Conference 2026

Follow-along glossary

A plain-language guide to words and abbreviations you may hear at the Rare Disease Research Conference. Find the session you are attending, then look up terms as you hear them.

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08:50 Welcome

RDCTN — Rare Disease Clinical Trial Network. An Irish network that brings researchers, healthcare professionals and patients together to improve rare disease clinical trials.

09:00 Session 1 | Orphan Medical Devices: Innovation and Regulation

Regulatory, industry and orphan-device perspectives

Orphan medical device — A medical device intended to diagnose, prevent or treat a condition affecting a very small number of people. EU guidance uses a threshold of no more than 12,000 people in the EU per year and also asks whether suitable alternatives are lacking or the device offers an expected benefit.

HPRA (Health Products Regulatory Authority) — Ireland's regulator for medicines, medical devices and other health products.

MDR (Medical Device Regulation) — EU Regulation 2017/745, which sets requirements for the safety and performance of medical devices.

Custom-made device — A medical device specially made for one person, following healthcare professional's written instructions about that person's needs.

CORE-MD — Coordinating Research and Evidence for Medical Devices. A European project that examined how to assess the safety and effectiveness of high-risk medical devices.

EC (European Commission) — The part of the European Union that proposes laws and carries out EU policies. It also makes certain EU-wide decisions about medicines.

EMA (European Medicines Agency) — The EU agency that assesses medicines and advises on whether they should be authorised for use.

FDA (Food and Drug Administration) — The US agency that regulates products including medicines and medical devices.

HealthTech Ireland — An Irish industry association for organisations that develop, make or supply medical devices, diagnostics and other health technologies.

HTA (health technology assessment) — A review of the benefits, risks, costs and wider effects of a treatment, test or health technology to help with healthcare decisions.

HUD (Humanitarian Use Device) — A US category for medical devices intended to help people with conditions affecting no more than 8,000 individuals in the United States each year.

IRDiRC (International Rare Diseases Research Consortium) — An international partnership of researchers, funders, companies and patient organisations working to speed up rare disease research.

IRDiRC Working Group on Medical Technologies — A group that has examined challenges in developing devices, implants and digital tools for rare diseases and children.

IVD (in vitro diagnostic) — A test carried out on a sample taken from the body, such as blood, saliva or tissue.

MDCG (Medical Device Coordination Group) — A group of experts from EU countries that helps guide how European medical device rules are put into practice.

Orphan Device Task Force — A group set up within the MDCG to develop guidance on medical devices intended for very small patient populations.

09:55 Trial Spotlights

Shared terms that could be useful across the three short trial presentations.

Clinical trial — A research study involving people that tests whether a treatment or other healthcare approach is safe and works.

PI (principal investigator) — The person with overall responsibility for leading a research study at a particular site.

SAE (serious adverse event) — An event during research that has serious consequences, such as requiring hospital admission or posing a threat to life. It is not necessarily caused by the treatment.

AE (adverse event) — Any unwanted health problem that happens during a study, whether or not it was caused by the treatment.

AR (adverse reaction) — An unwanted effect thought to be caused by a medicine or treatment.

CHI (Children's Health Ireland) — The organisation providing specialist hospital care for children in Ireland.

GCP (Good Clinical Practice) — International standards that help make clinical trials ethical, safe and reliable.

MMUH (Mater Misericordiae University Hospital) — A Dublin hospital that provides patient care and takes part in teaching and research.

NREC (National Research Ethics Committee) — An independent committee in Ireland that reviews certain research studies to protect participants' rights, safety and wellbeing.

PIL (participant information leaflet) — A document that explains a study so someone can decide whether to take part. Sometimes called a patient information leaflet.

09:55 EB Shield | Dr Fiona Browne

EB (epidermolysis bullosa) — A group of rare conditions that make the skin very fragile and easily damaged.

EBS (epidermolysis bullosa simplex) — A form of EB that mainly affects the outer layer of skin. Rubbing or minor injury can cause blisters.

Vehicle-controlled trial — A study that compares a medicine with the same cream, gel or other base without the active medicine, to see what difference the medicine makes.

10:05 PROSERA Study | Dr Sarah Cullivan

6MWD (six-minute walk distance) — The distance a person can walk in six minutes. It can help researchers understand how a condition or treatment affects physical ability.

PAH (pulmonary arterial hypertension) — High blood pressure in the blood vessels carrying blood from the heart to the lungs. It can make the heart work harder and cause breathlessness.

PVR (pulmonary vascular resistance) — A measure of how much resistance the blood meets as it flows through the vessels in the lungs.

CTD (connective tissue disease) — A group of conditions affecting tissues that support the body, such as skin, joints and blood vessels.

FC (functional class) — A way of describing how much a condition limits someone's usual physical activities and causes symptoms.

10:15 DysK Seed Fund Update | Dr Owen Killian

Cervical dystonia — A movement condition that causes neck muscles to tighten or contract without a person intending them to, sometimes changing the position of the head.

Pilot study — A small early study that tests whether a method or larger study is practical and helps researchers improve its design.

10:30 PPI Leadership | Shaping and Modelling Best Practice through Partnership

Dr Cassy Dinius

PPI (patient and public involvement) — Research carried out with patients and members of the public, rather than only about them. PPI partners help shape decisions about what is studied and how.

EDI (equality, diversity and inclusion) — Making involvement fair and accessible, valuing differences and ensuring that a wider range of people can contribute.

11:15 Session 2 | Innovation, Regulation & Data Systems

11:15 Innovative Methodologies for Rare Disease Clinical Trials – the RealiseD Approach

Prof Ralf-Dieter Hilgers

RCT (randomised controlled trial) — A study in which people are assigned by chance to different groups so researchers can compare treatments fairly.

N-of-1 trial — A study that compares treatments within one person, often over different time periods, to learn what works best for them.

Platform trial — A study designed to test several treatments using one shared trial structure. Treatments can sometimes be added or removed as evidence develops.

Cross-over trial — A study in which participants try more than one treatment, in a planned order, so the effects can be compared.

Parallel-group trial — A study in which different groups receive different treatments at the same time.

SWAT (study within a trial) — A smaller study built into a larger trial to test a better way of running research, such as how to invite or support participants.

11:35 Pathways to Rare Disease Therapies | Prof Claire Booth

Orphan drug — A medicine developed for a rare condition. Special rules can encourage companies to develop treatments for small patient groups.

OMD (orphan medicinal product designation) — A status given to a medicine under development for a rare condition when it meets certain criteria. It can bring support or incentives during development.

MA (marketing authorisation) — Official permission to place a medicine on the market after its quality, safety and benefits have been assessed.

MAH (marketing authorisation holder) — The company or other organisation legally responsible for a medicine that has marketing authorisation.

NCPE (National Centre for Pharmacoeconomics) — An Irish body that reviews whether medicines offer good value for the health service, alongside their expected benefits.

11:55 Health & Data Systems for Research & Clinical Trials | Prof Catherine Quinlan

EHR (electronic health record) — A digital record of someone's healthcare, such as appointments, medicines, test results and letters.

Interoperability — When different computer systems can share information and understand it. For example, two hospitals can exchange test results rather than asking a patient to repeat everything.

Orphacode — A code for a particular rare disease, helping hospitals and researchers record the same diagnosis consistently, even across different systems.

IHI (individual health identifier) — A unique number used in Ireland to help match health information to the right person, even when records come from different services.

Genomics — The study of a person's complete set of DNA, rather than looking at just one gene.

Genetic testing — Testing DNA for changes that may help explain a condition.
Privacy, access and responsible use of data

Data governance — The rules and decisions about who can use health information, why and how it is protected. For example, deciding who can see records for an approved study.

EHDS (European Health Data Space) — An EU framework designed to make health information easier to access and share securely, and to support approved uses for research.

GDPR (General Data Protection Regulation) — EU law that protects personal information and gives people rights over how their data is used.
Secondary use of data — Using existing health information for another approved purpose, such as research or planning services, with safeguards.

Consent — An informed choice to take part in research or allow information to be used in an agreed way. People should understand what they are agreeing to.

Pseudonymised data — Data where names and direct identifiers are replaced by codes. Someone holding a separate, secure key may still be able to link it to a person.

Federated analysis — A way of studying information held in different places while keeping the original data in each location and sharing only the findings.

Registry — An organised collection of information about people with a particular condition, often gathered over time to support research and care.

Real-world data — Information collected during everyday healthcare, rather than specifically for a clinical trial.

12:30 Patient Spotlight | My Clinical Trial Experience – Linda Sheehan

aPAP (autoimmune pulmonary alveolar proteinosis) — A rare lung condition in which a substance called surfactant builds up in the air sacs, making it harder for oxygen to pass into the blood.

Double-blind trial — A trial where neither the participants nor the research team know who is receiving which treatment during the blinded part of the study.

Placebo — A treatment that looks like the one being tested but does not contain its active ingredient. It helps researchers compare effects fairly.

CRC (clinical research centre) — A dedicated place or team that supports research with patients and clinical trials.

PWRD (person with a rare disease) — An abbreviation for someone living with a rare disease.
PRO (patient-reported outcome) — Information that comes directly from a patient about their health, symptoms or daily life, without a clinician interpreting it.
PROM (patient-reported outcome measure) — A questionnaire or other tool used to collect outcomes directly from patients.

PCOM (patient-centred outcome measure) — A measure designed to capture outcomes that matter most to patients, such as daily activities or quality of life.

QoL (quality of life) — How a person experiences their wellbeing and everyday life, including comfort, independence and activities they value.

Single-blind trial — A study where one side, usually the participant, does not know which treatment was assigned, but the other side does.

14:00 Research Spotlight | Advanced Therapy Medicinal Products – Prof Martina Hennessy

ATMP (advanced therapy medicinal product) — A type of medicine made using genes, cells or tissues. Some are being developed to treat rare diseases.

ATIMP (advanced therapy investigational medicinal product) — An advanced therapy medicine being tested in a clinical trial, before it is approved for wider use.

GTMP (gene therapy medicinal product) — A medicine that uses genetic material to treat or prevent a condition, for example by helping cells make a missing or faulty protein.

CRISPR — A gene-editing tool that lets scientists make targeted changes to DNA.

14:30 Session 3 | Efficiency & Effectiveness in Treating Rare Diseases

Endpoint — A result researchers measure to see whether a treatment works or is safe, such as walking distance, pain or hospital visits. The primary endpoint is the main result.

Phase 1 trial — An early clinical trial that mainly explores safety and how a treatment behaves in the body.

Phase 2 trial — A trial that explores whether a treatment seems to work and continues to assess safety and dose.

Phase 3 trial — A larger trial that tests how well a treatment works and compares its safety with other options.

Phase 4 study — Research carried out after a treatment has been approved, to learn more about its effects during wider use.

Preclinical study — Research done before testing a treatment in people, often using laboratory tests or experimental models.

14:40 REMEDi4ALL – Patient-centred medicines repurposing | Mr Anton Ussi

REMEDi4ALL — A European project helping researchers, patients and others explore new uses for existing medicines, including treatments for rare diseases.

Medicines repurposing — Testing whether a medicine already used for one condition could also help people with another condition.

16:00 Poster Lightning Talks

16:00 Warburg Micro Syndrome | Ms Ailbhe Conway

Warburg micro syndrome — An extremely rare inherited condition that can affect eyesight, movement and development.

Research model — A laboratory or other scientific system used to study how a condition develops and to explore possible treatments.

Mutation — A change in DNA. Some DNA changes affect how the body works and may be linked to disease.

16:05 ATP6V0A1 and cell function | Dr Kasia Whysall

ATP6V0A1 — A gene that provides instructions for part of a V-ATPase pump inside cells.

Autophagy — A process cells use to break down and recycle damaged or unwanted components.

V-ATPase — A tiny pump that helps keep the right level of acidity inside parts of our cells, allowing cells to break down and recycle materials.

Lysosome — A cell's recycling centre. It breaks down damaged or unwanted material so useful parts can be used again.

Mitochondria — The cell's energy factories: tiny structures that turn nutrients into energy the cell can use.

Nutrient sensing — How a cell detects available nutrients and adjusts what it does in response.

16:10 Patient-centred care and the patient voice | Dr Niamh Banville

Patient-centred care — Care that takes account of what matters to the individual, including their needs, preferences and everyday life.

Patient voice — Patients' own experiences, views and priorities, used to help shape care, education or research.

Professional identity formation — How students gradually develop their sense of what it means to be a healthcare professional, including their values and responsibilities.

16:15 Session 4 | Policy Driven Innovation

ERDERA (European Rare Diseases Research Alliance) — A European partnership that brings together researchers, clinicians, funders and patient organisations to improve rare disease research.

ERNs (European Reference Networks) — Networks linking specialist healthcare centres across Europe so experts can share knowledge about rare or complex conditions.

JARDIN (Joint Action on Integration of ERNs) — A European project helping connect European Reference Networks with national health services.

EJP RD (European Joint Programme on Rare Diseases) — A European rare disease research programme that came before ERDERA.

ECRIN (European Clinical Research Infrastructure Network) — A European organisation that helps researchers run clinical studies across different countries.

EURORDIS (Rare Diseases Europe) — A European alliance of patient organisations that advocates for people living with rare diseases.

16:25 National Rare Disease Strategy | Prof Atif Awan

NRDO (National Rare Disease Office) — The Irish office that supports coordination and information on rare diseases and specialist care.

NRDS (National Rare Disease Strategy) — Ireland's national strategy for improving rare disease services, care and coordination.

HSE (Health Service Executive) — The organisation responsible for providing public health and social care services in Ireland.

NRDP (National Rare Disease Plan) — Ireland's earlier national plan for rare diseases, published in 2014.

NCPRD (National Clinical Programme for Rare Diseases) — An HSE programme working to improve how rare diseases are identified, managed and treated.

DoH (Department of Health) — The Irish government department responsible for national health policy.

CoE (centre of expertise) — A hospital or specialist service with particular experience in caring for a certain group of conditions.

NGGO (National Genetics and Genomics Office) — An Irish national office supporting better use of genetics and genomics in healthcare.

16:35 Policy Driven Research – Northern Ireland | Prof AJ McKnight

NIRDP (Northern Ireland Rare Disease Partnership) — A group connecting patients, health professionals and researchers to improve rare disease care and awareness in Northern Ireland.

NHS (National Health Service) — The publicly funded health service in Northern Ireland and the rest of the UK.