Chromosomal microarray 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.

Why is my physician recommending genetic testing?

Your physician (or other healthcare provider) may be ordering genetic testing, as some of the symptoms of your child may be suggestive of a genetic condition. It is possible that this test may help diagnose a genetic condition. If a genetic condition is diagnosed it can lead to a change in treatment and an understanding on what to expect in the future. Early diagnosis can help with finding the correct therapies for your child and prevent further complications before they happenand/or stop any additional, unnecessary, diagnostic workup. Identification of a genetic disorder can help determine if there is a risk for future pregnancies.

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. Our genes are located within structures called chromosomes.

What is a chromosome?

Chromosomes are structures that contain all of the DNA in our cells. We have 23 pairs of chromosomes. One copy of each pair is inherited from our mother while the other copy is inherited from our father. As we have two copies of each chromosome, we therefore have two copies of every gene. Each pair of chromosome are unique. Chromosome 1 is the largest and the remaining chromosomes are named by descending size, chromosome 22 is the smallest. Chromosome pair 23 are called the sex chromosomes. In general, sex chromosomes determine the gender of the individual. A pair of XX typically result in a female while a pair of XY chromosome typically result in a male. All of the chromosomes are found in the nucleus of the cell.

What is a chromosomal microarray?

The test that your physician is recommending is called a chromosomal microarray. This test analyses the chromosomes for extra segments of DNA called “duplications” or missing segments of DNA called “deletions”. Deletions and duplications of the chromosomes are also called copy number variants.

Why pursue testing?

Your physician or genetics specialist may recommend a microarray if your child’s symptoms are suggestive of an underlying genetic condition. Identification of a chromosomal deletion or duplication can help determine if your child has a genetic condition. For an accurate diagnosis it is necessary to determine the size and location of the deletion or duplication. Chromosomal deletions and duplications are only identifiable through a chromosomal microarray.

Accurately identifying the cause of your child’s symptoms and finding a diagnosis can also help determine the best course of treatment and what to expect in the future for your child’s health.

How is testing performed?

Chromosomal microarray requires a blood sample. DNA is extracted from the sample and is then analysed via a microarray platform.

How long before the results are ready?

The results of a chromosomal microarray are generally ready in 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 –

Positive – A variant was identified which explains your child'ssymptoms.

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 patient's condition.*

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 as pathogenic or benign in the future.*

A microarray can determine if there are common ancestors in the family. In other words, this test can determine if the child's parents are related.

Sometimes a follow up parental sample is requested to aid in the interpretation of the results. Parental testing should not be considered a diagnostictest for the parent.

Incidental findings

It is possible that this test will identify a condition that is not related to your child's condition. For example this test may identify a variant that results in an increased risk for developing cancer or an increased risk for developing a cardiac condition. While this outcome is uncommon, it is a possibility.

Limitations of a chromosomal microarray

A microarray cannot detect all types of genetic changes. It is possible that your healthcare provider will suggest additional genetic testing.

Maternity and paternity analysis is notpart of chromosomal microarray testing.

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