Understanding Rare Disease Registries

A practical leaflet for patients and carers | RDW-01-08

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A rare disease registry brings together information about people with a condition or group of conditions. It may help researchers understand the disease, assess care or find people interested in future studies. This leaflet explains what a registry can offer, what taking part may involve and what to check before sharing information. Joining does not guarantee better care, treatment or a place in a clinical trial.

Different registries have different purposes

A registry may be run by a hospital, university, patient organisation, public health service or company. The name alone does not tell you what it collects or how it is used. Common purposes include: [1]

- Contact registry — stores contact details and limited information so people can be approached about research opportunities.
- Disease or natural history registry — gathers information on symptoms, diagnosis, treatment and how a condition changes over time. Natural history means the course of a condition; ask how treatments are taken into account.
- Care or treatment registry — records care and outcomes, sometimes including the longer-term safety and effects of a particular treatment.

These purposes can overlap. A patient registry is different from a clinical trial registry, which lists studies rather than serving as the patient database for your condition. A biobank stores biological samples, such as blood or tissue; some registries link to one. Ask what each part involves and whether it has separate choices or consent.

What the information can and cannot show

Well-designed registries can reveal patterns in symptoms, care and outcomes over time. But the people included may not represent everyone with the condition, and records can be incomplete. A difference between treatment groups may reflect differences in their health or care, rather than an effect of the treatment itself. Registry findings need careful interpretation; an observed association does not by itself prove cause and effect. [2]

Registries can support research without producing an immediate result for participants. Ask what the registry aims to achieve, what it has published and how it will report progress, including when findings are uncertain.

What taking part may involve

Check the practical requirements

You might be asked to complete questionnaires, allow access to selected medical records or provide updates over time. Some projects include clinic assessments, genetic information, samples or links to other health databases. Ask which activities are required, which are optional and how often you will be contacted.

Confirm who can join, whether a confirmed diagnosis is needed, which countries are covered and whether there are any costs. Ask about help with language, disability, internet access or completing forms. If a carer answers on someone's behalf, clarify whose information is being provided and what permission is needed.

Understand consent and other legal arrangements

For a registry that asks you to join, informed consent means being given understandable information about its purpose, what will happen, foreseeable risks and how your information will be used, then deciding freely. Ask questions, take time and keep a copy of the information and your agreement. Check whether you can choose separately about future contact, samples or additional research. [3]

Not every registry relies on an invitation to join. Some public health registries collect information from healthcare services under legal powers without asking for individual consent. NHS England's National Disease Registration Service is one example. Ask how your information reached the registry, the legal basis for using it and what choices or opt-outs apply. These arrangements differ by country. [4]

For children or people unable to make the decision themselves, ask who can give permission and how the person's own wishes will be considered. Ask what happens when a child becomes able to decide or reaches adulthood.

Know what privacy terms mean

Identifiable information can reveal who you are, directly or through a combination of details. Pseudonymised or coded information replaces direct identifiers, such as your name, with a code; additional information kept separately may allow it to be linked back to you. Anonymous information cannot reasonably be linked back to a person. Removing a name alone does not necessarily make a record anonymous. [5]

A rare diagnosis combined with age, location, family history or genetic information can make someone easier to identify. Ask who holds the code key, what safeguards are used and what information leaves the registry. No organisation should promise that privacy risks are impossible. Consider whether the explanation is clear enough for you to make a decision.

Questions before you share information

Who decides how the data are used

Ask who is accountable for the registry, how it is funded and who approves access. Check whether universities, charities, public bodies or companies can use the data, whether access involves payment and whether future commercial research is allowed. Ask if data will be linked to other records or transferred to another country, and what safeguards apply.

Find out whether patient representatives help oversee the registry, how concerns are handled and what happens to the data if funding ends or the operator changes. Participation does not automatically give you control over every future project; the written information should explain the limits.

What happens if you change your mind

Ask how to stop questionnaires or future contact and what happens to data or samples already provided. Withdrawal does not always mean deletion: previous analyses may not be reversible, anonymous data may no longer be traceable, and some research data may be retained under applicable law. Confirm the registry's rules and your local rights before agreeing. [6]

Ask how to correct mistakes or request a copy of your information. Keep the registry's contact details securely. For advice about your rights, contact its privacy lead or your country's data protection authority.

Finding and checking a registry

Your specialist team or a condition-specific patient organisation may know relevant registries. The European Directory of Registries, within the EU Rare Disease Platform, provides information about rare disease registries in Europe. A directory entry helps you find a registry; it is not a personal recommendation to join. [7]

Verify the operator and contact details through a trusted organisation before uploading records. Read the participant information and privacy notice. If several registries cover your condition, ask how their purposes differ, whether they share information and how they handle duplicate records.

A short checklist

☐ I understand the purpose, what is required and any costs or burdens.

☐ I know who can use my data, for what purposes and in which countries.

☐ I understand privacy safeguards and the limits of withdrawal or deletion.

☐ I know whether I will receive updates, study summaries or individual findings.

Key points

Check the purpose and data arrangements of each registry. Contributing information can support research, but personal benefit is uncertain. You do not need to send medical data to Rare Disease Watch to use this leaflet. A registry is not an emergency service or a replacement for your usual healthcare.

References and further reading

Sources are numbered in order of first mention. Select a title to open its source URL. Sources checked on 3 November 2026. National sources explain their own systems; rights and legal arrangements may differ elsewhere.

1. US National Center for Advancing Translational Sciences. Patient Registries toolkit

2. US Agency for Healthcare Research and Quality. Registry Design in Registries for Evaluating Patient Outcomes A User's Guide fourth edition 2020

3. US National Center for Advancing Translational Sciences. Protect Human Research Participants RaDaR

4. NHS England National Disease Registration Service. Collecting and keeping patient data safe

5. UK Health Research Authority. Step 1 Scoping What are the data requirements for your project

6. UK Health Research Authority. Data subject rights and research exemptions

7. European Commission Joint Research Centre. European Rare Disease Registry Infrastructure

Applying this information

Registry methods and safeguards affect how useful the information is and what risks participation carries. No single set of rules applies to every registry. Consent to take part and the legal basis for processing personal data are related but distinct; the registry should explain both where relevant.

Registry-specific eligibility, consent, data transfers, withdrawal and return of individual findings require confirmation from the responsible registry team. If individual health or genetic findings are returned, ask who has checked them and how a qualified clinician will help you understand them before acting on them.

https://rarediseasewatch.com/

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