Sequencing Success: Genomics for cancer research and care

CANCER RESEARCH UK

Glossary

Biomarker tests	A laboratory process that checks for certain genes or biomarkers that are a sign of disease, using a biological sample from the patient (e.g. tissue, blood). Test results can be used to inform precise diagnoses and treatment decisions. **Companion diagnostic tests** are tests that inform decisions about precision (personalised) treatments.
Biomarkers	A **biological marker** or characteristic that can be used to measure what's happening in the body - indicating the presence/severity of a disease or how someone is reacting to a treatment.
BRCA 1/2	**BReast CAncer gene 1** (*BRCA1*) and **Breast CAncer gene 2** (*BRCA2*) are genes involved in DNA repair. A variant in either of these genes reduces the body's ability to repair DNA, increasing the risk of developing breast and ovarian cancer, as well as other cancers.
Cancer vaccine	There are different types of **cancer vaccines**. Cancer treatment vaccines help your body's immune system recognise and attack cancer cells. They are a type of immunotherapy.
ctDNA	Circulating tumour DNA (or ctNDA) refers to cell-free DNA fragments derived from cancer cells.
Genetic variants (somatic, germline, constitutional, pathogenic)	A variant is any difference between the DNA sequence of two individuals' genomes or a reference genome. Variants can be harmless or disease-causing (pathogenic). Both germline and somatic variants have been implicated in many cancers. **Germline or constitutional variants** are present in all cells of a person's body, including egg and sperm cells. They can be passed from parent to offspring. **Somatic variants** are present only in specific cells and are generally not hereditary. These variants are acquired at some point during a person's lifetime, often due to errors during cell division.

	Pathogenic variants are changes in the DNA sequence that cause a person to have or be at increased risk of developing a certain disease, such as cancer. Knowing if a person has a pathogenic variant may help prevent, diagnose and treat the disease. Not everyone who has a pathogenic variant will develop the disease. (Also referred to as deleterious mutation, disease-causing mutation, predisposing mutation, and susceptibility gene mutation).
Genomic healthcare / medicine	**Genomic medicine** uses insights from a human's complete set of DNA (genome) to inform their healthcare. **Genomic healthcare** combines the science of genomics with information about a person's health to improve diagnosis and treatment.
Genomic profiling	**Genomic profiling** is analysis of an individual's DNA to identify genetic variations related to health, traits, or disease risk.
Genomic research	**Genomic research** is the study of the genome and its relationship with health and disease.
Genomics England	Set up by the Department of Health and Social Care to deliver the UK Government's 100,000 Genomes Project, **Genomics England** now works with NHS England to improve genomic testing for NHS patients, facilitate Whole Genome Sequencing (WGS) and store WGS data.
Genomics Partnership Wales	Partnership of stakeholders aiming to harness the potential of genomics to improve the health, wellbeing, and prosperity of the people of Wales. Member stakeholders include: **All Wales Medical Genomics Service (AWMGS)**: Hosted by Cardiff and Vale University Health Board at the University Hospital of Wales (UHW) in Cardiff, it provides clinical and laboratory genetics services to the population of Wales. **Wales Gene Park**: Fosters genetic and genomic research and translates innovative findings into the NHS. **Pathogen Genomics Unit**: Applies academic approaches to genomics in clinical settings.

GeNotes	**GeNotes** (Genomic Notes for clinicians) is a service set up by NHS England's National Genomics Education Programme which aims to provide quick, concise information to help healthcare professionals make the right genomic decision at each stage of the clinical pathway.
Germline genetic testing	Testing on non-cancerous cells that looks for variants associated with hereditary cancer risk, present in all cells of the body since birth.
GPwER / GPwSI	**General Practitioners with an Extended Role** (GPwER) [previously known as General Practitioners with a Special Interest (GPwSI)] are GPs who have undertaken a role that is beyond the scope of GP training and requires further training. The Royal College of General Practitioners does not accredit GPwER but supports its members to develop extended roles by providing best practice frameworks in areas we know are of interest to members, this includes a framework for GPwER in genomics.
Inherited risk	Genes are passed on from parent to child (inherited) during conception. If someone has a cancer-associated gene variant such as *BRCA1/2,* there is a chance they could pass it on to their child. Not everyone with a cancer-associated gene variant will go on to develop cancer, but they will be at higher risk of doing so.
International Standard Randomised Controlled Trial Number (ISRCTN)	**ISRCTN** is a publicly accessible clinical trial registry that accepts all research studies.
Large panel testing	**Large panel testing** looks at multiple genes at once. In the cancer context, relevant genes for a specific cancer type may be analysed together, or some laboratories use large panels with around 500 cancer genes as a standard.
Liquid biopsy	A test that analyses components of a liquid sample, such as blood or urine to detect the presence of cancer-related biomarkers.
Lynch syndrome	A hereditary cancer syndrome, associated with an increased lifetime risk of developing colorectal, endometrial and other cancers.
National Disease Registration Service (NDRS)	An NHS organisation that collects patient data on cancer, congenital anomalies and rare diseases. They provide analysis to support clinical teams, academics,

	charities and policy makers. Genetic data is collected from GLHs.
Next generation sequencing (NGS)	**Next-generation sequencing** is a collective name for technology that has led to faster and cheaper DNA sequencing.
NHS England Genomics Unit	The **Genomics Unit** is the national team with strategic oversight of commissioning the Genomic Medicine Service in England, funding and performance monitoring.
NHS Genomics Medicine Service (GMS)	The **GMS** is the arm of NHS England that delivers genomic testing and analysis. The following make up the NHS Genomics Medicine Service (GMS) in England: **NHS Genomic Laboratory Hubs (GLHs)**: Network working regionally across England to deliver the genomic tests on the National Test Directory to the NHS, alongside local laboratories. **NHS Genomic Medicine Service Alliances (GMSAs)**: Responsible for locally embedding genomics into routine care and ensuring equitable access to genomic tests for eligible patients in their region. **NHS Genomic Networks of Excellence (GNEs):** Partnerships of NHS, academia, third sector and industry that develop evidence and adoption processes for new genomic innovations and technology applications that will benefit patients. **National Genomic Test Directory (NTD)**: Details all genomic tests commissioned for rare diseases and cancer in England.
Non-WGS data	Genetic data that is just a segment of the genome, rather than the whole genome (WG).
Our Future Health	A research programme funded collaboratively between the public, charity and private sectors in the UK, looking at new ways to detect, treat and prevent a variety of diseases.
Penetrance	Penetrance measures how likely a gene variant is to cause a disease. A **high penetrance variant** is one where almost all individuals with this variant will go on to express the disease.

Pharmacogenomics	Combines pharmacology with genomics to use someone's genomic information to identify which drug will be most effective for them.
Precision (personalised) medicines / therapies	Choosing treatments to more specifically target someone's unique genetics or tumour profile.
Precision diagnostics	**Precision diagnostic** tools can be used to assess cancer risk, characterise and diagnose disease, and monitor long-term disease burden based on someone's unique genetics or tumour profile.
Scottish Strategic Network for Genomic Medicine (SSNGM)	Leadership and strategic oversight of genomics delivery in Scotland.
Scottish Test Advisory Group (SG-TAG)	Two groups sitting within the SSNGM that respectively advise on which genomic tests should be available on the Cancer and Rare and Inherited Diseases directories based on clinical viability and value for money.
Secure Data Environments (SDEs)	A term often used interchangeably with TRE. NHSE is currently setting up a network of SDEs in England.
Single panel testing	**Single panel testing** looks at a single gene, often when a clinician wishes to confirm or rule out a specific diagnosis or there is a family history of a certain genetic variant.
Standard of care testing	Genomic testing considered the proper and best practice by healthcare professionals.
Translational research	**Translational research** converts scientific discoveries into practical medical applications to improve health outcomes.
Trusted Research Environments (TREs)	These are highly secure computing environments where data is stored and then accessed by researchers. They are widely accepted to be the safest way to store and access health data. Examples include, Genomics England's Research Environment, UK Biobank and CRUK's TRE.
UK Biobank	A large-scale biomedical study and resulting database that can be accessed globally by approved researchers.
Welsh Health Specialised Services Committee (WHSSC)	WHSSC is responsible for planning of Specialised and Tertiary services to ensure the people of Wales have fair and equitable service access.
Whole Genome Sequencing (WGS)	Sequencing close to the entire genome, including both coding and non-coding DNA. WGS can capture information that might be missed with panel testing.