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PERSONALISED MOLECULAR DIAGNOSTICS > FOR SOMATIC DISEASE > FOR YOU

CYSTIC FIBROSIS

In Australia, 1 in 2,500 babies are born with Cystic Fibrosis (CF). On average 1 in 25 people carry the CF gene - most of whom are unaware that they are carriers.

What is Cystic Fibrosis?

Cystic fibrosis (CF), also known as mucoviscidosis, is an inherited disorder that causes severe damage to the lungs and digestive system. Cystic fibrosis affects the cells that produce mucus, sweat and digestive juices. These secreted fluids are normally thin and slippery, but in people with cystic fibrosis, a defective gene causes the secretions to become thick and sticky. Instead of acting as a lubricant, the secretions plug up tubes, ducts and passageways, especially in the lungs and pancreas.

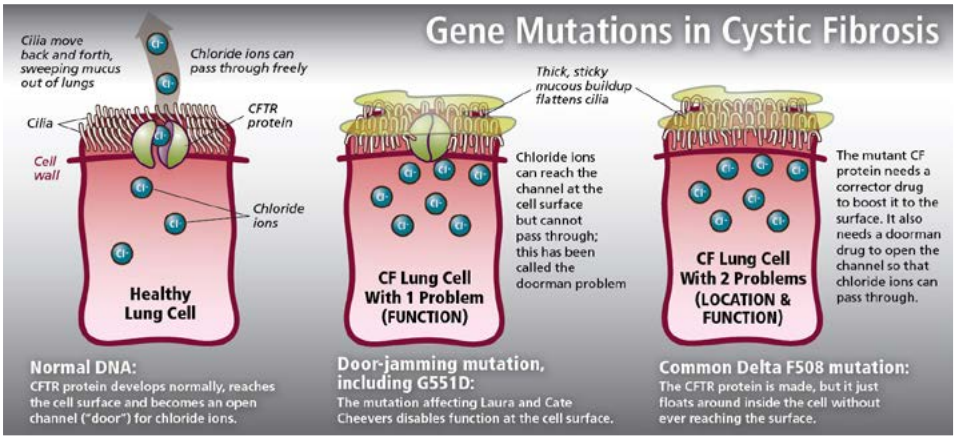
What are the effects of Cystic Fibrosis?

Cystic fibrosis is a condition which mainly affects the lungs and pancreas, but can affect other parts of the body, including the liver, nose and sinuses, reproductive organs, and sweat glands. Normally, cells in these parts of the body make mucus and other watery juices and secretions. CFTR-related disorders include congenital absence of the vas deferens (CAVD), the primary manifestation of which is male infertility.

What types of mutation causes CF?

CF is caused by a mutation in the gene cystic fibrosis transmembrane conductance regulator (CFTR).

The most common mutation, ΔF508, is a deletion (Δ signifying deletion) of three nucleotides that results in a loss of the amino acid phenylalanine (F) at the 508th position on the protein.



Who is at risk?

In Australia, one in 2,500 babies are born with CF, approximately one every four days. On average one in 25 people carry a defective CF gene - most of whom are unaware that they are carriers. Because carriers of CF are unaffected and show no symptoms they are not aware that CF may be a real risk. Any of us could be a carrier with no way of knowing. That's approximately 1 million unaware carriers.

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TESTING & DIAGNOSIS FOR CYSTIC FIBROSIS

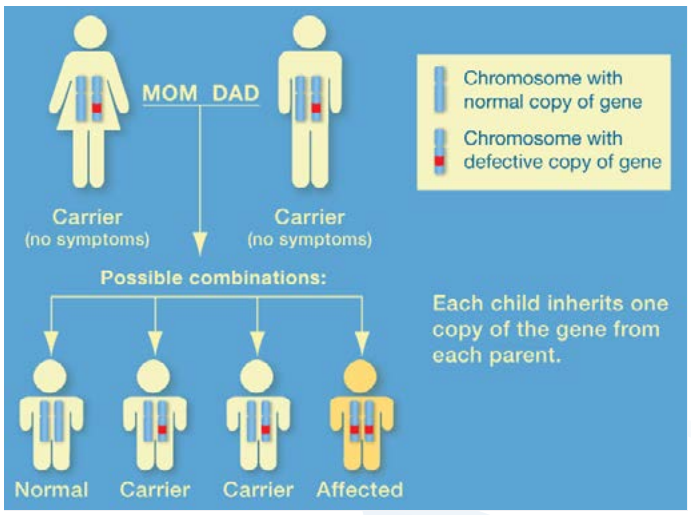
How is it inherited?

Cystic fibrosis is a recessive disorder, which means that both parents must pass on the defective gene for any of their children to get the disease. If a child inherits only one copy of the faulty gene, he or she will be a carrier.

People who have one normal CFTR gene and one faulty CFTR gene are CF carriers. CF carriers usually have no symptoms of CF and live normal lives. However, carriers can pass the faulty CFTR gene on to their children.

If you have a family history of CF or a partner who has CF (or a family history of it) and you're planning a pregnancy, you may want to find out whether you're a CF carrier. So what happens when two carriers have children...? Two carrier parents have a 25% chance of having a child with CF with each pregnancy.

Where an infant receives the CF gene from just one parent, he/she will not be born with CF, but will throughout life, be a symptomless carrier of the CFTR gene like his/her parents.



Molecular Testing

Over 1600 mutations occur in the CFTR gene; almost all are point mutations or small (1–84 bp) deletions where small pieces of DNA are missing. CAVD usually results from the combination of one severe CFTR mutation on one chromosome with either a mild CFTR mutation or the 5T allele on the other chromosome.

How is the CFTR test performed at GTLDNA?

CFTR is the only gene known to be associated with CF. DNA samples taken from a buccal swab of both parents and then DNA extracted from cells eg embryos are screened for specific defects in the CFTR gene. Our testing is performed using massive parallel sequencing (next generation sequencing) which allows for the analysis of exons, intron-exon boundaries, and UTRs that contain common mutations in the CFTR gene. This methodology with sequence analysis of all exons, intron/exon borders, promoter regions, and specific intronic regions detects more than 98% of CFTR mutations. If the molecular tests is negative and the patient has no family history of a CFTR-related disorder, the residual risk in a Caucasian person is 0.2% and 0.1% in an African American patient, with a molecular detection rate of 95%.

The residual risk for an unaffected sibling of an affected person (prior risk: 2 in 3) is 9.1% and for the unaffected sibling of a carrier (prior risk: 1 in 2) 4.8% with a molecular detection rate of 95%.

REFERENCES:
www.cysticfibrosis.org.au

Moskowitz, S.M., et al., Clinical practice and genetic counselling for cystic fibrosis and CFTR-related disorders. Genet Med, 2008. 10(12): p. 851-68.