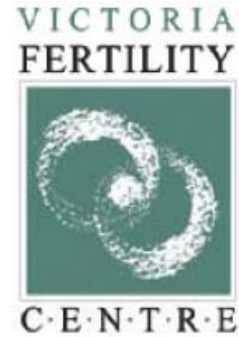


CYSTIC FIBROSIS

Cystic fibrosis is a chronic, significantly disabling illness that typically begins in the first few years of life. It causes problems with digestion and breathing due to thick mucus in the lungs and pancreas. It does not affect intelligence. Although there has been progress made in understanding and treating the condition, there is no cure. At the present time the median life expectancy is between 30 and 40 years.



Cystic fibrosis is a fairly common genetic disorder caused by mutations in the CFTR gene. In the Caucasian population it affects roughly 1 in 3000 children. We all have two copies of every gene, and for an individual to be affected with CF, he or she must inherit an abnormal copy of this gene from each parent. Individuals having only one copy of the abnormal gene and one copy of the normal gene are known as carriers. They themselves do not experience any problems.

The likelihood that you are a carrier depends on your ethnic background. Individuals of Northern European or Ashkenazi/Jewish ancestry have about a 1 in 25 chance of being a carrier. This is lower in other ethnic groups. In Hispanics the incidence is 1 in 50, in African Americans 1 in 65, and in Asians 1 in 90. If both parents are CF carriers – there is a 1 in 4 (25%) chance that the child will inherit two copies of the abnormal gene – and therefore develop cystic fibrosis.

Men who are carriers of the cystic fibrosis gene may sometimes have severely abnormal sperm counts, which can be caused by an abnormality in development of the tubules which drain the sperm-producing areas in the testes. This condition is called congenital bilateral absence of the vas deferens and usually presents with azoospermia (no sperm in the ejaculates). Although we can assist men with such problems by surgically retrieving sperm from the testicle or epididymis – it is important that such men are screened for cystic fibrosis.

The DNA test to detect the mutations (abnormality) causing cystic fibrosis is usually done on a blood specimen or a cheek swab. It can also be performed prenatally on amniotic fluid specimens. Because there are more than 900 different mutations within the cystic fibrosis gene, this test cannot detect every single possible mutation. However, it does have a 99% accuracy.

In summary therefore, the individuals who should be screened for cystic fibrosis include the following:

1. Individuals with a family history of cystic fibrosis.
2. Reproductive partners of individuals who have cystic fibrosis.
3. Men with azoospermia or severe oligospermia.
4. Optional – individuals of Northern European or Ashkenazi/Jewish ancestry.