Chromosome microarray
general information

Key points:

Microarray is a powerful way to look for genetic variations that can cause health and developmental problems. These changes are called copy number variants (CNVs).

CNVs - These variations are small segments of missing or extra DNA. Some have been linked with health problems and disability, while others are normal human variation. There are some CNVs that have an unknown or uncertain affect on health.

What is a microarray?

A microarray is a test that looks for extra or missing segments of DNA in a person's genome. A genome is the complete set of our genetic material. Our genetic material, or DNA is found in our body cells.

How does it work?

Our genome consists of thousands of genes. We should have two copies of every gene in each cell. A microarray helps determine how many copies of each gene are present in our DNA. More than two, or less than two, copies of genes can cause health and/or development problems. This is called copy number variation, and this is what a microarray tests for.

Why are microarrays useful?

A microarray test is ordered if a doctor suspects that extra or missing pieces of the genome may be the cause of health or developmental problems. Microarrays are a cheap, fast way of looking at almost the whole genome at the same time.

Up until a few years ago looking at chromosomes under a microscope was the only way to analyse the whole genome at low cost. However, chromosome analysis can only detect very large changes in the genome. Microarrays can detect much more subtle copy number variants than chromosome analysis and so are more effective at finding the cause of developmental or other health problems.

Does it test for everything?

No. A microarray only tests for variants in the DNA copy number. There are many other conditions in which there is a variant in a gene without any variation in the number of copies of that gene. In other situations the copy number variant may be too small to be detected by the microarray.

References: Testing for missing or extra segments of DNA Fact Sheet, The Centre for Genetics Education, NSW Health. www.genetics.edu.au

How are the results reported?

You can expect two possible results from a microarray test:

a) No copy number variant is found. This is the most common result, and it means that the condition in question remains unexplained by this test. It does not mean that the cause of the condition is not genetic.

OR

b) A copy number variant is found.

What happens if a CNV is found?

Not all copy number variants cause problems. Some copy number changes are innocent genetic changes within a person or family. Health professionals determine how likely the copy number variant is to cause problems by checking medical reports of other individuals with similar copy number variants and by examining the genes within the copy number change. There are several possible outcomes of this analysis.

a) The variant is known to cause the condition. The microarray test has found the cause of the individual's health problems.

b) The variant is of uncertain significance. This means we don't yet fully understand how the variant impacts on health and development. Although these variants may occur more frequently in individuals with health or developmental concerns, they also may be found in individuals without these concerns.

c) The variant is of unknown significance. When this occurs, further investigations are needed in order to try and clarify the result. Variants of unknown significance have generally not been reported previously.

d) The variant found involves genetic material that is unrelated to the condition being investigated, but is potentially associated with other future health concerns. This occurs infrequently but results in families receiving unexpected information. An incidental finding may require further follow up and counselling.

For variants of unknown or uncertain significance, testing of parents is useful. If one parent has the variant and has similar features as the child, then the variant is likely to be responsible. If one parent has the variant and doesn't have similar features as the child, then the variant is unlikely to be responsible (unless there is evidence in the medical literature to suggest otherwise).

Sometimes neither parent will have the variant. This is called a *de novo*, or a new variant. A *de novo* finding increases the probability that the variant is the cause of the condition in the child, but this variant may still have uncertain or unknown significance.

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