KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED IMMUNOREACTIVE TRYPSINOGEN AND PRESENCE OF CF GENE MUTATION (IRT/DNA)

CONDITION: CYSTIC FIBROSIS (CF)

***DIFFERENTIAL DIAGNOSIS**:* Cystic fibrosis (CF); gastrointestinal abnormalities are also causes of increased IRT.

METABOLIC 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.

ACTION TO BE TAKEN **IMMEDIATELY:**

- Contact family to inform them of the newborn screening result and ascertain clinical status (meconium ileus, failure to thrive, recurrent cough, wheezing and chronic abdominal pain).
- Schedule sweat chloride (sweat test) at a **CF Foundation accredited center**.
- If cystic fibrosis is confirmed, clinical evaluation and genetic counseling are indicated.
- For single mutation and normal sweat chloride, consider genetic counseling for carrier status.
- Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: If IRT is ≥ 170 ng/mL **OR** DNA screening panel shows one or more mutations, follow up with sweat chloride test at an **accredited CF center**. See below for contact information. Consider genetic counseling for infants with one mutation and normal 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.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

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