Primary ciliary dyskinesia 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 primary ciliary dyskinesia?

Primary ciliary dyskinesia (PCD) is a rare condition that affects the small "hair like" structures that line our lungs and other organs. These structures are called cilia. The cilia are responsible for moving small particles such as dust, bacteria or other foreign objects out of our lungs. They do these by moving in a wave like motion that sweeps the particles up towards the mouth to be expelled from the airway. When there is an issue with the functioning of the cilia, the small particles are not removed from the lungs. When this happens it can lead to infection and other medical complications.

PCD is when all the cilia are absent or are not functioning properly. PCD is a genetic condition, to understand how genetics impacts 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 genes responsible for PCD are important for the creation and functioning of the cilia.

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 help find the cause of a patients PCD. Identification of the genetic basis for PCD can help determine if there is a risk for future pregnancies. PCD can be caused by many genes. When genetic testing is ordered, all the known genes associated with PCD are analysed for changes within the genes that may stop them from functioning properly. We still do not know all of the genes that can cause PCD, therefore it is possible that we will not identify the genetic basis for this condition in your child.

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 PCD

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

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 implications for the healthcare of your child.

Limitations of genetic testing

The genetic basis for all cases of PCD is still not known, therefore it is possible that your child may have PCD but the genetic testing will be negative. A negative result does not rule out the possibility of a genetic basis for your child's condition.

However, research is ongoing in this area so it is possible that we will know the genetic basis for all patients with PCD in the future.

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