

Genetic Counseling



Your doctor will tell you if your baby is at risk for genetic disorders.

What is genetic counseling?

Genetic counseling helps us understand what things we may pass to our children through genes. Genes tell us about things we may inherit from our relatives.

- Genetic counselors help us to understand what risks could happen during pregnancy.
 - A disease that might be passed on to a baby.
 - A pregnancy that may not be normal.
 - Chances of having affected children.
- Genetic counselors are specially trained to help families understand:
 - Birth defects.
 - How genes control what is passed from one generation to another.
- Genetic counselors provide information and support for people who must make decisions about their pregnancy and childcare.

- A genetic counselor can help you understand:
 - An abnormal test result.
 - A family history of genetic disorders.
 - What it would mean to have a baby with this problem.
 - Counselors also provide emotional support during a possibly upsetting time.
 - Genetic counselors will not make decisions for you.
 - They will help you understand the facts so you can make a decision.

- If you meet with a genetic counselor, the counselor will do the following:
 - Talk with you about your family history.
 - Schedule your appointments for tests.
 - Explain more about the risks you already know about.
 - Explain what role genes play in these conditions.
 - Plan the discussion of test results with your doctor.

- Give you information to help you make your own decisions. For example, on the risk of having a child (or more children) with a genetic disorder.
- Refer you to community resources if you need them.

Who Should Receive Genetic Counseling?

- Your doctor will tell you if your baby is at risk for genetic disorders.
- The doctor will tell you the pros and cons of screening and diagnostic tests that may apply to your pregnancy.
- You may be referred to a genetic counselor.
- Anyone who has questions about genetic problems in their family should think about seeing a genetic counselor.
- This would include:
 - An inherited disorder or birth defect.
 - Women over the age of 35 who are pregnant or plan to become pregnant.
 - Those who already have a child with an inherited disorder.
 - Women who have had 2 or more miscarriages or babies who have died in infancy.
 - Couples who would like more information about genetic defects that occur frequently in their ethnic group.
 - Couples who are 1st cousins or closer.
 - Those who are concerned that their job or lifestyle may pose a risk to a pregnancy, including exposure to chemicals, infections, drugs, or radiation.
 - Pregnant women who have been told that their pregnancy may be at risk for complications or birth defects.
 - If there is a family history of genetic disorders; e.g., sibling or parent.

If a parent has the gene for a dominant condition, there is a 50% chance that the child will receive the same condition.

GENETICS AND INHERITANCE

- Every one of us is born with about 100,000 genes that direct the development of our bodies and how they work.
- These genes decide our hair and eye color, our type of build, and other traits.
- Genes are located on the chromosomes inside each human cell.

- We each receive 23 unpaired chromosomes from each one of our parents.
- When these chromosomes are paired, they create new cells with 46 chromosomes.
- This results in a genetically unique *blueprint* for each of us.

DOMINANT DISORDERS

- If a parent has the gene for a dominant condition, there is a 50% chance the child will receive the same condition.
- Some birth defects caused by dominant inheritance include:
 - Achondroplasia – a problem with bone growth such as dwarfism.
 - Huntington’s disease – a nervous system disorder that gets worse.
 - Marfan’s syndrome – a problem with connective tissue that can affect the heart, blood vessels, lungs, eyes, bones, and ligaments.
 - Neurofibromatosis – a problem of the nervous system.
 - High cholesterol that is inherited.
 - Some forms of glaucoma – a disease of the eye.
 - Polydactyly – extra fingers or toes.

RECESSIVE DISORDERS

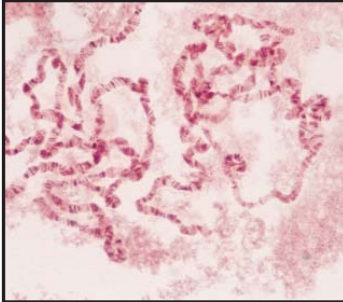
If both parents are carriers of recessive genes, there is a one in four chance that each of their children will inherit the disorder.

- Recessive genes are genes that are only expressed when both parents have the same ones.
 - There is a 1 in 4 chance that each of the children will inherit the disorder.
 - Each child born has a 25% chance of inheriting the disorder.
- If only 1 parent passes on the gene of the disorder:
 - The other parent with the normal gene will stop the condition from being inherited.
 - But, that child will be a carrier of the disorder.

Recessive disorders include:

- Sickle cell anemia – a blood disorder that mainly affects African Americans.
- Tay-Sachs disease – a nervous system disease that causes mental retardation and death, mainly in people of Eastern European Jewish heritage.

- Cystic fibrosis — a problem with the lungs and digestive system, mainly affecting Caucasians.
- Phenylketonuria — a problem that affects the functions of the body, mainly affecting Caucasians.



X-LINKED INHERITANCE

A problem coming from an abnormal gene located on the X chromosome.

- It is called an X-linked or sex-linked disorder.
- A mother who carries the abnormal gene has a 50% chance of passing the disorder to her child.
- The female child who has 2 of these X's will usually show no signs of the disorder.
- The male child who has 1 X and 1 Y chromosome will not have an extra X to use and will have the disorder.
- Some X-linked disorders include:
 - Hemophilia — a disorder where the blood has a problem clotting.
 - Duchenne's muscular dystrophy — a disorder that causes a muscle weakness that gets worse. It can be fatal.
 - Red-green color blindness.
- If you are concerned about a problem that someone in your family has, ask your doctor or genetic counselor about testing.

CHROMOSOME DISORDERS

- A chromosome disorder happens when the egg or sperm is forming.
 - This could be an extra chromosome or a missing chromosome.
 - Chances of a chromosomal disorder happening to the baby increase as the mother gets older.
- Down syndrome is a common chromosome disorder.
 - The child is not normal physically and is mentally retarded.
 - The child may also have heart problems.

MULTIFACTORAL INHERITANCE

- These problems seem to come from interaction of genes and problems in the environment.

- Examples of these types of problems are:
 - Cleft lip and/or palate
 - Clubfoot (talipes)
 - Asthma
 - Diabetes mellitus
 - Certain forms of cancer
 - High blood pressure (hypertension)

Screening Tests

MATERNAL SERUM ALPHA-FETOPROTEIN (MSAFP)

- MSAFP is a blood test that measures the amount of alpha-fetoprotein in the mother's system. The purpose of the test is to find people who may be carriers of the risk of birth defects.



- These people may need more testing.
- Results of this test are most accurate when done between the 16th and 18th week of pregnancy.
- The test can also be done between the 15th and 20th week.
- Results also depend on the mother's weight, whether or not she has diabetes, her race, and if she is carrying more than 1 baby.
- High levels may mean neural tube defects. Low levels may mean chromosome disorders.
- Sometimes abnormal levels are false alarms. If this happens, the doctor may want you to have a second test. An ultrasound may be performed. The doctor may ask you to have an amniocentesis.

ULTRASOUND

- An ultrasound can determine the baby's age.
- It can tell how many babies you are carrying.
- It can tell if the baby has a malformation like spina bifida, heart or kidney problems, and limb defects.
- Not all problems are seen with an ultrasound.

Diagnostic Tests and Procedures

AMNIOCENTESIS

- Amniocentesis is a procedure where a small amount of fluid is taken from the sac that the baby is in.

- The fluid is tested to see if everything is okay.
- It is performed between the 15th and the 18th week of pregnancy.
- It may be done later to determine if the baby's lungs mature.

- Reasons for an amniocentesis include:

- The mother is older.
- To test for Down syndrome or neural tube defects.



- To test for genetic disorders.
- To check for the baby's lung maturity.

- Risks of having an amniocentesis include:

- Lower abdominal cramping.
- Vaginal bleeding or spotting.
- Leakage of fluid from the vagina.
- Low risk that the procedure may cause a miscarriage.

- You should discuss this procedure with your doctor to see if you should have it. This is what to expect if you have the procedure:

- You will be lying down on an exam table.
- An ultrasound will be used to locate the area of fluid in your belly.
- Your belly will be cleaned with an antiseptic.
- An area will be numbed where the doctor will draw the fluid from your belly.
- After the procedure you will need to rest and follow your doctor's instructions.
- Results are usually available in about 2 weeks.

CHORIONIC VILLUS SAMPLING

- This test is done during the 10th week of pregnancy or later.
- A needle is inserted through the belly. The doctor can also use a slim tube through the vagina. A sample of tissue is taken from outside the sac where the baby develops.
- This tissue is analyzed for problems with the baby.
- This procedure is not as good as an amniocentesis.
- The risk of miscarriage appears to be higher.