Department of Health & Social Care

Rare Diseases Glossary

Glossary of commonly used terms and rare diseases initiatives

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Glossary of commonly used terms and rare diseases initiatives

Term	Definition
100,000 Genomes Project	A project that will sequence 100,000 genomes from around 70,000 people, the aim of which is to create a new genomic medicine service for the NHS, transforming the way in which people receive care.
All Wales Medical Genetics Service (AWMGS)	The All Wales Medical Genetics Service (AWMGS) provides specialist genetic services to individuals and families with, or concerned about, rare genetic conditions. The service is made up of clinical and laboratory services which together provide medical genetics services to the population of Wales.
All Wales Medicines Strategy Group (AWMSG)	The All Wales Medicines Strategy Group (AWMSG) was established in 2002 to provide advice on medicines management and prescribing to Welsh Government in an effective, efficient and transparent manner.
Care Opinion	Care Opinion (previously Patient Opinion) is a not-for-profit social enterprise that provides an online feedback service enabling people in Scotland to give real-time feedback, and engage in constructive dialogue with healthcare service providers about the services they, their families, or the people they care for, have received.
Congenital Anomaly Register and Information Service (CARIS)	CARIS was established in 1998 with the objective of assessing patterns of anomalies in Wales, including possible clusters and their causes. Information from CARIS informs planning of wider health services, including screening services.
Cross–border healthcare directive	This is an EU directive which sets out the conditions under which a patient may travel to another EU country to receive medical care and reimbursement. It covers healthcare costs, as well as the prescription and delivery of medications and medical devices. A directive is a legal act of the European Union, which requires member states to achieve a particular result without dictating the means of achieving that result.
Chief Medical Officer (CMO)	The Chief Medical Officer (CMO) acts as the UK government's principal medical adviser and the professional head of all directors of public health in local government.
Commissioning	Commissioning is the process used by health services and local authorities to: identify the need for local services; assess this need against the services and resources available from public, private and voluntary organisations; decide priorities; and set up contracts and service agreements to buy services. As part of the commissioning

	process, services are regularly evaluated.
Diagnostic Odyssey Task & Finish Group	A sub-committee of the UK Rare Disease Policy Board whose objective is to establish a process that will measure the time travelled in the diagnostic pathway for patients with particular rare diseases.
Discovery forum	Genomics England works with industry through its Discovery Forum. Created in July 2017, the Forum builds on the work of the GENE Consortium. The Discovery Forum provides a platform for collaboration and engagement between Genomics England, industry partners, academia, the NHS and the wider UK genomics landscape.
European Bioinformatics Institute (EBI)	The European Bioinformatics Institute (EMBL-EBI) forms part of EMBL, Europe's flagship laboratory for the life sciences, and shares data from life science experiments, performs basic research in computational biology and offers an extensive user training programme, supporting researchers in academia and industry.
European Reference Network	An organisation that help professionals and centres of expertise in different EU countries to share knowledge.
Exome Sequencing	The exome corresponds to a person's entire DNA that codes for proteins only – called genes. Only 2% of a person's DNA codes for proteins. Exome sequencing involves reading the protein-coding sections of a person's DNA.
Chief Medical Officer annual report 2016: Generation Genome	The report discusses the current state of genomic service provision in the NHS in England. It explores the potential of genomics to improve health and prevent ill-health. The report presents evidence and discourse around: screening; diagnosing rare diseases and the use of genomics in personalised prevention It makes recommendations to address gaps and widen access to genomic services aimed at Government, health system organisers, regulators, research and medical training bodies and healthcare professionals.
Genetic Alliance UK	A national charity working to improve the lives of patients and families affected by all types of genetic conditions.
Genomic Medicine Centre	One of 13 providers across England that are delivering the 100,000 Genomes Project.
Genomics Education Programme	The Genomics Education Programme (GEP) is the NHS's method of ensuring their staff have the knowledge, skills and experience to ensure that the health service remains a world leader in genomic and precision medicine – particularly for NHS England Genomic Medicine Centres (GMCs) and the contribution to the 100,000 Genomes Project.
Genomics England	The organisation set up to deliver the 100,000 Genomes Project.

Genomics England Clinical Interpretation Partnership	Researchers and clinicians, from both academia and the NHS, are working together to analyse data from the 100,000 Genomes Project. They are joined up through the Genomics England Clinical Interpretation Partnership (GeCIP).
Genomics England PanelApp	PanelApp is a knowledge base curated by Genomics England. This tool has information on thousands of genes that may be linked to rare diseases, as reported by expert doctors and researchers.
Health Data Finder for Research	The Health Data Finder for Research enables researchers to find information about the UK healthcare data sets that are available for research and to direct to the relevant data custodian experts to request access to these data sets. The aim of the Health Data Finder for Research is to help navigate the UK health data landscape.
Health Education England (HEE)	Health Education England (HEE) is the national leadership organisation for education, training and workforce development in the health sector.
Health Technology Appraisal	Health technology appraisals are recommendations on the use of new and existing medicines and treatments within the NHS.
Health Technology Wales (HTW)	Health Technology Wales (HTW) has been established to deliver a strategic, national approach to the identification, appraisal and adoption of new health technologies into health and care settings across NHS Wales.
Health Research Authority (HRA)	The HRA protects and promotes the interests of patients and the public in health research. They are one of a number of organisations that work together in the UK to regulate different aspects of health and social care research.
INVOLVE	INVOLVE was established in 1996 and is part of, and funded by, the National Institute for Health Research, to support active public involvement in NHS, public health and social care research. It is one of the few government funded programmes of its kind in the world.
Life Science Industrial Strategy (LSIS)	Published in August 2017, LSIS sets out recommendations for UK's world leading life sciences industry to drive growth, increase productivity, improve the use of data, reinforce our science base, deepen our skills and secure benefits for patients throughout the United Kingdom.
Life sciences: Sector Deal	The Life Sciences Sector Deal will help ensure new pioneering treatments and medical technologies are produced in the UK, improving patient lives and driving economic growth. The deal involves substantial investment from private and charitable sectors and significant commitments in research and development from the Government. https://www.gov.uk/government/publications/life-sciences-sector-deal
Medical Research Council (MRC)	The Medical Research Council (MRC) is a national funding agency dedicated to improving human health by supporting research across the

	entire spectrum of medical sciences, in universities and hospitals, in MRC units, centres and institutes in the UK, and in MRC units in Africa.
Medicines and Healthcare products Regulatory Agency (MHRA)	The Medicines and Healthcare products Regulatory Agency (MHRA) is responsible for protecting and promoting public health and patient safety by ensuring that medicines, healthcare products and medical equipment meet appropriate standards of safety, quality, performance and effectiveness, and are used safely.
National Centre for Biotechnology Information (NCBI)	The National Centre for Biotechnology Information provides access to biomedical and genomic information. https://www.ncbi.nlm.nih.gov/
National Congenital Anomaly and Rare Disease Registration Service	The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) records those people with congenital abnormalities and rare diseases across the whole of England. PHE has expanded congenital anomaly and rare disease registration to cover the whole population of England, to meet national requirements for high quality public health disease surveillance identified by the Chief Medical Officer. The creation of the NCARDRS is part of the UK Rare Disease Strategy and the Department of Health 2020 Vision on Rare Diseases.
National Genomics Board (NGB)	The National Genomics Board (NGB), chaired by a Minister, will facilitate collaboration and effective delivery of key actions. Its vision is 'to make sure that the UK remains the world's leading centre for genomic medical research, and to leverage this position to deliver quantifiable benefits for NHS patients and for the life sciences sector.' The NGB will oversee the implementation of the Generation Genome report; implement the genomics component of the Life Sciences Industrial Strategy and the Life Sciences Sector Deal; establishment of the NHS Genomics Service; to articulate the "genomics dream" of faster and better diagnosis and treatment - especially for patients with rare diseases and cancer - and propagate economic growth.
National Institute for Health Research (NIHR)	The National Institute for Health Research funds health and care research and translate discoveries into practical products, treatments, devices and procedures, involving patients and the public. NIHR ensures that the NHS is able to support the research of other funders to encourage broader investment in, and economic growth from, health research. NIHR works with charities and the life sciences industry to help patients gain earlier access to breakthrough treatments and trains and develops researchers to keep the nation at the forefront of international research.
National Institute for Health and Care Excellence (NICE)	The National Institute for Health and Care Excellence (NICE) in England provides national guidance and advice to improve health and social care. NICE has responsibility for the development of guidance for highly specialised technologies (HST). NICE's HST programme determines

	whether selected very rare disease treatments should be recommended for NHS-wide commissioning in England.
National Services Division	The National Services Division is responsible for commissioning and performance managing National Screening Programmes, Specialist Clinical Services and National Managed Clinical Networks on behalf of NHS Scotland.
Newborn Blood Spot Screening programme	Every baby is offered newborn blood spot screening, also known as the heel prick test, ideally when they are five days old. Newborn blood spot screening involves taking a blood sample to find out if your baby has one of nine rare but serious health conditions.
NIHR Biomedical Research Centres	Formed through partnerships between England's leading NHS organisations and universities, 20 NIHR Biomedical Research Centres (BRCs) conduct translational research to transform scientific breakthroughs into life-saving treatments for patients. Staffed by expert investigators and clinicians, NIHR BRCs are leaders in translating lab-based discoveries into new cutting edge treatments, technologies, diagnostics and other interventions in clinical settings.
NIHR BioResource	The NIHR BioResource is a panel of thousands of volunteers, both with and without health problems, who are willing to be approached to participate in research studies and trials on the basis of their phenotypic and genetic make-up to develop new treatments for a range of diseases and investigate the links between genes, the environment, health and disease.
NIHR BioResource – Rare Diseases	The NIHR BioResource – Rare Diseases is enrolling rare disease patients to the BioResource using a national consent model across 60 NHS Trusts.
NIHR BioResource for Translational Research for Common and Rare Diseases	During the 2017 to 2018 period, the NIHR BioResource, NIHR BioResource - Rare Diseases and the NIHR RD-TRC are being integrated. This will result in a newly formed NIHR BioResource for Translational Research for Common and Rare Diseases. This BioResource will provide a nationally accessible resource of volunteers from the general population as well as patients with common and rare diseases.
NIHR Office for Clinical Research Infrastructure (NOCRI)	The NIHR Office for Clinical Research Infrastructure (NOCRI) provides potential partners, including the life sciences industry and charities, with a direct and simplified route to a wide range of experimental medicine facilities and expert NIHR investigators. NOCRI works with organisations to help navigate this infrastructure and, where required, form partnerships and collaborations to bring new treatments to patients faster.
NIHR Rare Diseases Translational Research Collaboration (RD-TRC)	The Rare Diseases Translational Research Collaboration (RD-TRC) provides: research infrastructure to support fundamental discoveries and translational research on rare diseases; support for increasing research collaborations which lead to improved diagnosis; treatment and care; and, support for deep phenotyping (defined as the precise and comprehensive analysis of phenotypic abnormalities) of people with rare

	diseases.
Northern Ireland Patient & Client Council	The Patient and Client Council provides a powerful, independent voice for patients, clients, carers and communities on health and social care issues in Northern Ireland.
Northern Ireland Rare Disease Partnership (NIRDP)	The Northern Ireland Rare Disease Partnership (NIRDP) are a not for profit organisation bringing together those living with a rare disease and organisations representing them; clinicians and other health professionals; researchers and producers of specialist medicines and equipment; health policy makers and academics.
Orphanet	A reference source of information on rare diseases located at www.orpha.net
Office for Strategic Coordination of Health Research (OSCHR)	The Government currently funds health related research through two main routes, the Medical Research Council (MRC) and the National Institute for Health Research (NIHR). The 'Review of funding in UK health research' led by Sir David Cooksey in 2006 resulted in the creation of the independent Office for Strategic Coordination of Health Research (OSCHR) to 'monitor' the translational research activity funded by Government.
Patient Empowerment Group (PEG)	The Patient Empowerment Group (PEG) was established by the Rare Disease UK campaign to help monitor the implementation of the Strategy. The purpose of the group is to ensure that the patient voice is properly informed and effectively represented in the implementation of the Strategy.
Project ECHO	Project ECHO links expert inter-professional teams at an academic hub with primary care providers in local communities. The Project ECHO model was piloted in Northern Ireland with a Vasculitis patient cohort.
Public Health Agency (PHA)	The Public Health Agency (PHA) was established in April 2009 as part of the reforms to Health and Social Care (HSC) in Northern Ireland. The PHA is the major regional organisation for health protection and health and social wellbeing improvement. Their role also commits them to addressing the causes and associated inequalities of preventable ill-health and lack of wellbeing.
Public Health England (PHE)	Public Health England's (PHE) mission is to protect and improve the nation's health and wellbeing, and reduce health inequalities. They are therefore responsible for supporting local authorities and the NHS to plan and provide health and social care services such as immunisation and screening programmes, and to develop the public health system and its specialist workforce, amongst many other priorities.
Quality-adjusted life year (QALY)	A quality-adjusted life year (QALY) is a measure of the state of health of a person or group in which the benefits, in terms of length of life, are adjusted to reflect the quality of life. One QALY is equal to 1 year of life in perfect health.

Rare disease	The European definition of a rare disease is a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000. Rare diseases often require special efforts to co-ordinate care given by many different specialists and agencies.
Rare Diseases Advisory Group (RDAG)	The Rare Diseases Advisory Group (RDAG) is an NHS England committee that makes recommendations to NHS England and the devolved administrations of Scotland, Wales and Northern Ireland on developing and implementing the strategy for rare diseases and highly specialised services.
Rare Disease Collaborative Network / Centre (NHSE)	A recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes.
Rare Disease Implementation Oversight Group	The role of the Rare Disease Implementation Oversight Group (RDIOG) is to monitor the implementation of the Scottish Rare Disease Plan 'It's Not Rare to Have a Rare Disease' and to ensure that the 51 commitments in the Strategy are being met.
Rare Disease Policy Board	The Rare Diseases Policy Board is a committee of the Department of Health and Social Care in England that has responsibility for the coordination of policy development and meeting the commitments set out in the Strategy.
Rare Disease UK	The national campaign for people with rare diseases and all who support them.
Scottish Genomes Partnership (SGP)	The Scottish Genomes Partnership is a major Scotland-wide research programme between the Universities of Edinburgh, Glasgow, Aberdeen and Dundee, with NHS Scotland, NHS Lothian, NHS Greater Glasgow & Clyde, NHS Grampian and NHS Tayside. The SGP is a collaboration which is capitalising on investment in whole genome sequencing technology.
UK Clinical Trials Gateway (UKCTG)	The UK Clinical Trials Gateway (UKCTG) website pulls through information about clinical trials and other research from several different UK registers. https://www.ukctg.nihr.ac.uk/
UK Genomic Knowledge Base	The growing sum of knowledge being accrued as a result of the 100,000 Genomes Project when taken together with the existing research base.
UK National Screening Committee	The UK NSC advises ministers and the NHS in the 4 UK countries about all aspects of screening and supports implementation of screening programmes.
UK Rare Disease Forum	The UK Rare Disease Forum is a large stakeholder group, primarily focussing on the provision of stakeholder insight and advice on key issues, challenges and risks to delivery of the 51 commitments in the Strategy

UK Strategy for Rare Diseases	The UK Strategy for Rare Diseases was published in 2013 and contains over 50 commitments to ensure people living with a rare disease have access to the best evidence-based care and treatment that health and social services, working with charities, researchers and industry can provide.
Wales Gene Park	The Wales Gene Park is an infrastructure support group funded by Welsh Government through Health and Care Research Wales. Their mission is to: promote and facilitate Welsh medical genetic and genomic research and its application to improve health and wealth in Wales; and, to engage the public and health professionals to improve understanding of the opportunities and challenges arising through genetics and genomics.
Welsh Health Specialised Services Committee (WHSSC)	The Welsh Health Specialised Services Committee (WHSSC) is responsible for the joint planning of Specialised and Tertiary Services on behalf of Local Health Boards in Wales.
Welsh Rare Diseases Implementation Group (RDIG)	The implementation of the 'Welsh Implementation plan for Rare Diseases' is supported by a national Implementation Group which has representatives from every health board in Wales and other key stakeholder organisations.
Welsh Rare Disease Patient Network	In collaboration with the Wales Gene Park, Rare Disease UK established the Welsh Rare Disease Patient Network to engage patients, families and patient organisations to ensure the patient voice is properly informed, and effectively represented in the discussion and development of the implementation of the Strategy.
Whole Genome Sequencing (WGS)	A range of techniques aimed at determining the complete DNA sequence of an organism.