Reporting Title: Cystic Fibrosis

Test Definition: CF0001
Testing Location: Bridgeville, PA
Reporting Location: Melville, NY

Description:
Cystic fibrosis (CF), in the classic form, is a severe autosomal recessive disorder characterized by a varied degree of chronic obstructive lung disease and pancreatic enzyme insufficiency. The incidence of CF varies markedly among different populations, as does the mutation detection rate for the mutation screening assay. To date, over 1,500 mutations have been described within the CF gene, named cystic fibrosis transmembrane conductance regulator (CFTR). The most common mutation, deltaF508, accounts for approximately 67% of the mutations worldwide and approximately 70% to 75% in a North American Caucasian population. Most of the remaining mutations are rather rare, although some show a relatively higher prevalence in certain ethnic groups or in some atypical presentations of CF such as congenital bilateral absence of the vas deferens (CBAVD). Mutations detected include the 23 mutations recommended by the American College of Medical Genetics as well as 16 other mutations

Analytical Method(s):
DNA extracted from dried blood spots for the 23 ACMG/ACOG recommended mutations plus an additional 16 polymorphisms within the Cystic Fibrosis Transmembrane Regulator (CFTR) gene. These mutations were screened for using the Luminex xTAG® Cystic Fibrosis 39 Kit v2 after multiplex polymerase chain reaction (PCR). The test is performed by EGL Genetics, 2460 Mountain Industrial Boulevard, Tucker, GA 30084.

Patient Preparation:
Counsel patient on carrier screening for cystic fibrosis.

Specimen Requirements:
NTD Prenatal Test Requisition Form

Container/Tube: Dried Blood Spot Card

Specimen Volume: Minimum: 2 Spots, Preferred: 5 spots

Specimen Stability: Dried blood spots are stable at ambient temperature for 30 days.

Specimen Rejection Criteria: insufficient volume, layering, insufficient drying time

Additional Information:
1. Indications for Testing: General population screening of pregnant women. Male partner of woman identified as a cystic fibrosis carrier
2. Special Timing: None.

CPT Code: 81220

Reference Values:
An Interpretive Report will be provided
Supplemental Report:
No

Testing Algorithm:

Follow-up testing:
1. Mutation Identified: testing of Male Partner
2. Mutation Identified in both Parents: Counsel parents on risk of cystic fibrosis in patient and offer CVS or amniocentesis for diagnostic confirmation.

Consents/Authorizations:

1. A patient courtesy consent form is available for Cystic Fibrosis Carrier Testing.
2. A New York State informed consent acknowledgment form is required for Cystic Fibrosis Carrier Testing. This is to be signed by the healthcare provider and kept on file at Eurofins NTD

Disclaimer:

The xTAG® Cystic Fibrosis 39 kit v2 has been cleared by the U.S. Food and Drug Administration (FDA) for use with human whole blood samples. The performance characteristics of the test using dried blood spot samples have been determined by EGL Genetics, Inc. and has not been approved by the FDA.
Prenatal Screening Test Requisition Form Instructions

1. Account Information – Please enter ordering physician name and referring (if any) name and phone number, if applicable. A provider signature is required for patients with Medicaid.

2. Specimen Labels – Preprinted with the requisition number. Please enter the patient’s last and first name EXACTLY as they appear on the requisition form. Affix label(s) to patient specimen(s). Please complete date drawn and drawn-by fields.

3. Patient Information – For all tests, please complete patient’s weight, ethnicity and current pregnancy information. Complete additional patient history as appropriate for test(s) ordered. Please note, all patient information requested is used to ensure the most accurate risk assessment possible for your patient.

4. Gestational Age – Complete for tests other than First Trimester Screen I FB, Sequential Screen I FB, Preeclampsia Screen™ I T1 or Maternal Fetal Screening™ I T1 which require CSL (see section 2).

5. Biophysical Information – Complete this section for preeclampsia screening only.

6. Ultrasound Information – Please provide sonographer and supervisor names and credentialing numbers. Enter all ultrasound information as appropriate for test(s) ordered.

7. Test Requests – Tests are ordered by specimen type. Check all tests that apply and provide appropriate ICD codes.

8. Cell Free DNA – ECW/TCI physician and patient signatures are required.

9. Billing Information – Provide photocopy of front and back of insurance card or print the information in the required fields.

10. Patient Signature – Required for all tests.

Please call 1-888-NTD-LABS (683-5227) for further assistance.

Revision: 1
Effective date: May 18, 2018
Specimen Collection Instructions

Dried Blood Collection Procedure

1. Prepare material needed for sample collection and place on a clean dry surface. Please note, collection supplies have expiration dates, please confirm supplies are not expired prior to use.
   - NTD Prenatal Test Requisition Form
   - Alcohol
   - Sterile Gauze
   - Sterile lancet
   - 903™ Five Spot Collection Card
   - Pre-addressed, pre-paid business reply envelope

2. Complete the NTD Prenatal Screening Requisition Form ensuring all fields are completed accurately and legibly. Making sure the patient’s name appears on the specimen label exactly as it appears in the Patient Information section.

3. For first trimester screening, pregnancy is dated based on the CRL at the time of the nuchal translucency examination. This gestational age may sometimes vary from that estimated at the time of blood collection. Therefore, we recommend that specimens be drawn at 10 weeks or later to prevent recalculation of gestational age resulting in the patient being too early for testing at the time of blood draw.

4. For second trimester AFP dried blood screening test, pregnancy is dated based on EDC, with gestational age 15 – 21 weeks 6 days.

5. Remove the specimen label with patient name, from the requisition form and place it on the designated area on the 903™ Five Spot Collection Card.

Preparation and Site Selection is Vital to the Success of the Blood Collection Process

1. Choose either the “Ring” or “Middle” finger for the test because they are not as sensitive to pain as the index finger and bleed more easily.

2. Have patient wash hands thoroughly with warm soapy water while massaging the entire length of the finger for about 30 seconds to increase blood flow.

3. The incision should be made perpendicular to the finger print about 1/4 inch from the nail bed, at the point where the finger begins to curve (see Figure 1). Avoid the tip of the finger because of increased sensitivity and less robust bleeding.

Dried Blood Collection Process Using the Finger-Stick Lancet

1. Have patient remain seated during the blood collection process. To increase blood flow, instruct patient to rub hands together for 10 seconds and then allow the hand with the selected finger to rest for 30 seconds.

2. Open the flap of the 903™ Five Spot Collection Card to expose the blood spot circles and fold back away from the spots during the collection and drying process. (Fig. 2)

3. Cleanse the finger with an alcohol swab and wipe dry with sterile gauze.
   (Failure to allow alcohol to dry may adversely affect results.)

continued on back
Dried Blood Collection Process Using the Finger-Stick Lancet (continued)

4. Firmly press the tip of lancet on the selected puncture site. Puncture using a sterile lancet. (Fig. 3)

5. Immediately wipe away the first small drop of blood with sterile gauze.

6. Lower the hand to increase blood flow and allow a second, larger drop of blood to form. Apply gentle continuous pressure if necessary below the puncture incision site but avoid excessive massaging. Keep the drop balanced on the finger as it forms. When the drop is large enough and appears ready to roll, touch the underside of the card to the blood drop so it soaks into one of the circles. (Fig. 4)

7. Continue to collect blood drops as above until all five circles have been spotted.
   Note: USE ONE LARGE DROP OF BLOOD PER CIRCLE.
   - Do not overlay multiple drops.
   - Blood should saturate at least 75% of each circle on ECTH sites.
   - If more blood is needed, puncture another finger.

8. Following blood collection, gently press a dry sterile gauze pad on the incision site until bleeding stops, and then bandage the finger.

9. Allow blood spots to dry at room temperature for a minimum of 3 hours. Keep away from direct sunlight and heat.

Clean-Up Procedure

1. Lancets are designed to be used only once, please do not reuse.

2. Dispose of the used lancet, gauze, alcohol swabs and all other used materials.

Mail-In Procedure

1. After drying, close the protective flap on the 903™ Five Spot Blood Card and tuck under the front flap where the label has been affixed.

2. Staple the 903™ Five Spot Blood Card to the requisition in the designated area.

3. Fold the requisition and place with the 903™ Five Spot Blood Card and any applicable insurance copies into the postage paid transport envelope and seal.

4. Store the sealed specimens in a cool, dry place until transported.

5. Specimen Stability: Dried blood spots are stable at ambient temperature for 30 days.

6. Mail to Eurofins NTD using the pre-addressed, pre-paid business reply envelope.
Collection of the Dried Blood Sample using a DIFF-SAFE device:

Warning: Use only Red-Top Vacutainer® tubes or tubes with NO additives. Purple-Top tubes or tubes with additives produce significant interference with testing.

1. Collect 2-4 cc whole blood into a Red-Top VACUTAINER® tube.

   Note: Immediately transfer blood to the blood collection card using a syringe or DIFF-SAFE® blood dispenser before the blood has a chance to clot.

2. Holding the tube in upright position, INSERT the DIFF-SAFE cannula into the stopper.
3. INVERT the tube and position DIFF-SAFE on target surface (blood circle).
4. Gently PRESS tube downward until a drop forms at the tip of the DIFF-SAFE.
5. RELAX pressure as soon as the blood fills the circle and move to the next circle
6. Spot all 5 circles before the blood has a chance to clot.
7. Allow blood spots to dry at room temperature for a minimum of 3 hours. Keep away from direct sunlight and heat.
8. Store specimens in cool dry place until transported.

Special handling needs between time of collection and time received by laboratory

Allow blood spots to dry at room temperature for up to 3 hours in a horizontal position after collection. Keep away from direct sunlight and heat, and do not touch circles after blood application. After drying, place the Dried Blood Sample card in the transport envelope and seal. Store the sealed specimens in a cool, dry place until transported. Transport by regular postal mail or by FedEx.

Positive Patient Identification and Specimen Labeling

The phlebotomist is responsible for correctly identifying the patient using two unique patient identifiers that include the patient's complete first and last name, date of birth and/or medical record or hospital number.

Every patient must verbalize his/her name to the phlebotomist, if able to do so by asking "Would you please tell me (or spell) your name and birthdate.". It is unacceptable for the phlebotomist to ask the patient to confirm his/her name that was verbalized by the phlebotomist.

The Specimen MUST be labeled with two forms of patient ID that match EXACTLY with the information on the test requisition. The test Requisition has peel off labels preprinted with the Test Requisition Number that may act as one form of ID. Please print the patient's full first and last names on the label exactly as they appear on the requisition and affix to the specimen. Please retain the requisition number in your records and use if inquiring about test results.

If using your own specimen labels please include two unique patient identifiers that include the patient's complete first and last name, date of birth and/or medical record or hospital number and/or the test requisition number. All information must match exactly on the test requisition form.

Enter the name of the person drawing the specimen on the test requisition to allow for traceability.
### NTD Prenatal Screening Requisition Form

**Prenatal Screening Requisition**

**Physician Information**

- **Name**: [Redacted]
- **Address**: [Redacted]
- **Medical Record #:** [Redacted]
- **Physician Code**: [Redacted]

**Patient Information**

- **Date of Birth**: [Redacted]
- **Sex**: [Redacted]
- **Race**: [Redacted]
- **Twin**: [Redacted]
- **Smoker**: [Redacted]

**Biophysical Information**

**Ultrasound Information**

- **March Anti-Hemorrhage**: [Redacted]
- **NT Measurement**: [Redacted]
- **Right**: [Redacted]
- **Left**: [Redacted]

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**Billing Information**

- **Insurance Claim #:** [Redacted]
- **Financial Information #:** [Redacted]

**Specimen Labeling**

- **Date Drawn**: [Redacted]
- **Drawn By**: [Redacted]

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© This document is copyright of Eurofins Durin the actual use of this form, cover page need not be printed.
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Example Report

Cystic Fibrosis Carrier Testing Final Report

Patient Name: DOE, JANE
Date of Birth: 09/01/81
Gender: U
Date Received: 03/12/18
Date Collected: 03/10/18
Date of Service: 03/09/18
Ethnicity: Afr. Amer./Carib.
Patient ID #: 100EXP99999
Accession #: 999999

Mutations Analyzed

dF508 dI507 G542X G85E R117H 621+1G>T 711+1G>T
2785+5G>A 1078delT R334W R347P A455E 1717+1G>A R560T
B553X G551D 1808+1G>A 2184delA 3120+1G>A R1162X 3650delC
3849+10kbc-T W1128X N1303K 394delIT Y122X R347H V520F
A559T S549N S549R 1889+5G>T 2183AA>G 2307mAs Y1092X
M1101K S1255X 3876delA 3905insT

Result: No pathogenic variant detected.

Interpretation: A negative test result does not rule out carrier status of cystic fibrosis. The risk that this individual carries a cystic fibrosis mutation other than the ones tested depends on family history and ethnicity. If the patient’s reproductive partner is a CF carrier, genetic counseling and further testing are recommended.

Ethnicity | Assay Detection Rate | Carrier Risk Before Testing | Carrier Risk with a Negative Result
---|---|---|---
Ashkenazi Jewish | 94% | 1/24 | 1/400
Caucasian (non-Hispanic) | 89% | 1/25 | 1/227
Hispanic | 73% | 1/38 | 1/214
African American | 69% | 1/41 | 1/180
Asian American | 54% | 1/94 | 1/204

Methodology: This panel of 39 pathogenic variants in the CFTR gene includes the recommended ACMG panel of 23 common pathogenic variants. These variants were tested using the Luminex xTAG® Cystic Fibrosis 39 Kit v2 after multiplex polymerase chain reaction (PCR). The xTAG® Cystic Fibrosis 39 kit v2 has been cleared by the U.S. Food and Drug Administration (FDA) for use with human whole blood samples. The performance characteristics of the test using dried blood spot samples have been determined by EOL Genetics, Eurofins Clinical Diagnostics and have not been approved by the FDA. The test result should always be interpreted in the context of clinical presentation and family history.

References:
1) Assay detection rates were provided by the manufacturer (Luminex Corporation)

Jonathan B. Cernichello, Ph.D., Laboratory Director
Terrence W. Hallahan, Ph.D., Laboratory Director
The test was performed by EOL Genetics, 2400 Mountain Industrial Boulevard, Tucker, GA 30084
Tel: 855-831-7447, Fax 470-378-2250
Email: jbh@eolgenetics.com, JHC@eolgenetics.com
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This report was generated on 05/04/2018 09:31:28 PM
CSE
Physician Attestation to Obtain Informed Consent

Acknowledgement of Practitioner’s Obligation to Obtain Informed Consent for Genetic Testing

- I understand that The New York State Department of Health, Clinical Laboratory Standards of Practice requires NY-permitted laboratories to notify ordering practitioners that they are required to obtain informed consent for genetic testing from their patients.
- I am aware that Eurofins, NTD, LLC (PFI 3173) (“Eurofins”) provides a genetic test consent form that includes test-specific information to aid me in fulfilling my obligations to obtain informed consent.
- I understand that I have been apprised that I may obtain these forms in hard copy from Eurofins, and that the forms are also available on the Eurofins website (http://www.ntdlabs.com/providers/consent_forms.php).
- I acknowledge that this information has been made available to me for patient use in decision-making and the informed consent process.
- I acknowledge that I have reviewed and understand the informed consent for genetic testing form relevant to the testing I am ordering as described above and that the information contained in the form shall be conveyed to each patient and/or their guardian in obtaining full and effective informed consent for each genetic test I order.
- I confirm that I shall maintain documentation that I obtained informed consent for such testing in each of my patients’ medical charts.
- I confirm that I will make available and provide a copy of the signed patient consent form to Eurofins NTD, LLC upon request.

Practitioner Signature ___________________________ Date __________________

Printed Name ___________________________

Hospital, Facility or Clinic Name ___________________________

Address ___________________________

City, State, Zip ___________________________

Phone # ___________________________

Fax # ___________________________

NTD Account # ___________________________

Please Return to:
Eurofins NTD, LLC, 80 Ruland Road, Suite 1, Melville, NY 11780
Fax: 631-425-0864
Patient Informed Consent for Genetic Testing

- **MFM490 Spinal Muscular Atrophy: Carrier Screen**
  - Spinal muscular atrophy (SMA) is the second most common, autosomal recessive disorder in Caucasians, with a carrier frequency of 1/60. SMA is a common cause of infant death and is characterized by severe and progressive symmetrical muscle weakness and wasting, joint contractures, respiratory insufficiency, and feeding and sleep difficulties. It has variable age of onset and severity of symptoms.
  - Limitations:
    - This carrier assay tests for the common SMA deletion (exon 7 and 8 deletion) only. Single point mutations or variants other than the known gene will not be detected.
    - Approximately 26% of affected individuals will have a deletion on one chromosome with two normal SMN1 copies on the second chromosome. This assay will not detect these carriers.
    - This assay will not report alpha-spectrin deletions.

- **MFRAX Fragile X: CGG Repeat Analysis**
  - Fragile X syndrome is the most common inherited (X-linked) form of intellectual disability. Additional symptoms include autism, behavioral problems, and characteristic facial features in affected males. This test can also identify premutation carrier females who are at risk for developing premature ovarian insufficiency, characterized by infertility, early menopause, and other ovarian problems, and premutation carrier males who are at risk for developing late onset fragile X-associated tremor/ataxia syndrome characterized by problems with movement and thinking ability after approximately the age of 50.
  - Limitation:
    - Approximately 1/3 of cases of AMM-associated intellectual disability are due to mutations that cannot be detected by this test. Other testing such as sequencing and mutation/duplication of the FRAX gene may be warranted.

- **CF Cystic Fibrosis: CTR Common Mutation Panel [Dried blood sample submission only]**
  - CFTR Common Mutation Panel tests for only the 19 most common mutations found in a Caucasian population, while the expanded panel tests for 142 mutations that are found across all ethnic groups. Cystic fibrosis (CF) is an autosomal recessive disorder involving multiple organ systems. Classic CF primarily involves the respiratory and digestive systems, and may have a range of clinical severity. Thick mucus accumulation in the lungs leads to breathing difficulties, infection and poor digestion. The average life expectancy is in the 30s. Congential bilateral absence of the vas deferens (ABVD) is seen in men without pulmonary or digestive symptoms of CF, and results in absence of sperm in the semen.
  - Limitations: Pathogenic variants in regions other than the targeted area will not be detected by this analysis.

- **MM580 ACOG/ACMG Carrier Screen: Targeted Mutation Panel**
  - The ACOG/ACMG carrier screen tests for cystic fibrosis, spinal muscular atrophy, and eight other disorders common to those of Ashkenazi Jewish descent. This screen is limited to those conditions recommended by the American College of Obstetrics and Gynecology and the American College of Medical Genetics for screening during pregnancy.
  - Fragile X testing is not currently recommended by ACOG/ACMG unless there is a family history and therefore not included in this panel.
  - Fragile X testing may be ordered separately.
  - Limitations: Pathogenic variants in regions other than the targeted area will not be detected by this analysis.

- **MM480 Pan-Ethnic Carrier Screen: Targeted Mutation Panel**
  - The Pan-Ethnic Carrier Screen tests for 148 genes that cause autosomal recessive and X-linked conditions. Includes Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X syndrome, and 143 other genetic disorders related to intellectual disability, mobility impairment, visual impairment, joint and bone disorders, nervous system abnormalities, developmental delay, hearing loss, skin irregularities, and metabolic syndromes.
  - Includes only most common mutations in each disorder.
  - Limitations: Pathogenic variants in regions other than the targeted area will not be detected by this analysis.

- **MM470 Pan-Ethnic Carrier Screen: Gene Sequencing Panel**
  - The Pan-Ethnic Carrier Screen tests for 148 genes that cause autosomal recessive and X-linked conditions. Includes Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X syndrome, and 143 other genetic disorders related to intellectual disability, mobility impairment, visual impairment, joint and bone disorders, nervous system abnormalities, developmental delay, hearing loss, skin irregularities, and metabolic syndromes.
  - Looks at an entire coding region for more mutations and rarer mutations.
  - Limitations:
    - Due to technical issues the results may be inconclusive and the test might need repeating. Results may also be inconclusive due to identification of a variant of unknown significance.
Patient Informed Consent for Genetic Testing

General Information, Limitations, and Risks:

- This test is indicated for individuals or couples seeking to assess reproductive risk for a variety of conditions and individuals or couples that are considered at high risk due to ethnicity, family history, or clinical indication.
- Carriers of autosomal recessive disorders are typically healthy individuals but can pass on their genetic variants to children. If both partners are identified as carriers of the same genetic disorder, they have a 25% chance of having a child affected with that disorder with each pregnancy.
- Women who are carriers of X-linked conditions are typically healthy individuals but have a 50% chance of having a son who is affected with the condition or a 50% chance of having a daughter who is also a carrier. Carrier status may also be misleading.
- DNA testing requires a blood sample or saliva sample, both of which have risks associated with obtaining the sample. Additional samples may be needed if the sample is damaged in shipment or inaccurately submitted. In order to perform accurate prenatal testing, samples from the affected individual, parents, or additional family members may be required.
- DNA-based studies performed are specific to the condition(s) indicated above. The accuracy of genetic testing is limited by the methods employed, the clinical diagnosis, and the nature of the specific condition for which testing is requested. In some cases, the test may detect an abnormality, called a mutation, in the gene. In other cases, the test may be unable to identify an abnormality although an abnormality may not exist. This may be due to the current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of changes (mutations) in a gene.
- As with any complex genetic test, there is always a small possibility of a failure or error in sample analysis. Extensive measures are taken to try to avoid these errors. The methods are not 100% accurate due to the possibility of rare genetic variations in the DNA of an individual or due to the complexity of the testing itself. A low error rate, approximately 1 in 1000 samples, is generally estimated to exist in a laboratory.
- Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to the patient's ordering provider, who will then review and discuss the test results with me.
- Patient-identifying results and information at Eurofins NTD, LLC will remain confidential and may only be released to other parties with my expressed written consent or as permitted or required by applicable law.
- I understand that no tests other than those authorized shall be performed on my sample and that the sample shall be discarded within sixty days after taken. However, Eurofins NTD, LLC performs research and development studies to improve and validate existing and new tests and to advance biomedical knowledge. I and my heirs will not receive payments, benefits, or rights to any resulting products or discoveries. Patient permission is requested for the use of patient-de-identified sample in research and development studies and is entirely voluntary.
- If I have additional questions, I understand that I may wish to obtain further professional genetic counseling prior to consenting to this testing.

My signature below acknowledges my voluntary participation in the genetic tests ordered by my physician and verifies that I have been appropriately counseled about the testing process and the different possible outcomes.

Patient/Guardian Signature  Printed Name  Date

Physician/Counselor/ Clinician Statement:
I have explained DNA testing to the patient/parent/guardian. The consent form and limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test genetic counseling. I will use my independent professional judgment and the patient’s best interests in advising the patient/parent/guardian regarding DNA test results, the use and limitations of same, and any research study, clinical trial, drug, treatment or device brought to my attention by Eurofins NTD, LLC or others.

Healthcare Provider Signature  Printed Name  Date

A signed physician acknowledgement of NY informed consent for genetic testing must be on file at Eurofins NTD, LLC to permit testing and processing.

This Patient Informed Consent form is to be maintained by the physician in the patients' medical file. DO NOT Return to Eurofins, NTD, LLC.
Physician Information Brochure
Patient Information Brochure

What is cystic fibrosis?

Cystic fibrosis, or CF, is a life-threatening disease of certain glands in the body—in particular, the ones that produce mucus and sweat. CF is inherited, or passed genetically, from parents to children.

In healthy people, mucus helps to keep organs from drying out or getting infected. But in people who have CF, the mucus is thick and sticky. It can build up in organs—such as the lungs, pancreas, liver, intestines, and sinuses—causing infections and digestive problems.

CF also causes people to lose a lot of salt when they sweat. This can result in dehydration, increased heart rate, tiredness, weakness, and decreased blood pressure.

People who have CF are also at higher risk of developing diabetes, osteoporosis (a disease in which bones become fragile and likely to break), infertility (in men), and problems getting pregnant.

The average lifespan of a person who has CF is 37 years. CF does not affect intelligence.

How is CF inherited?

CF results when someone has 2 copies of an abnormal (mutated) gene. One is inherited from the mother and one is inherited from the father. This gene makes a protein that works incorrectly, producing thick, sticky mucus and salty sweat.

Each time 2 carriers conceive, there is a:

- 25% chance their child will have CF
- 50% chance their child will be a carrier of the abnormal CF gene, but not affected with CF
- 25% chance their child will not have CF and will not be a carrier of the abnormal CF gene

Could I be a carrier of CF?

A CF carrier is a person who has inherited one abnormal gene from one parent, and one normal gene from the other. Although CF carriers have no symptoms, they can pass the abnormal gene on to their children.

There are about 12 million CF carriers in the United States. Many of them don’t know they have this abnormal gene.

Who is at risk of developing CF?

CF affects about 1 in 3,500 Caucasians in the United States. There are about 36,000 people who have CF: about 1,000 new cases are diagnosed each year.

CF affects both males and females, including people from all racial and ethnic groups. CF is most common among those of Ashkenazi Jewish descent and Caucasians with a Northern European background; however, CF occurs in people of all ethnicities.

Why is CF testing recommended?

The American College of Obstetricians and Gynecologists (ACOG) recommends genetic carrier testing for CF be offered to all couples who are thinking about having a child or who are already pregnant. Testing may show whether one or both partners carry the abnormal CF gene.
Is CF testing required?

CF testing is not required. The decision to be tested is a personal choice and is up to the individual since they have been informed about the test. Talk to your doctor to figure out whether testing is right for you.

How is CF testing performed?

The CF test is a simple blood test. You or your doctor should provide information about your race, ethnicity, and any personal or family history of CF to help with the interpretation of the results.

Immediately after the blood collection is completed, the blood sample and test requisition should be sent directly to NTD Labs for testing. The results of the blood test are maintained at NTD Labs and made available to your physician 2 to 4 days after the sample is received. The test used at NTD Labs covers all mutations for which ACOG recommends testing.

What does a positive screen mean?

A positive screen means you have an abnormality in one of your CF genes, making you a CF carrier. The next step is to test your baby's father. If the test shows the father is not a carrier, there remains only a very slight possibility your child will inherit CF.

What does a negative screen mean?

A negative screen means no abnormalities were detected in your CF genes; therefore, your chance of having a baby with CF is low. There remains a small chance, however, that you could be a carrier because some gene abnormalities are rare and some have not yet been discovered.

What additional testing will be needed if both parents are found to be CF carriers?

If the mother and father are both CF carriers, prenatal diagnostic testing by chorionic villus sampling (CVS) or amniocentesis can be performed to determine if the baby has inherited CF. Talk to your doctor to find out if prenatal diagnostic testing is right for you.

Genetic testing for cystic fibrosis

What YOU need to know

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