## Informed Consent/Decline for Cystic Fibrosis Carrier Testing

I have read, or had read to me, the information in this brochure and I understand it. Before signing this form, I have had the opportunity to discuss CF testing further with my doctor, someone my doctor has designated, or to a genetics professional. I have all the information I want, and all my questions have been answered. I have decided that:

- 0 I do not want CF carrier testing.
- 0 I want CF carrier testing.

Patient Signature: _____

Date: _____

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

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Cystic Fibrosis Carrier Testing

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# Cystic Fibrosis Carrier Testing

The American College of Obstetricians and Gynecologists recommends that couples planning a pregnancy, or those already pregnant, be informed about cystic fibrosis (CF) and CF carrier testing. Testing can help determine if a couple is at risk of having a child with CF.

This brochure contains general information about CF, how it is inherited and CF carrier testing. It is our hope that you find this information helpful, but if you have any additional questions please contact your doctor or genetics professional.

## What is cystic fibrosis (CF)?

CF is a genetic disease that affects about 1 in 3,300 people in the United States. 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 33 years. CF does not affect intelligence.

## 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 abnormal CF gene, then that person is a carrier of CF. Having only one abnormal copy of the CF gene is not enough to cause the disease, so a CF carrier will not have any symptoms.

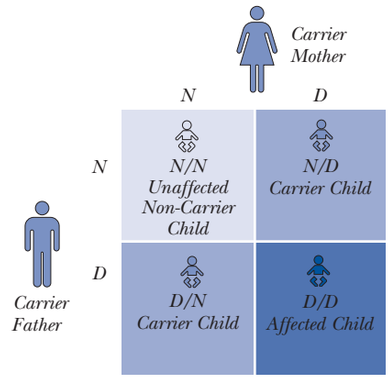
## How is CF inherited?

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

With each pregnancy there is a (See Table 1):

- 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 abnormal CF gene, and will be a carrier of CF
- 25% (1 out of 4) chance that the child will inherit two copies of an abnormal CF gene – one from each parent – and will be affected with the disease

## Table 1




*The above diagram shows that if both parents are carriers of the same disease gene (D), each child has a 25% chance of having the disease (D/D) and a 75% chance of not having the disease (N/N, N/D or D/N). The chances are the same for each pregnancy, no matter how many children a couple has, and are also the same for boys and girls.*

## Can anyone be a CF carrier?

Yes. 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 2). CF is more 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 someone in your family has CF, then your chance of being a carrier is increased. The chance is even greater if the person with CF is a close relative.

## Table 2

| Ethnicity | CF Carrier Risk in People with No Family History of CF |
|---|---|
| Caucasian | 1 in 25 |
| Ashkenazi Jewish | 1 in 26 |
| Hispanic | 1 in 46 |
| African American | 1 in 65 |
| Asian | 1 in 90 |

## What is the CF carrier test?

The CF carrier test provides specific information about whether or not individuals are CF carriers, and therefore about their risks of having a child affected with CF. The test requires a simple blood sample and results are usually ready within 5 to 10 days. If a couple has not yet conceived, one partner is usually tested first. If that person is found to be a CF carrier, the other partner should be tested. If a woman is already pregnant, both partners may be tested at the same time.

## What do the results mean?

CF carrier testing tells you if there is a greater chance that you are a carrier. A negative result significantly lowers, but does not completely eliminate, the risk of being a CF carrier. Extremely rare abnormalities (mutations) in the CF gene are not included in carrier testing.

## What additional testing will be needed if both parents are carriers?

If both partners are determined to be CF carriers, prenatal testing by chorionic villi sampling (CVS) or amniocentesis can be performed to determine whether or not the unborn baby has inherited the two abnormal CF genes. If a couple does not want prenatal diagnosis, then testing the baby for CF after birth is also an option.

## Is CF testing required?

No. The decision to accept or decline testing is entirely up to the individual or couple.

## Informed Consent/Decline for Cystic Fibrosis Carrier Testing

*You should be certain you understand the following points:*

1. The purpose of the test is to determine whether I am a carrier of one of the common CF mutations.
2. The decision to have CF carrier testing is completely mine.
3. The test does not detect all CF carriers.
4. If I am a carrier, testing my partner will help me learn more about the chance that our baby could have CF.
5. If one parent is a carrier and the other is not, it is still possible that the baby will have CF, but the chance is very small.
6. If both parents are carriers, prenatal testing can be done to find out whether or not the baby has inherited the CF gene.
7. The laboratory needs accurate information about my family history and ethnic background for the most accurate interpretation of the test results.
8. No other test will be performed and reported on my sample unless authorized by my doctor, and any unused portion of my original sample will be destroyed within two months of receipt of the sample by the laboratory.
9. The laboratory will disclose the test results ONLY to my doctor, or to his/her agent, unless otherwise authorized by me or required by law.

(Continued on other side)