Glossary

Wait, what was that term again? To keep things clear, we've put together a quick reference of common abbreviations. As always, if you're unsure about anything, just ask!

CHI	Children's Health Ireland (used to be Temple Street Children's University Hospital)
CTIS	Clinical Trials Information System
CISCRP	The Center for Information and Study on Clinical Research Participation Inc aims to make education about clinical research easy to understand and useful. They want to help people learn about the research process, appreciate the value of joining clinical trials, and improve the experience for patients and their families.
Clinical Trial	A research study that tests treatments or medicines. The goal is to find out if the treatment is safe and effective for patients. Volunteers are often divided into groups, with some receiving the new treatment and others getting a standard treatment or a placebo. This helps researchers compare results and learn how well the new treatment works. Clinical trials can be hard for rare diseases for a few reasons. There may be a small number of patients, rare diseases can be complex, and symptoms can vary a lot between patients.

COE	Centre of Expertise is a location, like a hospital or research centre, that has a lot of knowledge and experience in a particular area, such as rare diseases. These centres often have experts, advanced resources, and access to the latest treatments.
COI	Conflict of Interest, can be real, perceived or potential. When a person or group has direct or indirect interest in a matter they also influence or control.
CRISPR	CRISPR gene editing is a powerful technology that allows precise changes to DNA. Many rare diseases are caused by genetic mutations, CRISPR may fix these to treat or even cure some rare diseases.
Cross-Over	A type of study where groups of participants receive two or more interventions in a specific order. All participants receive all interventions at some point, but in a different order. For example, two-by-two cross-over involves two groups of participants. One group receives medicine A first, then medicine B. The other group receives medicine B first, then medicine A. During the trial, participants "cross over" to the other medicine.
CTN	Clinical Trial Network are groups of organisations, researchers, and healthcare providers that work together to conduct clinical trials. This can make it easier to recruit participants, gather data, and analyse results.
CTU	Clinical Trials Unit

DoH	Department of Health is responsible for overseeing healthcare services in Ireland. This includes creating policies and managing health programs to improve the health and wellbeing of people in the country.
ECRIN	European Clinical Research Infrastructure Network is a group that helps researchers across Europe run clinical trials. It provides support for studies that take place in multiple countries, making it easier for researchers to share knowledge, resources, and data. ECRIN can help coordinate clinical trials across Europe, which is important since rare disease patients are often spread across different countries.
EDI	Equality, Diversity And Inclusion is about making sure that everyone, no matter their background, has equal opportunities and is treated fairly. It focuses on valuing differences, whether it's race, gender, disability, or other factors, and creating environments where everyone feels respected and included. In healthcare and research, EDI ensures that all groups have a voice and access to resources.
EHR	Electronic Health Records are digital versions of a patient’s medical history and health information, like doctor visits, test results, and treatments. EHRs allow healthcare providers to easily access and share up-to-date information, improving communication and coordination of care. This is especially helpful for managing complex conditions, like rare diseases, where patients may see multiple specialists and need detailed, accurate records.
EJP-RD	European Joint Programme on Rare Diseases was a collaborative group across Europe that has been replaced by European Rare Diseases Research Alliance (ERDERA)

EMA	European Medicines Agency is the organisation that evaluates and approves medicines for use in the EU. They ensure medicines are safe, effective, and of high quality before they can be sold. For rare diseases, the EMA plays a key role in reviewing and approving treatments, including rare disease drugs, often referred to as "orphan medicines," that are designed for conditions affecting small numbers of people.
ERICA	European Rare Disease Research Coordination and Support Action is an initiative aimed at improving research efforts across Europe for rare diseases. ERICA coordinates research activities, shares knowledge, and provides support for projects focused on understanding and treating rare diseases. This increases the chances of finding new treatments for rare diseases across Europe.
ERDERA	European Rare Diseases Research Alliance is a collaborative group that brings together researchers, healthcare providers, and patient organisations across Europe to focus on rare disease research. Its goal is to unite efforts, share resources, and improve research outcomes to better understand rare diseases and develop new treatments. Formerly the European Joint Programme on Rare Diseases (EJP-RD).
ERN	European Reference Networks are special groups that connect hospitals across Europe. These networks work together to help with rare and complex diseases that need very specialised care. ERNs allow doctors from different countries to talk about cases involving rare and complicated diseases. They can offer advice on the best way to diagnose and treat these conditions. Patients can’t directly contact ERNs

	themselves. But with the patient's permission, their doctors can share information and get help from the ERNs according to national health rules. There are currently 24 ERNs, as of 2024, Ireland is part of 18 of these 24 ERNs.
EUPATI	European Patients' Academy on Therapeutic Innovation is an initiative that educates and empowers patients and the public about the medicine development process. It provides training and resources to help patients understand how new treatments are developed, evaluated, and brought to market.
EURORDIS	European Organisation for Rare Diseases is an organisation that represents patients with rare diseases across Europe. It works to raise awareness, promote research, and influence policy to improve the lives of people affected by rare conditions. EURORDIS connects patient organisations, advocates for access, and aims to ensure that patient voices are heard in decision-making processes at both national and European levels.
EuRRECa	European Registries for Rare Endocrine Conditions is a network that collects and shares information about rare endocrine disorders, which affect hormone-producing glands in the body. The registry helps track patient data, improve understanding of these conditions, and support research efforts.
FDA	Food and Drug Administration, the American federal agency responsible for ensuring the safety, efficacy, and security of various products, including food, drugs, cosmetics, and medical devices.

FORT	Future of Registries Taskforce is a limited timeline project to bring together key stakeholders across the patient registry and wider health information space to discuss the sustainability of patient registries in Ireland and jointly lay out solutions for the future.
GDPR	General Data Protection Regulation is an EU law that sets rules for how personal data is collected, used, and protected. This regulation is particularly important in healthcare, where sensitive information about patients needs to be protected.
HIQA	Health Information and Quality Authority is an Irish organisation that promotes high-quality health and social care. It sets standards, monitors services, and provides guidance to ensure that healthcare providers deliver safe and effective care to patients.
HPRA	Health Products Regulatory Authority is Ireland's agency responsible for regulating medicines, medical devices, and other health products. It ensures that these products are safe, effective, and of high quality before they can be used by patients.
HRB	Health Research Board is an Irish agency that funds and supports health research.
HRCI	Health Research Charities Ireland is a network of health charities that collaborate to promote and fund health research in Ireland. HRCI advocates for the interests of patients and researchers, aiming to enhance the quality and impact of health research.

HSE	Health Service Executive is Ireland's body responsible for delivering public health and social care services. It manages hospitals, clinics, and community health programs, aiming to provide accessible, quality care to all citizens.
ICTD	International Clinical Trials Day is held each May to celebrate and raise awareness of clinical trials. This date recognises the start of the first modern Clinical Trial. The first trial was designed by James Lind and found a cure for scurvy.
IP	Intellectual property refers to inventions, designs, brands, and artistic works. IP laws protect these creations, giving creators exclusive rights to use, sell, or licence their work.
IPHA	Irish Pharmaceutical Healthcare Association is an organisation that represents the research-based pharmaceutical industry in Ireland.
IPPOSI	Irish Platform for Patient Organisations, Science and Industry is a collaborative initiative that connects patient groups, researchers, and industry representatives. Its goal is to encourage conversations and partnerships that help involve patients in health research and the development of new treatments.
IRC	Irish Research Council was an agency that promoted and funded research across various disciplines in Ireland, including humanities and social sciences. In 2024 they were combined with Science Foundation Ireland to become Taighde Éireann.

IRDiRC	International Rare Diseases Research Consortium is a A global initiative that speeds up research and development for rare diseases. It brings together researchers, patient groups, and companies to work on scientific projects, share information, and promote good practices.
JARDIN	Joint Action to support the integration of European Reference Networks is a program that helps make sure ERNs work well with each European country's health system. JARDIN creates helpful guidelines, improves patient care, and links ERNs with local health services. They also work on sharing health data more effectively and supporting medical centres that are part of ERNs.
MMUH	Mater Misericordiae University Hospital is a major hospital located in Dublin. The hospital is also involved in teaching and research.
N-Of-1	A special kind of study that focuses on one person at a time. In this trial, a patient tries a treatment for a specific period, then switches to a different treatment. The researchers can compare how well each one works for that person. This approach is helpful for rare diseases because it helps doctors see what works best for that patient. It can provide valuable information even when there aren’t enough people for a larger trial.
NCPRD	The National Clinical Programme for Rare Diseases is an initiative by the HSE in Ireland that aims to improve how rare diseases are diagnosed, treated, and managed. It creates guidelines for doctors, encourages the best ways to provide care, and helps healthcare providers work together to make sure patients get good, coordinated care.

NCTO	National Clinical Trials Office oversees all the clinical trial networks.
NGGO	National Genetics and Genomics Office is important for rare diseases because many of these conditions are caused by changes in genes. By studying genetics, the office helps doctors understand why certain rare diseases happen and how to diagnose them. This can lead to better tests and treatments for patients.
NGS	Next Generation Sequencing is a technology used to quickly and accurately determine the sequence of DNA or RNA. It is particularly valuable in research and clinical settings for personalised medicine, genetic testing, and understanding complex conditions, including rare diseases.
NIRDP	Northern Ireland Rare Disease Partnership is an organisation that brings together patients, healthcare professionals, and researchers to improve awareness, diagnosis, and treatment of rare diseases in Northern Ireland.
NRDO	National Rare Disease Office is a government body in Ireland that focuses on improving the lives of people with rare diseases. It coordinates national policies, supports research, and collaborates with healthcare providers, patient organisations, and other stakeholders.
NRDP	National Rare Disease Plan was first published in 2014 and contained 46 recommendations. These included improving diagnosis, identifying centres of excellence, and setting up dedicated rare disease treatment pathways. The group leading this work is the National Rare Disease Steering Group. In 2024,

	the Department of Health opened a call for public and patient stakeholders of rare disease to provide recommendations that would go on to form the new National Rare Disease Strategy, to be delivered in 2025.
NRDS	National Rare Disease Strategy, formed by the National Rare Disease Steering Group and informed by the 2024 Department of Health call. To be delivered in 2025.
NRECs	National Research Ethics Committees are bodies that review and oversee research proposals to ensure they meet ethical standards. In Ireland, these committees assess studies involving human participants to protect their rights, safety, and wellbeing, helping to ensure that research is conducted responsibly and ethically.
NSAGGM	National Strategy for Accelerating Genetic and Genomic Medicine is a plan in Ireland that aims to speed up the use of genetics and genomics in healthcare. It focuses on improving access to genetic testing and services, enhancing research, and helping doctors use genetic information in patient care.
ODA	Orphan Drug Act is an American law to incentivize the development of drugs for rare diseases, often referred to as "orphan drugs". It provides financial and market-based incentives to pharmaceutical companies.
OMD	Orphan Medicine Designation is a status assigned to a medicine intended for use against a rare condition. The medicine must fulfil certain criteria for designation as an orphan medicine so that it can benefit from incentives such as protection from competition once on the market.

Orphacode	Orphacode is a unique identification system used to classify and identify rare diseases. Each rare disease is assigned a specific code, making it easier for researchers, healthcare professionals, and organisations to track and share information about these conditions.
Orphanet	Orphanet is a website for people with rare diseases. It can show patients where to find expert doctors and medical centres that specialise in their condition. They offer information about support groups and organisations that can provide emotional help and useful resources.
Orphan Drug	A medicine that has been developed specifically to treat a rare 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.
PAG	Patient Advisory Group is a team made up of patients and carers who provide input and feedback on healthcare practices and policies. Their goal is to ensure that the voices of patients are heard in decisions about treatments, services, and research.
PCOM	Patient Centred Outcome Measure is a way to find out what matters most to patients in their healthcare. It focuses on how patients feel about their health, treatments, and overall quality of life. For rare diseases, PCOMs are important because they help doctors understand the specific needs and experiences of patients who may not have many treatment options.

Phase I	The first phase in a clinical trial. New treatments are tested for the first time in a small group of people to evaluate a safe dosage range and identify side effects.
Phase II	The second phase in a clinical trial. If Phase I shows they are safe, treatments are tested in a larger group to monitor for any negative effects.
Phase III	The third phase in a clinical trial. If Phase II shows they are safe, treatments are tested on even larger populations and in different regions and countries.
Phase IV	The fourth phase in a clinical trial. After the treatment is approved researchers may study long-term risks, benefits and side effects during day-to-day use in the population.
PI	Principal Investigator is the lead researcher in a study or project. They are responsible for planning, managing, and overseeing the research.
PIL	Patient Information Leaflet or “PILL” is a document that provides potential participants with clear, comprehensive information about a research study.
Platform Trial	A type of study that tests multiple treatments at the same time for a particular condition. Platform trials also make it easier to adapt and add new treatments as they are developed. This is useful for rare diseases because it allows researchers to quickly compare different treatments using the same group of patients. It can help find the best option faster since there may not be enough people for separate trials for each treatment.

PPI	Patient and Public Involvement means including patients and the public in the planning and decision-making of healthcare and research projects. This involvement can lead to better treatments and services that truly address what patients want and need.
PPI Ignite	PPI Ignite National Network is a group in Ireland that focuses on improving how patients and the public are involved in health research. It connects different organisations and supports projects that promote patient and public involvement (PPI) in decision-making.
PREM	Patient Reported Experience Measure is a tool used to gather feedback directly from patients about their experiences with healthcare services. It helps healthcare providers understand how patients feel about their care, including aspects like communication, treatment effectiveness, and overall satisfaction.
PRO	Patient Reported Outcome is a report of a patient's health status directly from the patient, without interpretation by a clinician or anyone else . It captures the patient's perspective on their experiences with illness and treatment. PROs can include information about symptoms, functional status and quality of life.
PROM	Patient Reported Outcome Measure is a tool that collects PROs (see above) from patients about their health status.

PROMIS	PROM Information System is a collection of standardized, person-centered measures developed to evaluate and monitor various aspects of health.
QoL	Quality of Life refers to how good or satisfying someone's life is. It includes factors like happiness, comfort, and the ability to do everyday activities.
RAinDRoP	Rare Disease Research Partnership is a collaboration that brings together researchers, healthcare providers, and organisations to focus on improving knowledge and non-pharmacological treatments for rare diseases.
Rare Disease	A condition that affects less than 1 in 2,000 people. A disease may be considered rare in one region of the world but not in another. In Ireland, this definition does not include rare cancers, cystic fibrosis, or infectious diseases. Rare diseases are chronic, meaning they are often serious and progressive. 80% of rare diseases are genetic. Fewer than 5% have an approved treatment.
Rare Ireland	An organisation focused on supporting individuals and families affected by rare diseases in Ireland. They work to raise awareness about these conditions, provide information, resources, and advocacy to help improve the quality of life for those impacted by rare diseases.
RCPI	Royal College of Physicians of Ireland is a professional body that trains and supports doctors in Ireland.

RCSI	Royal College of Surgeons of Ireland is a professional body and also a university that trains and supports surgeons in Ireland. It offers education and training programs for medical students and surgical residents, focusing on high standards in surgical practice and advancing research.
RCT	Randomised Control Trial is a type of scientific experiment where participants are randomly assigned to different groups (typically an experimental group receiving a new treatment and a control group receiving a standard treatment or placebo). RCTs are considered the gold standard for evaluating effectiveness and safety.
RDCat	Rare Disease Research Catalyst Consortium is a project that brings together universities, hospitals, healthcare professionals, researchers, patients, and advocacy groups in Ireland to enhance research on rare diseases. RDCat aims to help find diagnoses for those with undiagnosed rare diseases and train the next generation of researchers to understand these conditions better. Lead researcher is Rachel Crowley and the Project Manager is Daniel Mikula.
RDCTN	Rare Disease Clinical Trial Network is a group that works with experts from around the world. We help make rare disease research better by focusing on what patients need and teaching new researchers.
RDI	Rare Diseases Ireland is a group that supports people with rare diseases in Ireland. RDI helps raise awareness, provides information, and connects patients and their families to resources and support services.
REO	Regional Executive Officer (see RHA below)

RHA	Regional Health Area, as of 2024, the HSE has created six new health regions. Each region is responsible for providing both hospital and community care for the people in that area.
Registry	A registry is a list or database that collects information about people with a specific condition. It helps researchers and doctors learn more about the condition, how it affects patients, and what treatments work best.
SFI	Science Foundation Ireland was a government organisation that supported scientific research and innovation in Ireland. In 2024, they joined with the Irish Research Council to become Taighde Éireann.
SLG	Shared Learning Group is an informal meeting of people who come together to share knowledge and ideas about important topics. They focus on learning from each other's experiences and finding ways to improve practices in their fields.
SMAB	Scientific and Medical Advisory Board is a group of experts who provide guidance and advice on scientific and medical issues. Members of the board usually include doctors, scientists, and researchers who have experience in their fields. Their main goal is to ensure that the organisation follows the latest science and provides the best possible care or information.
SVUH	St Vincent’s University Hospital is a major hospital located in Dublin. The hospital is also involved in teaching and research.

SWOT	Study Within A Trial is a type of study that looks at a particular aspect of a treatment, like its effectiveness or safety. This is different than a clinical trial which looks at a treatment as a whole. In rare diseases, SWOTs can be helpful because they provide information about how a treatment works for a small group of patients.
Taighde Éireann	Or “Research Ireland”, is a new organisation started in 2024 that invests in and funds research in Ireland. They are a restructure of Science Foundation Ireland and Irish Research Council.
TCD	Trinity College Dublin is a university, well-known for educating students and conducting research.
TMRN	Trials Methodology Research Network is an Irish group working to make trials better, more inclusive, and easier for everyone involved.
ToR	Terms of Reference are a set of guidelines that explain the purpose and structure of a group or project. They outline the goals, responsibilities, and rules that members need to follow. ToR help everyone understand what is expected, how decisions will be made, and the scope of the work involved.
TUH	Tallaght University Hospital is a major hospital located in Dublin. The hospital is also involved in teaching and research.
UCD	University College Dublin is a university, well-known for educating students and conducting research.

UHI	Unique Health Identifier is a special number assigned to individuals to help track their health records and medical information. This can improve patient care by making it easier to coordinate treatments, manage appointments, and reduce errors in medical records.
Ultra Rare	A condition that affects less than 1 in 100,000 people.
YPAG	Young Persons' Advisory Group is a team made up of young patients and carers who provide input and feedback on healthcare practices and policies. Their goal is to ensure that the voices of young patients are heard in decisions about treatments, services, and research.