

## A glossary explaining genetic and medical terms in plain English

Finding out more about genetics and the NHS can involve lots of jargon and technical words. This is a guide to some of these words and terms to help with understanding. When words in the explanation section are in bold text, it means you can find an explanation of them in the glossary.

### Genetic terms

Please note that this glossary only includes basic terms relating to genetics that you may wish to use in accessible information for people who are new to this topic.

Term	Explanation
<b>Autosomes</b>	The set of <b>chromosomes</b> that are the same in males and females. We have 22 pairs of autosomes in almost all of the cells in our body.
<b>Benign variant</b>	A genetic <b>variant</b> that does not contribute to a particular condition. We have thousands of benign variants that are responsible for making us all different.
<b>Blood test</b>	Testing a sample of blood, typically for the <b>diagnosis</b> of illness or to predict the chance of someone developing an illness in the future.
<b>Carrier</b>	An unaffected individual that carries an alteration in one of their two copies of a <b>gene</b> that is linked to a <b>recessive condition</b> .
<b>Chromosome</b>	Structures of <b>DNA</b> that contain our genetic information, made up of many <b>genes</b> . Humans have 22 pairs of <b>autosomes</b> and one pair of <b>sex chromosomes</b> .
<b>Chromosomal condition (often referred to as a disorder)</b>	A condition caused by an unexpected <b>chromosome</b> change. The chromosome change may have been inherited from a parent, or it may have happened for the first time when a baby was conceived, or during early development.
<b>Clinical trial</b>	Research studies evaluating the effectiveness and safety of medications or medical devices by monitoring their effects on large groups of people.
<b>Complex condition (often referred to as a disorder)</b>	Condition caused by the combined effect of many different <b>genes</b> , which often also interact with environmental and lifestyle factors such as diet. Also known as <b>multifactorial</b> or <b>polygenic</b> conditions.
<b>Developmental delay</b>	When a child takes significantly longer to reach certain developmental milestones than would be expected for their age (for example, rolling over, sitting up, walking and talking). Some children might not reach these milestones at all.
<b>Developmental regression</b>	When a child begins to lose previously acquired developmental milestones (for example, they can no longer roll over, sit up, walk or talk).

Term	Explanation
<b>Diagnosis</b>	Identifying that an individual has a particular health condition by considering their signs and symptoms, and in some cases the findings from <b>diagnostic tests</b> or <b>scans</b> .
<b>Diagnostic odyssey</b>	A term used to describe the journey from initial signs or onset of symptoms through to disease recognition and a final <b>diagnosis</b> .
<b>Diagnostic test</b>	Tests used to gather clinical or genetic information for the purpose of making a <b>diagnosis</b> .
<b>Disorder</b>	There is a clear preference from people living with genetic conditions and their families to use the word 'condition' rather than 'disorder'. We have included the term 'disorder' here because it is regularly used by health professionals and in medical literature.
<b>DNA</b>	The molecule that encodes genetic information. Like an instruction manual, our code is written in DNA.
<b>Dominant condition</b>	Condition caused when one of two copies of a particular gene contain an alteration that stops the gene working correctly.
<b>Gene</b>	A length of <b>DNA</b> that codes for a certain characteristic.
<b>Genetic counselling</b>	A service provided by trained healthcare professionals that offers information, support and advice about genetic conditions.
<b>Genetic condition</b>	A condition relating to or involving <b>genes</b> or <b>chromosomes</b> .
<b>Genetic testing</b>	A medical test that can identify an altered <b>gene</b> or <b>chromosome</b> that causes a genetic condition.
<b>Genetics</b>	The study of <b>genes</b> , genetic variation and inheritance of genetic traits.
<b>Genome</b>	The entire genetic material of an organism.
<b>Genotype</b>	The particular combination of genetic material that an individual has. If the genetic material contains one or more gene changes, this could increase the chance of the individual, or their children, developing a genetic condition.
<b>Multifactorial condition (often referred to as a disorder)</b>	See <b>complex condition</b> .
<b>Non-genetic condition</b>	A condition not relating to or involving <b>genes</b> or <b>chromosomes</b> .
<b>Pathogenic variant</b>	A genetic <b>variant</b> that results in a particular condition.
<b>Polygenic condition</b>	See <b>complex condition</b> .

Term	Explanation
(often referred to as a disorder)	
<b>Predictive test</b>	Testing used to identify genetic changes linked with a condition before a person shows symptoms.
<b>Rare condition</b>	A condition that affects fewer than 1 in 2,000 people.
<b>Recessive condition</b>	Condition caused when two copies of a particular gene contain an alteration that stops the gene working correctly.
<b>Reproductive options</b>	If you or a family member has a serious <b>genetic condition</b> , you may be concerned about the possibility of passing it on to your children. There are reproductive options available to help manage this situation and <b>genetic counselling</b> can help you make informed choices.
<b>Saliva test</b>	Testing a sample of saliva (commonly referred to as spit) to identify markers of a particular condition.
<b>Scan</b>	A detailed examination of inside the body or a part of the body using techniques such as x-ray, ultrasound, computerised tomography (CT) or magnetic resonance imaging (MRI).
<b>Sex chromosomes</b>	The <b>chromosomes</b> that determine your sex, XX in females and XY in males.
<b>Single gene condition (often referred to as a disorder)</b>	A genetic condition caused by only one <b>gene</b> .
<b>Syndrome without a name (SWAN)</b>	Sometimes an individual is suspected to have a genetic condition, but <b>genetic testing</b> has so far failed to identify the change in their <b>DNA</b> that has caused it. Sometimes the change is so rare it has never been seen before, or a change has been found but it is not yet possible to confirm if it has caused their condition (a <b>variant of uncertain significance</b> ). These are called undiagnosed conditions or syndromes without a name (SWAN).
<b>Tissue test</b>	The removal of a small amount of tissue which is then analysed in a laboratory to identify a particular condition.
<b>Ultra rare condition</b>	A condition that affects fewer than 1 in 50,000 people.
<b>Variant</b>	A variant is a change to a <b>gene</b> that could affect how it works. Some changes might not affect the way the gene works at all, other changes could be very significant for a person's health and lead to a genetic condition.
<b>Variant of uncertain significance (VUS)</b>	Sometimes a <b>variant</b> is found that scientists do not yet understand: it is not clear if the change to the <b>gene</b> is harmless, or if it will lead to a genetic <b>diagnosis</b> in future. There is more to learn about the variant, and we don't yet have enough information to be sure.

Term	Explanation
<b>Whole genome sequencing (WGS)</b>	Sequencing of an individual's entire genome and testing for a wide range of <b>variants</b> in a large number of <b>genes</b> at the same time.
<b>X-linked condition</b>	This type of condition is caused by an alteration in a gene carried in the X-chromosome. If an individual carries this type of gene alteration, their sex (whether they are female or male) may influence how likely they are to develop symptoms, or how severe their symptoms may be.

## Healthcare professional titles

Having a genetic condition might mean having to see a number of different healthcare professionals. Here is a guide to their roles.

<b>Allied health professional (AHP)</b>	A term for a health professional who is not a doctor or nurse. For example, physiotherapists, occupational therapists, and speech and language therapists.
<b>Audiologist</b>	A health professional who diagnoses, treats, and helps manage a hearing or balance condition.
<b>Clinical Geneticist</b>	A doctor based in a hospital with advanced training and clinical training in genetics.
<b>Clinical nurse manager</b>	A nurse responsible for the management of a ward or a unit.
<b>Clinical nurse specialist</b>	A nurse who has undertaken specialist training to become an expert in one area of healthcare.
<b>Clinical support worker (or auxiliary nurse)</b>	Assists registered nurses with patient care.
<b>Consultant</b>	A consultant is a senior doctor who practises in one of the medical specialities.
<b>District Nurse</b>	A nurse who looks after patients in the community.
<b>General Practitioner (GP)</b>	A doctor based in the community who treats patients with minor or chronic illnesses and refers those with serious conditions to a hospital.
<b>Genetic Counsellor</b>	A health professional trained to provide support, information and advice about genetic conditions.
<b>Genetic Nurse</b>	A registered nurse with a special education and training in genetics.
<b>Health Visitor</b>	A qualified and registered nurse or midwife who has chosen to gain additional training and qualifications. They mainly work in the

	community with children from birth to five years old and their families.
<b>Learning Disability Nurse</b>	A learning disability nurse helps individuals with developmental or intellectual disabilities to ensure their health needs are met.
<b>Midwife</b>	A qualified health professional who cares for women throughout pregnancy, birth, and during the postnatal period, as well as caring for newborn babies.
<b>Occupational Therapist (OT)</b>	A health professional who promotes health and wellbeing through the use of particular activities as an aid to self-management of a condition. OTs can help individuals with genetic conditions to develop gross and fine motor skills and provide equipment.
<b>Paediatrician</b>	A doctor specialising in the physical, mental and social health of children from birth to young adulthood.
<b>Pharmacist</b>	A health professional who is an expert in medicines and their use. They advise medical and nursing staff and provide information to patients.
<b>Physiotherapist</b>	A health professional who helps people affected by injury, illness or disability through movement and exercise, manual therapy, education and advice. They provide advice on pain management.
<b>Practice nurse</b>	A nurse who works in the community, usually in a GP surgery or local health centre.
<b>Physician Associate</b>	A health professional who works alongside doctors and surgeons providing medical care as an integral part of the multidisciplinary team.
<b>Psychiatrist</b>	A doctor who specialises in mental health.
<b>Psychologist</b>	A health professional dealing with the mind and behaviour.
<b>Specialist consultant</b>	A doctor who has completed advanced education and clinical training in a specific area of medicine.
<b>Speech and Language Therapist</b>	A health professional who addresses speech, language and communication problems. They can also help with issues relating to eating, feeding and swallowing.

## Medical specialities

Many doctors and healthcare professionals have expertise in a particular medical speciality. Here is a list and explanation of specialities you may experience.

Term	Speciality
<b>Cardiology</b>	Speciality dealing with conditions of the heart.

<b>Term</b>	<b>Speciality</b>
<b>Clinical neurophysiology</b>	Speciality dealing with the diagnosis and monitoring of neurological conditions by recording and interpreting electrical signals (for example, in epilepsy, nerve function or sleep disorders).
<b>Dermatology</b>	Speciality dealing with conditions of the skin.
<b>Endocrinology</b>	Speciality dealing with hormones and their effects on the body (for example, conditions such as diabetes).
<b>Gastroenterology</b>	Speciality dealing with conditions of the stomach and intestines.
<b>Genetics</b>	Speciality dealing with diagnosis and management of genetic conditions.
<b>Haematology</b>	Speciality dealing with conditions of the blood.
<b>Immunology</b>	Speciality dealing with conditions of the immune system.
<b>Nephrology</b>	Speciality dealing with conditions of the kidneys.
<b>Neurology</b>	Speciality dealing with conditions of the brain, spinal cord and nervous system. This can include some musculoskeletal conditions.
<b>Obstetrics and gynaecology</b>	Speciality dealing with pregnancy, childbirth, and care for the mother and baby for around the first six weeks after childbirth.
<b>Oncology</b>	Speciality dealing with prevention, diagnosis and treatment of cancers.
<b>Ophthalmology</b>	Speciality dealing with disorders and diseases of the eye.
<b>Orthopaedics</b>	Speciality dealing with bones and muscles.
<b>Otorhinolaryngology</b>	Speciality dealing with conditions of the ear, nose and throat.
<b>Paediatrics</b>	Speciality dealing with children and teenagers.
<b>Pharmacology</b>	Specialty dealing with how medicines work and how they affect our body.
<b>Podiatry</b>	Speciality dealing with foot, ankle and lower limbs.
<b>Psychiatry</b>	Specialty dealing with diagnosis, treatment and prevention of mental health conditions.
<b>Psychology</b>	Speciality dealing with the mind and behaviour.
<b>Radiology</b>	Speciality using medical imaging techniques including x-rays and scans.
<b>Respiratory medicine</b>	Speciality dealing with conditions of the respiratory system (breathing) and lungs.

Term	Speciality
<b>Rheumatology</b>	Speciality dealing with conditions that affect joints, tendons, ligaments, bones and muscles, including many types of arthritis.
<b>Urology</b>	Speciality dealing with conditions of the urinary (commonly known as wee) system.
<b>Vascular</b>	Speciality dealing with the circulatory system (which conveys blood around the body) and lymphatic system (which conveys water and other essentials around the body).

## Information from [Genomics England](#)

What is genomic or genetic testing? ([podcast](#) and [explainer video](#))

What is a genome? ([podcast](#) and [explainer video](#))

What is whole genome sequencing? ([podcast](#) and [explainer video](#) + [blog](#))

What's the difference between DNA and RNA? ([podcast](#) and [explainer video](#) + [blog](#))

What is a variant of uncertain significance? ([podcast](#) and [explainer video](#))

What is a rare condition? ([podcast](#) and [explainer video](#) + [blog](#))