



Physician ACT SHEET for POSTIVE Newborn Screening Result for CYSTIC FIBROSIS

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- **Contact family to inform them of the newborn screening result and to ascertain clinical status** (failure to thrive, recurrent cough, wheezing and chronic abdominal pain, etc.).
- **Contact an Accredited CF Center to schedule appointment** and to ask questions.
- **Arrange for the collection of a repeat dried blood spot filter paper specimen to be collected and tested at the screening lab.**
- Report findings to the newborn screening program.

Accredited CF Center contacts:

Nebraska Accredited CF Center (at UNMC in Omaha): 402 559-6275

Colorado Accredited CF Center (Children's in Denver): 720 777-6181

Meaning of the Screening Result: A positive screening result can be obtained on an initial specimen or on a repeat screen. An elevated immunoreactive trypsinogen (IRT) result in conjunction with two copies of DNA mutations is suggestive of cystic fibrosis.

Confirmation of Diagnosis: Genotype-phenotype associations are known for many CF mutations and combinations of mutations. Therefore, it may not be necessary to conduct a sweat test for some patients when this information is derived from the screen. However, it is important to repeat a dried blood spot filter paper specimen to verify the correct child has been identified. Referral to an accredited CF center is always recommended for comprehensive management of CF patients.

Condition Description: The CFTR protein regulates chloride transport that is important for function of lungs, upper respiratory tract, pancreas, liver, sweat glands and genitourinary tract.

Clinical Expectations: Deficient chloride transport in lungs leads to production of abnormally thick mucous leading to airway obstruction, neutrophil dominated inflammation and recurrent and progressive pulmonary infections. Pancreatic insufficiency is found in 80-90% of cases.

Additional Information:

- Cystic Fibrosis Foundation - www.cff.org
- New England Metabolic Consortium - www.childrenshospital.org/newenglandconsortium
- Gene Tests/Gene Clinics - www.genetests.org
- U.S. National Newborn Screening & Genetics Resource Center - www.genes-r-us.uthscsa.edu