CYSTIC FIBROSIS CARRIER SCREENING

Background
CF is an inherited genetic disease characterized by pulmonary and gastrointestinal symptoms of varying severity. People with CF have a thick, sticky mucous in the lungs causing persistent coughing, wheezing, and frequent lung infections. Some people with CF have difficulty gaining weight because of an abnormally low amount of pancreatic enzymes, which inhibits the digestion, and absorption of food. People with CF also have high levels of salt in their sweat. Men with CF often have infertility due to an absence of the vas deferens, the tube that carries sperm from the testes to the urethra.

CF symptoms may be either mild or severe. CF is not curable, but there are treatments that can increase the life span of patients with CF, although half of the people born with CF die by the age of 30 years, primarily from lung disease.

CF Genetics
People normally have two of each gene because each chromosome is found in pairs. In order to have CF, both genes must have a mutation making the gene on both chromosomes defective.

A CF carrier is someone without CF who has one normal gene, and one mutated or defective gene. If an individual is a CF carrier, the offspring of that individual have a higher chance of having CF. If both husband and wife are carriers, the chance of having a child with CF is 25%.

Carrier Screening
In 1997, The National Institutes of Health Consensus Development Conference on Genetic Testing for Cystic Fibrosis recommended that genetic screening to identify the carriers of CF should be offered to:
1. Adults with a positive family history of CF.
2. Partners of individuals with CF.
3. Couples in whom one or both partners are Caucasian, and are planning a pregnancy or seeking prenatal care.

Literally hundreds of mutations have been reported amongst individuals with CF. Fortunately however, only a few mutations account for the majority of individuals with CF. Since 1997, the American College of Medical Genetics has recommended a core panel consisting of a 25-mutation panel for general population cystic fibrosis carrier screening. In October 2001, the American College of Obstetricians and Gynecologists, in conjunction with the American College of Medical Genetics, published their clinical and laboratory guidelines for preconception and prenatal carrier screening for cystic fibrosis.

Based on these clinical guidelines, couples seeking infertility care who are Caucasian (one or both partners), and specifically of European Caucasian and Ashkenazi Jewish ethnicity, should be offered CF carrier screening. Carrier screening will detect 80% of carriers from European Caucasian backgrounds, and 97% of carriers of Ashkenazi Jewish decent.

Acknowledgement
I acknowledge I have been offered cystic fibrosis carrier screening.

☐ Yes, Please test me for cystic fibrosis
☐ No, I decline testing for cystic fibrosis. Should I decide to undergo screening I will contact you on a voluntary basis to be tested.

Signature: ___________________________ Date: ____________