



A Guide to Genetics And Your Health

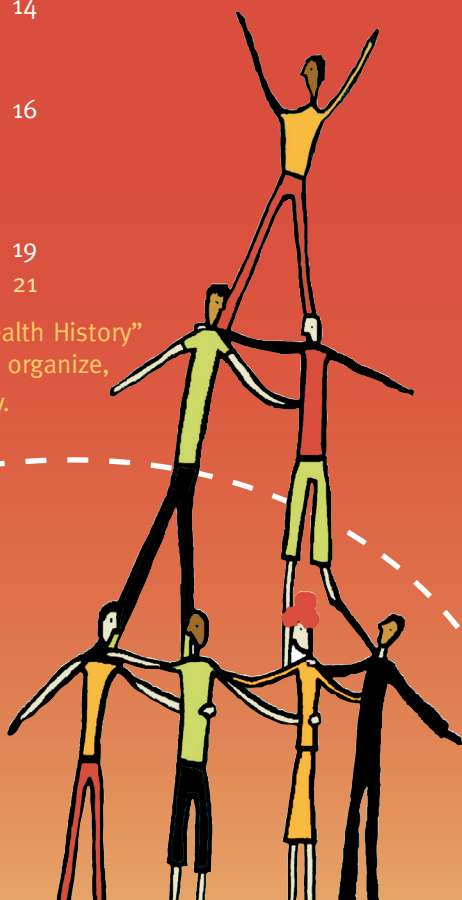
DOES IT RUN IN THE FAMILY?

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Read “Book 1: A Guide to Family Health History”
to learn more about how to collect, organize,
and share your family health history.



Why is genetics important to my family and me?

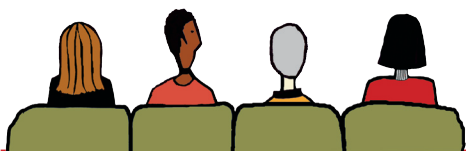


Genetics helps to explain:

- What makes you unique, or one of a kind
- Why family members look alike
- Why some diseases like diabetes or cancer run in families
- How learning your family health history can help you stay healthy
- Why you should bring your family health history to your healthcare provider

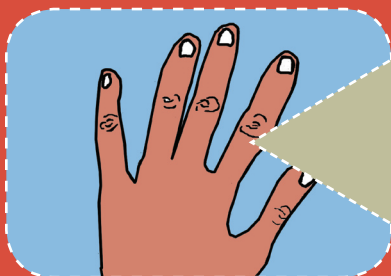
Taking time to learn about health and diseases that run in your family is worth it! It will help you understand your own health and make healthy choices.

What makes me unique?



Every person is unique. Part of what makes you unique is your genes. **Genes are the instructions inside each of your cells.** They control how you look and how your body works. Since everyone has slightly different genes, everyone has a different set of instructions. **Genes are one reason why you are unique!**

1. Hand



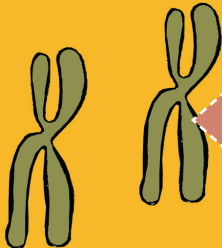
2. Cell



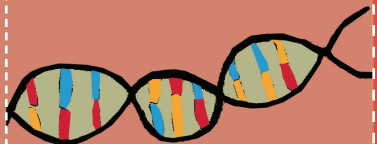
Tell me more about my genes

- A person has two copies of each gene, one from the mother and one from the father.
- Genes carry instructions that tell your cells how to work and grow.
- Cells are the building blocks of the body. Every part of your body is made up of billions of cells working together.
- Genes are arranged in structures called chromosomes. Humans have 23 pairs of chromosomes. Copies of the chromosomes are found in each cell.
- Chromosomes are made up of DNA. DNA is the special code in which the instructions in your genes are written.

3. Chromosomes



4. DNA



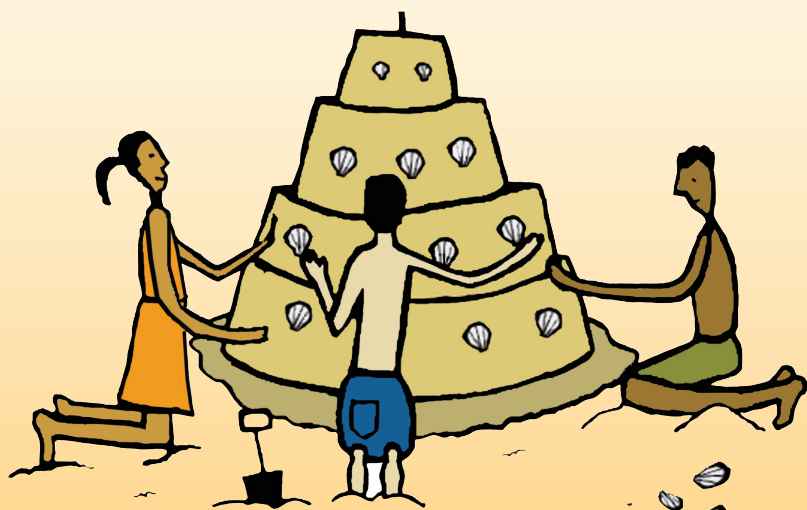


Why do family members have things in common?

Children inherit pairs of genes from their parents. A child gets one set of genes from the father and one set from the mother. These **genes can match up in many ways** to make different combinations. This is why many family members look a lot alike and others don't look like each other at all. Genes can also increase the risk in a family for getting certain health conditions.

Families also share habits, diet, and environment. These influence how healthy we are later in life.

You share a lot with your family—including what can make you sick.



Why do some diseases run in families?

Some diseases are caused when there is a change in the instructions in a gene. This is called a mutation. **Every person has many mutations.** Sometimes these changes have no effect or are even slightly helpful. But sometimes they can cause disease.

Most common diseases are caused by a combination of mutations, lifestyle choices, and your environment. **Even people with similar genes may or may not get an illness** if they make different choices or live in a different environment.

Common Disease: Diabetes

Changes in your genes passed on by your parents may make you more likely to develop type 2 diabetes. If you are active and eat a healthy diet, you may be able to lower your risk.

Visit page 10 to learn about some



Thousands of diseases are caused by a specific change in the DNA of a single gene. **Many of these diseases are rare.**

These conditions usually develop when an individual is born with a mutated gene.

**Single Gene Disorder:
Sickle Cell Anemia**

Sickle cell anemia is caused by a mutation in a single gene passed from each parent.

If a rare disease runs in your family, be sure to write it down. Do not forget to learn about common conditions that affect your family's health.

diseases that run in families.

How can knowing my family health history help me stay healthy?

Your family health history tells you which diseases run in your family. Health problems that develop at a younger age than usual can be a clue that your family has a higher risk. Though you cannot change your genes, you can change your behavior.

Knowing your family health history will help you:

- Identify risks due to shared genes.
- Understand better what lifestyle and environmental factors you share with your family.
- Understand how healthy lifestyle choices can reduce your risk of developing a disease.
- Talk to your family about your health.
- Tell your healthcare provider about the diseases that run in your family.

Remember

1. Share your family health history with your healthcare provider.
2. Ask if you can be screened for a disease that runs in your family.

Why should I take my family health history to my healthcare provider?

Your healthcare provider (doctor, nurse, or physician's assistant) may use your family health history and current health to figure out your risk for developing a disease. Your provider can then help decide which screenings you get and which medicines you might take.

Based on your family health history, a healthcare provider may order a **genetic test** or refer you to a genetic counselor or geneticist. Genetic tests can show if you have a gene change that increases your risk for disease. They can also tell if you have a gene change that you might pass on to your children. Your healthcare provider can help you:

- Understand the results of your tests.
- Learn of any treatments for a disease found by the test.

All newborn babies born in the U.S. and many other countries are tested for certain genetic diseases that may make them sick. This is called **newborn screening**. If the screening test finds a problem, a healthcare provider will help you understand what can be done to help the baby.

Diseases that run in the family

In the rest of this booklet, we give you examples of some common diseases that affect our communities and families. For each disease, we include information under the following headings:

- What is the disease?
- Who is at risk?
- Hints for health



Heart disease

Heart disease is the main cause of death in America in both men and women. There are many types of heart disease. Two of the most common types are coronary artery disease (CAD) and high blood pressure (hypertension).

WHAT IS CORONARY ARTERY DISEASE (CAD)?

- In CAD the arteries that supply blood to the heart muscle can get hard and narrow. The arteries narrow, or get smaller, because plaque and cholesterol build up on the inner walls.
- CAD gets worse over time. As the arteries get smaller, less blood gets to the heart, and less oxygen gets to the heart muscle. Very low levels of oxygen can cause chest pain or a heart attack.
- CAD is the most common cause of heart attacks among Americans.

Who is at risk?

- About 13 million Americans have CAD.
- Everyone has some risk for developing heart disease.
- CAD is caused by a combination of genes, lifestyle choices, and environment.
- For some people, a healthier diet and more activity can change cholesterol level and lower risk.
- Since your genes cannot be changed, some people need medicine to lower their risk of having a heart attack.

Hints for health

- Eat healthy meals.
- Get active and exercise regularly. Obesity increases your risk.
- Take your prescribed medications to control high cholesterol, high blood pressure, and diabetes.
- If you smoke, talk with your healthcare provider about quitting.

For more information, visit www.nhlbi.nih.gov/health/dci and click on “Coronary Artery Disease” or call the American Heart Association at 800-AHA-USA-1 (800-242-8721).

Heart disease continued

WHAT IS HIGH BLOOD PRESSURE?

- Blood pressure is a measure of how hard your heart is working to push the blood through your arteries, the blood vessels leaving your heart.
- There are two numbers in a blood pressure reading. A normal reading is about 120/80 (read as “120 over 80”). The first number is the force your heart uses to pump the blood. The second number is the pressure between heartbeats.
- High blood pressure means that your heart is working too hard. Over time, high blood pressure can cause kidney failure, heart attacks, strokes, and other health problems.

Who is at risk?

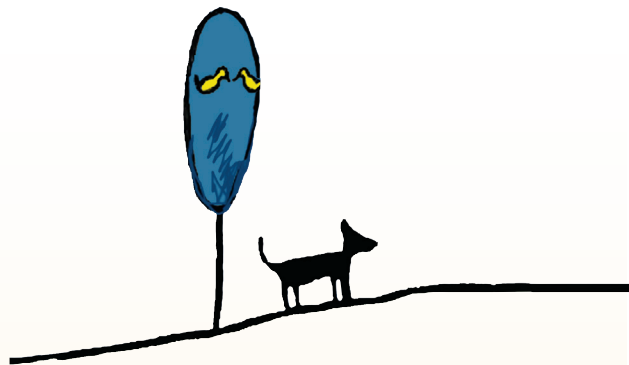
- About one in three adults has high blood pressure. Many do not even know it because there are no clear symptoms.
- A family history of high blood pressure increases your risk for getting it at a younger age.
- Risk increases with age, being overweight, or having a family history of hypertension.

Hints for health

- Eat less salt.
- Maintain a healthy weight.
- Manage your stress.
- Get active and exercise regularly.
- Limit the alcohol you drink.
- Get screening regularly.

For more information, visit www.nhlbi.nih.gov/health/dci and click on “High Blood Pressure” or call the American Heart Association at 800-AHA-USA-1 (800-242-8721).

Heart disease symptoms may not appear until the damage is already done. Talk to your family about heart disease today.



Asthma

WHAT IS ASTHMA?

- Asthma is a lung disease that causes repeated episodes of breathlessness, wheezing, coughing, and chest tightness. The episodes can range from mild to life threatening.
- Asthma episodes are caused by triggers. These are things like dust mites, animal dander, mold, pollen, cold air, exercise, stress, viral colds, allergies, tobacco smoke, and air pollutants.
- Some people have genes that control their response to these asthma triggers.

Who is at risk?

- Asthma affects about one in 10 children and one in 12 adults.
- Asthma is the main reason children end up in the emergency room and miss days of school.
- If you have parents, siblings, or children with asthma or allergies, you are more likely to get it.

Hints for health

- Avoid exposure to triggers.
- Use medication correctly.



For more information, visit www.nhlbi.nih.gov/health/dci and click on “Asthma” or call the American Lung Association at 800-548-8252.

Diabetes (sugar disease)

Diabetes is a serious, chronic disease in which blood sugar levels are above normal. Many people learn about their diabetes after problems develop. According to the American Diabetes Association, one out of three people who have type 2 diabetes do not know that they have the disease.

Symptoms occur when the body fails to change sugar and other food into energy. This happens when the body cannot make or use a hormone called insulin. Serious problems from diabetes can include blindness, kidney failure, and death. Diabetes can be detected early and treatment can prevent or delay these serious health problems. Both genetic and environmental factors such as diet and exercise plays an important role in getting the disease.

WHAT IS TYPE 1 DIABETES?

- Type 1 diabetes usually develops in young children or young adults.
- People with type 1 diabetes stop making their own insulin.

WHAT IS TYPE 2 DIABETES?

- Type 2 diabetes usually develops in people over 30 years of age. In recent years, more young people are getting it due to poor diet.
- Scientists are learning more about the specific genes involved in this type of diabetes.



Who is at risk?

- Diabetes affects about one in 13 people in the United States.
- Five to 10 percent of Americans with diabetes have type 1 diabetes.
- Children or siblings of people with diabetes are more likely to get diabetes.
- Obese people are more likely to get type 2 diabetes.
- Women who had a baby that weighed more than 9 pounds or who had gestational diabetes while pregnant are at risk.

Hints for health

- Eat more fruits and vegetables, less sugar and fat.
- Get active and exercise regularly.
- Lose weight if necessary.

For more information, visit www.ndep.nih.gov or call 800-860-8747.

Cancer

There are many types of cancer. Cancer is caused by the growth and spread of abnormal cells. Though your risk of getting cancer increases as you get older, genetic and environmental factors also cause people to be at a higher risk for certain types of cancer. Some of the most common cancers are breast cancer, lung cancer, and prostate cancer.

WHAT IS BREAST CANCER?

- Breast cancer is a type of cancer that forms in the tissues of the breast, usually the ducts.
- Breast cancer is one of the most common cancers among women. Although it is rare, men can also get breast cancer.
- Although it is rare, men can also get breast cancer.
- Most breast cancer can be treated if found early.

Who is at risk?

- One out of eight American women will develop breast cancer in her lifetime.
- Among Hispanic/Latina women, breast cancer is the most common type of cancer.
- Breast cancer risk is higher if a woman has close blood relatives who have had this disease. Both your mother's and father's family history of breast cancer is important.

Hints for health

- Women should do monthly breast self-exams.
- After age 40, women should get annual mammograms.
- Ask about genetic testing for high-risk families.
- Eat a healthy, balanced diet.
- Get active and exercise regularly.
- Limit the alcohol you drink.

For more information, visit www.cancer.gov/cancertopics and click on "Breast Cancer" or call 800-4-CANCER (800-422-6237).

WHAT IS LUNG CANCER?

- Lung cancer is the uncontrolled growth of abnormal cells in one or both of the lungs.

Who is at risk?

- Lung cancer is the leading cause of cancer death for both men and women.
- About 160,000 people died in the United States from lung cancer in 2007.
- Nearly 87 percent of lung cancer cases in the United States are smoking-related.

Hints for health

- Do not smoke.
- Avoid secondhand smoke.
- Find out about testing for radon and asbestos in your home and at work.

For more information, visit www.cancer.gov/cancertopics and click on “Lung Cancer” or call 800-4-CANCER (800-422-6237).



Cancer continued

WHAT IS PROSTATE CANCER?

- Prostate cancer develops in the male reproductive system. The prostate is a small gland near the bladder.
- Scientists do not yet know what causes prostate cancer.
- Doctors have a test to find out whether a man might have prostate cancer.

Who is at risk?

- Men of all ages can develop prostate cancer. However, more than eight out of 10 cases occur in men over the age of 65.
- Prostate cancer is the most common type of cancer diagnosed in Hispanic/Latino and African American men.
- Having a father or brother with prostate cancer more than doubles a man's risk for getting prostate cancer. The risk goes up with the number of relatives who have it, especially if the relatives were less than 50 years old when they got it.

Hints for health

- Follow a healthy diet.
- Exercise regularly.
- After age 50, have your prostate checked.

For more information, visit www.cancer.gov/cancertopics and click on "Prostate Cancer" or call 800-4-CANCER (800-422-6237).

Single gene disorders

WHAT ARE SINGLE GENE DISORDERS?

- Earlier in this booklet, you read about conditions caused by mutations in a single gene. These conditions are called single gene disorders.
- There are more than 6,000 single gene disorders. Combined, they occur in about 1 in 300 births.
- The symptoms of single gene disorders vary widely, but many of them run in families.
- Collecting your family health history for these conditions is important for diagnosis and management of the condition and for making reproductive choices.

Who is at risk?

- Every person is born with mutations. Most of these mutations will not cause disease on their own, but it is important to identify any that do.
- Depending on which gene is affected, single gene disorders can be passed down even when the mother and father do not show any symptoms.
- Some single gene disorders are identified during a pregnancy or soon after a child is born. Others will not be diagnosed until adulthood.
- The most harmful mutations may lead to a miscarriage or stillbirth. If you have a family history of miscarriages, this may be related to a genetic mutation.

Single gene disorders continued

Hints for health:

- If you have a family history of a single gene disorder, discuss it with your healthcare provider. Your provider may refer you to a specialist.
- Know which newborn screening tests are performed in your state.
- For thousands of conditions, advocacy organizations provide support services, information, and ways to get involved in the discovery of treatment options.

Visit Disease InfoSearch at www.geneticalliance.org to find out more.

For more information on single gene disorders, contact the Genetic and Rare Diseases (GARD) Information Center at gardinfo@nih.gov or 888-205-2311.



Resources

The “Does It Run In the Family?” toolkit includes two pieces that can help you summarize your health information for your provider—the family health portrait and healthcare provider card. You may also hear your healthcare provider call a Family Health Portrait a “pedigree.”

Each family and individual is unique and may have genetic diseases other than the major diseases listed here.

For more information visit:

Disease InfoSearch

www.geneticalliance.org

National Library of Medicine

www.nlm.nih.gov/services/genetics_resources.html

For More, information contact Cygenex, Inc.
info@cygenex.com 888-232-7761



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Genetic Alliance transforms health through genetics. We promote an environment of openness centered on the health of individuals, families, and communities. We bring together diverse stakeholders that create novel partnerships in advocacy; integrate individual, family, and community perspectives to improve health systems; and revolutionize access to information to enable translation of research into services and individualized decision making.



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