Patient Education

intermountainhealthcare.org

A Guide to Prenatal Testing

LIVING AND LEARNING TOGETHER

Intermountain® Healthcare

Most news is **good news.**

Most babies are born **without** major birth defects.

Early in your pregnancy, you'll need to make decisions about prenatal testing. Prenatal tests aim to detect the risk or presence of a birth defect or serious disease in your developing baby.

This guide gives you the facts you need to make decisions about testing. Spend some time with this guide. Take it home and read it carefully. At your next prenatal checkup, ask any remaining questions before making your decisions.

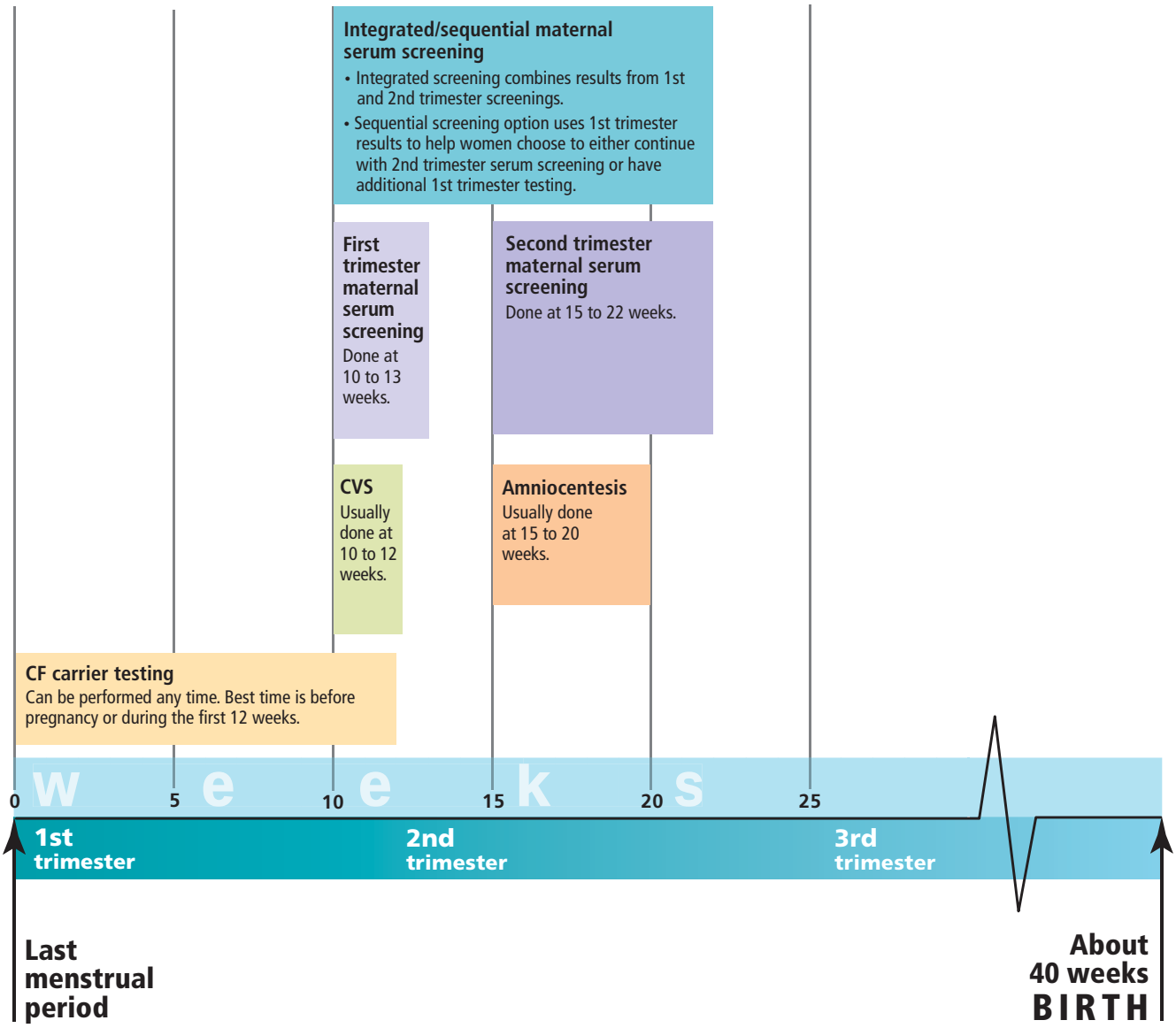
What's Inside:

At a glance

These two pages summarize the prenatal tests described in this guide and show you when each happens during pregnancy.

TESTING TIMELINE

When do different prenatal tests occur during pregnancy?



Cystic fibrosis (CF) carrier testing — *more on pages 7 and 8*

- This blood test tells whether you carry the altered gene that causes CF. Your baby is at greater risk for having CF only if both you and the baby's father are carriers. In this case, your baby has a 25% chance of having CF.
- Carrier testing does NOT tell you whether your baby actually has CF.
- Results apply not only to this pregnancy — but also to any future pregnancy that you and your partner have together.

1st trimester maternal serum screening — *more on pages 9 to 11*

- Paired with an ultrasound, results from this blood test calculate your chance of having a baby with Down syndrome.
- The screening does NOT tell you whether you baby has — or will have — Down syndrome. Rather, it places you in a higher or lower risk category for having a baby with this disorder.
- The screening can detect about 75% of pregnancies affected by Down syndrome.

2nd trimester maternal serum screening — *more on pages 9 to 11*

- Results from this blood test calculate your chance of having a baby with Down syndrome, trisomy 18, or a neural tube defect (NTD).
- The screening does NOT tell you whether you baby has — or will have — one of these problems. Rather, it places you in a higher or lower risk category for having a baby with the disorder.
- The screening can detect about 75% of pregnancies affected by Down syndrome, 70% of those affected by trisomy 18, and 85% of those affected by an NTD.

Integrated maternal serum screening — *more on pages 9 to 11*

- This test is a combination of 1st and 2nd trimester serum screenings. It looks at the two results together to create a clearer picture of your baby's risk for Down syndrome, trisomy 18, or a neural tube defect (NTD).
- As with the other screenings, the test does NOT tell you whether you baby has — or will have — one of these problems. It simply places you in a higher or lower risk category having a baby with the disorder.
- Sequential screening is a stepwise version of integrated screening. It uses 1st trimester results to help women choose to either continue with 2nd trimester serum screening or have additional 1st trimester testing.
- The screening can detect about 90% of pregnancies affected by Down syndrome, 90% of those affected by trisomy 18, and 80% of those affected by an NTD.

Chorionic villus sampling (CVS) — *more on pages 12 to 14*

- CVS requires a small sample of the placenta. The sample is then tested in a lab.
- Routine results tell you whether your baby has Down syndrome or trisomy 18. CVS can also check for other genetic disorders such as cystic fibrosis (CF).
- This test can be done as early as 10 weeks of pregnancy.
- CVS has a very low rate of pregnancy loss (miscarriage).
- CVS is 99% accurate in identifying genetic disorders.

Amniocentesis — *more on pages 12 to 14*

- Amniocentesis requires a small sample of the fluid surrounding your developing baby (the amniotic fluid). The sample is then tested in a lab.
- Routine results tell you whether your baby has a neural tube defect (NTD) or a genetic disorder such as Down syndrome or trisomy 18. Amniocentesis can also check for other genetic disorders such as cystic fibrosis (CF).
- Amniocentesis has a very low rate of pregnancy loss (miscarriage).
- Amniocentesis is 99% accurate in identifying genetic disorders.

COUNTING THE TIME IN YOUR PREGNANCY

Many women don't know the exact day they conceived. That's why doctors date pregnancies from the first day of the last menstrual period. The average length of pregnancy is 280 days (40 weeks) from the beginning of the last menstrual period.

The 40 weeks of pregnancy are split into 3 roughly equal parts, called trimesters. The prenatal tests discussed in this guide take place in the first and second trimesters of pregnancy.

Tests

Common prenatal options

This section gives more information about the prenatal testing options listed on the previous pages. As you read, keep in mind that you won't need to have every test described here. (Some of them check for the same conditions.) The other thing to remember is that no test is perfect. No test is 100% accurate, nor is it possible to test for every condition.

WHAT ABOUT ROUTINE TESTS DURING PREGNANCY?

In addition to the prenatal screenings and tests described in this booklet, there are also many routine checks and tests during pregnancy. These happen during your regular prenatal visits and help your doctor know how you and your growing baby are doing. Ask your doctor or midwife if you have questions about these routine tests:

- **URINE TESTS** to check for **urinary tract infections** and signs of **gestational diabetes** and **preeclampsia**.
- **CERVICAL AND VAGINAL TESTS** to check for signs of **cervical cancer**, **chlamydia**, and **gonorrhea**. At 34 weeks or later, you'll have a **Group B strep test** to see if you need treatment to protect your baby from this bacteria during birth.
- **A GLUCOSE SCREENING TEST**, usually done between 24 and 30 weeks, that checks for signs of **gestational diabetes.** This test requires you to drink a sweet liquid, and then have your blood drawn an hour later. If results show that your blood sugar is too high, your doctor can teach you how to control it.
- **OTHER BLOOD TESTS** to determine your **blood type** and **Rh factor**, check for anemia, and see if you have (or are at risk for) **rubella**, **chickenpox**, **syphilis**, or **hepatitis**.
- You may also have a blood test to see if you have **HIV**, the virus that causes AIDS. The Centers for Disease Control (CDC) recommends that ALL pregnant women be offered an HIV test. Why? Because if you have HIV, your baby can get it from you during pregnancy, labor, or breastfeeding. But if you get treatment, you can lower the chance that this will happen.

Cystic fibrosis (CF) carrier testing

CF carrier testing looks at the risk of you and your baby's father having a child with CF. CF is a serious genetic disorder. It can shorten life and require daily treatment. For more information on CF and your risk for having a child with CF, see page 15.

How is the test performed, and what does it tell me?

CF carrier testing is done in a laboratory, using a sample of your blood or saliva. It can be done at any time, even if you're not pregnant. If you choose this test during pregnancy, it's best to have it in the first trimester. That way, you'll have more time to make decisions about the pregnancy.

The carrier test looks to see if you carry the altered gene that causes CF. If the test shows that you do, the baby's father is tested as well. Your baby is at greater risk for having CF only if both you and the father are carriers. In this case, there is a 25% chance (1 in 4) of your baby having CF. (Your own health is not affected.)

What if results say that we're both carriers?

If you and the baby's father are both found to be carriers, you can choose to test your baby during pregnancy. Amniocentesis and chorionic villus sampling (CVS), prenatal tests described later in this guide, can tell whether the developing baby actually has CF. Or, you may choose to wait until after birth for this information. All newborns are routinely screened for certain disorders, including CF. These screenings detect most, but not all, cases of CF. For a diagnosis of CF in a newborn, further testing is required.

WHAT'S A *genetic disorder?*

A **genetic disorder** is a problem caused by a change in one of your genes, the basic "blueprints" that control your body's growth and function. These gene changes (mutations) are often inherited from parents who are carriers. But they can also happen if a gene is damaged. That's why a genetic disorder can occur in any pregnancy, even if there's no history of it in your family.

Genetic disorders can't be cured. Cystic fibrosis, Down syndrome, and trisomy 18 are all genetic disorders.

What are the risks of the CF carrier test?

The CF carrier test has no risks to you or to the pregnancy.

Who might choose CF carrier testing?

As with any testing, the decision to have CF carrier testing is personal. You might be influenced by your view of the risk based on ethnic background or family history (see page 15).

MORE FACTS ABOUT CF CARRIER TESTING

- Since the test is about you and the father's status as carriers, the results apply to any pregnancy you have together — not just this one. For example, if you're both carriers, any children you have in the future will face the same 25% chance of CF.
- The carrier test will find most — but not all — Caucasian (white) carriers. But the test's accuracy is less for other ethnic groups.
- You may want to check with your health insurance carrier to find out if CF carrier testing is covered.

Maternal serum screening

Maternal serum screening measures the levels of certain chemical markers in your blood while you're pregnant. It may also use information from ultrasound images. Analyzing this data together helps determine whether there's a high chance or a low chance that your pregnancy is affected by a condition such as Down syndrome, trisomy 18, or spina bifida.

There are different options for maternal serum screening. This section describes three of the most common options.

What are the options — and what do they tell me?

All of these screenings happen during the first or second trimester of pregnancy.

1. **First trimester screening** is done between the 10th and 13th week of pregnancy. It involves an ultrasound and a blood test. The ultrasound has two main uses. First, it's used to double-check the age of the developing baby (**gestational age**). Second, it's used to measure the fluid on the back of the baby's neck (the **nuchal translucency**). The ultrasound findings and the blood test results are analyzed together.

   First trimester screening estimates the chance that you'll have a baby with Down syndrome. It can't tell you whether or not your baby actually has the disorder. The screening detects about 75% of Down syndrome cases.

2. **Second trimester screening** is done between the 15th and 22nd week of pregnancy. Also called **quad screening**, this blood test measures the levels of four different markers in your blood. The screening is sometimes paired with an ultrasound.

   Second trimester screening estimates the risk of your baby having Down syndrome, trisomy 18, or a neural tube defect (NTD). It can also detect some problems with the placenta, the special tissue that joins you and your baby inside the womb (**uterus**). This screening can detect about 75% of Down syndrome pregnancies, 70% of trisomy 18 pregnancies, and 85% of pregnancies with NTDs.

WHAT IS A PREGNANCY ULTRASOUND?

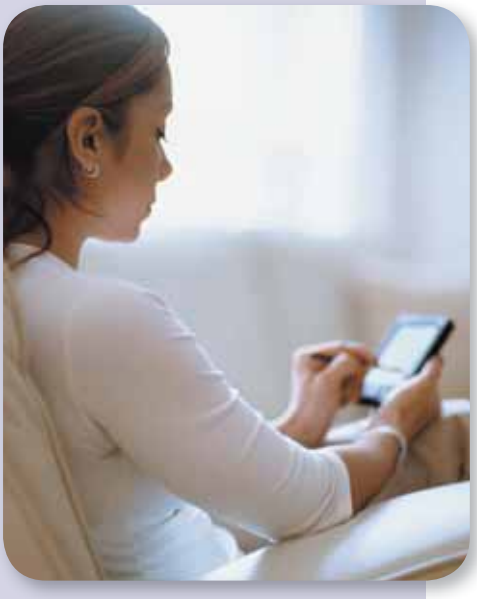
In pregnancy, **ultrasound** (sonography) is a way to capture images of your developing baby. It creates pictures by bouncing special sound waves off body structures. Doctors use ultrasound to measure the baby's size, check the gestational age, and look for some types of problems.

for more information...

See pages 16 and 17 to learn more about Down syndrome, trisomy 18, and neural tube defects (NTDs).

What else CAN SCREENING PICK UP?

Sometimes maternal serum screening can identify an increased risk for pregnancy complications. For example, a screening may identify poor growth of the baby, or too little fluid around the baby.



3 **Integrated screening** is a combination of first and second trimester screening. The results of integrated screening are based on the 10 to 13 week blood draw and ultrasound as well as the second blood test at 15 to 22 weeks.

You can get the integrated screening results after the second blood test. (And if you choose the integrated screening — but miss the second blood test — you can still get the results of the first trimester screening.)

Integrated screening estimates the risk for Down syndrome, trisomy 18, NTDs, and problems with the placenta. It's the most accurate of the maternal serum screening options for detecting Down syndrome. It can detect about 90% of Down syndrome pregnancies, 90% of trisomy 18 pregnancies, and 80% of pregnancies with NTDs.

Sequential screening is a stepwise approach to integrated screening. It involves undergoing the first part of integrated screening (the blood test and ultrasound) and then pursuing additional testing based on these early results. For example, the first results are reported as either:

- Less than 1 in 25 chance of genetic defect, in which case you continue with integrated screening and have the second blood test at 15 to 22 weeks
- Greater than 1 in 25 chance of genetic defect, in which case you are offered other options to evaluate your pregnancy (such as CVS or amniocentesis, tests discussed on pages 12 to 14)

Sequential screening lets women know if they are at very high risk (greater than 1 in 25 chance) for a disorder and allows them to learn more without waiting for second trimester test results.

What are the risks of maternal serum screening?

There are no risks to you or to the pregnancy from maternal serum screening.

Who might choose one of these screenings?

People choose maternal serum screening for many different reasons. Women who think they may be at increased risk (because of their age or family history) often choose it. Many like the fact that the screening gives information without any risk to the pregnancy. Some choose screening, with a plan to have further testing if the results suggest possible problems. Yet others decide that screening doesn't give them enough concrete information, and decide to forego the screenings.

How are screening results reported?

All three screening options give results as either ***screen positive*** or ***screen negative*** for a particular birth defect:

- If your screening result places you in a higher risk category for having a baby with the birth defect, you're said to have a **screen positive** result.
- If your screening result places you in a lower risk category, you're said to have a **screen negative** result.

Keep in mind that **a screen positive result doesn't mean that your baby has a problem**. It just means that there's a higher-than-average risk of a problem. Only further testing can tell for sure whether your pregnancy is affected.

What if I have a screen positive result?

If your screening result places you in a higher risk category for having a baby with a birth defect, you may decide that you want more concrete information. Here, your options depend on which screening you've chosen, and what your particular results are:

- For a first trimester screen positive result, you can choose a more detailed ultrasound, chorionic villus sampling (CVS), or amniocentesis.
- For a second trimester, integrated testing result, you may choose to have another, detailed ultrasound. Or, you can choose to have amniocentesis.

Whether you decide to have more testing, it's a good idea to talk with your doctor or genetic counselor about your results. They can help you understand the risk and your options.

RISK GROUPS — *not certainties*

Screenings assess the **risk** of birth defects, not the presence of birth defects. And, like all tests, they're not perfect. So you shouldn't view a screening result as an absolute indicator of your baby's health. Sometimes the screening will give a negative result when there's a problem. Sometimes the screening places someone in a higher risk group even when the pregnancy is normal. (This is then called a **false positive** result.)

But in spite of this uncertainty, serum screening is a popular choice for many pregnant women. Combined, integrated screening in particular has a high detection rate for Down syndrome, and a fairly small false positive rate.

for more information...

See page 18 to learn about how a genetic counselor may help you.

Chorionic villus sampling (CVS) and amniocentesis

CVS and amniocentesis are similar in many ways. Both are highly accurate ways to check for genetic disorders. They are both called invasive tests because they require going inside the mother's womb (uterus) to get the test sample. This section describes CVS and amniocentesis, their similarities, and their differences.

What is CVS?

CVS is a prenatal test done between 10 and 12 weeks of pregnancy. It involves testing a sample of the placenta, the special tissue that joins you and your developing baby.

CVS routinely checks for genetic disorders like Down syndrome and trisomy 18. It can also test for other genetic disorders like cystic fibrosis (CF). Talking to your doctor or genetic counselor will help determine whether CVS is right for you and what specific conditions it should test for.

How is CVS performed, and what does it tell me?

During CVS, a doctor inserts a hollow needle into the placenta. (To reach the placenta, the doctor can either go through your abdomen into your uterus, or up through the vagina and cervix.) Ultrasound shows the doctor where to safely put the needle. Once the needle is in place, the doctor withdraws a small sample of the placenta. The needle is then taken out, and the sample is sent to a laboratory for testing.

It takes about 7 days to get your results from CVS. The results are highly accurate. With more than 99% accuracy, CVS can tell you whether your baby has a genetic disorder.

What are the risks of CVS?

CVS can cause cramping, bleeding, and infection. It has a very low rate of pregnancy loss (miscarriage).

What is amniocentesis?

Amniocentesis is a test usually performed between 15 and 20 weeks of pregnancy. It involves testing a sample of the fluid that surrounds your developing baby (the amniotic fluid).

Amniocentesis routinely checks for neural tube defects (NTDs) and genetic disorders such as Down syndrome and trisomy 18. It can also test for other genetic disorders like cystic fibrosis (CF). Talking to your doctor or genetic counselor will help determine whether amniocentesis is right for you, and what specific conditions it should test for.

How is amniocentesis performed, and what does it tell me?

During amniocentesis, a doctor inserts a hollow needle through your abdomen into your uterus. Ultrasound shows the doctor where to safely put the needle. Once the needle is in place, the doctor withdraws a small amount of fluid. The needle is then taken out, and the sample is sent to a laboratory for testing.

It takes about 10 to 14 days to get your results from amniocentesis. The results are highly accurate. With more than 99% accuracy, amniocentesis can tell you whether your baby has a genetic disorder.

What are the risks of amniocentesis?

Although amniocentesis is fairly safe, it does carry risks. It can cause cramping, bleeding, and infection. It can cause leaking of the amniotic fluid. Amniocentesis has a very low rate of pregnancy loss (miscarriage).

A MATTER OF *timing*

One possible advantage of CVS is timing. CVS can give results between 12 and 15 weeks of pregnancy. Amniocentesis results are usually not available until 17 to 22 weeks.

Earlier results give you more time to consider your options if a genetic disorder is found. For some people, options include terminating the pregnancy. Doctors advise that a first trimester termination is often safer, more easily performed, less expensive, and less emotionally difficult than one done later.

Who usually has CVS or amniocentesis?

Women who choose CVS or amniocentesis are often those who:

- Are older at the time of the pregnancy, as the risk of genetic disorders increases with the mother's age
- Have a family history of a disorder that can be detected by CVS or amniocentesis
- Have had prenatal screening results suggesting an increased risk of a genetic disorder
- Seek reassurance about their pregnancies

How do you choose between these two tests?

Many women are choosing between CVS and amniocentesis. Both tests offer reliable information about whether a pregnancy is affected by a genetic disorder.

One possible advantage of CVS is timing. (See the sidebar at left). An advantage of amniocentesis is that it can help detect neural tube defects.

What if I get abnormal results?

If CVS or amniocentesis reveals a birth defect, first learn as much as you can about the condition and your options. Talk to your doctor or genetic counselor. You may also want to talk to parents of children with the same disorder. Learning as much as you can will help you make the best decision about the future.

Conditions

The diseases and disorders discussed in this guide

This section summarizes the conditions that prenatal testing may detect. It also discusses some of the factors that affect your risk for having a baby with one of these conditions.

Cystic fibrosis (CF)

Cystic fibrosis (CF) is the most common life-shortening genetic disease in the Caucasian (white) population. CF causes severe problems with breathing and digestion. It cannot be cured. It requires daily, lifelong treatment.

In the past, children with CF rarely lived to adulthood. Today, newer treatments allow people with CF to live longer and more comfortably. Now the average life span for someone with CF is about 34 years.

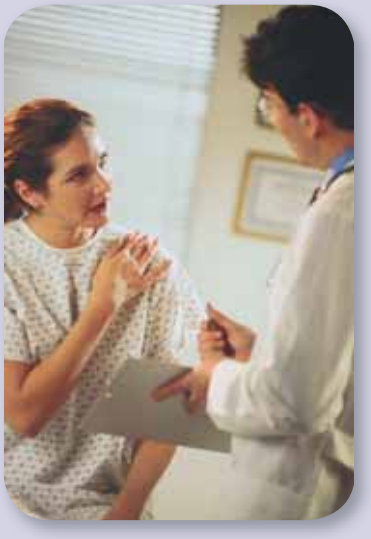
What affects my chances of having a baby with CF?

Your chances of having a baby with CF depend on whether you and the baby's father carry the altered gene that causes CF. This, in turn, may depend on your ethnic background. See the table below to get an idea of how common CF is in your ethnic group.

Family history is also a factor. If someone in your family has CF — or is known to be a carrier of the CF gene — you have an increased risk of having a baby with CF. Still, most couples who have a child with CF have no known family history of the disease.

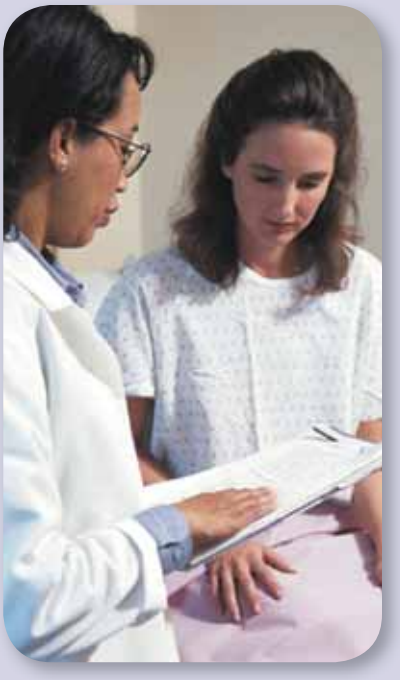
Ethnicity/Race	Chance of being a CF carrier	Babies born with CF in the US each year
European Caucasian	1 in 29	1 out of 3,300
Hispanic American	1 in 45	1 out of 9,000
African American	1 in 65	1 out of 15,300
Asian American	1 in 90	1 out of 32,000

Regardless of ethnicity, both parents must be carriers of the CF gene for their baby to develop CF. If both parents are carriers, their baby has a 25% chance (1 in 4) of having CF.



A CHANCE, A CHOICE

Birth defects can happen in any pregnancy. But there are factors that can affect the odds. Learning about these factors — and the disorders themselves — can help you decide about prenatal testing.



AGE AND RISK

Your risk for having a baby with Down syndrome increases as you age. Still, anyone — even younger mothers — can have a baby with this disorder. In fact, most babies with Down syndrome are born to women who are under age 35. (Younger women have far more babies.)

Down syndrome

Down syndrome is a genetic disorder and the most common cause of intellectual disability. Most children born with Down syndrome have mild or moderate intellectual disability and benefit from special education. Some are born with heart or bowel problems that can be corrected with surgery. As adults, people with Down syndrome may successfully hold a job, but they're unlikely to live independently. Life expectancy for people with Down syndrome is close to that of the general population.

What affects my chances of having a baby with Down syndrome?

Your risk for having a baby with Down syndrome is linked to your age during pregnancy. The older you are, the greater your chance of having a baby with Down syndrome (see the table below). Having a family history of Down syndrome may also increase the chance.

Mother's age	Risk of having a live baby with Down syndrome
20	1 in 1,667
25	1 in 1,250
30	1 in 952
33	1 in 602
35	1 in 378
37	1 in 224
40	1 in 106
41	1 in 82
42	1 in 63
43	1 in 49
44	1 in 38
45	1 in 30

Trisomy 18

Trisomy 18 is a genetic disorder that affects about 1 in 5,000 babies born in the U.S. It causes severe intellectual disability and physical defects. Most babies with trisomy 18 live for only a few weeks or months after birth.

What affects my chances of having a baby with trisomy 18?

As with Down syndrome, the chance for this disorder increases with the mother's age. But the chance for trisomy 18 at the ages listed in the table above is less than that for Down syndrome.

Neural tube defects (NTDs)

Neural tube defects (NTDs) are caused by problems in the development of the baby's brain, skull, backbone, or spinal cord during the early part of pregnancy. Two more common NTDs are **spina bifida** and **anencephaly**:

- **Spina bifida.** The effects of spina bifida vary a lot from person to person. Some people have only mild problems. Others have leg paralysis and lack of bladder or bowel control. Some people with spina bifida are intellectually disabled.
- **Anencephaly.** With this severe defect, the brain and head do not develop normally. Nearly all babies with anencephaly are stillborn or die soon after birth.

What affects my chances of having a baby with an NTD?

Anyone can have a child with an NTD. And since NTDs are probably caused by many factors, it's difficult to know the risk for any one pregnancy. Still, scientists have learned the following:

- Most babies with NTDs are born to women without a family history of the disease. But having this history does increase your baby's risk for a NTD. So does having diabetes or a seizure disorder treated by certain medications.
- Women who take a vitamin with 400 mcg (micrograms) of folic acid before and during their pregnancy have a much smaller chance of having a baby with an NTD or other birth defect.

TAKE A PILL!

Take a vitamin with at least 400 mcgs (micrograms) of folic acid every day…

- Before you're pregnant
- During your pregnancy
- After you're pregnant
- Always — if you have any chance of getting pregnant, on purpose or accidentally

Folic acid is important to help prevent NTDs in a developing baby. And it's good for you, too. So if you have any chance at all of becoming pregnant — take a pill!

Considerations

Things to think about as you decide about testing

The tips on this page can help you weigh your options and find more information if you need it.

Who you can talk to

It often helps to talk to others about your prenatal testing options. Your doctor or midwife can probably answer many of your questions. But you may also want to talk to a genetic counselor.

A **genetic counselor** is a medical professional who can analyze your family's medical history and help you understand your risk of having a baby with a particular disorder. A counselor can also clarify the pros and cons of different prenatal tests and help you interpret your results. Talk to your doctor if you're interested in meeting with a genetic counselor.

Questions to ask yourself

As you make decisions about testing, consider the questions below.

- What do I hope to learn from this test? Can the test give me what I'm looking for?
- How will I cope with waiting for results, or with any uncertainty in the results?
- Does the value of the test (the information it gives me) outweigh any risks?
- How do I interpret my risk for having a child with this disease or disorder? (Risk can be based on such things as your age, ethnic group, family history, etc.)
- How serious does this condition seem to me?
- What if the test identifies problems? What decisions might the news affect, and what choices might I make? For example, I may need to make decisions about:
  - Further testing or monitoring during the pregnancy
  - Continuing the pregnancy
  - Creating a delivery plan that works for me
  - Arrangements for special care or treatment at delivery
  - Plans for caring for a child with special needs
  - Meeting with a specialist to help coordinate care

WHERE TO LEARN MORE

Check out Intermountain Healthcare's Health Resources site:

intermountainhealthcare.org/health

Scroll to the bottom of the page to access Intermountain's Women and Newborn Services site.

Verification of Education

It's important that you understand the information in *A Guide to Prenatal Testing.*

If you have any questions about the tests described in this guide, please ask your health care provider before signing this form.

❏ **I have read and I understand the information in *A Guide to Prenatal Testing.***

I plan to choose the following:

- ❏ Cystic fibrosis (CF) carrier testing
- ❏ First trimester maternal serum screening
- ❏ Second trimester maternal serum screening
- ❏ Integrated maternal serum screening (sequential? ☐ yes ☐ no)
- ❏ Chorionic villus sampling (CVS) *(separate consent form required)*
- ❏ Amniocentesis *(separate consent form required)*
- ❏ None of the above

_____

Signed

_____

Date

To find this booklet and other resources, go to:

intermountainhealthcare.org/health

Also available in Spanish/También disponible en español

Patient and Provider Publications 801.442.2963 WN008 – 1/12