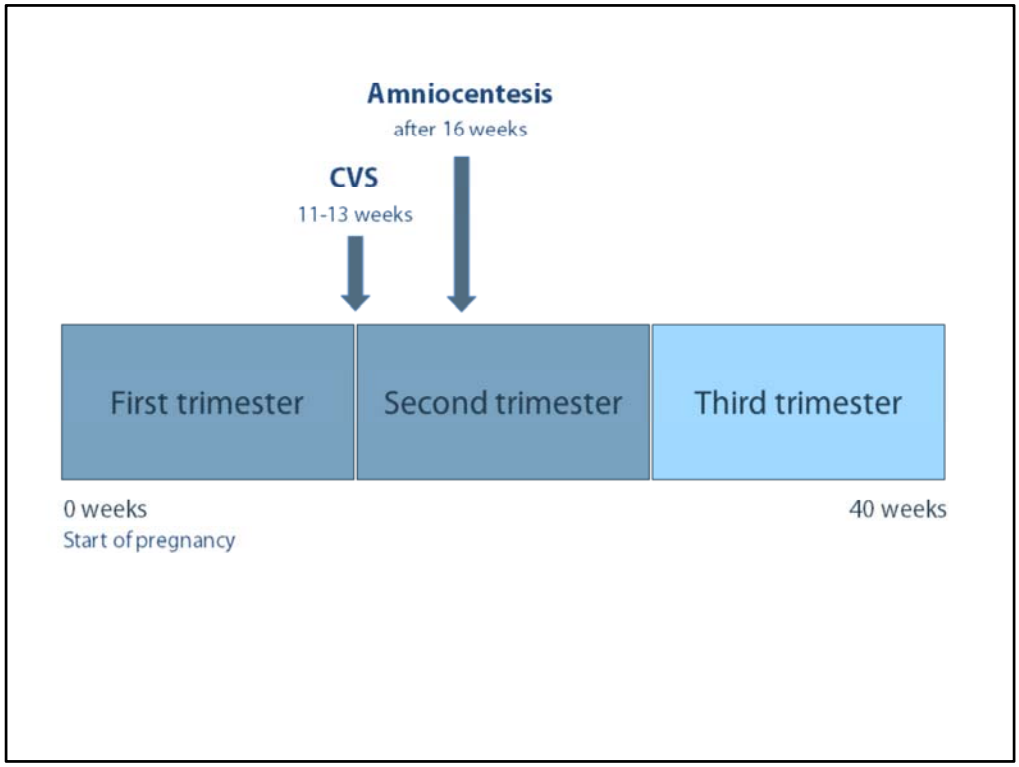
This information sheet will provide more information about reproductive options. This information sheet may be helpful if you or your partner are found to have an increased risk to have a child with a genetic condition.

The words underlined are explained in more detail in the genomics terms sheet.

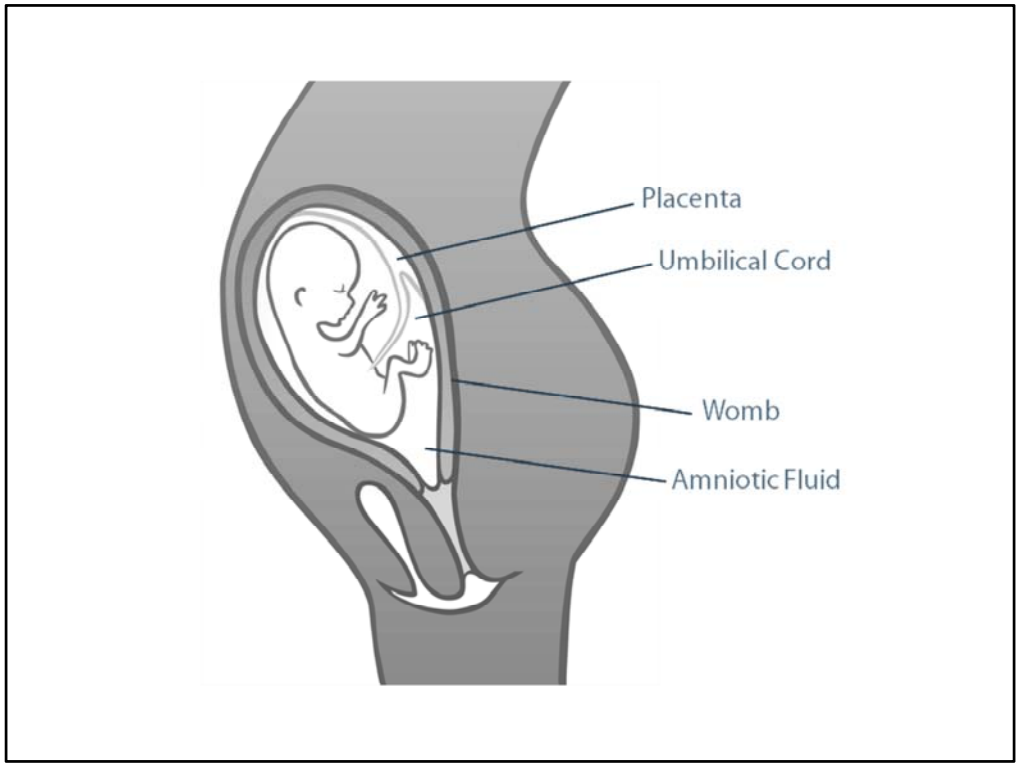
Couples or individuals who are at risk of having a child affected with a genetic condition may choose to pursue reproductive options to increase the likelihood of having a healthy child.

As discussed in the Carrier Screening video, when both parents are carriers of the same recessive genetic condition, there is a 1 in 4, or 25%, chance of having a child affected with the condition.

As discussed in the Personal Genomic Risk video, if one parent has or is a risk for dominant genetic condition, there is a 1 in 2, or 50%, chance of passing the genetic mutation on to a child and having a child affected or at risk to have the same genetic condition.

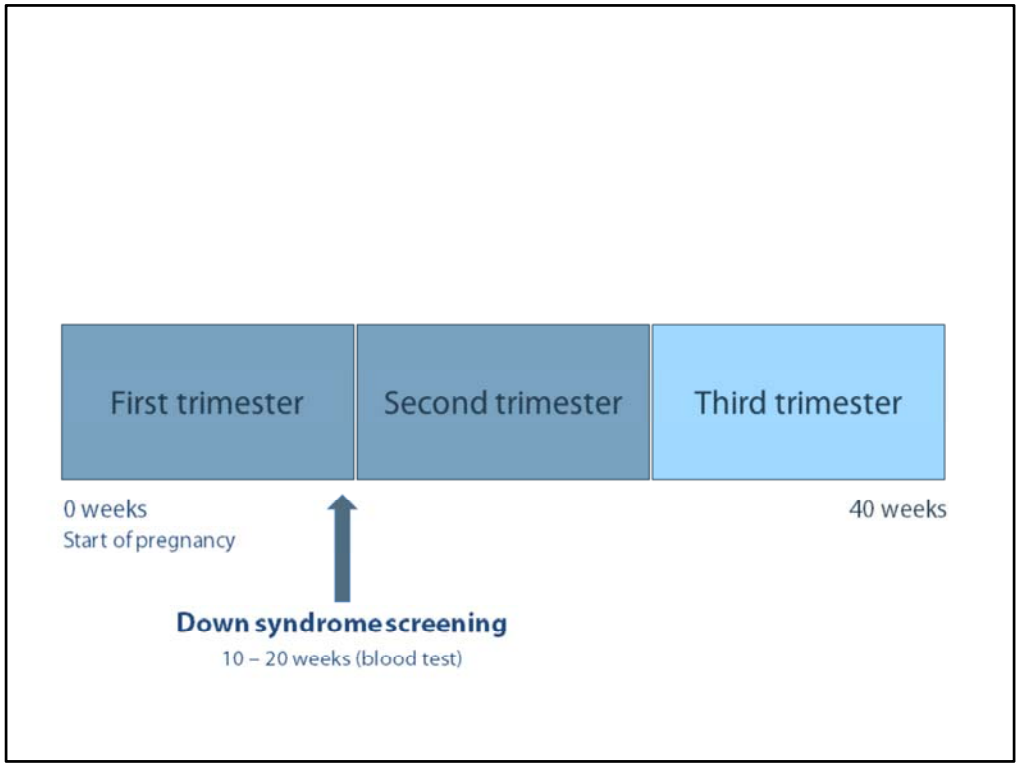


Prenatal genetic tests are available during the first and second trimesters of pregnancy to determine whether the a pregnancy is affected with a the genetic condition. Prenatal genetic testing is done either by chorionic villi sampling, CVS, at 11 to 13 weeks of pregnancy, or by amniocentesis after 16 weeks of pregnancy.

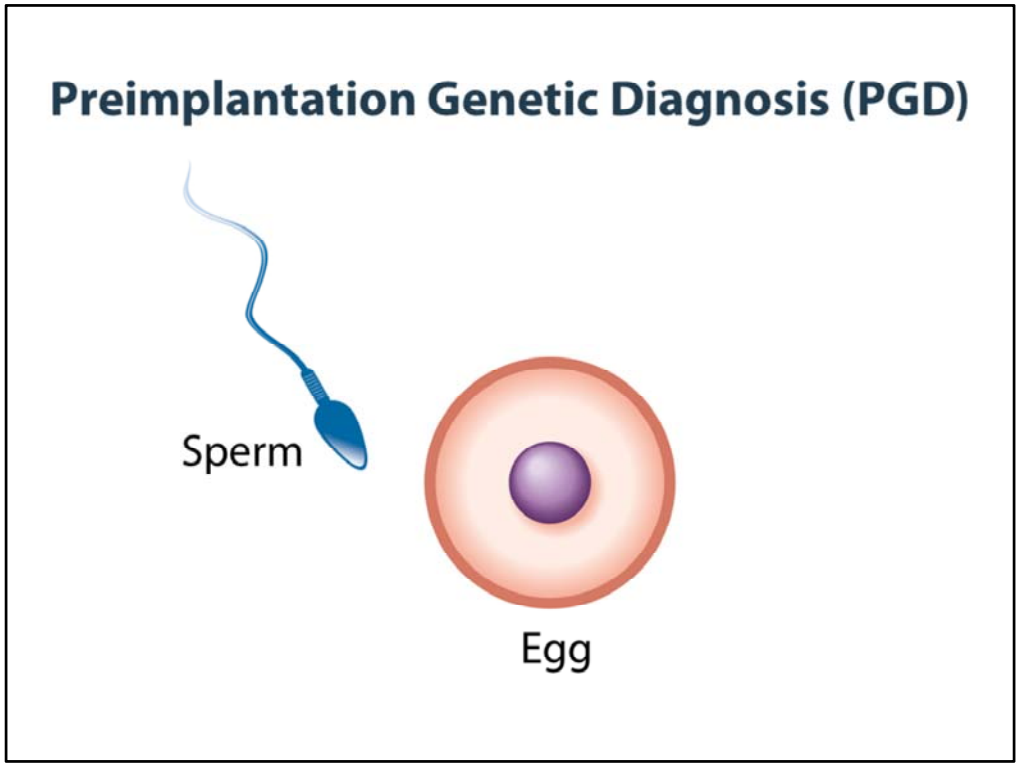


With these procedures, a small number of cells are removed from the placenta or amniotic fluid and tested for the specific genetic condition. With each of these procedures there is a small chance of miscarriage – less than 1%.

If you learn through this testing that the pregnancy is affected with a genetic condition, you can use this information to make a plan for the pregnancy



There are blood tests that are commonly offered during the pregnancy to screen for genetic conditions that are not passed down or inherited from the mother and father, including Down syndrome and other conditions. Presently, genetic conditions inherited from the mother and father cannot be screened for with a blood test and can only be detected by CVS or amniocentesis.



An alternative to prenatal genetic testing is a procedure called preimplantation genetic diagnosis, or PGD. This test occurs prior to pregnancy and requires in vitro fertilization, or IVF - the fertilization of the egg with the sperm outside of the woman. IVF is often used to help couples with infertility have a child but it can also be used to help couples who are at risk to have a child with a genetic condition.

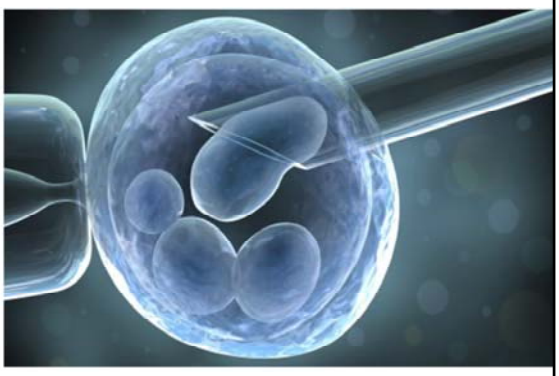
For this procedure, the woman takes hormones to induce the production of eggs. A sperm sample from the man is then used to fertilize the eggs to

make an embryo.

Preimplantation Genetic Diagnosis (PGD)

A procedure that ensures child will not have a specific genetic condition

Embryo



The embryos are then tested for the genetic condition, and only embryos that do not have the condition are put into the woman to start a pregnancy.

There is NO testing that is able to screen for all risk of birth defects

Even when couples choose to pursue reproductive options to prevent their children from having a specific genetic condition, there is still a chance of having a child with a birth defect or another genetic condition. There is no testing that is able to screen for all risk, so, as in all pregnancies, there remains some risk of having a child with a birth defect.

We understand this is complex information. If you'd like to discuss this information with a genetic expert, please click on the option on the website or

Email: **jw2500@columbia.edu**

Call: (212) 305-6987.

The following information sheets may also be helpful

- Introduction to Genetics
- Carrier Screening
- Personal Genomic Risk
- Pharmacogenetics
- Possible Genomic Screening Results