Prenatal Diagnosis Program
Prenatal Screenings and Diagnosis

What is prenatal diagnosis?

Prenatal diagnosis refers to the use of one or more tests to determine if a developing baby has a problem before birth. These problems include chromosomal abnormalities such as Down syndrome as well as many single gene disorders such as cystic fibrosis, Tay-Sachs disease, sickle cell disease or other birth defects. Other problems, concerns or information can also be discovered.

Many steps can be involved in prenatal diagnosis, and not everyone will choose the same path depending on their beliefs and concerns. In general, there are two broad categories of tests available: screening and diagnostic. Screening tests determine the risk of possible complications with the baby. Screening may include a careful family history, blood tests and an ultrasound exam. There is generally no risk to the pregnancy with screening tests. Diagnostic tests refer to the ability to accurately tell whether a fetus has a specific problem or not. Amniocentesis and chorionic villus sampling are commonly used diagnostic tests. These tests are very safe; however a small risk to the pregnancy is involved with diagnostic procedures. The benefits and risks of these options will be discussed in more detail in the next few pages.

Before any diagnostic tests are considered, the genetic counselor will meet with you, explain the benefits and the risks of each option, and help you make a decision with which you and your family are comfortable. The genetic counselor will also ask you and your partner questions concerning family health and medical history. There will be plenty of time to answer all of your questions and concerns.

It is not possible to test for all known birth defects. Therefore, prenatal diagnosis is only available if risk factors are identified because of family history, maternal age, ultrasound findings, ethnic background or other prenatal risks. Prenatal diagnosis may not be an option if a clinical test is not available for a specific condition.
Who should think about prenatal diagnosis?

Prenatal genetic counseling and diagnosis is generally offered to the following people:

- Women who are at increased risk due to ultrasound findings.
- Women who are at increased risk due to positive maternal serum screening (First Screen, Integrated Screen, QUAD screen, triple screen or AFP test).
- Women and men who have had a child with a chromosomal abnormality.
- Women and men who are carriers of a known chromosomal rearrangement.
- Women who are carriers of an X-linked disease such as hemophilia, muscular dystrophy or fragile X syndrome.
- Women and men who are carriers of a single gene disorder that can be detected prenatally.
- Women and men who have had a child with a birth defect of the spinal cord or brain such as spina bifida, an open neural tube defect or anencephaly.
- Women who have been exposed to medications, drugs or other dangerous substances during their current pregnancy.
- Women and men of certain ethnic backgrounds.
- Women who will be 34 or older when their baby is born.
Screening Tests

Screening tests are recommended for all pregnant women. They are safe, noninvasive tools used to gather information about the pregnancy. Examples of screening tests include maternal blood tests and ultrasound. These tests are best when they are combined with each other. Information gathered from screening tests can tell a woman if she has a lower or higher chance of having a baby with specific problems, commonly Down syndrome, Trisomy 18 (both are chromosome abnormalities), a heart defect, or open spina bifida (a birth defect caused by an opening of the spine or the skull). Screening tests can also tell if a pregnancy is at increased risk for fetal growth problems or stillbirth. Screening tests do not tell for sure whether the baby is healthy or not.

Ultrasound

An ultrasound examination uses sound waves to evaluate the developing baby. The results of studies have shown that ultrasound is safe for the developing baby and mother. Using ultrasound, the developing baby is measured to find out how far along the pregnancy is (the gestational age) and to follow growth and development. Most major birth defects are found by ultrasound during the first trimester nuchal translucency scan or the second trimester “level II” ultrasound. (More information on these tests is provided on the next page.) Unfortunately, ultrasound cannot find all birth defects or other problems, and a normal ultrasound cannot guarantee a healthy baby.

The gender of the baby may also be seen on ultrasound or discovered through other tests if there is a risk of a genetic condition that affects one gender more than the other. If we think we can tell the gender of the baby, we would be happy to share this information with you. Ultrasound is not perfect, but is generally very accurate in determining the gender of the baby. If you do not want to know the gender of your baby, we are also happy to keep this a secret.

We will provide you with pictures to take home and share with friends and family.
Three Types of Ultrasound

1. **Nuchal translucency (NT) measurement**
   If you are early in your pregnancy (between 11 weeks one day, and 13 weeks six days), a nuchal translucency (NT) measurement is recommended for all pregnant women. The NT is a normal fluid-filled space at the back of the developing baby’s neck. The NT measurement can be used alone to determine if there is an increased risk for a chromosome abnormality, structural heart defect or other birth defects, however the NT works best when combined with a maternal blood test to provide more accurate information about chromosomal problems, fetal growth problems or stillbirth.

2. **“High resolution” or “Level II” ultrasound**
   Later in pregnancy, typically between 16 and 22 weeks of gestation, a second ultrasound is also recommended for all pregnant women. This is helpful because the developing baby has grown, and overall growth and development is easier to see. The best time for the ultrasound is between 18 and 20 weeks, but it is still very good when done earlier or later than this. You may have heard this called a “high-resolution” or a “Level II” ultrasound. These terms just mean that our ultrasound machines, ultrasound technologists and doctors provide you with the most information possible in ultrasound technology. Measurements will be made to ensure the fetus is growing appropriately. We will also look at the developing baby’s heart, brain and other organs. Following your ultrasound, a specially trained doctor will review the ultrasound findings with you and answer any questions that you may have.

3. **Fetal echocardiogram**
   Fetal echocardiography is a specialized examination focused on the structure of your baby’s heart. Fetal echocardiogram is not recommended to all pregnant women unless the baby is at an increased risk of having a heart defect. A routine pregnancy ultrasound will see limited views of your baby’s heart, but a fetal echocardiogram is a very detailed evaluation by a specialist in fetal echocardiography. Early fetal echocardiograms are performed by certain specialists at 15 weeks of pregnancy. More traditional echocardiograms are performed between 20-22 weeks of pregnancy. Because a fetal echocardiogram is just a specialized ultrasound, it is safe for the mother and the fetus.
Blood screening tests

There are several types of blood screening tests available during pregnancy. First trimester screening includes the First Screen, which screens for Down syndrome and Trisomy 18 between 11.0-13.6 weeks of pregnancy. In addition, we are now offering the Sequential Screen, which screens for Down syndrome, Trisomy 18, and open spina bifida in a step-wise manner during both the first and second trimesters of pregnancy. This screening option is based on the results of a lengthy, multi-center study on evaluation of risk in early pregnancy (known as the FASTER trial) and is only available at select perinatal centers. Second trimester-only screening for Down syndrome, Trisomy 18, and open spina bifida is available with the Quadruple Marker Serum Screen. Each option has specific detection and “screen positive” rates, but all blood screen tests work best when combined with information from the baby’s ultrasound examinations. Your genetic counselor will discuss each option in detail at the time of your appointment.

A blood test showing increased risk to the baby is called ‘screen positive.’ A woman who has a blood test with a ‘screen positive’ result will be offered genetic counseling and diagnostic testing because of the increased chance that her baby might have Down syndrome, Trisomy 18, a heart defect, or open spina bifida. Diagnostic testing benefits and risks are discussed later in this brochure. A ‘screen positive’ result can cause worry. However, approximately 95% of women will be ‘screen negative’ and receive reassurance that the risks to the pregnancy are very low. If your result is ‘screen positive’ you can choose whether you want diagnostic testing to find out if the baby really has any one of these problems.

If a woman is ‘screen-negative,’ diagnostic testing is not recommended. Most women with a screen negative result will not have a baby with Down syndrome, Trisomy 18, a heart defect, or open spina bifida, however, because these tests are not 100% accurate, women who are ‘screen negative’ may undergo a diagnostic test if they desire one for extra reassurance.
Diagnostic Tests

Diagnostic tests can be performed to detect chromosome problems, such as Down syndrome or Trisomy 18. They may test for additional conditions if indicated by family history of other risk factors. Unlike screen tests, there is a small risk to the pregnancy with the diagnostic tests, including miscarriage and bleeding. The option to accept or decline diagnostic testing will always be yours.

Chorionic villus sampling

Chorionic villus sampling (CVS) is typically done between 10 and 13.9 weeks of pregnancy. Most commonly, CVS involves placing a thin plastic catheter (tube) through the opening of the cervix. Sometimes, a thin needle is inserted through the abdomen instead. With either approach, cells are taken from the placenta. Ultrasound is used to help guide the placement of the catheter or the needle.

CVS is typically performed to evaluate the chromosomes inside these cells to diagnose problems like Down syndrome. Other tests may be offered as indicated. Birth defects like spina bifida, which is not caused by chromosomal problems, cannot be detected with CVS.

CVS is a very accurate way to diagnose chromosomal problems. CVS is considered a safe procedure; however, there is a small risk to the pregnancy. There are other differences between the amniocentesis and CVS that the genetic counselor can discuss with you further. The option to accept or decline CVS will always be yours.

Amniocentesis

Amniocentesis is a procedure that uses a thin needle to remove a small amount (about two to three tablespoons) of the amniotic fluid surrounding the developing baby. During the amniocentesis, the developing baby and the amniotic fluid are seen by ultrasonography to help guide the needle placement. Amniocentesis is typically done between weeks 15 and 22, but can be performed later in special circumstances.
Amniotic fluid contains cells shed by the skin of the developing baby. The fluid sample is given immediately to the laboratory, where the cells are allowed to grow. Typically the chromosomes inside the cells are analyzed to diagnose problems like Down syndrome, and chemicals in the amniotic fluid are analyzed to diagnose spina bifida. Other tests may be offered as indicated.

Some Frequently Asked Questions

When will I get the results?
A chromosomal study from either amniocentesis or CVS takes between two and three weeks to complete. The time for results depends on how well the cells grow in the laboratory. A longer time for results does not increase the chance for an abnormal outcome. The results may also take longer if additional tests are indicated. It is very rare for cells not to grow in culture.

Do the results guarantee a normal baby?
Unfortunately, the answer to this question is no. There are no tests available that can guarantee that your baby will be born healthy and well. Prenatal diagnosis is aimed at finding chromosomal problems and certain birth defects. Some birth defects cannot be found using prenatal diagnosis. These can include blindness, deafness, some heart defects and many types of mental retardation. However, the chance that any of these problems would occur is low, and most babies are born healthy. Although prenatal diagnosis cannot rule out every possible birth defect, normal results are reassuring. Testing error, although extremely rare, can happen because the tests involved with prenatal diagnosis are very difficult and complicated.

What happens if a problem is found?
You and your doctor or midwife will be given a full report of the findings from the prenatal testing. If a problem is found, we will give you an idea of the nature of the problem and what it means for the health of the baby. Unfortunately, not all problems found during prenatal diagnosis can be fixed, however some problems can be repaired during the pregnancy or after birth. Additional counseling is provided by a perinatologist (a doctor who is specially trained to take care of the mother and fetus during pregnancy) and the rest of our qualified staff within the Prenatal Diagnosis Program.

What information will you need to know?
Family history is extremely important in prenatal diagnosis. Any information the mother and father of the baby can provide about themselves, their pregnancies, previous deliveries and complications, and family health and medical history will help in identifying risk factors for the pregnancy.
Suggested Sources for Online Information

OHSU Perinatal Center
www.OHSUwomenshealth.com/baby

Prenatal Diagnosis at OHSU
www.OHSUwomenshealth.com/prenatal

Ultrasound
www.ob-ultrasound.net

Amniocentesis and CVS
www.familydoctor.org

Amniocentesis and CVS first screen
www.genzyme.com

Chromosomes and genes
www.medgen.ubc.ca/wrobinson/mosaic/chromosome.htm\nwww.meta-library.net/biogloss/chrom-body.html

Trisomy 13 and 18
www.trisomy.org

Genetic conditions
www.yourgenesyourhealth.org
(Down syndrome, cystic fibrosis, sickle cell disease and other genetic conditions.)

March of Dimes
www.modimes.org
(Amniocentesis, CVS, maternal blood sampling, ultrasound, genetic counseling.)
Who We Are

OHSU Perinatal Center

The Perinatal Center’s multi-disciplinary team of perinatologists, genetic counselors, radiologists, and other highly skilled specialists are here to provide you with the highest, most comprehensive level of health care. Here, outstanding prenatal care is tailored to your individual needs, from preconception to your birth.

Our services

We provide consultations and develop management plans with your primary care physician, but we can also provide comprehensive prenatal and delivery services.

- Pre-pregnancy consultations
- Comprehensive prenatal care during pregnancy and delivery
- Antepartum testing
- Pregnancy consultations
- State-of-the-art ultrasound diagnosis and fetal therapy
- Prenatal diagnosis
- Genetic consultations
- Coordination of care with your physician or certified nurse-midwife

Our locations

The OHSU Perinatal Center offers services on Marquam Hill and South Waterfront. All locations provide free parking for patients. Please ask for precise directions when making your appointment.
OHSU Perinatal Center

From individual screenings to comprehensive care

We provide consultations and develop management plans with your primary care physician, but we can also provide comprehensive prenatal and delivery services.

Find more information online at:
www.OHSUwomenshealth.com/prenatal

Appointments: 503 418-4200

After hours: 503 494-8311

Hours: Monday through Friday, 8 a.m. - 5 p.m.