Family History (Fact Sheet from CDC) ★ Basic Genetic Information ★ Dominant and Recessive Genetic Diseases ★ X-linked Genetic Diseases ★ What is a Chromosome Abnormality? ★ Understanding Genetic Testing ★ Prenatal Diagnosis ★ Birth Defects & Congenital Abnormalities ★ Newborn Screening ★ Genetic Counseling
Family History Is Important for Your Health

adopted from CDC’s Family History Fact sheet

http://www.cdc.gov/genomics/public/famhix/fs.htm

Most of us know that we can reduce our risk of disease by eating a healthy diet, getting enough exercise, and not smoking. But did you know that your family history might be one of the strongest influences on your risk of developing heart disease, stroke, diabetes, or cancer? Even though you cannot change your genetic makeup, knowing your family history can help you reduce your risk of developing health problems.

Family members share their genes, as well as their environment, lifestyles and habits. Everyone can recognize traits that run in their family, such as curly hair, dimples, leanness or athletic ability. Risks for diseases such as asthma, diabetes, cancer, and heart disease also run in families. Everyone’s family history of disease is different. The key features of a family history that may increase risk are:

- Diseases that occur at an earlier age than expected (10 to 20 years before most people get the disease);
- Disease in more than one close relative;
- Disease that does not usually affect a certain gender (for example, breast cancer in a male);
- Certain combinations of diseases within a family (for example, breast and ovarian cancer, or heart disease and diabetes).

If your family has one or more of these features, your family history may hold important clues about your risk for disease. People with a family history of disease may have the most to gain from lifestyle changes and screening tests. You can’t change your genes, but you can change unhealthy behaviors, such as smoking, inactivity, and poor eating habits. In many cases, adopting a healthier lifestyle can reduce your risk for diseases that run in your family. Screening tests (such as mammograms and colorectal cancer screening) can detect diseases like cancers at an early stage when they are most treatable. Screening tests can also detect disease risk factors like high cholesterol and high blood pressure, which can be treated to reduce the chances of getting disease.
**Learning About Your Family History**

To learn about your family history:

- Ask questions,
- Talk at family gatherings, and
- Look at death certificates and family medical records, if possible.

Collect information about your grandparents, parents, aunts and uncles, nieces and nephews, siblings, and children. The type of information to collect includes:

- Major medical conditions and causes of death,
- Age of disease onset and age at death, and
- Ethnic background.

Write down the information and share it with your doctor. Your doctor will:

- Assess your disease risk based on your family history and other risk factors,
- Recommend lifestyle changes to help prevent disease, and
- Prescribe screening tests to detect disease early.

If your doctor notices a pattern of disease in your family, it may be a sign of an inherited form of disease that is passed on from generation to generation. Your doctor may refer you to a specialist who can help determine whether you have an inherited form of disease. Genetic testing may also help determine if you or your family members are at risk. Even with inherited forms of disease, steps can be taken to reduce your risk.

**What If You Have No Family History?**

Even if you don’t have a history of a particular health problem in your family, you could still be at risk. This is because

- Your lifestyle, personal medical history, and other factors influence your chances of getting a disease;
- You may be unaware of disease in some family members;
- You could have family members who died young, before they had a chance to develop chronic conditions such as heart disease, stroke, diabetes, or cancer.

Being aware of your family health history is an important part of a lifelong wellness plan.

**Where You Can Find More Information**

For more information on CDC’s Office of Genomics and Disease Prevention, visit [http://www.cdc.gov/genomics](http://www.cdc.gov/genomics).

The following websites provide additional information on family history:

Basic Genetic Information

• Cells are the body’s building blocks. Inside most cells is a nucleus—the center of the cell. The nucleus contains threadlike structures called chromosomes made up of smaller structures called genes.

• Genes direct the structure and function of your cells which make up all of your body’s organs and tissues.

• Genes are inherited from your parents and determine how you will look.

• Genes come in pairs. One gene comes from your father and one from your mother. This is why you look like your parents.

• Genes also contain instructions for how you age, what diseases you are at risk for or may get as you grow older, or what diseases you might pass down to your children.

• Some genes are stronger, or dominant, and they take over directing your body for that function.

• Some genes are weaker, or recessive, and need the presence of a like partner to become active and make a difference.

• Changes (also called mutations) can sometimes happen in a gene. Changes in a gene may cause cells or organs not to work correctly, leading to a disease. Changes in a gene may also lead to improvement in your body’s ability to cope with disease. Changes in the genes can be inherited from your parents or happen due to the environment you live in—the chemicals you are exposed to, through the air you breathe, the food you eat, or the water you drink.

• Whether the specific set of genes you inherited from your parents—or any changes that occur to them during your lifetime—promote health or produce disease may depend on both environmental and behavioral factors. Proper exercise and nutrition can help delay or prevent disease, while smoking and lack of exercise can increase your chances of disease.

• You can take special tests—genetic tests—to see what specific genes are present in your body. These tests can sometimes tell you what diseases you might have or might develop later, and what diseases you might pass along to your children.

• Newborn babies also take genetic tests to look for diseases that might harm the baby or cause mental retardation if they are not treated immediately. These tests are done just after the baby is born so that treatment can be started immediately to protect the baby from these diseases. If a disease is found, a doctor will help you understand what treatment your baby needs. Sometimes you may be asked to see a genetic counselor.
Dominant and Recessive Genetic Diseases

The basic laws of inheritance are important in order to understand how diseases are passed on in a family. For almost every gene, a person has two copies of each gene—one copy from your father and one copy from your mother. Changes to either copy of the gene or both copies of the gene can result in a wide range of effects. Some changes result in relatively minor or undetectable changes; these types of changes are often called single nucleotide polymorphisms (“snips”) or gene variations.

Other changes in a gene can result in changes to the corresponding protein which can lead to disease. These changes are often known as mutations. Diseases caused by mutations in a single gene are usually inherited in a simple pattern, depending on the location of the gene and whether one or two normal copies of the gene are needed. For certain functions in the body, you need two copies of a gene to work normally. For other functions, only one copy is necessary.

There are two major modes of inheritance for single-gene diseases: recessive and dominant. When a person inherits a mutation in one of the two copies of a gene, disease may develop if both copies are required for normal function. In this case, the mutated gene is dominant and the person develops a genetic disease. Dominantly inherited genetic diseases tend to occur in every generation of a family. Each affected person usually has one affected parent.

If a person inherits a mutation in one copy of a gene, but does not develop a disease, the mutated gene is recessive. For a recessively inherited disease to develop, both copies of the gene must be mutated. This can happen when both the mother and father carry a copy of the mutated gene and pass each copy onto the child, who will then have two copies of the mutated gene. Recessive genetic diseases are typically not seen in every generation of an affected family. The parents of an affected person are generally not affected.
X-linked Genetic Diseases

For genes located on the sex chromosomes (X or Y), the inheritance patterns are slightly different than for genes located on the other chromosomes (1-22). This is due to the fact that females carry two X chromosomes (XX) and males carry a single X and Y chromosome each (XY). Therefore, females carry two copies of each X-linked gene similar to all other genes, but males carry only one copy of X-linked and Y-linked genes.

Since males only have one X chromosome, any mutated gene on the X chromosome will result in disease. But because females have two copies of X-linked genes, diseases caused by mutated genes located on the X chromosome can be inherited in either a dominant or recessive manner. For X-linked dominant diseases, a mutation in one copy of an X-linked gene will result in disease. Families with an X-linked dominant disorder often have both affected males and affected females in each generation.

For X-linked recessive diseases to occur, both copies of the gene must be mutated in order for disease to occur in females. Families with an X-linked recessive disorder often have affected males, but rarely affected females, in each generation.

A striking characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons since they only pass on the Y chromosome. In contrast, affected mothers can pass the mutated X-linked gene to either their son or daughter.

![X-Linked Dominant Inheritance](image1)

![X-Linked Recessive Inheritance](image2)
What is a Chromosome Abnormality?

Almost every cell in our body contains 23 pairs of chromosomes for a total of 46 chromosomes. Half of the chromosomes come from our mother and the other half come from our father. The first 22 pairs (called autosomes) are numbered according to size—the largest chromosome is chromosome 1. The 23rd pair are the sex chromosomes X and Y—females have two X chromosomes and males have an X and Y chromosome each. All of the information that the body needs to work comes from the chromosomes. Each chromosome contains thousands of genes which direct the body’s development, growth, and chemical reactions.

Although almost everyone has a complete set of chromosomes, sometimes pieces of chromosomes can be switched or moved. In general, as long as all of the material is present, the majority of people with rearranged chromosomes do not develop any health problems. However, when sections of or entire chromosomes are missing or duplicated, miscarriage, infant death, or disease usually occurs. For example, an extra copy of chromosome 21 results in Down syndrome (trisomy 21).

Chromosome abnormalities usually happen as a result of an error when cells grow and divide. Errors can occur when eggs or sperm are formed, resulting in either too many chromosomes or not enough chromosomes. Or, errors can occur during the early developmental stages of the fetus, also resulting in an abnormal number of chromosomes. The age of the mother and certain environmental factors can increase the risk of a fetal chromosomal abnormality.

Testing can be performed to examine the chromosomes of the fetus. The two types of testing available are amniocentesis and chorionic villus sampling. In both cases, some cells from the fetus are grown and processed in the laboratory so that the chromosomes can be studied. Pictures of the chromosomes viewed under a microscope are taken and the chromosomes are then arranged by size and paired together. The picture of the arranged chromosomes is known as a karyotype, as seen above. The karyotype is evaluated for size and structure of the chromosomes.
Understanding Genetic Testing

Genetic testing involves examining a person’s DNA, found in blood or other tissues, for some abnormality linked to a disease or condition. DNA is actually a chemical alphabet composed of four units that make up all of the genes, or genetic material, found inside our cells. Genes are important for the body’s normal development and functioning. Each gene is unique due to the order of the four DNA units.

When a mistake happens affecting part or all of the gene, this can result in an abnormal function or change in the body, leading to disease. The mistake can be fairly large or very small, and different types of genetic tests are used to identify the specific gene abnormality.

The most common type of genetic testing is newborn screening. Almost every baby born in the United States has a blood sample tested for abnormal or missing genes or proteins. Early detection can allow the doctor to prescribe drugs or to place the baby on a specific diet in order to prevent or reduce the severity of a disease. Another type of testing, known as carrier testing, can help determine the risk of parents passing on a mutation to their child. Predictive or predispositional genetic testing can determine the risk of a healthy person developing a disease in the future. Finally, genetic tests can be used to look for gene abnormalities in persons suspected of having a genetic disease based on symptoms or family history.

Genetic testing is not always 100 percent accurate. Even when a genetic test positively detects a mutation, the test usually cannot determine when or what symptoms of the disease may show, which symptoms will occur first, how severe the disease will be, or how the disease will progress over time. If a test is negative, an individual may still be at risk for a disease. Therefore, it is important to speak to a health professional such as a genetic counselor to help you understand the benefits and risks of genetic testing and to answer any questions you may have before and after testing.

Genetic counselors are health professionals trained in the areas of medical genetics and counseling. Genetic counselors are trained to help persons as they consider testing, when they receive the results, and in the weeks and months afterward.

When deciding whether or not to have a genetic test for you or your child, several issues should be considered. In addition to the medical issues, genetic testing also raises some social, ethical, and legal issues you should be aware of. Below is a list of some of the issues you should discuss with your physician or genetic counselor:

- What treatments are available for this genetic disease?
- What impact would the genetic test results have on my family?
- What happens if the results are uncertain or inconclusive?
- What are the risks for future pregnancies?
- What is the cost of the test and will my insurance cover it?
- Who will have access to the test results?
- What emotional support services are available?
- Do other family members have a right to know the test results?
- What is the risk of discrimination by my employer or insurer?
Prenatal Diagnosis

Prenatal diagnosis refers to testing performed during a pregnancy. Prenatal diagnosis is helpful for determining the outcome of the pregnancy, planning for possible complications during delivery, planning for problems that may occur in the newborn, deciding whether to continue the pregnancy, and finding conditions that may affect future pregnancies.

A common reason for prenatal diagnosis is the mother’s age. According to professional guidelines, prenatal diagnosis should be offered to women who will be over the age of 35 years at the time of delivery because of an increased risk of having a child with a chromosome abnormality such as Down syndrome. Children with Down syndrome have a distinct facial appearance and mental retardation; however, the severity of the disease can vary greatly from child to child. The disease is caused by an extra copy of chromosome 21 (trisomy 21).

Other possible reasons for prenatal diagnosis include: a previous child with a genetic condition, a fetus known to be at risk for a genetic condition because both parents are mutation carriers, a family history of a genetic condition, a positive prenatal screening test (triple/quadruple/first trimester screen), or abnormal ultrasound findings.

Several types of prenatal diagnosis are available depending how far along the pregnancy is and what type of disorder is being tested. Chorionic villus sampling (CVS) and amniocentesis are two common procedures used to obtain a sample for further testing.

Amniocentesis and chorionic villus sampling are both invasive procedures that carry a risk of miscarriage (less than 0.5% for amniocentesis and about 1% for CVS). Amniocentesis involves removing a sample of amniotic fluid that surrounds the fetus by inserting a syringe through the abdomen. The technique is generally performed at 15 to 20 weeks gestation. In CVS, the fetal cells are removed from an area around the fetus known as the chorion with a syringe inserted through the cervix or abdomen. CVS can be performed as early as nine week’s gestation, but based on safety data, it is typically performed at 10 to 13 weeks’ gestation. This allows the results to be available at an earlier stage of pregnancy. Both amniocentesis and CVS samples contain fetal cells that can be grown in the laboratory for genetic testing.
Birth Defects/Congenital Abnormalities

A birth defect is a problem that happens while a fetus is developing prior to birth. Congenital abnormalities refer to features or conditions that a baby is born with, as opposed to conditions that develop later in life. About 1 in 33 babies is born with a birth defect in the U.S.

A birth defect may cause physical or mental disabilities. It can affect almost any part of the body and can range from mild to severe. Some birth defects can be corrected by surgery or other medical treatments and children can lead normal lives. But some birth defects are very severe and can cause death. Some birth defects are easily detected, such as a club foot or cleft lip, but others such as heart defects or hearing loss may require x-rays and special tests. Not all birth defects can be detected prenatally.

Some of the most common birth defects affect the heart. About 1 in every 200-300 babies is born with a heart defect. Depending on the type and severity of the heart defect, it may be correctable by surgery. Other common birth defects are called “neural tube” defects. These are due to abnormal development of the baby’s spine or brain and affect about 1 in 1,000 babies. These defects are sometimes very severe, causing early death. Birth defects of the lip and the roof of the mouth are also common. They are referred to as cleft lip and cleft palate and affect about 1 in 700-1,000 babies.

Many birth defects are caused by multiple factors—both genetic and environmental. For example, the risk of neural tube defects is increased in families with a history of neural tube defects, but the risk can be reduced with folic acid supplementation during early pregnancy. Uncontrolled medical conditions of the mother, such as diabetes, can also lead to birth defects such as heart defects. Some medicines such as Accutane are also known to cause birth defects.

To learn more about your risk of having a baby with a birth defect, please talk with your doctor or a genetic counselor. In particular, women should consult their doctor before becoming pregnant to begin multi-vitamin supplements containing folic acid, to get help with managing their medical conditions, to decide which medications are safe to take, and to avoid exposure to alcohol, drugs, and smoking.
Newborn Screening

Each year, approximately 98% of all children born in the United States (at least 4 million babies) are tested for a panel of diseases that, when detected and treated early, can lead to significant reduction in disease severity and possibly even prevention of the disease. Between 2,700 and 3,000 newborns test positive for one of these severe disorders each year.

Within 48 hours of a child’s birth, a sample of blood is obtained from a “heel stick.” The blood can be analyzed for more than 50 life-threatening diseases, including phenylketonuria (PKU), sickle cell disease, and hypothyroidism. The sample, called a “blood spot,” is tested at a state public health or other participating laboratory. Each state has its own panel of tests.

Newborn screening programs began in the U.S. in the 1960’s with the work of Dr. Robert Guthrie, who developed a screening test for PKU. PKU is an inherited metabolic disease that is caused by a mutation in an enzyme responsible for metabolism of the amino acid phenylalanine. Children who are identified early can avoid foods with phenylalanine, thereby avoiding buildup of the amino acid which can lead to brain damage and mental retardation. When Dr. Guthrie also introduced a system for collection and transportation of blood samples on filter paper, cost effective wide scale genetic screening became possible.

In general, newborn screening is performed for conditions that, when detected and treated early, can lead to significant reduction in disease severity and possibly even prevention of the disease. The panel of newborn diseases screened for varies from state to state, and decisions for adding or deleting tests involve many complex social, ethical, and political issues. Usually, newborn population screening disorders are selected based on disease prevalence, detectability, treatment availability, outcome, and overall cost effectiveness. It is possible to screen for many disorders at birth and soon more will be possible. The American College of Medical Genetics and the March of Dimes recommend that all babies be screened for a core panel of 29 disorders. About half of the state newborn screening programs have adopted this recommendation.

For specific information on newborn screening in New England, including contact information for each state, see the printable brochure, Newborn Screening Tests: They Could Save Your Baby’s Life, available at www.nergg.org/nbsbrochures.php
Genetic Counseling

Genetic counselors work as part of a health care team, providing information and support to individuals and families affected by or at risk for a genetic disorder. Genetic counselors are trained not only to present complex information about genetic risks, testing, and diagnosis, but also to provide supportive counseling as well as referrals to other sources of information and support.

Common reasons for seeing a genetic counselor include: pregnancy in a woman age 35 or older; family history of a genetic condition; or suspected diagnosis of a genetic condition in a fetus, child, or adult.

A genetic counselor may do any or all of the following during your appointment:

• Ask you questions about your medical/pregnancy history
• Create a picture of your family health history (a pedigree)
• Assess your genetic health risks
• Provide information about the genetic condition(s) affecting you or your family
• Discuss screening and/or preventive measures to address your genetic health risks
• Help you process the significance of your genetic risks or diagnosis
• Assess whether you might benefit from genetic testing
• Help you evaluate the pros and cons of undergoing genetic testing
• Explain genetic test results
• Refer you to relevant resources for further education and support

It is not uncommon for multiple genetic counseling appointments to occur, especially if genetic testing is performed and/or a diagnosis has been made. Depending on the reason for the visit(s), you may also see a geneticist, a physician specializing in genetics.

For more information on genetic counseling, or to find a genetic counselor near you, contact: The National Society of Genetic Counselors, (312) 321-6834, www.nsgc.org