

Suggested Follow-up for Cystic Fibrosis Elevated IRT

Possible Causes: Autosomal recessive disorder characterized by pulmonary obstruction often accompanied by pancreatic dysfunction. Elevated IRT can also occur in premature/stressed infants.

Next Steps if Abnormal: Repeat IRT screening on filter paper and send to the DHEC laboratory. No treatment necessary until result of repeat testing known. If IRT is still elevated in repeat specimen, refer to pediatric pulmonologist for sweat testing at a CF Foundation accredited care center. Further evaluation/testing is necessary to establish diagnosis.

Neonatal Presentation: Usually none. Meconium ileus or volvulus may occur in 5-10% of affected infants.

Emergency Treatment: None.

Standard Treatment: Chest physiotherapy to aid in airway clearance. Antibiotics/other medication to treat lung infections as needed. Pancreatic enzymes if indicated; vitamins; NaCl supplements. Close monitoring of growth parameters and use of nutritional supplements if needed to enhance/maintain appropriate growth/development.

Advice for Family: Provide basic information about CF. The handout, *When Baby Needs a Second Test for Cystic Fibrosis*, may be used for this purpose.

NOTE: All infants with meconium ileus should be thoroughly evaluated for CF regardless of the IRT result. A normal IRT does not rule out CF in these infants.

Internet Resources:

<http://www.cff.org/home/>

<http://www.genetests.org/query?dz=cf>

<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>