WHAT ARE THE CHANCES THAT THE TEST RESULT WILL BE ABNORMAL?

Pediatric microarray analysis identifies extra or missing chromosomal information in about 15 to 20% of children with developmental delays and in about 7% of children with an autism spectrum disorder.[4]

WHAT CAN I DO IF THE RESULT IS ABNORMAL?

Talk to your child's healthcare provider and/or a genetic counselor to learn what your results mean, understand your options and, if necessary, make a plan.

A definitive diagnosis can help you get the right support

- Your child's result may help their healthcare provider order additional tests or identify new treatments for your child.
- A definitive diagnosis may qualify your child for assistive services such as physical therapy or speech therapy at school.
- Your child's result may also help their healthcare provider decide if you, or other family members, could benefit from additional genetic testing and counseling as certain chromosomal conditions are inherited.

HOW DO I PAY FOR PEDIATRIC DIAGNOSTIC TESTING?

Insurance (US only)
Invitae is proud to be in network for more than 250 million patients in the United States and will contact insurance companies directly to coordinate coverage and payment on your behalf.

Financial assistance (US only)
For patients who do not have adequate coverage through insurance, Invitae offers patient-pay pricing. We may also be able to offer testing at limited or no expense to those who qualify for need-based assistance.

For complete billing information, please visit www.invitae.com/billing-info.

Questions? Visit www.invitae.com/contact-us for a complete list of contact information.

ABOUT INVITAE

Invitae is a genetics company whose mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people.

Invitae testing provides answers to essential health questions—understanding disease risk, guiding a healthy pregnancy, or finding a diagnosis—at high quality, fast turnaround, and low prices.

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):742-5.
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;25;77(17):1629-35.
3. Ellison JW *et al*. American Academy of Pediatrics: Clinical utility of chromosomal microarray analysis. *Pediatrics*. 2012;130e:e1085-95.
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(5):749-64.

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INVITAE

Pediatric diagnostic testing

WHAT IS PEDIATRIC DIAGNOSTIC TESTING?

Pediatric diagnostic tests can help you find answers by determining if your child's medical challenges are due to a condition caused by a chromosomal abnormality.

What is a chromosomal abnormality?
Chromosomal conditions occur when there is a change in the number, size, or structure of your child's chromosomes. This change in the amount or arrangement of the genetic information in the cells may affect growth, development, or the ability for body systems to function.

How do chromosomal conditions develop?
Usually, chromosomal changes are spontaneous and occur when the egg or sperm cells are forming during conception. Sometimes, however, chromosomal changes are inherited from one or both parents.

IS PEDIATRIC DIAGNOSTIC TESTING RIGHT FOR MY CHILD?

Pediatric diagnostic testing can help children with one or more of the following:

- developmental delays or intellectual disability
- birth defects
- unusual physical features
- autism spectrum disorders
- other health conditions like seizures

TESTING WITH INVITAE

Invitae offers several types of tests that can be used to diagnose medical conditions caused by chromosomal abnormalities. Your child's healthcare provider may choose to use microarray analysis, karyotype analysis, or both to help guide your child's medical care.

Pediatric microarray analysis
Microarray analysis is more comprehensive than karyotyping and can detect both large and small changes in a child's chromosomes. Therefore, the American College of Medical Genetics,[1] American Academy of Pediatrics,[2] and the American Academy of Neurology[3] all recommend chromosomal microarray testing over karyotyping. However, in some cases, you and your doctor may choose a karyotype test.

Karyotype analysis
This test looks at your baby's overall chromosomal structure to see if there are any abnormalities, such as extra or missing chromosomes. Karyotype tests can detect large chromosomal changes, like those that cause Down syndrome and fragile X syndrome, but they are not sensitive enough to uncover small changes that may have caused your child's medical condition.

HOW LONG WILL IT TAKE TO GET MY RESULTS?

Microarray results are ready in 10–12 days on average from the time the sample is received by Invitae.

Karyotype results are ready in 12 days on average from the time the sample is received by Invitae.

WHAT WILL MY CHILD'S RESULTS TELL ME?

Normal
Chromosomal abnormalities **were not detected** on your child's test. A normal result rules out most conditions caused by changes to the chromosomes. In some cases, your child's healthcare provider may recommend additional testing to look for genetic disorders that are not caused by chromosomal abnormalities.

Abnormal
Chromosomal abnormalities **were detected** on your test. Your child's healthcare provider will provide more information to help you understand the implications of the detected abnormality and will help make a personalized healthcare plan for your child.

Variant of uncertain significance (VUS)
In some situations, test results may be inconclusive. This means a variant (genetic change) was detected on your test, but there is not enough information to provide a conclusive result. Your healthcare provider may request blood samples for analysis from you and your reproductive partner to help clarify the result. VUS results are relatively common and are usually harmless.

How Invitae testing works

1. Talk to your child's healthcare provider.
Your child's healthcare provider will work with you to choose the right pediatric diagnostic test.

2. Provide a sample.
Pediatric diagnosis can be performed on a blood sample or on cheek cells that are swabbed from the inside of your child's mouth with a special kit called a buccal swab.

3. Get your results.
Your child's healthcare provider will receive a report that contains important details about your child's results.

4. Make your plan.
Pediatric diagnostic testing can provide answers that may guide your child's medical care. Discuss your child's test results with their healthcare provider, one of Invitae's genetic counselors, or both.