

## **SCREENING FOR CYSTIC FIBROSIS: INFORMATION FOR PARENTS**

### **What is Cystic Fibrosis?**

Cystic Fibrosis (CF) is a life-long, multisystem disease caused by a defective CF gene that makes it difficult for chloride to move through cells. Impaired chloride transport leads to thick, sticky mucus secretions in the lungs and digestive tract and to increased salt content in sweat. Children and adults with CF often experience malnutrition, poor growth, numerous infections, breathing difficulties, and eventually, permanent lung damage. Lung disease is the primary cause of death in most CF patients.

### **How do you get Cystic Fibrosis?**

CF is an autosomal recessive disease, meaning the fetus must inherit a defected CF gene from both parents in order to have the disease. Approximately 1 in 31 Americans is a symptomless carrier of the CF gene. Those at highest risk are people with a Northern European or Ashkenazi Jewish heritage.

### **What is the life expectancy of a child born today with CF?**

Approximately 30-40 years

### **What is the treatment for CF?**

At present, there is no cure for CF. Treatment consists of reducing symptoms and slowing progression of the disease so the patient's quality life is improved.

### **How is screening for CF performed? Can it detect all CF gene mutations?**

Screening for CF before or during pregnancy involves obtaining a sample of blood or saliva from the mother and determining if she is a carrier of the CF gene. If she is a carrier, the next step is to test the baby's father. If both mother and father are carriers of the CF gene, there is a 25% chance with each pregnancy that child will have CF. However, there are some rare mutations in the CF gene that the current screening test cannot find. Therefore, even with normal result from the screening test, you may still be a carrier.

### **If both of us are carriers of the CF gene, can we determine if our baby will have CF?**

Yes, through additional testing during pregnancy. Chorionic villus sampling (CVS), done around the 11th week of pregnancy, involves removing a small portion of the placenta. Amniocentesis, the most common method of testing for CF, is done around the 16th week of pregnancy and involves removing a small amount of fluid surrounding the baby with a thin needle inserted into the woman's uterus. The results of both of these tests are nearly 100% accurate about whether the baby will have the disease.

### **What are the risks of CVS and Amniocentesis?**

The risk of losing the pregnancy as the result of the procedure is 1:100 after CVS and 1:200 after Amniocentesis. These are risks that some women are not willing to take for a diagnostic procedure.

**If the diagnostic test shows that our baby 'will have CE, what are the options?**

CF is not a curable disease. There are treatments available to slow disease progression, but only after the baby is born. Therefore, the only two options after diagnosis are to continue the pregnancy and prepare for the addition to your family of a child with CF or to terminate the pregnancy. You should have open discussions with your family and your obstetrician before making a decision regarding screening and testing for CF.

**Where can I obtain more information about screening for CF?**

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

National Society of Genetic Counselors: [www.nsgc.org](http://www.nsgc.org)

MEDLINE plus: Cystic Fibrosis: [www.nlm.nih.gov/medlineplus/cysticfibrosis](http://www.nlm.nih.gov/medlineplus/cysticfibrosis)

American College of Obstetricians and Gynecologists: [www.acog.org](http://www.acog.org)