Cystic fibrosis genetic testing patient information

It is important that you discuss the benefits and limitations of genetic testing with your physician, a medical geneticist or a genetic counsellor before you have genetic testing.

What is cystic fibrosis?

Cystic fibrosis is a progressive genetic condition that affects multiple organ systems including the lungs and digestive tract. A gene called CFTR causes cystic fibrosis. A build of thick mucus that the body is not able to expel easily results in the complications associated with cystic fibrosis. The CFTR gene is responsible for the movement of chloride (A component of salt) in and out of our cells. This movement of chloride is important within the body to keep water on the cell surface. Retaining water on the cell surface results in the mucus being more dilute and therefore easier to expel from the body. A sweat test is one of the tests that physicians use to identify people who may have cystic fibrosis due to the increased level of chloride in the sweat. A more effective way to diagnosing a child with cystic fibrosis is through genetic testing. Genetic testing can identify changes (also called variants) in the CFTR gene.

An accurate diagnosis can help determine prognosis and recurrence risks. Cystic fibrosis is a genetic condition, to understand how genetics affects our health, it is important to understand the basics of genetics.

What is DNA?

DNA is the basic unit of genetics. It is the genetic material that we inherit from our parents and pass to our children. DNA is comprised of four separate molecules (chemicals) that are called bases. Each base is assigned a different letter. These letters are A (adenine), T (thymine), G (guanine), and C (cytosine). These four bases are joined together in very long strings of DNA within our cells. These long strings of DNA are called DNA sequences (also called the DNA code or the genetic code). DNA is what genes are composed of.

What is a gene?

A gene is a specific pattern of DNA bases; this pattern is called a DNA sequence. We have approximately 20,000 genes and each gene has a specific function within our body. Each gene creates a different protein, which are the building blocks for our body. Our genes determine how our bodies develop and function from hair and eye colour to how our brain cells communicate and how we develop during pregnancy. The gene that causes cystic fibrosis is called CFTR and is responsible for the movement of chloride within our cells

Why pursue genetic testing?

It is important to have an accurate diagnosis as it can it can help with finding the correct therapies for your child and prevent further complications before they happen. Genetic testing can confirm if a child had cystic fibrosis. Identification of the genetic basis for cystic fibrosis can help determine if there is a risk for future pregnancies.

How is testing performed?

In general, DNA testing is done from a blood sample, although less frequently other samples can be obtained from other sources such as saliva. DNA is extracted from the sample and is then analysed via sequencing machines.

How long before the results are ready?

The results of genetic testing are generally ready between 3-4 weeks. Depending on what you have agreed with you healthcare provider, results may be delivered over the phone or in person

Possible results –

There are three possible results that could be reported on your whole exome sequencing test report.

Positive – Genetic changes are identified which confirms the genetic diagnosis for cystic fibrosis

Negative – No variants were identified which could explain the patient's condition. *A negative result does not always rule out a genetic basis for cystic fibrosis.*

Uncertain – A variant is classified as uncertain when there is insufficient evidence to determine if the variant is either disease causing (pathogenic) or has no effect (benign). *As more research is conducted, it is possible that uncertain variants may be reclassified in the future.*

Unexpected results – In rare situations, it is possible that a genetic diagnosis, not directly related to your child's condition is identified. These findings could have implications for the healthcare of your child.

It is important to follow up with your physician, a medical geneticist or a genetic counsellor to discuss the results of your genetic testing.