

The background of the page is a repeating pattern of stylized chromosomes. Some are white on a teal background, while others are teal on a white background. The chromosomes are depicted as pairs of sister chromatids, some forming an 'X' shape and others as two parallel lines.

**FACTS ABOUT**  
**Amniocentesis & CVS**



KAISER PERMANENTE®

## **PRENATAL DIAGNOSIS FACTS**

Prenatal diagnostic procedures are medical tests done during the early part of pregnancy to detect certain birth defects. Amniocentesis and chorionic villus sampling (CVS) are two different diagnostic procedures. This booklet describes each procedure and the benefits and risks related to the procedures. Learning about the procedures may help you decide if you want to have prenatal diagnostic testing during your pregnancy. It is always your choice whether or not to have one of these tests.

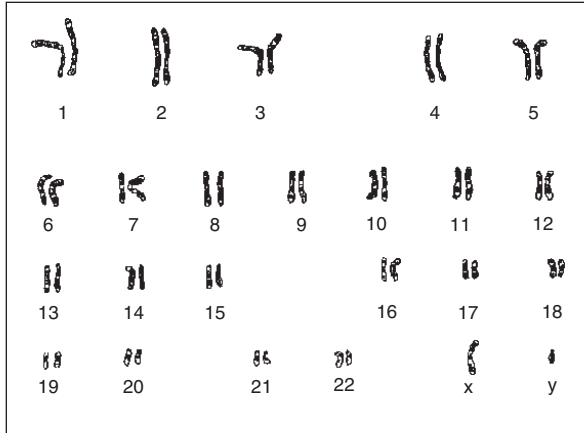
### **I. BIRTH DEFECTS: FREQUENCY AND CAUSES**

Birth defects generally occur in about 3 out of every 100 births. While there are many kinds of birth defects, not all can be diagnosed prior to the baby's birth. Chromosome abnormalities are one type of birth defect that can be diagnosed during pregnancy. Neural tube defects are another type of birth defect that can be found before birth. Amniocentesis and CVS are usually done to identify chromosome abnormalities. Amniocentesis can also help detect neural tube defects.

## II. CHROMOSOME ABNORMALITIES

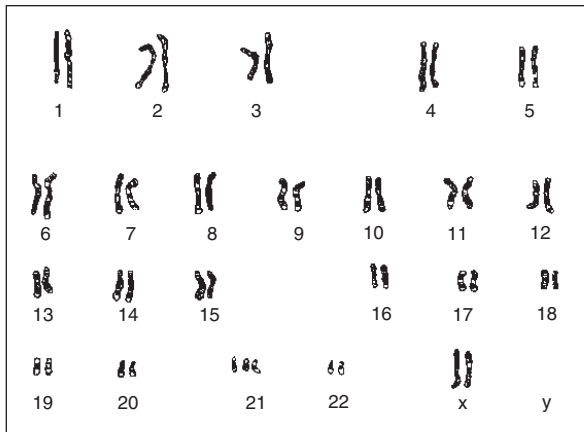
Chromosomes are the packages of genetic information found in every cell of the body. They contain the genes that are responsible for growth and development. Normally, there are 46 chromosomes in every cell.

**CHROMOSOMES OF A  
NORMAL MALE**



Extra or missing chromosomal material almost always causes mental retardation and physical abnormalities. Down syndrome is the most common chromosome disorder. It occurs when there is an extra copy of chromosome #21. Features of Down syndrome include moderate mental retardation and a typical facial appearance. About 40% of individuals with Down syndrome also have heart defects. Other chromosome abnormalities may be more or less severe than Down syndrome.

**CHROMOSOMES OF  
A FEMALE WITH  
DOWN SYNDROME**



### III. CHANCE OF HAVING A BABY WITH A CHROMOSOME ABNORMALITY.

Chromosome abnormalities happen unexpectedly. This type of birth defect usually does not run in families. It is possible for a woman of any age to have a baby with a chromosome abnormality, but the chance is greater as your age increases. The following chart shows the approximate chances of having a baby born with a chromosome abnormality at different ages:

<b>Mother's age at due date</b>	<b>Chance of chromosome disorder</b>	
20	1/525	
25	1/475	
26	1/475	
27	1/455	
28	1/435	
29	1/420	
30	1/400	(0.25%)
31	1/385	
32	1/325	
33	1/285	
34	1/250	
35	1/200	(0.5%)
36	1/165	
37	1/125	
38	1/100	(1%)
39	1/80	
40	1/65	
41	1/50	(2%)
42	1/40	
43	1/30	(3.3%)
44	1/25	
>45	1/20	(5%)

#### **IV. NEURAL TUBE DEFECTS**

Neural tube defects (NTDs), such as anencephaly and spina bifida, are problems in the formation of the skull and spine. NTDs are not related to your age. They happen in about 1 out of 1,000 births. Women who take a vitamin called folic acid before conception and in early pregnancy have a lower chance for NTDs. Most NTDs can be detected prenatally by ultrasound. Screening for NTDs is done by measuring the amount of a protein called alpha-fetoprotein (AFP). AFP is found in the amniotic fluid surrounding the fetus and in every pregnant woman's blood. Follow-up testing is offered if the level of AFP is higher than expected.

#### **V. ULTRASOUND**

Ultrasound is used for many different reasons during pregnancy. It can determine the fetal age, the number of fetuses, and the location of the fetus and the placenta. Some physical birth defects can also be found with ultrasound, but ultrasound cannot diagnose chromosome abnormalities in the fetus. During an amniocentesis or CVS procedure, ultrasound is used to guide the procedure. There is no known risk to the mother or the fetus from ultrasound.

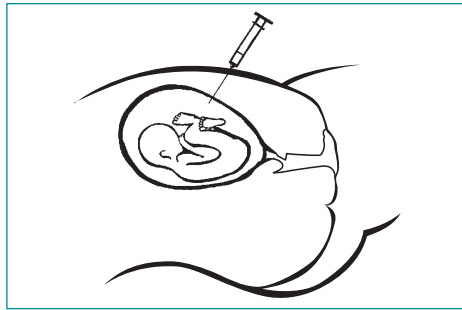


**FETAL ULTRASOUND**

## VI. PROCEDURES

**A. Amniocentesis** is generally performed between 15 and 20 weeks of pregnancy. A thin needle is inserted through the woman's abdomen and into the amniotic sac. A small amount of the amniotic fluid surrounding the fetus is removed. Fetal cells from the amniotic fluid are grown in the laboratory, and the fetal chromosomes are studied. The amount of AFP in the amniotic fluid is also measured to look for NTDs.

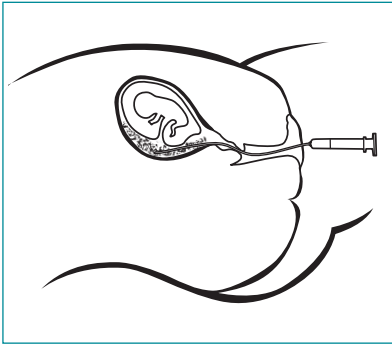
**Early amniocentesis** is the same procedure, but performed before the 15th week of pregnancy (usually during the 14th week of pregnancy).



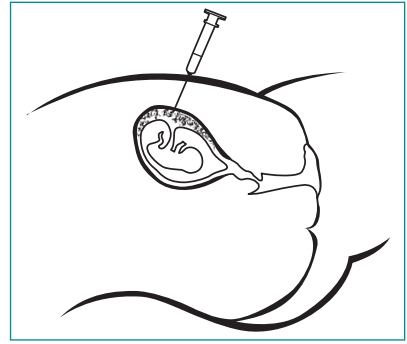
**AMNIOCENTESIS**

**B. Chorionic villus sampling (CVS)** is usually performed between 10 and 14 weeks of pregnancy. Cells from the developing placenta (chorionic villi) are removed and grown in the laboratory. Chromosomes in these cells are then studied. This test is done either transabdominally or transcervically, depending on the location of the placenta and the position of the uterus. The doctor determines which method is best.

- 1. Transabdominal CVS** is done by inserting a thin needle through the woman's abdominal wall into the uterus and removing a small sample of the placental tissue (chorionic villi).
- 2. Transcervical CVS** is done by inserting a thin flexible tube (catheter) through the vagina and cervix into the uterus and removing a small sample of the placental tissue (chorionic villi).



**TRANSCERVICAL CVS**



**TRANSABDOMINAL CVS**

CVS cannot detect neural tube defects (NTDs) since the amniotic fluid containing AFP is not sampled. However, AFP can be measured in a woman's blood between 15 and 20 weeks of pregnancy by a simple blood test.

## **VII. RESULTS**

Results from either the amniocentesis or CVS are ready in about two weeks. These tests are very accurate and detect approximately 99% of chromosome abnormalities. There are rare chromosome abnormalities that cannot be detected by these procedures.

Almost all NTDs can be detected by having an ultrasound (usually between 18 and 21 weeks in pregnancy) AND measuring AFP levels (either by amniocentesis or a blood test).

Chromosome abnormalities and NTDs are not curable. Should an abnormality be detected, the options are either to continue or terminate the pregnancy. In some cases, an early diagnosis of an abnormality can help a woman prepare for the birth of the baby and arrange the specialty care that may be needed at delivery.

## VIII. RISKS OF PROCEDURES

Throughout any pregnancy there is a risk for miscarriage and other complications. However, amniocentesis and CVS procedures pose some additional risks to the pregnancy.

- A. The increased risk of miscarriage due to either the amniocentesis or CVS procedure is less than 1 in 300. Early amniocentesis has a higher risk.
- B. When a woman is carrying two or more babies, the risk for miscarriage due to the amniocentesis or CVS procedure may be somewhat higher. In addition, attempts to obtain a CVS sample from each fetus may not be successful. Early amniocentesis is not routinely done on multiple gestations.
- C. The risk of injury to the fetus is minimal; however,
  - 1. Several years ago, it was thought that CVS may be associated with a risk for fetal limb abnormalities. More recent research has indicated that CVS procedures performed after 10 weeks of pregnancy do not pose an increased risk of limb defects.
  - 2. Studies have indicated that amniocentesis performed *before* 14 weeks of pregnancy is associated with an increased risk for fetal clubfoot. This risk does not appear to be significantly increased when the procedure is performed later. For this reason, early amniocentesis is generally not performed before 14 wks of pregnancy.
- D. Possible complications of either procedure include cramping, abdominal soreness, vaginal spotting, vaginal leakage of fluid, or infection. Women who have transcervical CVS are more likely to have some vaginal spotting.
- E. More than one attempt may be necessary to obtain an adequate amount of either amniotic fluid or chorionic villi (placental tissue). Multiple attempts are more common with CVS procedures.
- F. There is a minimal risk of Rh sensitization in Rh negative women. A Rhogam injection is given to Rh negative women after either procedure to effectively eliminate this risk.



G. A follow-up test may be needed in 2–3% of CVS procedures if cells fail to grow in the laboratory or if the results are unclear. The need for follow-up testing after amniocentesis is uncommon. Follow-up testing may mean a blood test for the pregnant woman or her partner. In some cases, another prenatal procedure is offered. A follow-up amniocentesis is offered in about 2% of CVS tests due to unclear (mosaic) results.

## IX. SCHEDULING AND ADDITIONAL INFORMATION

Please contact your area Genetics department as soon as possible regarding your decision about prenatal diagnostic testing or if you have questions. A genetic counselor can talk with you about these procedures.

<b>If you receive prenatal care at:</b>	<b>Contact</b>
Alameda, Antioch, Deer Valley, Fairfield, Hayward, Livermore, Martinez, Napa, Oakland, Park Shadelands, Pleasanton, Richmond, Vallejo, Walnut Creek	Kaiser Oakland Genetics Department (510) 752-6298
Davis, Elk Grove, Fair Oaks, Folsom, Point West, Rancho Cordova, Roseville, Sacramento, South Sacramento, Vacaville	Kaiser Sacramento Genetics Department (916) 614-4075
Daly City, Novato, Petaluma, Rohnert Park, San Francisco, San Rafael, Santa Rosa	Kaiser San Francisco Genetics Department (415) 833-2998
Campbell, Fremont, Gilroy, Milpitas, Mountain View, Redwood City, Santa Clara, San Jose	Kaiser San Jose Genetics Department (408) 972-3300
Clovis, Fresno, Oakhurst, Selma	Kaiser Fresno (Clovis) Genetics Department (559) 324-5330
Manteca, Modesto, Stockton, Tracy	Kaiser Modesto Genetics Department (866) 916-4075

## SUMMARY OF PRENATAL DIAGNOSTIC PROCEDURES

	<b>AMNIOCENTESIS</b>	<b>CVS</b>
<b>Procedure</b>	Amniotic fluid is removed by needle	Chorionic villi are removed by needle (transabdominal) or by catheter (transcervical), depending on location of placenta
<b>Timing</b>	15–20 weeks. Early amnio is done in the 14th week.	10–14 weeks
<b>Chromosome Abnormalities</b>	Greater than 99% detection rate	Greater than 99% detection rate
<b>Neural Tube Defects (NTDs)</b>	Screens for NTDs  AFP measured in amniotic fluid AND routine ultrasound provides detection rate of more than 95%	Does not screen for NTDs.  AFP blood test between 15 and 20 weeks AND routine ultrasound provides detection rate of more than 95%
<b>Results</b>	About 2 weeks	About 2 weeks
<b>Miscarriage Due To Procedure</b>	Less than 1 in 300 Early amnio has a higher risk	Less than 1 in 300
<b>Vaginal Bleeding</b>	Rare	Spotting may occur after transcervical CVS
<b>Cramping</b>	Occasional, usually mild	Occasional, usually mild
<b>Infection, Fluid Leakage</b>	Uncommon Fluid leakage may be slightly more common after early amniocentesis	Uncommon
<b>Injury to fetus</b>	Extremely rare	Extremely rare
<b>Frequency of Follow-up Testing</b>	Less than 1 in 300	2–3%
<b>Restriction of activities</b>	Limit physical activity for 24 hours after procedure (no sexual intercourse, strenuous exercise)	Limit physical activity for 24 hours after procedure (no sexual intercourse, strenuous exercise)

## NOTES

The information presented here is not intended to diagnose health problems or to take the place of professional medical care. If you have persistent health problems or if you have further questions, please consult your health care provider.