



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.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE 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.

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:

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:

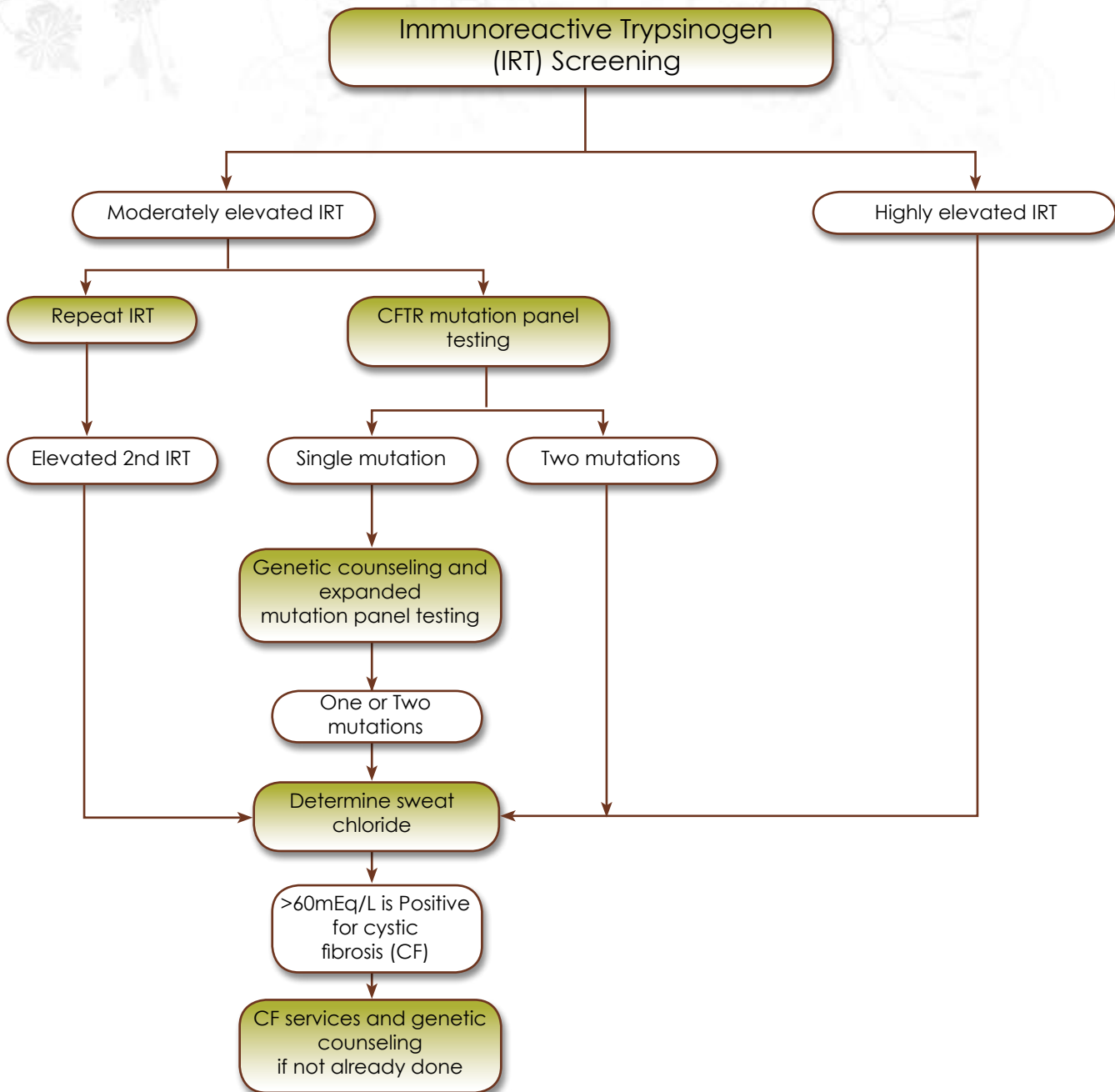
Deficient chloride transport in lungs causes production of abnormally thick mucous leading to airway obstruction, neutrophil dominated inflammation and recurrent and progressive pulmonary infections. Pancreatic insufficiency found in 80– 90% of cases.

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. 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





Immunoreactive Trypsinogen (IRT Elevated)



Actions are shown in shaded boxes; results are in the unshaded boxes.

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