**Pedigree analysis**

**Pedigree analysis**, to study the inheritance of genes in humans. Pedigree analysis is also useful when studying any population when progeny data from several generations is limited. Pedigree analysis is also useful when studying species with a long genration time.

A series of symbols are used to represent different aspects of a pedigree. Below are the principal symbols used when drawing a pedigree.



Once phenotypic data is collected from several generations and the pedigree is drawn, careful analysis will allow you to determine whether the trait is dominant or recessive. Here are some rules to follow.

For those traits exhibiting dominant gene action:

* affected individuals have at least one affected parent
* the phenotype generally appears every generation
* two unaffected parents only have unaffected offspring

The following is the pedigree of a trait contolled by dominant gene action.



And for those traits exhibiting recessive gene action:

* unaffected parents can have affected offspring
* affected progeny are both male and female

The following is the pedigree of a trait contolled by recessive gene action.



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**Karyotype**

**Karyotyping** is the process by which photographs of [chromosomes](https://en.wikipedia.org/wiki/Chromosome) are taken in order to determine the chromosome complement of an individual, including the number of chromosomes and any abnormalities. The term is also used for the complete set of chromosomes in a [species](https://en.wikipedia.org/wiki/Species) or in an individual organism[[1]](https://en.wikipedia.org/wiki/Karyotype%22%20%5Cl%20%22cite_note-1)[[2]](https://en.wikipedia.org/wiki/Karyotype#cite_note-White2-2)[[3]](https://en.wikipedia.org/wiki/Karyotype#cite_note-3) and for a test that detects this complement or measures the number.

Karyotypes describe the [chromosome count of an organism](https://en.wikipedia.org/wiki/List_of_organisms_by_chromosome_count) and what these chromosomes look like under a light [microscope](https://en.wikipedia.org/wiki/Microscope). Attention is paid to their length, the position of the [centromeres](https://en.wikipedia.org/wiki/Centromere%22%20%5Co%20%22Centromere), banding pattern, any differences between the [sex chromosomes](https://en.wikipedia.org/wiki/Sex_chromosome), and any other physical characteristics.[[4]](https://en.wikipedia.org/wiki/Karyotype#cite_note-4) The preparation and study of karyotypes is part of [cytogenetics](https://en.wikipedia.org/wiki/Cytogenetics%22%20%5Co%20%22Cytogenetics).



Karyogram of human male using [Giemsa](https://en.wikipedia.org/wiki/Giemsa%22%20%5Co%20%22Giemsa) staining

The study of whole sets of chromosomes is sometimes known as *karyology*. The chromosomes are depicted (by rearranging a photomicrograph) in a standard format known as a *karyogram* or *idiogram*: in pairs, ordered by size and position of centromere for chromosomes of the same size.

The basic number of chromosomes in the [somatic](https://en.wikipedia.org/wiki/Somatic_%28biology%29) cells of an individual or a species is called the *somatic number* and is designated *2n*. In the [germ-line](https://en.wikipedia.org/wiki/Germ-line) (the sex cells) the chromosome number is *n* (humans: n = 23). Thus, in [humans](https://en.wikipedia.org/wiki/Human_genome) 2n = 46.

So, in normal [diploid](https://en.wikipedia.org/wiki/Diploid) organisms, [autosomal](https://en.wikipedia.org/wiki/Autosomal%22%20%5Co%20%22Autosomal) chromosomes are present in two copies. There may, or may not, be [sex chromosomes](https://en.wikipedia.org/wiki/Sex_chromosomes). [Polyploid](https://en.wikipedia.org/wiki/Polyploidy%22%20%5Co%20%22Polyploidy) cells have multiple copies of chromosomes and [haploid](https://en.wikipedia.org/wiki/Ploidy) cells have single copies.

Karyotypes can be used for many purposes; such as to study [chromosomal aberrations](https://en.wikipedia.org/wiki/Chromosomal_aberration), [cellular](https://en.wikipedia.org/wiki/Cell_biology) function, [taxonomic](https://en.wikipedia.org/wiki/Taxonomy_%28biology%29) relationships, [medicine](https://en.wikipedia.org/wiki/Medicine) and to gather information about past [evolutionary](https://en.wikipedia.org/wiki/Evolutionary) events (*[karyosystematics](https://en.wikipedia.org/wiki/Systematics%22%20%5Co%20%22Systematics)*).

## Observations on karyotypes

### Staining

The study of karyotypes is made possible by [staining](https://en.wikipedia.org/wiki/Staining). Usually, a suitable [dye](https://en.wikipedia.org/wiki/Dye), such as [Giemsa](https://en.wikipedia.org/wiki/Giemsa%22%20%5Co%20%22Giemsa),[[19]](https://en.wikipedia.org/wiki/Karyotype#cite_note-19) is applied after [cells](https://en.wikipedia.org/wiki/Cell_%28biology%29) have been arrested during [cell division](https://en.wikipedia.org/wiki/Cell_division) by a solution of [colchicine](https://en.wikipedia.org/wiki/Colchicine%22%20%5Co%20%22Colchicine) usually in [metaphase](https://en.wikipedia.org/wiki/Metaphase) or [prometaphase](https://en.wikipedia.org/wiki/Prometaphase%22%20%5Co%20%22Prometaphase) when most condensed. In order for the [Giemsa](https://en.wikipedia.org/wiki/Giemsa%22%20%5Co%20%22Giemsa) stain to adhere correctly, all chromosomal proteins must be digested and removed. For humans, [white blood cells](https://en.wikipedia.org/wiki/White_blood_cells) are used most frequently because they are easily induced to divide and grow in [tissue culture](https://en.wikipedia.org/wiki/Tissue_culture).[[20]](https://en.wikipedia.org/wiki/Karyotype#cite_note-Gustashaw_K.M_1991-20) Sometimes observations may be made on non-dividing ([interphase](https://en.wikipedia.org/wiki/Interphase%22%20%5Co%20%22Interphase)) cells. The sex of an unborn [fetus](https://en.wikipedia.org/wiki/Fetus%22%20%5Co%20%22Fetus) can be determined by observation of interphase cells (see [amniotic centesis](https://en.wikipedia.org/wiki/Amniocentesis) and [Barr body](https://en.wikipedia.org/wiki/Barr_body)).

### Observations

Six different characteristics of karyotypes are usually observed and compared: Differences in absolute sizes of chromosomes. Chromosomes can vary in absolute size by as much as twenty-fold between genera of the same family. For example, the legumes [*Lotus tenuis*](https://en.wikipedia.org/wiki/Lotus_tenuis) and *[Vicia faba](https://en.wikipedia.org/wiki/Vicia_faba%22%20%5Co%20%22Vicia%20faba)* each have six pairs of chromosomes, yet *V. faba* chromosomes are many times larger. These differences probably reflect different amounts of DNA duplication.

1. Differences in the position of [centromeres](https://en.wikipedia.org/wiki/Centromeres%22%20%5Co%20%22Centromeres). These differences probably came about through [translocations](https://en.wikipedia.org/wiki/Translocations).
2. Differences in relative size of chromosomes. These differences probably arose from segmental interchange of unequal lengths.
3. Differences in basic number of chromosomes. These differences could have resulted from successive unequal translocations which removed all the essential genetic material from a chromosome, permitting its loss without penalty to the organism (the dislocation hypothesis) or through fusion. Humans have one pair fewer chromosomes than the great apes. Human chromosome 2 appears to have resulted from the fusion of two ancestral chromosomes, and many of the genes of those two original chromosomes have been translocated to other chromosomes.
4. Differences in number and position of satellites. Satellites are small bodies attached to a chromosome by a thin thread.
5. Differences in degree and distribution of [heterochromatic](https://en.wikipedia.org/wiki/Heterochromatin) regions. Heterochromatin stains darker than [euchromatin](https://en.wikipedia.org/wiki/Euchromatin%22%20%5Co%20%22Euchromatin). Heterochromatin is packed tighter. Heterochromatin consists mainly of genetically inactive and repetitive DNA sequences as well as containing a larger amount of [Adenine](https://en.wikipedia.org/wiki/Adenine)-[Thymine](https://en.wikipedia.org/wiki/Thymine) pairs. Euchromatin is usually under active transcription and stains much lighter as it has less affinity for the [giemsa](https://en.wikipedia.org/wiki/Giemsa%22%20%5Co%20%22Giemsa) stain.[[22]](https://en.wikipedia.org/wiki/Karyotype#cite_note-ReferenceA-22) Euchromatin regions contain larger amounts of [Guanine](https://en.wikipedia.org/wiki/Guanine)-[Cytosine](https://en.wikipedia.org/wiki/Cytosine) pairs. The staining technique using [giemsa](https://en.wikipedia.org/wiki/Giemsa%22%20%5Co%20%22Giemsa) staining is called [G banding](https://en.wikipedia.org/wiki/G_banding) and therefore produces the typical "G-Bands".A full account of a karyotype may therefore include the number, type, shape and banding of the chromosomes, as well as other cytogenetic information.

Variation is often found:

1. between the sexes,
2. between the [germ-line](https://en.wikipedia.org/wiki/Germ-line) and [soma](https://en.wikipedia.org/wiki/Somatic_%28biology%29) (between [gametes](https://en.wikipedia.org/wiki/Gametes) and the rest of the body),
3. between members of a population ([chromosome polymorphism](https://en.wikipedia.org/wiki/Polymorphism_%28biology%29)),
4. in [geographic specialization](https://en.wikipedia.org/wiki/Allopatric_speciation), and
5. in [mosaics](https://en.wikipedia.org/wiki/Mosaic_%28genetics%29) or otherwise abnormal individuals.
6. Human karyotype

human karyotype (male)

The typical human karyotypes contain 22 pairs of [autosomal](https://en.wikipedia.org/wiki/Autosome%22%20%5Co%20%22Autosome) chromosomes and one pair of [sex chromosomes](https://en.wikipedia.org/wiki/Sex_chromosomes) (allosomes). The most common karyotypes for [females](https://en.wikipedia.org/wiki/Females) contain two [X chromosomes](https://en.wikipedia.org/wiki/X_chromosome) and are denoted 46,XX; [males](https://en.wikipedia.org/wiki/Males) usually have both an X and a [Y chromosome](https://en.wikipedia.org/wiki/Y_chromosome) denoted 46,XY. Approximately 1.7% percent of humans are [intersex](https://en.wikipedia.org/wiki/Intersex), sometimes due to variations in sex chromosomes. Some variations in karyotype, whether to autosomes or allosomes, cause [developmental abnormalities](https://en.wikipedia.org/wiki/Karyotype#Chromosome_abnormalities).

**Inborn errors of metabolism**

Inborn errors of metabolism are rare genetic (inherited) disorders in which the body cannot properly turn food into energy. The disorders are usually caused by defects in specific proteins (enzymes) that help break down (metabolize) parts of food.

A food product that is not broken down into energy can build up in the body and cause a wide range of symptoms. Several inborn errors of metabolism cause developmental delays or other medical problems if they are not controlled.

There are many different types of inborn errors of metabolism.

A few of them are:

* [Fructose intolerance](https://medlineplus.gov/ency/article/000359.htm)
* [Galactosemia](https://medlineplus.gov/ency/article/000366.htm)
* [Maple sugar urine disease](https://medlineplus.gov/ency/article/000373.htm) (MSUD)
* [Phenylketonuria](https://medlineplus.gov/ency/article/001166.htm) (PKU)

[Newborn screening tests](https://medlineplus.gov/ency/article/007257.htm) can identify some of these disorders.

# Galactosemia

Galactosemia is a condition in which the body is unable to use ([metabolize](https://medlineplus.gov/ency/article/002257.htm)) the simple sugar galactose.

## Causes

Galactosemia is an inherited disorder. This means it is passed down through families. If both parents carry a nonworking copy of the gene that can cause galactosemia, each of their children has a 25% (1 in 4) chance of being affected with it.

There are 3 forms of the disease:

* Galactose-1 phosphate uridyl transferase (GALT) deficiency: Classic galactosemia, the most common and most severe form
* Deficiency of galactose kinase (GALK)
* Deficiency of galactose-6-phosphate epimerase (GALE)

People with galactosemia are unable to fully break down the simple sugar galactose. Galactose makes up one half of lactose, the sugar found in milk.

If an infant with galactosemia is given milk, substances made from galactose build up in the infant's system. These substances damage the liver, brain, kidneys, and eyes.

People with galactosemia cannot tolerate any form of milk (human or animal). They must be careful about eating other foods containing galactose.

# Hereditary fructose intolerance

Hereditary fructose intolerance is a disorder in which a person lacks the protein needed to break down fructose. Fructose is a fruit sugar that naturally occurs in the body. Man-made fructose is used as a sweetener in many foods, including baby food and drinks.

## Causes

This condition occurs when the body is missing an enzyme called aldolase B. This substance is needed to break down fructose.

If a person without this substance eats fructose or sucrose (cane or beet sugar, table sugar), complicated chemical changes occur in the body. The body cannot change its stored form of sugar (glycogen) into glucose. As a result, blood sugar falls and dangerous substances build up in the liver.

Hereditary fructose intolerance is inherited, which means it can be passed down through families. If both parents carry a nonworking copy of the aldolase B gene, each of their children has a 25% (1 in 4) chance of being affected.

# Phenylketonuria

Phenylketonuria (PKU) is a rare condition in which a baby is born without the ability to properly break down an amino acid called phenylalanine.

## Causes

Phenylketonuria (PKU) is inherited, which means it is passed down through families. Both parents must pass on a nonworking copy of the gene in order for a baby to have the condition. When this is the case, their children have a 1 in 4 chance of being affected.

Babies with PKU are missing an enzyme called phenylalanine hydroxylase. It is needed to break down the essential amino acid phenylalanine. Phenylalanine is found in foods that contain protein.

Without the enzyme, levels of phenylalanine build up in the body. This buildup can harm the central nervous system and cause brain damage.

# Maple syrup urine disease

Maple syrup urine disease (MSUD) is a disorder in which the body cannot break down certain parts of proteins. The urine of people with this condition can smell like maple syrup.

## Causes

Maple syrup urine disease (MSUD) is inherited, which means it is passed down through families. It is caused by a defect in 1 of 3 genes. People with this condition cannot break down the [amino acids](https://medlineplus.gov/ency/article/002222.htm) leucine, isoleucine, and valine. This leads to a buildup of these chemicals in the blood.

In the most severe form, MSUD can damage the brain during times of physical stress (such as infection, fever, or not eating for a long time).

Some types of MSUD are mild or come and go. Even in the mildest form, repeated periods of physical stress can cause [mental disability](https://medlineplus.gov/ency/article/001523.htm) and high levels of leucine to build up.

# Genetic Diseases

A genetic disease is any disease caused by an abnormality in the genetic [**makeup**](https://www.medicinenet.com/beauty_quiz/quiz.htm) of an individual. The genetic abnormality can range from minuscule to major -- from a discrete mutation in a single base in the DNA of a single gene to a gross chromosomal abnormality involving the addition or subtraction of an entire chromosome or set of chromosomes. Some people inherit genetic disorders from the parents, while acquired changes or mutations in a preexisting gene or group of genes cause other genetic diseases. Genetic mutations can occur either randomly or due to some environmental exposure.

There are a number of different types of genetic disorders (inherited) and include:

1. Single gene inheritance
2. Multifactorial inheritance
3. Chromosome abnormalities

### single gene inheritance disorders

Single gene inheritance is also called Mendelian or monogenetic inheritance. Changes or mutations that occur in the DNA sequence of a single gene cause this type of inheritance. There are thousands of known single-gene disorders. These disorders are known as monogenetic disorders (disorders of a single gene).

Single-gene disorders have different patterns of genetic inheritance, including

* autosomal dominant inheritance, in which only one copy of a defective gene (from either parent) is necessary to cause the condition;
* autosomal recessive inheritance, in which two copies of a defective gene (one from each parent) are necessary to cause the condition; and
* X-linked inheritance, in which the defective gene is present on the female, or X-chromosome. X-linked inheritance may be dominant or recessive.

Some examples of single-gene disorders include

1. [**cystic fibrosis**](https://www.medicinenet.com/cystic_fibrosis/article.htm),
2. alpha- and beta-thalassemias,
3. [**sickle cell anemia**](https://www.medicinenet.com/sickle_cell/article.htm) ([**sickle cell disease**](https://www.medicinenet.com/sickle_cell/article.htm)),
4. [**Marfan syndrome**](https://www.medicinenet.com/marfan_syndrome/article.htm),
5. [**fragile X syndrome**](https://www.medicinenet.com/fragile_x_syndrome/article.htm),
6. Huntington's disease, and
7. [**hemochromatosis**](https://www.medicinenet.com/iron_overload/article.htm).

### Sickle cell anemia (disease, SCD)



* Sickle cell **[anemia](https://www.medicinenet.com/anemia/article.htm)** (SCD) is an [**inherited**](https://www.medicinenet.com/genetic_disease/article.htm) disorder of the **[hemoglobin](https://www.medicinenet.com/hemoglobin/article.htm)** in blood.
* Sickle cell **[anemia](https://www.medicinenet.com/anemia/symptoms.htm)** requires the inheritance of two sickle cell genes.
* Sickle cell trait, which is the inheritance of one sickle gene, almost never causes problems.
* Virtually all of the major symptoms of sickle cell anemia are the direct result of the abnormally shaped sickled [**red blood cells**](https://www.medicinenet.com/complete_blood_count/article.htm) blocking the flow of blood.
* The current treatment of sickle cell anemia is directed primarily toward managing the individual features of the illness as they occur.

Sickle cell anemia (sickle cell disease) is a disorder of the blood caused by an inherited abnormal hemoglobin (the oxygen-carrying protein within the red blood cells). The abnormal hemoglobin causes distorted (sickled appearing under a microscope) red blood cells. The sickled red blood cells are fragile and prone to rupture. When the number of red blood cells decreases from rupture (**[hemolysis](https://www.medicinenet.com/hemolysis/symptoms.htm)**), anemia is the result. This condition is referred to as sickle cell anemia. The irregular sickled cells can also block blood vessels causing tissue and organ damage and [**pain**](https://www.medicinenet.com/pain_management/article.htm).

Sickle cell anemia is one of the most common inherited blood anemias. The disease primarily affects Africans and African Americans. It is estimated that in the United States, some 90,000 to 100,000 Americans are afflicted with sickle cell anemia. Overall, current estimates are that one in 500 U.S. African American births is affected with sickle cell anemia.

Virtually all of the major symptoms of sickle cell anemia are the direct result of the abnormally shaped, sickled red blood cells blocking the flow of blood that circulates through the tissues of the body. The tissues with impaired circulation suffer damage from lack of oxygen. Damage to tissues and organs of the body can cause severe disability in patients with sickle cell anemia. The patients endure episodes of intermittent "crises" of variable frequency and severity, depending on the degree of organ involvement.

The major features and symptoms of sickle cell anemia include:

1. [**Fatigue**](https://www.medicinenet.com/fatigue/article.htm) and anemia
2. [**Pain**](https://www.medicinenet.com/pain_quiz/quiz.htm) crises
3. Dactylitis (swelling and inflammation of the hands and/or [**feet**](https://www.medicinenet.com/feet_facts_quiz/quiz.htm)) and [**arthritis**](https://www.medicinenet.com/arthritis/article.htm)
4. [**Bacterial infections**](https://www.medicinenet.com/bacterial_infections_101_pictures_slideshow/article.htm)
5. Sudden pooling of blood in the spleen and [**liver**](https://www.medicinenet.com/liver_anatomy_and_function/article.htm) [**congestion**](https://www.medicinenet.com/treating_congestion/article.htm)
6. Lung and [**heart**](https://www.medicinenet.com/heart_how_the_heart_works/article.htm) injury
7. Leg ulcers
8. [**Aseptic necrosis**](https://www.medicinenet.com/aseptic_necrosis/article.htm) and bone infarcts (death of portions of bone)
9. Eye damage

Some features of sickle cell anemia that can occur at any age include:

1. [**Fatigue**](https://www.medicinenet.com/causes_of_fatigue_pictures_slideshow/article.htm)
2. Anemia
3. Pain crises
4. Bone infarcts

Many features typically occur in certain age groups.

Infants with sickle cell anemia do not develop symptoms in the first few months of life because the hemoglobin produced by the developing fetus (fetal hemoglobin) protects the red blood cells from sickling. This fetal hemoglobin is absent in the red blood cells that are produced after birth so that by 5 months of age, the sickling of the red blood cells is prominent and symptoms begin.

Infants and younger children can suffer signs and symptoms of;

1. [**fever**](https://www.medicinenet.com/aches_pain_fever/article.htm),
2. [**abdominal pain**](https://www.medicinenet.com/abdominal_pain_causes_remedies_treatment/article.htm),
3. pneumococcal bacterial infections,
4. painful swellings of the hands and feet (dactylitis), and
5. splenic sequestration.

Adolescents (preteens and [**teens**](https://www.medicinenet.com/teenagers/article.htm)) and young adults more commonly develop:

1. Leg ulcers
2. Aseptic necrosis
3. Eye damage

Symptoms in adult typically are intermittent pain episodes due to injury of bone, muscle, or internal organs.

**Huntington's disease**

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.

Medications are available to help manage the symptoms of Huntington's disease. But treatments can't prevent the physical, mental and behavioral decline associated with the condition.

### Products & Services

* [Book: Mayo Clinic Family Health Book, 5th Edition](https://order.store.mayoclinic.com/books/gnweb43?utm_source=MC-DotOrg-PS&utm_medium=Link&utm_campaign=FamilyHealth-Book&utm_content=FHB)

## Symptoms

Huntington's disease usually causes movement, cognitive and psychiatric disorders with a wide spectrum of signs and symptoms. Which symptoms appear first varies greatly from person to person. Some symptoms appear more dominant or have a greater effect on functional ability, but that can change throughout the course of the disease.

### Movement disorders

The movement disorders associated with Huntington's disease can include both involuntary movement problems and impairments in voluntary movements, such as:

* Involuntary jerking or writhing movements (chorea)
* Muscle problems, such as rigidity or muscle contracture (dystonia)
* Slow or abnormal eye movements
* Impaired gait, posture and balance
* Difficulty with speech or swallowing

Impairments in voluntary movements — rather than the involuntary movements — may have a greater impact on a person's ability to work, perform daily activities, communicate and remain independent.

### Cognitive disorders

Cognitive impairments often associated with Huntington's disease include:

* Difficulty organizing, prioritizing or focusing on tasks
* Lack of flexibility or the tendency to get stuck on a thought, behavior or action (perseveration)
* Lack of impulse control that can result in outbursts, acting without thinking and sexual promiscuity
* Lack of awareness of one's own behaviors and abilities
* Slowness in processing thoughts or ''finding'' words
* Difficulty in learning new information

### Psychiatric disorders

The most common psychiatric disorder associated with Huntington's disease is depression. This isn't simply a reaction to receiving a diagnosis of Huntington's disease. Instead, depression appears to occur because of injury to the brain and subsequent changes in brain function. Signs and symptoms may include:

* Feelings of irritability, sadness or apathy
* Social withdrawal
* Insomnia
* Fatigue and loss of energy
* Frequent thoughts of death, dying or suicide

Other common psychiatric disorders include:

* **Obsessive-compulsive disorder,** a condition marked by recurrent, intrusive thoughts and repetitive behaviors
* **Mania,** which can cause elevated mood, overactivity, impulsive behavior and inflated self-esteem
* **Bipolar disorder,** a condition with alternating episodes of depression and mania

In addition to the above disorders, weight loss is common in people with Huntington's disease, especially as the disease progresses.

### Symptoms of juvenile Huntington's disease

The start and progression of Huntington's disease in younger people may be slightly different from that in adults. Problems that often present early in the course of the disease include:

#### Behavioral changes

* Difficulty paying attention
* Rapid, significant drop in overall school performance
* Behavioral problems

#### Physical changes

* Contracted and rigid muscles that affect gait (especially in young children)
* Tremors or slight involuntary movements
* Frequent falls or clumsiness
* Seizures

## Causes

**Autosomal dominant inheritance patternOpen pop-up dialog box**

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.

## Complications

After Huntington's disease starts, a person's functional abilities gradually worsen over time. The rate of disease progression and duration varies. The time from disease emergence to death is often about 10 to 30 years. Juvenile Huntington's disease usually results in death within 10 years after symptoms develop.

The clinical depression associated with Huntington's disease may increase the risk of suicide. Some research suggests that the greater risk of suicide occurs before a diagnosis is made and in the middle stages of the disease when a person starts to lose independence.

Eventually, a person with Huntington's disease requires help with all activities of daily living and care. Late in the disease, he or she will likely be confined to a bed and unable to speak. Someone with Huntington's disease is generally able to understand language and has an awareness of family and friends, though some won't recognize family members.

Common causes of death include:

* Pneumonia or other infections
* Injuries related to falls
* Complications related to the inability to swallow

**In vitro fertilization**

People with a known family history of Huntington's disease are understandably concerned about whether they may pass the Huntington gene on to their children. These people may consider genetic testing and family planning options.

If an at-risk parent is considering genetic testing, it can be helpful to meet with a genetic counselor. A genetic counselor will discuss the potential risks of a positive test result, which would indicate that the parent will develop the disease. Also, couples will need to make additional choices about whether to have children or to consider alternatives, such as prenatal testing for the gene or in vitro fertilization with donor sperm or eggs.

Another option for couples is in vitro fertilization and preimplantation genetic diagnosis. In this process, eggs are removed from the ovaries and fertilized with the father's sperm in a laboratory. The embryos are tested for presence of the Huntington gene, and only those testing negative for the Huntington gene are implanted in the mother's uterus.

**Thalassemia**

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.

### Products & Services

* [Book: Mayo Clinic Family Health Book, 5th Edition](https://order.store.mayoclinic.com/books/gnweb43?utm_source=MC-DotOrg-PS&utm_medium=Link&utm_campaign=FamilyHealth-Book&utm_content=FHB)
* Symptoms

There are several types of thalassemia. The signs and symptoms you have depend on the type and severity of your condition.

Thalassemia signs and symptoms can include:

* Fatigue
* Weakness
* Pale or yellowish skin
* Facial bone deformities
* Slow growth
* Abdominal swelling
* Dark urine

Some babies show signs and symptoms of thalassemia at birth; others develop them during the first two years of life. Some people who have only one affected hemoglobin gene don't have thalassemia symptoms.

[**Request an Appointment at Mayo Clinic**](https://www.mayoclinic.org/appointments)

## Causes

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.

### Alpha-thalassemia

Four genes are involved in making the alpha hemoglobin chain. You get two from each of your parents. If you inherit:

* **One mutated gene,** you'll have no signs or symptoms of thalassemia. But you are a carrier of the disease and can pass it on to your children.
* **Two mutated genes,** your thalassemia signs and symptoms will be mild. This condition might be called alpha-thalassemia trait.
* **Three mutated genes,** your signs and symptoms will be moderate to severe.

Inheriting four mutated genes is rare and usually results in stillbirth. Babies born with this condition often die shortly after birth or require lifelong transfusion therapy. In rare cases, a child born with this condition can be treated with transfusions and a stem cell transplant.

### Beta-thalassemia

Two genes are involved in making the beta hemoglobin chain. You get one from each of your parents. If you inherit:

* **One mutated gene,** you'll have mild signs and symptoms. This condition is called thalassemia minor or beta-thalassemia.
* **Two mutated genes,** your signs and symptoms will be moderate to severe. This condition is called thalassemia major, or Cooley anemia.

Babies born with two defective beta hemoglobin genes usually are healthy at birth but develop signs and symptoms within the first two years of life. A milder form, called thalassemia intermedia, also can result from two mutated genes.

## Risk factors

Factors that increase your risk of thalassemia include:

* **Family history of thalassemia.** Thalassemia is passed from parents to children through mutated hemoglobin genes.
* **Certain ancestry.** Thalassemia occurs most often in African Americans and in people of Mediterranean and Southeast Asian descent.

## Complications

Possible complications of moderate to severe thalassemia include:

* **Iron overload.** People with thalassemia can get too much iron in their bodies, either from the disease or from frequent blood transfusions. Too much iron can result in damage to your heart, liver and endocrine system, which includes hormone-producing glands that regulate processes throughout your body.
* **Infection.** People with thalassemia have an increased risk of infection. This is especially true if you've had your spleen removed.

In cases of severe thalassemia, the following complications can occur:

* **Bone deformities.** Thalassemia can make your bone marrow expand, which causes your bones to widen. This can result in abnormal bone structure, especially in your face and skull. Bone marrow expansion also makes bones thin and brittle, increasing the chance of broken bones.
* **Enlarged spleen.** The spleen helps your body fight infection and filter unwanted material, such as old or damaged blood cells. Thalassemia is often accompanied by the destruction of a large number of red blood cells. This causes your spleen to enlarge and work harder than normal.

An enlarged spleen can make anemia worse, and it can reduce the life of transfused red blood cells. If your spleen grows too big, your doctor might suggest surgery to remove it.

* **Slowed growth rates.** Anemia can both slow a child's growth and delay puberty.
* **Heart problems.** Congestive heart failure and abnormal heart rhythms can be associated with severe thalassemia.

## Prevention

In most cases, you can't prevent thalassemia. If you have thalassemia, or if you carry a thalassemia gene, consider talking with a genetic counselor for guidance if you want to have children.

There is a form of assisted reproductive technology diagnosis, which screens an embryo in its early stages for genetic mutations combined with in vitro fertilization. This might help parents who have thalassemia or who are carriers of a defective hemoglobin gene have healthy babies.

The procedure involves retrieving mature eggs and fertilizing them with sperm in a dish in a laboratory. The embryos are tested for the defective genes, and only those without genetic defects are implanted into the uterus.

**Brachydactyly**

Brachydactyly is a shortening of the fingers and toes due to unusually short bones. This is an inherited condition, and in most cases does not present any problems for the person who has it. There are different types of brachydactyly, based on which bones are shortened. This condition can also be a symptom of other genetic disorders.

Unless there is an accompanying disorder that produces symptoms, or the shortened digits impair the use of hands and feet, there is no treatment needed for brachydactyly.

## Symptoms of brachydactyly

The signs of brachydactyly are usually present at birth, but it’s possible that shortened limbs become more obvious with growth and development. The main symptom of brachydactyly is fingers, toes, or both that are shorter than normal. Unless you have another condition associated with brachydactyly, you should not feel any pain or have any other symptoms.

The shortened fingers and toes of brachydactyly may cause you to have difficulty with grip. If the brachydactyly is severe in the feet, you may have trouble walking. These symptoms are rare, however, when there is no other condition present.

## Causes of brachydactyly

Brachydactyly is an inherited condition, which makes genetics the main cause. If you have shortened fingers or toes, other members of your family most likely also have the condition. It is an autosomal dominant condition, which means you only need one parent with the gene to inherit the condition. It’s thought that two different mutations in a certain gene contribute to brachydactyly.

In some cases, it’s possible that brachydactyly is caused by exposure to medications that the mother takes during pregnancy. It may also be caused by blood flow problems to the hand and feet, especially in developing babies.

It’s possible that your brachydactyly is symptomatic of a genetic syndrome. This is much rarer. If so, you will have other symptoms besides the shortened fingers or toes. Brachydactyly might be caused by Down syndrome or Cushing’s syndrome, for example.

## Types of brachydactyly

The different types of brachydactyly are categorized by the bones and digits affected.

### Type A

Type A brachydactyly is the shortening of the middle phalanges. These are the finger bones that are the second from the end of each digit. Type A is further classified by finger types. These are as follows:

* Type A1: The middle phalanges of all the fingers are shortened.
* Type A2: The index finger and sometimes the little finger are shortened.
* Type A3: Only the little finger is shortened.

### Type B

Type B brachydactyly affects the ends of the index through little fingers. The last bone on each finger is shortened or completely missing. The nails are also absent. The same occurs in the toes. The thumb bones are always intact but often flattened and/or split.

### Type C

Type C is rare and affects the index, middle, and little fingers. The middle phalanges, as in type A, are shortened, but the ring finger is often not affected and is the longest finger on the hand.

### Type D

Type D brachydactyly is considered to be common and affects only the thumbs. The end bones of the thumbs are shortened but all the fingers are normal.

### Type E

Type E brachydactyly is a rare form if it is not accompanied by another disorder. It is characterized by shortened metacarpals and metatarsals. These are the bones in the hands and feet that are third and fourth from the end of the digits. The result is small hands or feet.

## Diagnosis of brachydactyly

A careful examination of the hands and feet by a doctor may be enough to diagnose brachydactyly. X-rays can also be used to see which bones are shortened and to diagnose the type of brachydactyly. In mild cases, an X-ray may be the only way to tell that the condition is present.

To determine if brachydactyly is part of a syndrome, a full skeletal X-ray may be done. This can help determine if other bones in the body are abnormal, which suggests a syndrome. Genetic testing may also be necessary to determine if the syndrome is present.

## Treatment for brachydactyly

In a large majority of cases of brachydactyly, no treatment is necessary. If your condition is not a part of another syndrome, you should be healthy and will have no medical concerns related to your hands and feet.

In rare cases, brachydactyly may be severe enough to present problems with functionality. You may have trouble gripping things or walking normally. In these instances, physical therapy can help. Physical therapy can improve the range of motion and improve both strength and functionality of the affected areas.

### Surgery

In extreme and very rare cases, surgery may be used to treat brachydactyly.

Plastic surgery may be used for cosmetic purposes, or in rare cases, to improve functionality. Many who need surgery will have brachydactyly along with another condition. Surgery may include an osteomy, which cuts the bone. This can contribute to [“gradual lengthening”Trusted Source](https://www.ncbi.nlm.nih.gov/pubmed/21407065) of the shortened fingers.

## Risk factors

Brachydactyly is typically an inherited condition. If you have a family member with brachydactyly, your risk for also having it is much higher.

If your child is born with Down syndrome, their risk for brachydactyly is higher.

Women are more likely to develop brachydactyly than men. This may be partially because women are more likely to experience the full expression of the trait than men. This makes it more noticeable in them.

## Complications

Most people with brachydactyly won’t experience any significant complications that hinder their daily lives. In some cases, if the brachydactyly is severe enough, it can limit the functioning of the hand or difficulty walking. Surgery and physical therapy may be used to improve functioning.

**Diabetes**

Diabetes mellitus, commonly known as diabetes, is a metabolic disease that causes high blood sugar. The hormone insulin moves sugar from the blood into your cells to be stored or used for energy. With diabetes, your body either doesn’t make enough insulin or can’t effectively use the insulin it does make.

Untreated high blood sugar from diabetes can damage your nerves, eyes, kidneys, and other organs.

There are a few different types of diabetes:

* [Type 1 diabetes](https://www.healthline.com/health/type-1-diabetes-causes-symtoms-treatments) is an [autoimmune disease](https://www.healthline.com/health/autoimmune-disorders). The immune system attacks and destroys cells in the [pancreas](https://www.healthline.com/human-body-maps/pancreas), where insulin is made. It’s unclear what causes this attack. About [10 percent](https://www.idf.org/aboutdiabetes/what-is-diabetes/types-of-diabetes.html) of people with diabetes have this type.
* [Type 2 diabetes](https://www.healthline.com/health/type-2-diabetes) occurs when your body becomes resistant to [insulin](https://www.healthline.com/health/type-2-diabetes/insulin), and sugar builds up in your blood.
* [Prediabetes](https://www.healthline.com/health/type-2-diabetes/what-is-prediabetes) occurs when your blood sugar is higher than normal, but it’s not high enough for a diagnosis of type 2 diabetes.
* [Gestational diabetes](https://www.healthline.com/health/gestational-diabetes) is high blood sugar during pregnancy. Insulin-blocking hormones produced by the placenta cause this type of diabetes.

A rare condition called [diabetes insipidus](https://www.healthline.com/health/type-2-diabetes/diabetes-insipidus) is not related to diabetes mellitus, although it has a similar name. It’s a different condition in which your kidneys remove too much fluid from your body.

Each type of diabetes has unique symptoms, causes, and treatments. [Learn more about how these types differ from one another.](https://www.healthline.com/health/diabetes/types-of-diabetes)

## Symptoms of diabetes

Diabetes symptoms are caused by rising blood sugar.

### General symptoms

The general symptoms of diabetes include:

* increased hunger
* increased thirst
* weight loss
* [frequent urination](https://www.healthline.com/health/frequent-urination-diabetes)
* [blurry vision](https://www.healthline.com/health/diabetes/blurry-vision)
* [extreme fatigue](https://www.healthline.com/health/diabetes/fatigue)
* [sores that don’t heal](https://www.healthline.com/health/diabetes/diabetes-and-wound-healing)

### Symptoms in men

In addition to the general symptoms of diabetes, [men with diabetes](https://www.healthline.com/health/recognizing-diabetes-symptoms-men) may have a [decreased sex drive](https://www.healthline.com/health/type-2-diabetes/sex-health), [erectile dysfunction (ED)](https://www.healthline.com/health/type-2-diabetes/type-2-and-erectile-dysfunction), and poor muscle strength.

### Symptoms in women

[Women with diabetes](https://www.healthline.com/health/diabetes/symptoms-in-women) can also have symptoms such as [urinary tract infections](https://www.healthline.com/health/urinary-tract-infection-adults), [yeast infections](https://www.healthline.com/health/diabetes/diabetes-and-yeast-infections), and dry, itchy skin.

### Type 1 diabetes

Symptoms of type 1 diabetes can include:

* extreme hunger
* increased thirst
* unintentional weight loss
* [frequent urination](https://www.healthline.com/health/urination-excessive-volume)
* blurry vision
* tiredness

It may also result in mood changes.

### Type 2 diabetes

Symptoms of type 2 diabetes can include:

* increased hunger
* increased thirst
* increased urination
* blurry vision
* tiredness
* sores that are slow to heal

It may also cause recurring infections. This is because elevated glucose levels make it harder for the body to heal.

### Gestational diabetes

Most women with gestational diabetes don’t have any symptoms. The condition is often detected during a routine blood sugar test or oral glucose tolerance test that is usually performed between the 24th and 28th weeks of gestation.

In rare cases, a woman with gestational diabetes will also experience increased thirst or urination.

### The bottom line

Diabetes symptoms can be so mild that they’re hard to spot at first. [Learn which signs should prompt a trip to the doctor.](https://www.healthline.com/health/diabetes-symptoms)

## Causes of diabetes

Different causes are associated with each type of diabetes.

### Type 1 diabetes

Doctors don’t know exactly what causes type 1 diabetes. For some reason, the immune system mistakenly attacks and destroys insulin-producing beta cells in the [pancreas](https://www.healthline.com/health/diabetes-and-pancreas).

Genes may play a role in some people. It’s also possible that a virus sets off the immune system attack.

### Type 2 diabetes

Type 2 diabetes stems from a combination of [genetics](https://www.healthline.com/health/type-2-diabetes/genetics) and lifestyle factors. Being overweight or [obese](https://www.healthline.com/health/obesity) increases your risk too. Carrying extra weight, especially [in your belly](https://www.healthline.com/nutrition/20-tips-to-lose-belly-fat), makes your cells more resistant to the effects of insulin on your blood sugar.

This condition runs in families. Family members share genes that make them more likely to get type 2 diabetes and to be overweight.

### Gestational diabetes

Gestational diabetes is the result of hormonal changes during pregnancy. The placenta produces hormones that make a pregnant woman’s cells less sensitive to the effects of insulin. This can cause high blood sugar during pregnancy.

Women who are [overweight when they get pregnant](https://www.healthline.com/health/pregnancy/obesity) or who [gain too much weight during their pregnancy](https://www.healthline.com/health-news/gaining-too-much-weight-during-pregnancy) are more likely to get gestational diabetes.

### The bottom line

Both genes and environmental factors play a role in triggering diabetes. [Get more information here on the causes of diabetes.](https://www.healthline.com/health/diabetes-causes)

## Diabetes risk factors

Certain factors increase your risk for diabetes.

### Type 1 diabetes

You’re more likely to get type 1 diabetes if you’re a child or teenager, you have [a parent or sibling](https://www.healthline.com/health/family-health-history-day) with the condition, or you carry certain genes that are linked to the disease.

### Type 2 diabetes

Your risk for type 2 diabetes increases if you:

* are overweight
* are age 45 or older
* have a parent or sibling with the condition
* aren’t physically active
* have had gestational diabetes
* have prediabetes
* have [high blood pressure](https://www.healthline.com/health/type-2-diabetes/hypertension), [high cholesterol](https://www.healthline.com/health/high-cholesterol/treating-with-statins/guide-to-diabetes-and-high-cholesterol), or [high triglycerides](https://www.healthline.com/health/triglyceride-level)
* have African American, Hispanic or Latino American, Alaska Native, Pacific Islander, American Indian, or Asian American ancestry

### Gestational diabetes

Your risk for gestational diabetes increases if you:

* are overweight
* are over age 25
* had gestational diabetes during a past pregnancy
* have given birth to a baby weighing [more than 9 pounds](https://www.healthline.com/health/macrosomia)
* have a family history of type 2 diabetes
* have [polycystic ovary syndrome (PCOS)](https://www.healthline.com/health/diabetes/are-pcos-and-diabetes-connected)

### The bottom line

Your family, environment, and preexisting medical conditions can all affect your odds of developing diabetes. [Find out which risks you can control and which ones you can’t.](https://www.healthline.com/health/diabetes-risk-factors)

## Diabetes complications

High blood sugar damages organs and tissues throughout your body. The higher your blood sugar is and the longer you live with it, the greater your risk for complications.

Complications associated with diabetes include:

* [heart disease](https://www.healthline.com/health/type-2-diabetes/understanding-cv-disease-diabetes), [heart attack](https://www.healthline.com/health/heart-attack), and [stroke](https://www.healthline.com/health/diabetes/diabetes-and-stroke)
* [neuropathy](https://www.healthline.com/health/type-2-diabetes/diabetic-neuropathy)
* [nephropathy](https://www.healthline.com/health/type-2-diabetes/nephropathy)
* [retinopathy](https://www.healthline.com/health/type-2-diabetes/retinopathy) and [vision loss](https://www.healthline.com/health/diabetes/diabetic-eye-exam)
* [hearing loss](https://www.healthline.com/health/type-2-diabetes/hearing-loss)
* [foot damage](https://www.healthline.com/health/type-2-diabetes/feet) such as infections and sores that don’t heal
* [skin conditions](https://www.healthline.com/health/type-2-diabetes/skin-problems) such as [bacterial](https://www.healthline.com/health/cellulitis) and [fungal](https://www.healthline.com/health/fungal-infection) infections
* [depression](https://www.healthline.com/health/type-2-diabetes/depression)
* [dementia](https://www.healthline.com/health/dementia-risk-factors)

### Gestational diabetes

Uncontrolled gestational diabetes can lead to problems that affect both the mother and baby. Complications affecting the baby can include:

* [premature birth](https://www.healthline.com/health/pregnancy/premature-infant)
* [higher-than-normal weight at birth](https://www.healthline.com/health/parenting/average-baby-weight)
* increased risk for type 2 diabetes later in life
* [low blood sugar](https://www.healthline.com/health/hypoglycemia)
* [jaundice](https://www.healthline.com/health/newborn-jaundice)
* stillbirth

The mother can develop complications such as high blood pressure ([preeclampsia](https://www.healthline.com/health/preeclampsia)) or type 2 diabetes. She may also require [cesarean delivery](https://www.healthline.com/health/c-section), commonly referred to as a C-section.

The mother’s risk of gestational diabetes in future pregnancies also increases.

### The bottom line

Diabetes can lead to serious medical complications, but you can manage the condition with medications and lifestyle changes. [Avoid the most common diabetes complications with these helpful tips.](https://www.healthline.com/health/type-2-diabetes/6-complications-diabetes)

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## Treatment of diabetes

Doctors treat diabetes with a few different medications. Some of these drugs are taken [by mouth](https://www.healthline.com/health/diabetes/diabetes-pills-vs-insulin), while others are available as [injections](https://www.healthline.com/health/intravenous-medication-administration).

### Type 1 diabetes

[Insulin](https://www.healthline.com/health/diabetes/insulin-injection) is the main treatment for type 1 diabetes. It replaces the hormone your body isn’t able to produce.

There are four types of insulin that are most commonly used. They’re differentiated by how quickly they start to work, and how long their effects last:

* Rapid-acting insulin starts to work within 15 minutes and its effects last for 3 to 4 hours.
* Short-acting insulin starts to work within 30 minutes and lasts 6 to 8 hours.
* Intermediate-acting insulin starts to work within 1 to 2 hours and lasts 12 to 18 hours.
* Long-acting insulin starts to work a few hours after injection and lasts 24 hours or longer.

### Type 2 diabetes

Diet and exercise can help some people manage type 2 diabetes. If lifestyle changes aren’t enough to lower your blood sugar, you’ll need to take medication.