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Informed Consent/Decline for Reveal SNP Microarray – Pediatric Testing

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References

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What is chromosome analysis?

Chromosome analysis involves looking at a person’s chromosomes under a microscope. Most people have 46 chromosomes in each cell of their body; 23 are inherited from their mother, and 23 are inherited from their father. Chromosomes are made up of DNA, which contains a code that tells each cell in the body how to function.

Chromosome analysis can detect certain missing or extra pieces of DNA, which can be a whole chromosome or just a small part of one. It can also tell if the DNA in the chromosome has been rearranged. Changes in the amount or structure of a person’s DNA may be a cause of intellectual disabilities, certain birth defects, developmental delay, or autism spectrum disorders. This brochure is designed to answer some of your questions about chromosome analysis and the Reveal SNP Microarray.

Why might my doctor order the Reveal SNP Microarray for me?

Your doctor may decide Reveal SNP Microarray could provide valuable information to help diagnose a problem that might not be detected by routine chromosome testing.5

What kind of sample is needed for Reveal SNP Microarray?

DNA is obtained from a sample of about 1 teaspoonful of your child’s blood.

What might an abnormal test result mean?

Any changes found in your child’s DNA—and the possible health effects of those changes, if known—will be discussed with you by your doctor or genetic counselor. Follow-up testing on DNA from you and the child’s other parent may be requested to find out whether a DNA change was inherited or is a new change in your child.

Sometimes a DNA change may be found and the effect it could have on a person’s health is unknown. Other times a DNA change may be found that will have no effect on a person’s health. Researchers are still trying to determine the possible health effects of all the DNA changes that can be detected with microarray testing.

What are the limitations of SNP microarray testing?

Not all intellectual disabilities and health problems present at birth have a known genetic cause.

The amount of change in your child’s DNA may be too small to be detected by the test.

Structural changes to DNA that do not result in gains or losses of genetic material (called balanced rearrangements) cannot be detected.

Some people have DNA changes in some, but not all of their cells. This is called mosaicism. Microarrays cannot always detect these when the percent of abnormal cells is low.

A normal Reveal SNP Microarray result (one in which no DNA changes are found) may be the result of one of these limitations, or it may mean there is truly no abnormality in your child’s DNA.

What if I have more questions about Reveal SNP Microarray?

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- This test may not provide conclusive results for a number of reasons. Some of the reasons are: 1) the need to test other family members; 2) individual genetic variation; and/or 3) technical reasons.
- This testing can give information about who is, or is not, the mother or father of a child. I agree to provide a family history to the best of my knowledge.
- All test results are confidential and will be released only to the ordering physician or that physician’s designated representative, or for further treatment, payment, or health care operations.
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Chromosome analysis can detect certain missing or extra pieces of DNA, whether it’s a whole chromosome or just a small part of one. It can also tell if the DNA in a chromosome has been rearranged. Changes in the amount or structure of a person’s DNA may be a sign that something is wrong with the person’s chromosomes.

Chromosome analysis can detect certain missing or extra pieces of DNA, whether it’s a whole chromosome or just a small part of one. Changes in the amount or structure of a person’s DNA may be a sign that something is wrong with the person’s chromosomes. Some changes may be small enough to be seen under a microscope. This allows for the detection of smaller changes in the amount of DNA, as well.

In addition to detecting small gains or losses of genetic material, Reveal SNP Microarray can also show if a pair of chromosomes came from just one of the patient’s parents or whether a patient’s parents are related. These events may explain certain genetic disorders.

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What is the Reveal SNP Microarray test and what are its advantages?

Reveal SNP Microarray is an advanced technique that makes it possible to examine a patient’s chromosomes in greater detail than can be seen under a microscope. This allows for the detection of smaller changes in the amount of DNA.

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What might an abnormal test result mean?

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Sometimes a DNA change may be found and the effect it could have on a person’s health is unknown. Other times a DNA change may be found that will have no effect on a person’s health. Researchers are still trying to determine the possible health effects of all the DNA changes that can be detected with microarray testing.

What are the limitations of SNP microarray testing?

Not all intellectual disabilities and health problems present at birth have a known genetic cause.

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