



FACTS ABOUT
Amniocentesis & CVS

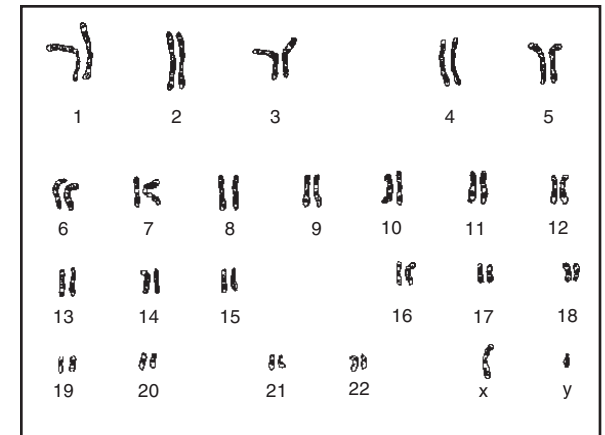


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II. CHROMOSOMAL ABNORMALITIES

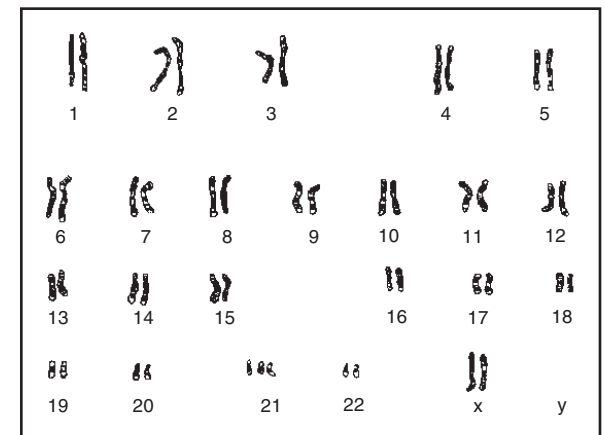
Chromosomes are the packages of genetic information found in every cell of the body. They contain the genes which are responsible for growth and development. Normally there are 46 chromosomes in every cell.

CHROMOSOMES OF A
NORMAL MALE



Extra or missing chromosomal material almost always causes mental retardation and physical abnormalities. Down Syndrome is the most common chromosomal disorder. It occurs when there is an extra #21 chromosome. 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 less common chromosomal abnormalities may be more or less severe than Down Syndrome.

CHROMOSOMES OF
A FEMALE WITH
DOWN SYNDROME



PRENATAL DIAGNOSIS FACTS

Prenatal diagnostic procedures are medical tests which can be performed during the early part of pregnancy to detect certain birth defects. The tests are offered when there is a higher than usual chance for the baby to have these birth defects. Amniocentesis and chorionic villus sampling (CVS) are two procedures currently available to detect specific birth defects. These procedures will be described and the benefits and risks of each will be presented to help you determine if you would like to have prenatal diagnostic testing done. It is your choice whether or not to have a procedure.

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, only a few can be diagnosed prior to the baby's birth. Abnormalities of the chromosomes are one type of birth defect. Neural tube defects which are abnormalities in the development of the brain or spine are another type of birth defect. Amniocentesis and CVS are performed most often in order to identify chromosomal abnormalities. Amniocentesis can also detect neural tube defects.

III. CHANCE OF HAVING A BABY WITH A CHROMOSOMAL ABNORMALITY

Although it is possible for a woman of any age to have a baby with a chromosomal abnormality, the chance is greater as her age increases. The following chart shows the approximate chances of having a baby with a chromosomal abnormality:

Mother's Age At Due Date	Chance of Chromosomal Disorder	
20	1/525	
25	1/475	
30	1/400	(0.25%)
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 (NTDs)

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 a woman's age. They occur in about 1 out of 1,000 births. Taking a vitamin called folic acid before conception and in early pregnancy decreases the risk for NTDs. The majority of NTDs can be detected prenatally by measuring the amount of a protein called alpha-fetoprotein (AFP) in either the amniotic fluid surrounding the fetus or in the pregnant woman's blood.

V. ULTRASOUND

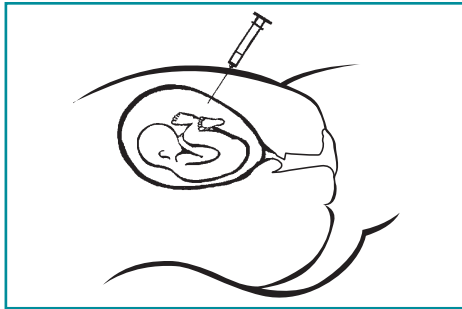
Ultrasound is used to determine the fetal age, the location of the fetus and the placenta, twin pregnancies, and some physical abnormalities of the fetus. There is no known risk to the mother or fetus from ultrasound. Amniocentesis and CVS are done with ultrasound guidance. Ultrasound does not diagnose chromosome abnormalities in the fetus.



FETAL ULTRASOUND

VI. PROCEDURES

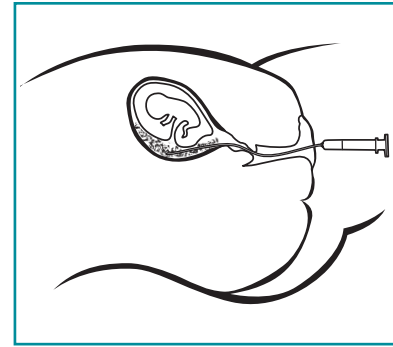
- A. **Amniocentesis** is generally performed between 15 and 20 weeks of pregnancy. A thin needle is inserted through the woman's abdominal wall and into the amniotic sac. A small amount of the amniotic fluid surrounding the fetus is removed. Fetal cells from the amniotic fluid are grown in the laboratory, 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 is generally performed in the 14th week of pregnancy.



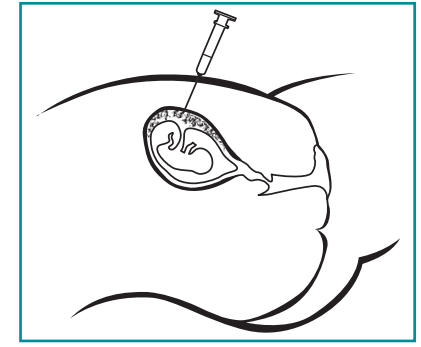
AMNIOCENTESIS

- B. **Chorionic villus sampling (CVS)** is usually performed between 10 and 12 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 transcervically or transabdominally, depending on the location of the placenta. The doctor determines which method is best.

1. **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).
2. **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).



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 the woman's blood between 15 and 20 weeks of pregnancy by a simple blood test. A follow-up ultrasound or amniocentesis is offered if the level of AFP is elevated.

VII. RESULTS

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

Chromosomal abnormalities and NTDs are not curable. Should an abnormality be detected, the options are either to continue or terminate the pregnancy.

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 the amniocentesis procedure is between 1 in 300 and 1 in 500. Early amniocentesis may have a higher risk. The risk of miscarriage due to CVS is less than 1 in 100.
- B. When a woman is carrying 2 or more babies, the risk for miscarriage due to the amniocentesis procedure is somewhat higher. CVS and early amniocentesis are not routinely performed in these pregnancies.
- C. The risk of injury to the fetus is minimal; however,
 1. Some studies have suggested that CVS may be associated with a risk for fetal limb abnormalities. Most studies, however, seem to indicate that procedures performed at 10 weeks of pregnancy or greater pose little if any increased risk.
 2. Some studies have suggested that early 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 after 14 weeks 0 days.
- D. Possible complications of either procedure include cramping, soreness, spotting, vaginal leakage of fluid, or infection. Women who have transcervical CVS are more likely to have some 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 in CVS procedures.
- F. There is a minimal risk of Rh sensitization in Rh negative women. A Rhogam injection is offered to Rh negative women after either procedure to effectively eliminate this risk.

- G. If cells fail to grow in the laboratory or if results are unclear, a follow-up test may be necessary. The need for follow-up procedures after the amniocentesis is uncommon. Follow-up tests are needed more often after CVS.
 1. In 3–5% of CVS cases, a follow-up amniocentesis will be recommended due to unclear results.
 2. In up to 15% of CVS cases, a maternal blood sample will be requested to confirm that the test result reflects fetal chromosomal status, rather than that of the mother.

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 any questions.

If you receive your prenatal care at:	Contact
Alameda, Antioch, Deer Valley, Fairfield, Livermore, Martinez, Napa, Oakland, Park Shadelands, Pleasanton, Richmond, Vallejo, Walnut Creek	Kaiser Oakland Genetics Department (510) 752-6298
Davis, Elk Grove, Fair Oaks, Folsom, Manteca, Modesto, Point West, Rancho Cordova, Roseville, Sacramento, South Sacramento, Stockton, Tracy, 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
Fremont, Gilroy, Hayward, Milpitas, Mountain View, Redwood City, Santa Clara, Santa Teresa (San Jose)	Kaiser San Jose Genetics Department (408) 972-3300
Clovis, Fresno, Selma	Kaiser Fresno Genetics Department (559) 324-5330

SUMMARY OF PRENATAL DIAGNOSTIC PROCEDURES

	AMNIOCENTESIS	CVS
Procedure	Amniotic fluid is removed by needle	Chorionic villi are removed by catheter (transcervical) or by needle (trans-abdominal), depending on location of placenta
Timing	15–20 weeks. Early amnio is done in the 14th week	10–12 weeks
Chromosomal Abnormalities	Greater than 99.8% detection rate	Greater than 99% detection rate
Neural Tube Defects (NTDs)	AFP measured in amniotic fluid, with detection rate of 90–95%	Does not detect NTDs. Maternal AFP blood test offered between 15 and 20 weeks, with detection rate of 80–85%
Results	About 2 weeks	About 2 weeks
Miscarriage Due To Procedure	1 in 300 to 1 in 500 (0.3%–0.2%) Early amnio may have a higher risk	Less than 1 in 100 (1%)
Vaginal Bleeding	Rare	Spotting may occur after transcervical CVS
Cramping	Occasional, usually mild	Occasional, usually mild
Infection, Fluid Leakage	Uncommon Fluid leakage may be slightly more common after early amniocentesis	Uncommon
Injury to Fetus	Extremely rare	Extremely rare
Frequency of Repeat Procedure	<1 in 300 (0.3%)	3–5%
Restriction of Activities	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.