Neonatal Respiratory Distress 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 Neonatal Respiratory Distress?

It is estimated that up to 7% of all newborns may have some degree of respiratory distress. Neonatal respiratory distress is recognized as one or more signs of increased work of breathing, such as tachypnea, nasal flaring, chest retractions, or grunting. Several conditions can include respiratory distress as part of their diagnosis. Some of these conditions may also have involvement of other organ systems. It is important to have an accurate diagnosis to ensure the most effective treatment is identified. Depending on the presentation and clinical findings, a genetic diagnosis has been found in 20 – 70% of patients with neonatal respiratory distress. We offer genetic testing for the multiple genetic causes of neonatal respiratory distress. 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. There are multiple genes that can cause HPAH.

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 patient's respiratory distress. Identification of the genetic basis for respiratory distress can help determine if there is a risk for future pregnancies. Neonatal respiratory distress can be caused by many conditions. When genetic testing is ordered, all the known genes associated with these conditions are analysed for changes within the genes that may stop them from functioning properly.

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 basis for your child's condition

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

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.