What Is Microarray Testing?

The human body contains genetic information that is bundled into packages called chromosomes. Genetic information within each chromosome tells a person's body how to grow and develop. Changes in the amount of genetic information, such as extra or missing pieces of a chromosome, may cause medical conditions such as birth defects, delays in early development, learning difficulties, autism or other health issues.

A microarray test is an advanced technique used to look at a person's chromosomes to detect changes in the amount of genetic information. Microarray testing can detect very small chromosomal changes that cannot be seen using traditional methods, which look at the whole chromosome structure such as a karyotype.

About Us

We are a genetic testing laboratory whose mission is to provide specialized genetic tests that make a difference. Our testing services provide patients and physicians with information that can assist in making better- informed healthcare decisions.

Understanding Pediatric Microarray Analysis

Shared Understanding.

Better Care.

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Why Is Pediatric Microarray Testing Being Recommended for My Child?

The American College of Medical Genetics (ACMG)[1], American Academy of Pediatrics (AAP)[2] and the American Academy of Neurology (AAN)[3] all recommend chromosomal microarray testing for individuals with one or more of the following:

- Developmental delays or intellectual disability
- Birth defects
- Unusual physical features
- Autism spectrum disorders

Your child's doctor may be considering microarray testing because your child has one or more of these conditions.

Identifying a chromosomal abnormality by microarray testing can be helpful in several ways:

- Your child's doctor may be able to test for other health conditions associated with your child's result.
- Your child now may be eligible to receive services such as physical therapy or speech therapy at school.
- It may be important for you to know whether there is a genetic cause for your child's medical problems when considering future pregnancies or the health of other family members.

How Is Pediatric Microarray Testing Performed?

The CombiSNP™ Array for Pediatric Diagnosis can be performed on a blood sample or on cheek cells that are swabbed from the mouth with a special kit.

When possible, a blood sample is preferred, because this sample type provides greater flexibility for confirmatory and/or family studies if an abnormality is identified. Two small (4mL) tubes of blood will be drawn and sent to our lab for analysis.

If obtaining blood is problematic, we also offer the convenient option of sending a buccal (cheek) swab, which is obtained by sweeping a small foam-tipped applicator along the patient's lower gums to collect cells from the mouth for testing. If an abnormality is identified, a blood sample may be requested for confirmatory and/or family studies.

What Are the Types of Test Results?

There are typically three types of microarray analysis test results:

Normal: A normal test result means that microarray testing did not detect extra or missing large portions of genetic information. A normal test result rules out most conditions caused by changes to the chromosomes, but not all genetic disorders. The doctor may recommend additional tests based on the results of the microarray.

Abnormal: An abnormal test result means that microarray testing did detect extra or missing genetic information. If your child's test result is abnormal, his/her doctor will provide more specific information about the condition.

Variant of Uncertain Significance (VOUS): A VOUS test result means that the microarray testing identified a change that has been rarely, or perhaps never before detected. The meaning of VOUS test results may be unclear. In these situations, blood samples from the mother and the father of the child may be needed to help better understand the result.

After speaking to your physician, you can elect for your VOUS result to be stored in a de-identified public database for future reference and research. This may allow for better treatment for other children with a similar finding in the future.

1. Manning M and Hudgins L. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. Genet Med. 2010; 12(11):742-745.
2. Michelson DJ, et al. Evidence report: Genetic and metabolic testing on children with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. Neurology. 2011 Oct 25;77(17):1629-35.
3. Ellison JW, et al. American Academy of Pediatrics: Clinical utility of chromosomal microarray analysis. Pediatrics. 2012;130e:e1085-e1095.
4. Miller DT et al. Consensus Statement: Chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet. 2010; 86(%):749-764

What Are the Chances That the Test Result Will Be Abnormal?

Microarray testing identifies extra or missing chromosomal information in about 15-20% of children with developmental delays and/or birth defects and in about 7% of children with autism or an autism spectrum disorder.[4]

How Long Will It Take to Get the Test Results?

Microarray testing typically takes approximately 7 days from the time the blood sample is received by our laboratory. The results will be returned to the doctor who ordered the test, and your child's doctor will review the test results with you.

Will My Insurance Cover Microarray Testing?

Some insurance companies will pay the full cost of microarray testing, while others may pay only a portion of the cost. CombiMatrix accepts samples from all patients with commercial health insurance coverage and will bill your insurance company directly on your behalf.

We strive to minimize your financial burden and offer a flexible patient payment plan to help make microarray testing affordable for you and your family.

Do You Have Additional Questions?

If you still have questions or want to learn more, ask your doctor, genetic counselor or other healthcare provider.

Additional information is also available on our website at www.combimatrix.com.