HEALTH BASICS

Autosomal dominant

Autosomal dominant describes how some gene variants can pass through families.

WHAT IT IS
An inheritance pattern involving one copy of a gene variant

WHERE IT COMES UP
Genetic test reports, family histories, and inherited-condition diagnoses

WHY IT MATTERS
It can estimate relatives' chance of sharing a variant

WHAT IT IS NOT
Not a disease or a diagnosis

WHO TO ASK
A genetic counselor or clinical genetics service

HOW IT IS CHECKED
Family history and condition-specific genetic testing

Overview

Autosomal dominant is a pattern of genetic inheritance. It describes how a particular gene variant may pass from a parent to a child. It is not a disease or a diagnosis.

"Autosomal" means that the gene is on one of the numbered chromosomes. People usually have 22 pairs of numbered chromosomes, called autosomes, plus a pair of sex chromosomes.

"Dominant" means that one altered copy of a gene can be enough to affect a person's health. It does not mean that the condition is more severe, more common, or stronger than conditions with other inheritance patterns.

The meaning of an autosomal dominant result depends on the gene, the exact variant, and the condition linked to it. Some variants have major health effects. Others have mild effects, do not affect everyone who carries them, or remain uncertain after testing.

How it works

People usually have two copies of each autosomal gene. One copy comes from the egg and the other from the sperm.

In a typical autosomal dominant situation, one parent has one altered copy and one usual copy of the gene. For each pregnancy, that parent passes on only one of those copies. This creates:

These chances start again with every pregnancy. Having one child who inherits the variant does not change the chance for the next child. The calculation also assumes that the parent has one altered copy and that the other biological parent does not have a relevant variant in the same gene.

People of any sex can inherit and pass on an autosomal dominant variant. A father can pass one to a son, which helps distinguish this pattern from some forms of X-linked inheritance.

Not every autosomal dominant variant is inherited from a parent. A new, or de novo , variant can arise in an egg, sperm, or early embryo. The child may then be the first person in the family known to have it.

A person can also have mosaicism , meaning the variant is present in some cells but not others. If it is present in egg or sperm cells, it may sometimes be passed on even when a parent's blood test does not find it.

Two other terms can affect what an inherited variant means:

Because of these factors, a family tree may not show a clear pattern even when a condition is autosomal dominant.

- A 1 in 2 chance of passing on the altered copy
- A 1 in 2 chance of passing on the usual copy
- Penetrance is the proportion of people with a variant who develop its associated features
- Variable expression means those features can differ between relatives with the same variant

Why it matters for your health

Knowing that a condition is autosomal dominant can help estimate whether biological relatives may share the same genetic variant. It can also guide decisions about genetic counseling, testing, health monitoring, and pregnancy planning.

Examples of conditions that can follow this pattern include Huntington disease, Marfan syndrome, familial hypercholesterolemia, and autosomal dominant polycystic kidney disease. These are separate conditions with different health effects and care needs. The inheritance label alone does not tell you which condition is present or how it will affect someone.

The exact wording of a genetic test result matters. Laboratories may classify variants as pathogenic, likely pathogenic, of uncertain significance, likely benign, or benign. A variant of

uncertain significance , often shortened to VUS, is not the same as a confirmed disease-causing result. It usually cannot establish a diagnosis or reliably predict relatives' health by itself.

Genetic testing also has limits. A negative result may not rule out a condition if the relevant gene is unknown, the test did not examine every type of genetic change, or no confirmed family variant was available for comparison.

What can go wrong

The main risks come from misunderstanding or applying the inheritance pattern without enough information.

A "50% chance" may be mistaken for a statement that half of a family's children must inherit the variant. In reality, each pregnancy is a separate event. It is possible for several children, one child, or no children to inherit it.

A missing family history can also be misleading. A variant may be new in the family, relatives may not have been tested, or the condition may not affect everyone who carries the variant. Small families and incomplete health records can make the pattern harder to see.

Another concern is treating an uncertain result as a diagnosis. A VUS may later be reclassified as more evidence becomes available. Medical or family decisions should not usually be based on that result alone without condition-specific interpretation.

The consequences of missing a confirmed result depend on the associated condition. For example, familial hypercholesterolemia relates to very high cholesterol and early heart disease, while Marfan syndrome can involve the body's main artery. Autosomal dominant polycystic kidney disease affects the kidneys, and Huntington disease affects the brain. Each requires its own assessment rather than a general "autosomal dominant" care plan.

The inheritance term itself never creates an emergency. Reviewing a genetic report, arranging counseling, or discussing testing for relatives is usually routine follow-up. Urgent or emergency care is based on symptoms of the specific condition. If you have a diagnosed condition with an emergency plan, follow that plan. Sudden severe chest or back pain, fainting, severe breathing difficulty, or signs of a stroke require emergency assessment regardless of genetic history.

What you can do

If "autosomal dominant" appears in your test result or family records, gather the exact information rather than relying on the label alone.

Useful steps include:

A genetic counselor or clinical genetics service can explain the result and draw a family tree covering several generations. They can also discuss the limits of testing and the possible effects on relatives.

Testing relatives for a known family variant is called cascade testing . It is usually more focused than broad genetic testing because the laboratory already knows which variant to look for.

If you are considering pregnancy, a genetics service can explain available options. These may include testing during pregnancy, testing embryos created through in vitro fertilization, donor eggs or sperm, adoption, or choosing not to test. These are personal options, not required steps.

If you only encountered the term in general reading and have no related diagnosis, family history, or genetic result, there may be nothing you need to do.

- Keep a copy of the full laboratory report
- Record the gene name, exact variant, and laboratory classification
- Ask which condition the result is linked to
- Ask whether the finding is confirmed or still uncertain
- Write down relevant diagnoses and test results in biological relatives
- Ask whether testing should begin with a relative who has the condition
- Ask whether relatives need targeted testing for the known family variant

Common questions

Does autosomal dominant mean I have a genetic disease?

No. It is an inheritance pattern. Whether you have a condition requires information about your health, family history, and any genetic findings.

If my parent has an autosomal dominant condition, is my chance always 50%?

A 1 in 2 chance is typical when the parent has one disease-associated copy and one usual copy of the gene. Unusual situations, such as mosaicism or two altered copies, can change the calculation.

Can a person carry the variant without knowing?

Yes. Some associated conditions begin later in life, cause subtle features, or have reduced penetrance. Other people may already have signs that were not recognized as part of an

inherited condition.

Can an autosomal dominant condition skip a generation?

The variant does not literally jump over someone. A person between two affected generations may carry it without clear features, may never develop features, or may not have had formal assessment. Family information may also be incomplete.

Does a negative genetic test mean my relatives are not at risk?

Not always. Its meaning depends on what was tested and whether a confirmed family variant was known. A relative who tests negative for a known familial pathogenic variant generally has a different interpretation from someone whose broad test found no explanation.

Should children be tested?

This depends on the specific condition and whether results would change care during childhood. A genetics service can explain the medical and personal issues for that particular family.

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