

Newborn Screening ACT Sheet

[Elevated Immunoreactive Trypsinogen (IRT) + 1 DNA Variant]

Cystic Fibrosis (CF)

Differential Diagnosis: Cystic fibrosis carrier; Cystic fibrosis; CRMS (CFTR-Related Metabolic Syndrome).

Condition Description: Cystic fibrosis is a multisystem disorder affecting all races and ethnicities. It is caused by pathogenic variants in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene, an epithelial ion channel protein which maintains salt and water balance within cells. In individuals with CF, pathogenic variants in the *CFTR* gene cause thickened mucus, blocking the pulmonary and gastrointestinal systems. An elevated IRT level is a nonspecific marker of pancreatic stress and may be elevated due to CF, CF carrier status, or other reasons. Newborns with CF may or may not be symptomatic. A newborn with one *CFTR* variant may be a CF carrier or may have CF if the second variant is not identified by newborn screening. The majority of CF carriers are asymptomatic.

You Should Take the Following Actions:

- Inform the family of the newborn screening result.
- Ascertain clinical status (cough, abdominal pain, dehydration, frequent or oily stools) and inquire about family history of CF.
- Consult with CF specialist and/or contact newborn screening program for assistance within 24 hours.
- Evaluate the newborn (poor weight gain, dehydration, cough, respiratory distress, meconium ileus, abdominal pain, jaundice, abnormal stools).
- Refer promptly to CF Center for sweat testing, clinical evaluation, and genetic counseling as recommended by specialist.
- Provide family with basic information about CF.
- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: [Sweat Chloride Testing performed at a CF Care Center accredited by the CF Foundation](#) : may confirm the diagnosis. [Additional Molecular Genetic Testing:](#) may be required to confirm the diagnosis.

Clinical Considerations: Cystic fibrosis is a progressive disorder affecting all races and ethnicities in which abnormal salt regulation causes the accumulation of thickened mucus within the body. The symptoms are variable, and may include nasal polyps, growth delays, recurrent sinus and lung infections, diabetes, and malnutrition. Pancreatic insufficiency is found in 80 – 90% of cases.

A newborn with one *CFTR* variant may be a CF carrier or may have CF if a second variant is present but was not identified by newborn screening. Sweat chloride testing will identify most but not all individuals with CF. Management may include antibiotics, *CFTR* modulators, pulmonary and nutritional support. Although affected males are generally infertile, assisted reproductive technology can be used to obtain sperm for fertilization. Due to improved medical treatments, the majority of individuals with CF survive well into adulthood.

Additional Information:

[How to Communicate Newborn Screening Results](#)

[Gene Reviews](#)

[Medline Plus](#)

[Condition Information for Families- HRSA Newborn Screening Clearinghouse](#)

[Cystic Fibrosis Foundation](#)

[Clinicaltrials.gov](#)

Referral (local, state, regional, and national):

[Find a Genetics Clinic Directory](#)

[Genetic Testing Registry](#)

[Find a CF Care Center](#)

Disclaimer: This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

Local Resources *(Insert Local Website Links)*
State Resource Site *(Insert Website Information)*

Name	
URL	
Comments	

Local Resource Site *(Insert Website Information)*

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

Additional Information

How to Communicate Newborn Screening Results

- <https://bit.ly/NBSResultsHRSA>

Gene Reviews

- <https://www.ncbi.nlm.nih.gov/books/NBK1250/>

Medline Plus

- <https://medlineplus.gov/genetics/condition/cystic-fibrosis/>

Condition Information for Families- HRSA Newborn Screening Clearinghouse

- <https://newbornscreening.hrsa.gov/conditions/cystic-fibrosis>

Cystic Fibrosis Foundation

- <https://www.cff.org/>

Clinicaltrials.gov

- <https://clinicaltrials.gov/>

Referral (local, state, regional and national)

Find a Genetics Clinic Directory

- <https://clinics.acmg.net>

Genetic Testing Registry

- <https://www.ncbi.nlm.nih.gov/gtr/>

Find a CF Care Center

- https://apps.cff.org/ccd?_ga=2.114810501.381175015.1706627402-180508355.1696535979