

GENETICS FORUM: Glossary of genetic terminology

Adult-onset A change in a person's DNA that increases his/her risk for

condition: developing a specific health condition later in life, often in middle

age (40s-60s).

<u>Carrier Status:</u> An individual has a DNA change for a health condition but does not

display symptoms. In order to have the health condition, an individual must have inherited DNA changes from both parents. An

individual must have inherited DNA changes from both parents. An individual having one normal gene and one DNA change does not have the disease. Two carriers may produce children with the

health condition.

<u>Carrier screening:</u> A type of genetic testing performed on people who display no

symptoms for a genetic disorder but may be at risk for passing it on

to their children.

<u>Cell:</u> The basic building block of living things found in all of the tissues

and organs in your body.

Chromosome: An organized section of DNA found in all of person's cells. Humans

have 23 pairs of chromosomes. Each parent gives one chromosome to each pair so that a child gets half of their chromosomes from

their mother and half from their father.

DNA: (Deoxyribonucleic Acid) The chemical name for the molecule that

carries genetic instructions in all living things.

DNA Change: This refers to a change in a DNA sequence, often called a mutation

or variant. These changes can be present when a person is born or can happen during their lifetime. Changes can result from DNA copying mistakes made during cell division, exposure to radiation,

chemicals, or infection by viruses.

Dominant: A DNA change that is almost always expressed, even if only one

copy is present.

Genome A laboratory technique used to determine the exact sequence of

sequencing: DNA, including any changes in a DNA sequence.

Gene: The basic physical unit of inheritance. Genes are passed from

parents to children and contain the information needed to specify personal characteristics. Humans have approximately 20,000 genes

on their chromosomes.



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Genetic
Information
Nondiscrimination
Act (GINA)

Federal legislation that makes it unlawful to discriminate against individuals on the basis of their genetic information in regard to health insurance and employment.

Genetics: The study of genes and heredity (see **Heredity**) in an organism.

Genetic testing: A laboratory method that looks at DNA changes in your genes.

Genetic tests are used to diagnose genetic conditions, identify carriers and individuals at increased risk. Genetic test results can be used to make decisions about screening, management of care and

potential response to medications.

Genetic variation: Every person has a unique set of DNA. We share much of our

DNA with other humans and with every living thing. The small set of DNA changes that differ between humans is called genetic variation. This is what gives you your traits (see **Trait**).

Genome: All of a person's genetic information.

Genomics: The study of the entire genome of an organism.

Heredity: The transmission of personal characteristics from one generation

to the next through the genes.

Human Genome

Project:

An international project that mapped and sequenced the entire

human genome. It was completed in April 2003.

Inherited: Passed down from parents to children through generations. The

genes present in the parents are passed down to their children

through egg or sperm cells.

Medically

This is when a change in a person's DNA increases the risk for developing a specific health condition, where actions can be tak

developing a specific health condition, where actions can be taken to prevent, delay, or reduce symptoms of the health condition.

Nucleotide: The building blocks of DNA that contain one of the letters A, T, G

or C.

Pathogenic: This is when a DNA change causes a health condition.



genome:

findings:

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Pharmacogenetics: The study of the interaction of an individual's genetic makeup and

response to a drug.

Protein: Proteins are required for the structure, function, and regulation of

the body's cells, tissues, and organs; each protein has unique functions. Examples are hormones, enzymes, and antibodies.

Recessive: Two DNA changes are needed in the same gene one from your

mom and one from your dad. For a male, if one copy is present on the X chromosome (see **Chromosome**) he gets from his mother

this is also recessive.

Reference This is a representative set of DNA sequences. When a new

person has their genome sequenced, it is compared to a reference

genome to see what is unique to that person.

Risk: In the area of genetics, this is the probability that an individual will

develop a particular health condition. Both genes and environment

influence risk.

Secondary These are health conditions that you did not know you have a

higher chance of developing. They are discovered unintentionally and are unrelated to the reason the genetic test is being performed.

These are sometimes called "incidental findings".

<u>Targeted</u> This is when medication or medical procedures are given differently

treatment: to people who have the same health condition but different DNA

changes.

Trait: A specific characteristic like height, eye color, personality, or

abilities. This also includes personal risk for a health condition. Traits are determined by genes or by the environment or, more commonly, by interactions between genes and the environment.

commonly, by interactions between genes and the environment

Variant of unknownScientists and doctors do not know what many DNA changes that can be found by genome sequencing mean. A VUS is a DNA change

significance (VUS): that may or may not lead to a health condition or other harmful

trait (see **Trait**) but at this time, we do not know enough information about the DNA change to predict what it means.