to three tiers of testing before results are reported.

First Tier: All specimens are tested using immunoreactive trypsinogen (IRT).

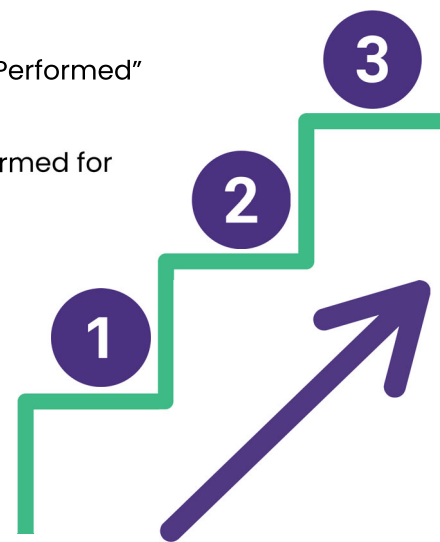
If the IRT results are <50 ng/mL, CF will report as “Within Normal Limits–DNA Not Performed” on the newborn screening report.

Second Tier: DNA analysis of the 74 most common CF-causing variants is performed for the following reasons:

- Top 4% of daily IRT results
- All specimens with IRT ≥50 ng/mL
- All specimens with meconium ileus reported

Third Tier: Next Generation Sequencing (NGS) is performed for the following reasons:

- A CF-causing variant was detected during second tier testing
- No variant was detected on second tier and the IRT is ≥160 ng/mL
- Second tier results were inconclusive



REPORTABLE VARIANTS

A reportable variant is classified as either pathogenic, likely pathogenic, variant of uncertain significance, or variant of varying clinical consequence.

If **no reportable variants** are detected after all three tiers of testing, results are considered **within normal limits**. CF will reflect as “Within Normal Limits– No Reportable Variants Detected” on the newborn screening report.

If a **single reportable variant** is detected after all three tiers of testing, the **infant is considered to be a CF carrier**. The newborn screening report will reflect the IRT value and single variant with the pathogenicity listed. Letters and information sheets are mailed to the parent and primary care provider on record.

If **multiple reportable variants** are detected after all three tiers of testing, the **infant will be referred by the NBS Program to a contracted CF referral center** for diagnostic evaluation. The newborn screening report will reflect the IRT value and multiple variants with the most severe pathogenicity listed.

See reverse for additional information.

SPECIAL CONSIDERATIONS FOR CYSTIC FIBROSIS SCREENING RESULTS

- IRT levels can fluctuate and may not require DNA analysis on all specimens; however, DNA results do not change. Any CF variant results detected on any specimen are valid and should be treated as such.
- While Florida's variant panel can detect known CF-causing variants, it is possible for an infant to have a variant(s) not identified through screening. Physician discretion is advised if signs and symptoms are present, or there is a family history of CF.
- Prenatal testing through commercial laboratories is often limited to the most common CF-causing variants, so it is possible for newborn screening to detect a variant not included on prenatal testing panels.

ADDITIONAL CYSTIC FIBROSIS RESOURCES

All NBS Program CF referral centers have received Cystic Fibrosis Foundation pediatric accreditation. Should additional testing be desired, please contact the referral center in your area. A map of all NBS Program referral centers may be found under the Health Care Providers tab on the NBS Program's website:

FloridaHealth.gov/NewbornScreening.

NBS PROGRAM CYSTIC FIBROSIS REFERRAL CENTERS

Johns Hopkins All Children's Hospital

St. Petersburg
727-767-4146

Lee Health, Golisano Children's Hospital

Ft. Myers
239-437-5500

Memorial Healthcare System, Joe DiMaggio Children's Hospital

Hollywood
954-265-3665

Nemours Children's Health

Jacksonville
904-697-3600

Nemours Children's Health

Orlando
689-208-5205 (Ask for CF Team)

Nemours Children's Health

Pensacola
850-505-4700

Nicklaus Children's Hospital

Miami
305-669-5864

Orlando Health, Arnold Palmer Children's Hospital

Orlando
321-841-6350

University of Florida

Gainesville
352-273-8380

University of Miami

Miami
305-243-6162

University of South Florida

Tampa
813-821-8029

Florida Department of Health

Division of Children's Medical Services
Florida Newborn Screening Program
850-245-4201 | CMS.NBS@FLHealth.gov

For additional questions, please call the Newborn
Screening Follow-up Program Nursing Unit at
866-804-9166.

For additional information, visit
FloridaHealth.gov/NewbornScreening.