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
A chromosomal abnormality was 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 PREGNATAL 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.

Patient pay
You have the option to pay $450 for prenatal diagnostic genetic testing. This option requires upfront payment before test results are released. In addition, your clinician must place the order online and provide your e-mail address so we can send you a link to pay online using a credit card.

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.

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 recommend diagnostic testing if a previous screening test revealed an increased risk for chromosomal conditions.

What is a chromosomal condition?
Chromosomal conditions occur when there is a change in the number, size, or structure of your baby’s genetic information (chromosomes). The developing baby is sensitive to such changes and depending on the type of change, it may affect baby’s 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. Some chromosomal conditions occur more frequently with increasing maternal age. Occasionally, a baby may inherit a chromosomal change from one or both a parents.

IS PRENATAL DIAGNOSTIC TESTING RIGHT FOR ME?
The American College of Obstetricians and Gynecologists (ACOG) recommends that women, regardless of age, be offered screening and/or diagnostic testing for chromosomal conditions during pregnancy. Invitae supports testing for all women who want access to their genetic information. Prenatal diagnosis may be offered to women who have an increased chance of having a baby with a chromosomal condition due to:

- an abnormal blood screening result, such as non-invasive prenatal screening (NIPS)
- one or more fetal structural abnormalities identified by ultrasound
- a history of an affected pregnancy or a previous child with a chromosomal condition

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 offer various testing options based on your personal history and situation.

Prenatal microarray analysis
Microarray analysis counts the number of chromosomes that are present and whether there are any extra or missing pieces. This test can also detect small changes (called microdeletions and microduplications) that cannot be identified by karyotyping alone. For this reason, ACOG recommends that anyone who is considering prenatal diagnosis for chromosomal conditions be offered the option of microarray analysis.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, but they are not sensitive enough to uncover small changes that may also lead to a chromosomal disorder.

Rapid FISH analysis
Fluorescence in situ hybridization (also known as FISH) is a test that offers a rapid answer to how many copies of several specific chromosomes are present in your baby, including chromosomes 13, 18, 21, X, and Y. This test is generally followed by another more comprehensive chromosomal test, like a karyotype or 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 a thin plastic tube through your cervix (an ultrasound is used to guide the needle or tube 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 CVS and amniocentesis. 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.