

NIH Recommends CF Screening For Pregnant Couples

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The National Institutes of Health (NIH) convened an independent panel of scientists that recommended screening for cystic fibrosis (CF) carrier status be offered to “adults with positive family history of CF, to partners of people with CF, to couples currently planning a pregnancy, and to couples seeking prenatal care.”

Cystic fibrosis is an inherited disorder which occurs in about one in 2,500 live births in North American Caucasians of European ancestry with a lower frequency in other ethnic groups.

It is an autosomal recessive genetic disorder. Autosomal means the abnormal gene is not on a sex chromosome and recessive means the effect of the gene would only be seen when a baby inherits an abnormal gene from both parents and does not inherit a normal dominant gene. If both parents are found to be CF carriers, they will have a one in four chance of having a baby with CF.

In individuals affected with CF, the glands that produce mucous, sweat, and intestinal secretions do not function properly resulting in abnormally thick secretions. These thick secretions, present in the lungs and intestines, cause problems with breathing and digestion.

Common respiratory signs of CF may include coughing, breathing problems, respiratory infections, and lung damage. Digestive signs are poor weight gain and bulky, foul-smelling stools. In addition, the skin has a salty taste. While treatment has improved, death due to CF may occur in early childhood or as a young adult.

A DNA blood test can identify carriers of the most common CF gene mutations. There are many CF gene mutations, most of them rare, which cannot be detected in routine testing. Because there are many gene mutations that cause CF, testing is most reliable if the specific mutations are known for the affected relative. Even if the mutations are not known, carrier testing may be performed. Current DNA tests will detect about 90 percent of CF carriers in North American Caucasians, 95 percent in Ashkenazi Jews, and less in other ethnic groups. Carrier tests may be completed within one to three weeks. Prenatal tests take approximately three weeks.

Individuals who are concerned about the risk of carrying the CF gene are encouraged to discuss genetic testing with their physicians and to consider having genetic counseling. During counseling, a detailed family history will be reviewed and appropriate testing determined for each individual situation. Call for information.

GeneCare Medical Genetics Center, in Chapel Hill, offers high quality genetic evaluation, counseling, and laboratory services, including prenatal diagnosis—the process of evaluating the developing fetus when there may be an increased risk for a specific abnormality. More information: (919) 942-0021, or 1-800-277-4363. Web site: www.genecare.com.