Cystic Fibrosis (CF) Fact Sheet for Health Care Providers

Interpreting an Abnormal CF Newborn Screening Result - 2014

What is the screening methodology utilized by the Public Health Laboratory Service, Oklahoma State Department of Health?

A two-tier screen is utilized:

1. Immunoreactive trypsinogen (IRT) level is tested on each filter paper.
2. If the IRT level is elevated, DNA mutation analysis is performed.

Note: Please see http://nsp.health.ok.gov/ for a list of the specific mutations included in the newborn screening panel. Not all specimens receive DNA testing.

What is the normal range for CF screening?

An IRT < 57 ng/ml.

How are abnormal screen results reported?

The IRT level and # of mutations detected will be reported:

Zero mutations, IRT: ≥57 ng/ml

- Infants with this result have approximately a 1% chance of having cystic fibrosis.
- Follow-up or sweat testing is recommended **only if** clinically indicated or if there is a family history of cystic fibrosis.

One mutation, IRT: ≥57 ng/ml

- This result is most consistent with CF carrier status. CF carriers do not need any special medical care.
- **However**, some infants with this result will be found to have CF. This is because the mutation panel used cannot detect all known mutations in the CF gene.
- Newborn screening sweat testing is necessary to distinguish between those infants who are carriers and those who are affected.

Two mutations, **IRT: ≥57 ng/ml**

- This result is consistent with a diagnosis of CF.
- Newborn screening sweat testing is needed for confirmation.
- Some mutations are associated with milder/atypical CF and borderline sweat test results.

Since the screen includes DNA testing, is further testing needed?

Yes! FREE sweat testing and genetic counseling is needed and available at the Newborn Screening Program confirmatory sweat testing sites:

Oklahoma City: OU Medical Center Laboratory Client Services (405) 271-5330
Tulsa: Children's Hospital at Saint Francis Laboratory Scheduling (918) 502-2280

What is CF?

- Cystic fibrosis is an autosomal recessive genetic condition characterized by chronic pulmonary disease and gastrointestinal abnormalities. Most infants with CF are born to parents who are both unknowingly asymptomatic CF carriers and have NO known history of CF in the family.
- Early symptoms of CF include meconium ileus, recurrent cough, wheezing, chronic abdominal pain, loose stools and failure to thrive.

How accurate is CF newborn screening?

Screening for CF results in more *false negatives* than are documented for other established newborn screening tests. Screening tests are not diagnostic, false positives are expected.

What is my role in screening?

You have been listed as the infant's planned Health Care Provider on the filter paper requisition and are required by the Newborn Screening Program (NSP) *Regulations* to arrange follow-up testing as recommended.

For more information or assistance, call (405) 271-6617 or 1-800-766-2223, then press option 2.