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. CF affects multiple body systems and is associated with progressive damage to respiratory and digestive systems.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family 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).
- Contact CF Center for consultation with CF specialist.
- Determine sweat chloride (sweat test) through experienced sweat test laboratory.
- If cystic fibrosis is confirmed, clinical evaluation and genetic counseling are indicated.
- Report findings to newborn screening program.

Diagnostic Evaluation: Varies with screening test. Infants with highly elevated immunoreactive trypsinogen (IRT) may be considered screen positive. Elevated IRT results are followed with second tier tests for either additional IRT measurement or CFTR mutation panels. If screen positive, follow up with sweat chloride test to confirm diagnosis.

Clinical Considerations: 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. Some males may be infertile in adulthood.

Additional Information:

Gene Reviews
Cystic Fibrosis Foundation
OMIM
Genetics Home Reference
American College of Medical Genetics

Referral (local, state, regional and national):

Testing
Clinical Services
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Reviews
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=cf

Cystic Fibrosis Foundation
http://www.cff.org/AboutCF/

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

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

American College of Medical Genetics
http://www.acmg.net/StaticContent/StaticPages/CF_Mutation.pdf

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests

Clinical Services
http://www.cff.org/LivingWithCF/CareCenterNetwork/CFFoundation-accreditedCareCenters/

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

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