Disease Name	**Cystic Fibrosis**
Acronym	CF
Disease Classification	Genetic Disorder Formerly known as cystic fibrosis of the pancreas, this entity has increasingly been labeled simply 'cystic fibrosis.' Manifestations relate not only to the disruption of exocrine function of the pancreas but also to intestinal glands (meconium ileus), biliary tree (biliary cirrhosis), bronchial glands (chronic bronchopulmonary infection with emphysema), and sweat glands (high sweat electrolyte with depletion in a hot environment). Infertility occurs in males and females.
Symptom onset	Usually within the first year of life. A small number, however, are not diagnosed until age 18 or older. These patients usually have a milder form of the disease.
Symptoms	Infants with CF have a variety of symptoms including: meconium ilieus, liver disease, pancreatic insufficiency, pulmonary disease or an excessive appetite but poor weight gain; and greasy, bulky stools. Symptoms vary from person to person due, in part, to the more than 1,000 mutations of the CF gene. The sweat test is the standard diagnostic test for CF. A sweat test should be performed at a CF Foundation-accredited care center where strict guidelines are followed to ensure accurate results. This simple and painless procedure measures the amount of salt in the sweat. A high salt level indicates CF.
Treatment	The treatment of CF depends upon the stage of the disease and the organs involved. Clearing mucus from the lungs is an important part of the daily CF treatment regimen. In addition, approximately 90 percent of all people with CF take pancreatic enzyme supplements to help them absorb food in digestion.
Inheritance	Autosomal recessive
General population incidence	1 in 2,500 white live births and 1 in 17,000 African American live births.
OMIM Link **Genetests Link**	http://www.ncbi.nlm.nih.gov/omim/219700 www.genetests.org
Support Group	Cystic Fibrosis Foundation http://www.cff.org/home National Organization for Rare Diseases http://www.rarediseases.org

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