

# Reveal<sup>SM</sup>

*SNP Microarray*



*Pediatric*

 **Integrated**  
GENETICS  
.....  
LabCorp Specialty Testing Group



# Reveal<sup>SM</sup>

## *SNP Microarray*

**Reveal<sup>SM</sup> SNP Microarray is a test that analyzes chromosomes for changes that can explain certain birth defects, developmental delay, or intellectual disability, and autism spectrum disorders. This brochure is designed to answer some of your questions about chromosome analysis and the Reveal SNP Microarray.**

### **What is chromosome analysis?**

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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. <sup>1,2,3</sup>



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

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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.<sup>4,5</sup>

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 closely related.<sup>4,5</sup> These events may explain certain genetic disorders.<sup>2,4,5</sup>

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

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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.<sup>5</sup>

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## **What kind of sample is needed for Reveal SNP Microarray?**

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

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## **What might an abnormal test result mean?**

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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.

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Sometimes a DNA change may be found and the effect it could have on a person's health is unknown.<sup>3</sup> 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?**

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- 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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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](http://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](http://www.nsgc.org).

## **Informed Consent/Decline for Reveal<sup>SM</sup> SNP Microarray – Pediatric 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 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.
- Procedures to obtain blood may have associated risks, such as bruising from blood collection. These risks have been explained to me.
- An additional blood sample may have to be obtained in the absence of results or if the results are inconclusive.
- 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.

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## Informed Consent/Decline for Reveal SNP Microarray – Pediatric 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 the 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.

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

**(Date)**

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**(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) Sutcliff, JS. Insights into the pathogenesis of autism. *Science* 2008; 321:208-9.
- 3) 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.
- 4) 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.
- 5) 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.
- 6) 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](http://www.mytestingoptions.com)  
[www.integratedgenetics.com](http://www.integratedgenetics.com)



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