RARE DISEASE
CLINICAL TRIAL NETWORK

HRCI Health Research Charities Ireland

HRB Health Research Board

UCD DUBLIN

Glossary of Terms

We want to make sure everything we say is easy to understand, so we try not to use too much complicated language or abbreviations. But, sometimes, they might pop up. That's why we put together this list of common abbreviations, if anything still doesn't make sense, just ask!

This glossary was co-created with insights from the UCD Clinical Research Centre and individuals living with rare diseases. We extend our appreciation to the following entities whose contributions inspired this compilation: Health Research Charities Ireland, Health Research Board, and Prof. Rachel Crowley.

BME	Black & Minority Ethnic
CHI	Children's Health Ireland
COI	Conflict of Interest
CRISPR	CRISPR gene editing
CTI	Cancer Trials Ireland
DoH	Department of Health
ECRIN	European Clinical Research Infrastructure Network
EDI	Equality, Diversity And Inclusion
EHR	Electronic Health Record
EJPRD	European Joint Programme on Rare disease
EMA	European Medicines Agency
EPAG	European Patient Advocacy Group
ERICA	European Rare Disease Research Coordination and Support Action
ERDERA	European Rare Diseases Research Alliance
ERN	European Reference Networks
EUPATI	European Patients' Academy on Therapeutic Innovation
EURORDIS	European Organisation for Rare Diseases
EuRRECa	European Registries for Rare Endocrine Conditions (covers EndoERN and ERN BOND)
GDPR	General Data Protection Regulation
HIQA	Health Information and Quality Authority
HPRA	Health Products Regulatory Authority
HRB	Health Research Board
HRCI	Health Research Charities Ireland
HSE	Health Service Executive
IP	Intellectual property
IPHA	Irish Pharmaceutical Healthcare Association

IPPOSI	Irish Platform for Patient Organisations, Science and Industry
IRC	Irish Research Council
IRDiRC	International Rare Diseases Research Consortium
MMUH	Mater Misericordiae University Hospital
NCTO	National Clinical Trials Office
NGS	Next Generation Sequencing
NIHR	National Institute for Health Research (UK)
NIRDP	Northern Ireland Rare Disease Partnership
NRDO	National Rare Disease Office
NRDP	National Rare Disease Plan
NRECs	National Research Ethics Committees
ORPHA	ORPHAcodes
ORPHANET	Orphanet
PCOM	Patient Centred Outcome Measure
PI	Principal Investigator
PPI	Patient and Public Involvement
PPI Ignite	PPI Ignite National Network
PPI SLG	Patient and Public Involvement Shared Learning Group
PREM	Patient Reported Experience Measure
PROM	Patient Reported Outcome Measure
QoL	Quality of Life
RAinDRoP	Rare Disease Research Partnership
RCPI	Royal College of Physicians of Ireland
RCSI	Royal College of Surgeons of Ireland
RDCat	Rare Disease Research and Innovation Catalyst Award
RDCTN	Rare Disease Clinical Trial Network
RDD	Rare Disease Day
RDF	Rare Disease Forum
RDI	Rare Diseases Ireland
REC	Research Ethics Committee
SFI	Science Foundation Ireland
SLG	Shared Learning Group
SMAB	Scientific and Medical Advisory Board
SVUH	St Vincent’s University Hospital
TCD	Trinity College Dublin
TMRN	Trials Methodology Research Network
ToR	Terms of Reference
TUH	Tallaght University Hospital
UCD	University College Dublin
UHI	Unique Health Identifier
YPAG	Young Persons’ Advisory Group

Autosomal dominant genetic conditions	These are conditions whereby a person needs only to inherit one changed copy (alteration) of the gene in order to be affected by the condition, or become affected by the condition later in life. The changed gene is dominant over the normal gene.
Autosomal recessive genetic conditions	These are conditions whereby a person has to inherit two changed copies (alterations) of the gene (a changed copy from each parent) to be affected by the condition. A person who has only one copy of the changed gene will be an unaffected carrier.
Autosomes	We have 23 pairs of chromosomes. Pairs number 1 to 22 are called autosomes and look the same in men and women. Pair number 23 are different in men and women and are called the sex chromosomes.
Carrier of a changed gene	A person who is generally not affected with the condition (at that moment), but carries one copy of a changed gene. In the case of recessive conditions, the person will not usually be affected; in the case of dominant conditions, the person may become affected at a later stage.
Chromosome disorder	These are conditions that affect the structure of a chromosome, where a piece of chromosome material can be missing or extra or a whole chromosome is missing or extra.
Chromosome testing	This is called karyotyping and involves looking at the overall structure of the chromosomes to check for large pieces of missing or extra chromosome material. This test cannot show small or subtle changes in chromosomes.
Gene alteration or mutation	A change in a gene that is sometimes also known as a gene alteration. Sometimes when a gene is changed, its information is altered so it does not work properly. This may cause a genetic condition.
Genetic condition	A condition or disease caused by an abnormality in a gene or chromosome.
Genetic test	A test which can help identify if there is a change in a particular gene or chromosome. It is usually a blood or tissue test.
Genotype	Genotype is one of three factors that determine phenotype, along with inherited factors, epigenetic factors and non-inherited environmental factors.
Hereditary condition	One that is inherited (passed down through families).
Metabolic disorder	Occurs when abnormal chemical reactions in your body disrupt the metabolic process. When this happens, you might have too much of some substances or too little of other ones that you need to stay healthy. A metabolic disorder can either be inherited or acquired and can affect major organs of the body.
Orphan Disease and Drugs	An orphan drug is a pharmaceutical agent that has been developed specifically to treat a rare medical condition, the condition itself being referred to as an orphan disease. The reimbursement costs are often higher than other

	drugs because of high research and development costs and the relatively limited number of patients that can benefit from an ‘orphan drug’.
Patient Registry	Patient registries capture information systematically over long periods of time, at a population level (e.g. a particular disease) and with a view to answering current and future research questions
Phenotype	In genetics, the phenotype of an organism is the composite of the organism’s observable characteristics.
Predictive testing	A genetic test for a condition that may or will occur later in life. This testing is available to healthy individuals who are pre-symptomatic (no signs and symptoms of condition) but who are at risk of the condition due to their family history.
Teratogens/Teratogenic	Drugs that cause birth defects – these can be prescribed drugs with unintended side effects or the misuse of recreational drugs, including alcohol.
Ultra-Rare Disease	A disease is generally considered to be ultra-rare if it affects less than 1 in 100,000.