

Cystic Fibrosis (CF)

Differential Diagnosis: Cystic Fibrosis (Elevated IRT)

Condition Description: Cystic Fibrosis (CF) is an inherited metabolic disorder that affects the exocrine glands in multiple organ systems in the body. The respiratory and digestive systems are usually the most severely affected. A mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene causes abnormal movement of chloride and sodium ions in cells in the lungs and gastrointestinal tract. Thickened secretions develop in the lungs, which block small airways and are easily infected with bacteria. Many people with CF have pancreatic insufficiency, which results from thickened secretions blocking the pancreatic duct and preventing pancreatic enzymes from entering into the gastrointestinal tract. Most infants with CF are born to parents who are asymptomatic CF carriers with no known family history.

Take the Following Actions:

- ◆ **Contact the family to inform them of the newborn screening result and to determine clinical status (meconium ileus; recurrent cough; wheezing; recurrent abdominal pain; greasy, bulky, foul-smelling stools; and/or failure to thrive).**
- ◆ **Obtain sweat test at a CF Center. A reference list for sweat testing facilities is available from DSHS.**
- ◆ **If CF is confirmed, perform clinical evaluation and refer to a CF Center. Genetic counseling is also indicated.**
- ◆ **Report findings to Newborn Screening Program.**

Confirmation of Diagnosis: Infants with very highly elevated immunoreactive trypsinogen (IRT) may be considered screen-positive. If the IRT levels are elevated and one or two mutations have been identified by the DNA panel, refer patient for sweat testing at a CF Center. If one or two mutations are identified, consider a referral for genetic counseling.

Clinical Expectations: Abnormally thick secretions accumulate in the lungs, clogging the bronchi and leading to airway obstruction, neutrophil-dominated inflammation, and chronic pulmonary infections. In the digestive system, the thickened secretions disrupt the passage of digestive enzymes and block the absorption of essential nutrients. Pancreatic insufficiency occurs in 80 – 90% of cases. Due to chronic lung infections and the progressive loss of lung function, CF leads to decreased life expectancy. With proper, lifelong treatment, most people with CF live into their 30s. However, with new treatments and research the life expectancy may be greater.

Additional Information:

Cystic Fibrosis Foundation
www.cff.org

March of Dimes
www.marchofdimes.com/pnhec/4439_1213.asp

Online Mendelian Inheritance in Man (OMIM)
www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=219700

U.S. National Library of Medicine, Genetics Home Reference
www.ghr.nlm.nih.gov/condition=cysticfibrosis

U.S. National Library of Medicine and National Institutes of Health, MedlinePlus
www.nlm.nih.gov/medlineplus/ency/article/000107.htm
(Cystic fibrosis)
www.nlm.nih.gov/medlineplus/ency/article/002437.htm
(Nutritional considerations)

Source: American College of Medical Genetics.