

**7. How long do the test results take?**

508 FIRST (if positive).....<1 week  
CF AMPLIFIED.....3-5 weeks  
Family-specific mutation(s).....10-14 days

**8. My child had a positive test result and now the clinic wants to test us, his parents. Why?**

Family studies may be recommended when we diagnose a patient with two CF mutations. The purpose of testing each parent is to see if these mutations were inherited together on the same chromosome, which weighs against a diagnosis of typical CF, or inherited from each parent on opposite chromosomes, which supports a diagnosis of CF. Testing parents and other relatives for specific mutations is much quicker (10-14 days) and less expensive than analysis of the entire CF gene.

Another benefit of parental testing is that it enables parents to tell their own blood relatives, if they wish, which specific mutation they may also be at risk for carrying. These relatives can then choose the appropriate follow-up test if they want to know their carrier status.

**9. What if I have questions now or after my result is ready?**

We encourage you to discuss with your medical care provider which genetic test is most appropriate. With his or her agreement, Ambry's genetic counselors can answer many of your questions. We are very pleased to provide information, supporting materials, and literature, and can best serve you when working closely with your physician.

**Diagnostic  
Genetic Testing  
for Cystic Fibrosis**

**at**



**Questions and Answers  
for  
Patients and Families**

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### **1. How does Ambry Genetics' testing differ from other CF DNA tests? How many mutations are tested?**

Our test checks the entire CF gene and detects ~99% of mutations in patients of all ethnicities, making it the most comprehensive test available. There are over 1500 CF mutations, and we can detect >99% of these along with others that have not yet been discovered. Most other CF tests only look for a limited number of mutations, and their detection rates depend partly on the patient's ethnicity as shown in the chart in the next section.

### **2. What DNA test options are available to diagnose CF?**

- The Ambry Test®: CF AMPLIFIED™ analyzes the entire coding sequence of the CF gene, which is called *CFTR*. We also look for large rearrangements of the gene, such as duplications and deletions, that cannot be detected by sequence analysis alone.
- 508 FIRST™ is an option to look first for deltaF508, the most common CF mutation. If deltaF508 explains the diagnosis, the testing is completed very inexpensively in less than one week. If not, we continue with the CF AMPLIFIED test to examine the entire gene.

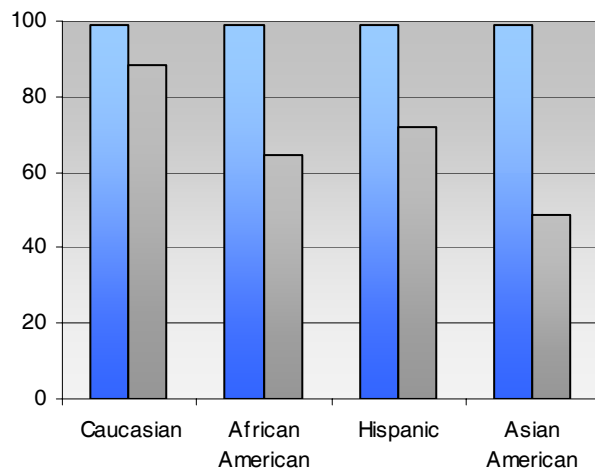
### **3. My child already had a DNA test for CF. Does she need to be tested again?**

Generally if your child had two mutations identified in her first test, there is no need to test again. If not, then it is possible CF AMPLIFIED could detect the other mutation (s). This is a question to review with your child's medical care provider.

### **4. What are the benefits of testing at Ambry Genetics?**

Ambry Genetics has unsurpassed experience in full gene sequence analysis in CF. We began offering this test in 2001 and have tested over 10,000 patients. This is especially important when interpreting unusual results. We have been first in the field twice since then with the introduction of tests for deletions/duplications and a modifier of CF lung disease.

Ambry Genetics has genetic counselors, knowledgeable client service representatives, and billing specialists to help you, your doctors, and their staff. We can help determine your insurance coverage before testing and control your costs through the Patient Care Plan (see #6). We also provide an electronic newsletter. Please see our website to sign up ([www.ambrygen.com](http://www.ambrygen.com)).



#### **Detection Rate (%) in Various Ethnicities**

- = Ambry Test: CF AMPLIFIED
- = Panel of 23 Common Mutations

### **5. What testing is available for relatives of the patient? Who should be tested?**

Once mutation(s) are identified in the patient, any blood relative can be tested specifically for the same mutation(s). This testing is less costly and has a shorter turn-around-time. Since CF mutations may be inherited from either side of the family, relatives of a patient may choose to have a full sequence analysis for extra reassurance. Genetics professionals can provide information about testing options to best address your individual situation.

Spouses and partners of people who carry a CF mutation may use Ambry Genetics' testing to more precisely determine the risk they also carry a CF mutation. With any test, there remains a small chance the patient carries an undetected mutation even if the result is negative. This "revised risk" is much lower after Ambry Genetics' testing than after common mutation panels. The high detection rate allows for a significantly greater reduction of the risk of carrying a CF mutation.

### **6. Does insurance cover the test?**

We have excellent experience with insurance reimbursement through contracts with several insurance plans, including Medicaid from many states. We can verify your coverage for testing before your appointment and let you know what the estimated charges will be. Also, we help control your costs through our Patient Care Plan which may reduce your out-of-pocket expense. We also offer discounts when payment is made at the time of service.