

## Cystic Fibrosis (CF)

### Action required

Contact CF Center before 1 month of age.

### Issues to discuss with CF specialist

- Evaluation of infant
  - § Sweat chloride test
  - § Based on baby's weight, age, and clinical situation, when should baby be seen

### False Positives

**Most children with positive results are CF carriers.** Screening result is considered positive if elevated immunoreactive trypsinogen and one CF mutation is seen. Screening result can be impacted by transfusion. Screening results are not impacted by baby's age at time of screening.

### Review with family

Family has **not** been notified of result by MDH.

Discuss with family that most babies with positive newborn screens do not actually have CF, but that follow-up is important to determine if the baby is affected and can benefit from treatment. Contact CF Center to coordinate sweat test and evaluation. Expect infant to be stable at clinic visit.

Prompt follow-up and interventions are critical.

### NICU issues

Positive newborn screens are seen more commonly in babies who spend time in the NICU. Sweat tests for newborns who spent time in the NICU should be scheduled around 1 month corrected gestational age. For infants < 1800g, sweat testing is indicated if any one of the newborn screens is positive.

Newborn screens cannot be accurately interpreted if collected after transfusion or after 2 months of age.

### Clinical summary

Cystic Fibrosis (CF) is an autosomal recessive disorder that results from defective activity of the CF transmembrane conductance regulator (CFTR protein). Newborns are typically asymptomatic, though some may present with a meconium ileus or poor weight gain. If an infant is not screened and/or left untreated, symptoms begin to appear in childhood and can include progressive lung disease, pancreatic insufficiency, and poor nutritional uptake/weight gain.

Affected children require life-long enzyme treatment, respiratory therapy, and monitoring by both primary care and specialty providers.

**Incidence:** ~ 1/3,200; most common in Caucasian individuals

### Clinical expectations

If treated promptly, children with cystic fibrosis can be expected to develop and gain weight appropriately.

Even with treatment, bacterial endobronchitis, pancreatic insufficiency, and infertility can occur.

Affected children should be monitored for pulmonary function and proper growth and weight gain.

### Resources

**GeneTests:** [www.genetests.org](http://www.genetests.org)

**OMIM:** [www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM](http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM)

**ACT Sheets:** [www.acmg.net/resources/policies/ACT/condition-analyte-links.htm](http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm)

**MN Newborn Screening Program:**  
[www.health.state.mn.us/newbornscreening](http://www.health.state.mn.us/newbornscreening)