Rare diseases GO FAIR!

A networking initiative to foster the implementation of principles to make rare disease data Findable, Accessible, Interoperable and Reusable for humans and computers (FAIR)

**Our target:** speeding up progress towards early diagnosis and new treatments of rare diseases.

**Challenge:** clinicians, patients and researchers should be able to perform analysis across data systems efficiently and without error. Even though rare disease data are collected everywhere, data are often not compatible, difficult to find and usage restrictions are often unclear.

**Solution:** Global Open FAIR implementation networks: community networks to help each other choose and adopt standards to implement FAIR. We introduce the initiative to form a GO FAIR implementation network for the rare disease community: RDs GO FAIR!

**Patients** FAIR enables data custodians to take responsibility for making rare disease data as reusable as possible. Patients drive the culture change that this requires. The network helps organise workshops and webinars with patients.

---

**Metadata**

- To be Findable: F1. Data and metadata are assigned a globally unique and persistent identifier. F2. Data are described with rich metadata (see R1).
- To be Accessible: A1. Data and metadata are retrievable by their identifier using an endorsed communications protocol A1.1. The protocol is open, free, and universally implementable. A1.2. The protocol allows for an authentication and authorisation procedure, where necessary.
- To be Interoperable: I1. Data and metadata use a formal, accessible, shared, and broadly applicable language for knowledge representation. I2. Data and metadata use vocabularies that follow FAIR principles. I3. Data and metadata include qualified references to other data and metadata.
- To be Reusable: R1. Data and metadata are released with a clear and accessible data usage license. R1.1. Data and metadata are richly described with a plurality of accurate and relevant attributes. R1.2. Data and metadata are associated with detailed provenance. R1.3. Data and metadata meet domain-relevant community standards.

---

**Background** In 2014, global leaders in data science conceived the FAIR principles to reduce the cost of cross-resource data analysis. Preliminary estimates for the Research Data Alliance surpass 10 billion € per year for not having FAIR principles in place in Europe. The FAIR principles are endorsed by EU infrastructures, the NIH, the G20 and G7, and IRdIRC for rare diseases. The GO FAIR concept was born from discussions about how to implement the European Open Science Cloud (with a global scope). RDs GO FAIR is one of several initiatives that receive organisational support from GO FAIR international support & coordination offices. For a quick introduction to the concept of a FAIR infrastructure for health, search for “Personal health: FAIR infrastructure for health” on Vimeo or YouTube.

Help us create a powerful network of FAIR and secure rare disease data sources! Express your interest in the RDs GO FAIR initiative by contacting the members of the seed group or visit https://www.go-fair.org/implementation-networks/overview/go-fair-rare-disease/