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Prenatal Testing During Pregnancy

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Learn the basics of the prenatal tests and screenings you'll be offered throughout your pregnancy.

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Throughout your pregnancy, you'll probably be poked, prodded and pressed by practitioners more than ever before. Fortunately, many of these prenatal screenings and tests — like blood tests, urinalysis and ultrasounds — are routine, pain-free and absolutely beneficial.

Prenatal tests offer the most accurate view possible of your and your baby's health, and can catch many pregnancy complications early on. Some prenatal tests could be life-saving, while others provide information that can tell you more about how your baby is developing, such as whether he may have a genetic condition.

But most of these tests will confirm that everything is just fine — or can be made so with simple, standard follow-up care. Discuss early on with your doctor what tests are right for you so you can schedule them during the proper times in your pregnancy.

Types of prenatal tests and screenings

Some of the tests that you'll take during pregnancy will help your doctor monitor your health, while others will give you more information about your baby's health.

Here are the different types of prenatal tests that can give you more information about your child, including their risk of having a genetic or chromosomal condition:

- **Screenings.** Prenatal screenings, such as blood tests, can help you identify the likelihood of a baby having a genetic or chromosomal condition, but they cannot actually diagnose the disorder. If the results of these tests indicate a high risk for an abnormality, your doctor may suggest a diagnostic test to confirm or rule out the finding.
- **Diagnostic tests.** If the results of a prenatal screening indicate an increased risk of a disorder, you'll likely get a diagnostic test, like an amniocentesis or chorionic villus sampling (CVS), next. These tests will examine the chromosomes of your baby (either through a sample of amniotic fluid or cells from the developing placenta), rather than your own. These tests can diagnose some conditions with nearly 100 percent accuracy.

Prenatal screening and testing

Some prenatal screenings and tests are par for the pregnancy course, and offered to all pregnant women.

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- **Urine tests.** You started off your pregnancy by peeing on an at-home pregnancy test — so it's only fitting that you'll continue peeing in a cup at pretty much every prenatal visit. Why? Urinalysis helps your doctor or midwife check you for two potentially dangerous pregnancy complications: high blood pressure (preeclampsia) and

gestational diabetes. Both are treatable, but can be dangerous if they're not identified. The good news: Providing a urine sample is risk-free, so drink up before each appointment!

- **Initial blood work.** At your first prenatal visit, you'll roll up your sleeve for a blood sample, which will be tested for conditions that could affect your pregnancy and delivery. Specifically, your provider will check for anemia, along with your blood type, hCG levels (a hormone that's produced by the fetus), immunity to certain diseases and Rh factor, which diagnoses Rh disease, another condition that can be easily managed as long as your provider knows about it in advance. You may also receive a screening for cystic fibrosis, Tay-Sachs, sickle cell anemia, spinal muscular atrophy, thalassemia and other genetic conditions, if you weren't screened before conception.
- **Pap smear.** Your midwife or doctor might do a Pap smear (or Pap test) at your first prenatal appointment to screen for abnormal cervical cells. You'll also likely be screened for sexually transmitted infections (STIs) including gonorrhea, syphilis, hepatitis B, HIV and chlamydia. If you test positive for a bacterial infection, your doctor will prescribe pregnancy-safe antibiotics for you and possibly your partner — because if you give birth with an untreated infection, your baby is at risk of dangerous infections too, including pneumonia.
- **Genetic carrier screening**. A carrier screening can reveal if you have a gene for (or, are a "carrier" of) genetic conditions such as cystic fibrosis or spinal muscular atrophy. Your doctor will likely take blood or a swab of saliva from the inside of your cheek and send the sample to a lab. The results will either be "positive" (meaning, you are a carrier for the condition) or "negative" (meaning, you aren't a carrier for the condition). While the test can be done before you get pregnant, some women choose to have the test done during pregnancy.

First trimester genetic screening and diagnostic testing

Starting around week 10, pregnant women can receive first trimester screening and tests for certain chromosomal conditions. If the screening results indicate that your baby may have an abnormality, you can receive follow-up diagnostic testing to confirm or eliminate the diagnosis.

By opting for early screening and testing, you'll have more time to make medical decisions during your pregnancy and after delivery.

- **Noninvasive prenatal testing (NIPT).** NIPT (i.e. a noninvasive prenatal screening, or NIPS) is a blood test that analyzes DNA fragments that are circulating in a woman's blood (also called cell-free DNA, or cfNDA). The test, which can be done around week 10 of pregnancy, can screen for certain chromosomal abnormalities, including Down Syndrome, trisomy 18 and trisomy 23.
- **Nuchal translucency screening (NT).** The nuchal translucency (NT) screening is an ultrasound that's performed around weeks 10 to 13. This routine test measures the nuchal fold, an area of fluid behind the baby's developing neck. Babies with increased fluid and swelling in this spot have a higher risk of chromosome abnormalities, such as Down syndrome. The hormonal levels drawn at the time of the NT can also indicate an elevated risk of preeclampsia.
- **Chorionic villus sampling (CVS).** If an NIPT or NT shows that your baby has a higher than usual risk for certain congenital conditions, your doctor might recommend a diagnostic test called a chorionic villus sampling (CVS). This test will take a small sample of the placenta, which contains baby's genes, and can confirm or rule out that your child has the chromosomal condition. CVS is typically done between week 10 and week 13 of pregnancy.

Second trimester genetic screening and diagnostic testing

The results from your first-trimester screening can be combined with others that are done during the second-trimester — including the quad screen and an amniocentesis — to give you a more accurate understanding of your child's health.

During the second trimester, pregnant women also receive a glucose screening and an anatomy scan.

- **Quad screen.** The quad (or quadruple) screen is a blood test that measures four substances in a mother's bloodstream: alpha-fetoprotein (AFP), a protein made by the baby; human chorionic gonadotropin (hCG), a hormone made by the placenta; unconjugated estriol (uE3), a form of estrogen made by the placenta and baby; and inhibin A, a hormone made by the placenta. The results, taken together, can determine the likelihood that your child has a neural tube defect or a chromosomal condition such Down syndrome or trisomy 18. The test is typically done between week 14 and week 22 of pregnancy.
- **Amniocentesis.** An amniocentesis, or "amnio," for short, is a diagnostic test that can detect hundreds of genetic and chromosomal conditions, including Tay-Sachs disease and Down syndrome, as well as neural tube defects. During the procedure, which is normally done between week 16 and 20, a long, thin needle will be inserted into your abdomen and uterus, and a few tablespoons or amniotic fluid will be withdrawn. The fluid will be sent to a lab, and the results made available within one day to several weeks.
- **Anatomy scan.** Given during the second trimester at around 20 weeks, the anatomy scan — also known as a level 2 ultrasound — makes sure baby is growing and developing as he should. During the ultrasound, the technician will move a wand over your abdomen to generate a 2D (or even 3D or 4D) image of your growing baby. She'll also take baby's measurements, examine his heart and organs, and check your placenta and amniotic fluid levels. And, unless you want to be surprised, you'll also learn the sex of your baby during this scan. Some higher risk patients may be referred for an early anatomy sonogram at around 16 or 17 weeks. For high-risk patients, if there is an abnormality found, then they can make a decision to do an amniocentesis before 20 weeks.
- **Glucose screening.** A glucose screening is recommended for virtually all moms-to-be between weeks 24 and 28 of pregnancy. The test can detect gestational diabetes, a type of diabetes that occurs during pregnancy and affects an estimated 6 to 9 percent of all expecting women. For the test, you'll drink a special sugary beverage that tastes like flat soda, then give a blood sample. If the test reveals that your blood sugar levels are higher than expected, you'll take a second glucose tolerance test that involves fasting for at least 8 hours before drinking another sugary beverage and having your blood tested several times over a 3-hour period to confirm or rule out the diagnosis.

Third trimester screening and testing

Starting at 36 weeks, you'll have weekly checkups with your doctor, which will continue until you go into labor. Unless your doctor has told you otherwise, you may only need to have one more screening: the group B strep test. Some women, however, may also need to have other tests, including routine biophysical profiles or nonstress tests.

- **Nonstress test.** If you have a high-risk pregnancy — for example, you have gestational diabetes or your baby is measuring small for his gestational age — or you have gone beyond your due date, your doctor may want to monitor his heart rate with a nonstress test (NST). During an NST, a fetal monitor will be placed around your belly for about 20 to 40 minutes. If the results are "reactive," your baby's heart rate went up occasionally, which

is normal. If the results are "non-reactive," your baby's heart rate didn't go up enough times, and your doctor may recommend follow-up testing with a biophysical profile.

- **Biophysical profile.** If the results of one test, such as the nonstress test, are unclear, doctors sometimes order a biophysical profile (BPP) to determine if labor should be induced. The test uses an ultrasound to measure your amniotic fluid levels and evaluate your baby's heart rate, movements, tone (i.e. the ability to flex a limb) and breathing (or, if he takes "practice breaths" of amniotic fluid). If the results are normal, your baby is likely fine, but you may need to have another test later on. If not, you may need to deliver early.
- **Group B strep test.** About 25 percent of women have group B strep, a type of bacteria that's found in the vagina and rectum. Group B strep is harmless to you, but it could cause an infection in your baby when he's exposed to the bacteria during childbirth. Your practitioner will offer this risk-free test toward the end of your pregnancy, usually around week 36. It involves swabbing your vagina and rectum during a pelvic exam. If you do have group B strep, you can take antibiotics to protect your baby during labor.

Questions to ask about prenatal testing

Thanks to major medical advances over the past few decades, it's now possible for parents to learn their baby's risk for a number of medical conditions as early as a few months into the pregnancy.

If you're thinking about being screened for chromosomal disorders, here are a few questions to consider:

1. Which prenatal genetic screening tests do you recommend for me and why?

Your doctor will likely perform a preliminary chromosome screening for some disorders as part of the first trimester blood work. She may also offer more advanced screening, like NIPT, for patients whose babies have an increased risk for chromosome disorders. Those include moms who are 35 or older, moms who previously had a child with a genetic disorder, or those with a family history of certain conditions. You should ask about all your options.

2. Will my insurance cover this test?

Insurance companies are covering more essential prenatal care than ever before. If you have certain risk factors for chromosome abnormalities and your doctor orders a test, your insurance may cover part or even all of the cost. If you're unsure of whether your insurance company will cover a prenatal screening or diagnostic test, call your insurer first.

3. Are there any risks?

Physically speaking, screenings that only require a blood or saliva sample are practically risk-free, but some diagnostic tests, such as amnios, carry a small risk of miscarriage. More commonly, undergoing genetic and chromosomal tests can cause anxiety and emotional challenges, regardless of the results.

It may help to speak with genetic counselor before having a prenatal test; he or she can help you weigh the benefits of the test with the downsides.

4. What conditions are — and aren't — screened by this test?

Some prenatal screenings, like quad screenings and first trimester blood work, assess a fetus' risk for chromosomal conditions. More advanced technologies, like NIPT, can determine a fetus' risk of up to hundreds of other disorders,

including Turner syndrome (a female baby who's missing an X chromosome) and triploidy (a baby who has an extra set of chromosomes in each cell).

But not all screenings and tests look at the same chromosomes, which means that they aren't all testing for the same conditions. And chromosomal screenings can't assess your baby's risk for genetic disorders like sickle cell anemia or neural tube defects like spina bifida; congenital heart defects (which can be identified with a nuchal translucency screening); or developmental disorders.

In the end, you may opt for a combination of screenings, which can take place during different phases of pregnancy.

5. How accurate are the results?

Not all screenings and tests have the same level of accuracy, so it's important to discuss how precise each recommended screening is at assessing your baby's risk of a chromosomal disorder. NIPT, for example, can tell you with over 99 percent accuracy whether your child is at risk for having Down syndrome, while quad screenings are only about 80 percent accurate.

6. What are my options if my test results indicate a high risk for a genetic or chromosome disorders?

Your doctor likely has a standard protocol if a screening shows a mom-to-be is at high risk for a chromosomal disorder, so he or she will know how to help you understand the next steps before you even get started.

When a prenatal screening indicates that your child is at risk for a genetic or chromosome disorder, your doctor may suggest a diagnostic test, such as an amnio or CVS, to diagnose the condition. If a diagnostic test yields a positive result, your doctor might refer you to a genetic counselor, who can talk you through all your options in more detail.

Your practitioner may recommend a specialized birth facility for labor and delivery that can better address your specific needs or recommend medical interventions which, if performed immediately after birth, can improve your baby's quality of life.

Preparing yourself for prenatal tests

Bottom line? While testing can be stressful, information can be powerful — especially when it comes to you or your baby's health. The results of all these tests will allow you to make better health care decisions, and in many cases, get treatment to solve or manage unexpected conditions.

You can ease the stress you might feel by talking openly with your doctor or midwife. Ask what tests and screenings they plan to conduct and when. Make sure you understand which ones are routine and which ones are optional (or "opt-in"). Although most are covered by insurance, some aren't if you're not considered high-risk, so get that information up front.

And don't be afraid to ask plenty of questions at every prenatal visit. Your practitioner has done these tests a thousand times and may accidentally forget to go over all the details. Speak up and ask what a screening is for if you're confused, how it will work, any risks associated with it, and when you can expect results. Your relationship with your doctor or midwife should be a partnership, so take an active part in it.

From the What to Expect editorial team and Heidi Murkoff, author of What to Expect When You're Expecting. Health information on this site is based on peer-reviewed medical journals and highly respected health organizations and institutions including ACOG (American College of Obstetricians and Gynecologists), CDC (Centers for Disease Control and Prevention) and AAP (American Academy of Pediatrics), as well as the What to Expect books by Heidi Murkoff.

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