HOW LONG WILL IT TAKE TO GET MY RESULTS?

Most prenatal microarray results are ready 5–7 days on average after samples arrive at Invitae's lab. However, it is common for this test to take place after the completion of a fetal karyotype test, which typically takes 10–12 days on average.

WHAT WILL MY RESULTS TELL ME?

Normal

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

Abnormal

Chromosomal abnormalities were detected on your test. Your healthcare provider will have additional information to help you understand implications of the detected abnormality, if any, and will help you better understand next steps.

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 DO I PAY FOR MY PRENATAL DIAGNOSTIC TEST?

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. ACOG Practice Bulletin No. 163 (replaces practice bulletin No. 77, January 2007): Screening for Fetal Aneuploidy. Practice Bulletin No. 163. American College of Obstetricians and Gynecologists. *Obstet Gynecol*. 2016; 127(5):e123-37.

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INVITAE

Prenatal diagnostic testing

WHAT IS PRENATAL DIAGNOSTIC TESTING?

A prenatal diagnostic test is performed during pregnancy to determine if your baby is affected by certain conditions caused by chromosomal changes. Healthcare providers frequently recommended diagnostic testing if a previous screening test revealed an increased risk for chromosomal conditions or if you are over the age of 35.

What is a chromosomal condition?
Chromosomal conditions occur when there is a change in the number, size, or structure of your baby'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 PRENATAL DIAGNOSTIC TESTING RIGHT FOR ME?

Invitae believes all women should be offered the option of prenatal diagnostic testing. In some cases, your healthcare provider may recommend a prenatal diagnostic test because of an abnormal finding on an ultrasound or because you are considered to have a high-risk pregnancy.

Prenatal diagnostic testing is recommended for all women who:

- want access to their genetic information
- are over 35 years of age
- have experienced multiple miscarriages
- previously had a child with a medical condition; particularly one associated with chromosomal changes
- had an abnormal maternal blood test during pregnancy **or**
- discovered an abnormal finding on an ultrasound or karyotype test

TESTING WITH INVITAE

Invitae offers several types of tests that can be used to diagnose medical conditions caused by chromosomal abnormalities. Your healthcare provider may choose to use a karyotype analysis, fluorescent in situ hybridization (FISH) analysis, prenatal microarray analysis, or a combination of the three.

Prenatal microarray analysis
Microarray analysis is more comprehensive than karyotyping and can detect both large and small changes in a baby's chromosomes. Therefore, the American College of Medical Genetics recommends offering all pregnant women undergoing prenatal diagnosis the option of microarray analysis over karyotyping, regardless of maternal age.[1]

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 also lead to disease.

FISH analysis
FISH is used to quickly count the number of chromosomes in your baby's cells. It is useful when you and your healthcare provider need test results back quickly. This test is generally followed by another chromosomal test, like a microarray analysis.

HOW ARE SAMPLES COLLECTED?

Samples for prenatal diagnosis are usually collected using one of two procedures.

Chorionic villus sampling (CVS) is a first-trimester procedure performed to collect a small sample of cells from the placenta using a thin needle inserted into your belly or cervix (an ultrasound is used to guide the needle safely).

Amniocentesis is a second-trimester procedure performed to collect amniotic fluid using a thin needle inserted into your belly (an ultrasound is used to guide the needle safely).

While all pregnancies carry risk of miscarriage, there is a small additional risk (less than 1%) associated with testing due to the invasive nature of these procedures. Your healthcare provider can help you understand the process and risks involved.

How Invitae testing works

1. Select your tests.
Work with your healthcare provider to choose the tests that are right for you.

2. Provide a sample.
Your healthcare provider will collect a sample and ship it to Invitae's laboratory for analysis.

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

4. Make your plan.
Discuss your results with your healthcare provider to understand their implications and learn what you need to do next.