

Indiana Newborn Screening Program Cystic Fibrosis Newborn Screening Action Sheet for Healthcare Practitioners

Your patient has a positive newborn screening result for cystic fibrosis.

This infant is at increased risk for cystic fibrosis (CF). During newborn screening, all infants with an immunoreactive trypsin (IRT) level above the laboratory-designated cutoff are screened for changes within the *CFTR* gene. **A positive screen indicates at least one gene change for CF has been identified.**

Positive for one gene change

This infant has an approximately 1/20 – 1/30 chance of having cystic fibrosis. Approximately 95% of infants in this category will be asymptomatic carriers of a CF gene change. Those infants with CF have a second gene change that was not included in the screening analysis.

1. **Contact the family.** Report the newborn screening result.
2. **Schedule the infant for sweat chloride testing at an accredited CF Center.** Alert the Center of the referral and any pertinent clinical information. When discussing the infant's clinical history, **be sure to inform the CF Center that this child had a positive newborn screen.**
3. **Recommend genetic counseling.** The positive newborn screening indicates the patient is likely a carrier for CF. A genetic counselor will review the parents' risks for future pregnancies.

Positive for two gene changes

This infant most likely has cystic fibrosis.

1. **Contact the family.** Report the newborn screening results.
2. **Refer the infant to an accredited CF Center for further testing and evaluation.** This may include a sweat chloride test and evaluation for signs and symptoms of CF. Genetic counseling will also be available.
3. **Contact the CF Center.** Alert the Center of the referral and any pertinent clinical information. When discussing the infant's clinical history, **be sure to inform the CF Center that this child had a positive newborn screen.**

IMPORTANT POINTS

- Term infants may be scheduled for sweat testing **after 48 hours of age.** The likelihood that the sweat test result on a young infant will be valid is good, with sufficient sweat for analysis noted in over 90% of infants.
- Newborn screening is **not** a diagnostic test and will **not** detect all affected infants. Infants that have gene changes not included in the DNA testing panel may be missed. Currently, there are over 1,400 different gene changes for CF; Indiana is testing for 44 of the most common gene changes. Infants or older children with signs or symptoms of CF should be sent for sweat chloride testing **even if they are screen negative in the newborn period.**
- Although CF is more common in Caucasians, it affects all races, including those of Hispanic, African-American, and Oriental descent.