Reveal[SM]

SNP Microarray

Prenatal

Integrated GENETICS

LabCorp Specialty Testing Group

Reveal[SM] SNP Microarray

Reveal[SM] SNP Microarray is a test that analyzes chromosomes for changes that can explain certain kinds of birth defects. This brochure is designed to answer some of your questions about chromosome analysis and the Reveal SNP Microarray.

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, whether it's 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 and autism spectrum disorders.[1,2]

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 much greater detail than can be seen under a microscope. This allows for the detection of smaller changes in the amount of DNA.[3,4]

In addition to detecting small gains or losses of genetic material, Reveal SNP Microarray can also show whether a pair of chromosomes in the fetus came from just one parent (instead of both parents) and whether the genes originated from parents who are related. These events may help explain certain genetic disorders.[2,3,4]

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

There are many reasons a physician may order Reveal SNP Microarray, including, but not limited to:

- When there is a known family or personal history of chromosomal problems or the presence of disorders associated with chromosomal problems.
- When a fetal ultrasound exam has revealed a birth defect in your baby and the routine chromosome analysis is normal.
- To get more information when routine chromosome analysis has revealed the presence of some abnormality, but it is unclear whether or not this will cause problems for your baby.

What kind of sample is needed for Reveal SNP Microarray?

Chromosome analysis can be performed on a sample of amniotic fluid (the fluid that surrounds your developing baby) or a small piece of tissue (chorionic villi) removed from the placenta (the organ that connects your baby to you). Cells from the sample are grown under special laboratory conditions to prepare for the analysis. In routine analysis, a cytogeneticist studies the cells for extra, missing or rearranged chromosomes or pieces of DNA. When Reveal SNP Microarray is performed, a cytogeneticist studies the DNA from these same samples in greater detail to look for smaller changes (extra or missing DNA) that might be present.

What might an abnormal test result mean?

Any changes found in your fetus' 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 father of the fetus may be requested to find out whether a DNA change was inherited or is a new change in the fetus.

Sometimes a DNA change may be found and the effect it could have on a person's health is unknown.[2] 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.[5]

What are the limitations of SNP microarray testing?

- Not all birth defects have a known genetic cause.
- The amount of change in your fetus' DNA may be too small to be detected even by the microarray test.
- Structural changes to DNA that do not result in gains or losses of genetic material (called balanced rearrangements) cannot be detected.
- Some fetuses 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 fetus' DNA.

What if I have more questions about Reveal SNP Microarray?

If you have questions or want more information about Reveal SNP Microarray, ask your doctor, genetic counselor or other genetics health care provider. Information is also available on our patient education website, www.mytestingoptions.com. Additionally, you may search an online address book provided by the National Society of Genetic Counselors for a genetic counselor in your area at www.nsgc.org.

Informed Consent/Decline for Reveal[SM] SNP Microarray – Prenatal Testing

I understand the following and freely give my consent to have this genetic test performed.

I have had an opportunity to read the information provided in this brochure and/or my physician has explained the risks, benefits and limitations for the tests ordered. I am aware that genetic counseling is available to me both before and after testing.

It has been explained to me that:

- The test may provide an indication of risk for me or my child.
- The ability of genetic testing to provide risk information or a diagnosis varies with the type of test. I have been provided with information about the detection rate, and/or my health care provider has discussed it with me in detail.
- This test may not provide informative results for a number of reasons. Some of the reasons are: 1) the need to test other family members; 2) individual genetic variation; 3) maternal contamination of prenatal samples; and/or 4) technical reasons.
- This testing may give information about whether the parents are related. 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.
- Procedures to obtain blood or tissue samples may have associated risks, such as bruising from blood collection. Other procedures, such as prenatal diagnosis, may have additional risks. These risks have been explained to me.
- An additional amniotic fluid or CVS tissue sample may have to be obtained in extremely rare cases.
- All samples are coded with unique identifying information to assure quality and confidentiality.
- My/my child's sample will be securely stored in case re-testing is necessary. Samples are stored according to applicable federal, state, and professional regulations. If no regulation applies, samples will be stored no longer than 60 days from the collection date. At the end of that time, the sample will be destroyed unless otherwise specified on the reverse.

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Informed Consent/Decline for Reveal SNP Microarray – Prenatal Testing

Check one:

☐ I agree to allow my/my child's blood or tissue sample provided for genetic testing to be used for the purpose of (diagnosis/research) (development/quality control). I understand that if I agree, any information identifying me/my child will be kept confidential so that it will not be possible to determine from whom the sample was drawn.

☐ I request that the sample be destroyed immediately after testing. I understand that it will not be available if re-testing is required.

All rights to the samples will belong to the laboratory conducting the testing. There will be no compensation in the event of an invention resulting from research and development using this sample.

Your signature on this form indicates that you understand to your satisfaction the information about genetic testing and agree to have the test(s) done. In no way does this waive your legal rights or release anyone from their legal and professional responsibilities. If you have further questions concerning matters related to this consent, you may wish to seek professional genetic counseling prior to signing this form. Consultation with a medical geneticist, genetic counselor or your referring physician also may be warranted after the test has been completed.

(Signature of patient or legal guardian) **(Date)**

(Signature of health care provider) **(Date)**

References

1) McKinlay Gardner, RJM, Sutherland, GR. Elements of medical cytogenetics. In: McKinlay Gardner RD, Sutherland GR, eds. *Chromosome Abnormalities and Genetic Counseling*. 3rd ed. New York, NY: Oxford University Press; 2004:3-20.
2) Sagoo, S et al. Array CGH in patients with learning disability (mental retardation) and congenital anomalies: Updated systematic review and meta-analysis of 19 studies and 13,926 subjects. *Genet Med* 2009; 11:139-46.
3) Tepperberg, J et al. Genomic imbalances, UPD and consanguinity identified by whole genome SNP microarray analysis. Abstract presented at: Annual Clinical Genetics Meeting; March 25-29, 2009; Tampa, FL.
4) Schwartz, S et al. SNP array detection of additional clinically significant chromosomal abnormalities in patients with established cytogenetics abnormalities: An important factor in phenotypic variability. Abstract presented at: Annual Clinical Genetics Meeting; March 25-29, 2009; Tampa, FL.
5) Rosenfeld, J et al. Development of an extensive array CGH database as a free resource for large scale collaborative research. Abstract presented at: Annual Clinical Genetics Meeting; March 25-29, 2009; Tampa, FL.

About Integrated Genetics

Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years.

This brochure is provided by Integrated Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web sites:
www.mytestingoptions.com
www.integratedgenetics.com

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