Cystic Fibrosis Carrier Screening

Cystic fibrosis (CF) is the most common potentially life threatening autosomal recessive genetic condition found in some populations. It is a progressive illness that causes thick mucus in the lungs and intestinal tract and is associated with frequent respiratory infections, obstructed pancreatic and liver ducts and impaired protein digestion. It also affects the sweat glands, and nearly always causes infertility in affected males. It does not affect intelligence. Due to the development of effective treatments, the life expectancy for people with classic CF has improved greatly, with a median survival now of 37 years. About 15% of people with CF have a mild atypical form of the disease and live an average of 56 years.

In 2001, it became the standard of care to offer all pregnant patients in the United States a carrier screening blood test for CF, regardless of family history. The test is only done once, and does not need to be repeated during subsequent pregnancies. The carrier rates for different populations are detailed in the table below. If a pregnant woman is found to be a carrier, testing is then offered to the father of the baby. If both parents are carriers, and both pass the altered genes to their child, the child will have cystic fibrosis. The chance of that happening is 25% for each pregnancy. The diagnostic tests available after a positive screening result are either CVS or amniocentesis.

CF is caused by mutations on chromosome number 7. Over 1700 different mutations have been identified. Testing identifies only the most common mutations. Therefore, a negative screening test result greatly reduces but does not eliminate the chance of being a carrier and having an affected child. If there is a family history of cystic fibrosis, it is extremely helpful to know the exact mutation if that information is available.

To put the risks in perspective, with a carrier rate of 1 in 25, the prevalence is only 1 in 2500-3300 live births. Although there are over 1700 known mutations, most occur at frequencies of less than 0.1%.

Table 1. Cystic Fibrosis Detection and Carrier Rates Before and After Testing

Racial or Ethnic Group	Detection Rate (%)	Carrier Risk Before Testing	Approximate Carrier Risk After Negative Test Result
Ashkenazi Jewish	94	1/24	1/380
Non-Hispanic white	88	1/25	1/200
Hispanic white	72	1/58	1/200
African American	64	1/61	1/170
Asian American	49	1/94	1/180