

Newborn Screening for Cystic Fibrosis Educational Materials for Healthcare Practitioners

OVERVIEW

In October 2007, the State of Indiana added cystic fibrosis (CF) to its newborn screening panel. CF newborn screening is a two-step process performed on a standard Guthrie card. The first step involves analysis of immunoreactive trypsin (IRT). Specimens that have IRT levels in the top 5 percent are considered abnormal and trigger a second test that checks for mutations within the *CFTR* gene. The screen is considered “positive” for CF if one or more mutations from the panel are detected. At this step, the healthcare practitioner is notified of the positive test by the Indiana University Newborn Screening Laboratory.

What does an abnormal CF newborn screen mean?

1. When two mutations are identified, the test is presumptive for the diagnosis of CF. **Infants who screen positive for two mutations should be referred directly to a Cystic Fibrosis Foundation (CFF) accredited center for prompt evaluation and treatment. Part of this evaluation will include a sweat test to verify the diagnosis of CF. A separate sweat test will not need to be arranged unless requested by the CF Center.** Genetic counseling is also recommended at this time.
2. If one mutation is identified, there is approximately a 5% chance that the child has CF. Most of these screened infants will be carriers of a single *CFTR* mutation. It is possible that the child has a second mutation, not included in the newborn screening DNA panel, and thus has CF. A sweat test is needed to determine whether this infant has CF. **The referral for a sweat test should be completed within the first two weeks after the primary care physician has been notified of the positive CF newborn screen.** Genetic counseling for the family is also strongly recommended at this point, since at least one parent must also carry a *CFTR* mutation. The parents can receive genetic testing to determine their carrier status during a genetic counseling appointment. A list of genetic services centers in Indiana is included in this packet for your convenience.

Additional Information about CF and Newborn Screening Follow-Up

- CF is most common in Caucasians, but is present in all ethnic groups. Currently, over 1,400 disease-causing *CFTR* mutations have been recognized. The sweat test is more sensitive than the DNA analysis because of the high number of unidentifiable mutations. CF is inherited in an autosomal recessive pattern; however, over 75% of newly-diagnosed patients do not have a family history of CF.
- When informing parents of the positive CF newborn screen and the need for a sweat test, reassure them that **most infants who have a CF newborn screen positive for one mutation do not have CF.** The parent handout titled “Positive CF Newborn Screen – One Gene Change” may be helpful in further explaining the newborn screen and the sweat test. Many questions may also be answered by the genetic counselor.
- **Any infant that has a newborn screen positive for CF should be assessed for signs of malabsorption.** Symptoms can include loose and frequent stools, dehydration and/or poor weight gain. Respiratory problems are uncommon in this age group, but should be reviewed.

Why Screen for CF?

Although there is no cure for CF, there are multiple interventions that improve nutritional and respiratory outcomes and overall quality of life. Several studies have shown that infants diagnosed early through newborn screening have an improved nutritional outcome when compared with those children with a delayed diagnosis.

Newborn screening may also impact family planning for affected individuals. After meeting with a genetic counselor, parents may elect to have further testing that would determine their specific carrier status. This information may impact future plans for prenatal diagnosis or other pregnancy management issues.