YOUR VALUES
YOUR PREFERENCES
YOUR CHOICE

Allina Health

Considering Your Options For Prenatal Genetic Testing

Screening Tests (First Trimester Screening, Second Trimester Screening, Sequential or Integrated Screening, Cell Free DNA Screening) and Diagnostic Tests (Chorionic Villus Sampling, Amniocentesis)

Understanding Prenatal Genetic Testing

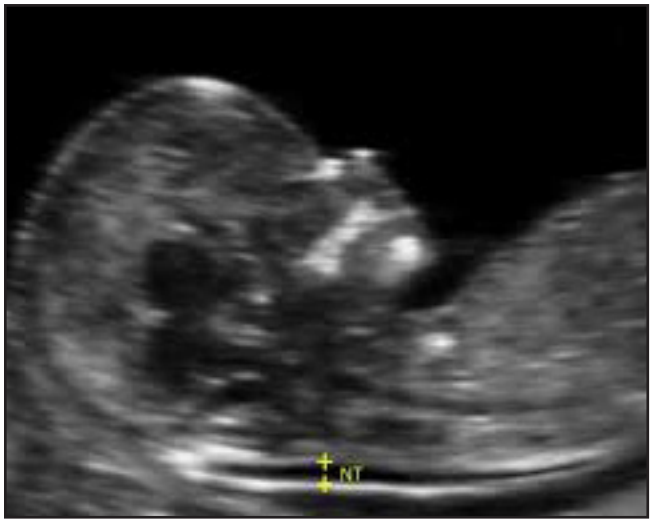
Prenatal Tests

There are many prenatal genetic tests that may be offered to you during your pregnancy.

These tests can give you information about whether there may be a higher than expected chance for certain developmental problems. These tests can give you information about whether there may be a higher than expected chance for chromosome aneuploidy, a cause of developmental problems for a baby.

A normal chromosome count is 46. Chromosome aneuploidy occurs when there is an extra (47) or a missing (45) chromosome for the pregnancy.

Any woman has the chance to have a baby with chromosome aneuploidy, but this risk increases as the woman ages.



Genetic testing can help identify pregnancies that have a higher risk of having a birth defect or genetic condition. No test can identify all types of genetic conditions, birth defects and developmental conditions.

Photo courtesy of "Increased Fetal Nuchal Translucency Thickness and Normal Karyotype: Prenatal and Postnatal Outcome," by Ksenija Gersak, Darija M. Strah and Maja Pohar-Perme. Submitted: April 26th 2012. Reviewed: September 6th 2012. Published: March 6th 2013. DOI: 10.5772/53112.

Risk Categories

In general, women fall into one of two categories of risk:

- **low (or average) risk**: A woman is of low risk if her age at the time of delivery is less than 35 years old.
- **increased (or high) risk**: A woman is of increased risk if her age at the time of delivery is 35 years old or higher, if she had a pregnancy with a chromosome aneuploidy, or if an ultrasound during pregnancy shows a birth defect (such as a heart defect or spina bifida).

Types of Tests

There are two types of tests available to all pregnant women:

- **screening tests:** These tests can help identify pregnancies that have a higher risk of having a birth defect or genetic condition. If a test result is abnormal, it means you are at a higher risk. It does not mean your baby has a genetic condition. Most women who have an abnormal screening test result have healthy babies.
- **diagnostic tests:** These tests can help identify pregnancies that have a genetic condition. Most diagnostic tests are more than 99 percent accurate.

There are no tests that can identify all types of genetic conditions, birth defects and developmental conditions. Tests cannot guarantee the delivery of a healthy baby.

Your Options

How to Use This Decision Aid

The following pages detail common screens available to you during your pregnancy. The testing options you and your health care provider talk about may vary based upon how far along you are in your pregnancy, how much information you want and if the testing is available.

If you are at low risk, the most common screens are either a first trimester, sequential or quadruple screen. If you are at increased risk, the most common screen is cell free DNA screening.

Diagnostic testing is available to you no matter which risk category you are in.

You have access to both screening and diagnostic tests. To learn more, see pages 3 to 5.

Abnormal screen results

Each screen has a different detection rate, or ability to identify, a baby with a genetic condition. If any screen comes back abnormal or "positive" it is important to remember this does not mean your baby has the condition, only that there is an increased risk, or a higher chance, that your baby may have the condition. There are both false positive and false negative results. You will be offered other tests if a screen is abnormal.

Screening Tests

There are many types of screening test options available. The most common birth defects addressed by prenatal screens are:

- Down syndrome (trisomy 21), a condition that causes mild to moderate delays in mental development
- trisomy 18, a condition that causes severe developmental disabilities and birth defects
- trisomy 13, a condition that causes severe developmental disabilities and birth defects
- spina bifida, an opening into the baby's spine.

It is important to remember each test is different and has a different detection rate for finding genetic conditions prenatally.

If a screening test comes back positive, it does not mean your baby has the condition. It means your baby has a higher chance to have the condition.

Your Options (continued)

Some people will have an increased risk for a genetic condition and their babies do not have one. Some people will have a reassuring low risk screen and their babies have a genetic condition.

First trimester screening

First trimester screening is done between 10 and 14 weeks of pregnancy. This screening test includes a blood draw and an ultrasound. The blood test measures proteins made by your placenta. The ultrasound is used to measure the thickness of the fluid at the back of your baby's neck. These tests will tell you if your baby is at a higher risk for having Down syndrome or trisomy 18. These tests do not screen for spina bifida.

Second trimester screening

Second trimester screening is done between 15 and 20 weeks of pregnancy. This blood test measures proteins made by your placenta. It can also detect pregnancies at an increased risk for spina bifida. If your provider has a concern about spina bifida, you may have a detailed ultrasound and be offered a diagnostic test called amniocentesis.

Sequential or integrated screening

A sequential or integrated screening test combines the results from the first and second trimester screens. Of all the screens for people at average risk, these have the highest accuracy for Down syndrome or trisomy 18 risk information.

Cell free DNA screening

Cell free DNA screening is a blood test that measures pieces of DNA in your blood to determine the risk for several genetic conditions such as Down syndrome, trisomy 18 and trisomy 13. This test does not screen for spina bifida.

You may see or hear ads that use terms like "99 percent accurate" but it's important to know that this test is a screen. There are both false positive and false negative results. Not all insurance companies cover this screen so ask your insurance provider if this screen is covered by your plan.

Your Options (continued)

Diagnostic Tests

Diagnostic tests can determine if there is a specific genetic condition present for your baby. These tests are mostly safe, but there is a small risk of a complication (side effect), such as miscarriage. Fewer than 1 in 500 women will have a complication.

Chorionic villus sampling (CVS)

CVS is the earliest diagnostic test that is available. It is usually done between 10 and 14 weeks of pregnancy. It is done with ultrasound either by:

- **transcervical**: an ultrasound guides a catheter (thin plastic tube) into your vagina, through the cervix, into your uterus and to the placenta
- **transabdominal**: an ultrasound guides a thin needle into your lower abdomen and to the placenta.

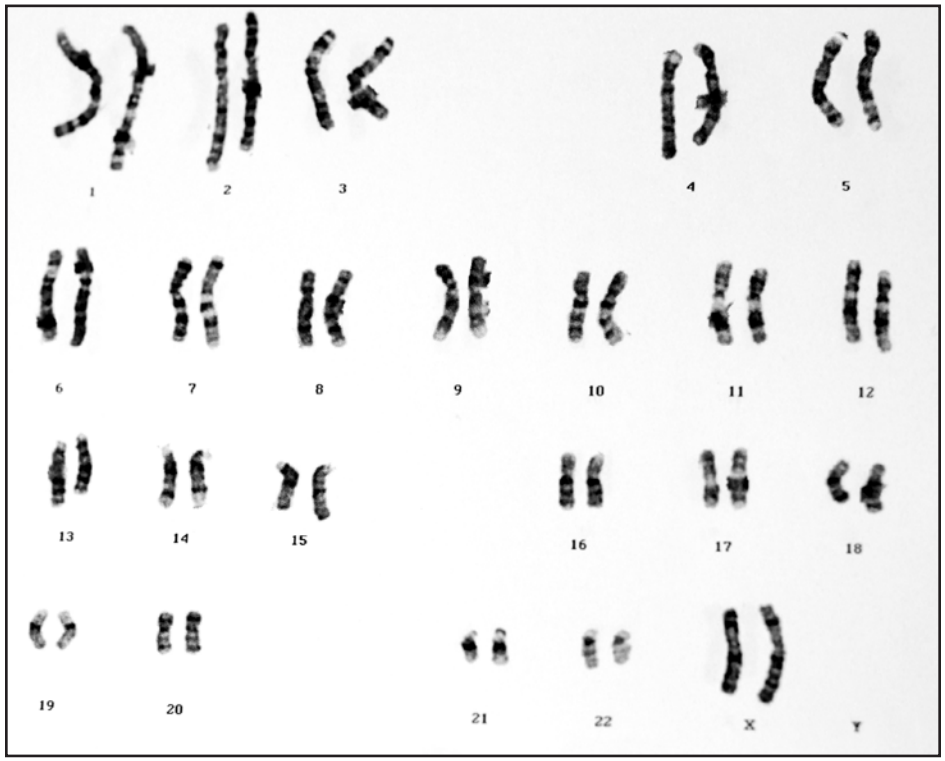
A small sample of chorionic villi (piece of the placenta) will be removed in either CVS procedure and sent to the lab for testing.

CVS is more than 98 percent accurate in finding most chromosome or genetic concerns.

Amniocentesis

The most common type of diagnostic test is an amniocentesis. This test is done after 15 weeks of pregnancy. It uses an ultrasound to guide a thin needle into your lower abdomen and into the amniotic sac. A small sample of amniotic fluid is removed and sent to a lab for testing.

Amniocentesis is more than 99 percent accurate in finding most chromosome or genetic concerns.



The normal chromosome count is 46 (23 from the woman and 23 from the man).

Comparing Your Options

	First Trimester Screening	Second Trimester Screening	Sequential or Integrated Screening
Advantages	■ This test is done early in the pregnancy. ■ This is a single test. ■ Blood work may give more information about the health of the placenta during pregnancy.	■ No ultrasound is needed. ■ It screens for open neural tube defects, like spina bifida. ■ Blood work may give more information about the health of the placenta during pregnancy.	■ This has the highest detection rate*, or accuracy, of the serum screening options. ■ It screens for open neural tube defects, like spina bifida. ■ Blood work may give more information about the health of the placenta during pregnancy.
— *What is most important to you?*			
Disadvantages	■ The detection rate*, or accuracy, is slightly lower than the sequential or integrated screen. ■ An ultrasound is required. ■ If you use insulin, a risk cannot be provided for trisomy 18.	■ The detection rate*, or accuracy, is the lowest of all serum screening options. ■ If you use insulin, a risk cannot be provided for trisomy 18.	■ Two samples are needed so you will need 2 appointments. ■ An ultrasound is required. ■ If you use insulin, a risk cannot be provided for trisomy 18.
— *What is most important to you?*			

***Detection rate is the ability to correctly categorize an individual at increased risk. It does not guarantee that either the test is accurate or that a condition is present.**

Comparing Your Options (continued)

	Cell Free DNA Screening	CVS	Amniocentesis
Advantages	■ This test has the highest detection rate*, or accuracy, for Down syndrome if you are older than age 35 at delivery or if you have given birth to a child with Down syndrome. ■ It can be done any time after you are 10 weeks pregnant.	■ This test gives answers (yes or no) to the question of certain genetic conditions. ■ It gives you time to prepare and manage your pregnancy decisions.	■ This test gives answers (yes or no) to the question of certain genetic conditions. ■ It gives you time to prepare and manage your pregnancy decisions.
— *What is most important to you?*			
Disadvantages	■ Your insurance provider may not cover this test. ■ There is an increased chance for a false result if you have average risk (younger than age 35 at delivery). ■ There is an increased chance for a false result for conditions other than Down syndrome. ■ There is a higher chance for a "no result" if you are obese. ■ Blood work cannot give more information about the health of your placenta.	■ There is a very small risk for complication such as miscarriage, preterm delivery or infection. ■ There is a small chance (less than 2%) for an unclear result which would need a follow-up amniocentesis test.	■ There is a very small risk for complication such as miscarriage, preterm delivery or infection.
— *What is most important to you?*			

My Preferences

Questions	My Thoughts
As you think about your options of screening or diagnostic tests, what are your fears or concerns?	
If your baby has a genetic condition, would you prefer to find out at delivery or before delivery?	
As you think about your options, what are your hopes or goals?	
Which of these options, at this time, do you feel fits best with your pregnancy plan?	
Is there anything that may get in the way of you having prenatal genetic test?	

My Decision at This Time

❑ first trimester screen ❑ second trimester screen ❑ sequential or integrated screen

❑ cell free DNA screen ❑ chorionic villi sampling ❑ amniocentesis ❑ undecided

Next Steps

Questions for Your Health Care Provider

1. Is there a risk of my baby being born with a birth defect or genetic condition?
2. Do I need to have screening or diagnostic testing?
3. If I have a high risk, or abnormal screen, what are the next steps? Where can I have the screening or testing done?
4. Would my pregnancy be managed differently if I were high risk, or if my baby had a birth defect or genetic condition?
5. What is the best test for me?
6. What else should I consider before getting screened or tested?
7. Is my pregnancy still at risk if I do not have any family history of a genetic condition or birth defect?
8. Other questions: _____

Next Steps (continued)

Insurance Coverage

Health care benefits change and differ from plan to plan and provider to provider. It is important for you to understand your health care benefits before your delivery.

Call your insurance provider and find out exactly what is and is not covered under your plan, and how much you have to pay yourself. The phone number should be on your membership card.

Check with your insurance provider if you have any questions about your coverage.

Notes

Resources

- Allina Health.org
  allinahealth.org/pregnancy
- The American College of Obstetricians and Gynecologists
  acog.org

Allina Health Account

Sign up for an Allina Health account online to get:

- better communication with your clinic, hospital and provider
- faster answers
- online access to you and your loved one's health information anytime.

With your account, you can:

- read visit notes and follow-up instructions
- view and pay bills
- refill a prescription
- view immunizations and medicines
- set and track health goals
- receive lab results sooner
- do an online visit for common conditions
- manage another person's care
- write a health care directive
- email your care team*
- schedule appointments*.

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ob-ah-78891 (5/19)