**Sickle cell anemia** is one of a group of inherited disorders known as sickle cell disease. It affects the shape of red blood cells, which carry oxygen to all parts of the body.

Red blood cells are usually round and flexible, so they move easily through blood vessels. In sickle cell anemia, some red blood cells are shaped like sickles or crescent moons. These sickle cells also become rigid and sticky, which can slow or block blood flow.

Sickle cell anemia is caused by a change in the gene that tells the body to make the iron-rich compound in red blood cells called hemoglobin. Hemoglobin enables red blood cells to carry oxygen from the lungs throughout the body. The hemoglobin associated with sickle cell anemia causes red blood cells to become rigid, sticky and misshapen.

For a child to be affected, both mother and father must carry one copy of the sickle cell gene — also known as sickle cell trait — and pass both copies of the altered form to the child.

If only one parent passes the sickle cell gene to the child, that child will have the sickle cell trait. With one typical hemoglobin gene and one altered form of the gene, people with the sickle cell trait make both typical hemoglobin and sickle cell hemoglobin.

**Phenylketonuria** (fen-ul-key-toe-NU-ree-uh), also called PKU, is a rare inherited disorder that causes an amino acid called phenylalanine to build up in the body. PKU is caused by a defect in the gene that helps create the enzyme needed to break down phenylalanine.

Without the enzyme necessary to process phenylalanine, a dangerous buildup can develop when a person with PKU eats foods that contain protein or eats aspartame, an artificial sweetener. This can eventually lead to serious health problems.

For the rest of their lives, people with PKU — babies, children and adults — need to follow a diet that limits phenylalanine, which is found mostly in foods that contain protein.

A defective gene (genetic mutation) causes PKU, which can be mild, moderate or severe. In a person with PKU, this defective gene causes a lack of or deficiency of the enzyme that's needed to process phenylalanine, an amino acid.

A dangerous buildup of phenylalanine can develop when a person with PKU eats protein-rich foods, such as milk, cheese, nuts or meat, and even grains such as bread and pasta, or eats aspartame, an artificial sweetener. This buildup of phenylalanine results in damage to nerve cells in the brain.

**Cystic fibrosis** (CF) is an inherited disorder that causes severe damage to the lungs, digestive system and other organs in the body.

Cystic fibrosis affects the cells that produce mucus, sweat and digestive juices. These secreted fluids are normally thin and slippery. But in people with CF, a defective gene causes the secretions to become sticky and thick. Instead of acting as lubricants, the secretions plug up tubes, ducts and passageways, especially in the lungs and pancreas.

Although cystic fibrosis is progressive and requires daily care, people with CF are usually able to attend school and work. They often have a better quality of life than people with CF had in previous decades.

In cystic fibrosis, a defect (mutation) in a gene — the cystic fibrosis transmembrane conductance regulator (CFTR) gene — changes a protein that regulates the movement of salt in and out of cells. The result is thick, sticky mucus in the respiratory, digestive and reproductive systems, as well as increased salt in sweat.

Many different defects can occur in the gene. The type of gene mutation is associated with the severity of the condition.

Children need to inherit one copy of the gene from each parent in order to have the disease. If children inherit only one copy, they won't develop cystic fibrosis. However, they will be carriers and could pass the gene to their own children

**Tay-Sachs disease** is a rare genetic disorder passed from parents to child. It's caused by the absence of an enzyme that helps break down fatty substances. These fatty substances, called gangliosides, build up to toxic levels in the brain and spinal cord and affect the function of the nerve cells.

In the most common and severe form of Tay-Sachs disease, signs and symptoms start to show up at about 3 to 6 months of age. As the disease progresses, development slows and muscles begin to weaken. Over time, this leads to seizures, vision and hearing loss, paralysis, and other major issues. Children with this form of Tay-Sachs disease typically live only a few years.

Less commonly, some children have the juvenile form of Tay-Sachs disease and may live into their teen years. Rarely, some adults have a late-onset form of Tay-Sachs disease which is often less severe than forms that begin in childhood.

Tay-Sachs disease is a genetic disorder that is passed from parents to their children. It occurs when a child inherits a flaw (mutation) in the HEXA gene from both parents.

The genetic change that causes Tay-Sachs disease results in a deficiency of the enzyme beta-hexosaminidase A. This enzyme is required to break down the fatty substance GM2 ganglioside. The buildup of fatty substances damages nerve cells in the brain and spinal cord. Severity and age of onset of the disease relates to how much enzyme is still produced.

**Thalassemia** (thal-uh-SEE-me-uh) is an inherited blood disorder that causes your body to have less hemoglobin than normal. Hemoglobin enables red blood cells to carry oxygen. Thalassemia can cause anemia, leaving you fatigued.

If you have mild thalassemia, you might not need treatment. But more severe forms might require regular blood transfusions. You can take steps to cope with fatigue, such as choosing a healthy diet and exercising regularly.

Thalassemia is caused by mutations in the DNA of cells that make hemoglobin — the substance in red blood cells that carries oxygen throughout your body. The mutations associated with thalassemia are passed from parents to children.

Hemoglobin molecules are made of chains called alpha and beta chains that can be affected by mutations. In thalassemia, the production of either the alpha or beta chains are reduced, resulting in either alpha-thalassemia or beta-thalassemia.

In alpha-thalassemia, the severity of thalassemia you have depends on the number of gene mutations you inherit from your parents. The more mutated genes, the more severe your thalassemia.

In beta-thalassemia, the severity of thalassemia you have depends on which part of the hemoglobin molecule is affected.

**Huntington's disease** is a rare, inherited disease that causes the progressive breakdown (degeneration) of nerve cells in the brain. Huntington's disease has a broad impact on a person's functional abilities and usually results in movement, thinking (cognitive) and psychiatric disorders.

Huntington's disease symptoms can develop at any time, but they often first appear when people are in their 30s or 40s. If the condition develops before age 20, it's called juvenile Huntington's disease. When Huntington's develops early, symptoms are somewhat different and the disease may progress faster.

Huntington's disease is caused by an inherited defect in a single gene. Huntington's disease is an autosomal dominant disorder, which means that a person needs only one copy of the defective gene to develop the disorder.

With the exception of genes on the sex chromosomes, a person inherits two copies of every gene — one copy from each parent. A parent with a defective gene could pass along the defective copy of the gene or the healthy copy. Each child in the family, therefore, has a 50% chance of inheriting the gene that causes the genetic disorder

Mutations in the [*HTT*](https://medlineplus.gov/genetics/gene/htt/) gene cause Huntington disease. The *HTT* gene provides instructions for making a protein called huntingtin. Although the function of this protein is unclear, it appears to play an important role in nerve cells (neurons) in the brain.

**Pseudo**[**achondroplasia**](https://medlineplus.gov/genetics/condition/achondroplasia/) is an inherited disorder of bone growth. It was once thought to be related to another disorder of bone growth called achondroplasia, but without that disorder's characteristic facial features. More research has demonstrated that pseudoachondroplasia is a separate disorder.

All people with pseudoachondroplasia have short stature. The average height of adult males with this condition is 120 centimeters (3 feet, 11 inches), and the average height of adult females is 116 centimeters (3 feet, 9 inches). Individuals with pseudoachondroplasia are not unusually short at birth; by the age of two, their growth rate falls below the standard growth curve.

Other characteristic features of pseudoachondroplasia include short arms and legs; a waddling walk; joint pain in childhood that progresses to a joint disease known as [osteoarthritis](https://medlineplus.gov/genetics/condition/osteoarthritis/); an unusually large range of joint movement ([hyperextensibility](https://medlineplus.gov/images/PX0001B8_PRESENTATION.jpeg)) in the hands, knees, and ankles; and a limited range of motion at the elbows and hips. Some people with pseudoachondroplasia have legs that turn outward or inward (valgus or varus deformity). Sometimes, one leg turns outward and the other inward, which is called windswept deformity. Some affected individuals have a spine that curves to the side ([scoliosis](https://medlineplus.gov/images/PX0000T0_PRESENTATION.jpeg)) or an abnormally curved lower back ([lordosis](https://medlineplus.gov/images/PX000138_PRESENTATION.jpeg)). People with pseudoachondroplasia have normal facial features, head size, and intelligenc

Mutations in the [*COMP*](https://medlineplus.gov/genetics/gene/comp/) gene cause pseudoachondroplasia. This gene provides instructions for making a protein that is essential for the normal development of cartilage and for its conversion to bone. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears.

The COMP protein is normally found in the spaces between cartilage-forming cells called chondrocytes, where it interacts with other proteins. *COMP* gene mutations result in the production of an abnormal COMP protein that cannot be transported out of the cell. The abnormal protein builds up inside the chondrocyte and ultimately leads to early cell death. Early death of the chondrocytes prevents normal bone growth and causes the short stature and bone abnormalities seen in pseudoachondroplasia.

**Marfan syndrome** is an inherited disorder that affects connective tissue — the fibers that support and anchor your organs and other structures in your body. Marfan syndrome most commonly affects the heart, eyes, blood vessels and skeleton.

People with Marfan syndrome are usually tall and thin with unusually long arms, legs, fingers and toes. The damage caused by Marfan syndrome can be mild or severe. If your aorta — the large blood vessel that carries blood from your heart to the rest of your body — is affected, the condition can become life-threatening.

Treatment usually includes medications to keep your blood pressure low to reduce the strain on your aorta. Regular monitoring to check for damage progression is vital. Many people with Marfan syndrome eventually require preventive surgery to repair the aorta.

**Duchenne muscular dystrophy** is a form of [muscular dystrophy](https://medlineplus.gov/ency/article/001190.htm). It worsens quickly. Other muscular dystrophies (including [Becker muscular dystrophy](https://medlineplus.gov/ency/article/000706.htm)) get worse much more slowly.

Duchenne muscular dystrophy is caused by a defective gene for dystrophin (a [protein](https://medlineplus.gov/ency/article/002467.htm) in the muscles). However, it often occurs in people without a known family history of the condition.

The condition most often affects boys due to the way the disease is inherited. The sons of women who are carriers of the disease (women with a defective gene, but no symptoms themselves) each have a 50% chance of having the disease. The daughters each have a 50% chance of being carriers. Very rarely, a female can be affected by the disease.

**Glucose-6-phosphate dehydrogenase deficiency** is a genetic disorder that affects red blood cells, which carry oxygen from the lungs to tissues throughout the body. In affected individuals, a defect in an enzyme called glucose-6-phosphate dehydrogenase causes red blood cells to break down prematurely. This destruction of red blood cells is called hemolysis.

The most common medical problem associated with glucose-6-phosphate dehydrogenase deficiency is hemolytic anemia, which occurs when red blood cells are destroyed faster than the body can replace them. This type of anemia leads to paleness, yellowing of the skin and whites of the eyes (jaundice), dark urine, fatigue, shortness of breath, and a rapid heart rate. In people with glucose-6-phosphate dehydrogenase deficiency, hemolytic anemia is most often triggered by bacterial or viral infections or by certain drugs

**Hemophilia A** is a hereditary [bleeding disorder](https://medlineplus.gov/ency/article/001304.htm) caused by a lack of blood clotting factor VIII. Without enough factor VIII, the blood cannot clot properly to control bleeding.

**Causes**

When you bleed, a series of reactions take place in the body that helps blood clots form. This process is called the coagulation cascade. It involves special proteins called coagulation, or clotting, factors. You may have a higher chance of excess bleeding if one or more of these factors are missing or are not functioning like they should.

Factor VIII (eight) is one such coagulation factor. Hemophilia A is the result of the body not making enough factor VIII.

Hemophilia A is caused by an inherited [X-linked recessive](https://medlineplus.gov/ency/article/002051.htm) trait, with the defective gene located on the X chromosome. Females have two copies of the X chromosome. So if the factor VIII gene on one chromosome does not work, the gene on the other chromosome can do the job of making enough factor VIII.

Males have only one X chromosome. If the factor VIII gene is missing on a boy's X chromosome, he will have hemophilia A. For this reason, most people with hemophilia A are male

**Hemophilia B** is a hereditary bleeding disorder caused by a lack of blood clotting factor IX. Without enough factor IX, the blood cannot clot properly to control bleeding.

**Causes**

When you bleed, a series of reactions take place in the body that helps blood clots form. This process is called the coagulation cascade. It involves special proteins called coagulation, or clotting factors. You may have a higher chance of excess bleeding if one or more of these factors are missing or are not functioning like they should.

Factor IX (nine) is one such coagulation factor. Hemophilia B is the result of the body not making enough factor IX. Hemophilia B is caused by an inherited [X-linked recessive](https://medlineplus.gov/ency/article/002051.htm) trait, with the defective gene located on the X chromosome.

Females have two copies of the X chromosome. If the factor IX gene on one chromosome does not work, the gene on the other chromosome can do the job of making enough factor IX.

Males have only one X chromosome. If the factor IX gene is missing on a boy's X chromosome, he will have Hemophilia B. For this reason, most people with hemophilia B are male.