

International course on rare disease registries and FAIRification of data at the source

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Introduction

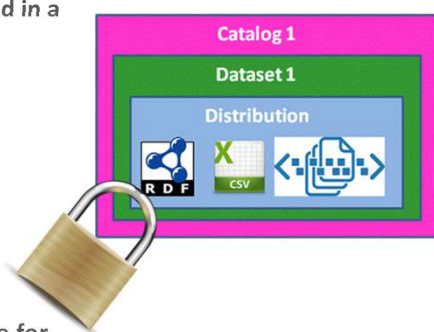
Rare disease registries represent key resources to improve diagnosis, patient management and to advance research. Since 2013 the National Centre for Rare Diseases - Istituto Superiore di Sanità (CNMR-ISS), Rome, Italy, has organised and held an annual International Summer School on Rare Disease and Orphan drug registries. The scope is to promote the establishment of rare disease registries following the Findable, Accessible, Interoperable, and Reusable (FAIR) principles in compliance with IRDiRC and EU recommendations. The course has recently been co-organized with other organisations.

Findable:

- F1.** (meta)data are assigned a globally unique and persistent identifier;
- F2.** data are described with rich metadata;
- F3.** metadata clearly and explicitly include the identifier of the data it describes;
- F4.** (meta)data are registered or indexed in a searchable resource;

Interoperable:

- I1.** (meta)data use a formal, accessible, shared, and broadly applicable language for knowledge representation.
- I2.** (meta)data use vocabularies that follow FAIR principles;
- I3.** (meta)data include qualified references to other (meta)data;



The 15 guiding principles of being FAIR for humans and computers and a FAIR resource, such as a registry, under control for example of the local data manager

Materials and Methods

The Course is open to health professionals, researchers, medical specialists, registry curators, database managers and representatives of patients associations involved in or intend to establish a rare disease registry, including inside European Reference Networks. The course has evolved over the years. The didactic approach shifted from being mainly based on lectures to the highly interactive One Day-One Problem Problem-based Learning. During the course there is also a hands-on tutorial (Bring Your Own Data, BYOD) where participants work with IT-trainers to make specific sample datasets FAIR.

Accessible:

- A1.** (meta)data are retrievable by their identifier using a standardized communications protocol;
- A1.1** the protocol is open, free, and universally implementable;
- A1.2.** the protocol allows for an authentication and authorization procedure, where necessary;
- A2.** metadata are accessible, even when the data are no longer available;

Reusable:

- R1.** meta(data) are richly described with a plurality of accurate and relevant attributes;
- R1.1.** (meta)data are released with a clear and accessible data usage license;
- R1.2.** (meta)data are associated with detailed provenance;
- R1.3.** (meta)data meet domain-relevant community standards;

Results and Conclusions

At the International Summer School, the attendees learn what resources are needed to establish a rare disease registry, and get to know strategies to ensure (i) long term sustainability of the registry, (ii) quality framework, (iii) legal and ethical issues, and (iv) the application of FAIR data principles to strengthen the use of registry data. Since the start of the course in 2013, we have received positive feedback and despite the limited number of places, the interest in the course has increased.

Acknowledgments

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