

Empowering the use of registries by applying FAIR principles: lessons learned and future possibilities

Optimierung der Nutzbarkeit von Daten für
Menschen, die mit einer (seltenen) Krankheit leben

Marco Roos

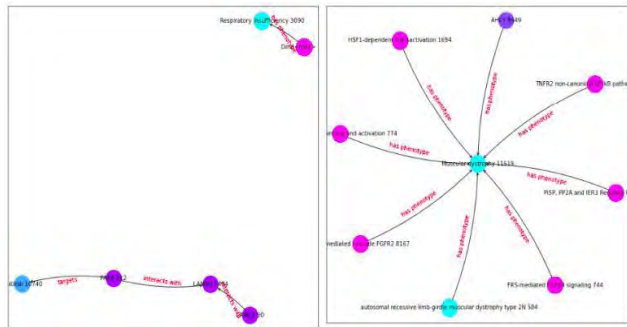
Human Genetics Department, LUMC

BERLIN, MAI 9, 2023

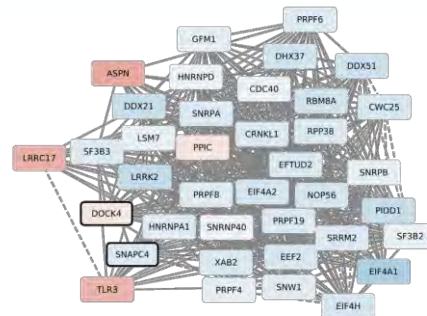


Why FAIR? – AI/ML/Data integration for rare diseases

Preliminary results kindly made accessible by Pablo Perdomo, Carmen Reep, Daphne Wijnbergen, Núria Queralt-Rosinach, Eleni Mina, Annika Jacobsen, Katy Wolstencroft



Ontological hypotheses generated from DMD knowledge graph of multiple sources by ML algorithm



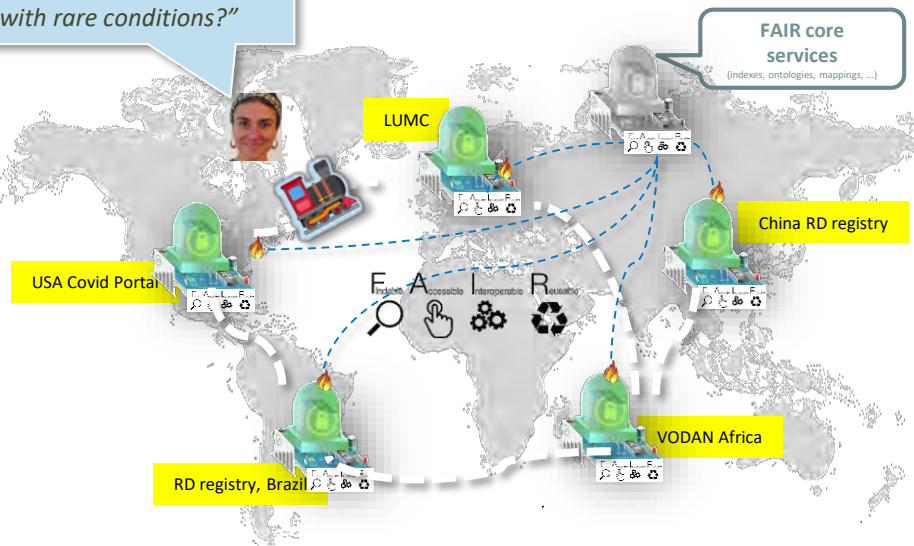
Subnetwork extracted from IBM transcriptomics networks & additional knowledge characterized by ‘RNA splicing’

drug URI	drug label
https://identifiers.org/chembl:CHEMBL29097	CHEMBL29097
https://identifiers.org/chembl:CHEMBL8260	BAICALEIN
https://identifiers.org/chembl:CHEMBL221137	EMBELIN
https://identifiers.org/chembl:CHEMBL267345	AMPHOTERICIN B
https://identifiers.org/chembl:CHEMBL308688	5,7-DIMETHOXYISOFLAVONE
https://identifiers.org/chembl:CHEMBL2110660	IGMESINE
https://identifiers.org/chembl:CHEMBL275809	FR-122047
https://identifiers.org/chembl:CHEMBL161343	ARACHIDONOYL GLYCINE
https://identifiers.org/chembl:CHEMBL585	TRIAMTERENE
https://identifiers.org/chembl:CHEMBL1269845	CHEMBL1269845

Prioritised drug targets for HD learned from knowledge graph

Globally Find, Access, Interoperate, & legitimately Reuse data for data science for patient benefit

*“What will my **machine learning algorithm** learn about Long-Covid predisposition for patients with rare conditions?”*



Outline

- Motivation to making registry data more findable, accessible, interoperable, and reusable for computational use and short introduction machine understandable Linked Data
- FAIR implementation effort Virtual Platform European Joint Programme Rare Diseases
- Next challenge

Where is your balance?

Effort on Data
“Having data”



Effort on Usability
“Reuse by others”

A question

- How many types of treatment are given to patients who are experiencing “difficulty in swallowing” (Dysphagia) as a symptom around the world?
 - ...in the TMF auditorium?

How long would it take to get the answer?

- Seconds
- Weeks or months
- Years
- Forever

Is this our balance?

- Weeks or months
- Years

Effort on Data
“Having data”



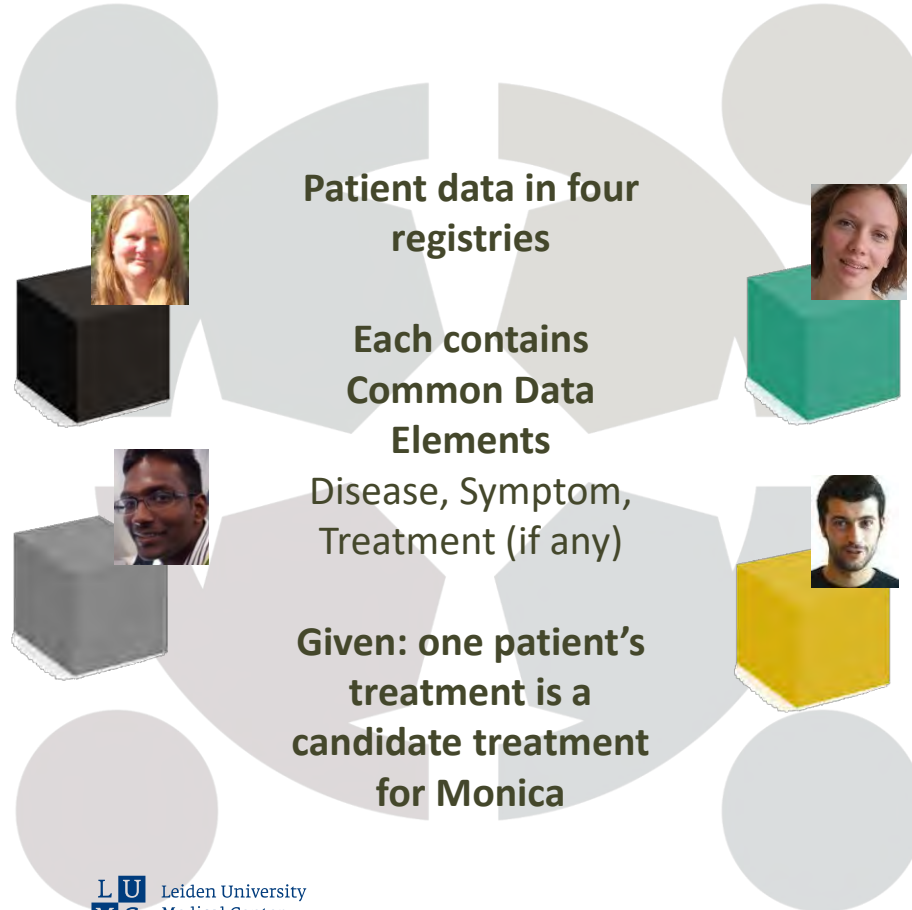
Effort on Usability
“Reuse by others”

Data linking for machines in action

If your (quality of) life depended on other people's data

Any resemblance
with real people is
purely coincidental

Disclaimer: mock
data!!!!



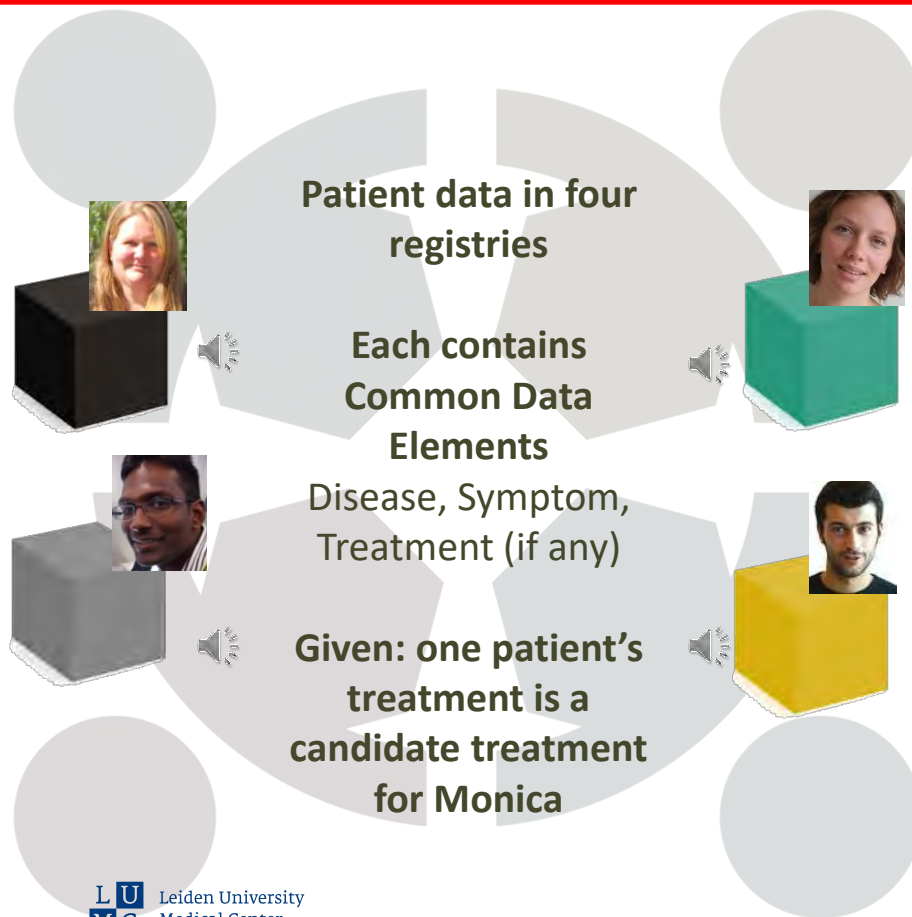
What is the candidate treatment for Monica?



Game: if your life depended on other people's data

Any resemblance
with real people is
purely coincidental

Disclaimer: mock
data!!!!



*What is the
candidate
treatment for
Monica?*



Find the treatment experiment

 Monika	 Annika	 Rajaram	 Pietro
Krankheit Ringbildung Chromosom 14, Salaam-Anfälle, (Keine Behandlung)	Ring-14-sjúkumynd, sankta Vitusar dansur, eingin viðgerð	பெர்ரி நோய்க்குறி, வலிப்பு தாக்குதல்கள், லாமோட்ரைஜின்	sindrome Perry, sbalzi d'umore estremi, ossalato

Find the treatment experiment

Monika 	Annika 	Rajaram 	Pietro 
Krankheit Ringbildung Chromosom 14, Salaam-Anfälle, (Keine Behandlung)	Ring-14-sjúkumynd, sankta Vitusar dansur, eingin viðgerð	பெர்ரி நோய்க்குறி, வலிப்பு தாக்குதல்கள், லாமோட்ரைஜின்	sindrome Perry, sbalzi d'umore estremi, ossalato
Ring-14 disease, Salaam seizures, (no treatment)	Ring-14 syndrome, Chorea, (no treatment)	Perry syndrome, Epileptic attacks, lamotrigine	Perry syndrome, extreme mood swings, oxalate

Find the treatment experiment

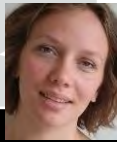
Machine understandability starts with human agreement on the semantics standards follow from there (Ontologies, OMOP, FHIR, etc.)



Monika

Krankheit Ringbildung Chromosom 14, Salaam-Anfälle, (Keine Behandlung)

Monika *has disease* Ring-14 disease, and *has phenotype* Salaam seizures



Annika

Ring-14-sjúkumynd, sankta Vitusar dansur, eingin viðgerð

Annika *has disease* Ring-14 disease, and *has phenotype* Chorea



Rajaram

பெர்ரி நோய்க்குறி, வலிப்பு தாக்குதல்கள், லாமோட்ரைஜின்

Rajaram *has disease* Perry syndrome, and *has phenotype* Epileptic seizures. Epileptic seizures *are treated by* lamotrigine



Pietro

sindrome Perry, sbalzi d'umore estremi, ossalato

Pietro *has disease* Ring-14 disease, and *has phenotype* Extreme mood swings. Extreme mood swings *are treated by* the drug Oxalate

Find the treatment experiment

Translate to ontological model with globally unique IDs (URIs)

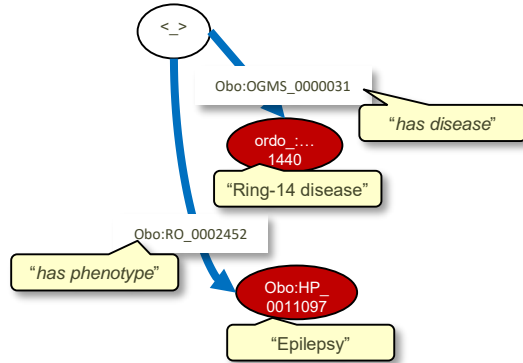
Monika	Annika	Rajaram	Pietro
Monika <i>has disease</i> Ring-14 disease, and <i>has phenotype</i> Salaam seizures	Annika <i>has disease</i> Ring-14 disease, and <i>has phenotype</i> Chorea	Rajaram <i>has disease</i> Perry syndrome, and <i>has phenotype</i> Epileptic seizures. Epileptic seizures <i>are treated by</i> lamotrigine	Pietro <i>has disease</i> Ring-14 disease, and <i>has phenotype</i> Extreme mood swings. Extreme mood swings <i>are treated by</i> the drug Oxalate
<_> obo:OGMS_0000031 ordo:Orphanet_1440 obo:RO_0002452 obo:HP_0011097.	<_> obo:OGMS_0000031 ordo:Orphanet_1440, obo:RO_0002452 obo:HP_0002072.	<_> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0011097 obo:RO_0002302 obo:CHEBI_33237	<_> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0000720 obo:RO_0002302 obo:CHEBI_132952

obo: <http://purl.obolibrary.org/obo/>
ordo: <http://www.orpha.net/ORDO/>

Unambiguous machine understandable semantics for:
 Person *has disease* Disease,
 Person *has phenotype* Phenotype,
 Phenotype *is treated by* Treatment

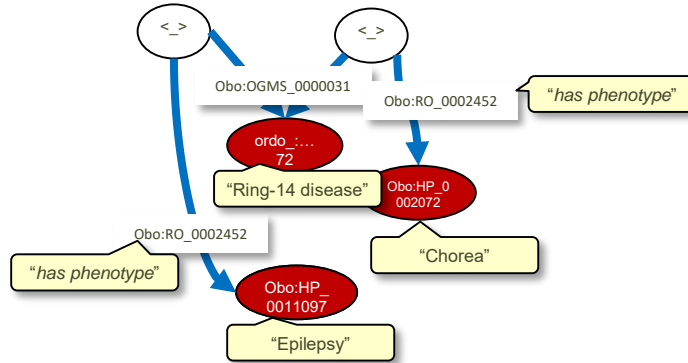
Explore combinations!

Monika	Annika	Rajaram	Pietro
<-> obo:OGMS_0000031 ordo:Orphanet_1440 obo:RO_0002452 obo:HP_0011097.	<-> obo:OGMS_0000031 ordo:Orphanet_1440, obo:RO_0002452 obo:HP_0002072.	<-> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0011097 obo:RO_0002302 obo:CHEBI_33237	<-> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0000720 obo:RO_0002302 obo:CHEBI_132952



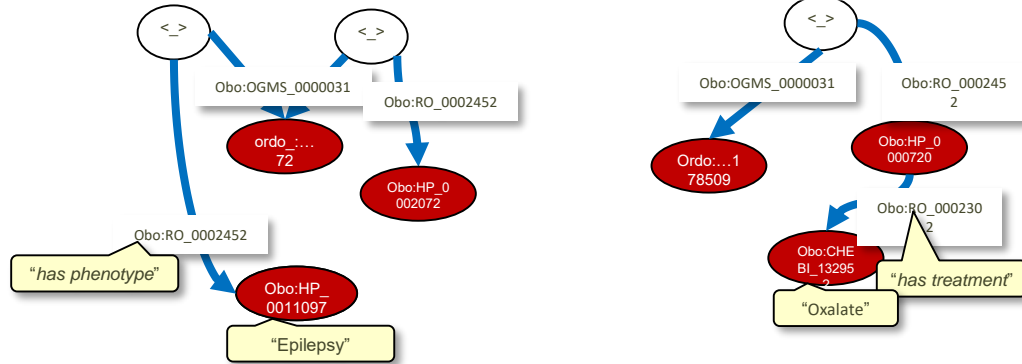
Explore combinations!

Monika	Annika	Rajaram	Pietro
<-> obo:OGMS_0000031 ordo:Orphanet_1440 obo:RO_0002452 obo:HP_0011097	<-> obo:OGMS_0000031 ordo:Orphanet_1440 , obo:RO_0002452 obo:HP_0002072	<-> obo:OGMS_0000031 ordo:Orphanet_178509 , obo:RO_0002452 obo:HP_0011097 obo:RO_0002302 obo:CHEBI_33237	<-> obo:OGMS_0000031 ordo:Orphanet_178509 , obo:RO_0002452 obo:HP_0000720 obo:RO_0002302 obo:CHEBI_132952



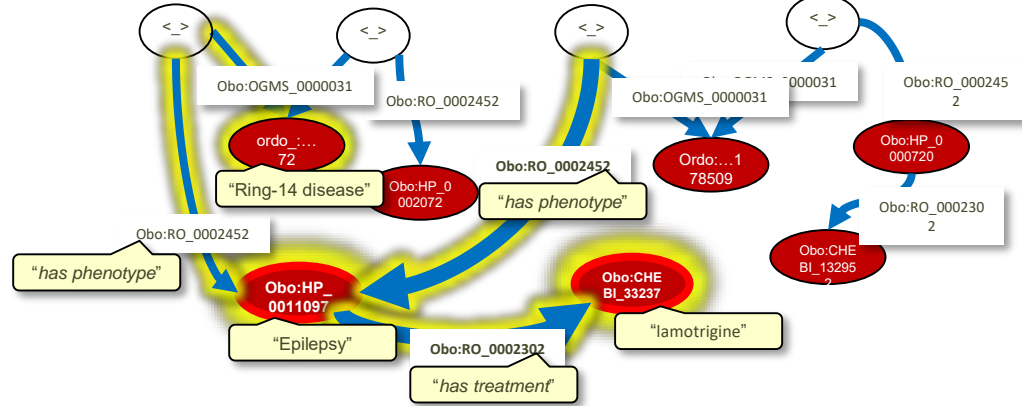
Explore combinations!

Monika	Annika	Rajaram	Pietro
<-> obo:OGMS_0000031 ordo:Orphanet_1440 obo:RO_0002452 obo:HP_0011097	<-> obo:OGMS_0000031 ordo:Orphanet_1440 , obo:RO_0002452 obo:HP_0002072	<-> obo:OGMS_0000031 ordo:Orphanet_178509 , obo:RO_0002452 obo:HP_0011097 obo:RO_0002302 obo:CHEBI_33237	<-> obo:OGMS_0000031 ordo:Orphanet_178509 , obo:RO_0002452 obo:HP_0000720 obo:RO_0002302 obo:CHEBI_132952



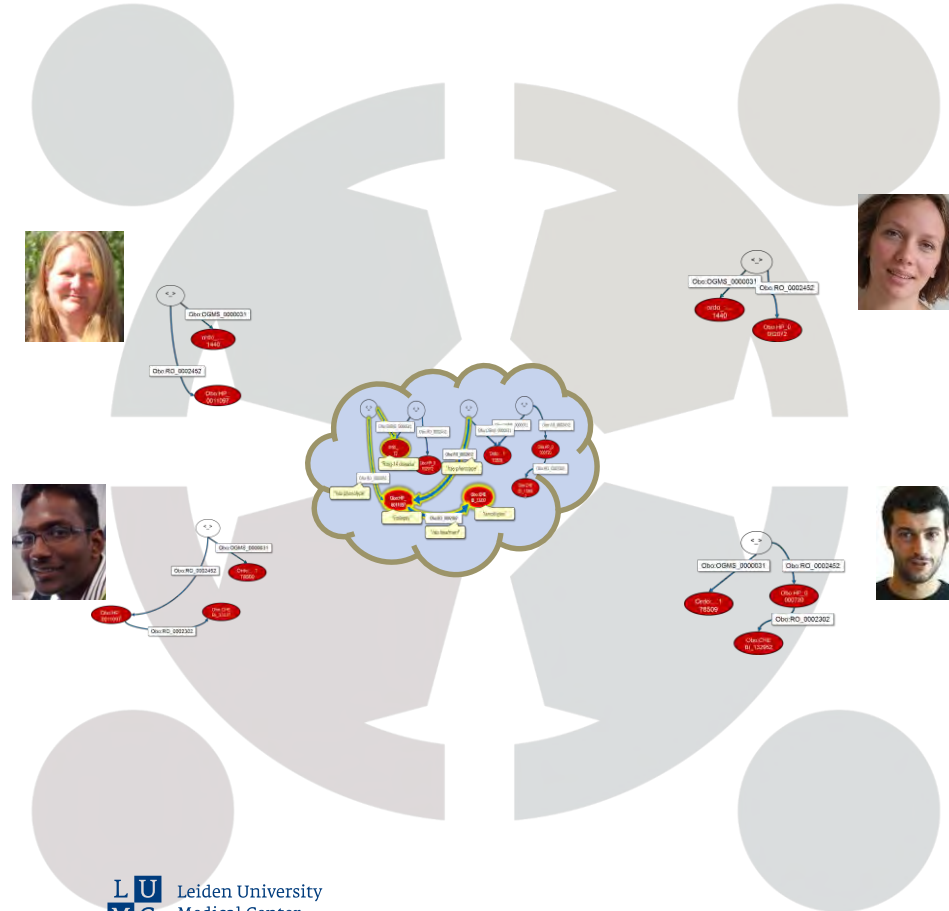
The solution!

Monika	Annika	Rajaram	Pietro
<-> obo:OGMS_0000031 ordo:Orphanet_1440 obo:RO_0002452 obo:HP_0011097.	<-> obo:OGMS_0000031 ordo:Orphanet_1440, obo:RO_0002452 obo:HP_0002072.	<-> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0011097 obo:RO_0002302 obo:CHEBI_33237	<-> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0000720 obo:RO_0002302 obo:CHEBI_132952

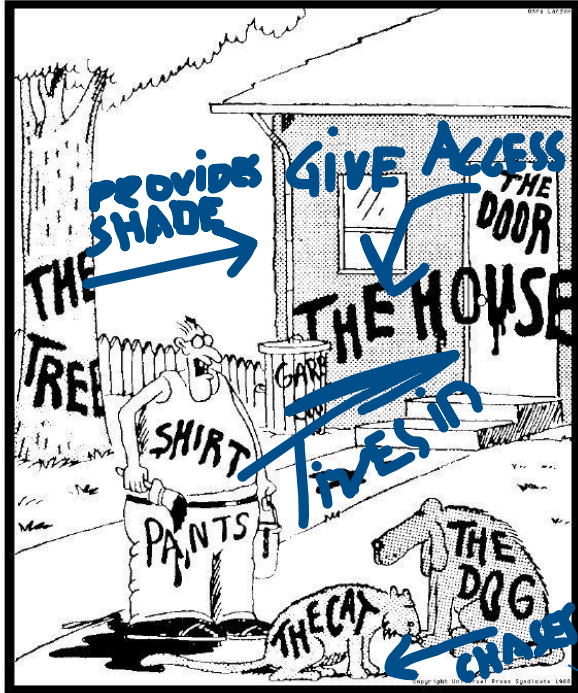


FAIR registries

They form a
virtual
knowledge graph
(i.e. not necessarily
in one location)



Observation of FAIR game



“Now! *That* should clear up a few things around here!”

...in a language that computers understand

- Common data elements, nor data quality resolved the lack of interoperability
- Data elements and their relations were converted in ontology-qualified identifiers ‘*at source*’
- That creates a virtual knowledge graph containing solutions across resources
- *A focus on FAIR may contribute more to (re)usability of data than a focus on data as such*

Silos, silos, silos



Silos, silos, silos



Absolute need to combine



Achieving the global vision



Usability of data for rare disease care and research must be brought to higher, global levels

Lorentz workshop 2014

Birth of the FAIR principles



Barend Mons (LUMC/UL)
Initiator FAIR principles
President CODATA



Lorentz Workshop, Leiden, 2014

Meeting of global leaders in data science

Motivation: data use & reuse *unacceptably* inefficient and poorly reproducible

Concluding that data should be

Findable, Accessible, Interoperable, Reusable
*for humans and machines**

*In 2016 refined in terms of 15 FAIR guiding principles for scientific data management and stewardship

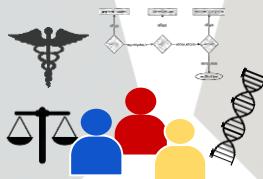
[Wilkinson *et al.*, 2016 <https://www.nature.com/articles/sdata201618>]

Research & Development of FAIRification

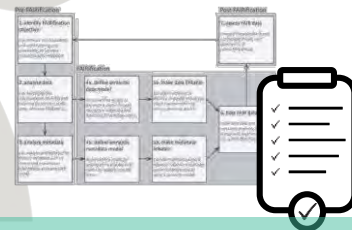
Experience from FAIRification projects and workshops in RD domain since 2014



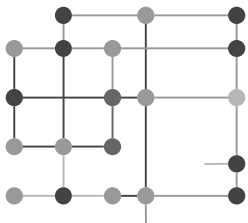
Training



Organising expertise



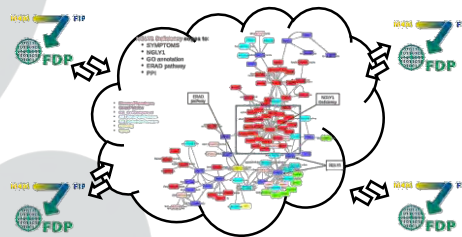
Guidelines & metrics



Align with infrastructure



Tools



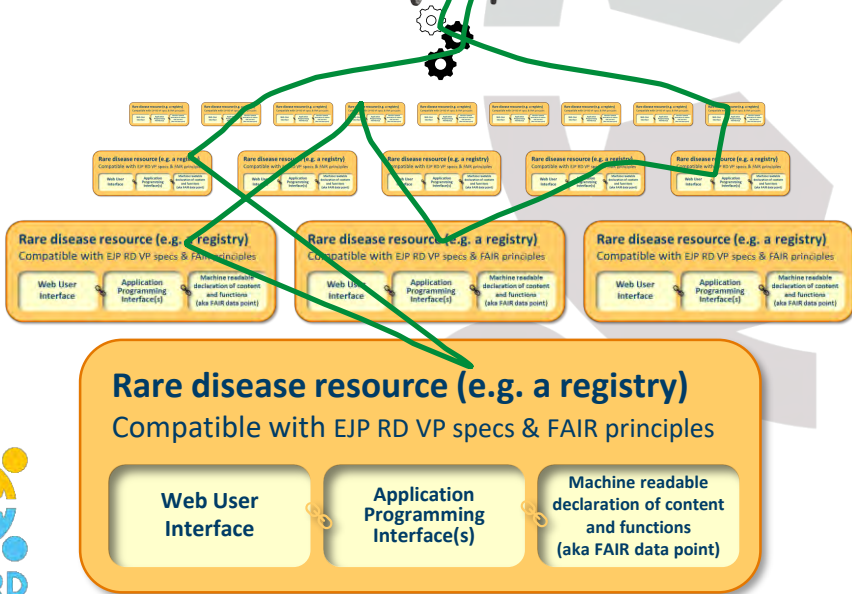
FAIR exploitation



EUROPEAN JOINT PROGRAMME RARE DISEASES

Development of the Virtual Platform network
of federated FAIR resources

EJP RD FAIR Virtual Platform to contribute to other people's research for benefit of Rare Disease patients



- ❁ Rare disease patients would like you to contribute to the VP network
- ❁ Resources create the Virtual Platform network
- ❁ Resources declare about their content and functions *in a language that computers understand*
- ❁ Analysis algorithms make use of FAIRness, researchers make use of these algorithms

FAIR for machines

example: web interface Duchenne Data Platform

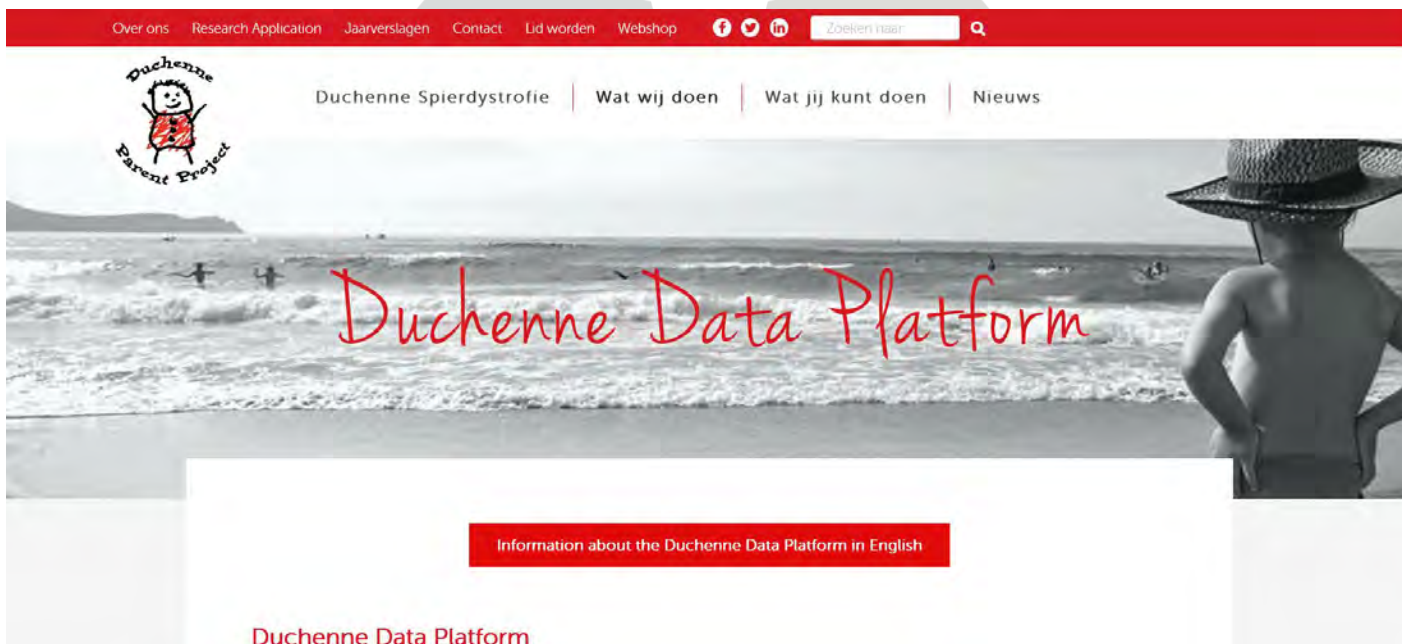
Rare disease resource (e.g. a registry)

Compatible with EJP RD VP specs & FAIR principles

Web User Interface

Application Programming Interface(s)

Machine understandable declaration of content



Duchenne Data Platform

API for engineers... (a special type of human)

A resource discovery API (programmatic access)

Rare disease resource (e.g. a registry)

Compatible with EJP RD VP specs & FAIR principles

Web User Interface

Application Programming Interface(s)

Machine understandable declaration of content

The screenshot shows the GitHub repository page for 'ejp-rd-vp / query_builder_api'. The repository is owned by 'davidReinert' and has 99 commits. The main page displays the repository name, navigation tabs (Code, Issues, Pull requests, Actions, Projects, Wiki, Security, Insights), and a list of files including 'versions', 'LICENSE', and 'README.md'. The 'About' section states: 'This project focusses on the query builder API.' Below this, the 'Query Builder API' section is visible, with the text: 'This project focusses on the query builder API. It will build up the catalogue APIs.'

The screenshot shows the 'specification.yaml' file in the 'query_builder_api / versions / v1.0' directory. The file is 288 lines long, 8.74 KB, and was last committed by 'davidReinert' on 2 Jul. The file content is as follows:

```
1 openapi: "3.0.0"
2 info:
3   version: "1.0"
4   title: "Federated Resource Query API Specification"
5   description: >-
6     "Federated Resource Query API is a web service for rare disease data sharing
7     that can be queried for information about specific rare disease data."
8   contact:
9     name: "EJP-RD Pillar 2 Query Builder Work Focus"
10  #license:
11  #name: "Apache 2.0"
12  #url: "http://www.apache.org/licenses/LICENSE-2.0.html"
13  externalDocs:
14    description: "EJP-RD Website"
```

Resource description for machines

example: DCAT metadata & metadata provisioning service ('FAIR Data Point')

Rare disease resource (e.g. a registry)

Compatible with EJP RD VP specs & FAIR principles

Web User Interface

Application Programming Interface(s)

Machine understandable declaration of content

FAIR Data Point

Search: Search for Data Point... Log in

DPP FAIR Data Point

FAIR Data Point to host description of Duchenne disease resources.

Metadata Issued	Metadata Modified
20.05.2021	21.05.2021

Catalogs

Duchenne Data Platform Catalog

The Duchenne Data Platform (DDP) Catalog

HP_0000708	HP_0000750	HP_0000819
HP_0002014	HP_0002019	HP_0002020
HP_0003044	HP_0003270	HP_0005997
HP_0008981	HP_0009473	HP_0011675
HP_0033333	HP_0033454	MP_0012106
NCIT_C115789	NCIT_C115805	NCIT_C121

```

{
  "@graph": [
    {
      "@id": "https://w3id.org/duchenne-fdp",
      "@type": [
        "http://www.re3data.org/schema/3-0#Repository",
        "http://www.w3.org/ns/dcat#Resource"
      ],
      "http://purl.org/dc/terms/accessRights": [
        {
          "@id": "https://w3id.org/duchenne-fdp#accessRights"
        }
      ],
      "http://purl.org/dc/terms/conformsTo": [
        {
          "@id": "https://www.purl.org/fairtools/fdp/schema/0.1/fdpMetadata"
        }
      ],
      "http://purl.org/dc/terms/description": [
        {
          "@value": "FAIR Data Point to host description of Duchenne disease resources."
        }
      ],
      "http://purl.org/dc/terms/hasVersion": [
    
```

W3C-recommended Data Catalog Vocabulary

Data elements for machines

Modelling pattern for data elements, aka "DCAT" for data elements

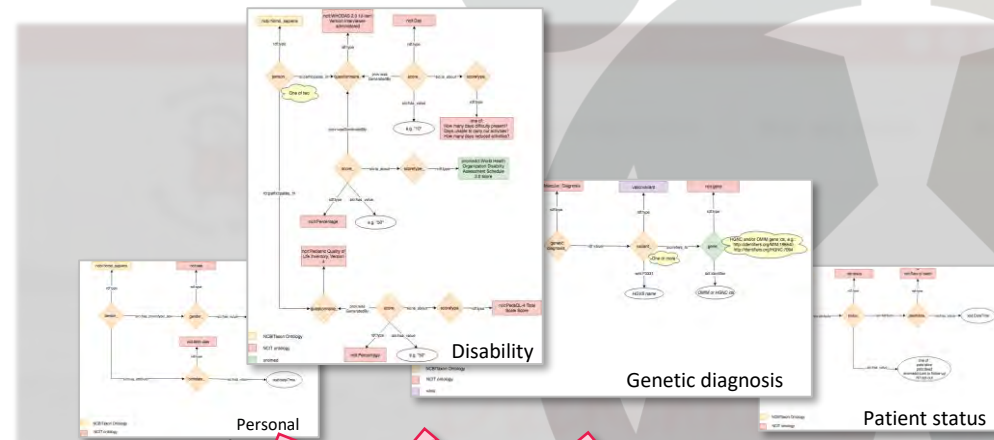
Rare disease resource (e.g. a registry)

Compatible with EJP RD VP specs & FAIR principles

Web User Interface

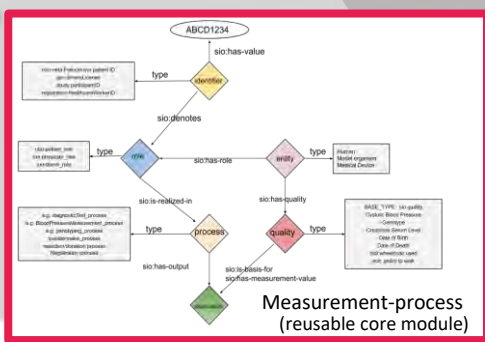
Application Programming Interface(s)

Machine understandable declaration of content



Semantic modelling pattern for machine understandable observations

- Extendable model *'for machines'* using standard ontologies, applied *at source*
- Modules available for 16 common data elements for RD patient registries
- Mappings/conversions, e.g. to FHIR, OMOP, C-DISC, OBO Foundry, GA4GH



<new data element>

Data access conditions for machines

Modelling common consent elements for machines

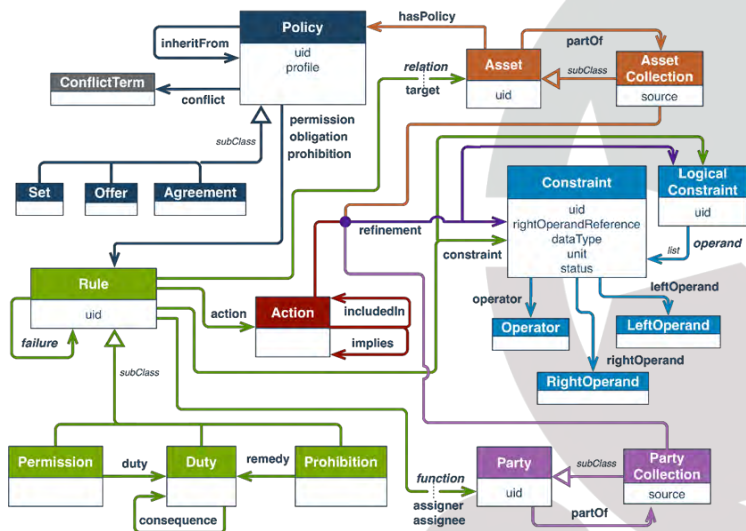
Rare disease resource (e.g. a registry)

Compatible with EJP RD VP specs & FAIR principles

Web User Interface

Application Programming Interface(s)

Machine understandable declaration of content



- ‘Common consent elements (IRDiRC Task Force)
- Developing machine understandable version for triaging access requests (W3C Open Digital Rights Language – ODRL)
- (Contact me if you are interested in a cross-project working group)

Light weight index generated from FAIR metadata

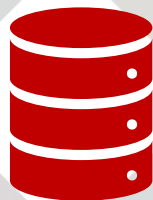
EJP METADATA
DCAT « Template »

EJP METADATA
DCAT « Template »

EJP METADATA
DCAT « Template »



Index generated
from metadata



« Virtual Platform Portal »



Ask for
Available
sources

Fetch metadata
to be displayed

Obtain
endpoint URLs
to perform
query API

FAIR metadata provisioning services
« FAIR Data Point » *at source*

Virtual Platform Network

Virtual Platform Portal

Applications for humans...

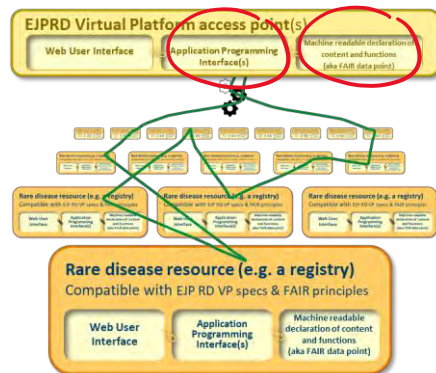
example: resource finders on vp.ejprardiseases.org

FAIR Data Point
Metadata for machines

FAIR Data Points

Filter: All 5 Act

Endpoint	Registration
https://w3id.org/ctsr-fdp	27-02-2023, 09:23:29
https://w3id.org/smarcare-fdp	27-02-2023, 10:34:25
http://w3id.org/ero-euro-rmd-fdp	27-02-2023, 09:15:55
https://w3id.org/fairvasc-fdp/	15-03-2023, 16:14:48



A screenshot of the EIPRD Resource Discovery interface. The header shows "EUROPEAN JOINT PROGRAMME RARE DISEASES" and "Resource Discovery". The search bar contains "Autosomal dominant polycystic kidney disease [730]". Below the search bar, there are filters for "Resource Type(s)", "Countries", "Gender(s)", and "Age Range". The "Results" section shows two entries: "ADPKD [730]" and "ADPKD [730]".

Or, ... create your own computational workflow or user application

EJP RD effort on getting there...

Your data
in original
form

FAIRification



FAIR VP compliant resource with FAIR data

- Web User Interface
- Application Programming Interface(s)
- Machine readable declaration of content and functions (aka FAIR data point)

Your data
in original
form

Ontological Information about resource & access

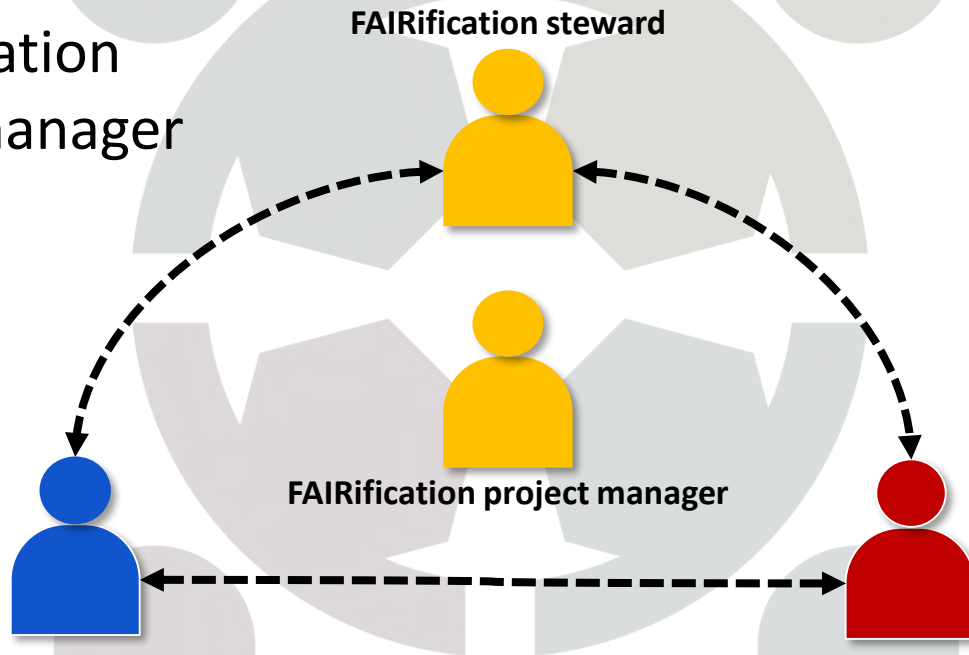
Ontological model describing data elements

R&D of FAIRification

Organising expertise



3 party collaboration
+ FAIR project manager

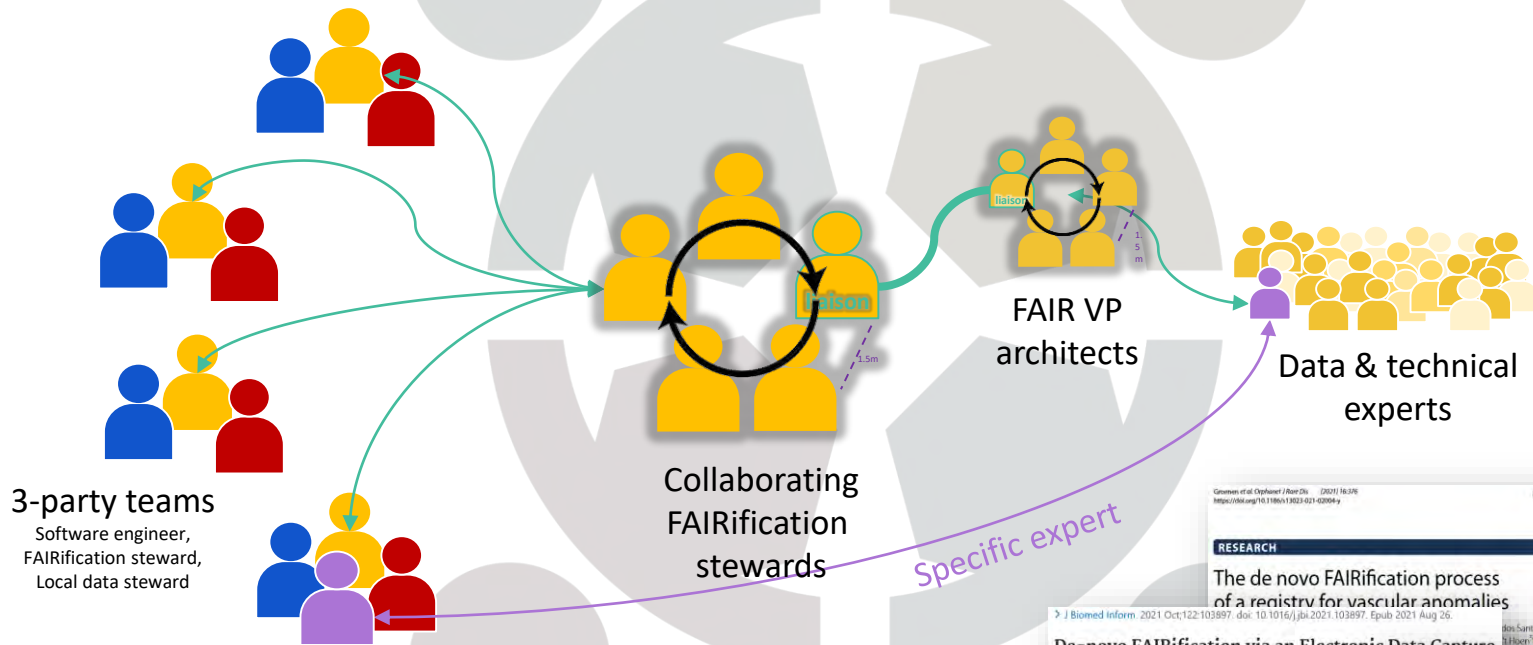


Tasks	Task	Period (months)	Associated Disease Deliverable	Comments
Start of preparation phase	Define driving research questions and target data for FAIRification (use cases)	1	M1	
	Technical assessment of requirements for the BYOD	1-2	M2	
	Select BYOD experts	3	M2	-Disease D- experts -Disease D- Address experts FAIR data experts
	Prepare internal reference ontology for BYOD	2-4	M2	
End of preparation phase	BYOD -Disease D- data owners and FAIR Data experts	4	M3	
Start of implementation phase	Technical assessment of requirements for implementing a FAIR Data Point for the -Disease D- resource	4-5	M4, D1	
	Develop and test -Disease D- FAIR Data Point	4-11	M4, D1	Agile development including pre-releases of FFPs for testing
	Design and execute data analysis for testing FAIR Data Points as substrate for knowledge discovery on -Disease D- (proof-of-concept)	5-11	M5	Enabled by subsampling research queries
End of	Release documented	11-12	D1	By -Disease D-

Roadmap & cost estimate



Network of FAIRification stewards



Orphanet Journal of Rare Diseases

RESEARCH Open Access

The de novo FAIRification process of a registry for vascular anomalies

J Biomed Inform 2021 Oct;122:103997. doi: 10.1016/j.jbi.2021.103997. Epub 2021 Aug 26.

De-novo FAIRification via an Electronic Data Capture system by automated transformation of electronic Case Report Forms into machine-readable data

Orphanet J Rare Dis. 2022 Dec 14;17(1):436. doi: 10.1186/s13023-022-02558-5.

Towards FAIRification of sensitive and fragmented rare disease patient data: challenges and solutions in European reference network registries

Martijn G Kerstloot¹, Annika Jacobsen², Karlijn H J Groenen³, Bruna Rajaram Kaliyaperumal², Amreen Abu-Hanna², Ronald Cornet⁵, Pei Leo Schultze Kool⁴, Derk L Arts⁷, Bruna Dos Santos Vieira^{8, 1, 2}, César H Bernabe⁸, Shoun Zhang^{8, 4, 9}, Heitham Abaza⁸, Nirupama Beris^{4, 9}, Alberto Cámara⁸, Ronald Cornet^{4, 5}, Clemence M A Le Cornec⁸, Peter A C 't Hoen⁴, Franz Schaefer⁸, K Joeri van der Walde⁸, Morris A Swertz⁸, Mark D Wilkinson⁷, Annika Jacobsen^{4, 10}, Marco Ross^{4, 11}

Incrementally contributing to the Virtual Platform



Onboarding guidance document (ongoing)

Onboarding F2F and follow-up workshops

Level 1

- Metadata discovery
- basic description of resource
- overview of content

What is it?

Level 2

- Discovery of limited parts of datasets
- output: yes/no, counts, etc..

What does it contain?

Level 3

- Querying of datasets
- record-level querying

Answer my complex question

Level 4

- Federated Analysis
- Answer advanced research questions

Tooling by the EJP RD

Your data
in original
form

FAIRification

- DIY by **VP & FDP specifications**
- CSV to FAIR by **FAIR-in-a-box**
- Create API by **Beacon-in-a-box**
- Out of the box FAIR generation by **VP specs-compliant registry software** such as MOLGENIS, CastorEDC, Duchenne Data Platform
- Question-based guidance by **smart guidance tool** for data stewards
- Project management guidelines



FAIR VP compliant resource with FAIR data

- Web User Interface
- Application Programming Interface(s)
- Machine readable declaration of content and functions (aka FAIR data point)

Your data
in original
form

Ontological Information about resource & access

Ontological model describing data elements



**WORLD
DUCHENNE
ORGANIZATION**
UPPMD

**INTEROPERABILITY
ACHIEVED BETWEEN
TWO FAIR REGISTRIES**

2023



**INTEROPERABILITY
ACHIEVED BETWEEN
FIVE FAIR REGISTRIES**



- DUCHENNE
FAIR DATA
DECLARATION**
1. Patient derived or generated data are not owned by those who collect them, and should not be generally accessible to the general data base. Research (academic, commercial and health professionals) are excluded.
 2. To create the central source of data, the data owner (UPPMD, Research, Government and Healthcare (i.e. FAIR) by medical professionals, patients and caregivers) are involved.
 3. The central source of data should be supported at all levels, by professional and scientific organizations working on the data collection, and the data use in the future, and also by patients and caregivers. The data should be shared in a secure and regulated manner (FAIR data management and security) as shown in the regulatory guidance. There is a need to set up an infrastructure about the data protection and their importance (FAIR Aware).
 4. Genetic data should be shared in a secure and safe way to ensure the confidentiality of sensitive associated with genetic data, and to prevent genetic tests at possible future. We are committed to contribute in the future to the development of a central source for the diseases we suffer from.
 5. Research, life cycle, allow identification of the individual associated with genetic information and to share it for research and clinical practice in the largest possible scope.
 6. Techniques and tools should be developed to enable 'fairness' concerning the collection, storage and handling of genetic information, commercial, academic and regulatory to form a 'social health' around all stakeholders and care.
 7. Regulations should optimally enable the sharing of key information and define what can and cannot be published in a central source in the largest possible scope.
 8. The FAIR should actively discourage publishing of health sensitive information, data and/or genetic information publicly. There are very strict requirements and standards for both research and care and by patients, families, data and information should be published in a secure and safe way in a central source for others in a small-scale circle.
 9. Funding agencies should have good data ownership following the FAIR principles outlined in their own conditions.
 10. The role of FAIR improve data management should also be wider than: they should include data for research for better clinical practice, care services and its contribution for those in their circles, will not be shared, such as the research results, and based on genetic information.
 11. Health insurance organizations and governments should use FAIR and central data to ensure the transparency of health information, which will be used to improve data use in research, care and the development of health care and the response to genetic data very different healthcare approaches for it.

**FAIR
TEAM**



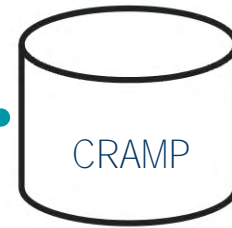
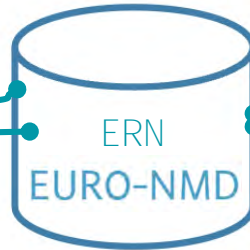
2020

**Duchenne
Data
Platform**

**FAIR
STATUS
ACHIEVED
2021**



2022



FAIR

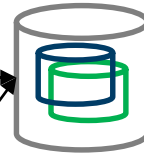
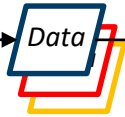
2019

My conclusion

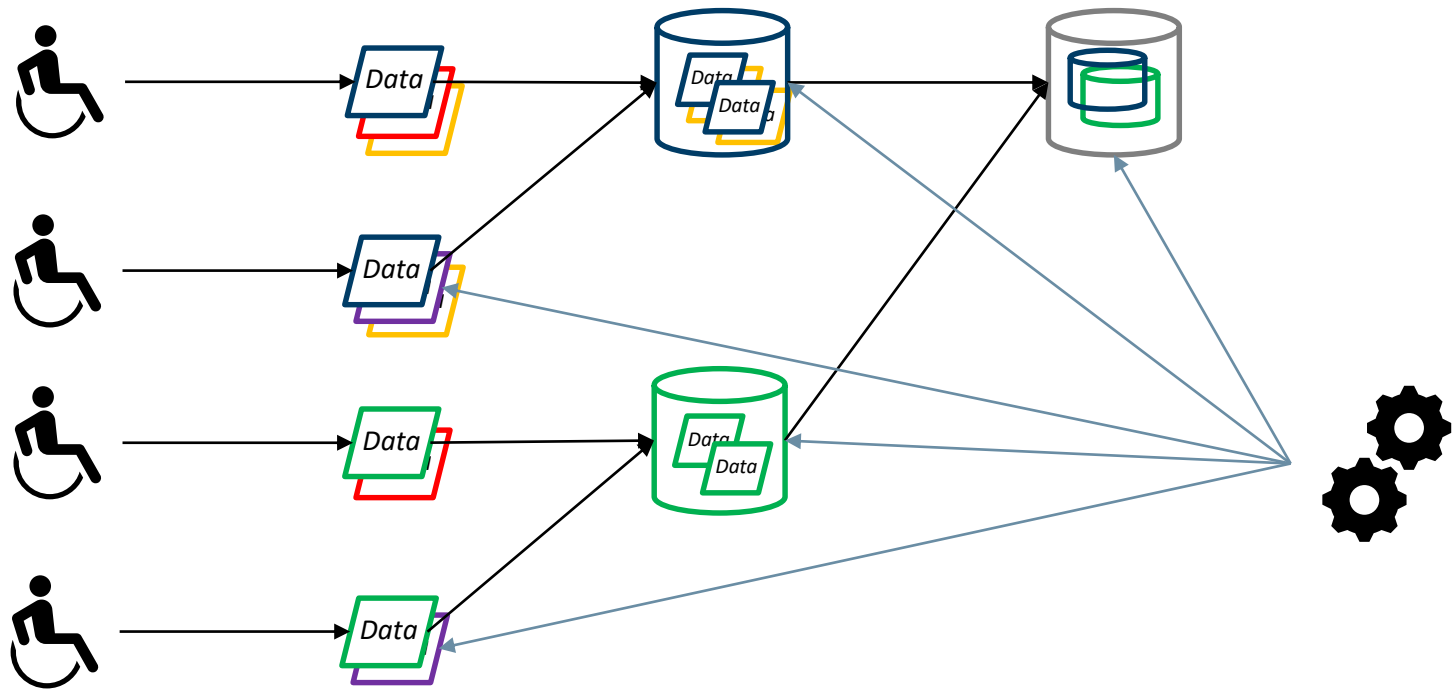


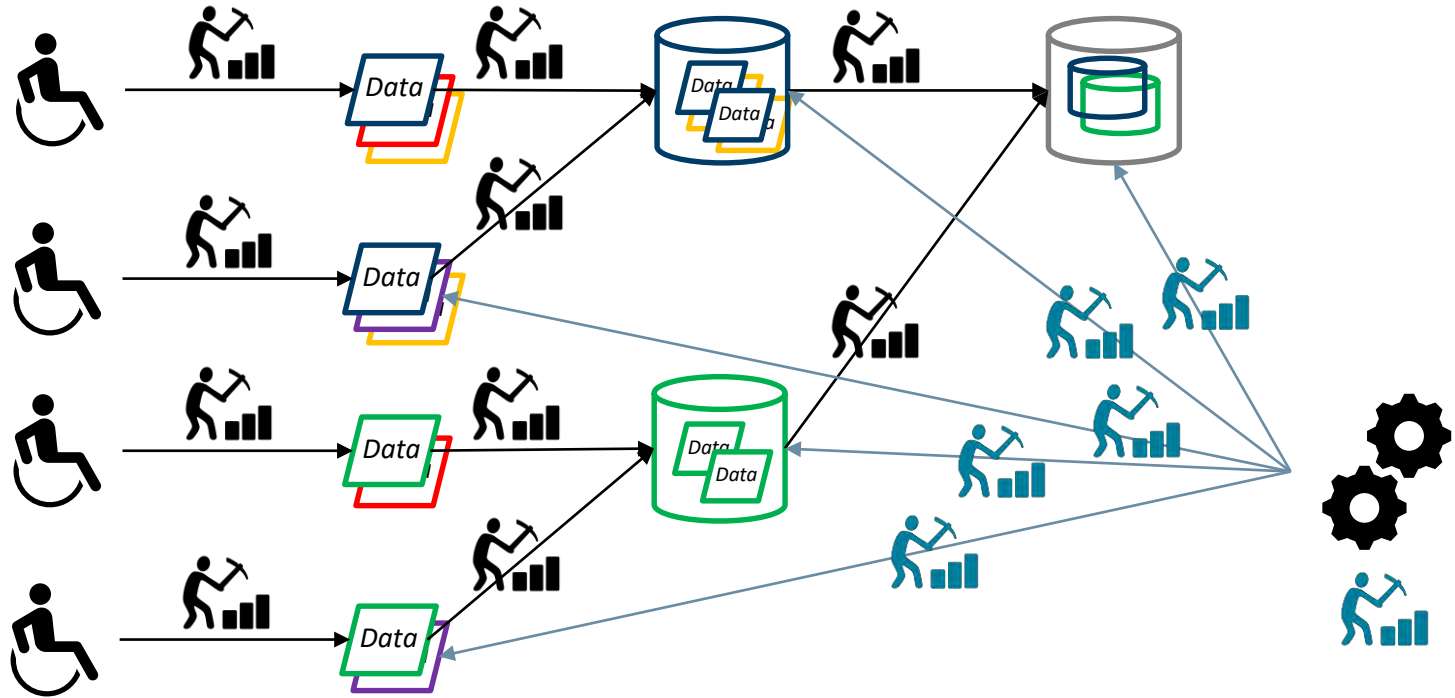
Lizanne, Duchenne
Muscular Dystrophy
patient, endorses
FAIR principles

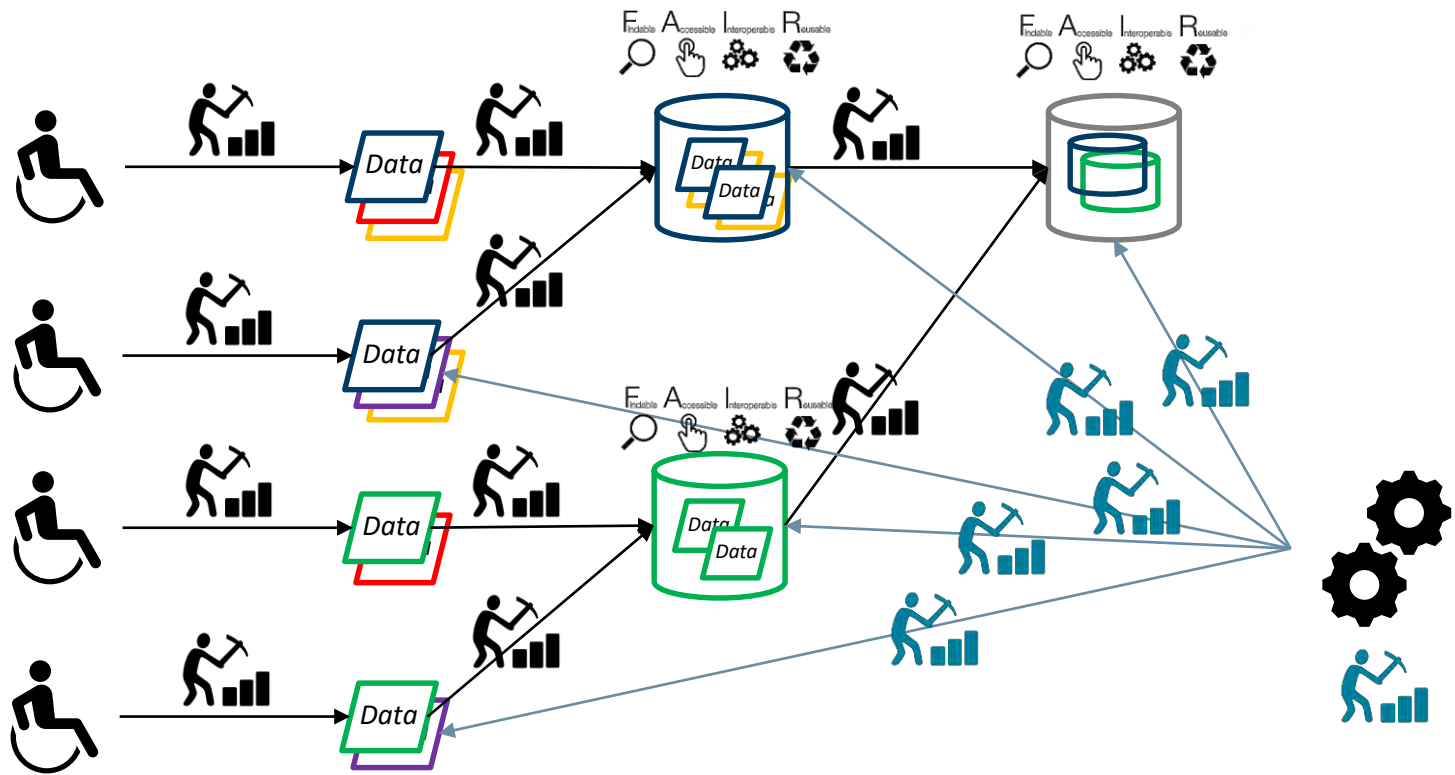
- Having data is not the same as having optimally usable data
- FAIR principles exist entirely to make data as usable as possible, for trusted algorithms, under defined – but efficiently processable – conditions
- Patients will benefit if data generators and data collectors shift balance to effort on making data FAIR as much as they do on generating/collecting data
- Be persistent: it may take years before we reach a critical mass of FAIR data (NB: patient organisations will not wait forever...)
- Future challenge: move FAIR closer to the source → automate creation of registries

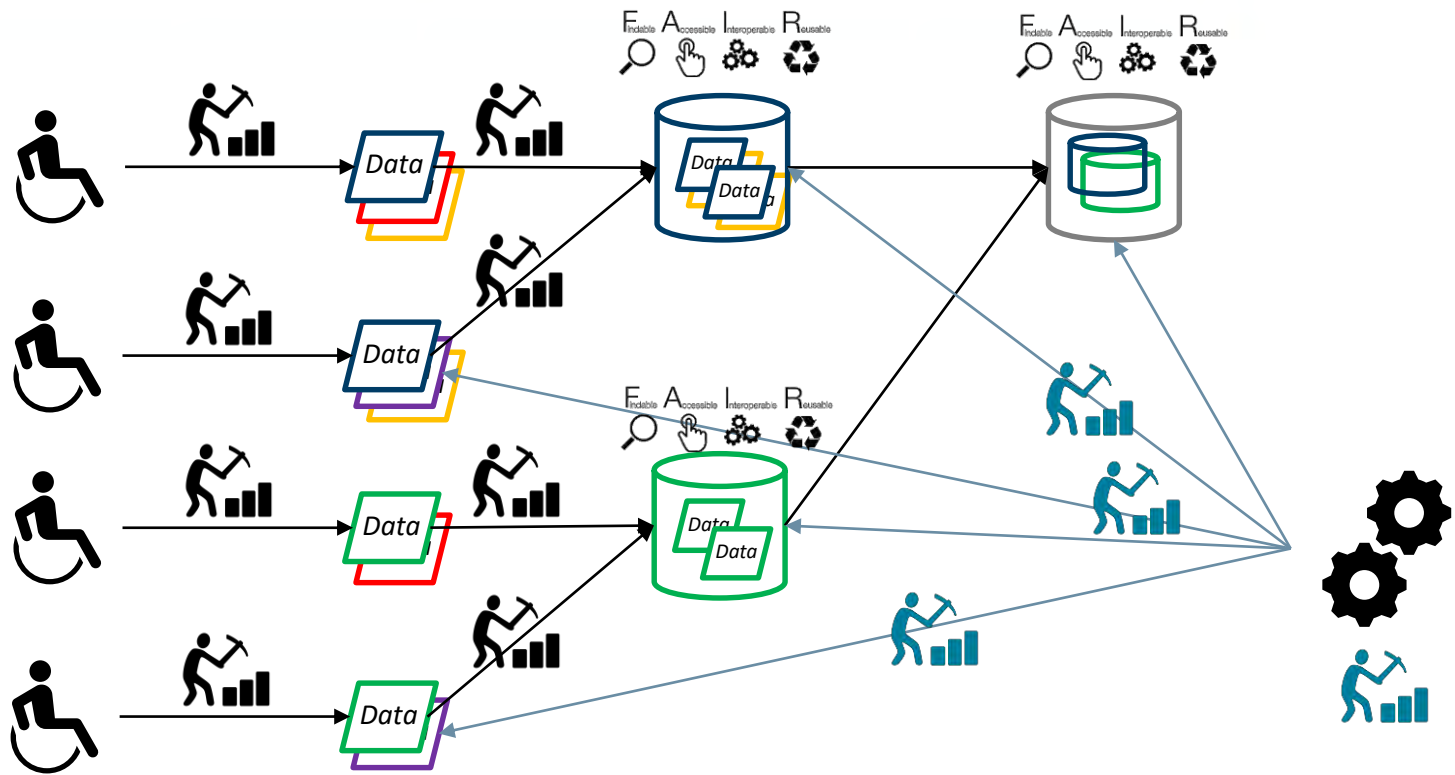


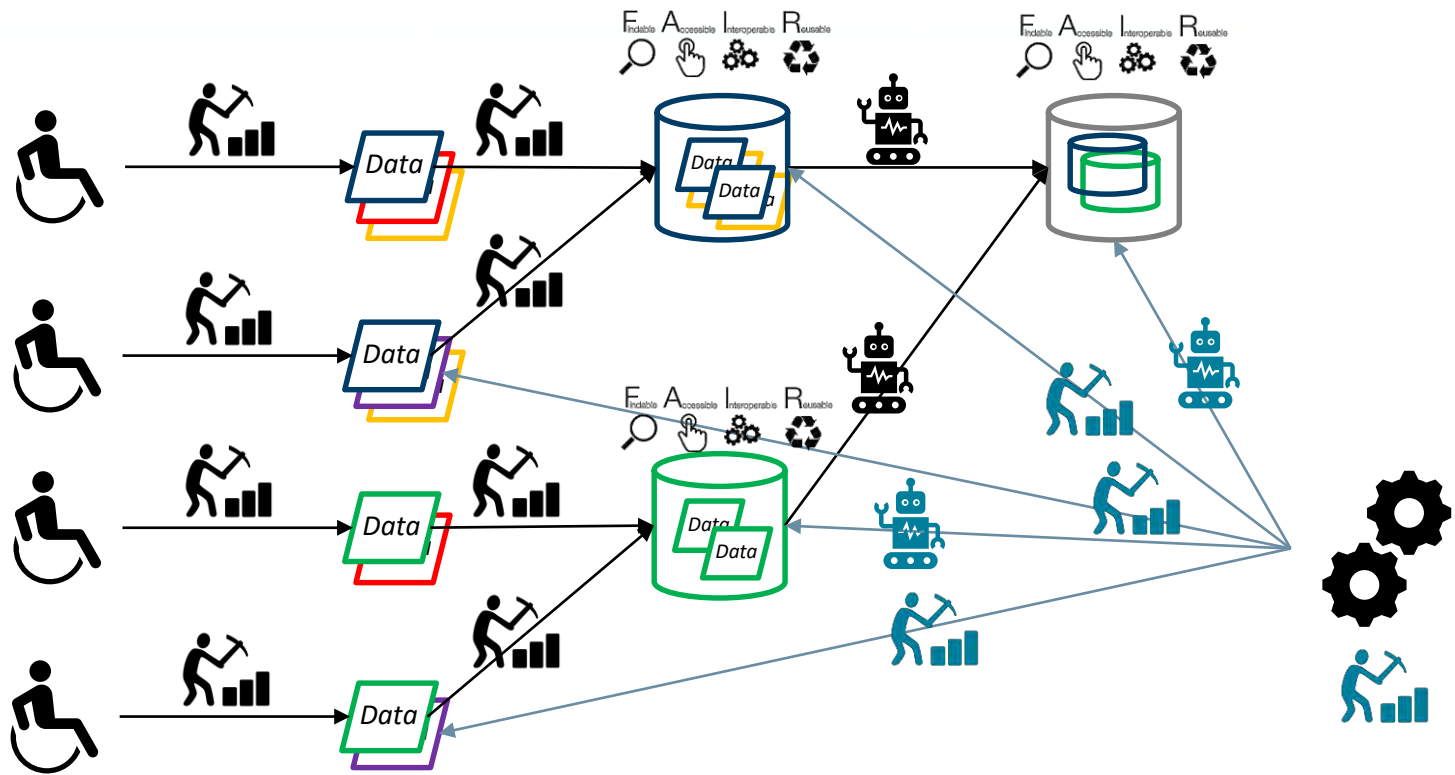
Filter, (pseudo)anonymise, aggregate

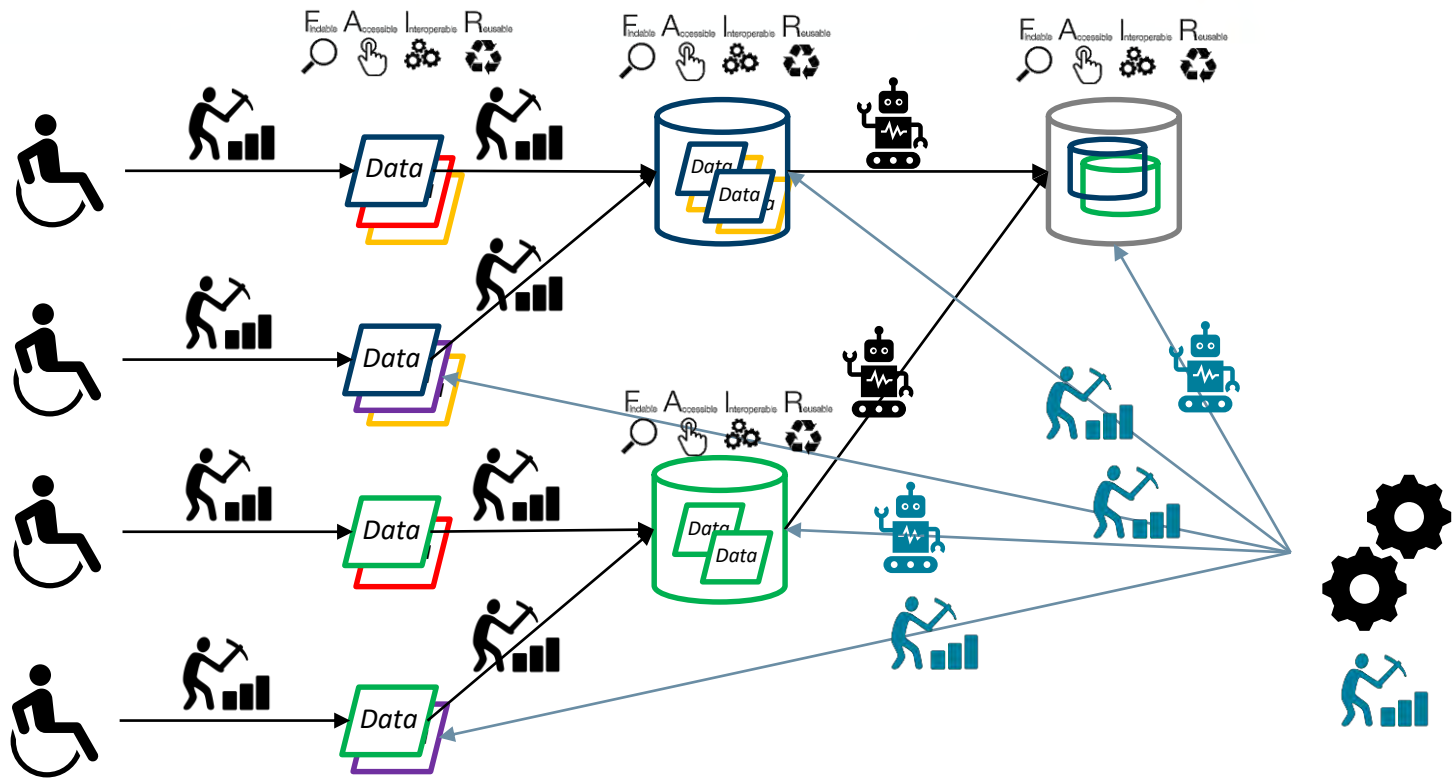


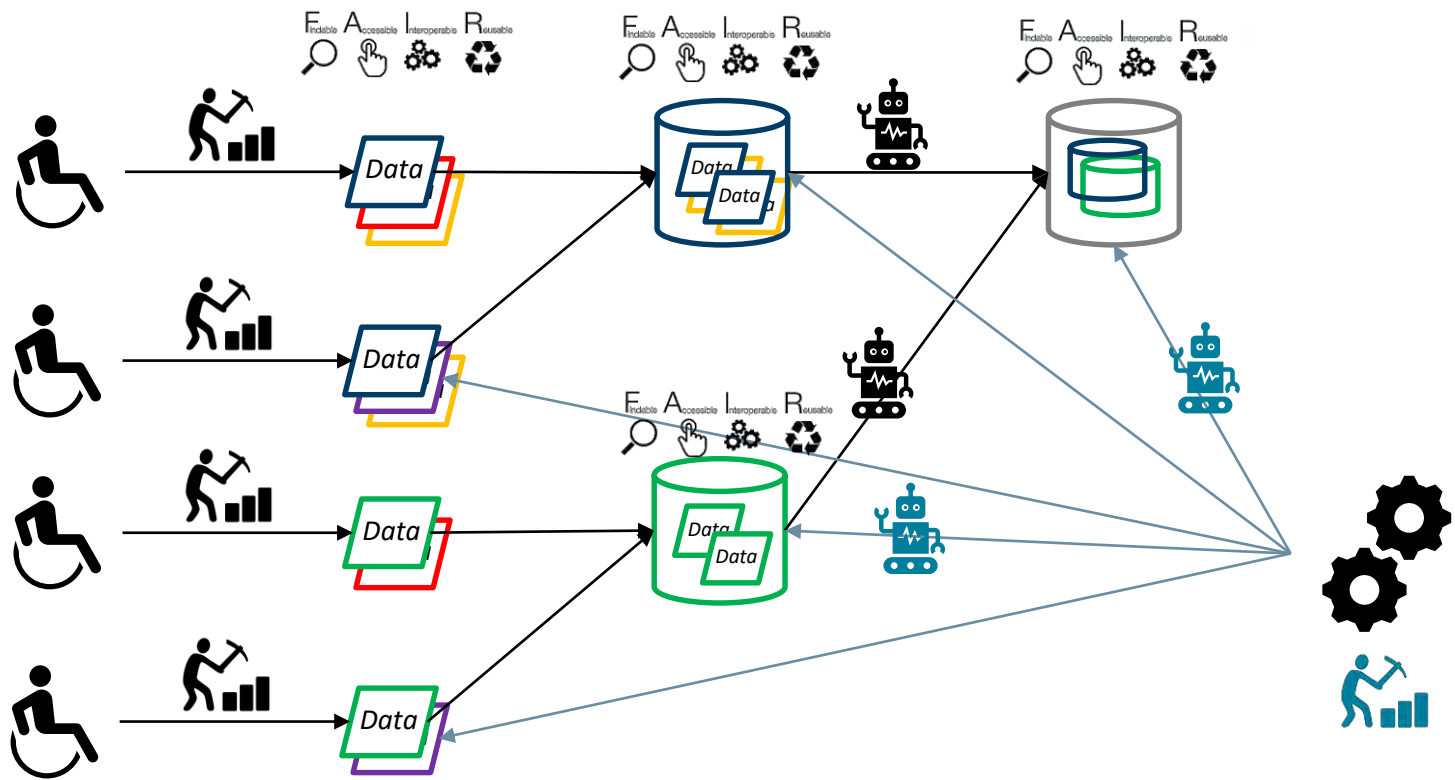


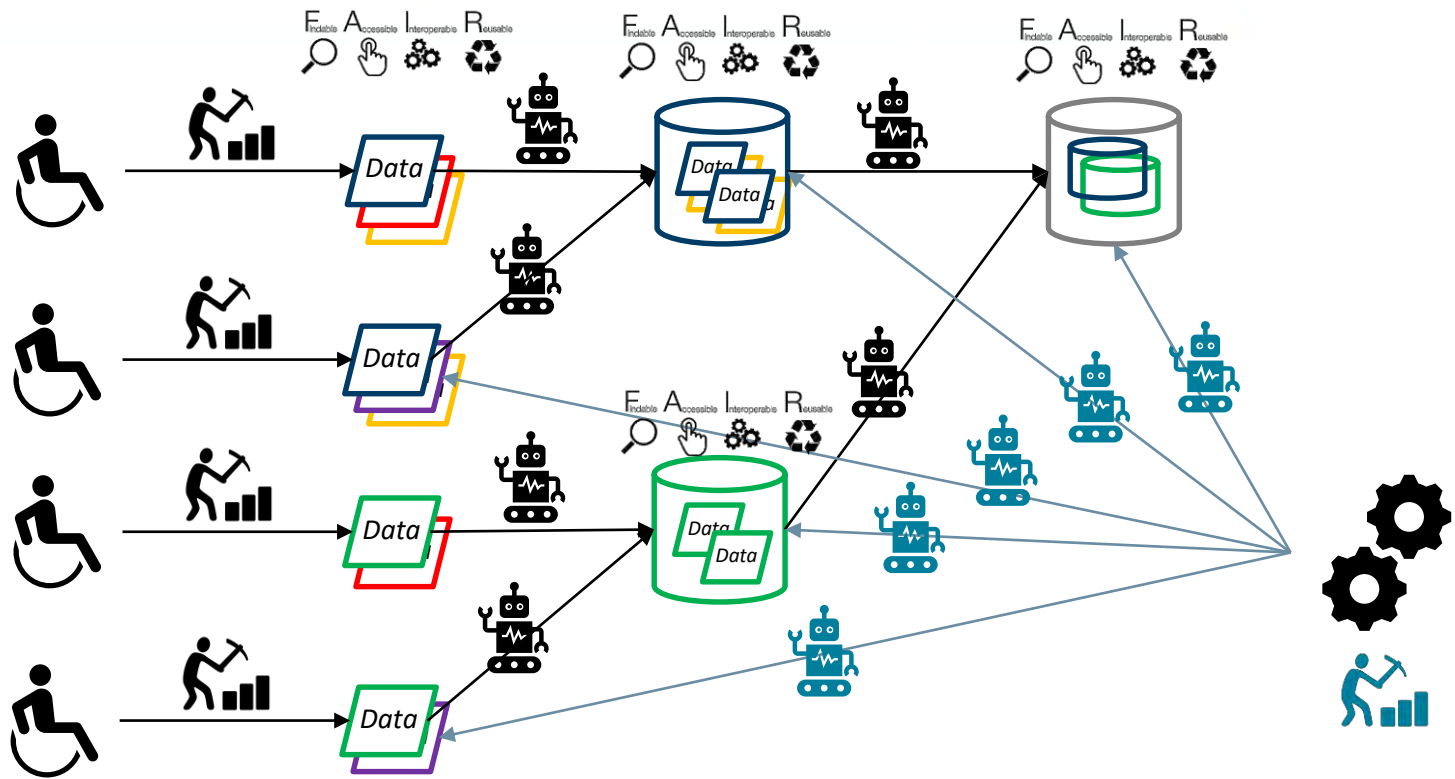




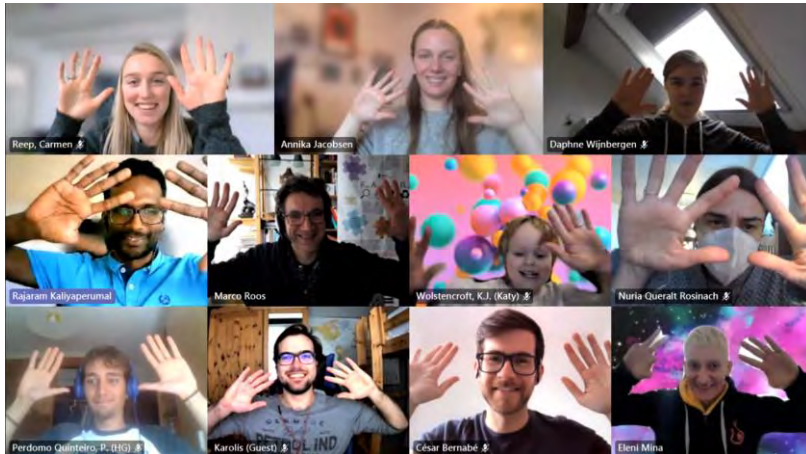








Thank you



Special thanks to the Biosemantics group, rare disease patient representatives, the Duchenne community, The European Joint programme rare diseases, ELIXIR, RD-Connect, the Semantic Web community, many, many more...

Vielen Dank für Ihre Aufmerksamkeit