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Glossary for HI Information Sheets

We have compiled a ‘glossary' which helps to explain (or will help you navigate) some of the more technical words which we have used in our information sheets and family resources. Any term highlighted in blue in this document is defined.

Allele

An allele is a specific version of a gene. Each person inherits two alleles for each gene, one from their Mum and one from their Dad. They are responsible for differences in traits including non-disease traits like eye colour or blood type.

Anaerobic

Anaerobic exercise is high-intensity exercise where your muscles work so hard that they don’t get enough oxygen to help make energy so the muscles can move. Instead, during this exercise the muscles make energy from glucose (sugar) that they have stored up in the body.

Benign/Likely Benign Variant

Please also see the entry in the glossary under ‘variant’. Genetic changes are put into categories based on how likely they are to be responsible for causing a condition. If a genetic change has been investigated before and we know it is part of our normal genetic variation and doesn’t cause a disease, we call it a benign or likely benign variant.

Beta Cells

Beta cells are cells in your pancreas that produce insulin. They detect rising blood sugar after you eat and release insulin that then moves the sugar from your blood into your cells so they can be used for energy.

Carrier

A carrier is someone who has a recessive variant within a disease gene but does not show any symptoms because they have another copy of the gene without the genetic change which compensates for the recessive variant. This means they are usually heterozygous. While carriers themselves are unaffected, they can still pass the recessive variant to their children.

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Cell

We are made up of trillions of cells and they are considered the building blocks of life. They contain genetic information in the form of DNA, and they have lots of functions that make sure our bodies work properly. Cells can work individually or can work together and form organs.

Characteristic

This refers to distinctive features of a person like eye colour or blood type that can be passed down from a parent to their child. Another term that can be used to describe a characteristic is a 'trait'.

Chromosome

Within each cell, the DNA forms structures called chromosomes. A person has 46 chromosomes in each cell; 23 of them inherited from their mother, and 23 of them inherited from their father. These chromosomes are numbered 1 to 22 not including the sex chromosomes. For the sex chromosomes, a male will usually have an X and Y chromosome, and a female will usually have two X chromosomes.

Clinical Geneticist

A clinical geneticist is a medically trained doctor that diagnoses, manages, and helps treat genetic conditions. They provide personalised care and guidance for individuals and their families affected by inherited conditions.

Congenital

This means that the cause of a condition has been present at or since the birth of a child.

De novo Variant

This is a genetic change that happens for the first time in an individual and is not inherited from either parent.

DNA

DNA (or deoxyribonucleic acid) is the molecule that acts as an instruction manual that tells our cells what to do. These instructions are split up into sections called genes.

Dominant Variant

Dominant variants cause disease when only one copy of the gene has the genetic change.

Duplicated

This means a section of DNA has been copied which can lead to a disease. These can be whole genes or just sections of gene.

Endocrine

This refers to anything to do with the endocrine system. The endocrine system is a group of glands that produce hormones that regulate how our bodies work and grow.

Gene

A gene is a part of DNA that carries the instructions for making proteins. Each person will have 2 copies of a gene, one inherited from their Mum and one inherited from their Dad.

Genetic

Genetics means how characteristics can be passed from parents to their children.

Genetic Counsellor

A genetic counsellor is a healthcare professional that specialises in genetics and provides support to both individuals and their families on genetic testing and the risk of inherited conditions. They help people to make informed decisions about their health in relation to genetics.

Glycaemic Index

The glycaemic index is a ranking of carbohydrate foods based on how quickly they raise blood sugar (glucose) levels after they have been eaten. Foods with a low glycaemic index include oats or legumes, and foods with a high glycaemic index include bread and potatoes.

Heterozygous

This means an individual has 2 different versions (alleles) of a section of DNA.

Homozygous

This means an individual has 2 identical versions (alleles) of a section of DNA.

Hormones

Hormones are chemical messengers that travel around the body to control or regulate different biological processes.

Hyperammonaemia

This is where you have a high level of a molecule called ammonia in your blood. This is often seen in people with HI due to genetic changes in the *GLUD1* gene.

Hyperinsulinism

This is a condition where the pancreas produces too much insulin, causing your blood sugar levels to drop very low (hypoglycaemia).

Hypoglycaemia

This is a condition where your blood sugar (glucose) levels drop too low, meaning your body lacks enough energy to function properly.

Hypotension

This is the medical word for low blood pressure.

Inherit/Inherited

In biology, this means that a genetic characteristic has been passed from a parent or both parents to their child. There are lots of different forms of inheritance, including; dominant, recessive, mosaic, and other rarer forms.

Insulin

Insulin is a hormone that is produced in the pancreas that controls the amount of sugar (also known as glucose) in the blood. Too much insulin (hyperinsulinism) can cause hypoglycaemia, and too little insulin can cause diabetes.

Lesion

A lesion is a part of an organ or tissue that is not working properly or is damaged usually caused by a disease or an injury.

Mosaic Variants

This means that only some cells in the body will have the genetic change, and the rest of the cells in the body will not have the genetic change and work properly.

Neuromuscular

This refers to the connection between the nervous system and the muscles in your body. The neuromuscular system lets you do things like walking, breathing, and keeping your posture.

Pancreas

The pancreas is an organ behind the stomach and it has two main functions. The first is to release molecules that help you to break down the food you eat, and the second is to produce hormones like insulin that help to control blood sugar levels.

Pathogenic/Likely Pathogenic Variant

Please also see the entry in the glossary under ‘variant’. Genetic changes are put into categories based on how likely they are to be responsible for causing a condition. If a genetic

change has been investigated before and we know it is likely to cause the condition, we call it a pathogenic or likely pathogenic variant.

Penetrance

This refers to the number of people with a specific genetic change that actually show symptoms of a disease the genetic change is known to cause. If every person with the genetic change shows symptoms, it is called complete penetrance. If not everyone with the genetic change shows symptoms it is called variable penetrance.

Pituitary Gland

This is a small group of cells (called a gland) at the bottom of the brain that produces hormones and regulates different processes in your body.

Proteins

Proteins are molecules that are built using the instructions from DNA and do different things within your cells and around your body. Proteins are essentials for your body to work properly.

Recessive Variant

A recessive variant is a genetic change where both copies of a gene need to be changed to cause a disease.

Sporadic

A sporadic variant is a random change to your DNA that hasn't come from a parent.

Variable

Variable means something can be different or can change.

Variable Expressivity

Variable expressivity is where people with the same genetic change will develop the condition but they may show differences in the severity of their symptoms. This is different

from variable penetrance because with variable expressivity, everyone who has the genetic change will show symptoms.

Variable Penetrance

This is where not everyone with the genetic change will develop the condition. This happens because of various genetic, environmental, and lifestyle factors.

Variant

A variant is a permanent change in the DNA sequence. They can be inherited from parents or can happen during your lifetime. These changes can have no effect on you (this is called a benign variant), they can have an effect on you (this is called a pathogenic variant), or we might not know what effect they might have (this is called a variant of uncertain significance or a VUS).

Variant of Uncertain Significance

Please also see the entry in the glossary under 'variant'. Genetic changes are put into categories based on how likely they are to be responsible for causing a condition. If we don't have enough evidence to be confident that the genetic change is the cause of a condition or is just part of our normal genetic variation, it is called a variant of uncertain significance, or VUS. This can't be used to diagnose a condition, so your doctor should treat you based on your symptoms. Getting a VUS result is not uncommon and may be reclassified if we find out new information about it.

Variation

Variation in biology is talking about the differences between individuals. In terms of genetics, variation is talking about differences in DNA sequences that can be passed from parents to their children.