Autosomal Dominant Traits and Disorders

by Sophia

WHAT'S COVERED

In this lesson, you will learn about the characteristics and examples of autosomal dominant traits and disorders. Specifically, this lesson will cover:

1. Cause of Autosomal Dominant Traits

Autosomal dominant traits or disorders are caused by the inheritance of at least one dominant allele on a person's autosomes. As you learned in a previous lesson, autosomes are the chromosomes found in your cells, excluding the sex chromosomes.

For each trait that you have, you possess two alleles for that trait, one inherited from your mother and one inherited from your father. For a person to express an autosomal dominant trait, they need to have at least one dominant allele.

This means the person's genotypes could be either big A-little a (Aa) or big A-big A (AA). You only need one dominant allele to phenotypically express an autosomal dominant trait; you can be homozygous dominant or heterozygous and still have that particular autosomal dominant trait.

TERM TO KNOW

Autosomal Dominant

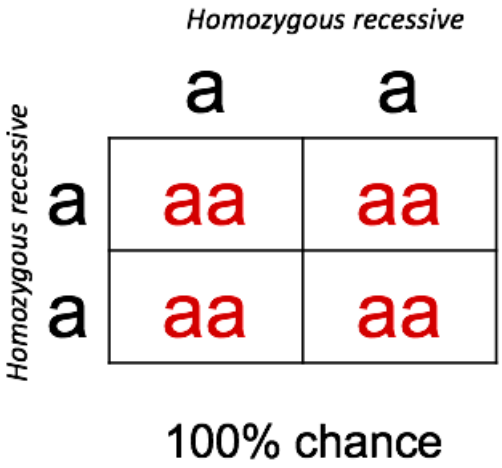
A trait or disorder caused by the inheritance of at least one dominant allele on an autosome.

2. Punnett Squares with Autosomal Dominant Disorders

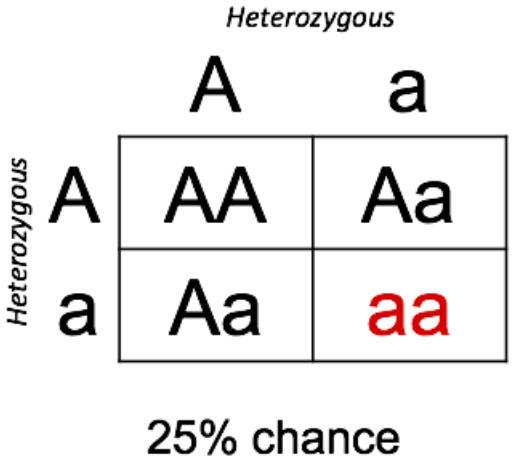
Some traits are controlled by recessive alleles, and some traits are controlled by dominant alleles. Punnett squares are thus one way to see how the genotypes of parents can affect the outcome of their children.

↱ EXAMPLE If a parent possesses at least one dominant allele, that parent will have that trait. The only way that 100% of their potential children would be normal is if both parents were homozygous

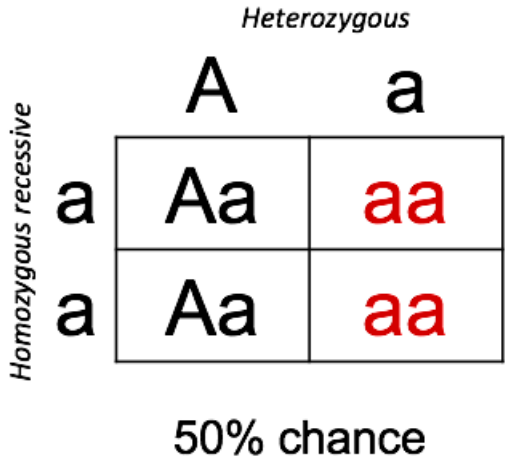
recessive. In the Punnett square below, you can see that if both parents are homozygous recessive, meaning neither possesses a single dominant allele, then they have a 100% chance of having all normal children. In this case, the normal trait would be homozygous recessive; however, you only need one dominant allele to possess a dominant trait.



↱ EXAMPLE Say both parents are heterozygous, meaning they have an autosomal trait or disorder because they both have at least one dominant allele. If you look at the Punnett Square below, you'll notice that because they also each possess a recessive allele for the normal condition, they actually have a 25% chance of having a child who is unaffected by the disease.



↱ EXAMPLE Now say one parent is heterozygous, and the other parent is homozygous recessive. The first parent contains one allele for that autosomal dominant trait—that parent possesses the trait—while the second parent is completely normal. As the Punnett square shows, this combination of parent alleles means the parents have a 50% chance of having totally normal children.



3. Common Autosomal Dominant Disorders

The following are examples of autosomal dominant disorders:

- **Huntington's Disease**: A disease that can be inherited, but the symptoms don't show up until adulthood. Often, people don't realize that they have Huntington's disease until they've already reproduced and passed it on to their children.
- **Marfan Syndrome**: This disorder causes a weakening of the aorta, meaning that the aorta can rupture over time. Generally, people with this disorder are very, very tall and lanky. However, the weakening of the aorta is the most serious aspect of Marfan syndrome since rupturing can occur with intense physical activity.
- **Achondroplasia**: This disorder results in a person who has short arms and legs and who is short overall. Most people with this type of disorder only get to be about 4 to 4.5 feet tall, so achondroplasia affects a person's height, and then—as a result—causes shortness of the arms and legs.
- **Familial Hypercholesterolemia**: This disorder leads to high blood cholesterol. This is because the cholesterol in the blood of people with this disorder won't bind to LDLs, which is the first step in removing the cholesterol from the body. Therefore, most people with familial hypercholesterolemia often don't have as long of a lifespan as other people as a result of their very high cholesterol.

TERMS TO KNOW

Huntington's Disease

An autosomal dominant disorder which does not show up until later in life often after the gene has been passed onto offspring; it is a neurodegenerative disorder that causes motor and cognitive impairment and eventually becomes fatal.

Marfan Syndrome

A genetic disorder of connective tissue that causes people to have a certain appearance: being abnormally tall with long limbs and digits; it can also affect other connective tissues such as heart valves, and can be fatal.

Achondroplasia

An autosomal dominant disorder in which the person is abnormally short in stature with short arms and legs.

Familial Hypercholesterolemia
An autosomal dominant disorder in which a person has chronic high cholesterol.

SUMMARY

In this lesson, you learned that the **cause of autosomal dominant traits** or disorders is the inheritance of at least one dominant allele on an autosome. In other words, if one parent has one dominant allele, there is a chance that the children will have the dominant trait as well, even though the other parent does not have that dominant allele, which can be shown in a **Punnett square**.

You now understand that some common examples of **autosomal dominant disorders** are Huntington's disease, Marfan syndrome, achondroplasia, and familial hypercholesterolemia.

Keep up the learning and have a great day!

Source: ADAPTED FROM SOPHIA TUTORIAL BY AMANDA SODERLIND.

TERMS TO KNOW

Achondroplasia
An autosomal dominant disorder in which the person is abnormally short in stature with short arms and legs.

Autosomal Dominant
A trait or disorder caused by the inheritance of at least one dominant allele on an autosome.

Familial Hypercholesterolemia
An autosomal dominant disorder in which a person has chronic high cholesterol.

Huntington's Disease
An autosomal dominant disorder which does not show up until later in life, often after the gene has been passed onto offspring. It is a neurodegenerative disorder that causes motor and cognitive impairment and eventually becomes fatal.

Marfan Syndrome
A genetic disorder of connective tissue that causes people to have a certain appearance: being abnormally tall with long limbs and digits. It can also affect other connective tissues such as heart valves, and can be fatal.