

# CYSTIC FIBROSIS



## **CYSTIC FIBROSIS**

Cystic fibrosis is an inherited disease that affects the lungs and digestive system. A defective gene and its protein product cause the body to produce unusually thick, sticky mucus that:

- clogs the lungs and leads to life-threatening lung infections; and
- obstructs the pancreas and stops natural enzymes from helping the body break down and absorb food.

### **Clinical Features in Children with Untreated Cystic Fibrosis**

If left untreated the abnormalities in the CF protein may cause lung and digestive problems, often beginning in early infancy. Recurrent pneumonia and failure to thrive are common and death may occur at an average age of 30-35 years.

### **Causes of Cystic Fibrosis**

Mutations in the *CFTR* gene can alter the structure, function, or production of a cyclic adenosine-5'-monophosphate (AMP)--dependent transmembrane chloride channel protein that is critical for normal functioning of multiple organs. The organs and systems that are affected in CF include the lungs and upper respiratory tract, gastrointestinal tract, pancreas, liver, sweat glands, and genitourinary tract.

### **Laboratory Screening Tests**

The Kansas State Laboratory screens for cystic fibrosis by testing the immuno-reactive trypsinogen (IRT) level in a baby's blood. This is performed off of the blood spot card collected shortly after birth. An elevated IRT level can be a sign of cystic fibrosis. When a baby has an elevated IRT level, a repeat blood spot card is requested and tested. If the second test is still elevated, the infant is referred to a Cystic Fibrosis Foundation accredited facility for a sweat test, which is the confirmatory test for cystic fibrosis.

It should be noted that the IRT levels decrease with the age of a child, so it is important to repeat an initially elevated IRT before two weeks of age.

### **Confirmatory Testing**

The standard confirmatory test for cystic fibrosis is a sweat chloride test. This should be performed at a Cystic Fibrosis Foundation accredited facility. If the initial sweat test is normal, a repeat sweat test at 6 months of age may be recommended by the cystic fibrosis specialist. If there is not enough sweat generated for the sweat chloride test to be accurate, DNA testing should be considered. Consultation with a cystic fibrosis specialist is recommended, as there are various DNA cystic fibrosis panels available.

If an infant has a family history of cystic fibrosis or has meconium ileus but the newborn screening is normal, further evaluation of the infant should be performed, including a sweat test to eliminate the possibility of cystic fibrosis.

## **Treatment for Cystic Fibrosis**

Newborn screening for CF allows for earlier diagnosis and treatment. Studies have shown that newborns who receive treatment early have an improved nutritional status compared to individuals who were diagnosed later. Infants frequently require pancreatic enzyme supplements and water soluble forms of fat soluble vitamins. Supplemental feedings may be required in breastfed infants to achieve normal growth. Infants should be closely monitored for respiratory symptoms. Special treatments for draining sputum - including physical therapy for the chest, physical exercise, and aerosols - are also important. Dietary therapy, which emphasizes the replacement of deficient digestive enzymes, is also critical.

## **Screening Practice Considerations**

The screening test for cystic fibrosis in Kansas is IRT/IRT, which means that the initial and repeat testing is done by measuring the levels of immuno reactive trypsinogen (IRT). If the initial IRT level is elevated, a repeat screening should be done within two weeks of birth as the IRT levels can decrease with age. If the repeat IRT test is not done by two weeks of age, healthcare providers should consider doing DNA testing or a sweat test as the second screen.

If the second IRT is performed within two weeks and is still elevated, a sweat test should be performed at a Cystic Fibrosis Foundation accredited facility. The sweat test is considered the confirmatory test for cystic fibrosis. The CF center may recommend a second sweat test at 6 months of age even if the initial sweat test is normal.

If an infant has a family history of cystic fibrosis or has meconium ileus but the newborn screening is normal, further evaluation of the infant should be performed, including a sweat test to eliminate the possibility of cystic fibrosis.

Sweat tests should be performed at a Cystic Fibrosis Foundation accredited center. See clinics listed below.

## **Medical Consultants for Children with Cystic Fibrosis**

Medical consultants are available to provide consultation for the follow-up, evaluation and long-term management of children with biotinidase deficiency through the State of Kansas.

Dr. Mitzi Scotten  
KU Pediatric Cystic Fibrosis Center  
Kansas City, KS  
Office: 913-588-6224  
Email: [mscotten@kumc.edu](mailto:mscotten@kumc.edu)  
KU sweat test scheduling: 913-588-6224

Dr. Maria Riva  
Wichita Cystic Fibrosis Clinic  
Wichita, KS  
Office: 316-962-2301  
Wichita sweat test scheduling: 316-268-6800

**Overview of Follow-Up Procedure  
Abnormal Cystic Fibrosis Newborn Screening**

- 1) Newborn Screening follow-up team reviews the laboratory reports that are faxed overnight from the lab or records the information provided per telephone call from the laboratory on a white phone information sheet.
- 2) If the IRT result is  $< 60.0$  ng/mL: the results are considered **normal**.
  - a) Follow-up team does not receive results.
  - b) Lab will fax or mail results to doctor listed on NBS card.
- 3) If the IRT result is  $\geq 60.0$  ng/mL the results are considered **elevated**.
  - a) Lab will contact follow-up team via phone with baby's information and test results. Follow-up team will document information on white phone slip.
  - b) Follow-up team will print out baby's information from DHEL database on a yellow sheet of paper, attach the white phone information sheet and write "IRT" with result on yellow sheet.
  - c) Follow-up team will enter data into Access database under CF.
  - d) Follow-up team will call healthcare provider listed on report and:
    - i) Verify that they are seeing the baby.
    - ii) Inform them of results.
    - iii) Ask them to get a repeat NBS card submitted to the KS lab within 1-3 days.
    - iv) Confirm doctor's fax number.
    - v) Inform them that a letter will be faxed to their office with the results and instructions.
  - e) Follow-up team will print CF First Elevated Results letter and fax (or mail, if no fax) to healthcare provider.
  - f) Follow-up team will print CF parent letter and CF parent information sheet and mail to baby's parents to inform them that their child has an abnormal result. NOTE: If infant is in the NICU, no parent letter is sent.
  - g) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 1 month from date of letter.
  - h) Follow-up team will enter data into Excel spreadsheet "Presumptive Totals" located on the "H" drive.
  - i) Lab will fax or mail results to doctor listed on NBS card.
  - j) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 1 month from date of letter. NOTE: Name changes are documented on the lab report. Surname changes are also documented in WebIZ as an alias.
  - k) When complete, paperwork is filed by infant's date of birth.

- 4) If the **repeat** IRT result is > 60.0 ng/mL, the results are considered **presumptive**.
  - a) Lab will contact follow-up team via phone with baby's information and test results. Follow-up team will document information on white phone slip.
  - b) Follow-up team will print out baby's information from DHEL database on a green sheet of paper, attach the white phone information sheet and write "IRT" with result on green sheet.
  - c) Follow-up team will enter data into Access database under CF.
  - d) Follow-up team will call healthcare provider listed on report and:
    - i) Verify that they are seeing the baby.
    - ii) Inform them of results.
    - iii) Ask them to notify parents and arrange appointment with specialist for a sweat test.
    - iv) Confirm doctor's fax number.
    - v) Inform them that a letter will be faxed to their office with the results and instructions.
  - e) Follow-up team will print CF Abnormal Repeat Results letter and CF physician report form and fax (or mail, if no fax) to healthcare provider.
  - f) Follow-up team will print CF parent letter and CF parent information sheet and mail to baby's parents to inform them that their child has an abnormal result. NOTE: If infant is in the NICU, no parent letter is sent.
  - g) Follow-up team will enter data into Excel spreadsheet "Presumptive Totals" located on the "H" drive.
  - h) Lab will fax or mail results to doctor listed on NBS card.
  - i) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 1 month from date of letter. NOTE: Name changes are documented on the copy of the lab report. Surname changes are also documented in WebIZ as an alias.
  - j) When complete, paperwork is filed by infant's date of birth.



# KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

## NEWBORN SCREENING ACT SHEET

**SCREEN FOR:** ELEVATED IMMUNOREACTIVE TRYPSINOGEN (IRT/IRT)

**CONDITION:** CYSTIC FIBROSIS (CF)

**DIFFERENTIAL DIAGNOSIS:** Cystic fibrosis (CF); gastrointestinal abnormalities are also causes of increased IRT.

**METABOLIC DESCRIPTION:** the cystic fibrosis transmembrane conductance regulator (CFTR) protein regulates chloride transport that is important for function of lungs, upper respiratory tract, pancreas, liver, sweat glands and genitourinary tract.

### ACTION TO BE TAKEN IMMEDIATELY:

- Contact family to inform them of the newborn screening result and ascertain clinical status (meconium ileus, failure to thrive, recurrent cough, wheezing and chronic abdominal pain).
- Repeat newborn screening test for IRT within 1-3 days.
- If second IRT level is elevated, determine sweat chloride (sweat test) at a CF Foundation accredited center. If sweat test is normal, repeat the test at 6 months of age.
- If cystic fibrosis is confirmed, clinical evaluation and genetic counseling are indicated.
- Report findings to newborn screening program.

**CONFIRMATION OF DIAGNOSIS:** All initial elevated IRT results should be repeated, preferably within two weeks of age as IRT levels can drop over time. If second screen is positive, follow up with sweat test at an accredited CF center. If the sweat test is normal, the infant should be evaluated at a CF center and the sweat test repeated at 6 months of age. See below for contact information.

**CLINICAL EXPECTATIONS:** Deficient chloride transport in lungs causes production of abnormally thick mucous leading to airway obstruction, neutrophil dominated inflammation and recurrent and progressive pulmonary infections. Pancreatic insufficiency found in 80-90% of cases.

**REPORTING:** Report diagnostic result to family and Kansas NBS program.

### SPECIALISTS:

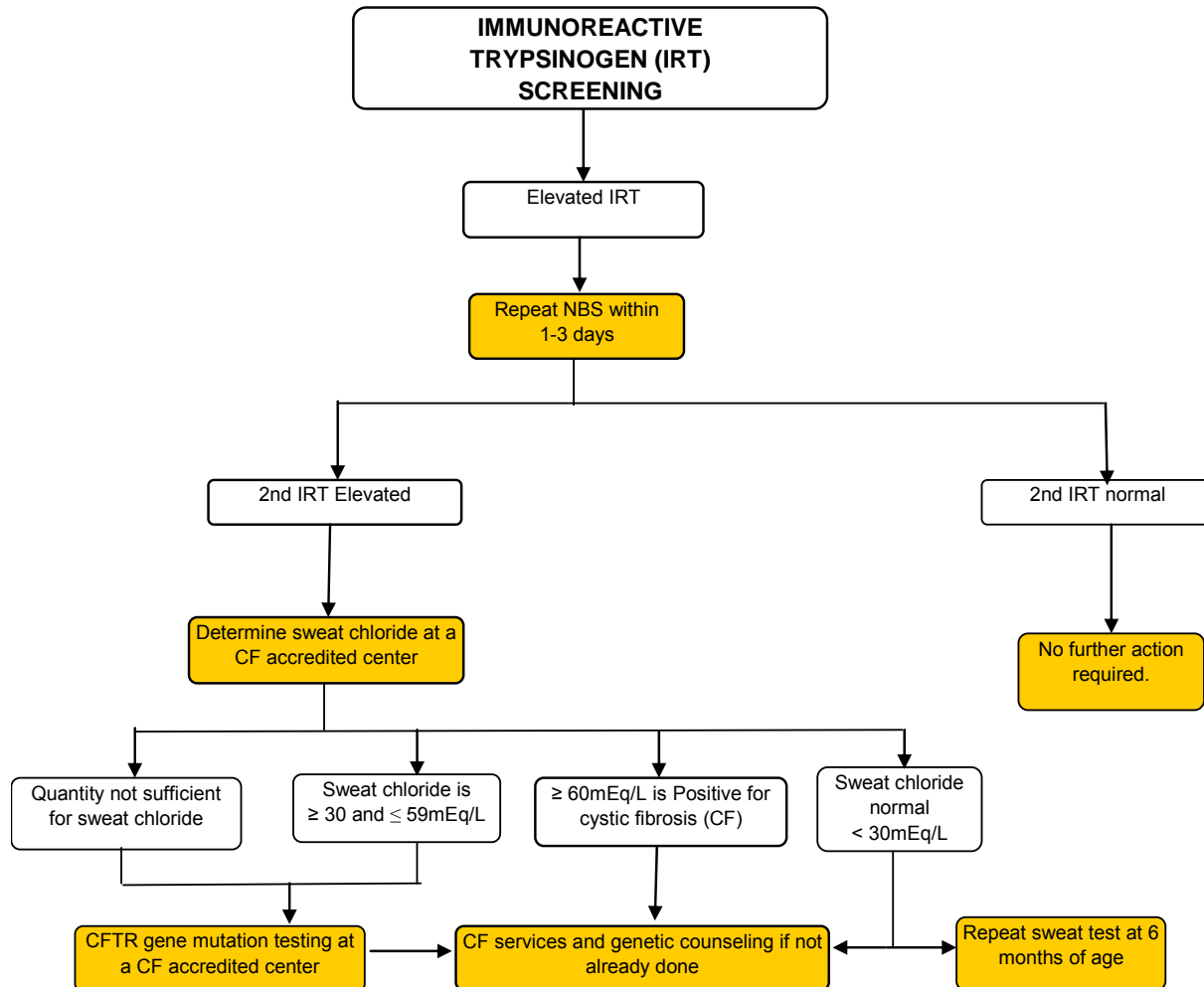
Dr. Mitzi Scotten  
KU Pediatric Cystic Fibrosis Center  
Kansas City, KS  
Office: 913-588-6224  
Email: mscotten@kumc.edu  
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**DISCLAIMER:** These standards and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines



## ELEVATED IMMUNOREACTIVE TRYPSINOGEN (IRT)



Action steps are shown in gold (shaded) boxes; results are in plain boxes.

### Abbreviations/Key

CF = Cystic fibrosis

CFTR = Cystic fibrosis transmembrane conductance regulator

IRT = Immunoreactive trypsinogen

NBS = Newborn Screening

DISCLAIMER: These algorithms and guidelines were adapted from the American College of Medical Genetics algorithm sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines

EXAMPLE OF 1<sup>ST</sup> ELEVATED IRT PHYSICIAN'S LETTER



Mark Parkinson, Governor  
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH  
AND ENVIRONMENT

[www.kdheks.gov](http://www.kdheks.gov)

**Elevated Immunoreactive Trypsinogen (IRT) Lab Report  
Kansas Newborn Screening Program**

Date

Doctor's Name  
Address Line 1  
Address Line 2

RE: Baby's Name  
DOB: xx/xx/xxxx

MOTHER'S NAME: Mother's Name  
MOTHER'S PHONE: xxx-xxx-xxxx

Specimen date: xx/xx/xxxx

**Immunoreactive trypsinogen level:** result ng/mL  
Expected range: <60.0 ng/mL

This elevated immunoreactive trypsinogen activity can be indicative of cystic fibrosis.

The final newborn screening lab report will be sent when all testing is completed.

**RECOMMENDATION:**

Repeat the screening test within 1 to 3 days and send the specimen to the State lab for analysis. (If baby was transfused, repeat screening on day 4 after transfusion.) If the repeat screening also shows an elevated IRT level, a sweat test should be scheduled at a CF Foundation accredited lab. Immediate consultation with one of the consultants listed below is essential if the sweat test is abnormal; call to arrange an appointment if indicated.

***In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.***

For consultation, please contact:

Dr. Mitzi Scotten  
KU Pediatric Cystic Fibrosis Center  
Kansas City, KS  
Office: 913-588-6224  
Email: [msscotten@kumc.edu](mailto:msscotten@kumc.edu)  
KU sweat test scheduling: 913-588-6224

Dr. Maria Riva  
Wichita Cystic Fibrosis Center  
Wichita, KS  
Office: 316-962-2301  
Wichita sweat test scheduling: 316-268-6800

Additional information is available on the Kansas Newborn Screening Website at:  
**[http://www.kdheks.gov/newborn\\_screening/info\\_professionals.htm](http://www.kdheks.gov/newborn_screening/info_professionals.htm)**

You may contact the Newborn Screening Program at (785) 291-3363 or 1-800-332-6262 if you have any questions or concerns.

EXAMPLE OF 2<sup>ND</sup> ELEVATED IRT PHYSICIAN'S LETTER



Mark Parkinson, Governor  
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH  
AND ENVIRONMENT

[www.kdheks.gov](http://www.kdheks.gov)

**Second Elevated Immunoreactive Trypsinogen (IRT) Lab Report  
Kansas Newborn Screening Program**

Date

Doctor's Name  
Address Line 1  
Address Line 2

RE: Baby's Name  
DOB: xx/xx/xxxx

MOTHER'S NAME: Mother's Name  
MOTHER'S PHONE: xxx-xxx-xxxx

1<sup>st</sup> Specimen date: xx/xx/xxxx  
2<sup>nd</sup> Specimen date: xx/xx/xxxx

**Immunoreactive trypsinogen level:** Result  
**Immunoreactive trypsinogen level:** Result  
Expected range: < 60.0 ng/ml

This elevated immunoreactive trypsinogen activity is highly suggestive of cystic fibrosis (CF).

**RECOMMENDATION:**

This repeat screening also shows an elevated IRT level. A sweat test should be scheduled at a CF Foundation accredited lab. Immediate consultation with one of the consultants listed below if the sweat test is abnormal. Please call to arrange an appointment if indicated.

The final newborn screening lab report will be sent when all testing is completed.

***In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.***

For consultation, please contact:

Dr. Mitzi Scotten  
KU Pediatric Cystic Fibrosis Center  
Kansas City, KS  
Office: 913-588-6224  
KUMC sweat test scheduling: 913-588-6224

Dr. Maria Riva  
Wichita Cystic Fibrosis Center  
Wichita, KS  
Office: 316-962-2301  
Wichita sweat test scheduling: 316-268-6800

**\*\*PLEASE COMPLETE AND RETURN THE ENCLOSED PHYSICIAN REPORTING FORM WHEN FOLLOW UP IS COMPLETE \*\***

Additional information is available on the Kansas Newborn Screening Website at:  
[http://www.kdheks.gov/newborn\\_screening/info\\_professionals.htm](http://www.kdheks.gov/newborn_screening/info_professionals.htm).

You may contact the Newborn Screening Program at (785) 291-3363 or 1-800-332-6262 if you have any questions or concerns.

EXAMPLE OF CYSTIC FIBROSIS (CF) PHYSICIAN'S REPORT FORM



Mark Parkinson, Governor  
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH  
AND ENVIRONMENT

www.kdheks.gov

**CYSTIC FIBROSIS (CF) NEWBORN SCREENING  
PHYSICIAN REPORTING FORM**

**\*\*Return this form When Follow-Up is Complete \*\***

Date

Doctor's Name  
Address Line 1  
Address Line 2

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**If this infant is not a current patient of this practice, record name and contact information for Primary Care Physician and return this form.**

RE: Baby's Name  
DOB: xx/xx/xxxx

---

**Baby's name if different than listed**

**LAB RESULTS:** (please fill in and attach copy of specialist's report)

Sweat chloride: \_\_\_\_\_ DNA results: \_\_\_\_\_

Additional lab results: \_\_\_\_\_

**DIAGNOSIS EXCLUDED:** Date Excluded: \_\_\_\_\_

Baby does **NOT** have cystic fibrosis

**DIAGNOSIS CONFIRMED:** Date Diagnosis Confirmed: \_\_\_\_\_

Baby has cystic fibrosis

Date treatment began: \_\_\_\_\_

Baby is a carrier for cystic fibrosis

Baby referred to specialist (please attach copy of specialist's report)

Name of specialist: \_\_\_\_\_

**FORM CONTINUES ON BACK**

Kansas Newborn Screening Program





## **Cystic Fibrosis Information for Health Professionals**

Mutations in the *CFTR* gene can alter the structure, function, or production of a cyclic adenosine-5'-monophosphate (AMP)--dependent transmembrane chloride channel protein that is critical for normal functioning of multiple organs. The organs and systems that are affected in CF include the lungs and upper respiratory tract, gastrointestinal tract, pancreas, liver, sweat glands, and genitourinary tract.

### ✓ **Clinical Symptoms**

The symptoms and severity of cystic fibrosis (CF) vary between individuals. At birth, most newborns appear healthy, but 15-20% of newborns with CF will have meconium ileus. Abnormal secretions produced by the pancreas gland result in malabsorption of nutrients causing diarrhea, vomiting, dehydration, abdominal distension, poor growth, poor weight gain, and abnormal stools in most children. Respiratory symptoms may not be present for years. Thick mucus secretion in the lungs will cause chronic coughing and wheezing. Individuals are highly susceptible to respiratory infections.

### ✓ **Incidence**

In the United States, CF occurs in 1:3200 Caucasians, 1:15,000 African Americans, and 1:30,000 Asians.

### ✓ **Genetics of Cystic Fibrosis**

Mutations in the *CFTR* gene cause cystic fibrosis. The *CFTR* gene provides instructions for making a channel that transports chloride ions in to and out of cells. The flow of chloride ions helps control the movement of water in tissues, which is necessary for the production of thin, freely flowing mucus.

Mutations in the *CFTR* gene disrupt the function of the chloride channels, preventing them from regulating the flow of chloride ions and water across cell membranes. As a result, cells that line the passageways of the lungs, pancreas, and other organs produce mucus that is unusually thick and sticky. This mucus clogs the airways and glands, causing the characteristic signs and symptoms of cystic fibrosis.

### ✓ **Inheritance Patterns**

Cystic fibrosis is inherited in an autosomal recessive pattern. Parents of a child diagnosed with cystic fibrosis are unaffected. These individuals are carriers of the condition and have one normal *CFTR* gene and one abnormal *CFTR* gene. Each pregnancy between carrier parents has a 25% chance of producing a child affected with CF, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

### ✓ **Treatment**

Newborn screening for CF allows for earlier diagnosis and treatment. Studies have shown that newborns who receive treatment early have an improved nutritional status compared to individuals who were diagnosed later. Infants frequently require pancreatic enzyme supplements and water soluble forms of fat soluble vitamins. Supplemental feedings may be required in breastfed infants to achieve normal growth. Infants should be closely monitored for respiratory symptoms. Special treatments for draining sputum - including physical therapy for the chest, physical exercise, and aerosols - are also important. Dietary therapy, which emphasizes the replacement of deficient digestive enzymes, is also critical.

✓ **Screening Methodology**

Testing methodology used in newborn screening to detect Cystic Fibrosis is a single-tier immunoreactive trypsinogen (IRT) assay. A positive newborn screen indicates an elevated immunoreactive trypsinogen level.

✓ **What to do After Receiving Presumptive Positive Cystic Fibrosis Screening Results**

- **Clinical Evaluation: Common findings include meconium ileus, failure to thrive, recurrent cough, wheezing, and chronic abdominal pain; however, patients are frequently asymptomatic.**
- **Laboratory: Repeat immunoreactive trypsinogen (IRT) assay.**
- **All newborns with a second positive IRT screen or history of meconium ileus or a positive family history of cystic fibrosis should have a sweat test done at a CF Foundation accredited laboratory.**
- **An abnormal sweat test is diagnostic for cystic fibrosis.**
- **Infants with a second positive IRT level and normal initial sweat test should be evaluated at a CF Center in 2-4 months and have a repeat sweat test at a CF Foundation accredited laboratory at 6 months of age.**
- **Call KS Newborn Screening Program at 785-291-3363 with questions about results.**
- **Report Clinical Findings to Newborn Screening Program at 785-291-3363.**

✓ **Confirmation of Diagnosis**

Sweat testing at a CF Foundation accredited laboratory is required to diagnose or confirm CF. DNA testing may be necessary in some cases.

✓ **Communication of Results to Parents**

**If a baby has a presumptive positive cystic fibrosis newborn screening result, additional testing needs to be performed to confirm a diagnosis.** In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.

If a baby is diagnosed with cystic fibrosis, the following points should be conveyed to parents:

- ***The baby needs to be followed at a CF center.***
- ***Treatment is life-long. Infant does not need urgent treatment; they will receive advice about treatment from a specialist at the CF center.***
- ***Compliance with treatment is necessary for the best outcome.***
- ***Parents who have a child with cystic fibrosis have a 25% chance with each pregnancy of having another affected child.***
- ***Prenatal diagnosis by molecular genetic testing may be available from laboratories offering custom prenatal genetic testing.***

For consultation, contact:

Dr. Mitzi Scotten  
KU Pediatric Cystic Fibrosis Center  
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Office: 913-588-6224  
Email: [msscotten@kumc.edu](mailto:msscotten@kumc.edu)  
KU sweat test scheduling: 913-588-6224

Dr. Maria Riva  
Wichita Cystic Fibrosis Clinic  
Wichita, KS  
Office: 316-962-2301  
Wichita sweat test scheduling: 316-268-6800

01/16/09

EXAMPLE OF PARENT LETTER FOR CYSTIC FIBROSIS (CF)



Mark Parkinson, Governor  
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH  
AND ENVIRONMENT

[www.kdheks.gov](http://www.kdheks.gov)

Date

Mother's Name  
Address Line 1  
Address Line 2

RE: Baby's Name                      DOB: xx/xx/xxxx

Dear Parent:

Best wishes on the birth of your baby! Shortly after your baby was born, a small blood sample was taken for a test called the Newborn Screen. This test helps parents find out if their baby has certain health problems. A baby can look healthy, but may have a harmful illness that can be found by doing this blood test.

The result of your baby's blood test shows that more testing needs to be done for cystic fibrosis. ***This does not necessarily mean your child is ill.***

**THIS IS WHAT YOU NEED TO DO NOW:**

1. Call your baby's doctor. Say that you have received a letter stating that your baby's Newborn Screen test was not normal. Set up a time for your baby to have a second test done as soon as you can.
2. We have < Doctor's Name > listed as your baby's doctor, and we have notified him/her of your baby's test result. If this is NOT your baby's doctor, please call the Newborn Screening Program at 785-296-0109 so we can contact the right doctor.

**DO NOT DELAY. YOUR BABY'S HEALTH DEPENDS ON YOU.**

If your baby does not have a doctor, or if you have questions about this letter, please call Kansas Newborn Screening at 785-296-0109.

Sincerely,

Jamey Kendall BSN, RN  
Kansas Newborn Screening  
Follow-up Coordinator

Linda A. Williams, MT(ASCP)  
Kansas Newborn Screening  
Follow-up Coordinator



## Cystic Fibrosis Information for Parents

### ➤ Overview

Cystic fibrosis (CF) is an inherited condition. It affects organs in the body, particularly the lungs and digestive system, which become clogged with sticky mucus, making it difficult to breathe and digest food.

### ➤ What is cystic fibrosis?

Cystic fibrosis is an inherited disease of the mucus glands that affects many body systems. The disorder's most common signs and symptoms include progressive damage to the respiratory system and chronic digestive system problems.

Mucus is a slippery substance that lubricates and protects the linings of the airways, digestive system, reproductive system, and other organs and tissues. In people with cystic fibrosis, the body produces mucus that is abnormally thick and sticky. This can obstruct the airways, leading to severe problems with breathing and bacterial infections in the lungs. These infections cause chronic coughing, wheezing, and inflammation. Over time, mucus buildup and infections result in permanent lung damage, including the formation of scar tissue (fibrosis) and cysts in the lungs.

Most people with CF also have digestive problems because the abnormal mucus interferes with the function of the pancreas.

### ➤ Why is newborn screening done for cystic fibrosis?

Newborn screening is done for CF so that babies with this condition can be diagnosed quickly. Research studies conducted over the past two decades have shown that early intervention with nutritional therapies provides distinct benefits including improved height, weight and cognitive function for people with CF. These therapies may impact respiratory function and life expectancy, and reduce hospitalizations.

### ➤ Does a positive result from the Kansas Newborn Screening Lab mean that my baby has cystic fibrosis?

No, not necessarily. Newborn screening tests the baby's level of immunoreactive trypsinogen, but additional tests will need to be done to determine if the baby has CF or not. Kansas has adopted rigorous screening cut-offs in order to identify children with CF as early as possible. With early diagnosis and treatment, long-term health outcomes are improved. Most of the initial positive CF screens will repeat as normal; however **repeat screening is essential to identify children who truly have cystic fibrosis.**

### ➤ How common is cystic fibrosis?

In the United States, CF occurs in 1:3200 Caucasians, 1:15,000 African Americans, and 1:30,000 Asians.

### ➤ How is cystic fibrosis inherited?

Cystic fibrosis is inherited in an autosomal recessive pattern. Parents of a child diagnosed with CF are unaffected. These individuals are carriers of the condition and have one normal CFTR gene and one abnormal CFTR gene. Each pregnancy between carrier parents has a 25% chance of producing a child affected with CF, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

➤ **What are the signs and symptoms of cystic fibrosis?**

Signs and symptoms of CF usually appear in babies under the age of 12 months. However, some children may not develop signs and symptoms of cystic fibrosis until they reach preschool age. In some newborns, the first sign may be a blockage of their intestines (meconium ileus). Other signs in newborns may include:

- Failure to grow
- Bulky and greasy stools
- Frequent respiratory infections

Cystic fibrosis symptoms in children and young adults may include:

- Salty taste to the skin. People with CF tend to have higher than normal amounts of salt (sodium chloride) in their sweat. This may be one of the first signs parents notice because they can taste the salt when they kiss their child.
- Blockage in the bowels.
- Foul-smelling, greasy stools.
- Coughing or wheezing.
- Delayed growth.
- Thick sputum. It's easy for parents to overlook this sign because young children tend to swallow their sputum rather than cough it up.

➤ **How is cystic fibrosis diagnosed?**

**Any initial abnormal newborn screening result requires an immediate repeat newborn screening test. If the second test also is abnormal, the infant should have a sweat test arranged by your doctor.**

➤ **Is there a cure for cystic fibrosis?**

No, there is no cure for cystic fibrosis. The pace of CF science suggests that there is good reason to feel optimistic about the future.

➤ **How is cystic fibrosis treated?**

- Common treatments include lung therapies and breathing treatments, as well as antibiotics, to minimize the risk of lung infections. Complications within the pancreas can be managed by controlling the patient's diet, and providing supplements of pancreatic enzymes to add to food.

➤ **Where can I get additional information?**

Cystic Fibrosis Foundation at [www.cff.org](http://www.cff.org)

[www.cysticfibrosis.com](http://www.cysticfibrosis.com)

GeneTests at <http://www.genetests.org/>

03/23/09