



Newborn Screening ACT Sheet

[Elevated IRT +/- DNA]

Cystic Fibrosis

Differential diagnosis: Cystic fibrosis (CF); gastrointestinal abnormalities are also causes of increased IRT.

Condition Description: The cystic fibrosis transmembrane conductance regulator (CFTR) protein regulates chloride transport that is important for function of lungs, upper respiratory tract, pancreas, liver, sweat glands and genitourinary tract.

You Should Take the Following Actions:

- Contact family and primary care physician to inform them of the newborn screening result and to ascertain clinical status (meconium ileus, failure to thrive, recurrent cough, wheezing and chronic abdominal pain).
- Determine sweat chloride (sweat test).
- If cystic fibrosis is confirmed, clinical evaluation, and genetic counseling are indicated.
- Report findings to newborn screening program.

Confirmation of Diagnosis: Varies with screening test. Infants with highly elevated immunoreactive trypsinogen (IRT) may be considered screen positive. Moderately elevated IRT results are followed with second tier tests for either IRT or CFTR mutation panels. If screen positive, follow up with sweat test.

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

Additional Information:

(Click on the name to take you to the website. Complete URLs are listed in the Appendix)

[Gene Tests/Gene Clinics](#)

[Cystic Fibrosis Foundation](#)

[OMIM](#)

[Genetics Home Reference](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical](#)

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality clinical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines 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 healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.

State Resource Site(insert state newborn screening program website information)

Name
URL
Comments

Local Resource Sites (insert local and regional newborn screening websites information)

Name
URL
Comments

Name
URL
Comments

Name
URL
Comments

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Tests/Gene Clinics

<http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=9BCW795RasXO3&gry=&fcn=y&fw=8eae&filename=/profiles/cf/index.html>

Cystic Fibrosis Foundation http://www.cff.org/about_cf/what_is_cf/

OMIM <http://www.ncbi.nlm.nih.gov/entrez/dispmim.cgi?id=219700>

Genetics Home Reference <http://ghr.nlm.nih.gov/condition=cysticfibrosis>

Referral (local, state, regional and national):

Testing

http://www.genetests.org/servlet/access?prg=j&db=genetests&site=gt&id=8888891&fcn=c&qry=2220&res=nous&res=nointl&key=9BCW795RasXO3&show_flag=c

Clinical <http://www.cysticfibrosis.com/centers.html>

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