

## Information for Primary Care Providers on NBS CF Results of:

### **Elevated IRT and Negative CFTR DNA Panel**

- A Cystic Fibrosis NBS result with elevated IRT and Negative CFTR DNA Panel results is interpreted as **Screen Negative for Cystic Fibrosis**; no further action or follow-up is required.
- An elevated IRT (Immunoreactive Trypsinogen) is **not** diagnostic for CF. The majority of elevated IRT results are false positives.
- Trypsinogen, a precursor to the enzyme trypsin, is produced in the pancreas. Blockage of pancreatic ducts causes elevations of trypsinogen in the blood of most newborns with CF. Newborns who do **not** have CF can have temporary elevations of trypsinogen; their levels become normal within a few weeks.
- Some studies have found high IRT levels associated with asphyxia during birth (measured by Apgar score)<sup>1-3</sup>, chronic fetal distress<sup>4</sup>, infection or other illnesses requiring neonatal intensive care<sup>5</sup>. How these factors influence IRT levels is not completely understood.
- Elevated IRT levels may be also be due to the biological and genetic variability of newborns<sup>1,2,6,7</sup>.
- The chance that an infant with an elevated IRT and negative CFTR DNA panel actually has CF is low.
- If the baby has symptoms of CF, both parents are known carriers of CF or if there is a family history of CF, consultation with a CF Center specialist about diagnostic testing and evaluation for the infant is recommended.

Symptoms of CF in the first few months of life may include some of the following:

- ◇ meconium ileus
  - ◇ very salty sweat
  - ◇ failure to thrive, slow growth
  - ◇ recurrent respiratory infections, including respiratory syncytial virus (RSV) infections
  - ◇ malnutrition (vitamin deficiencies, malabsorption)
  - ◇ frequent runny stools
- For all the disorders screened for, the newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider. A negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of the disorders screened for.

#### **References**

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