Of course, a woman may choose not to have any testing at all. If a woman is unsure whether or not she should have CVS, she should speak with her doctor, a genetic counselor or other specialist in genetics.

In addition to the above choices that everyone has, people with additional specific risks may choose to have CVS. Specific risks include:

**Family history of a chromosome abnormality or inherited disease**
If someone in the family, especially a close relative, is known to have a chromosome abnormality or inherited disease, there may be increased risk for another child to be born in the family with the same condition.

**The parents are carriers of an inherited disease**
If the parents of the unborn baby have been found to be carriers of an inherited disease, the pregnancy is at increased risk for the child to be affected. Many inherited diseases today can be diagnosed prenatally.

**Abnormal maternal serum screening test**
Screening tests performed on a sample of blood from a pregnant woman can identify pregnancies at risk for the common chromosome abnormalities, including Down syndrome and open neural tube defects. When the screening results show that a pregnancy has a high risk for one of these problems, CVS for diagnostic testing is recommended.

**How safe is CVS?**
Although CVS is a routine procedure, it does have some risks:

**Miscarriage**
Miscarriage is uncommon, but is the most serious risk of CVS. Some miscarriages would happen anyway, but a few are caused by the procedure. The average risk for miscarriage after CVS is about 1 in 100.

**Bleeding, cramping, or leaking of fluid from the vagina**
These symptoms are fairly common after CVS, but do not usually result in a miscarriage. However, a woman having these symptoms should call her doctor for advice.

**Infection**
CVS is performed under sterile conditions, and therefore infection is rare. However, a woman with fever or any flu-like symptoms after CVS should call her doctor at once.

**Harm to the fetus**
CVS can increase the risk for birth defects, such as undeveloped fingers, toes or limbs. However, when CVS is performed later than the 9th week of pregnancy, limb defects occur in less than 1 in 3,000 pregnancies.

**Repeat testing**
Occasionally, the doctor cannot obtain enough chorionic villi or the testing cannot be performed for some other reason. Neither of these situations indicate that something is necessarily wrong with the pregnancy, but the CVS may have to be repeated (or amniocentesis performed later in the pregnancy) in order to get results.

**Options after testing**
Most women who have CVS have normal test results. The few people who turn out to have abnormal results will need to speak with their doctors or with a genetic specialist about what their results mean and what to do going forward.

Deciding whether or not to have CVS usually depends upon your particular risk factors for certain conditions, your concerns, your family situation and your feelings. Your health care provider can help to answer any additional questions you may have. All decisions about whether or not to have genetic testing are up to you.

**About Integrated Genetics**
Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years.

This brochure is provided by Integrated Genetics as an educational service for physicians and their patients. For more information on our genetic testing and counseling services, please visit our web sites:

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**A procedure that allows a pregnancy to be tested for certain kinds of birth defects**

**Integrated Genetics**

Client Services
800-848-4436
Chorionic Villi Sampling

Chorionic villi sampling (CVS) is a procedure that allows a pregnancy to be tested for certain kinds of birth defects. Since you are considering having a CVS, this brochure will help to answer some of your questions about the procedure and the tests that can be done.

What is CVS?

CVS is the withdrawal of a small amount of chorionic tissue from the developing placenta. CVS is usually performed in a doctor's office or a hospital between the 10th and 12th week of pregnancy, counting from the first day of the last menstrual period.

Before the procedure, the doctor performs an ultrasound or sonogram, which shows a picture of the uterus, the placenta and the fetus on a screen. Depending on the position of the placenta and fetus, the doctor will use one of two approaches to obtain the chorionic villi sample, either through the cervical canal (transcervical) or through the abdomen (transabdominal).

- With the transcervical approach, the vagina is held in an open position with a speculum. Then, a thin tube is gently inserted through the cervix to reach the chorionic tissue (see Image A).
- With the transabdominal approach, the doctor inserts a thin needle through the woman’s abdominal skin into the uterus to reach the chorionic tissue (see Image B).

Using gentle suction, a very small amount of chorionic villi is removed (see drawing). Some women say CVS does not hurt at all, while others feel pressure or cramping during the procedure. Often, people find waiting for the test results is the most difficult part.

Sometimes CVS cannot be performed for reasons such as the presence of an infection, an unusually shaped uterus or an unusual fetal or placental position. If CVS is not possible, another procedure for prenatal diagnosis, called amniocentesis, can usually be done about 4–6 weeks later in the pregnancy.

What tests can be done on chorionic villi?

Different tests can be performed on a chorionic villi sample, depending upon why a particular pregnancy is at risk.

Testing for chromosome abnormalities

Most people who decide to have CVS are at risk for having a baby with a chromosome abnormality. Chromosomes are tiny packages of genetic material, present in every cell of the body, which contain the information needed for a fertilized egg cell to turn into a human being. The amniotic fluid contains cells that have been shed from the fetus. Normally in each human cell there are 46 chromosomes. A missing or an extra chromosome, or a missing or an extra part of a chromosome, causes many changes in the way an unborn baby develops, and almost always leads to serious physical birth defects, mental retardation or both.

The most common chromosome abnormality is Down syndrome, caused by an extra #21 chromosome. People with Down syndrome have a distinct physical appearance, varying degrees of mental retardation and certain birth defects such as congenital heart disease. The most common chromosome abnormalities account for about two-thirds of all the chromosome abnormalities that can be detected by CVS. The uncommon ones usually involve missing or extra parts of chromosomes.

The standard laboratory testing detects over 99% of all chromosome abnormalities. The results of chromosome analysis from CVS are usually available within seven days.

Testing for inherited genetic diseases

A woman may have CVS because her pregnancy is known to be at risk for one of the inherited diseases that can be tested for prenatally. Examples include Tay-Sachs disease, cystic fibrosis and sickle cell disease. Test results are usually available in 1–5 weeks, depending on the test performed.

How accurate is CVS?

Since the chorionic tissue and the fetus originate from the same fertilized cell, they are usually genetically the same. However, in approximately 1 in 100 cases, the sample shows a mixture of normal and abnormal cells. If this happens, a follow-up amniocentesis may be indicated. There is also an estimated 1 in 1,000 chance that, even though the CVS results are normal, the fetus may have a chromosome abnormality. It is also possible that some maternal cells may be present in the sample and may interfere with the results.

What cannot be tested with CVS?

Unlike amniocentesis, CVS cannot be used to test for open neural tube defects, such as spina bifida and anencephaly. These defects, in which the brain and spinal cord have not developed properly, occur in about 1 in every 1,500 births. It is usually recommended that a woman undergoing CVS also have the blood test called the maternal serum alphafetoprotein (MSAFP) screening test at about 16–18 weeks of pregnancy. This test identifies most (but not all) pregnancies at risk for open neural tube defects.

Neither CVS nor amniocentesis is able to test for the kinds of birth defects that do not have a known cause. A few examples are cleft lip, non-specific mental retardation and most heart defects. Every pregnancy has some risk, approximately 3–5%, for these kinds of problems.

Who should consider CVS?

The American College of Obstetricians and Gynecologists recommends that all women, regardless of age, consider the option of prenatal diagnosis (CVS or amniocentesis) for the detection of chromosome abnormalities.

In the past, only older women were offered prenatal diagnosis because the risk for the most common chromosome abnormalities was known to increase with maternal age (see chart below). However, there are now non-invasive screening tests done on a sample of a woman’s blood that can more accurately identify her specific risk for these common chromosome abnormalities. Therefore, a woman may choose one of the following options: to have a blood test to screen for the common chromosome abnormalities followed by CVS if the screening test shows an increased risk; or to have diagnostic testing by CVS first, without screening.

Maternal Age Risks for Chromosome Disorders in Pregnancy

(Numbers are approximate)