Cystic Fibrosis

The facts

- Every 4 days an Australian child is born with Cystic Fibrosis
- Cystic Fibrosis (CF) is the most common life threatening condition affecting Australian newborns
- Most Cystic Fibrosis carriers do not know they carry the disorder – they are completely healthy adults with no known family history of the disorder
- If both parents are CF carriers then the chance of having a child with CF is 1 in 4
- A simple blood test can identify if you are a CF gene carrier
- 1 in 25 Caucasian Australians are carriers of a CF gene variant

Cystic Fibrosis (CF) is an inherited genetic disorder that permanently affects practically all organs in the body, but has major ongoing implications for the respiratory and digestive systems.

The gene involved in CF regulates the production of a protein in cells that control the movement of salt in and out of cells. CF causes the formation of thick mucus that traps bacteria, resulting in infections that damage both the lungs and the gut.

A person with CF requires medical treatment throughout their entire life. Daily physiotherapy is required for their lungs and frequent courses of antibiotics are required to treat bacterial infections.

Despite modern treatment there is currently no cure and CF sufferers have a reduced life expectancy, often needing extreme treatments like a heart and lung transplant.

Cystic Fibrosis is a recessive genetic condition, so for a child to have CF both parents must be carriers of the CF gene. When two CF carriers have a child there is a 1 in 4 chance their child will be affected by Cystic Fibrosis.

The gene associated with CF (CFTR) was only identified in 1989 and the majority of adults have never been tested, so their carrier status is largely unknown.

Male CF carriers frequently have a variant gene that is associated with infertility, and CF carrier rates are higher in infertile couples.

For a child to have CF, both parents must be carriers of the gene variant responsible for CF. When two CF carriers have a child there is:

- A 1 in 4 chance their child will be affected by CF
- A 1 in 2 chance their child will be a carrier of a CF gene variant though not affected by CF, and
- A 1 in 4 chance the child will not inherit any gene variant and therefore not be affected by CF or at risk of passing a CF gene variant on to future generations.

CF screening not only provides you with an accurate assessment of your chances of having a baby with CF, but also enables your doctor to determine the most effective fertility investigations and treatments for your situation.

A Cystic Fibrosis Screening blood test is available at Virtus Diagnostics and recommended for one partner initially. If the test is positive identifying you as a carrier of CF the specialist will recommend your partner also be tested. If both members of a couple are shown to be carriers, referral for genetic counselling can be arranged.

Accuracy of testing

The current screening tests detect most carriers but they cannot detect every possible change in these genes that may cause the disorder. There is still a small chance that you may be a carrier.

While there is a range of providers offering CF this screen is the most comprehensive CF screen identifying over 1000 gene variants.

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