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
- ...

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Research
@ Semantic Interoperability
Of Rare Disease Data
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/