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Research

@ Semantic Interoperability

Of Rare Disease Data



Objective of Rare Disease (RD) Implementation Network

RD (analytical) tools are available through public FAIR repositories, and can adequately address the requirements for rare disease data.

- Develop FAIR standards
- Foster software solutions
- ...

- Define metadata
- Maintain, train, and support resources
- ...

- Provide sufficient data samples
- Preserve data privacy
- ...

GO CHANGE – Rare Disease

Doing

- Current focus - Transformation between Different Information Models

Build RDF graphs with instances for each model
Training: Learn RDF schemas, and SHACL
Skill: Python: *rdflib* package

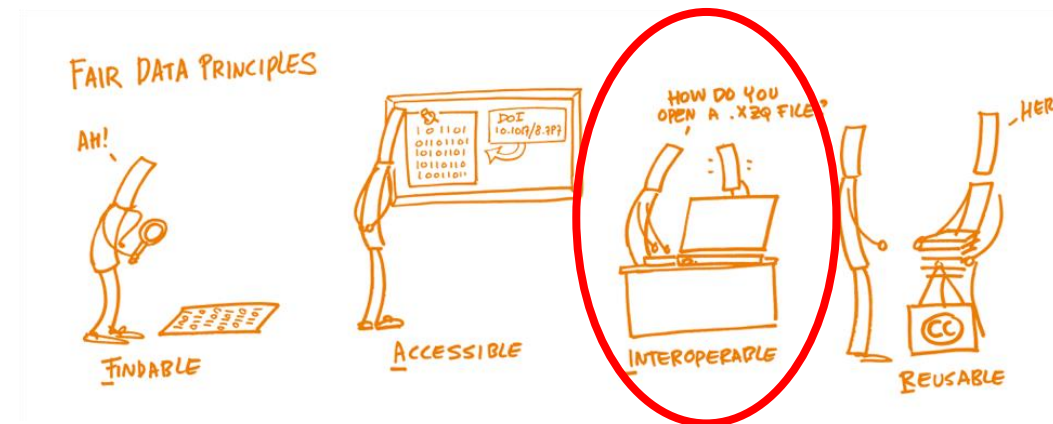
Qualitatively and quantitatively compare them.
Training: Comprehend structures of each model
Skill: SPARQL

Planning

- To evaluate the transformation
- To scale up (instances and information models)
- To develop the tool for automating the transformation

Training and skill:

- Understand how FAIR principles are conformed in each process;
- Clarify relationship between different FAIR artifacts.



Source: <https://www.openaire.eu/how-to-make-your-data-fair>

Are you interested in joining Rare Diseases IN?

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<https://www.go-fair.org/implementation-networks/overview/rare-diseases/>