Thinking about having an amniocentesis (amnio) or chorionic villus sampling (CVS) to see if your baby has Down syndrome or a related disorder?

This brochure explains a new DNA blood test that you may also want to think about before you decide about CVS or amnio.

Your doctor's office may have told you that your baby is at "high-risk" for having Down syndrome (trisomy 21), trisomy 13, or trisomy 18. These are serious conditions caused by an extra chromosome. You might be at high-risk because of your age, a positive serum screening test, an unusual ultrasound, or perhaps family history. The only way to tell for sure whether there is an extra chromosome is to have a CVS or amnio. These are known as "invasive" tests because a doctor has to remove cells from inside you to study your baby's chromosomes.

There is a new, DNA-based blood test that was studied by doctors and researchers at Women & Infants Hospital in 2011. The test is now available to pregnant women, and it may be a good first step if you are considered at high-risk for carrying a baby with a chromosome disorder.

Locations

For the convenience of our patients and referring health care providers, Women & Infants offers prenatal diagnosis services at several locations, including:

- **Prenatal Diagnosis Center**
  Bay Tower Medical Center
  101 Plain Street
  6th Floor
  Providence, RI 02905
- **North Dartmouth**
  Prenatal Diagnosis Services
  WIH Faculty Physicians, Inc.
  300 A Faunce Corner Road
  North Dartmouth, MA 02747
- **South County Hospital**
  95 Kenyon Avenue
  Wakefield, RI 02879
- **WIH Faculty Physicians, Inc.**
  119 Sachem Street
  Norwich, CT 06360

For directions to any of these sites, check our website at womenandinfants.org.

Stephen Carr, MD
Director, Prenatal Diagnosis Center

Barbara M. O'Brien, MD
Director, Perinatal Genetics
Co-Director, Prenatal Diagnosis Center

For more information

If you have questions or suggestions, please contact the Prenatal Diagnosis Center by calling (401) 453-7510. Our fax number is (401) 453-7517.

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DNA testing for Down syndrome and related disorders

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What is this new, DNA-based blood test?

This new DNA blood test does not take the place of CVS or amnio, but it can reduce the number of women who will need to decide about having those tests. Using blood taken from your arm, the test can correctly identify 99 out of 100 pregnancies with Down syndrome. It can also find almost all pregnancies with trisomy 13 and trisomy 18.

What will this new blood test tell me?

The test looks to see if your blood contains extra material from certain chromosomes. It is normal to find pieces of your baby's genetic material (DNA) as well as pieces of your own genetic material in your blood. Too many extra pieces from chromosome 21 means that your baby probably has Down syndrome (trisomy 21). Too many extra pieces from chromosome 18 or 13 means that your baby probably has trisomy 18 or trisomy 13.

What if my test is positive (abnormal)?

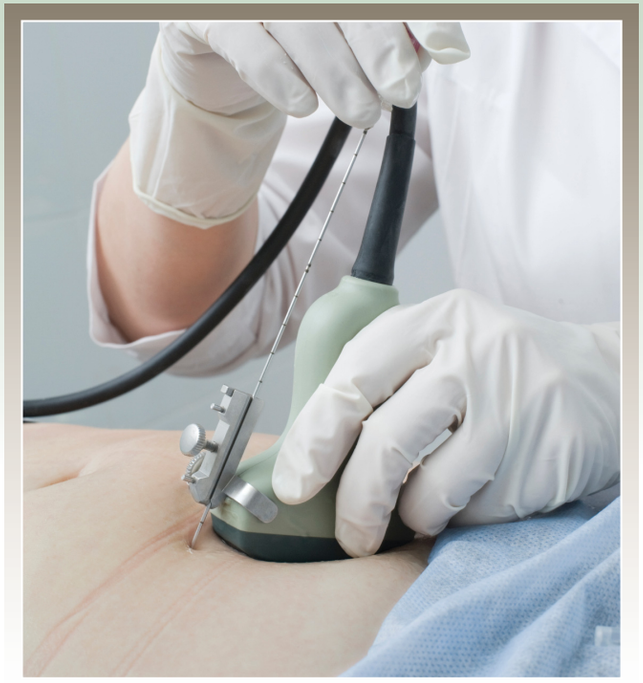
A positive test result means that there is a very high chance that your baby has Down syndrome, trisomy 18, or trisomy 13. A small number of women – 1 in 500 – will have a positive test result even though the baby does not have any of these conditions. When the test result is positive, the only way to tell for sure whether your baby is affected is to have an amnio or CVS. Your health care provider or genetic counselor will talk with you about this.

What if my blood test is negative (normal)?

A negative test may mean that the chance that your baby has Down syndrome, trisomy 18, or trisomy 13 is less than 1 in 2000. You would no longer be considered at "high-risk."

How will I know whether my results are normal or not?

The results of this blood test may take up to two weeks to get back to your doctor or genetic counselor. They will let you know what the results are and whether any other testing may be helpful. If you decide to have a CVS or amnio, those results may take a week or more as well.



If I decide not to have the blood test, what are my choices?

If you choose to have an amnio or CVS instead of having the blood test, you'll know in about a week whether or not your baby has a chromosome condition.

If you choose not to have any testing, you may not know until delivery whether or not your baby has a chromosome problem.

Are CVS and amniocentesis safe?

CVS is done early in pregnancy and takes a small sample of placental cells made by the baby. **Amniocentesis** is done later in the pregnancy and uses a needle to take a small sample of fluid and cells that surround the baby. While these tests are very dependable, many women find them to be uncomfortable, and there is a small chance that CVS or amnio may cause pregnancy loss — a miscarriage. Although this chance is about 1 in 300 to 1 in 600 or less, it's a chance that some women do not want to take.

As you decide about testing, you may want to consider:

- If you prefer an invasive test that will tell for sure if your baby has Down syndrome, trisomy 18, or trisomy 13 or if you prefer a blood test that may change your risk from "high" to "low."
- How you feel about the risk that comes with amnio or CVS (ask your doctor or genetic counselor if it is 1 in 200 at your prenatal diagnosis center)
- That you must be at least 10 weeks pregnant to have the blood test. Also, remember that you may need to wait up to two weeks for the results.
- That the blood test won't have an answer for 1 woman in about 125 because there is not enough DNA in the blood sample
- That only CVS or amnio, not the blood test, can tell you about some other genetic conditions that your baby might have
- What you would do if any of your tests results are positive
- Whether you can afford the cost of the new blood test. Because this is a new test, some insurance companies do not pay for it. Even if they do, you might still have to pay some of the cost yourself. CVS and amnio are usually paid for by insurance or Medicaid.

It is always a good idea to talk with your health care provider or genetic counselor about your choices and any questions that you may have. Make your decision only after you have all the information that you need. If your insurance won't pay for the testing, ask your doctor or genetic counselor if the lab has a financial aid plan. If you choose to have any testing, only you and your health care provider or genetic counselor will know the results. This information cannot be shared with anyone else without your permission.

Remember that you can choose not to have any testing at all.

For more information about Down syndrome (trisomy 21), trisomy 18, or trisomy 13, please speak with your doctor or genetic counselor, or visit womenandinfants.org.