

DOES IT RUN IN THE FAMILY?

2

A Guide for Understanding Genetics and Health



ALPHA-1 FOUNDATION

Contents

Why is genetics important to my family and me?	1
What makes me unique?	2
Tell me more about my genes	3
Why do family members have things in common?	4
Why do some diseases run in families?	6
How can family health history help me stay healthy?	8
Why should I bring my family health history to my healthcare provider?	9
Diseases that run in the family	10
Heart disease	11
<i>Coronary artery disease</i>	
<i>High blood pressure</i>	
Asthma	13
Diabetes	14
<i>Type 1</i>	
<i>Type 2</i>	
Cancer	16
<i>Breast</i>	
<i>Lung</i>	
<i>Prostate</i>	
Alpha-1 Antitrypsin Deficiency	19
Resources	21



Why is genetics important to my family and me?



Genetics helps to explain:

- What makes you unique
- Why family members have traits in common
- 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 in many ways. Part of what makes you unique is in your genes. **Genes are the instructions inside each of your cells.** These instructions influence 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



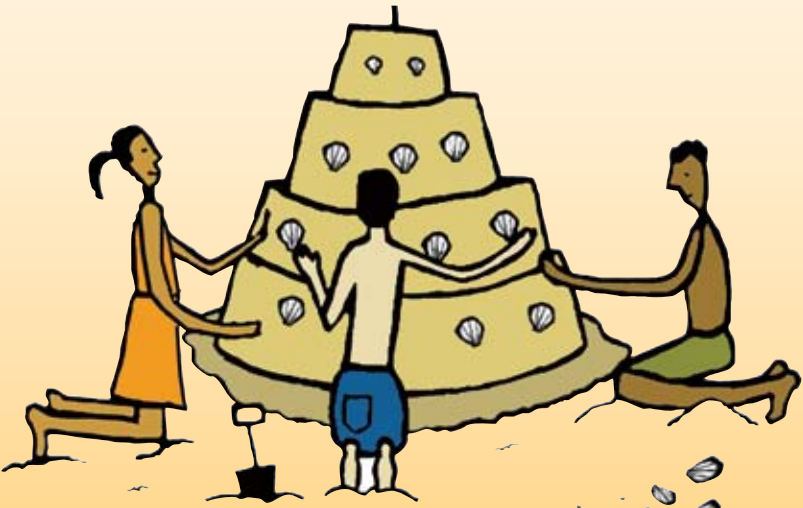


Why do family members have things in common?

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 determine similarities in appearance, but they may also lead to a risk in the family for developing certain health conditions.

Families also share habits, diet, and environment. These experiences might influence how healthy we are later on 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 develop 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.

Alpha 1-Antitrypsin Deficiency

Alpha-1 is caused by a mutation in a single gene passed from each parent.

Even if a rare disease runs in your family, don't forget to learn about more common conditions that affect your family's health.

diseases that run in families.

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

Family health history gives you an idea of which diseases run in your family. Health problems that develop at a younger age than usual can be a clue that your family has higher risk. Though you can't 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.
- Summarize your health information to give to your healthcare provider.

Remember

1. Share your family health history with your healthcare provider.
2. Ask if screening is available for a disease 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. A specific test can show whether you are affected by or at risk for a disease and which mutations you might pass along 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 if not treated. This is called **newborn screening**. If the screening test finds a problem, a healthcare provider or specialist 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 provide you with 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 diseases are the main cause of death in America in both men and women. Two of the most common diseases that involve the heart 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 because of a buildup of plaque or cholesterol on the inner walls.
- CAD gets worse over time. As the heart gets less blood, less oxygen is delivered to the heart muscle. When the heart gets very little oxygen, you can develop 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 genetic background, lifestyle choices, and your environment.
- For some people, a healthier diet and increased activity can change cholesterol level and lower risk.
- Since your genetic background cannot be changed, some people need additional medical assistance such as medication to lower their risk of having a heart attack.

Hints for health

- Eat healthy, nutritious 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.
- There are two numbers in a blood pressure reading. A normal reading is about 120/80 (read as “120 over 80”). The first number measures the force your heart uses to pump the blood. The second number measures 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?

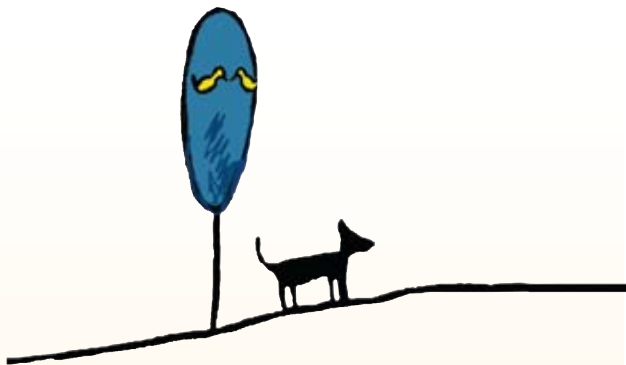
- Approximately one in three adults has high blood pressure. Many do not even know it because there are no clear symptoms.
- A family history of high blood pressure increases your risk for developing it at a younger age.
- Greater risk comes with increasing age, being overweight, or having a family history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

Hints for health

- Decrease the amount of salt you eat.
- 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 many triggers, including dust mites, animal dander, mold, pollen, cold air, exercise, stress, viral colds, allergies, tobacco smoke, and air pollutants.
- Genes control some of an individual's response to these asthma triggers.

Who's at risk?

- Asthma affects approximately one in 10 children and one in 12 adults.
- Asthma affects many children and 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 develop 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. Unfortunately, many people learn about their diabetes after complications develop. According to the American Diabetes Association, one-third of those affected by type 2 diabetes are unaware that they have the disease.

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

WHAT IS TYPE 1 DIABETES?

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

WHAT IS TYPE 2 DIABETES?

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



Who's at risk?

- Diabetes affects approximately one in 14 people in the United States.
- Five to 10 percent of Americans who are diagnosed with diabetes have type 1 diabetes.
- Children or siblings of individuals with diabetes are more likely to develop it themselves.
- Obese people have a greater risk for 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.
- Most breast cancer is treatable if found early.

Who is at risk?

- One out of eight American women will develop breast cancer in their lifetime.
- Among Hispanic/Latina women, breast cancer is the most common type of cancer.
- Breast cancer risk is higher among women whose close blood relatives 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.
- More than 150,000 people died in the United States from lung cancer in 2005.
- 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 is a disease in which cancer develops in the male reproductive system, specifically in a small gland near the bladder called the prostate.
- Scientists do not yet know what causes prostate cancer, but doctors can use certain tests to determine 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 of developing this disease. The risk increases with the number of relatives who have it, especially if the relatives were young (less than 50 years old) when they got it.

Hints for health

- Get regular screenings.
- 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).

Alpha-1 Antitrypsin Deficiency

WHAT IS ALPHA-1 ANTITRYPSIN DEFICIENCY?

- Alpha-1 Antitrypsin Deficiency (Alpha-1) is a hereditary condition that is passed from parents to their children through genes. There are many variations of the disorder that affect lung and liver function differently.
- This condition may result in serious lung disease in adults and/or liver disease in infants, children and adults.
- Alpha-1 occurs when there is a severe lack of a protein in the blood called alpha-1 antitrypsin (AAT) that is mainly produced in the liver.

Who is at risk?

- Everyone with emphysema, chronic obstructive pulmonary disease (COPD), chronic bronchitis or asthma that is not completely reversed after aggressive treatment
- Individuals with bronchiectasis
- Newborns, children and adults with unexplained liver disease
- Individuals with a family history of liver disease
- Blood relatives of a person diagnosed with Alpha-1
- Anyone with panniculitis, a skin disease

Alpha-1 Antitrypsin Deficiency continued

Hints for health:

- Create an exercise program (under medical supervision).
- Create a nutrition program (under medical supervision).
- Think about your health behavior (smoking, alcohol use, and excess weight).
- Avoid risk factors:
 - Stop smoking and avoid secondhand smoke as much as possible.
 - Avoid being around dust and fumes.

For more information, visit www.alphaone.org or call the Alpha-1 Foundation at 877-2-CURE-A1 (877-228-7321).



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

Alpha-1 Foundation

www.alphaone.org

1-877-2-CURE-A1 or 1-877-228-7321



Alpha-1 Foundation
2937 SW 27th Ave, Ste 302
Miami, FL 33133
www.alphaone.org
1-877-2-CURE-A1

The Alpha-1 Foundation is dedicated to providing the leadership and resources that will result in increased research, improved health, worldwide detection, and a cure for Alpha-1 Antitrypsin Deficiency.



4301 Connecticut Ave. NW, Suite 404, Washington, DC 20008-2369
Phone: (202) 966-5557 Fax: (202) 966-8553 info@geneticalliance.org

*Funded in part by a grant (U33 MC06836) from the Maternal and Child Health Bureau,
Health Resources and Services Administration.*