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/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/amnio is usually paid for by insurance or Medicaid.

Image of
patient talking with provider

It's 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 unless you agree

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

This new test was studied in 2011 by doctors at Women & Infants Hospital of Rhode Island. You can read more about this test at **www.ipmms.org**.

For more information about Down syndrome (trisomy 21), trisomy 18, or trisomy 13, please ask your doctor or genetic counselor.

PRENATAL DIAGNOSTIC CENTER

Your Institution

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

Image of
mother and newborn

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

TEL:

Why is this new DNA blood test being offered to me?

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 amniocentesis. These are known as "invasive" tests because a doctor has to remove cells from inside you to study your baby's chromosomes.

Are CVS and amniocentesis safe?

Image of amniocentesis

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 200 or less, it's a chance that some women do not want to take.

What about this new DNA blood test ?

There is a new test using blood taken from your arm that can lower the chance that you'll have to consider CVS or amnio. This new blood test has no chance of causing pregnancy loss, and it can correctly identify 99 out of 100 pregnancies with Down syndrome. It can also find almost all pregnancies with trisomy 13 and trisomy 18. This new DNA blood test does not take the place of CVS or amnio, but can reduce the number of women who will need to decide about having those tests.

Image of venipuncture

What will this new blood test tell me?

The test looks to see if your blood contains extra material from certain chromosomes. It's normal for your blood to have pieces of your baby's genetic material in it as well as yours. 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 high chance that your baby has Down syndrome, trisomy 18, or trisomy 13. There is a small chance – 1 in 10 - that 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 2 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. There is a small chance that the amnio or CVS may cause a miscarriage.

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

As you decide about testing, you may want to consider

- if you prefer an invasive test that will tell for sure that 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/CVS (ask your doctor or genetic counselor if it is 1 in 200 where you are)
- that you must be at least 10 weeks along to have the blood test. (Be prepared to wait up to