



## Routine Tests During Pregnancy

**D**uring pregnancy, certain lab tests are recommended for all women. Although most women have healthy pregnancies and healthy babies, routine tests can help detect possible problems. Additional tests may be suggested depending on your medical history, family or ethnic background, or previous test results. The sooner a problem is found, the sooner it can be treated and managed. This pamphlet explains

- why certain tests are done during pregnancy
- tests that are performed early in pregnancy
- tests that are performed later in pregnancy
- testing for birth defects

### Why Tests Are Done During Pregnancy

A number of lab tests are suggested for all women as part of routine *prenatal care*. Some of these tests are performed on a blood sample. Others use urine or a sample taken from your *vagina, cervix, or rectum*. These tests can help find conditions that can increase the risk of complications for you and your *fetus*. If problems are found, many can be treated during pregnancy.

### Tests Performed Early in Pregnancy

The following routine lab tests are done early in pregnancy:

- Complete blood count (CBC)—The CBC counts the numbers of different types of *cells* that make up your blood. The number of red blood cells
- Blood type—Just as there are different major blood groups, such as type A and type B, there also is an *Rh factor*. The Rh factor is a protein that can be present on the surface of red blood cells. Most people have the Rh factor—they are Rh positive. Others do not have the Rh factor—they are Rh negative. If the fetus is Rh positive and the woman is Rh negative, the woman's body can make *antibodies* against the Rh factor that can damage the fetus's red blood cells. Problems usually do not occur in a first pregnancy when only a small number of antibodies have a problem with blood clotting.
- can show whether you have a certain type of *anemia*. The number of white blood cells shows how many disease-fighting cells are in your blood, and the number of platelets can reveal whether you

are made, but they can occur in a later pregnancy. These problems can be prevented by giving *Rh immunoglobulin* during pregnancy. During the first trimester of pregnancy, you will have a blood test to find out your blood type and whether you are Rh positive or Rh negative. If you are Rh negative, your blood also will be tested to see if you have made antibodies to the Rh factor. If you already have made a certain number of Rh antibodies, you may need special tests and monitoring throughout pregnancy. The baby also may need treatment after birth.

- **Urinalysis**—Your urine may be tested for red blood cells (to see if you have urinary tract disease), white blood cells (to see if you have a urinary tract infection), and *glucose* (high levels may be a sign of *diabetes mellitus*). The amount of protein also is measured. The protein level early in pregnancy can be compared with levels later in pregnancy. High protein levels may be a sign of *preeclampsia*, a serious complication that usually occurs later in pregnancy or after the baby is born.
- **Urine culture**—This test looks for *bacteria* in your urine, which can be a sign of a urinary tract infection. Sometimes these infections do not cause symptoms. Your urine will be tested early in pregnancy and later in pregnancy. If your test result shows that you have bacteria in your urine, you will be treated with *antibiotics*. After you finish treatment, you may have a repeat test to see if the bacteria are gone.
- **Rubella**—Your blood is tested to check whether you have had a past infection with rubella (sometimes called German measles) or if you have been vaccinated against this disease. Rubella can cause birth defects if a woman is infected during pregnancy. If you had this infection before or you have been vaccinated, you are not likely to get it again—you are *immune* to the disease. If your blood test shows you are not immune, you should avoid anyone who has the disease while you are pregnant because it is highly contagious. The vaccine contains a live virus and is not recommended for pregnant women. If you have not had the vaccine, you should get it after the baby is born, even if you are breastfeeding.
- **Hepatitis**—Hepatitis B and hepatitis C are viruses that infect the liver. Pregnant women who are infected with hepatitis B or hepatitis C can pass the virus to their babies. All pregnant women are tested for hepatitis B virus infection. If you have risk factors, you also may be tested for the hepatitis C virus. If you are infected with either virus, you may need special care during pregnancy. Your baby also may need special care after birth. You still can breastfeed if you have either infection. A vaccine that protects against hepatitis B is available. It is given in a series of three shots, with the first dose given to the baby within a few hours of birth.

- **Sexually transmitted infections (STIs)**—All pregnant women are tested for *syphilis* and *chlamydia* early in pregnancy. Tests for these STIs may be repeated later in pregnancy if you have certain risk factors. Syphilis and chlamydia can cause complications for you and your baby. If you have either of these STIs, you will be treated during pregnancy and tested again to see if the treatment has worked. Your sex partner or partners also should be treated. If you have risk factors for *gonorrhea* (you are aged 25 years or younger or you live in an area where gonorrhea is common), you also will be tested for this STI.
- **Human immunodeficiency virus (HIV)**—This virus attacks cells of the body's immune system and causes *acquired immunodeficiency syndrome (AIDS)*. If you are infected with HIV, there is a chance you could pass it to your baby. While you are pregnant, you can be given medication that can greatly reduce this risk. You also can get specialized care to ensure that you stay as healthy as possible throughout your pregnancy.
- **Tuberculosis (TB)**—Women at high risk of TB (for example, women who are infected with HIV or who live in close contact with someone who has TB) should be tested for this infection.

#### Tests Performed Later in Pregnancy

Some tests that were done earlier in pregnancy may be repeated later in pregnancy in certain situations. The following tests are done later in pregnancy:

- **CBC**—This blood test may be repeated to check for anemia and other problems.
- **Rh antibody test**—If you are Rh negative, your blood will be tested again for Rh antibodies between 28 weeks and 29 weeks of pregnancy. If you do not have Rh antibodies, you will receive Rh immunoglobulin. This shot prevents you from making antibodies during the rest of your pregnancy. If you have Rh antibodies, you may need special care.
- **Glucose screening test**—This screening test measures the level of glucose (sugar) in your blood. A high glucose level may be a sign of *gestational diabetes*, which can cause problems during pregnancy. For this test, you drink a special sugar mixture. An hour later, a blood sample is taken and sent to a lab. If your glucose level is high, you will have another type of glucose test to confirm the results. This test usually is done between 24 weeks and 28 weeks of pregnancy. If you have risk factors for diabetes or had gestational diabetes in a previous pregnancy, screening may be done in the first trimester of pregnancy.
- **Group B streptococci (GBS)**—GBS is a type of bacteria that lives in the vagina and rectum. Many women carry GBS and do not have any symptoms. GBS can be passed to a baby during birth. Most babies who get GBS from their mothers do not have any problems. A few, however, become sick. This illness can cause

serious health problems and even death in newborn babies. GBS usually can be detected with a routine screening test that is given between 35 weeks and 37 weeks of pregnancy. For this test, a swab is used to take samples from your vagina and rectum. If you are found to carry the bacteria, antibiotics can be given during labor to help prevent the baby from becoming infected.

### Testing for Birth Defects

Most babies with birth defects are born to couples without risk factors. However, the risk of birth defects is higher when certain factors are present, such as a personal or family history of birth defects, belonging to certain ethnic groups, maternal age of 35 years or older, or having preexisting diabetes. Screening for birth defects begins by assessing your risk factors. Early in your pregnancy, your obstetrician or other health care professional may give you a list of questions to find out whether you have risk factors. If you do have risk factors, you may want to visit a *genetic counselor* for more detailed information about your risks.

Whether you want to be tested is a personal choice. Some couples would rather not know if they are at risk or whether their child will have a disorder, but others want to know in advance. Knowing beforehand allows the option of deciding not to continue the pregnancy. If you choose to continue the pregnancy, it can give you time to prepare for having a child with a particular disorder and to organize the medical care that your child may need. Your health care professional or a genetic counselor can discuss the options with you and help you decide.

### Types of Birth Defects

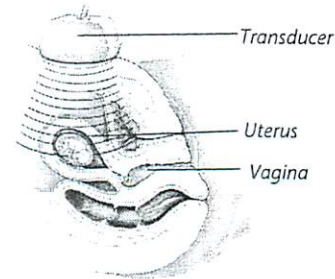
There are different types of birth defects. Some are caused by missing or extra *chromosomes*. This condition is called *aneuploidy*. The most common aneuploidy is called a *trisomy*, a condition in which there is an extra chromosome. One of the most common trisomies is *trisomy 21 (Down syndrome)*. Other trisomies include *trisomy 13* and *trisomy 18*. A *monosomy* is a condition in which there is a missing chromosome. A common monosomy is *Turner syndrome*, in which an *X chromosome* is missing in a female.

Other disorders are caused by defective *genes*. A defect in a gene is called a *mutation*. These disorders can be passed down by parents to their children. Some genetic disorders are more common in certain races and ethnic groups, such as *sickle cell disease* (African American), *cystic fibrosis* (non-Hispanic white), and *Tay-Sachs disease* (Ashkenazi Jewish, French Canadian, and Cajun descent). Mutations can occur on any of the chromosomes.

Birth defects also may be caused by exposure to harmful agents, such as medications, chemicals, and infections. Some birth defects may be caused by a combination of all of these factors. For about 70% of babies born with birth defects, it is not known for certain what caused the defect.

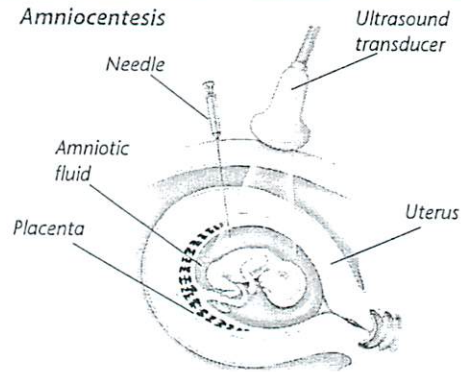
### Testing for Birth Defects

#### Ultrasound Exam



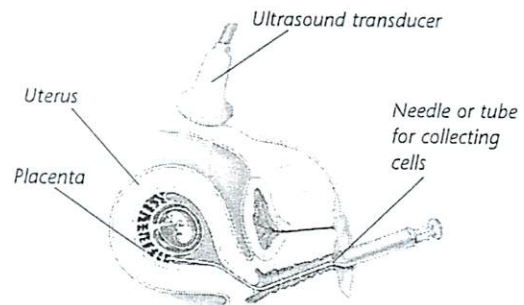
Energy in the form of sound waves is produced by a transducer. The sound waves are reflected off of the fetus. The reflected sound waves are changed into pictures that are shown on a video screen.

#### Amniocentesis



A small sample of amniotic fluid is removed with a needle to be studied.

#### Chorionic Villus Sampling



A small sample of cells (chorionic villi) is taken from the placenta to be studied.

### Tests for Birth Defects

A number of tests are available to help detect possible birth defects. A screening test for women at an increased risk of having a baby with certain birth defects also is available (see box).

**Carrier Screening Tests.** *Carrier* tests are screening tests that can show if a person carries a gene for a certain disorder, such as cystic fibrosis. Carrier tests can be done before or during pregnancy. Carrier testing often is recommended if you or your partner have a

genetic disorder, have a child with a genetic disorder, have a family history of a genetic disorder, or belong to an ethnic group that has an increased risk of specific disorders. Even if you do not have a family history, you still can request carrier screening for some disorders. Also, cystic fibrosis carrier screening is offered to all women of reproductive age because it is one of the most common inherited disorders. Once you know your carrier status for a particular disorder, you do not need to be tested again in a later pregnancy.

**Screening Tests.** Screening tests are done during pregnancy to assess the risk that the fetus has certain common birth defects. These include Down syndrome, Turner syndrome, and a few other chromosomal disorders as well as a group of non-genetic birth defects called *neural tube defects*. Screening tests include an ultrasound exam and blood tests that measure the levels of certain substances in the mother's blood. Screening tests cannot tell whether the baby actually has these disorders, only whether there is an increased risk that the baby will have the disorders. If you have a positive screening test result, diagnostic testing is needed to see if a disorder actually is present. There are no risks to the unborn baby from having these screening tests.

**Diagnostic Tests.** Diagnostic tests are available that can detect many, but not all, inherited defects and chromosomal disorders. These tests include *amniocentesis*, *chorionic villus sampling*, and a targeted ultrasound exam. If a screening test shows an increased risk of a birth defect, diagnostic tests may be done to help determine if a specific birth defect is present. Diagnostic testing may be done instead of screening if a couple has a family history of a birth defect, belongs to a certain ethnic group, or if the couple already has a child with a birth defect. Diagnostic tests also are available as a first choice for all pregnant women, including those who do not have risk factors. Some diagnostic tests carry risks, including a small risk of pregnancy loss.

#### Cell-Free DNA Test

A test called the cell-free DNA test uses a sample of a pregnant woman's blood to screen for three of the most common genetic disorders in the fetus: trisomy 13, trisomy 18, and trisomy 21. The screening test can be done as early as the tenth week of pregnancy. Test results can be available in about 1 week. Although the cell-free DNA test is not a diagnostic test, it is highly accurate. It is recommended that cell-free DNA testing be offered only to pregnant women at increased risk of having a child with trisomy 13, 18, or 21. Women who are 35 years and older, women who have a child with a trisomy, and women carrying a fetus that shows abnormalities on an *ultrasound exam* are at increased risk. The cell-free DNA test is not recommended for low-risk women.

#### Finally...

Lab tests are used throughout pregnancy to help find problems that could pose a risk to your health and to the health of your baby. Some tests are given routinely to all pregnant women. Others are given based on risk factors, family or ethnic background, or other test results. If test results reveal a possible problem, it often is possible to treat or manage the problem to reduce the risk of complications. Various types of tests for birth defects also are available. These tests are voluntary. Talk to your obstetrician or other health care professional if you have questions about routine lab tests or testing for birth defects.

#### Glossary

**Acquired Immunodeficiency Syndrome (AIDS):** A group of signs and symptoms, usually of severe infections, occurring in a person whose immune system has been damaged by infection with human immunodeficiency virus (HIV).

**Amniocentesis:** A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus.

**Anemia:** Abnormally low levels of blood or red blood cells in the bloodstream. Most cases are caused by iron deficiency, or lack of iron.

**Aneuploidy:** Having an abnormal number of chromosomes.

**Antibiotics:** Drugs that treat certain types of infections.

**Antibodies:** Proteins in the blood produced in reaction to foreign substances, such as bacteria and viruses that cause infection.

**Bacteria:** One-celled organisms that can cause infections in the human body.

**Carrier:** A person who shows no signs of a particular disorder but could pass the gene on to his or her children.

**Cells:** The smallest units of a structure in the body; the building blocks for all parts of the body.

**Cervix:** The lower, narrow end of the uterus at the top of the vagina.

**Chlamydia:** A sexually transmitted infection caused by bacteria that can lead to pelvic inflammatory disease and infertility.

**Chorionic Villus Sampling:** A procedure in which a small sample of cells is taken from the placenta and tested.

**Chromosomes:** Structures that are located inside each cell in the body and contain the genes that determine a person's physical makeup.

**Cystic Fibrosis:** An inherited disorder that causes problems in digestion and breathing.

**Diabetes Mellitus:** A condition in which the levels

of sugar in the blood are too high.

**Fetus:** The developing organism in the uterus from the ninth week of pregnancy until the end of pregnancy.

**Genes:** Segments of DNA that contain instructions for the development of a person's physical traits and control of the processes in the body. They are the basic units of heredity and can be passed down from parent to offspring.

**Genetic Counselor:** A health care professional with special training in genetics and counseling who can provide expert advice about genetic disorders and prenatal testing.

**Gestational Diabetes:** Diabetes that arises during pregnancy.

**Glucose:** A sugar that is present in the blood and is the body's main source of fuel.

**Gonorrhea:** A sexually transmitted infection that may lead to pelvic inflammatory infection, infertility, and arthritis.

**Human Immunodeficiency Virus (HIV):** A virus that attacks certain cells of the body's immune system and causes acquired immunodeficiency syndrome (AIDS).

**Immune:** Protected against infectious disease.

**Monosomy:** A condition in which there is a missing chromosome.

**Mutation:** A permanent change in a gene that can be passed on from parent to child.

**Neural Tube Defects:** Birth defects that result from incomplete development of the brain, spinal cord, or their coverings.

**Placenta:** Tissue that provides nourishment to and takes waste away from the fetus.

**Preeclampsia:** A disorder that can occur during pregnancy or after childbirth in which there is high blood pressure and other signs of organ injury, such as an abnormal amount of protein in the urine, a low number of platelets, abnormal kidney or liver function, pain over the upper abdomen, fluid in the lungs, or severe headache or changes in vision.

**Prenatal Care:** A program of care for a pregnant woman before the birth of her baby.

**Rectum:** The last part of the digestive tract.

**Rh Factor:** A protein that can be present on the surface of red blood cells.

**Rh Immunoglobulin:** A substance given to prevent an Rh-negative person's antibody response to Rh-positive blood cells.

**Sexually Transmitted Infections (STIs):** Infections that are spread by sexual contact, including chlamydia, gonorrhea, human papillomavirus infection, herpes, syphilis, and infection with human immunodeficiency virus (HIV, the cause of acquired immunodeficiency syndrome [AIDS]).

**Sickle Cell Disease:** An inherited disorder in which red blood cells have a crescent shape, causing chronic anemia and episodes of pain. It occurs most

often in African Americans.

**Syphilis:** A sexually transmitted infection that is caused by an organism called *Treponema pallidum*; it may cause major health problems or death in its later stages.

**Tay-Sachs Disease:** An inherited birth defect that causes intellectual disability, blindness, seizures, and death, usually by age 5 years. It most commonly affects people of Eastern European Jewish (Ashkenazi Jews), Cajun, and French Canadian descent, but it can occur in anyone.

**Trimester:** Any of the three 3-month periods into which pregnancy is divided.

**Trisomy:** A condition in which there is an extra chromosome.

**Trisomy 13 (Patau Syndrome):** A chromosomal disorder that causes serious problems with the brain and heart as well as extra fingers and toes, cleft palate and lip, and other defects. Most infants with trisomy 13 die within the first year of life.

**Trisomy 18 (Edwards Syndrome):** A chromosomal disorder that causes severe intellectual disability and serious physical problems such as a small head, heart defects, and deafness. Most infants with trisomy 18 die before birth or within the first month of life.

**Trisomy 21 (Down Syndrome):** A genetic disorder in which abnormal features of the face and body, medical problems such as heart defects, and intellectual disability occur. Many children with Down syndrome live to adulthood.

**Tuberculosis (TB):** A disease caused by bacteria that usually affects the lungs but also can affect other organs in the body. If not treated, it can be fatal.

**Turner Syndrome:** A condition affecting females in which there is a missing or damaged X chromosome. It causes a webbed neck, short height, and heart problems but usually does not cause developmental delays.

**Ultrasound Exam:** A test in which sound waves are used to examine internal structures. During pregnancy, it can be used to examine the fetus.

**Vagina:** A tube-like structure surrounded by muscles leading from the uterus to the outside of the body.

**X Chromosome:** One of two chromosomes that determine a person's sex.