Informed Consent/Decline for Cystic Fibrosis Carrier Screening

(Continued from other side)

My signature below indicates that I have read, or had read to me, the above information and I understand it. I have also read or had explained to me the specific disease(s) or conditions(s) tested for, and the specific test(s) I am having, including the test descriptions, principles, and limitations. I have had the opportunity to discuss the purposes and possible risks of this testing with my doctor or someone my doctor has designated. I know that genetic counseling is available to me before and after the testing.

I have all the information I want, and all my questions have been answered.

I have decided that:

☐ **I want CF carrier screening.**

☐ **I do not want CF carrier screening.**

Patient Signature

Date

Obtained by

This model informed consent form is provided by Integrated Genetics as a courtesy to physicians and their patients.

1. Update on Carrier Screening for Cystic Fibrosis. ACOG Committee Opinion Number 486. April 2011.
2. Cystic Fibrosis Foundation Patient Registry 2009
3. Rohlfs, E, et al. Cystic Fibrosis Carrier Testing in an Ethnically Diverse US Population. *Clin Chem* 2011; 57(6):841–848.

About Integrated Genetics

Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years.

This brochure is provided by Integrated Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web sites:
www.mytestingoptions.com
www.integratedgenetics.com

Integrated GENETICS
LabCorp Specialty Testing Group

Integrated Genetics Client Services
(800) 848-4436

Cystic Fibrosis Carrier Screening

A common chronic, life-shortening inherited disease of children and young adults

Integrated GENETICS
LabCorp Specialty Testing Group

Cystic Fibrosis Carrier Screening

The American College of Obstetricians and Gynecologists supports that couples planning a pregnancy, or those already pregnant, be offered cystic fibrosis (CF) carrier screening. Screening can help determine if a couple is at risk of having a child with CF.[1]

This brochure contains general information about CF, how it is inherited and CF carrier screening.

If you have any additional questions or are seeking CF testing due to the presence of CF symptoms, male infertility, abnormal prenatal ultrasound findings, a family history of CF, or for testing of an unborn baby, please contact your doctor or genetics professional for additional information.

What is cystic fibrosis?

CF is a genetic disease that affects about 1 in 3,500 births each year in the United States.[2] CF causes the body to produce abnormally thick mucus, leading to life threatening lung infections, digestion problems, diarrhea, poor growth and male infertility. Symptoms of the disease range from mild to severe. Individuals with mild CF may reach adulthood and be unaware that they have the disease. However, the average life span for individuals with CF is 37 years. CF does not affect intelligence.[1]

What is a CF carrier?

Every person has two copies of a gene, one inherited from each parent. If a person has one normal CF gene and one altered CF gene, then that person is a carrier of CF. Having only one altered copy of the CF gene is not enough to cause the disease, so a CF carrier does not have symptoms.

How is CF inherited?

If both parents are carriers of an altered CF gene, then there is a chance that each parent will pass an altered gene on to their child. Having two copies of an altered CF gene results in CF. With each such pregnancy there is a:

- 25% (1 out of 4) chance that the child will inherit two normal CF genes, and will be neither a carrier, nor affected with the disease
- 50% (2 out of 4) chance that the child will inherit one normal and one altered CF gene, and will be a carrier of CF
- 25% (1 out of 4) chance that the child will inherit two copies of an altered CF gene, one from each parent, and will be affected with the disease

Can anyone be a CF carrier?

Yes. CF can occur in any ethnic background. If there is no one in your family with CF, your risk for being a CF carrier is determined by your ethnic background (See Table 1). CF is common among people of Caucasian and Ashkenazi Jewish descent. The disease is less common among those of Hispanic, African American, Native American or Asian backgrounds.

If a relative of yours has CF, or is known to be a carrier of CF, your chance of being a carrier is greater based on your family history.

Table 1

Ethnicity	CF Carrier Risk in People with No Family History of CF[1]
Caucasian	1 in 25
Ashkenazi Jewish	1 in 24
Hispanic	1 in 58
African American	1 in 61
Asian	1 in 94

What is the purpose of CF carrier screening?

The purpose of CF carrier screening is to see if a couple is at increased risk for having a child with CF. The screening test requires a sample of blood or mouthwash and results are usually ready within 5 to 10 days. If a couple has not yet conceived, one partner is usually tested first. If a woman is already pregnant, a couple may opt to be tested at the same time.

What do the results mean?

A negative result significantly lowers, but does not completely eliminate, the risk of being a CF carrier. Less common alterations (mutations) in the CF gene are not included in carrier screening. For most individuals, no further testing is needed. Testing for rare CF mutations is available if indicated.

If your testing identifies one altered CF gene, then you are a carrier. Since both parents must be carriers for the baby to be at risk for CF, your partner should be tested.

Rarely, DNA testing may identify two altered genes in a healthy individual. When this occurs, further medical evaluation and testing of additional family members may be recommended.[1,3]

What if both my partner and I are CF carriers?

If both you and your partner are found to be carriers of an altered CF gene, there is a 25% (1/4) chance with each pregnancy that the child will be affected with CF. In this situation, you may be referred for genetic counseling. There are several choices couples in your situation can make when thinking about possible future pregnancies. Some couples decide to:

- Have prenatal testing, such as amniocentesis or chronic villi sampling, to determine whether or not the unborn baby has inherited the two altered CF genes
- Accept this level of risk and have children without further testing
- Go through in vitro fertilization and test the embryos using preimplantation genetic diagnosis (PGD)
- Adopt children
- Use donor sperm or donor eggs
- Not have children

If a couple does not want prenatal diagnosis, then newborn screening for CF is available in all states.

Is CF screening required?

No. The decision to accept or decline screening is entirely up to you.

Informed Consent/Decline for Cystic Fibrosis Carrier Screening

You should be certain you understand the following points:

1. The purpose of my genetic test is to determine whether I, or my fetus if fetal testing is ordered, have mutation(s) known to be associated with cystic fibrosis (CF).
2. This testing is done on a small sample of blood; in some cases a mouthwash sample can be used. For the fetus, testing is done on amniotic fluid, CVS or fetal blood.
3. Mutations are often different in different populations. I understand that the laboratory needs accurate information about my family history and ethnic background for the most accurate interpretation of the test results.
4. When CF testing shows a mutation, then the person is a carrier or is affected with CF. Consulting a doctor or genetic counselor is recommended to learn the full meaning of the results and to learn if the additional testing might be necessary.
5. When the CF testing does not show a known mutation, the chance that the person is a carrier or is affected is reduced. There is still a chance to be a carrier or to be affected because the current testing cannot find all the possible changes within a gene.
6. In some families, CF testing might discover non-paternity (someone who is not the real father), or some other previously unknown information about family relationships, such as adoption.
7. In the case of twins or other multiple fetuses, the results may pertain to only one of the fetuses.
8. In the case of abnormal diagnostic results, the decision to continue or to terminate the pregnancy is entirely mine.
9. The decision to consent to, or to refuse any of the above procedures/testing is entirely mine.
10. No test(s) will be performed and reported on my sample other than those authorized by my doctor; and any unused portion of my original sample will be destroyed within 2 months of receipt of the sample by the laboratory.
11. My doctor may release my pregnancy outcome or ultrasound and amniocentesis results to Esoterix Genetic Laboratories, LLC to be used for statistical analysis of the laboratory's performance.
12. Esoterix Genetic Laboratories, LLC will disclose the test results ONLY to the doctor named below, or to his/her agent, unless otherwise authorized by the patient or required by law.

(Continued on other side)