Caring Choice
Women's Center
Obstetrics & Gynecology

Cystic Fibrosis Screening in Pregnancy

Cystic fibrosis (CF) is a genetic disease affecting approximately 30,000 children and adults in the United States. It is most prevalent in Caucasians. A defective gene causes the body to produce abnormally thick, sticky mucus that clogs the lungs and leads to life-threatening lung infections. These thick secretions also obstruct the pancreas, preventing digestive enzymes from reaching the intestines to help digest food. Cystic fibrosis can also cause liver damage and causes infertility in most effected males.

Because it is a ***recessive*** condition, an individual must inherit two defective CF genes, one from each parent, to have CF. More than 10 million Americans are unknowing, symptomless carriers of a single defective CF gene. In the US, the carrier frequency and disease frequencies vary with ethnicity as follows:

	carrier frequency	**disease frequency**
Ashkenazi Jewish:	1:24	1: 2270
Caucasians:	1:25	1: 2500
Hispanic:	1:58	1:13500
African American:	1:61	1:15100
Asian American:	1:94	1:35100

Each time two carriers conceive there is a 25 percent chance that their child will have CF; a 50 percent chance that the child will be a carrier of the CF gene; and a 25 percent chance that the child will be a non-carrier.

Treatment of CF depends upon the stage of the disease and the organs involved. Chest physical therapy and mucous thinning drugs aid in clearing mucus from the lungs. Lung infections are treated with antibiotics. Pancreatic enzyme supplements aid in digestion. At present, the median age of survival for a person with CF is in the mid-30s. 15% of individuals with CF have a mild form with an average expected lifespan of 56. The life expectancy of a child born today with CF is over 40.

Cystic Fibrosis Screening

For women and men who desire to know if they are carriers of an abnormal cystic fibrosis gene, genetic screening is available. Cells are obtained from either mouthwashes or blood samples. The ultimate objective of a couple seeking CF screening is to be assured they are not both carriers. If both parents are carriers and wish to know if their baby has cystic fibrosis they can be offered chorionic villous biopsy after 11 weeks gestation or amniocentesis after 15 weeks gestation.

The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) recommend the following:

1. Information about cystic fibrosis screening should be made available to all couples. It is reasonable to offer cystic fibrosis carrier screening to all couples regardless of race or ethnicity as an alternative to selective screening.
2. Cystic fibrosis screening should be offered before conception or early in pregnancy when both partners are of Caucasian, European, or Ashkenazi Jewish ethnicity.
3. When both partners are cystic fibrosis carriers, genetic counseling is recommended to review prenatal testing and reproductive options. Prenatal diagnosis by chorionic villous sampling or amniocentesis, using DNA-based testing of the fetal cells, should be offered.

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