

The Centre for Genetics Education, NSW Health Testing for missing or extra segments of DNA

As part of an investigation into the developmental or health concern you have about your child or yourself, your doctor or genetics specialist has suggested a genetic test that looks for extra or missing DNA segments.

The information below will help you understand how this test works and what information you will receive from it. Knowing this information might help you better understand the health problem that is affecting your child or yourself.

Our DNA

In all the cells of our body our DNA is arranged into chromosomes (long strings of genes). Our DNA contains coded information that is used by the cells to make our bodies work and grow. The DNA in our cells makes up our *genome*.

There are usually 46 chromosomes in each cell. There are 22 chromosomes, which come in pairs (one copy of each chromosome is inherited from each of our parents) making 44. Additionally, if the person is a male, there will be an X and a Y chromosome. If the person is a female, there will be two copies of the X chromosome.

Why would I consider having this genetic test?

Our genome needs to contain the right number of chromosomes and the right amount of DNA within the chromosomes, so that our body's systems can work as they should. Having more or less DNA than usual can cause health or developmental concerns.

If it is suspected that developmental or health concerns could be caused by extra or missing segments of the DNA in the chromosomes, or whole chromosomes, then genetic testing might be suggested.

How is this test done?

This test is done by examining a sample of your DNA that is usually obtained from a blood test. The test will examine segments of DNA along each chromosome to see if there is more or less DNA than would normally be expected. Tens of thousands of segments are examined at the same time.

As there are usually two copies of each chromosome, there should be two copies of each segment of DNA. The test will first look for a variation in the number of copies of the segments (*copy number variant*). That is, are there more or less than the expected two copies of each DNA segment?

When the test finds a copy number variant, the laboratory will check which genes it contains. Understanding the genes that are present in extra copies or are missing can provide information about:

- The cause of the developmental or health concern
- How symptoms might develop over time
- The possibility of the condition affecting future children.

Possible test results

1) No copy number variant is found.

The cause of the health or developmental concern remains unexplained. This is the most common result.

2) A copy number variant is found that explains the health or developmental concern.

Sometimes additional testing will be suggested to confirm the result by another method or testing of the parents to see if the variant is only present in the child or was inherited from a parent.

3) A copy number variant is found but its impact on health or development is unclear.

This type of result is called a *variant of unknown significance*. There are a number of steps needed to further investigate this result.

Additional testing will be necessary to confirm the result by another method. Also, testing of parents will usually be requested to see if the variant is only present in the child or was inherited from a parent. The results of this additional testing will be:

o *One of the parents also has the copy number variant*

- If the parent has the same condition as the child, the variant is likely to be the cause of the health or developmental concern.
- If the parent does not have the same condition as the child, the variant is unlikely to be the cause of the health or developmental concern. However, further testing of other family members might be needed to confirm this interpretation of the finding.

o *Neither of the parents has the copy number variant*

This means the variant must have occurred during or soon after the child's conception (a *de novo variant*). Two interpretations are possible:

- The variant is considered to be the cause of the developmental or health concerns being tested for.

OR

- We do not understand how the variant might relate to the developmental or health concern being tested for. Therefore the result remains of uncertain significance.

4) A copy number variant is found that contains a gene or genes unrelated to the developmental or health concerns, but which could potentially cause other health problems in the future.

This will be a rare result. The genes located in the extra or missing DNA segment are known to be associated with other health or developmental concerns. This information could be significant for your child's, or your, future health.

It is important to discuss what type of information you will receive from this test with your doctor.

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This document was first produced February 2010; updated May 2010