FAIR DATA PRINCIPLES FOR CROSS-RESOURCE QUESTIONS IN EUROPEAN REFERENCE NETWORKS

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Special acknowledgement: Barend Mons, rare disease patients and representatives, participants of rare disease BYODs and hackathons

To promote and support FAIR RD Registries

This edition dedicated to ERNs.

Are you interested?
Send an e-mail to:
rareregistries-school@iss.it

RD Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Selection panel

Why FAIR data? Interactive experiment: find a treatment for Monica	What can users do with FAIR data? Demo tool to answer questions across biobanks & registries	What is FAIR?	How do I start? Bring Your Own Data workshops
How does FAIR relate to ontologies such as ORDO and HPO?	Who does what to make my data FAIR?	How does FAIRification work? (Technical!)	Why FAIR when we define common data elements?
Has this shown benefit? Example from cancer domain	How do individual persons fit in?	…	Role of ERNs

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Why “Interoperability”

RD-Activists say

- …

Knowing you…

- Research
- Health care
- Policy
- IT / Data management
- Other

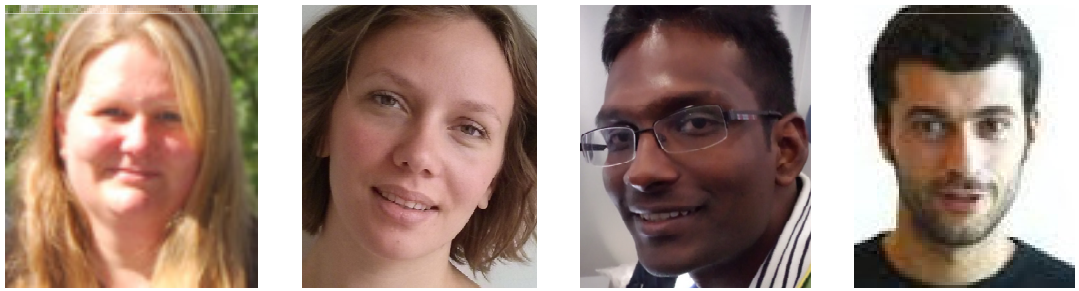
- Producer of data
- Consumer of data
  - Attempted to work with other people's data
- None

Why FAIR?

- Enable questions across resources
- Facilitate linking of **other people's data** to your data
- Lower preparatory effort before data analysis
  (now: ~ 6 months per data set, non-reusable)

Experiment: find the treatment!

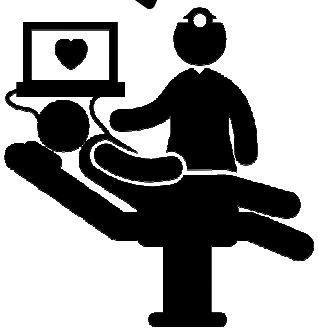
‘Patients’ with a disease, a phenotype, and some with a treatment

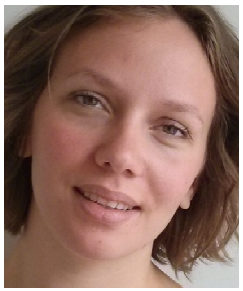


Disclaimer: mock examples!!!!

Experiment: find the treatment!

Which treatment <?T> ***is treating*** phenotype <?P> that is also a phenotype for Monica who ***has disease*** <?D>





Find the treatment experiment

Monika 	Annika 	Rajaram 	Pietro 	FAIR
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	Local registry

Find the treatment experiment

Monika	Annika	Rajaram	Pietro	FAIR
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	Local registry
Ring-14 disease, Salaam seizures, (no treatment)	Ring-14 syndrome, Chorea, (no treatment)	Perry syndrome, Epileptic attacks, lamotrigine	Perry syndrome, extreme mood swings, oxalate	English

Find the treatment experiment

Monika	Annika	Rajaram	Pietro	FAIR
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	Local registry
Ring-14 disease, Salaam seizures, (no treatment)	Ring-14 syndrome, Chorea, (no treatment)	Perry syndrome, Epileptic attacks, lamotrigine	Perry syndrome, extreme mood swings, oxalate	English

Find the treatment experiment

Which treatment <?T> *is treating* phenotype <?P> in a person who *has disease* Ring-14 disease and *has phenotype* <?P>

Monika	Annika	Rajaram	Pietro	FAIR
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	Local registry
Ring-14 disease, Salaam seizures, (no treatment)	Ring-14 syndrome, Chorea, (no treatment)	Perry syndrome, Epileptic attacks, lamotrigine	Perry syndrome, extreme mood swings, oxalate	English

Scenario

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

Monika	Annika	Rajaram	Pietro	FAIR
Ring-14 disease, Salaam seizures, (no treatment)	Ring-14 disease, Chorea, (no treatment)	Perry syndrome, Epileptic attacks, lamotrigine	Perry syndrome, extreme mood swings, oxalate	English
ORPHA1440, HP:0011097	ORPHA1440, HP:0011097	ORPHA178509, HP:0011097, CHEBI_6367	ORPHA178509, HP:0000720 CHEBI_132952	Coded

Putting the pieces together

Which treatment <?T> ***is treating*** phenotype <?P> in any person who ***has disease*** Ring-14 disease and ***has phenotype*** <?P>

ORPHA178509, HP:0000720, CHEBI_132952

ORPHA1440, HP:0011097

ORPHA178509, HP:0011097, CHEBI_6367

ORPHA1440, HP:0002072

Scenario

Monika	Annika	Rajaram	Pietro	FAIR
ORPHA1440, HP:0011097	ORPHA72, HP:00027072	ORPHA178509, HP:0011097, CHEBI_6367	ORPHA178509, HP:0000720 CHEBI_132952	Coded
Monika has disease Ring-14 disease, and has phenotype Salaam seizures	Annika has disease Ring-14 disease, and has phenotype Chorea	Rajaram has disease Perry syndrome, and has phenotype Epileptic seizures. Epileptic seizures are treated by lamotrigine	Pietro has disease Ring-14 disease, and has phenotype Extreme mood swings. Extreme mood swings are treated by the drug Oxalate	full meaning

Scenario

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

Monika	Annika	Rajaram	Pietro	FAIR
Monika has disease Ring-14 disease, and has phenotype Salaam seizures	Annika has disease Ring-14 disease, and has phenotype Chorea	Rajaram has disease Perry syndrome, and has phenotype Epileptic seizures. Epileptic seizures are treated by the drug lamotrigine	Pietro has disease Ring-14 disease, and has phenotype Extreme mood swings. Extreme mood swings are treated by the drug Oxalate	Full meaning
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Monika	Annika	Rajaram	Pietro
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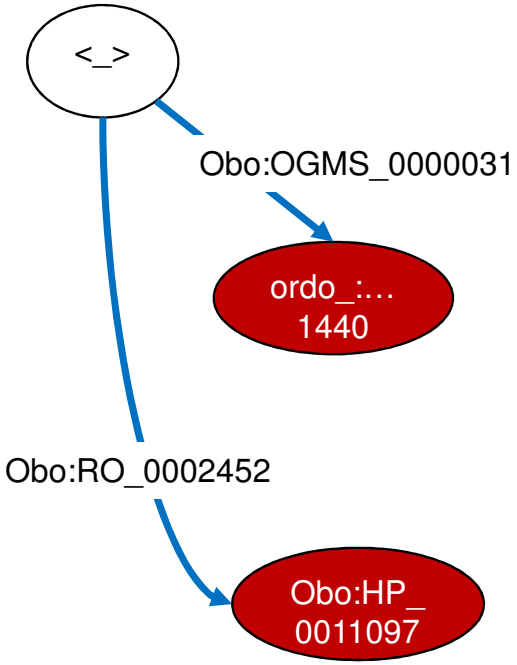

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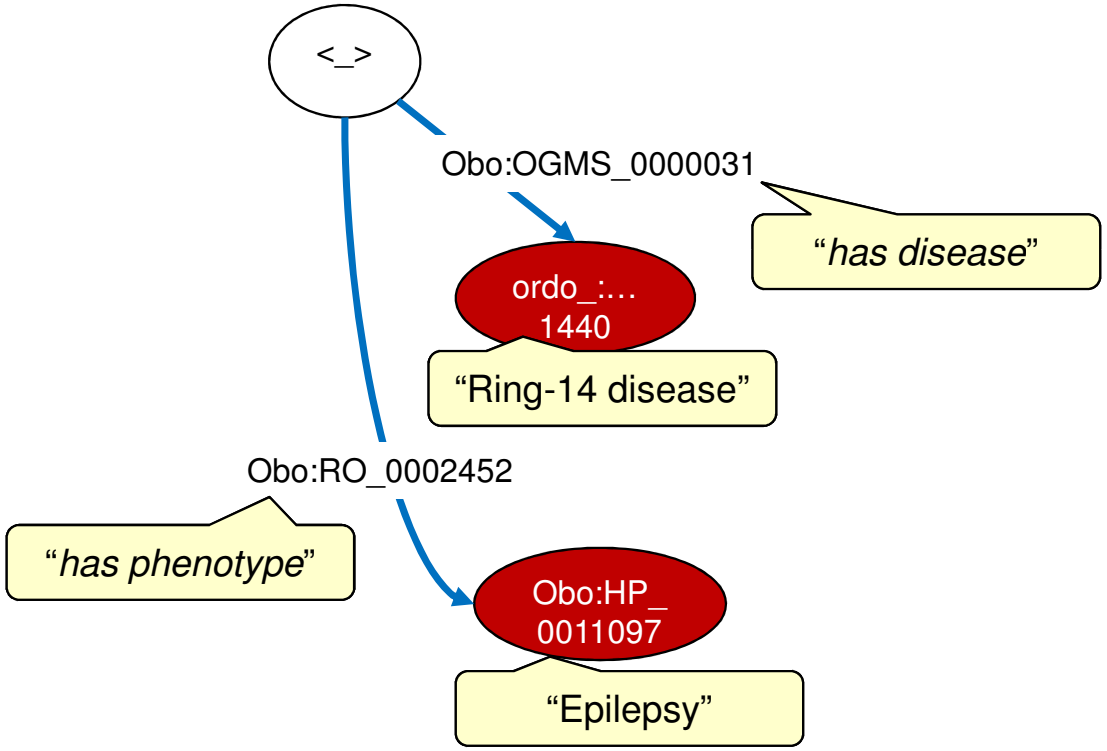

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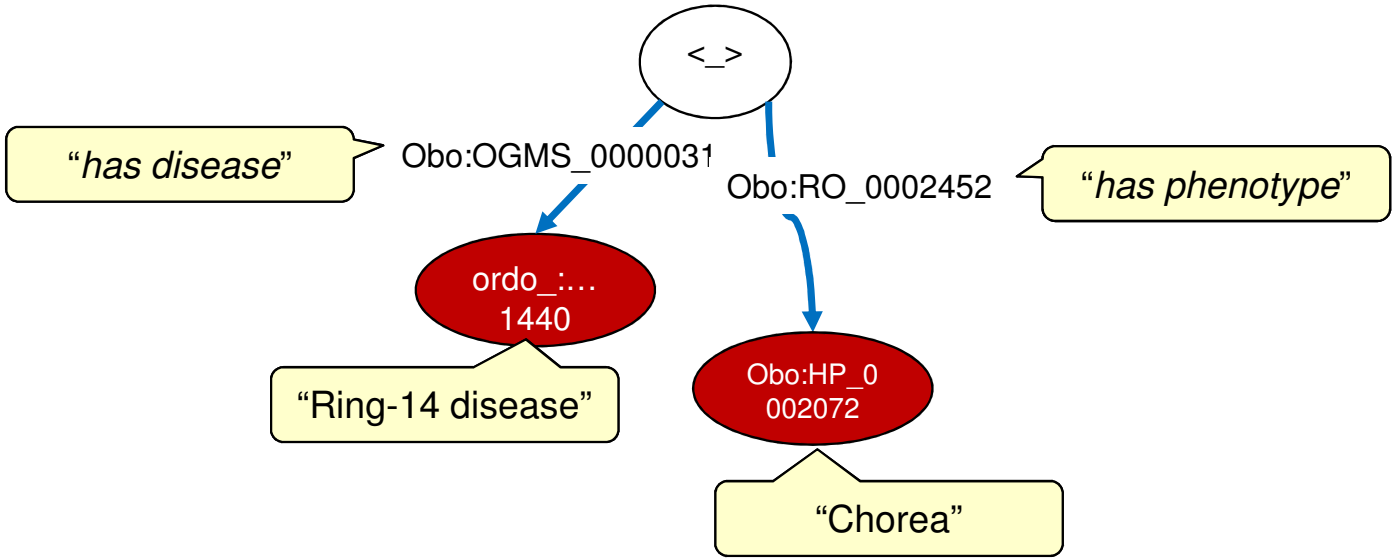

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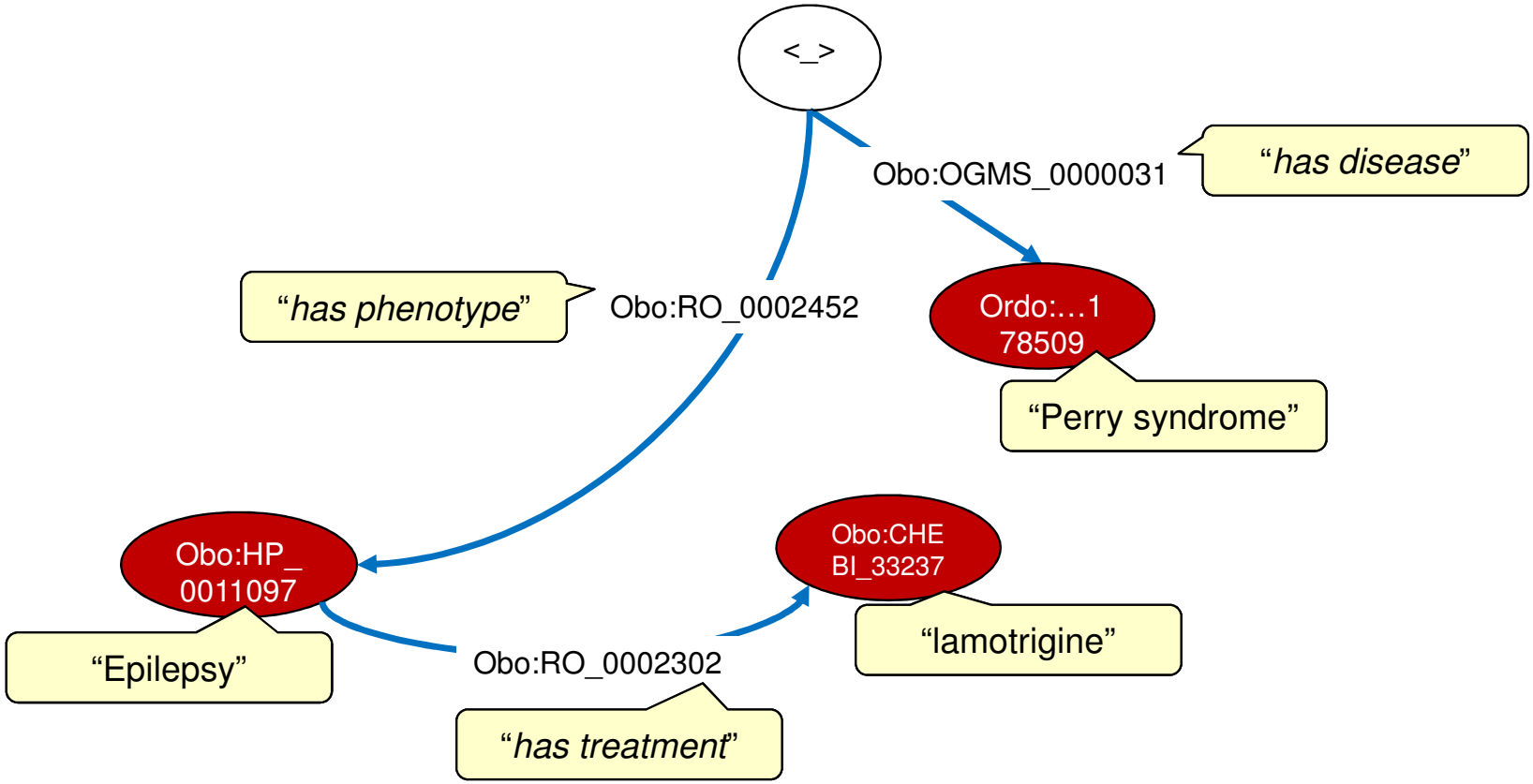

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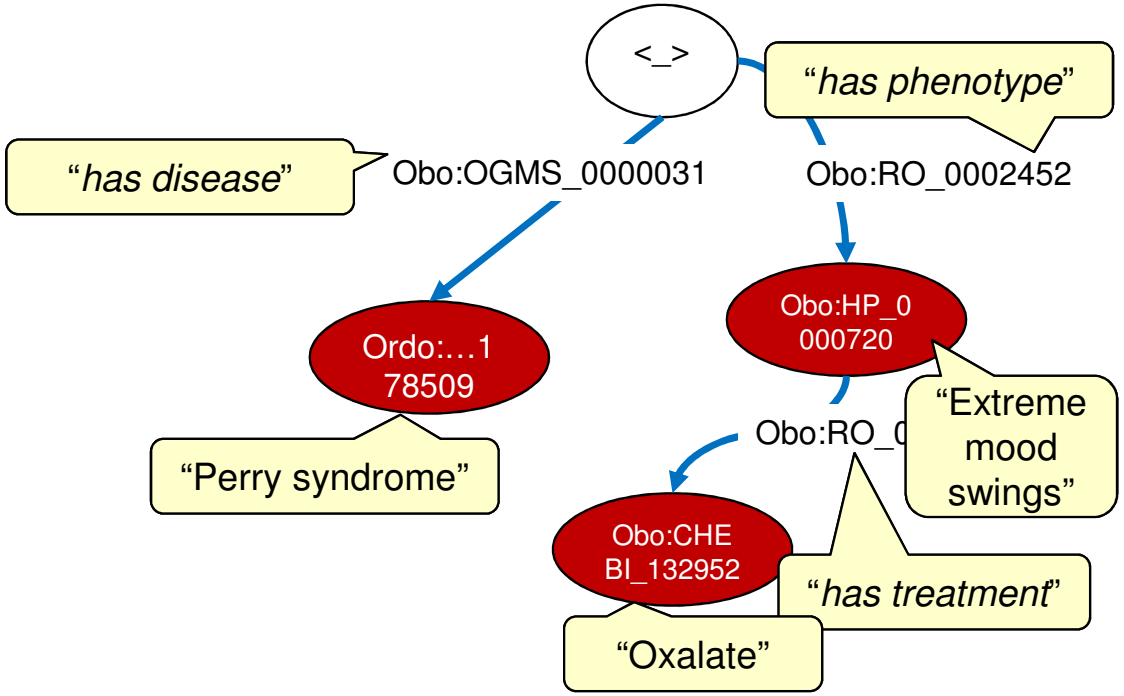
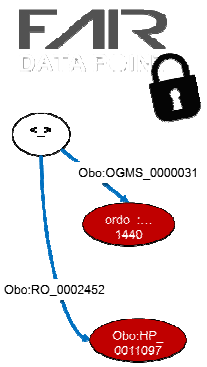
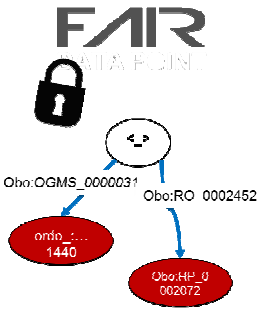
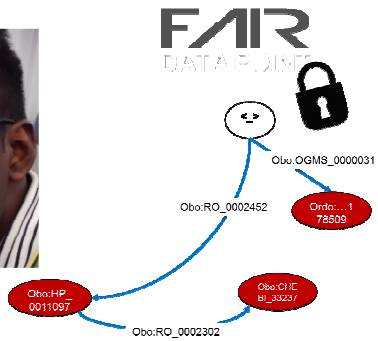


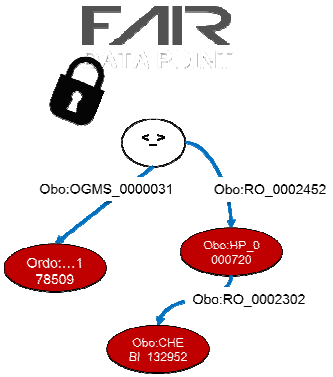
FAIR data landscape





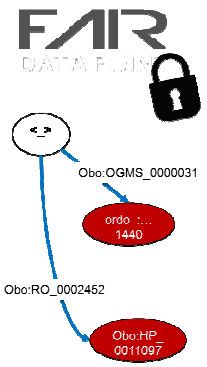
At this stage we have four **independently FAIR*** data sources, under control of the local data manager
(e.g. HCP, patient organisation, patient)



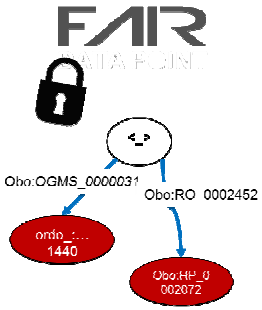


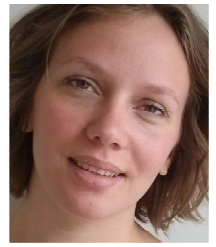
* Strictly speaking, the **interoperability** aspect mostly

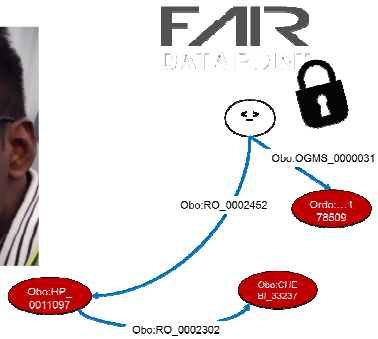
FAIR data landscape

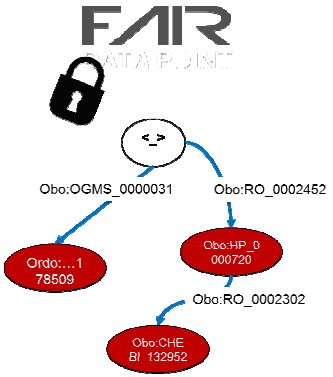


Data at each source is **self-explaining** through global standards that a computer program can understand









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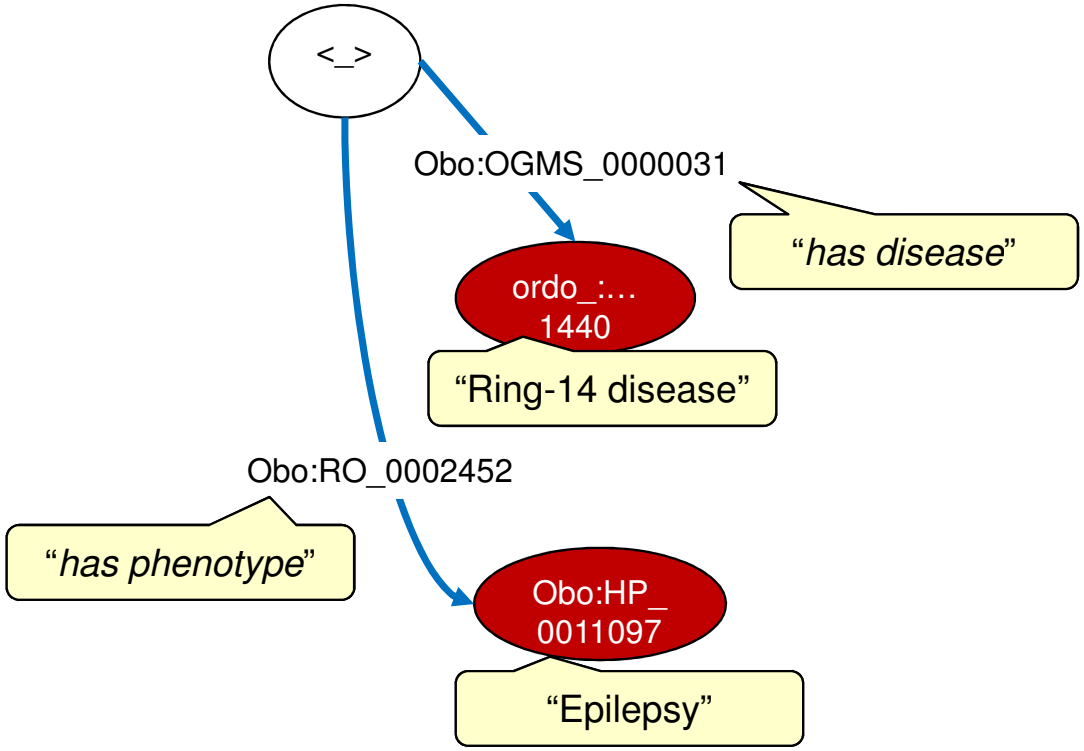

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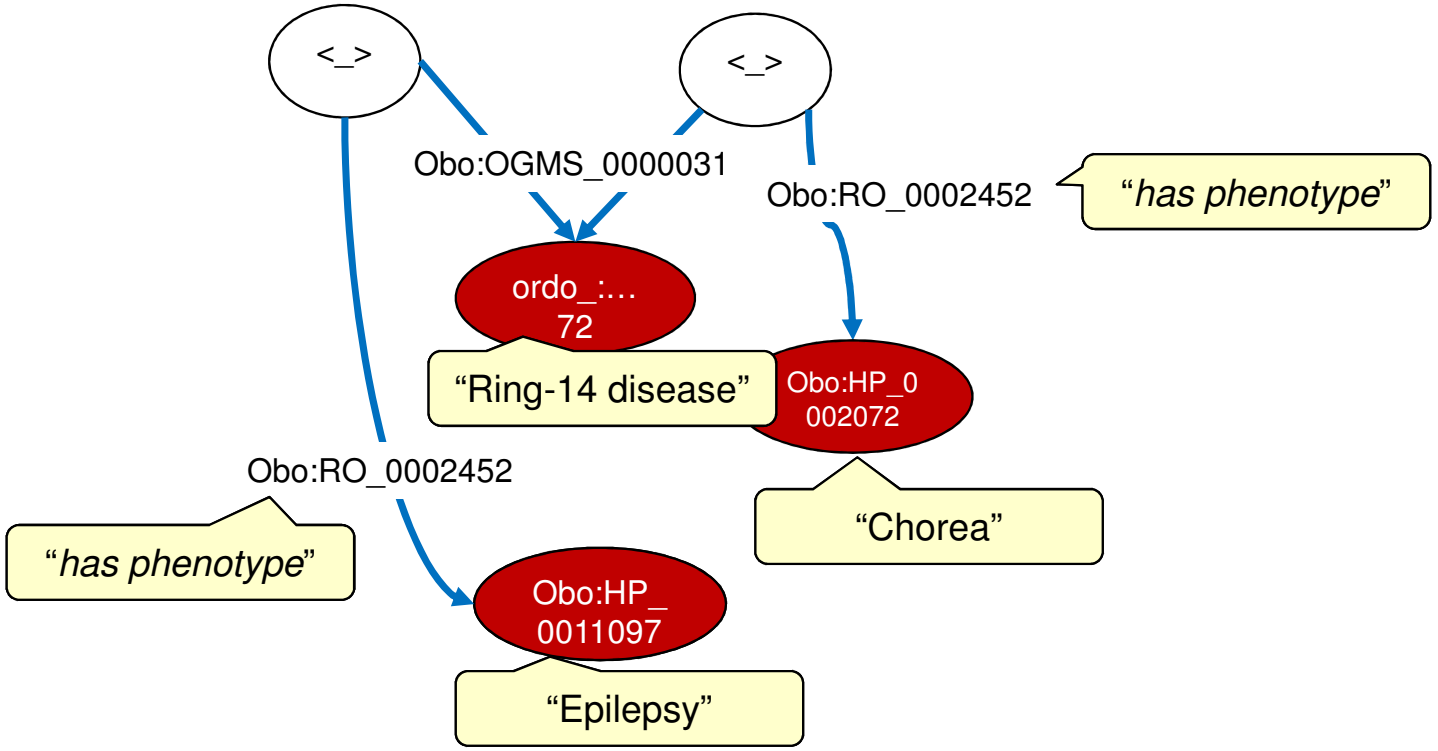

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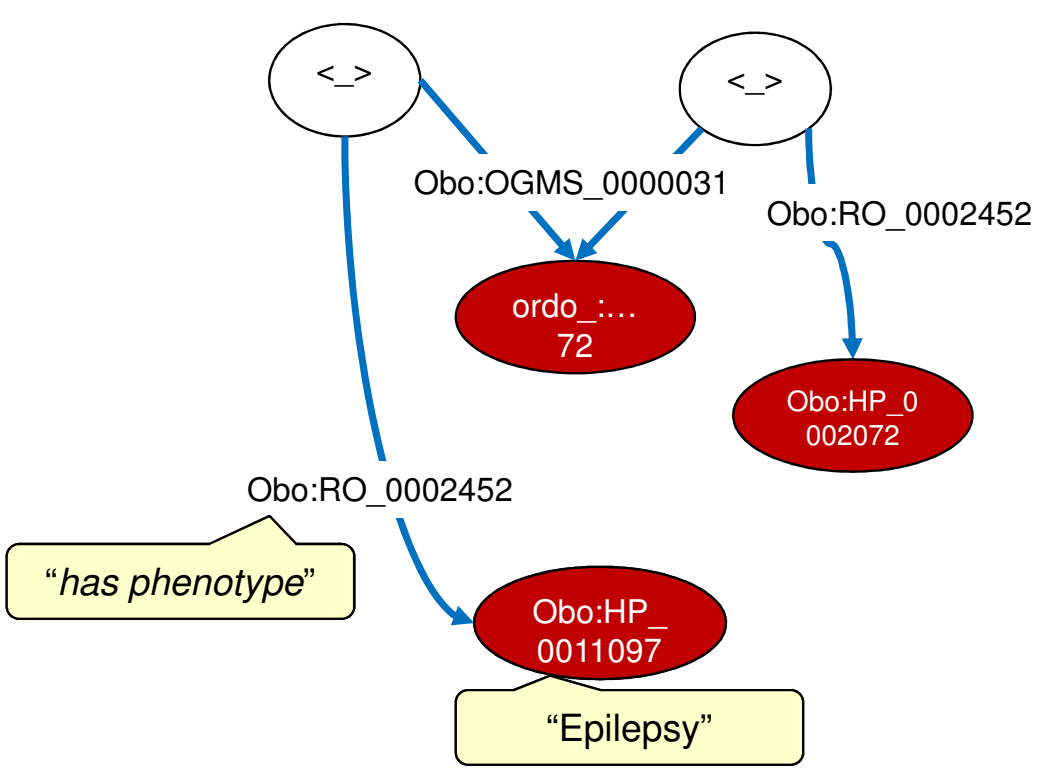

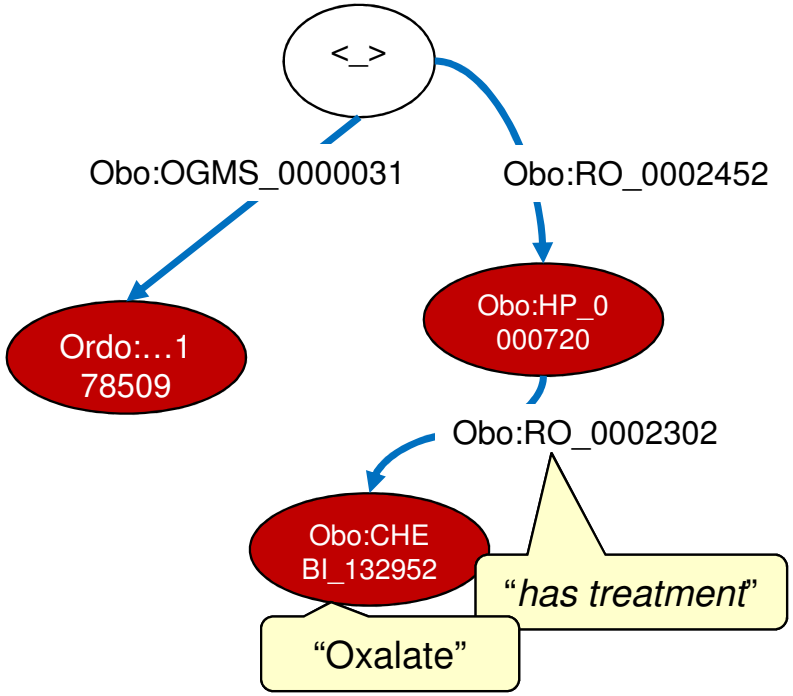

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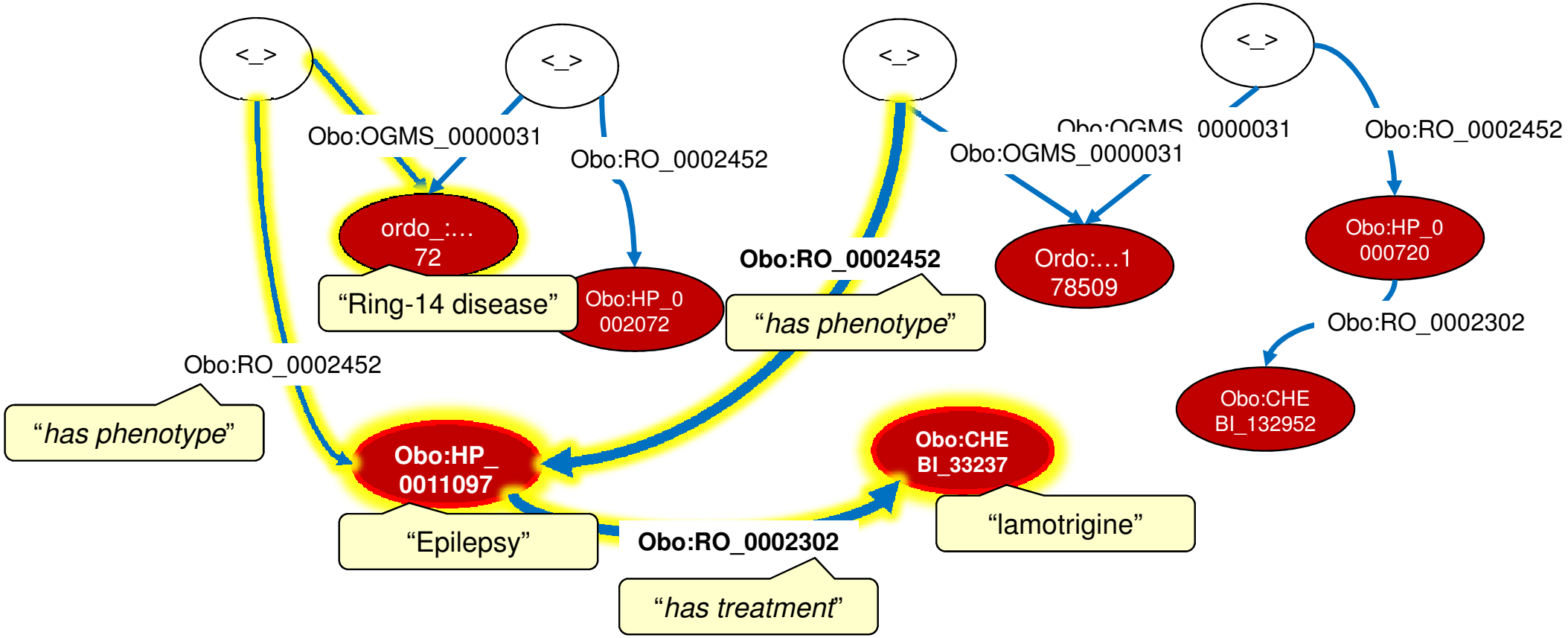

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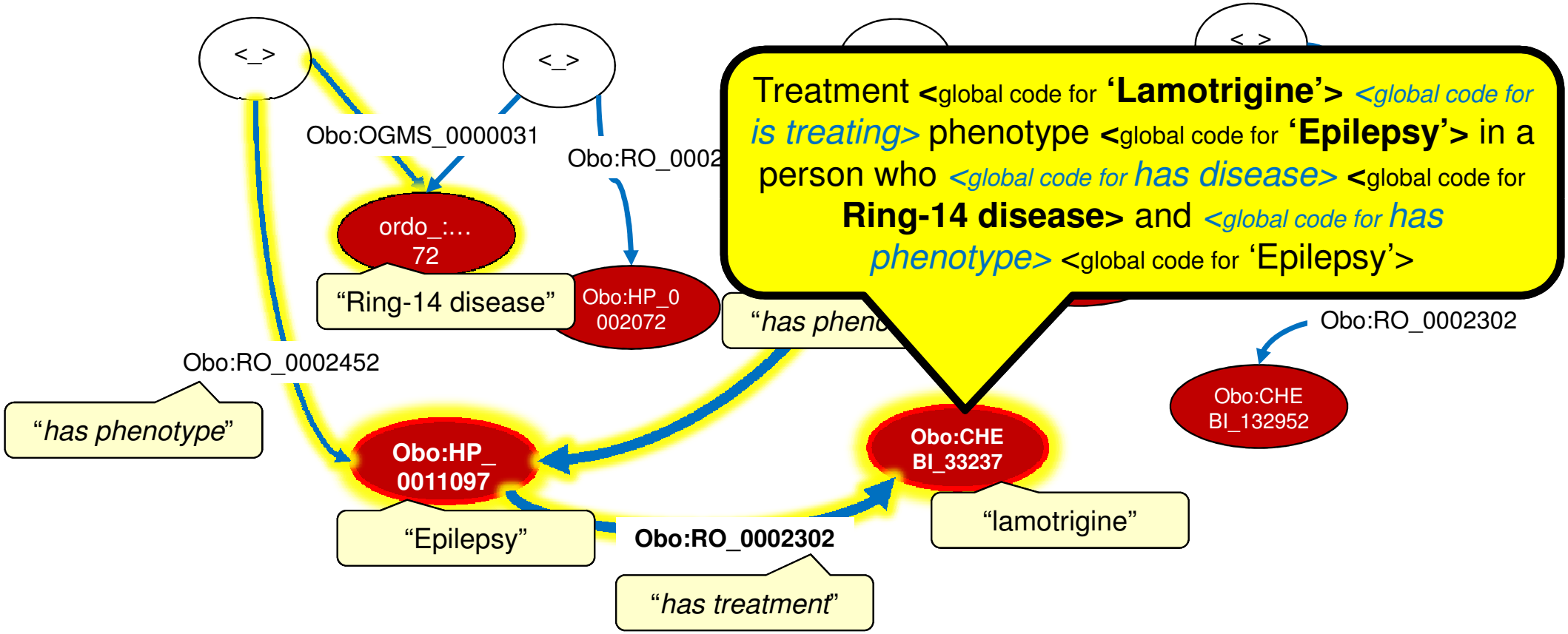
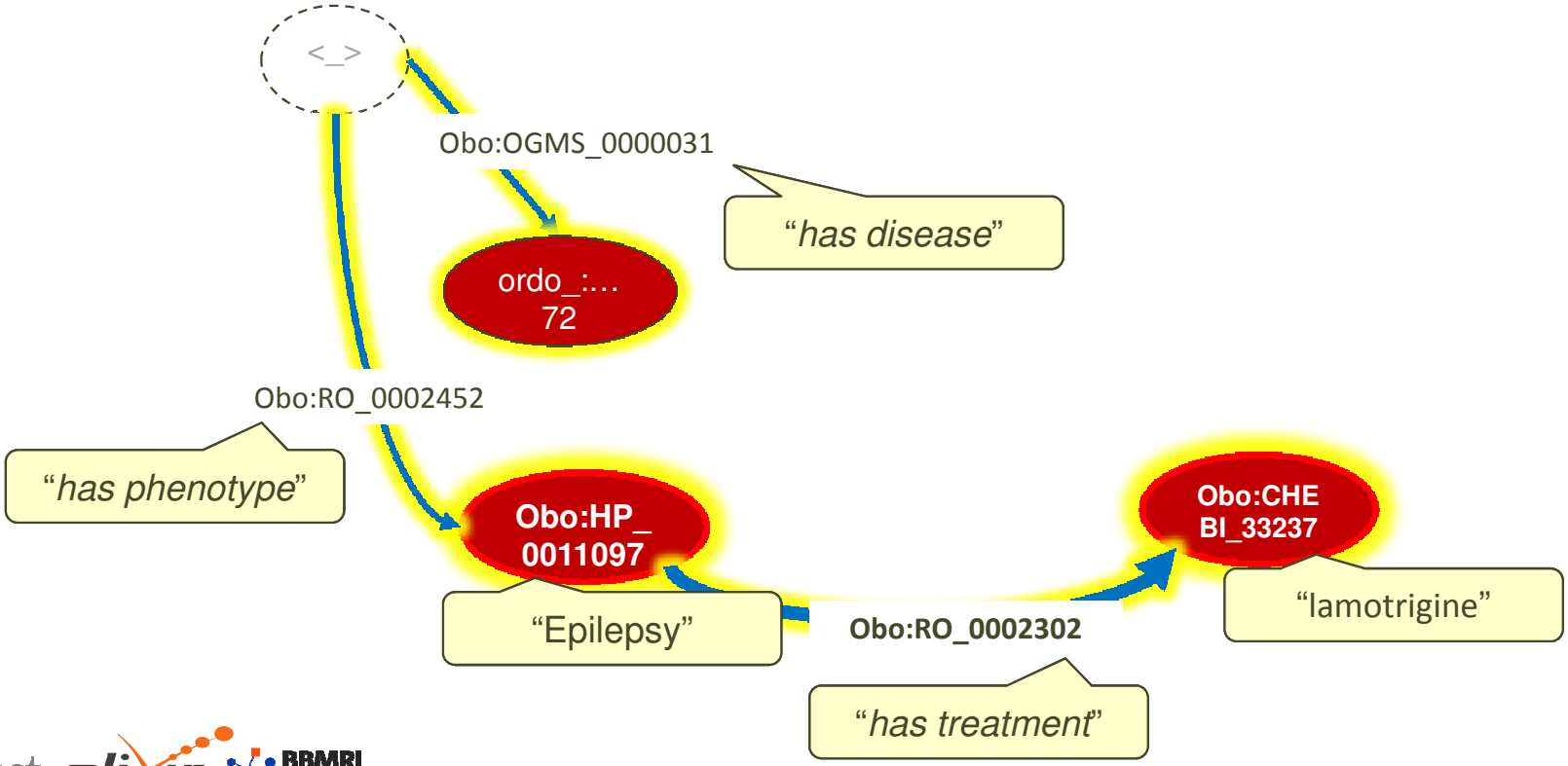


Result

Disease | global machine readable code (URI)		**Treatment** | URI		**Phenotype** | URI	
Ring-14 disease	ordo:Orphanet_1440	Lamotrigine	Obo:CHEBI_33237	Epilepsy	Obo:HP_0011097



RD-Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Results: codes are references to more information!

Disease | global machine readable code (URI)		**Treatment** | URI		**Phenotype** | URI	
Ring-14 disease	ordo:Orphanet_1440	Lamotrigine	Obo:CHEBI_33237	Epilepsy	Obo:HP_0011097

1. http://purl.obolibrary.org/obo/CHEBI_6367 **(CHEBI)**:
   - lamotrigine *in Ontobee*: CHEBI, DRON
   - 6-(2,3-dichlorophenyl)-1,2,4-tri
   - 0 *in Ontobee*: CHEBI
   - InChI=1S/C9H7Cl2N5/c10-5-3
   - 255.008 *in Ontobee*: CHEBI
   - 256.09100 *in Ontobee*: CHEBI
   - 3,5-diamino-6-(2,3-dichlorophenyl)-1,2,4-triazine *in Ontobee*: CHEBI, DRON
   - C9H7Cl2N5 *in Ontobee*: CHEBI, DRON
   - Lamictal *in Ontobee*: CHEBI, DRON
   - Nc1nnc(c(N)n1)-c1cccc(Cl)c1Cl *in Ontobee*: CHEBI, DRON
   - PYZRQGJRPPTADH-UHFFFAOYSA-N *in Ontobee*: CHEBI
   - lamotrigine *in Ontobee*: CHEBI, DRON
   - lamotriginum *in Ontobee*: CHEBI, DRON
   - InChIKey=PYZRQGJRPPTADH-UHFFFAOYSA-N *in Ontobee*: DRON

Class:lamotrigine Definition: A member of the class of 1,2,4-triazines in which the triazene skeleton is substituted by amino groups at positions 3 and 5, and by a 2,3-dichlorophenyl group at position 6.

1. http://purl.obolibrary.org/obo/HP_0011097 **(HP)**:
   - Epileptic spasms *in Ontobee*:
   - Salaam convulsions *in Ontobe*
   - Salaam seizures *in Ontobee*: H
   - West syndrome *in Ontobee*: H

Class:Epileptic spasms Definition: A sudden flexion, extension or mixed extension-flexion of predominantly proximal and truncal muscles which is usually more sustained than a myoclonic movement but not as sustained as a tonic seizure.

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Scenario

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

Monika	Annika	Rajaram	Pietro	FAIR
Monika has disease Ring-14 disease, and has phenotype Salaam seizures	Annika has disease Ring-14 disease, and has phenotype Chorea	Rajaram has disease Perry syndrome, and has phenotype Epileptic seizures. Epileptic seizures are treated by the drug lamotrigine	Pietro has disease Ring-14 disease, and has phenotype Extreme mood swings. Extreme mood swings are treated by the drug Oxalate	Full meaning
<_> obo:OGMS_0000031 ordo:Orphanet_1440 obo:RO_0002452 obo:HP_0011097.	<_> obo:OGMS_0000031 ordo:Orphanet_72, obo:RO_0002452 obo:HP_0002072.	<_> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0011097 obo:RO_0002302 :CHEBI_33237	<_> obo:OGMS_0000031 ordo:Orphanet_178509, obo:RO_0002452 obo:HP_0000720 obo:RO_0002302 obo:CHEBI_132952	Interoperable & Machine readable

Is there sensitive data in these data?

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Results: codes are references to more information!

Disease | global machine readable code (URI)		**Treatment** | URI		**Phenotype** | URI	
Ring-14 disease	ordo:Orphanet_1440	Lamotrigine	Obo:CHEBI_33237	Epilepsy	Obo:HP_0011097

Is there sensitive data in these data?

Results: codes are references to more information!

Disease | global machine readable code (URI)		**Treatment** | URI		**Phenotype** | URI	
Ring-14 disease	ordo:Orphanet_1440	Lamotrigine	Obo:CHEBI_33237	Epilepsy	Obo:HP_0011097

The ‘personal health train’ exploits this: algorithms travel to secure (FAIR) local data; no sensitive data is exposed outside of the FAIR data points

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Answer questions across rare disease resources
Demonstration web tool, biobanks and registries

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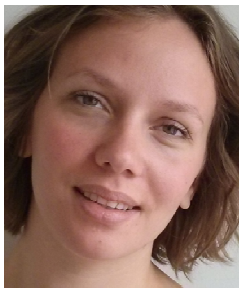
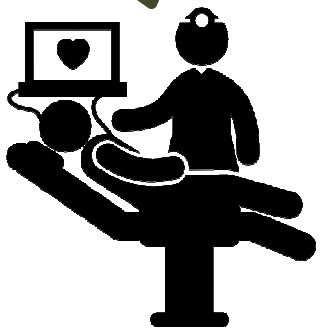
BBMRI
Biobanking and Biomolecular Resources Research Infrastructure
RD Connect

Experiment 1: find a treatment?

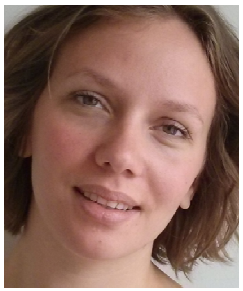
Which **treatment** <?T> *is treating* **phenotype** <?P> in a person who *has disease* Ring-14 **disease** and *has phenotype* <?P>

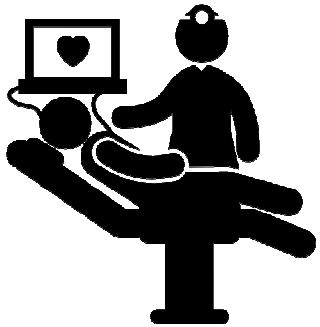



FAIR data point: gateway to cross-resource questions

Where can I obtain **blood samples** from donors that have an **abnormality of head or neck**?

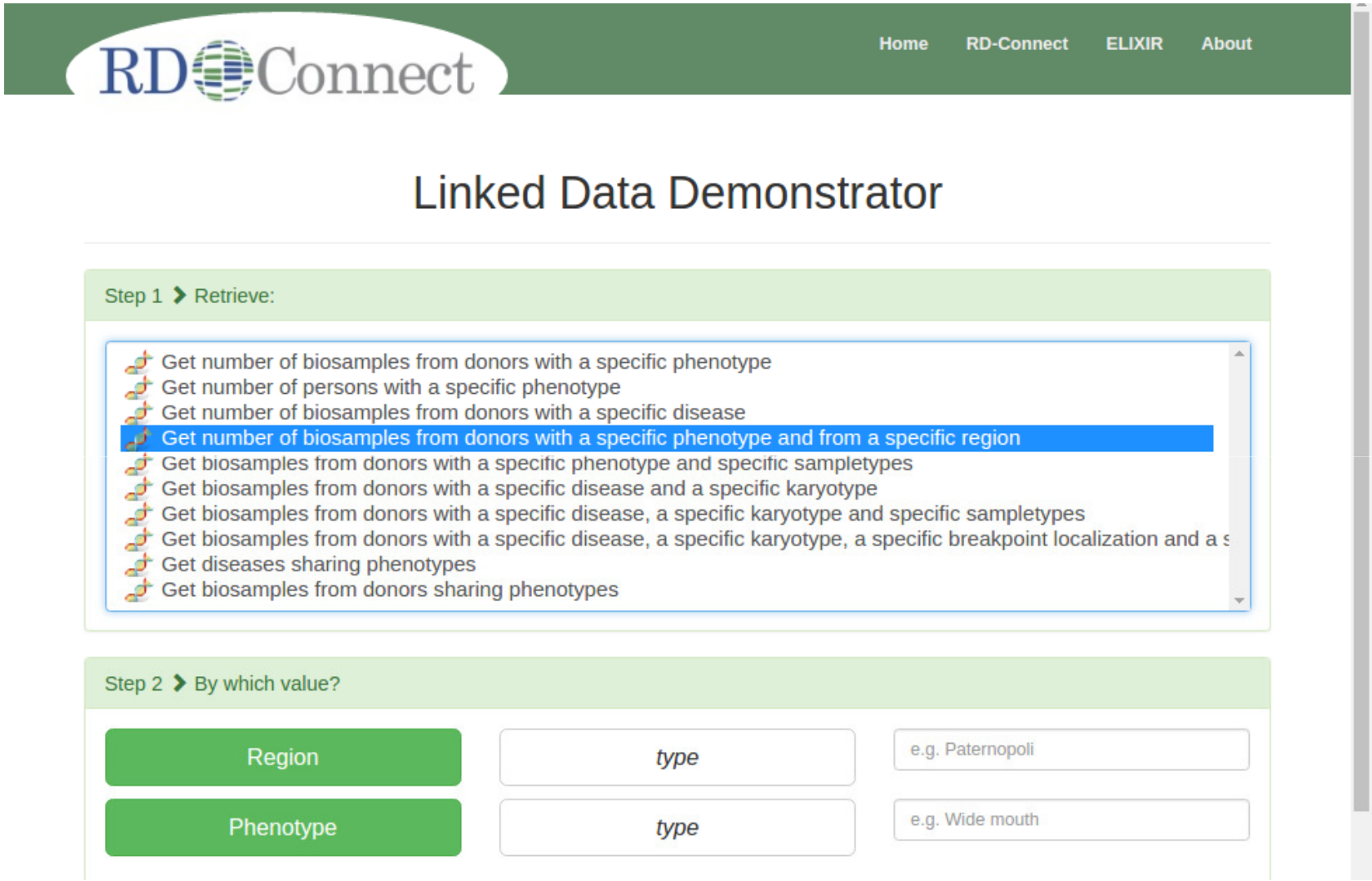



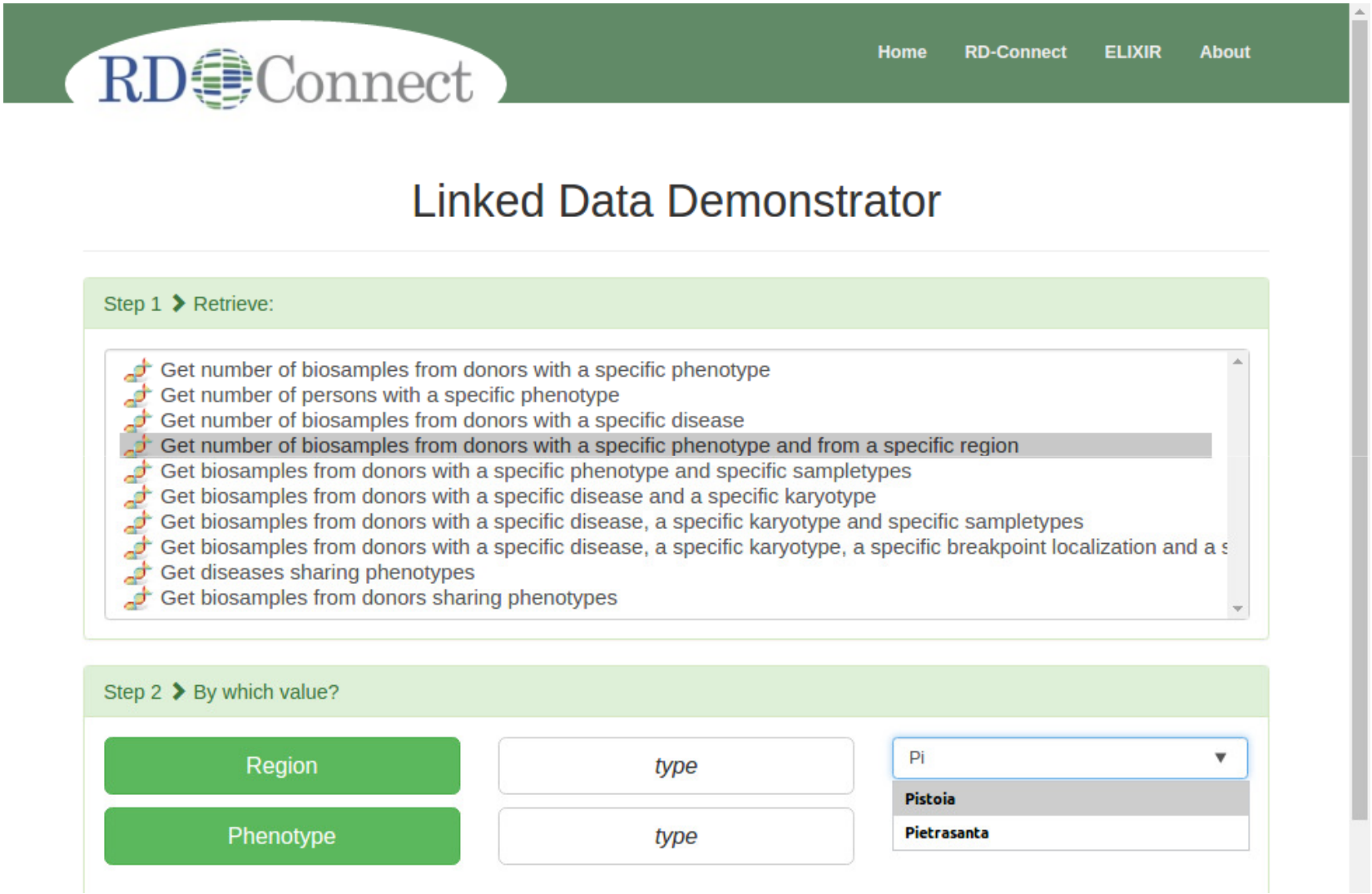



Linked Data Demonstrator

Step 1 > Retrieve:

- Get number of biosamples from donors with a specific phenotype
- Get number of persons with a specific phenotype
- Get number of biosamples from donors with a specific disease
- Get number of biosamples from donors with a specific phenotype and from a specific region
- Get biosamples from donors with a specific phenotype and specific sampletypes
- Get biosamples from donors with a specific disease and a specific karyotype
- Get biosamples from donors with a specific disease, a specific karyotype and specific sampletypes
- Get biosamples from donors with a specific disease, a specific karyotype, a specific breakpoint localization and a s
- Get diseases sharing phenotypes
- Get biosamples from donors sharing phenotypes


RD Connect
Home
RD-Connect
ELIXIR
About
Linked Data Demonstrator
Step 1 > Retrieve:
Get number of biosamples from donors with a specific phenotype
Get number of persons with a specific phenotype
Get number of biosamples from donors with a specific disease
Get number of biosamples from donors with a specific phenotype and from a specific region
Get biosamples from donors with a specific phenotype and specific sampletypes
Get biosamples from donors with a specific disease and a specific karyotype
Get biosamples from donors with a specific disease, a specific karyotype and specific sampletypes
Get biosamples from donors with a specific disease, a specific karyotype, a specific breakpoint localization and a s
Get diseases sharing phenotypes
Get biosamples from donors sharing phenotypes
Step 2 > By which value?
Region
type
e.g. Paternopoli
Phenotype
type
e.g. Wide mouth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Home
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Linked Data Demonstrator
Step 1 ❯ Retrieve:
Get number of biosamples from donors with a specific phenotype
Get number of persons with a specific phenotype
Get number of biosamples from donors with a specific disease
Get number of biosamples from donors with a specific phenotype and from a specific region
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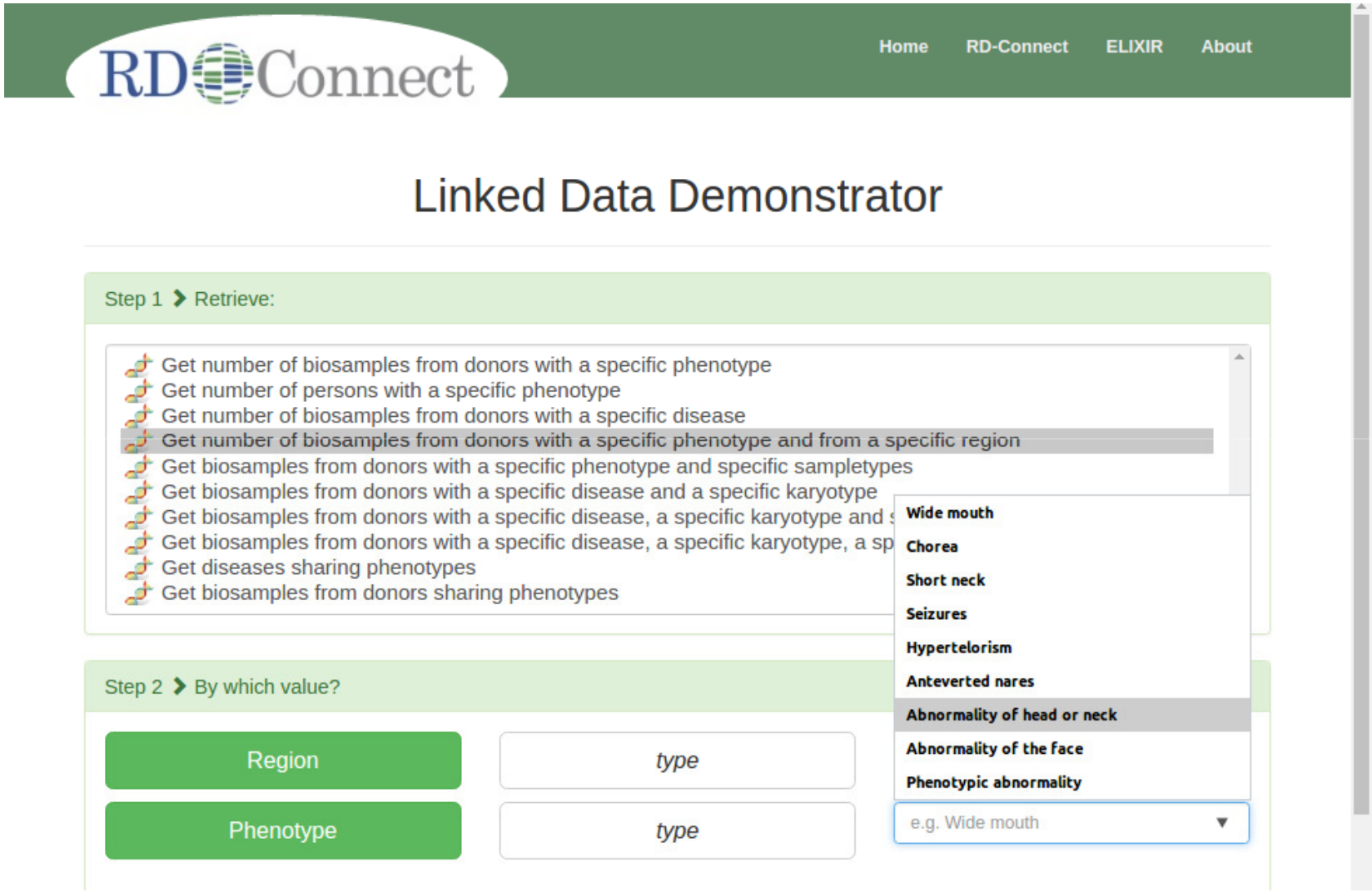
Get biosamples from donors with a specific disease and a specific karyotype
Get biosamples from donors with a specific disease, a specific karyotype and specific sampletypes
Get biosamples from donors with a specific disease, a specific karyotype, a specific breakpoint localization and a s
Get diseases sharing phenotypes
Get biosamples from donors sharing phenotypes
Step 2 ❯ By which value?
Region
type
Pi
Pistoia
Pietrasanta
Phenotype
type


RD Connect
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Get number of biosamples from donors with a specific disease
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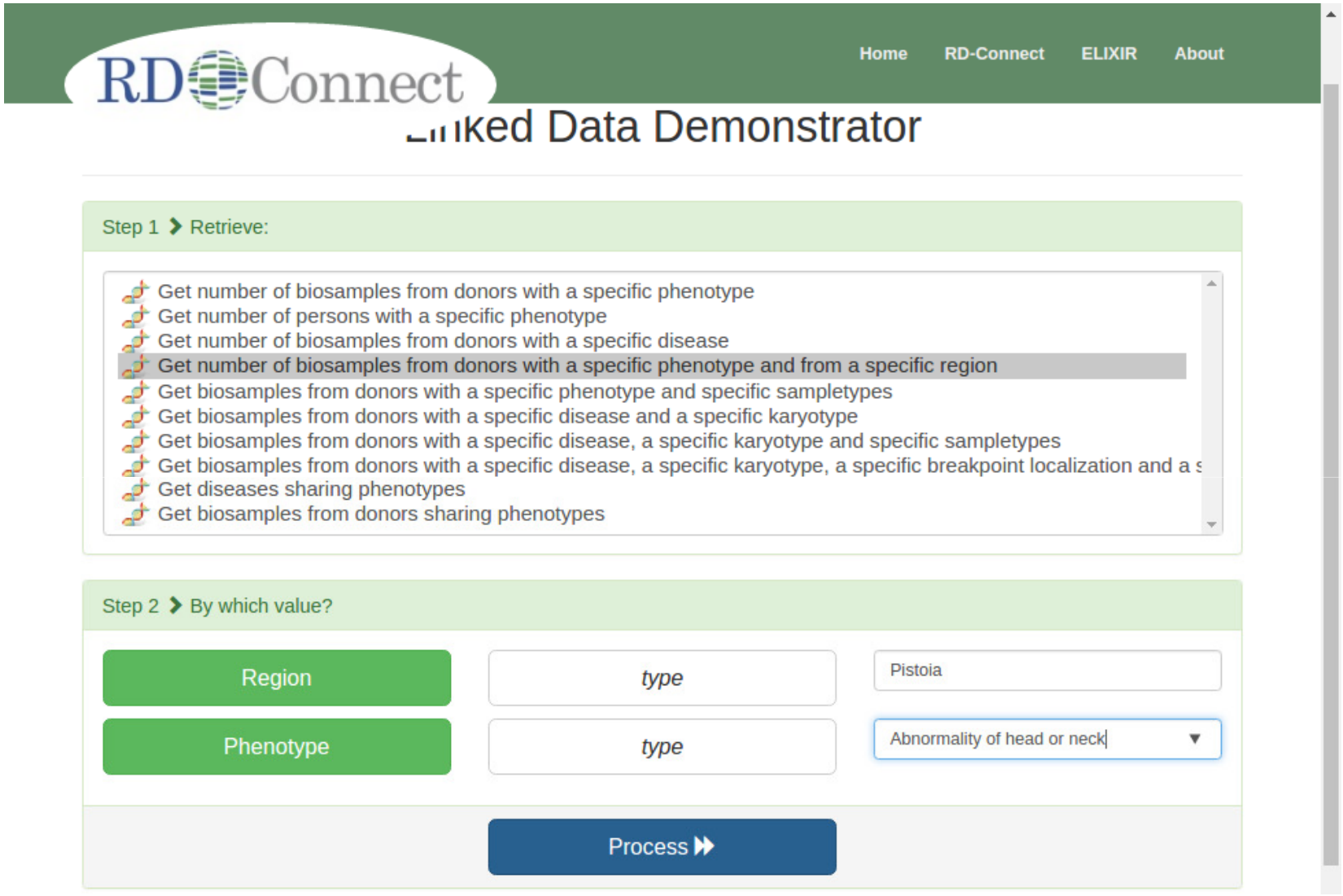
Get biosamples from donors with a specific phenotype and specific sampletypes
Get biosamples from donors with a specific disease and a specific karyotype
Get biosamples from donors with a specific disease, a specific karyotype and s
Get biosamples from donors with a specific disease, a specific karyotype, a sp
Get diseases sharing phenotypes
Get biosamples from donors sharing phenotypes
Wide mouth
Chorea
Short neck
Seizures
Hypertelorism
Anteverted nares
Abnormality of head or neck
Abnormality of the face
Phenotypic abnormality
Step 2 > By which value?
Region
type
Phenotype
type
e.g. Wide mouth


RD Connect
Home
RD-Connect
ELIXIR
About
Linked Data Demonstrator
Step 1 Retrieve:
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Get number of persons with a specific phenotype
Get number of biosamples from donors with a specific disease
Get number of biosamples from donors with a specific phenotype and from a specific region
Get biosamples from donors with a specific phenotype and specific sampletypes
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Get biosamples from donors with a specific disease, a specific karyotype, a specific breakpoint localization and a
Get diseases sharing phenotypes
Get biosamples from donors sharing phenotypes
Step 2 By which value?
Region
type
Pistoia
Phenotype
type
Abnormality of head or neck
Process

Process

Step 3 › Result:

numberOfSamples	phenotype	disease	biobank	registry	region
5	Downslanted palpebral fissures	Ring chromosome 14	Galliera Genetic Bank	Ring14 Clinical database	Pistoia
5	Anteverted nares	Ring chromosome 14	Galliera Genetic Bank	Ring14 Clinical database	Pistoia
1	Mandibular prognathia	Angelman syndrome	Galliera Genetic Bank	Tuscany registry of congenital defects	Pistoia
3	Depressed nasal bridge	Ataxia-telangiectasia	Biobank of the Institute of Rare Diseases Research/Institute of Health Carlos III (IIER-ISCIII)	CoF-AT study: a French cohort on ataxia-telangiectasia	Pistoia
5	Depressed nasal bridge	Ring chromosome 14	Galliera Genetic Bank	Ring14 Clinical database	Pistoia
2	Anteverted nares	Ataxia-telangiectasia	Biobank of the Institute of Rare Diseases Research/Institute of Health Carlos III (IIER-ISCIII)	CoF-AT study: a French cohort on ataxia-telangiectasia	Pistoia

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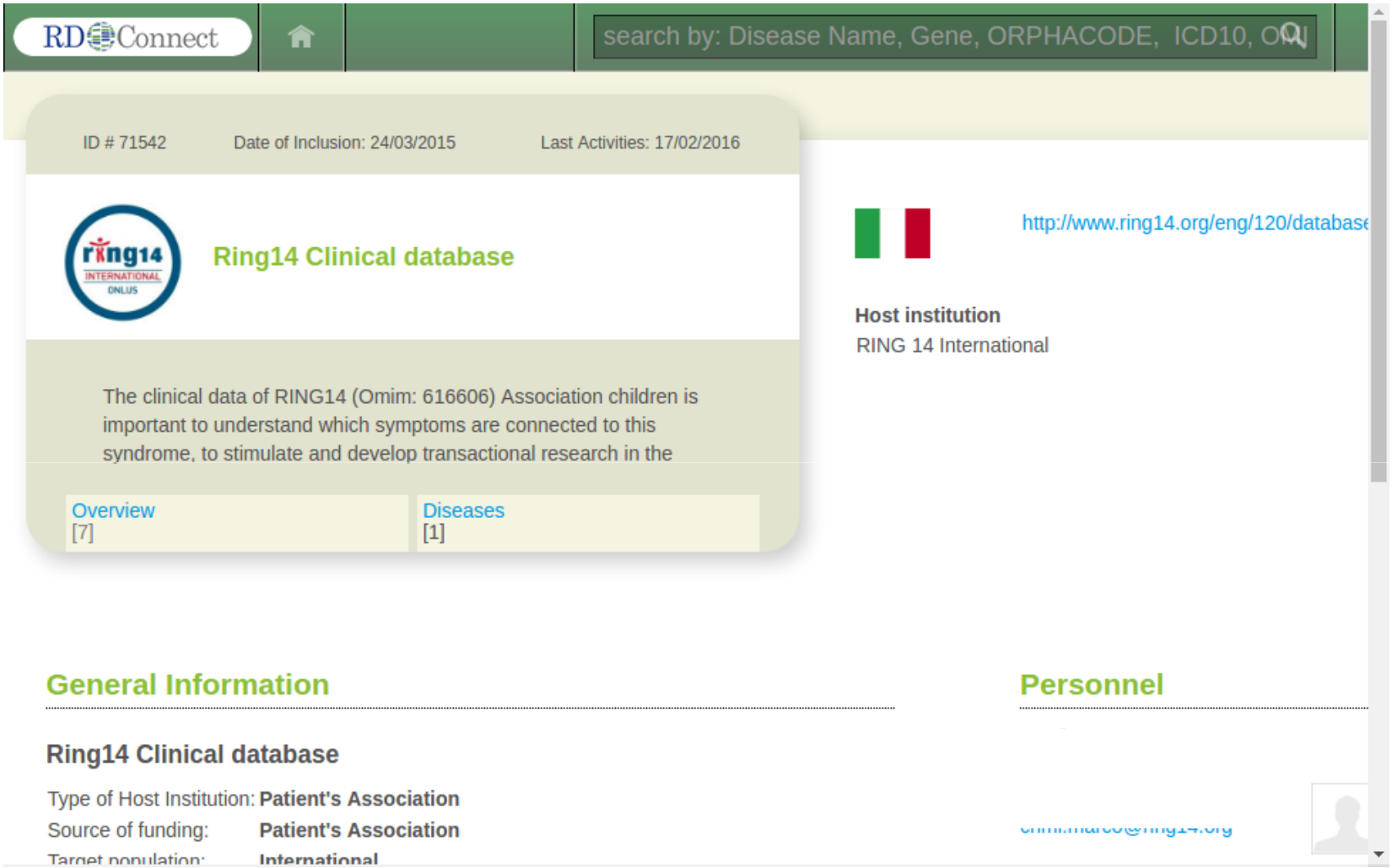
RD Connect
search by: Disease Name, Gene, ORPHACODE, ICD10, OM
ID # 71542
Date of Inclusion: 24/03/2015
Last Activities: 17/02/2016
Ring14 Clinical database
The clinical data of RING14 (Omim: 616606) Association children is important to understand which symptoms are connected to this syndrome, to stimulate and develop transactional research in the
Overview
[7]
Diseases
[1]
http://www.ring14.org/eng/120/database
Host institution
RING 14 International
General Information
Personnel
Ring14 Clinical database
Type of Host Institution: Patient's Association
Source of funding: Patient's Association
Target population: International

numberOfSamples	phenotype	disease	biobank	registry	region
5	Downslanted palpebral fissures	Ring chromosome 14	Galliera Genetic Bank	Ring14 Clinical database	Pistoia
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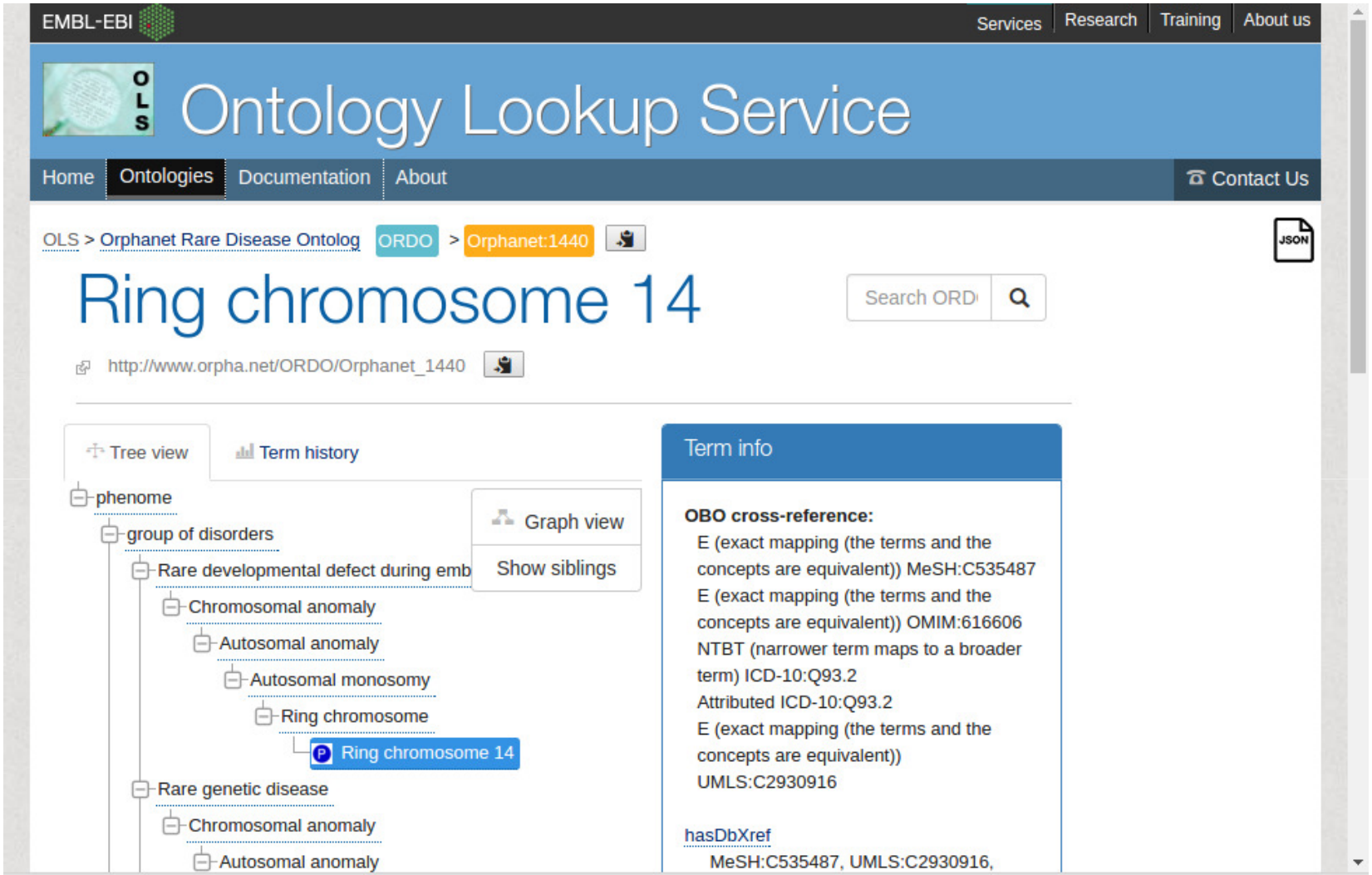
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OLS > Orphanet Rare Disease Ontolog
ORDO
> Orphanet:1440
JSON
Ring chromosome 14
Search ORD
http://www.orpha.net/ORDO/Orphanet_1440
Tree view
Term history
phenome
group of disorders
Rare developmental defect during emb
Chromosomal anomaly
Autosomal anomaly
Autosomal monosomy
Ring chromosome
Ring chromosome 14
Rare genetic disease
Chromosomal anomaly
Autosomal anomaly
Graph view
Show siblings
Term info
OBO cross-reference:
E (exact mapping (the terms and the concepts are equivalent)) MeSH:C535487
E (exact mapping (the terms and the concepts are equivalent)) OMIM:616606
NTBT (narrower term maps to a broader term) ICD-10:Q93.2
Attributed ICD-10:Q93.2
E (exact mapping (the terms and the concepts are equivalent)) UMLS:C2930916
hasDbXref
MeSH:C535487, UMLS:C2930916,

Process

Step 3 › Result:

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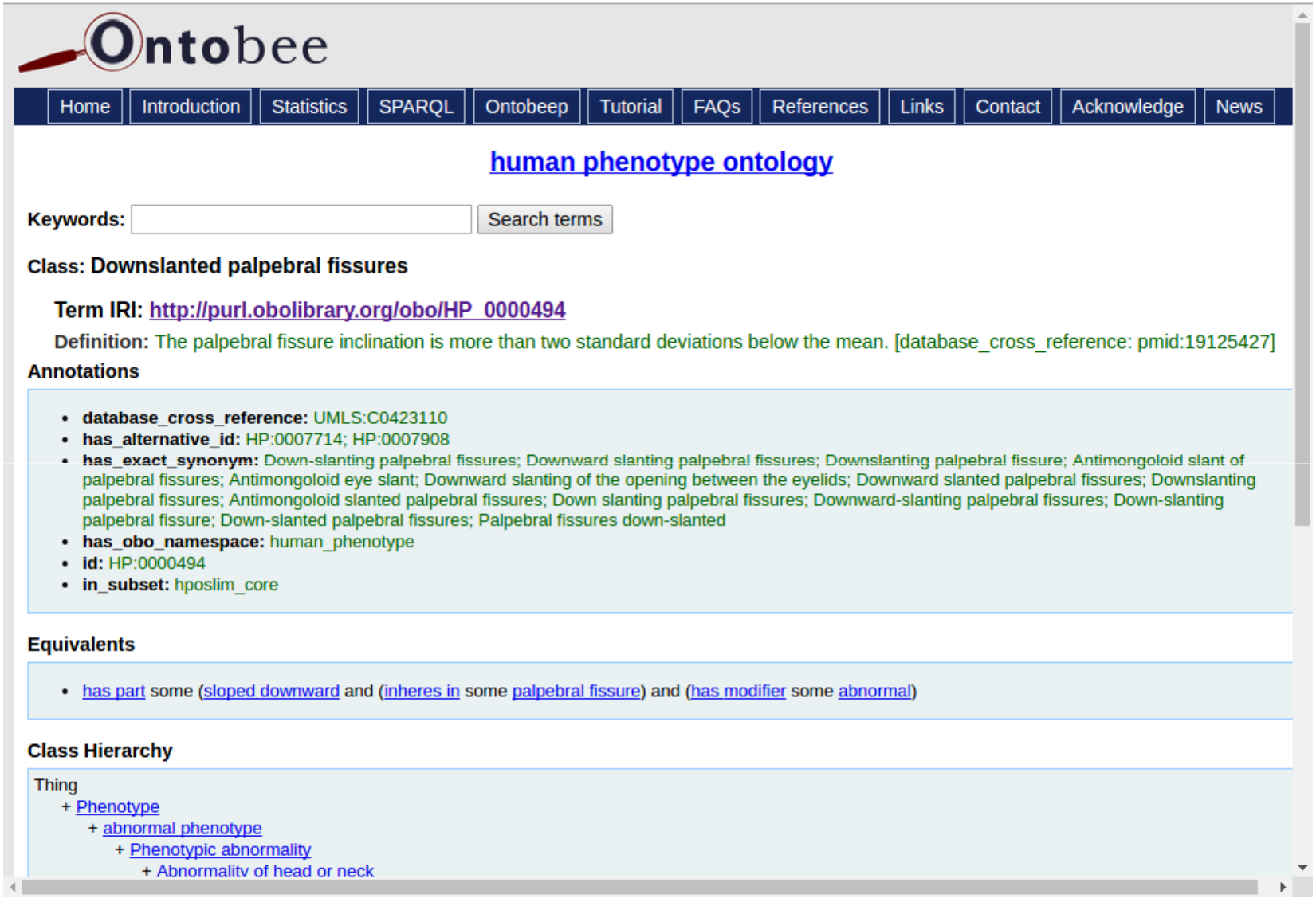
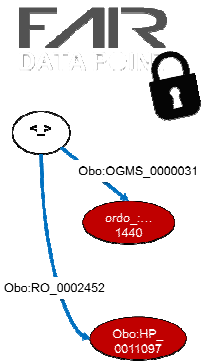
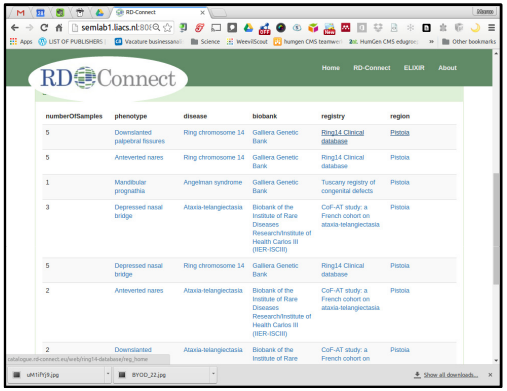
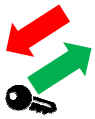
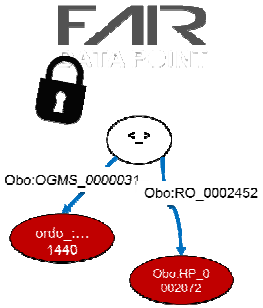

Ontobee
Home
Introduction
Statistics
SPARQL
Ontobeep
Tutorial
FAQs
References
Links
Contact
Acknowledge
News
human phenotype ontology
Keywords:
Search terms
Class: Downslanted palpebral fissures
Term IRI: http://purl.obolibrary.org/obo/HP_0000494
Definition: The palpebral fissure inclination is more than two standard deviations below the mean. [database_cross_reference: pmid:19125427]
Annotations
database_cross_reference: UMLS:C0423110
has_alternative_id: HP:0007714; HP:0007908
has_exact_synonym: Down-slanting palpebral fissures; Downward slanting palpebral fissures; Downslanting palpebral fissure; Antimongoloid slant of palpebral fissures; Antimongoloid eye slant; Downward slanting of the opening between the eyelids; Downward slanted palpebral fissures; Downslanting palpebral fissures; Antimongoloid slanted palpebral fissures; Down slanting palpebral fissures; Downward-slanting palpebral fissures; Down-slanting palpebral fissure; Down-slanted palpebral fissures; Palpebral fissures down-slanted
has_obo_namespace: human_phenotype
id: HP:0000494
in_subset: hposlim_core
Equivalents
has part some (sloped downward and (inheres in some palpebral fissure) and (has modifier some abnormal)
Class Hierarchy
Thing
+ Phenotype
+ abnormal phenotype
+ Phenotypic abnormality
+ Abnormality of head or neck

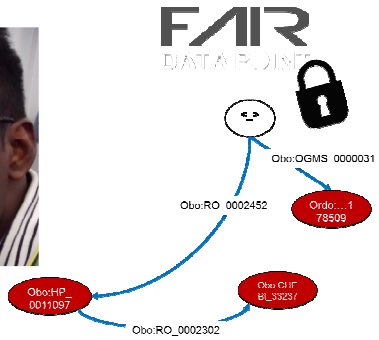
FAIR architecture (simplified)

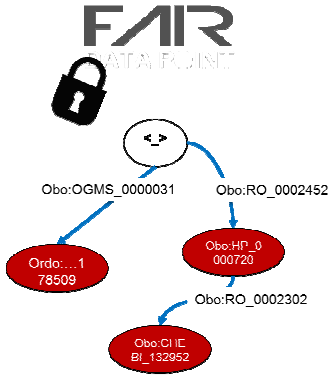






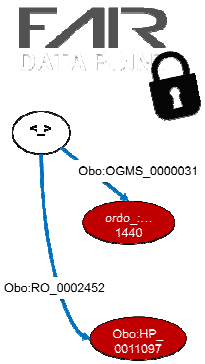


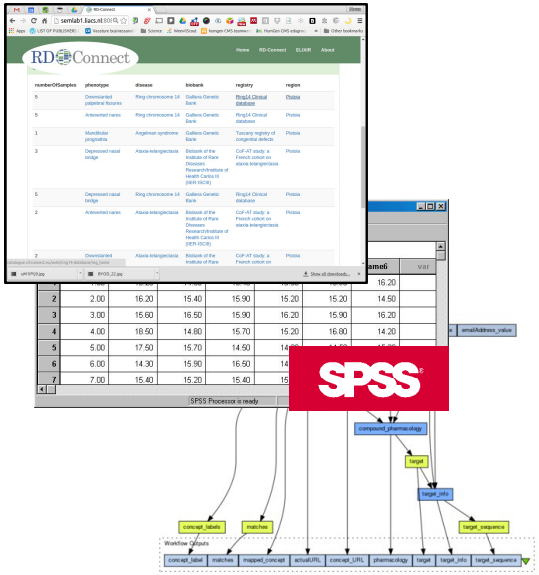


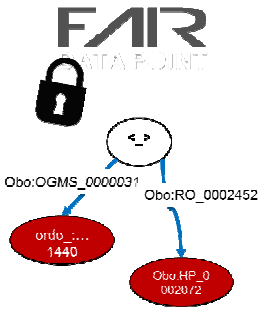


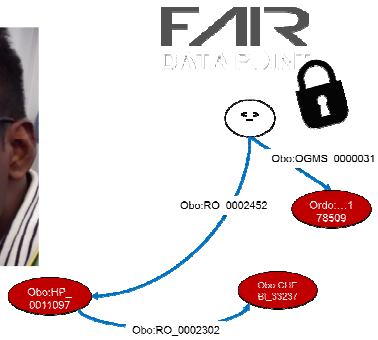
Locally controlled '**FAIR data points**'

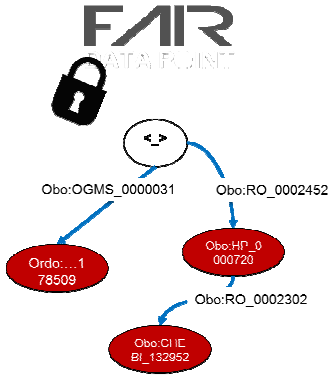
FAIR architecture (simplified)





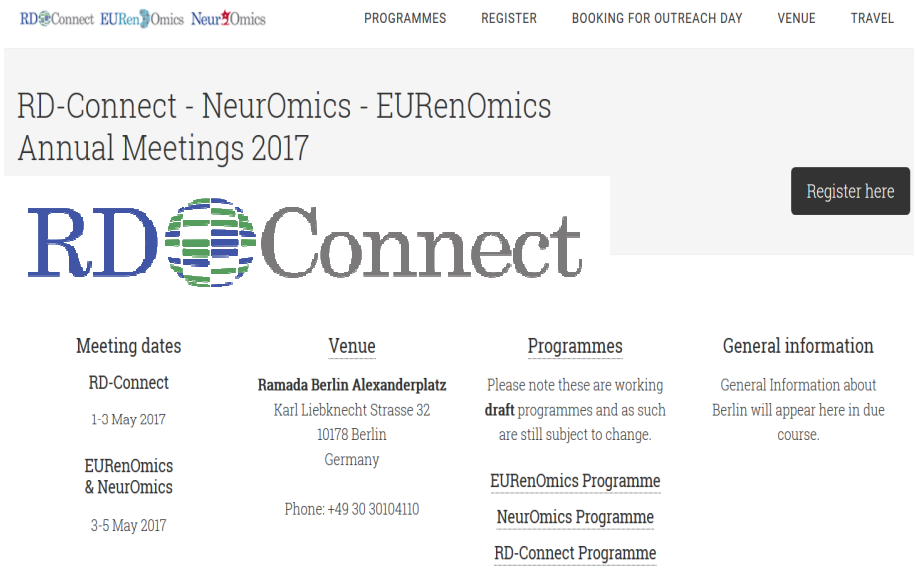






Locally controlled '**FAIR data points**'

More on FAIR for rare diseases…



RD-Connect annual meeting
http://rd-connect.eu/event/rd-connect-annual-meeting/

E-Rare Workshop on Data sharing and harmonization
http://www.erare.eu/events/e-rare-workshop-data-sharing-and-harmonization

To promote and support FAIR RD Registries

This edition dedicated to ERNs.

Are you interested?
Send an e-mail to:
rareregistries-school@iss.it

Who does what when making data FAIR?

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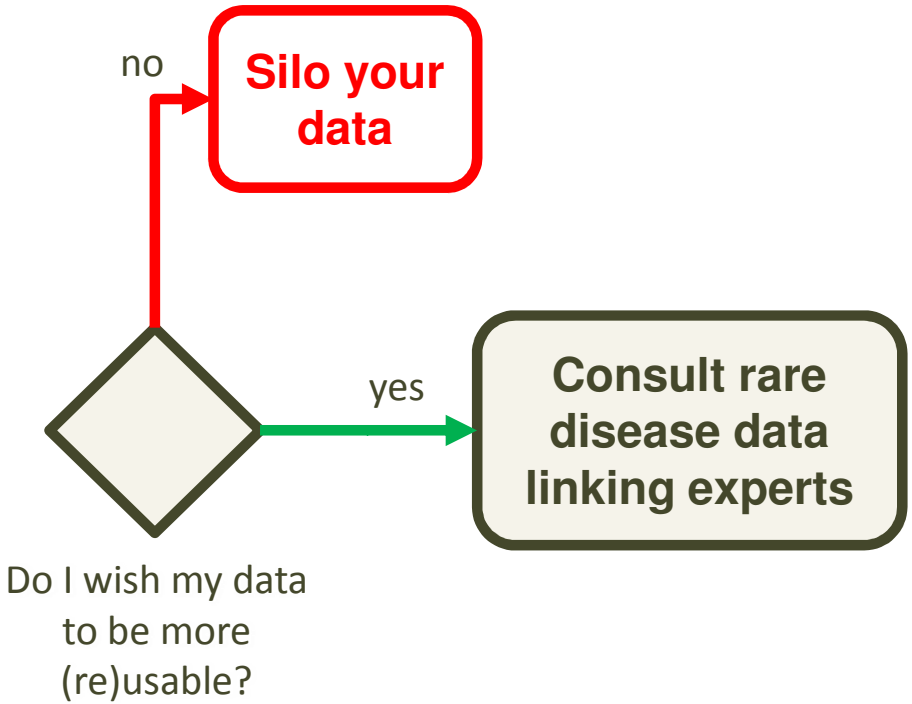
BBMRI Biobanking and Biomolecular Resources Research Infrastructure
RD Connect

Adopting FAIR principles



fair-rd-info@elixir-europe.org

Claudio Carta, Liaison/consultant FAIR training, BYOD workshops, FAIRification in RD domain

Mark Thompson, FAIR software architect, link to FAIR engineering team

David van Enckevort, technical lead RD data linkage plan, link to RD software engineers

Mascha Jansen, Coordinator FAIR data projects & BYODs for DTL/ELIXIR-NL

Annika Jacobsen, ‘Roving’ FAIR data expert for rare diseases

Marco Roos, ELIXIR infrastructure for rare diseases, project lead RD data linkage plan

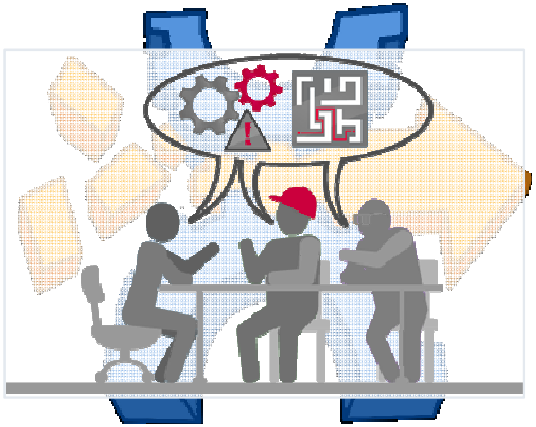
Rare disease data linkage plan

FAIR principles applied to rare disease data and software

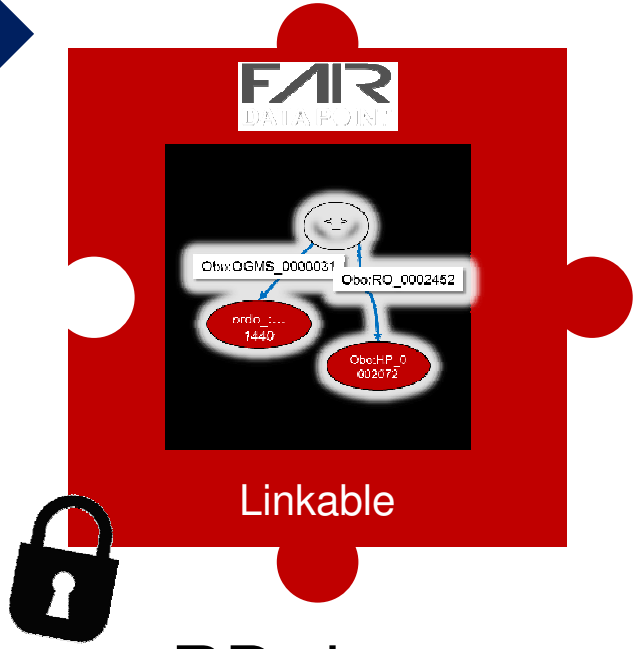
RD DATA

RD data custodian

Knowledge exchange

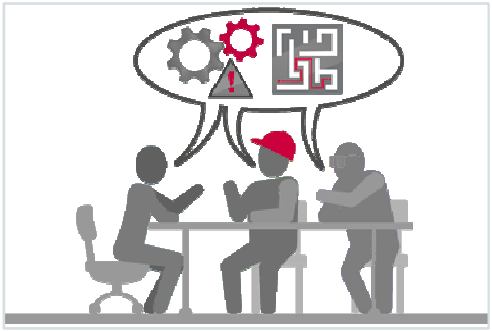


Domain experts & FAIR data stewards together



RD data custodian

FAIRification steps



Annika and Andra
'roving' FAIR data experts
for the rare disease case

1. Define at least one 'driving user question'
   BYOD: main goal; FAIR data project: minimum test
2. Disambiguate data with global codes
   Apply 'Linked Data' to make machine-readable
3. Make accessible via a FAIR data point
   Add license + standard terms about data set
4. Test: answer driving user question

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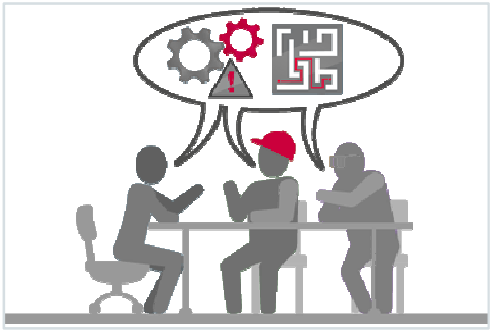
FAIRification steps: who does what?



Annika and Andra
FAIR data experts
for the rare disease case

1. Define at least one ‘driving user question’
   FAIR data steward + case expert(s)
2. Disambiguate data with global codes
   FAIR data expert (+ local trainee – preferred!)
3. Make dataset accessible via a FAIR data point
   FAIR data expert + case expert
   FAIR software developers + local IT expert(s)
4. Answer driving user question
   Case expert + FAIR data expert

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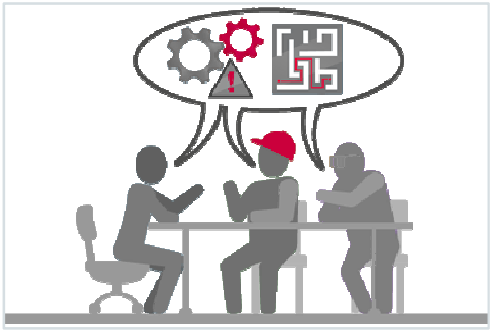
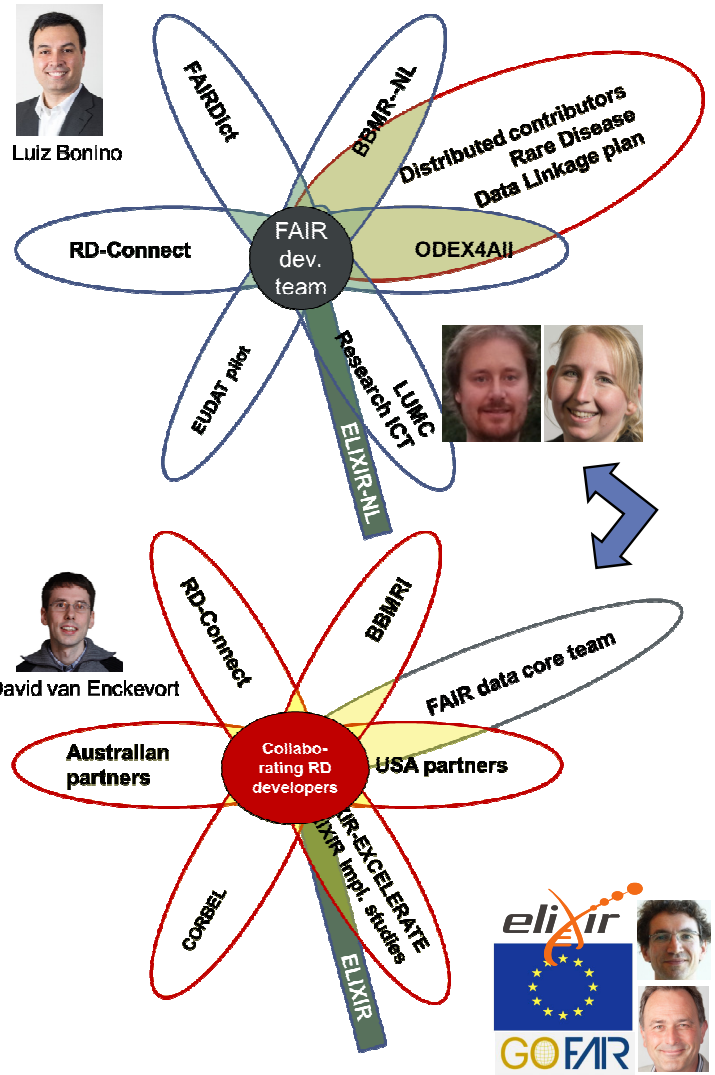
Example: NMD experts in FAIR NMD case



NMD experts contribute to

1. Defining at least one ‘driving user question’
2. Disambiguating data with global codes
3. Testing FAIR features of RDRF (registry software)
4. Describing NMD data **set**s for a FAIR data point
5. Testing NMD FAIR data point(s)
6. Optional: deep data analytics with FAIR data

FAIR data teams that we linked to rare disease case



FAIR Data engineering team
Luiz Bonino
Rajaram Kaliyaperumal
Kees Burger
Nuno Nunes
Shamanou van Leeuwen
Mark Thompson

DTL FAIR DATA ENGINEER TEAM

Skunk team / FAIR metrics
Mark Wilkinson
Michel Dumontier

Collaborating RD developers
David van Enckevort, Annika Jacobsen (LUMC), Andra Waagmeester; Heimo Muller, Robert Reihs (Graz, Austria); Pedro Sernadella, Jose Oliveira (Aveiro, Portugal); Marc Hanauer, Ana Rath (Orphanet, Paris); Roxana Merino (Karolinska, Sweden); Matthias Brochausen (USA); Developers of Castor, RDRF, OSSE, MolGenis;

Liaisons/case owners: Rachel Thompson, Libby Wood, Claudio Carta (Rome, Italy), Domenica Taruscio (Rome, Italy), Marco Crimi, Estrella Gomes, Marina Mordenti, Freddie Ehrhart

Elixir: Team Evelo (Maastricht), Team Goble (Manchester), Team Gut/Beltran (CNAG), Team Parkinson (EBI), Team Poch (Strassbourg);

GoFAIR: Barend Mons; **DTL**: Mascha Jansen, Celia van Gelder, Erik Schultes, Albert Mons

Patients and patient organisations

Starting point: Bring Your Own Data workshop

(& annual summer school on rare disease registries)

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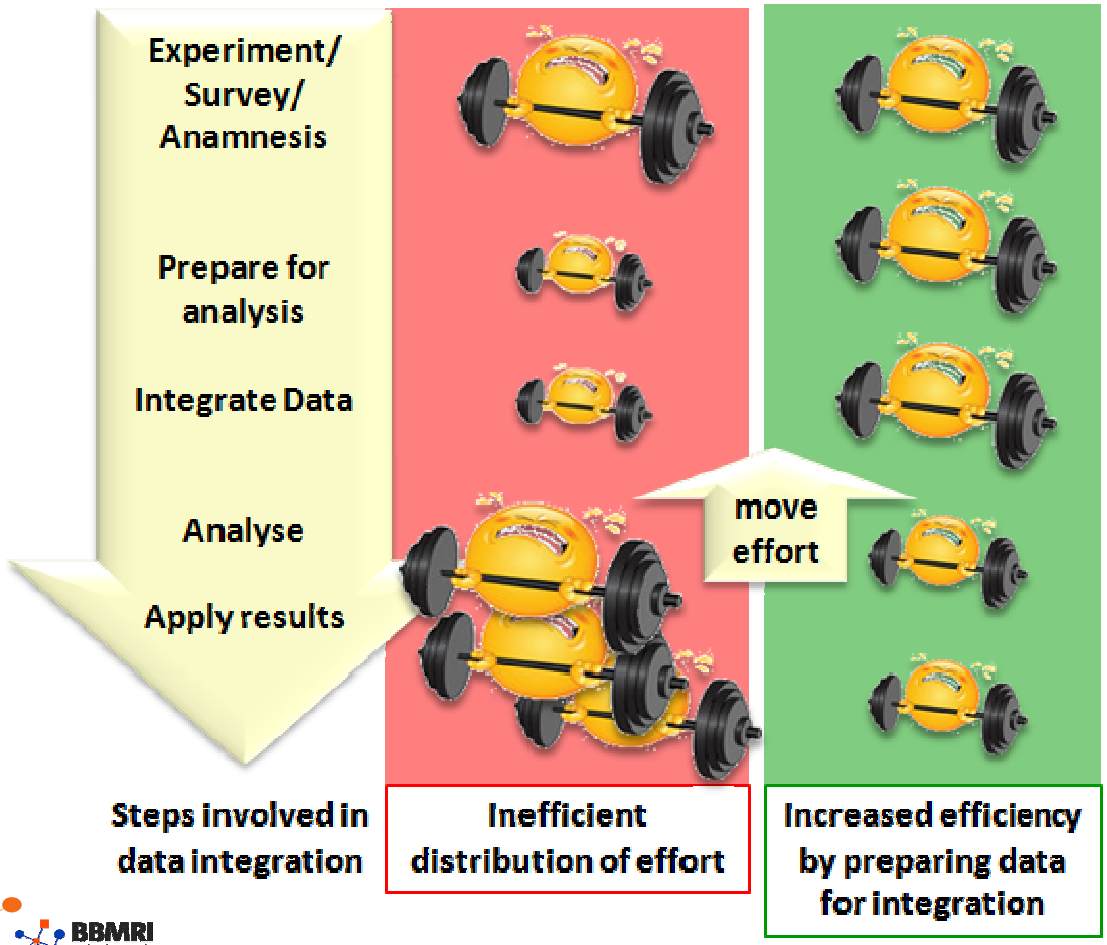
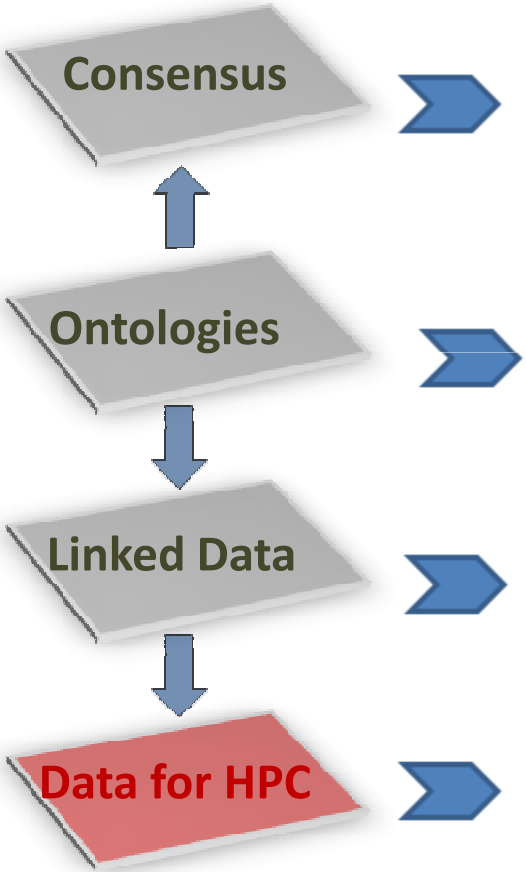
BYOD workshops and “FAIRification" protocols



BYOD workshops and “FAIRification" protocols



Harmonize what is measured and how

Harmonize classification and relations (meaning)

Harmonize how we make it computable

Transform for HPC

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Bring Your Own Data workshops

General Roadmap:

Preparation

(i) Attendees Subscripition
(ii) Technical organization
(iii) Attendees selction
(iv) Analysis of the items of the datasets provided by participants
(v) Technical video conference among trainers
(vi) Tentative output data
(vii) Data hosting software
(viii) Shared documents among trainers and attendees
(ix) Video conference Trainers and Attendees

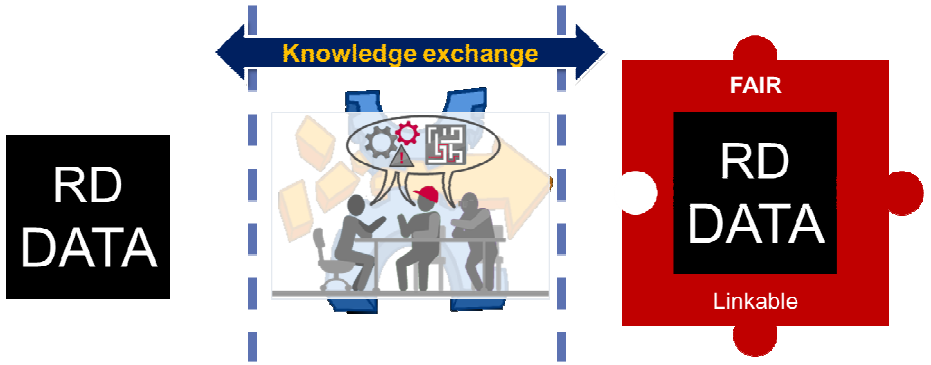
Bring Your Own Data workshops

General Roadmap: Preparation → Execution → Follow up

Execution

(i) Introduction: ontology, data linkage , FAIR data
(ii) Use case Introductions
(iii) Driving questions, cross resources
(iv) Workgroups division
(v) Answer driving questions cross resources
(vi) Working sessions
(vii) Final report from each group:
- Use case presentations
- Self–sketching the "FAIRification" workflow

(viii) Evaluation



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Bring Your Own Data workshops

General Roadmap:

(i) Report difficulties
(ii) Clarifications with trainers

Survey after a BYOD

The aspects that attendees appreciated most of the BYOD were:

- the possibility to work in a friendly environment
-- to exchange information with other registries
-- widen one's research horizons
-- learn a new methodology that can be useful for own work.

To promote and support FAIR RD Registries

Co-organized events, held in ISS (Rome, Italy) to promote and support the establishment of registries FAIR for humans and computers are:

> International Summer School on Rare Disease and Orphan Drug Registries, since 2013

> "Specific" Bring Your Own Data workshops to link RD-Registries, since 2014

Are there **biosamples** of donors with **phenotype** "K" and **mutations** in gene ""ZZ""?

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To promote and support FAIR RD Registries

Open to health professionals, researchers, medical specialists, registry curators, database managers and representatives of patients associations
who are involved in or intend to establish a rare disease registry, mainly inside a European Reference Network.

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To promote and support FAIR RD Registries

This edition dedicated to ERNs.

We should have:

- **a registry curator** (clinician)
- **a data manager** or IT-expert
- **An ePAG representative,** patient, or representative

Are you interested?
Send an e-mail to:
rareregistries-school@iss.it

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How does FAIR relate to recommended ontologies ORDO and HPO?

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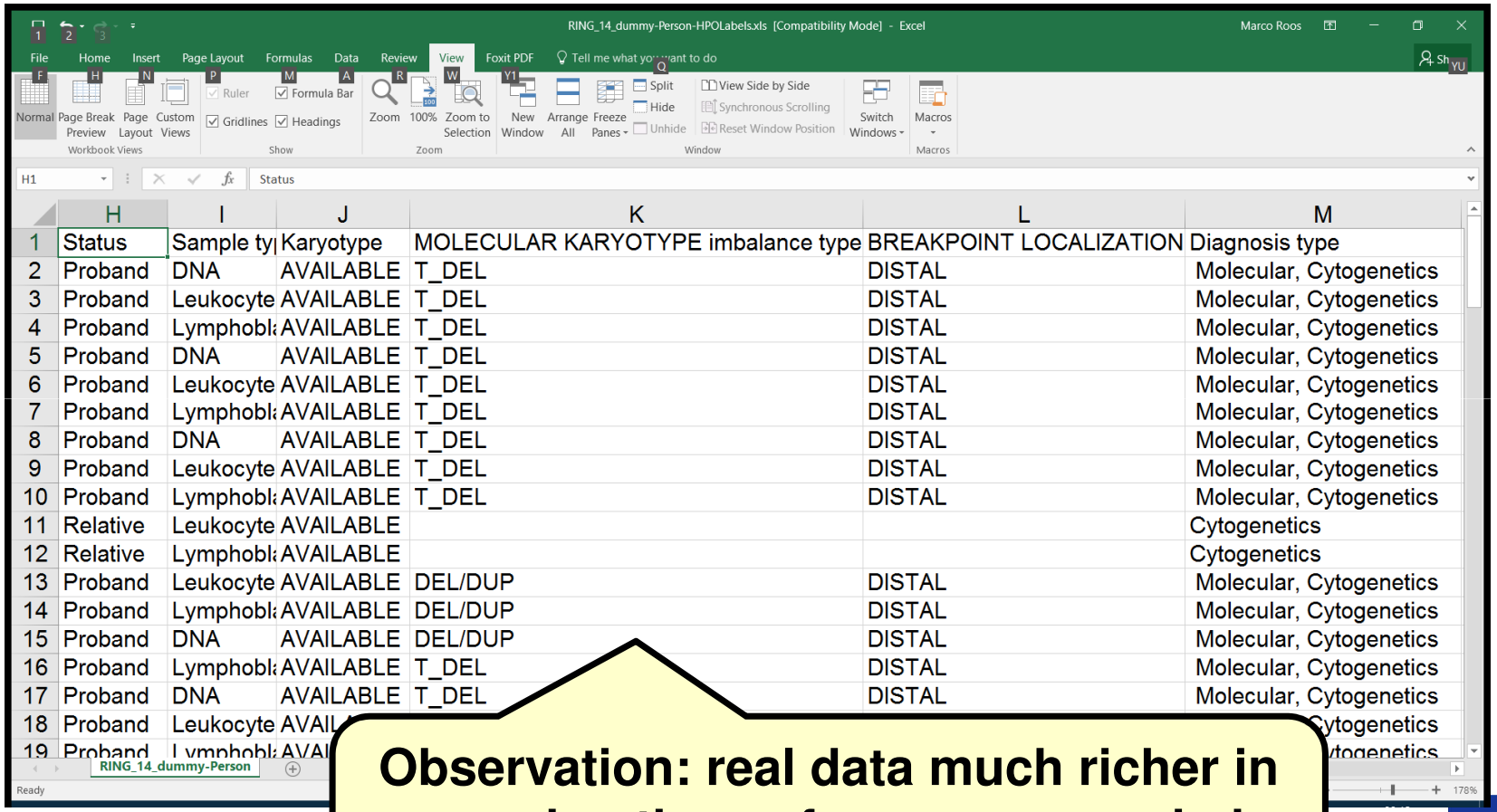
Real data



RING_14_dummy-Person-HPOLabels.xls [Compatibility Mode] - Excel

Marco Roos

File Home Insert Page Layout Formulas Data Review View Foxit PDF Tell me what you want to do

Normal Page Break Preview Page Layout Custom Views | Ruler Gridlines Formula Bar Headings | Zoom 100% Zoom to Selection | New Window Arrange All Freeze Panes Split Hide Unhide View Side by Side Synchronous Scrolling Reset Window Position | Switch Windows | Macros

Workbook Views Show Zoom Window Macros

H1 Status

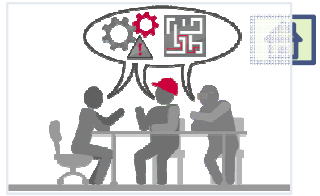
	H	I	J	K	L	M
1	Status	Sample ty	Karyotype	MOLECULAR KARYOTYPE imbalance type	BREAKPOINT LOCALIZATION	Diagnosis type
2	Proband	DNA	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
3	Proband	Leukocyte	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
4	Proband	Lymphobl	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
5	Proband	DNA	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
6	Proband	Leukocyte	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
7	Proband	Lymphobl	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
8	Proband	DNA	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
9	Proband	Leukocyte	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
10	Proband	Lymphobl	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
11	Relative	Leukocyte	AVAILABLE			Cytogenetics
12	Relative	Lymphobl	AVAILABLE			Cytogenetics
13	Proband	Leukocyte	AVAILABLE	DEL/DUP	DISTAL	Molecular, Cytogenetics
14	Proband	Lymphobl	AVAILABLE	DEL/DUP	DISTAL	Molecular, Cytogenetics
15	Proband	DNA	AVAILABLE	DEL/DUP	DISTAL	Molecular, Cytogenetics
16	Proband	Lymphobl	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
17	Proband	DNA	AVAILABLE	T_DEL	DISTAL	Molecular, Cytogenetics
18	Proband	Leukocyte	AVAIL			ytogenetics
19	Proband	Lymphobl	AVAI			ytogenetics

RING_14_dummy-Person

Ready 178%

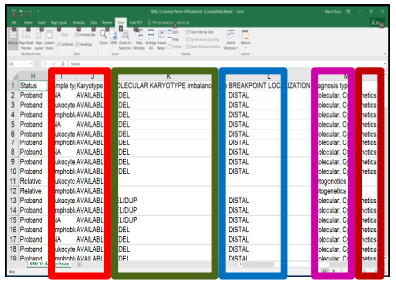
Observation: real data much richer in meaning than a few recommended ontologies / coding systems

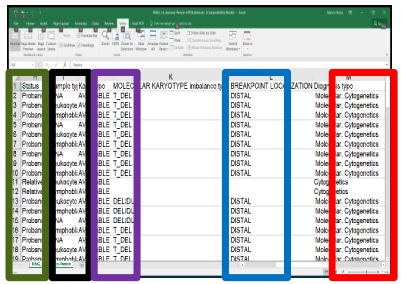
Making data linkable

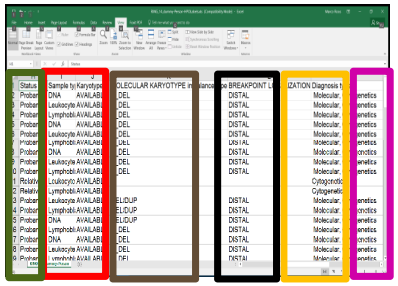


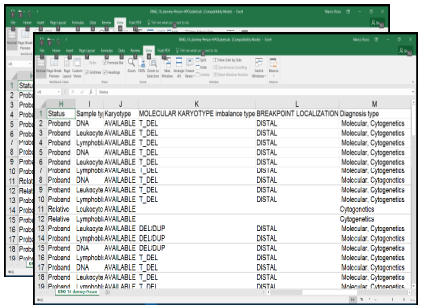
Result:

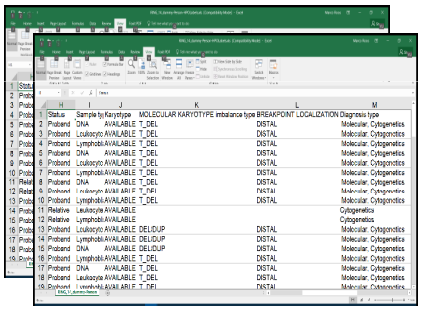
- **Most** parts interoperable
- Sources can serve **large** number of use cases (including unforeseen)
- **Relations** coded
- Probably need help

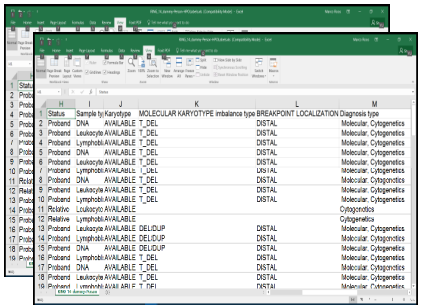












Driving user questions

Driving user questions

Driving user questions

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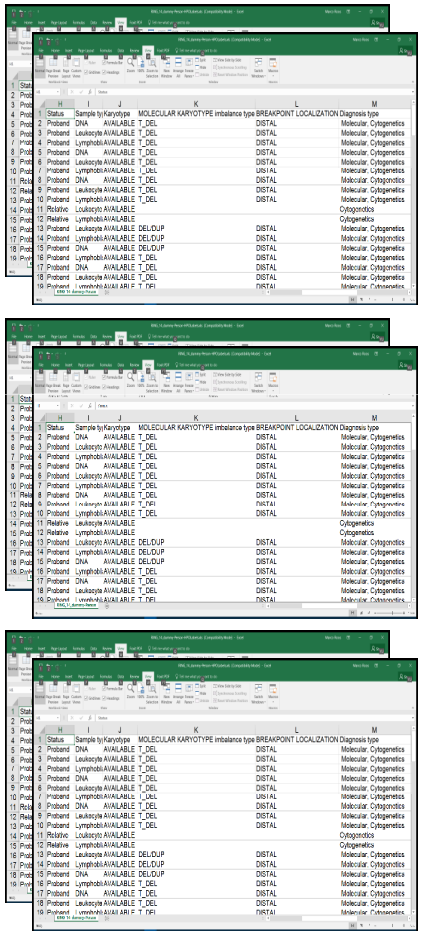
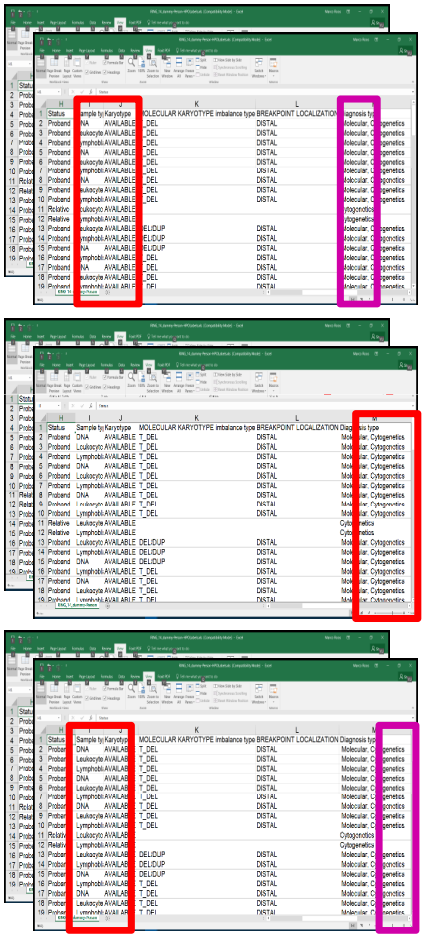
Recode to standard codes (e.g. HPO, ORDO)

Expert-certified ontology recommendations

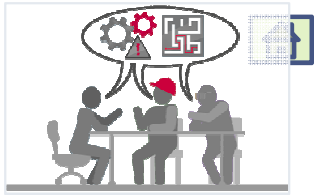




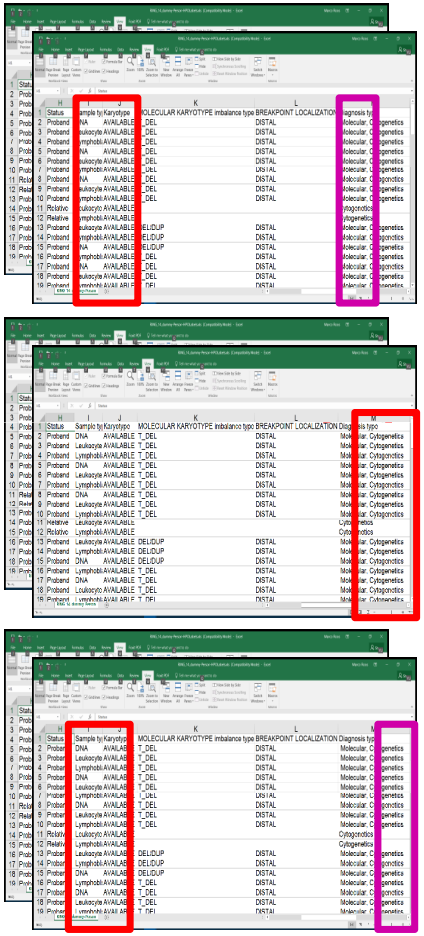
Result:

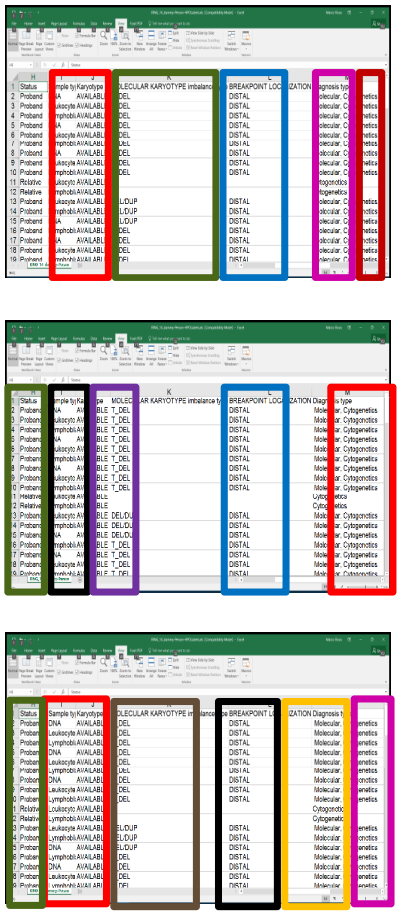
- **Some** parts interoperable
- **Many** sources can comply with **limited** number of use cases
- **Relations** usually not coded
- Probably doable without much help

Adopting FAIR principles



Expert-certified ontology recommendations





Driving user questions

Driving user questions

Driving user questions

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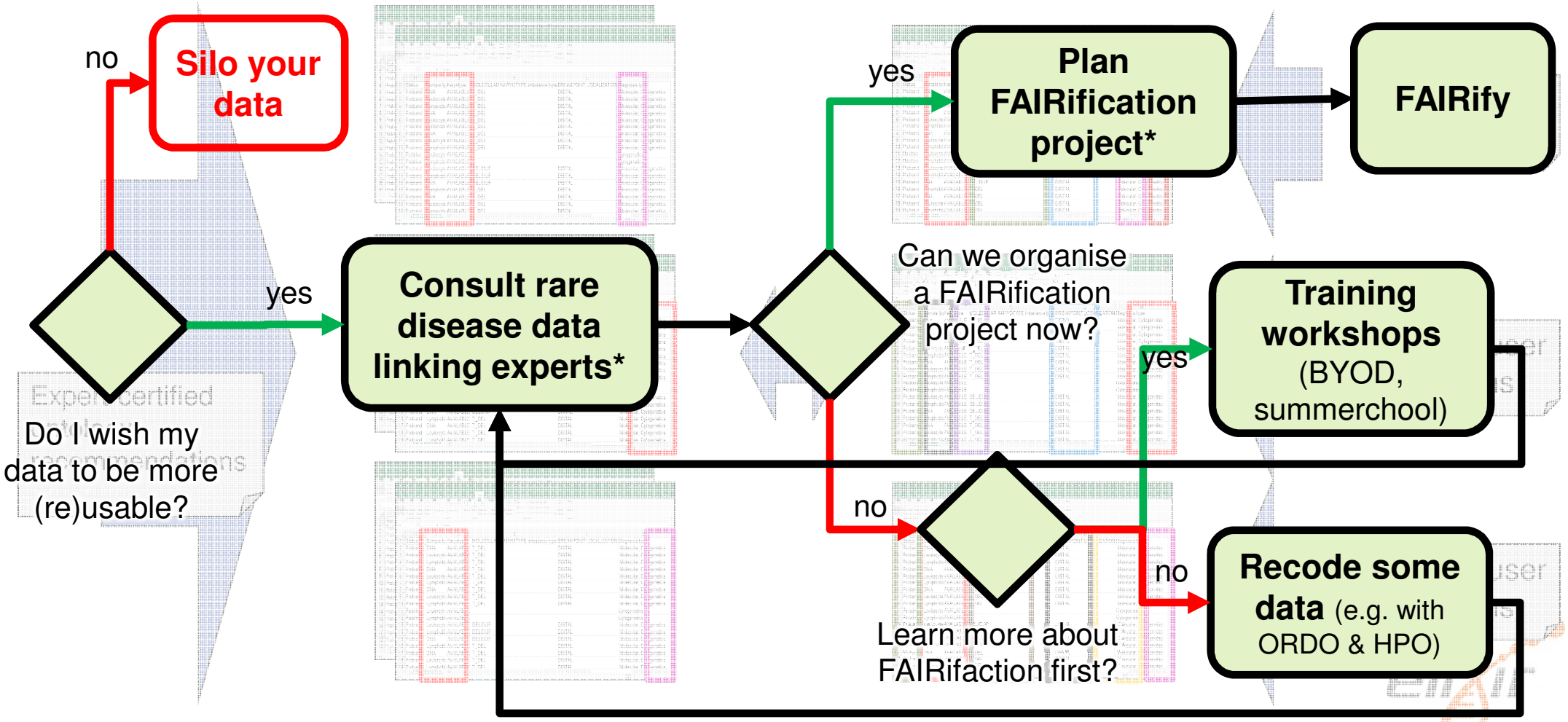
Adopting FAIR principles



* Contact: fair-rd-info@elixir-europe.org or Mascha.Jansen@dtls.nl

Starting point: Bring Your Own Data workshop

- Hands-on learning experience to promote and support the establishment of FAIR resources.
- Introduce "FAIRification" protocols.
- Demonstrate how easy it becomes to answer difficult queries across distributed resources.
- Provide a mechanism to data owners who would like to add value to their data by preparing them for data integration and computational analysis, but are unfamiliar with basic techniques to make data FAIR.

We define common data elements, why should I need FAIR for interoperability?

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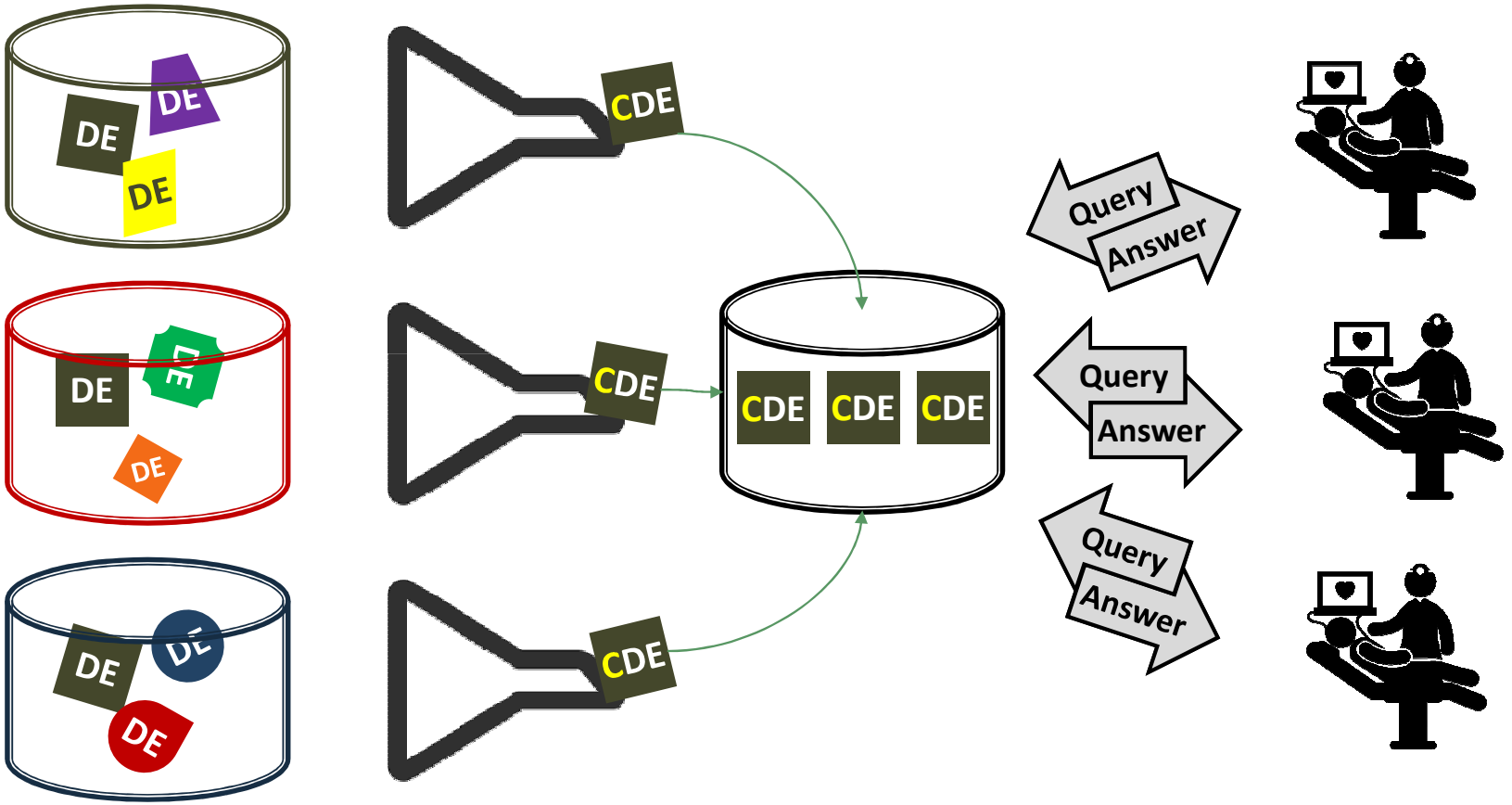
Central store of minimum/common data elements



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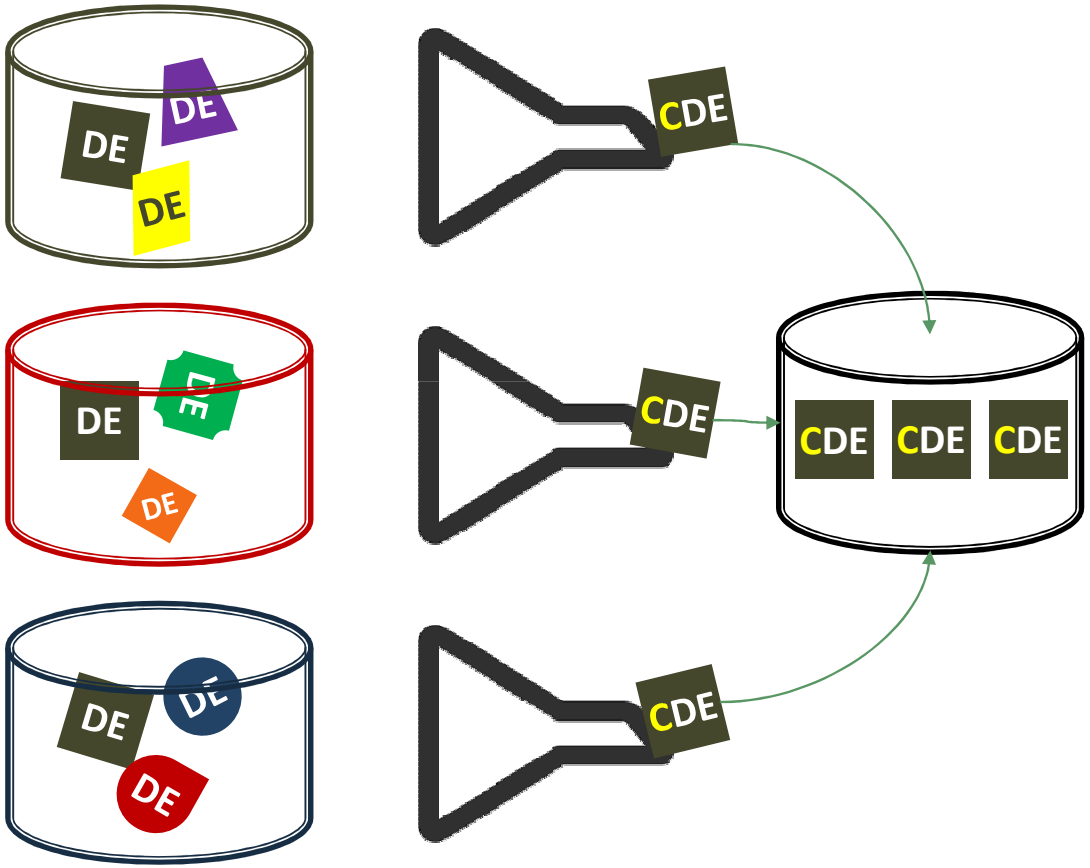
Central store of minimum/common data elements



Pros

- **More** of the same (e.g. for counting)
- Familiar technology

Cons

- More of **the same**
- **Limited** by legal constraints (ELSI)

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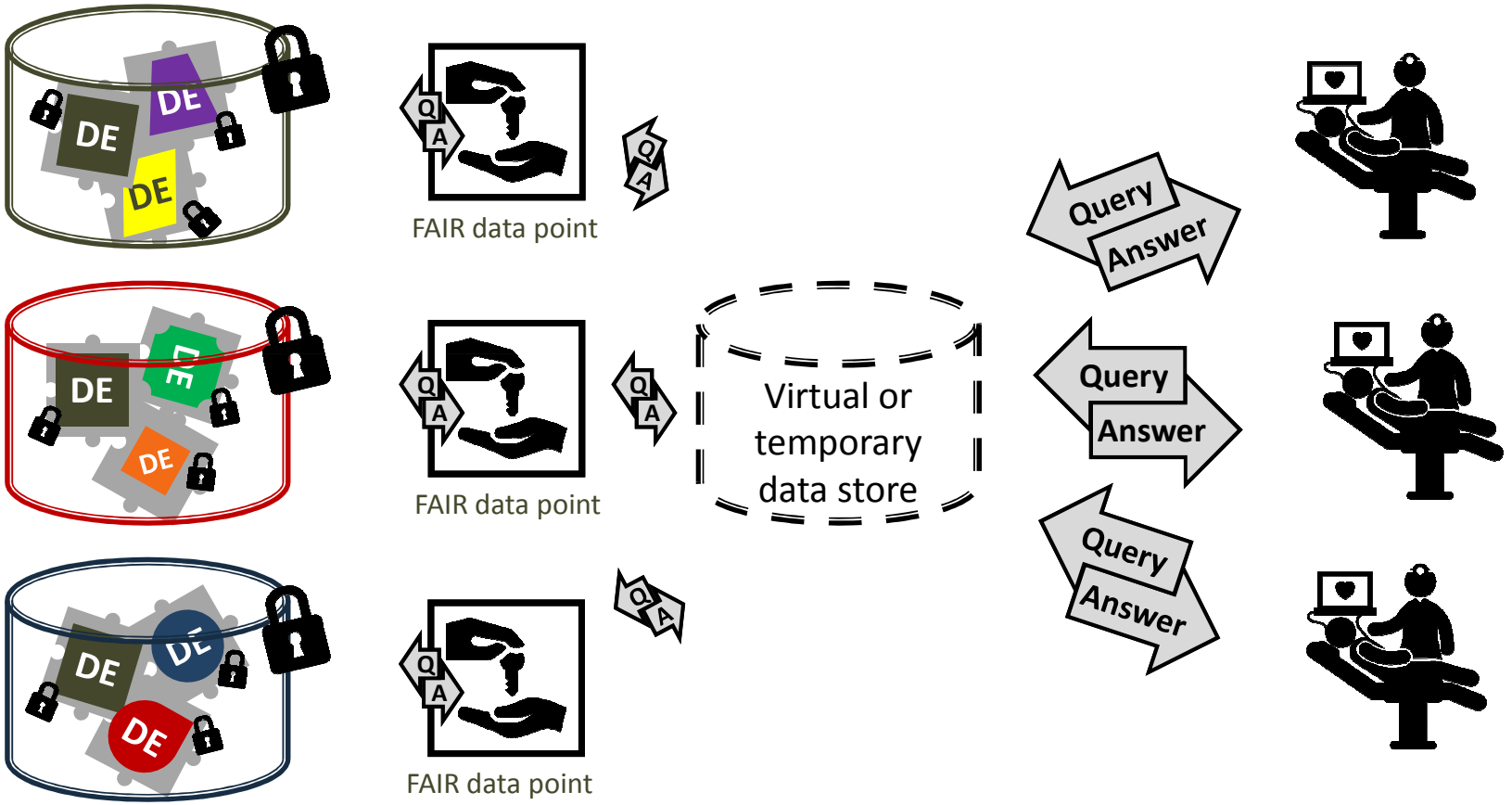
Findable, Accessible, Interoperable, Reusable

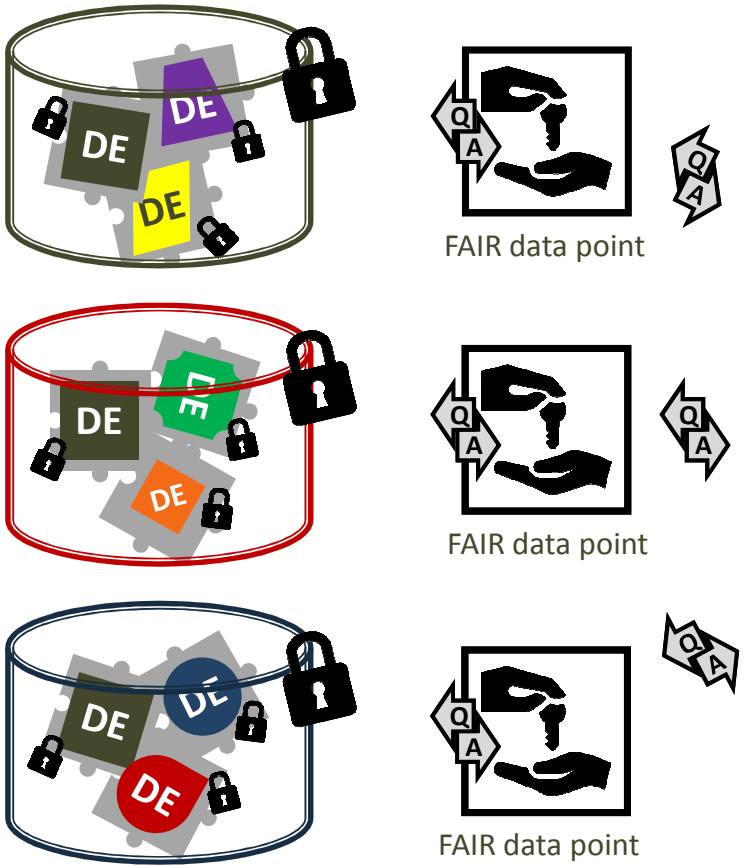
at the source



RD-Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Findable, Accessible, Interoperable, Reusable

at the source



Pro

- **More** of **more**
- Mitigates ELSI
- More useable *at the source*

Cons

- More complex
- + responsibility *at the source*
- Newer, partly under development

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FAIRification

OpenRefine-FAIRifier, Ontology Lookup Service, BioSharing.org

Tech alert!!

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FAIRification options

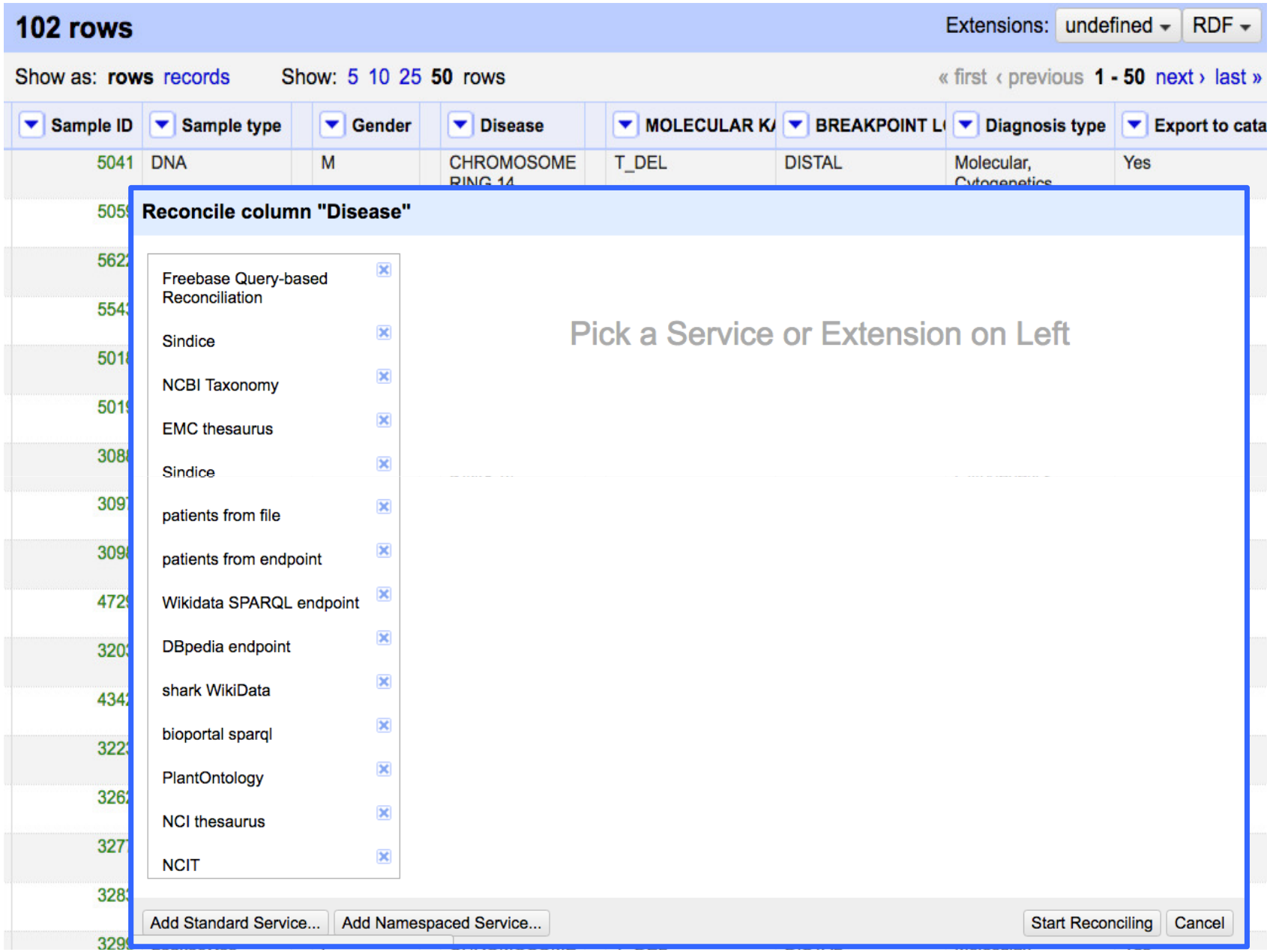
- OpenRefine-FAIRifier: general purpose FAIRifier tool
- <tool-X>+FAIR: tool with FAIRification built-in

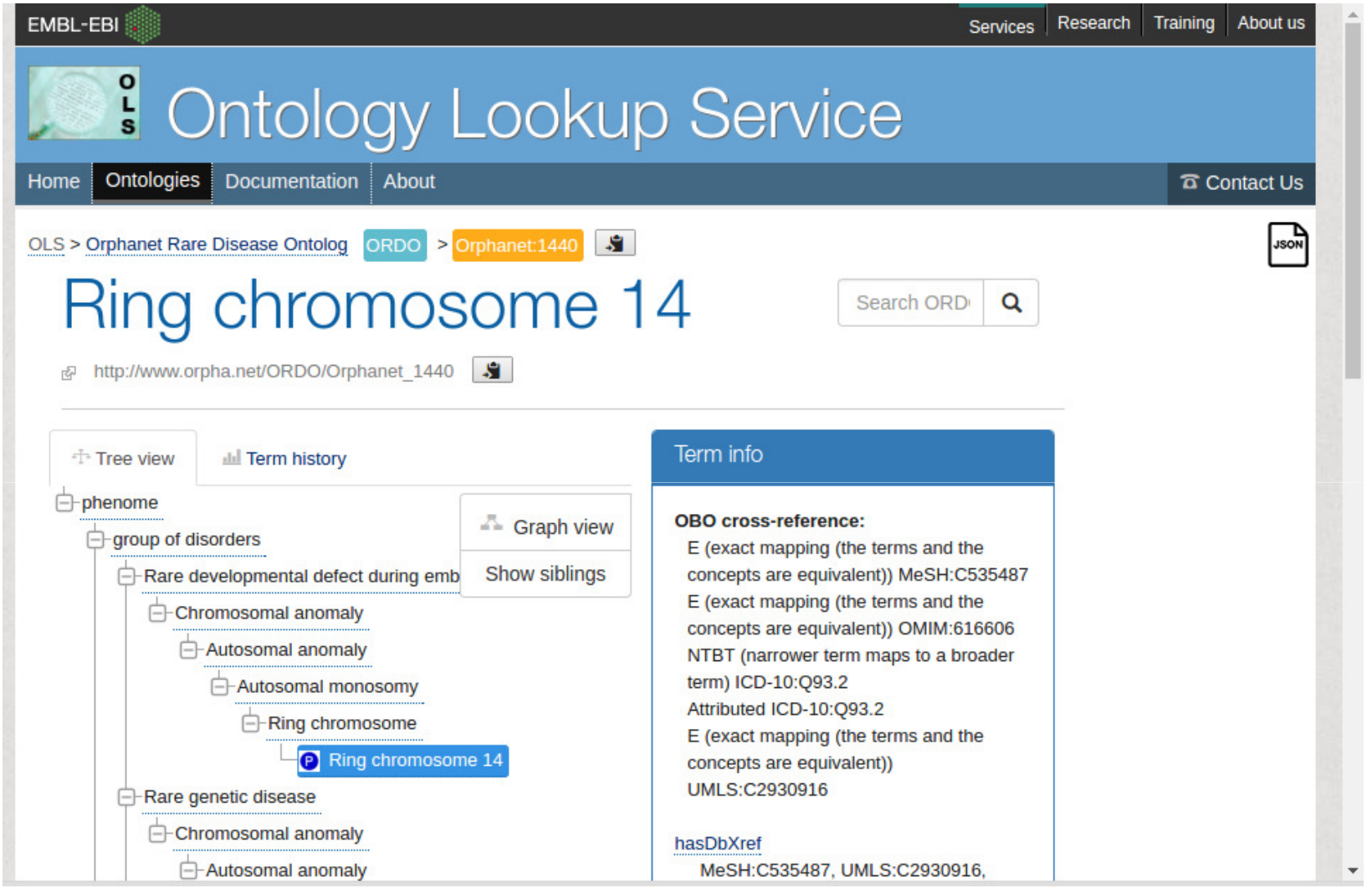
  (work in progress for MolGenis, ID-cards and selected registry tools)

102 rows Extensions: undefined RDF

Show as: **rows** records Show: 5 10 25 **50** rows « first ‹ previous **1 - 50** next › last »

Sample ID	Sample type	Gender	Disease	MOLECULAR K/	BREAKPOINT L(	Diagnosis type	Export to cata
5041	DNA	M	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
5059	Leukocytes	M	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
5622	Lymphoblast	M	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
5543	DNA	M edit	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
5018	Leukocytes	M	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
5019	Lymphoblast	M	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
3088	DNA	F	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
3097	Leukocytes	F	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
3098	Lymphoblast	F	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
4729	Leukocytes	F	CHROMOSOME RING 14			Cytogenetics	Yes
3203	Lymphoblast	F	CHROMOSOME RING 14			Cytogenetics	Yes
4342	Leukocytes	M	CHROMOSOME RING 14	DEL/DUP	DISTAL	Molecular, Cytogenetics	Yes
3223	Lymphoblast	M	CHROMOSOME RING 14	DEL/DUP	DISTAL	Molecular, Cytogenetics	Yes
3262	DNA	M	CHROMOSOME RING 14	DEL/DUP	DISTAL	Molecular, Cytogenetics	Yes
3277	Lymphoblast	F	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
3283	DNA	F	CHROMOSOME RING 14	T_DEL	DISTAL	Molecular, Cytogenetics	Yes
3299	Leukocytes	F	CHROMOSOME	T_DEL	DISTAL	Molecular,	Yes


102 rows
Extensions: undefined RDF
Show as: rows records
Show: 5 10 25 50 rows
« first ‹ previous 1 - 50 next › last »
Sample ID
Sample type
Gender
Disease
MOLECULAR K
BREAKPOINT L
Diagnosis type
Export to cata
5041
DNA
M
CHROMOSOME
T_DEL
DISTAL
Molecular,
Yes
Reconcile column "Disease"
Freebase Query-based Reconciliation
Sindice
NCBI Taxonomy
EMC thesaurus
Sindice
patients from file
patients from endpoint
Wikidata SPARQL endpoint
DBpedia endpoint
shark WikiData
bioportal sparql
PlantOntology
NCI thesaurus
NCIT
Pick a Service or Extension on Left
Add Standard Service...
Add Namespaced Service...
Start Reconciling
Cancel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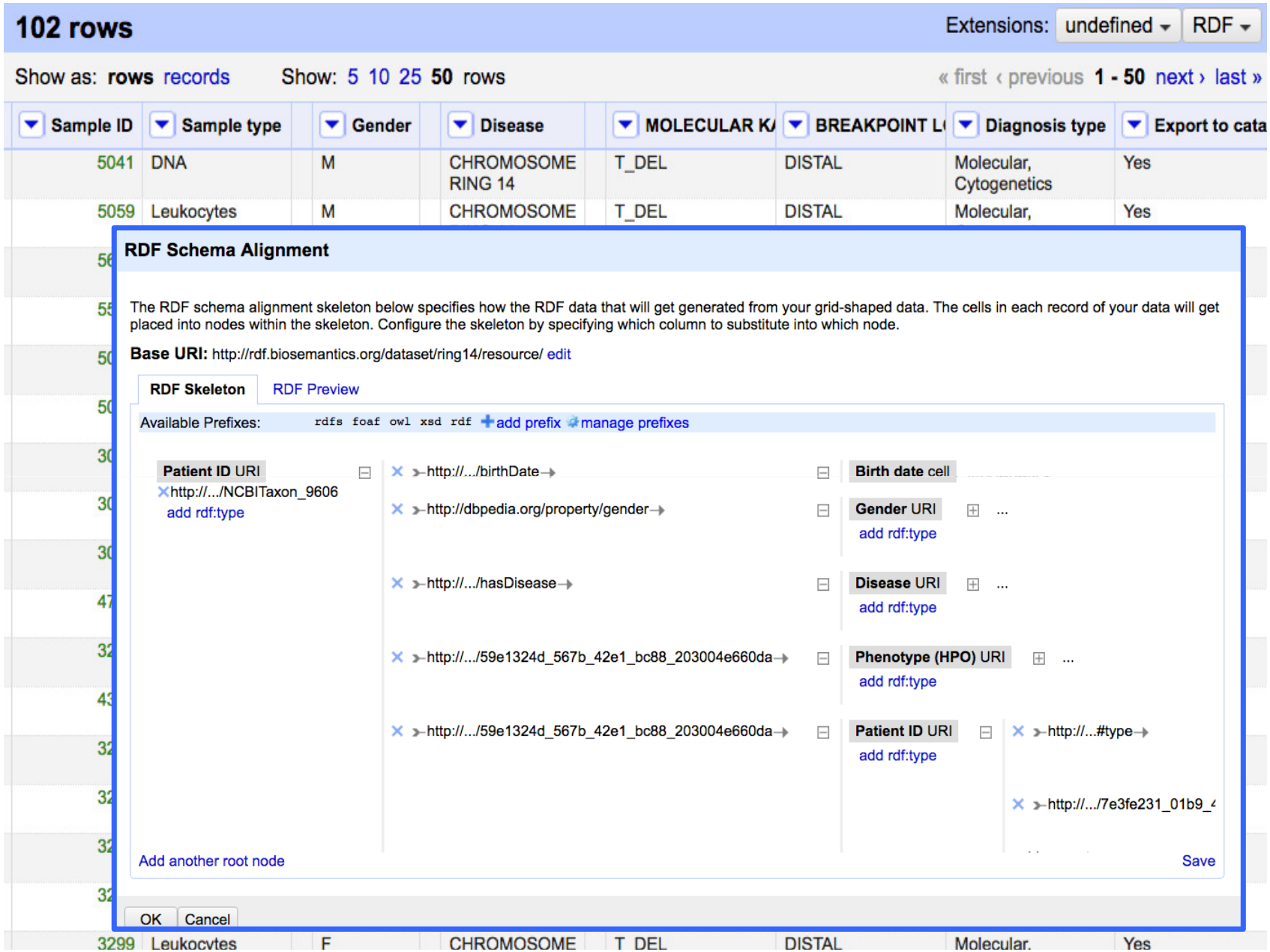
ORDO
> Orphanet:1440
JSON
Ring chromosome 14
Search ORD
http://www.orpha.net/ORDO/Orphanet_1440
Tree view
Term history
phenome
group of disorders
Rare developmental defect during emb
Chromosomal anomaly
Autosomal anomaly
Autosomal monosomy
Ring chromosome
Ring chromosome 14
Rare genetic disease
Chromosomal anomaly
Autosomal anomaly
Graph view
Show siblings
Term info
OBO cross-reference:
E (exact mapping (the terms and the concepts are equivalent)) MeSH:C535487
E (exact mapping (the terms and the concepts are equivalent)) OMIM:616606
NTBT (narrower term maps to a broader term) ICD-10:Q93.2
Attributed ICD-10:Q93.2
E (exact mapping (the terms and the concepts are equivalent)) UMLS:C2930916
hasDbXref
MeSH:C535487, UMLS:C2930916,


102 rows
Extensions: undefined RDF
Show as: rows records
Show: 5 10 25 50 rows
« first ‹ previous 1 - 50 next › last »
Sample ID
Sample type
Gender
Disease
MOLECULAR K
BREAKPOINT L
Diagnosis type
Export to cata
5041 DNA M CHROMOSOME RING 14 T_DEL DISTAL Molecular, Cytogenetics Yes
5059 Leukocytes M CHROMOSOME T_DEL DISTAL Molecular, Yes
RDF Schema Alignment
The RDF schema alignment skeleton below specifies how the RDF data that will get generated from your grid-shaped data. The cells in each record of your data will get placed into nodes within the skeleton. Configure the skeleton by specifying which column to substitute into which node.
Base URI: http://rdf.biosemantics.org/dataset/ring14/resource/ edit
RDF Skeleton
RDF Preview
Available Prefixes: rdfs foaf owl xsd rdf add prefix manage prefixes
Patient ID URI
http://.../NCBITaxon_9606
add rdf:type
http://.../birthDate
Birth date cell
http://dbpedia.org/property/gender
Gender URI
add rdf:type
http://.../hasDisease
Disease URI
add rdf:type
http://.../59e1324d_567b_42e1_bc88_203004e660da
Phenotype (HPO) URI
add rdf:type
http://.../59e1324d_567b_42e1_bc88_203004e660da
Patient ID URI
add rdf:type
http://...#type
http://.../7e3fe231_01b9_
Add another root node
Save
OK
Cancel
3299 Leukocytes F CHROMOSOME T_DEL DISTAL Molecular, Yes

102 rows Extensions: undefined | RDF

Show as: **rows** records Show: 5 10 25 **50** rows « first ‹ previous **1 - 50** next › las

Sample ID	Sample type	Gender	Disease	MOLECULAR KARYOTYPE
5041	DNA	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
5059	Leukocytes	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
5622	Lymphoblast	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
5543	DNA	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
5018	Leukocytes	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
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3088	DNA	http://dbpedia.org/resource/Female	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
3097	Leukocytes	http://dbpedia.org/resource/Female	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
3098	Lymphoblast	http://dbpedia.org/resource/Female	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/5998bfd4_721c_45c2_8
4729	Leukocytes	http://dbpedia.org/resource/Female	http://www.orpha.net/ORDO/Orphanet_1440	
3203	Lymphoblast	http://dbpedia.org/resource/Female	http://www.orpha.net/ORDO/Orphanet_1440	
4342	Leukocytes	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/de2bc1a8_3cdf_4086_9
3223	Lymphoblast	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/de2bc1a8_3cdf_4086_9
3262	DNA	http://dbpedia.org/resource/Male	http://www.orpha.net/ORDO/Orphanet_1440	http://rdf.biosemantics.org/ontolo connect/de2bc1a8_3cdf_4086_9
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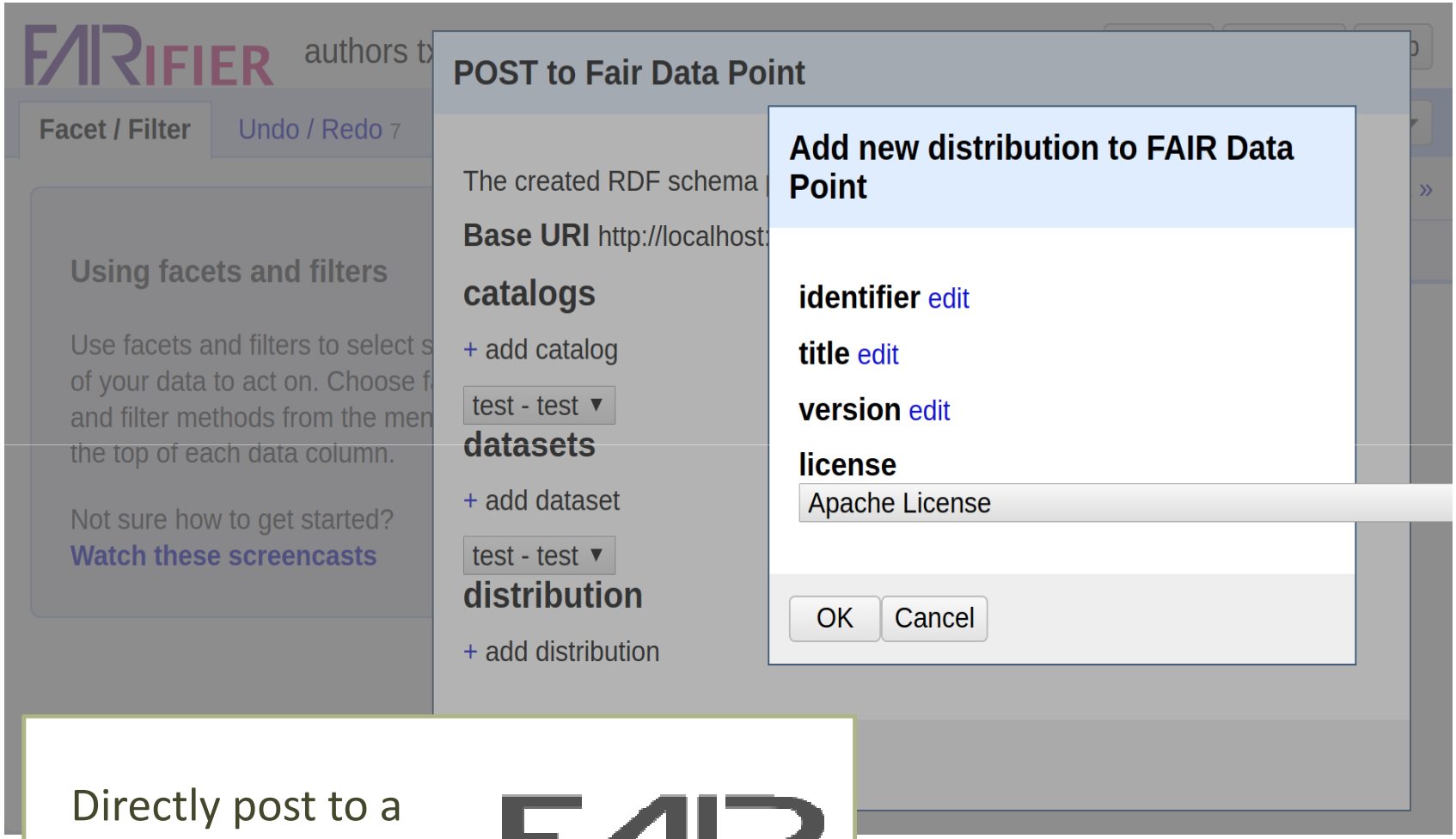
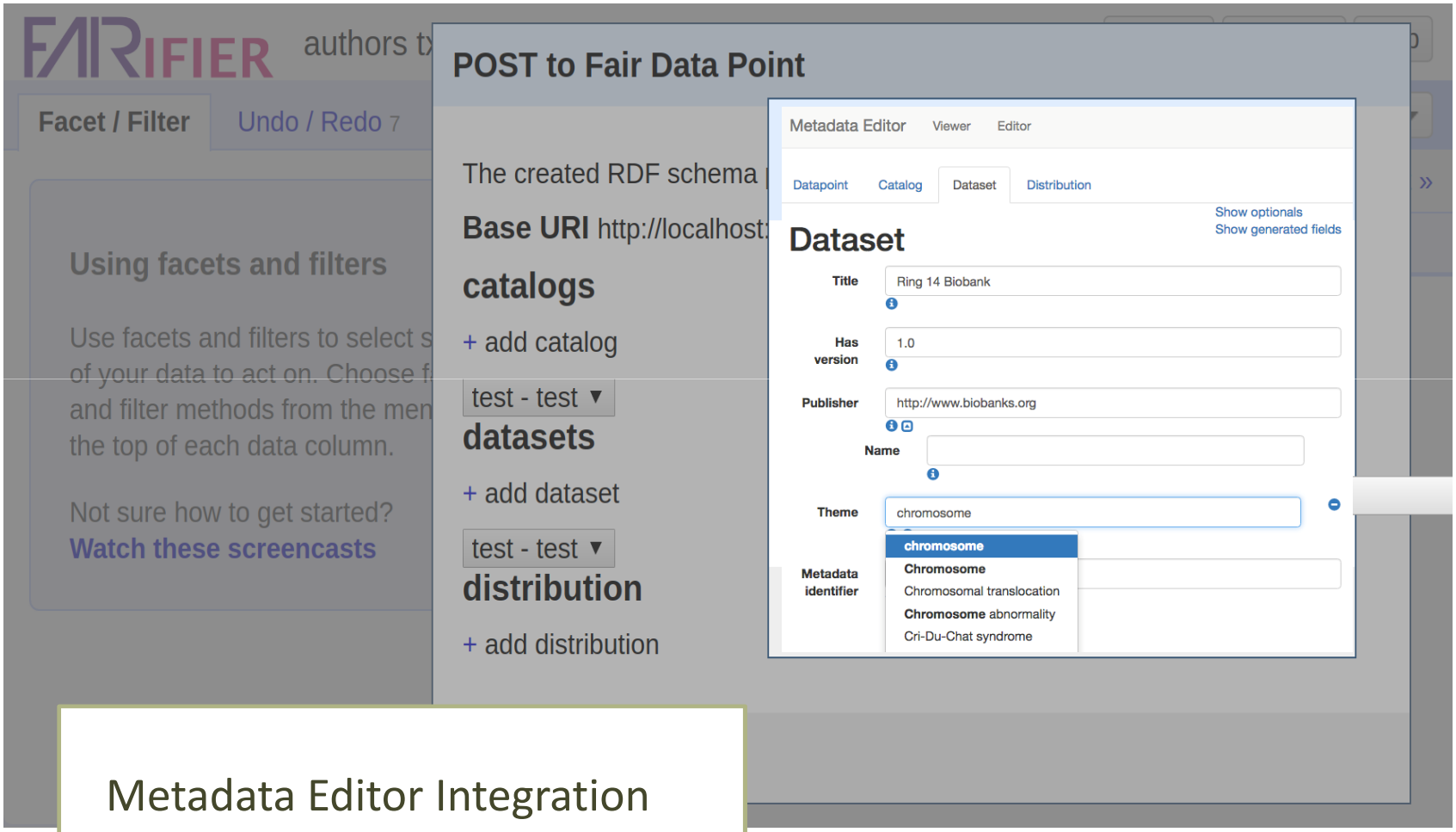


Directly post to a FAIR DATA POINT

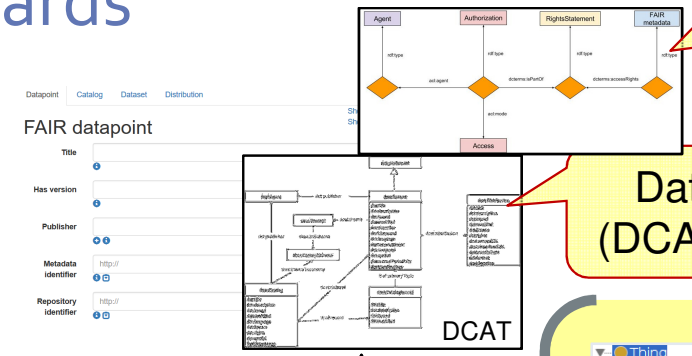


Metadata Editor Integration

FAIR DATA POINT Centred around standards

FIND, ACCESS

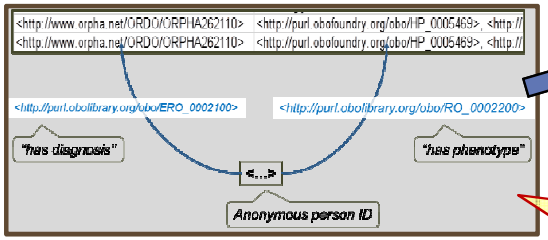
INTEROPERATE
REUSE



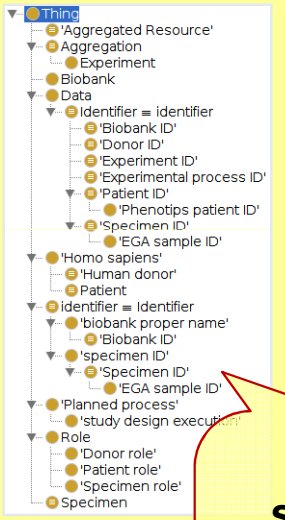
Access control model

Dataset description (DCAT, bioschema.org)

Data access protocol (REST)



Disorder	Symptoms
Chromosome 14 disease	Epileptic attacks as young child
Deletion of part of the long arm of chromosome 14	Learning deficit and growth problems



Linkable data distribution

Original tabular data distribution

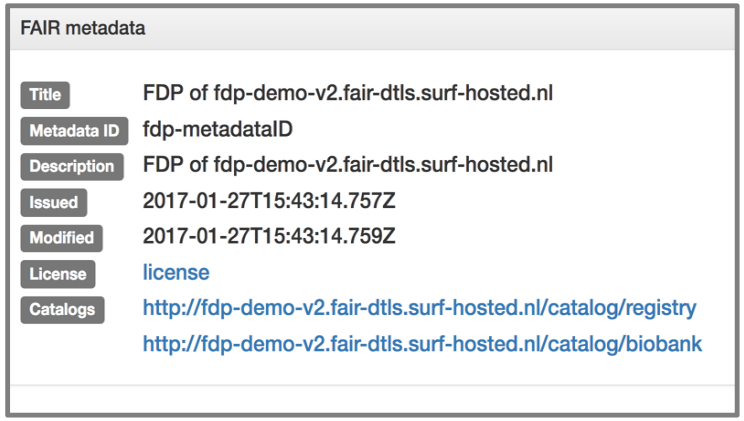
Shared semantic archetype
ORDO, HPO, OBIB, ORE, EDAM, IAO
https://biosharing.org/bsg-s000676

FAIR DATA POINT

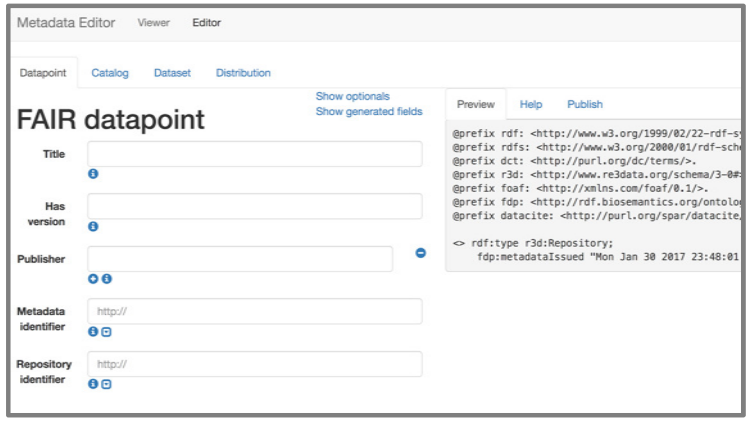
Human

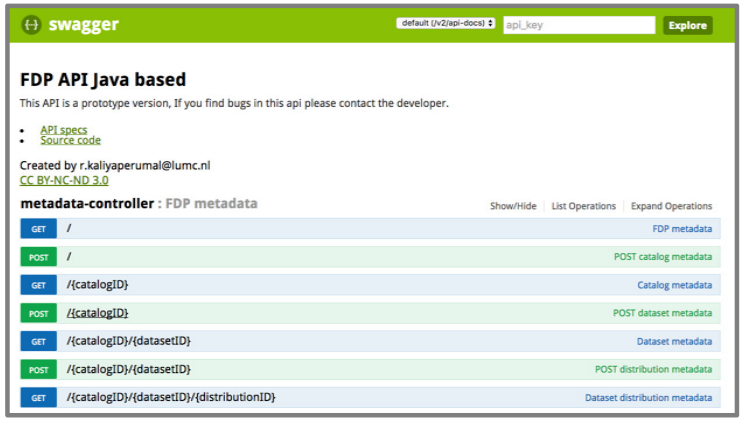
Machine

Read

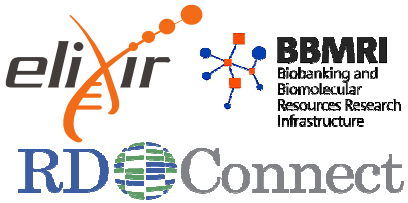


Write





What is 'FAIR'


elixir
BBMRI
Biobanking and
Biomolecular
Resources Research
Infrastructure
RD Connect

What is FAIR?

- Data can be found via search tools or in indexes
- Enabling low-cost cross-resource questions & analysis by preparing data *at the source*
- Data access is transparent
- Origin of data is transparent (provenance, audit trails)
- Meaning of data is transparent
- For humans and computers

Guiding principles for FAIR data
Human ←→ Computer

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;

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;

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;

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;

What is FAIR **not**? (1/2)

- A **standard**
  - **Principles** that are **implemented by existing standards**
- A **specific technology** or format (e.g. Linked Data or RDF)
  - Communities choose technologies to **implement FAIR**, such as RDF
- About **humans** finding, accessing, integrating, reusing
  - Computer-readability is an essential aspect of FAIR
- **Open**
  - Access 'under well-defined conditions' (conditions are transparent and computer readable)

RD Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

What is FAIR not? (2/2)

- A data management **system**
  - An **additional component** to enable cross-resource questions/analysis
  - A FAIR data port built from FAIR data services is a system
- Trivial to implement
  - Nobody can claim 100% FAIRness
  - FAIR data expertise required → **seek collaboration with FAIR data experts**
- An analysis tool
  - a **substrate** for more efficient cross-resource questions/analysis
- Made for high performance computing
  - Reduces the (much larger) cost of **making data interoperable**

Role of ERNs

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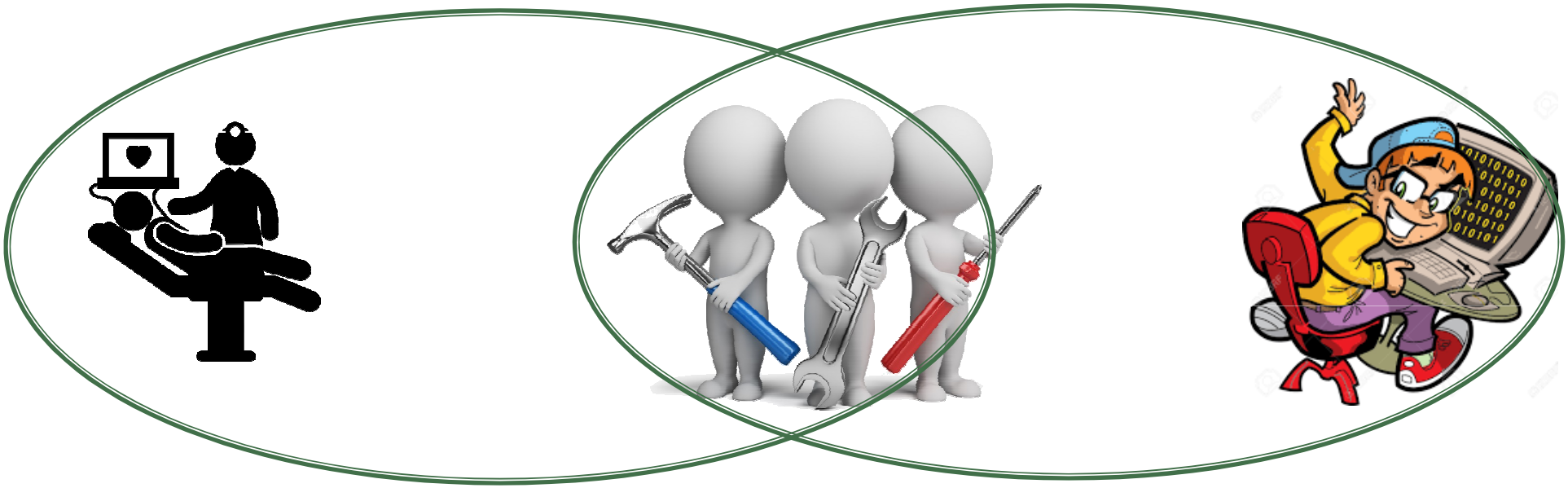
RD Connect

Role of ERNs

- Stimulate working on **local data quality** via FAIRification
  - Copy running FAIR projects at HCPs as a paradigm for your HCP
  - Add FAIR guiding principles to list of requirements
    (e.g. in a tender for a data management system for your HCP/registry)
- Help make FAIR service sustainable
  - Indicate how you can contribute to organising a FAIR support service for ERNs/Rare Diseases
- Express your interest via fair-rd-info@elixir-europe.org

RD-Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Preliminary model for service for ERNs
based on rare disease data linkage plan



Stakeholders

FAIR data stewards (the service)

Infrastructure developers (Ontologies, mapping services, FAIR software)

Collaboration and financing

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Has this shown proven benefit?

Example in cancer domain (André Dekker, Maastro Clinic, NL)

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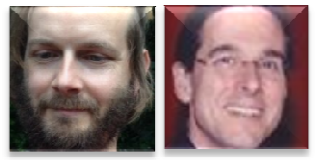
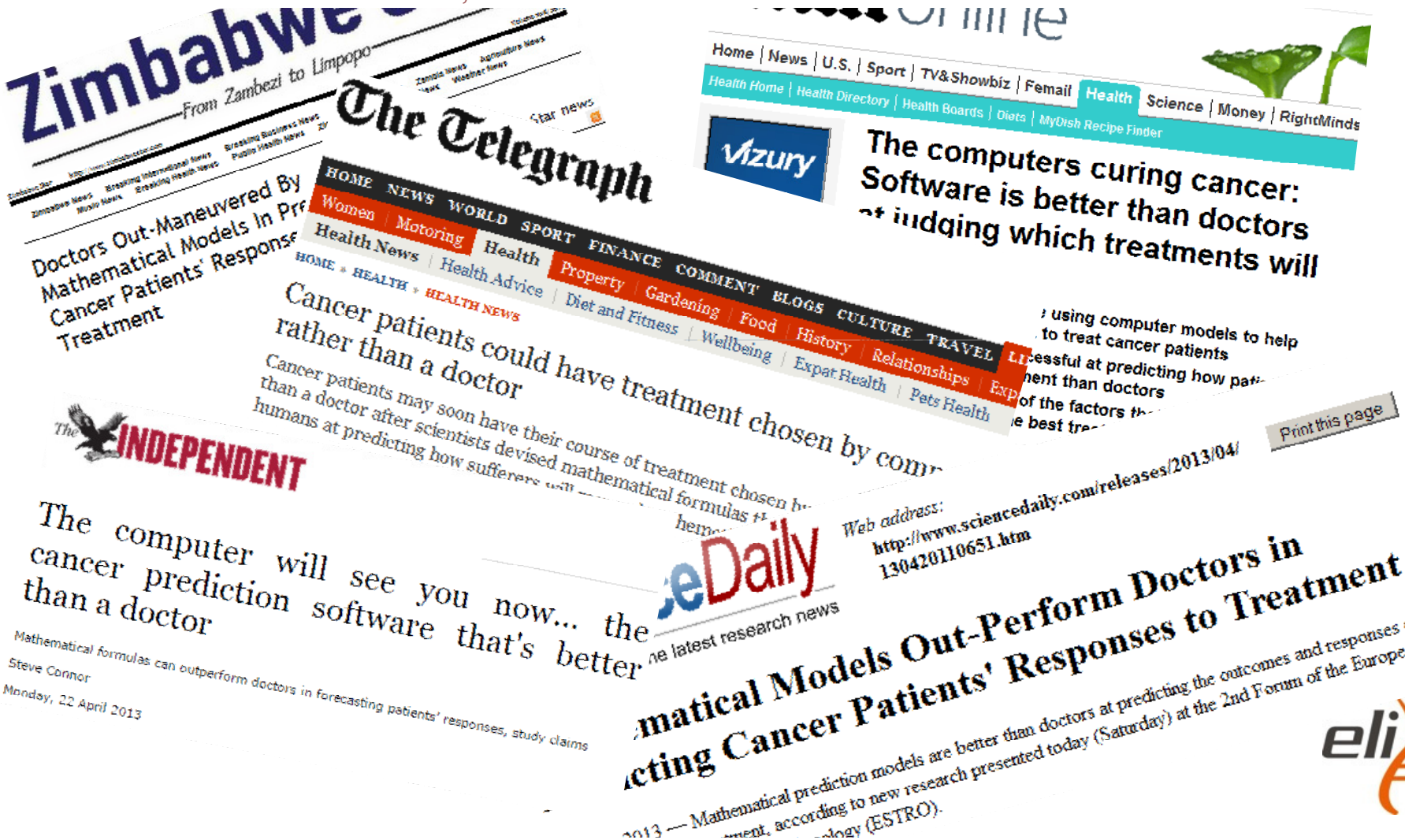
Example

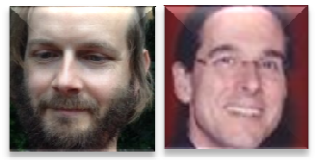
Improving cancer treatment effect prediction

Courtesy of Andre Dekker and Scott Marshall, Maastro Clinic





Andre Dekker's example
Better prediction of treatment prognosis

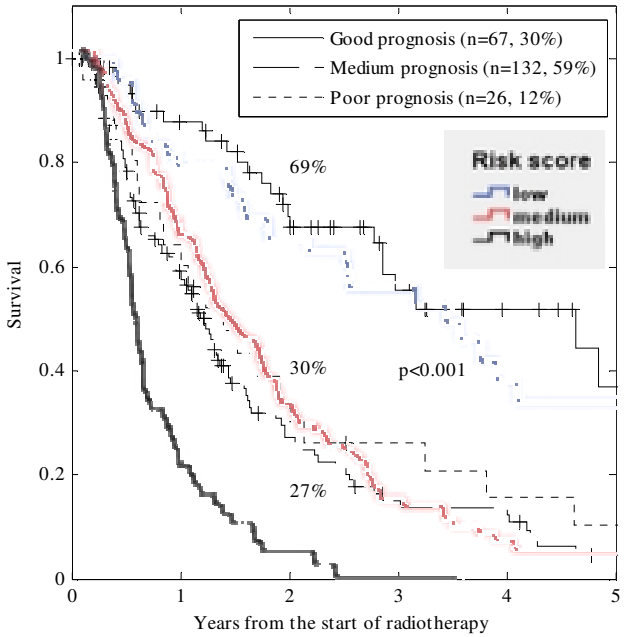


Australia

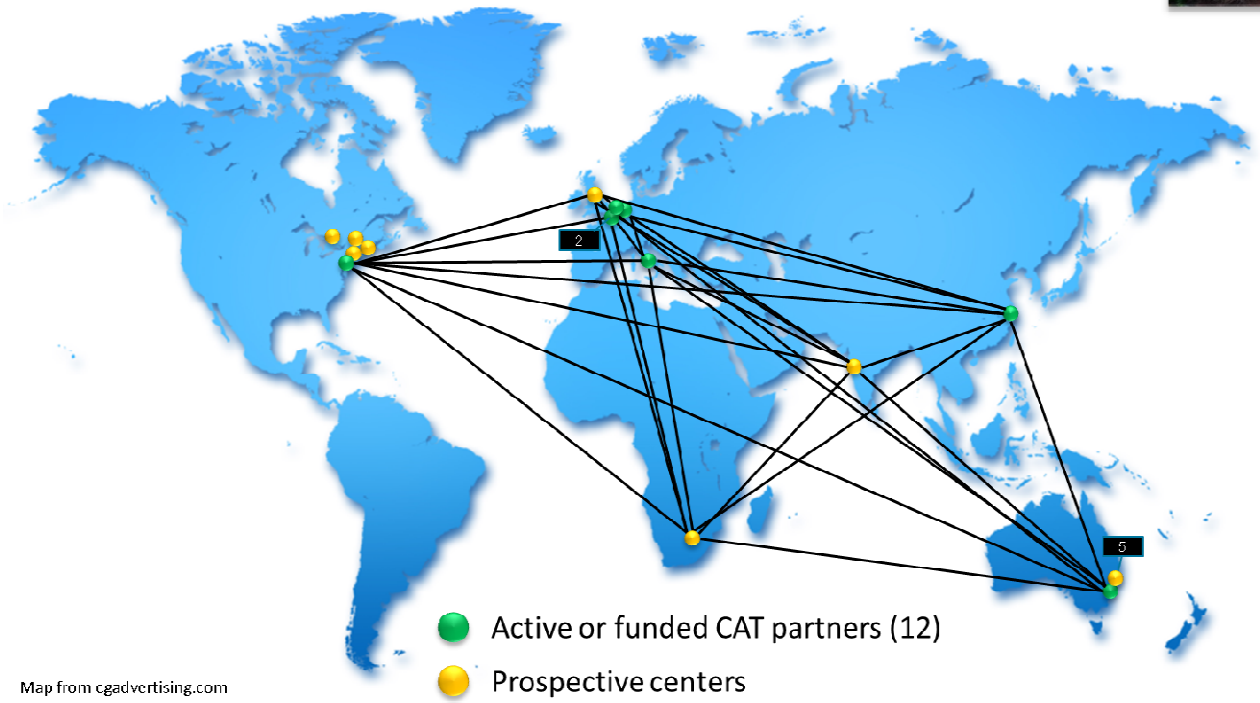
Rethink palliative treatments in good prognosis patients

Netherlands

Rethink curative treatments in poor prognosis patients



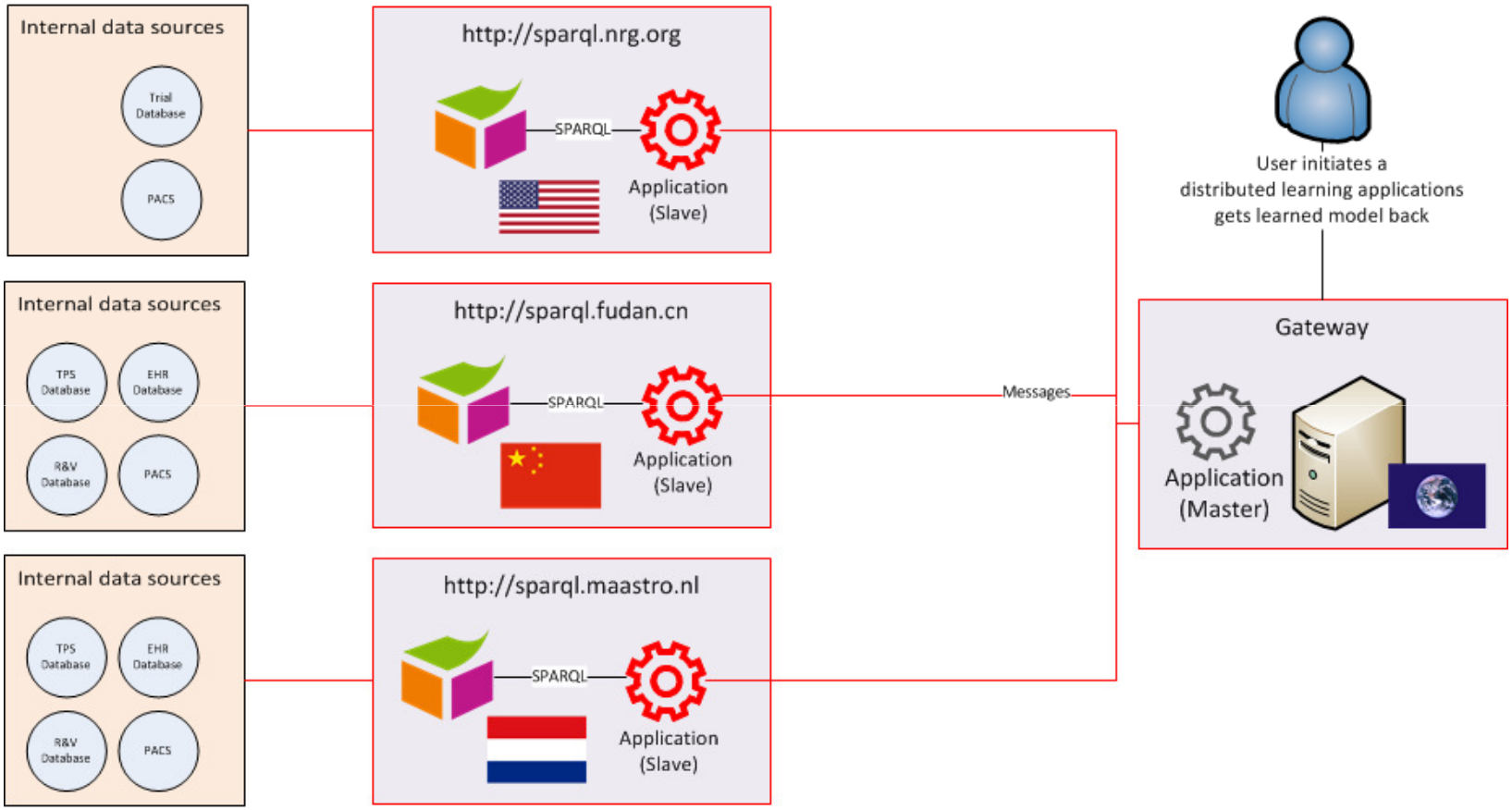
Method



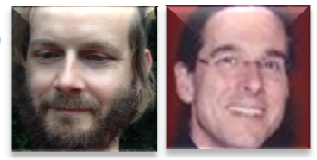
Learning algorithm on data from multiple hospitals….

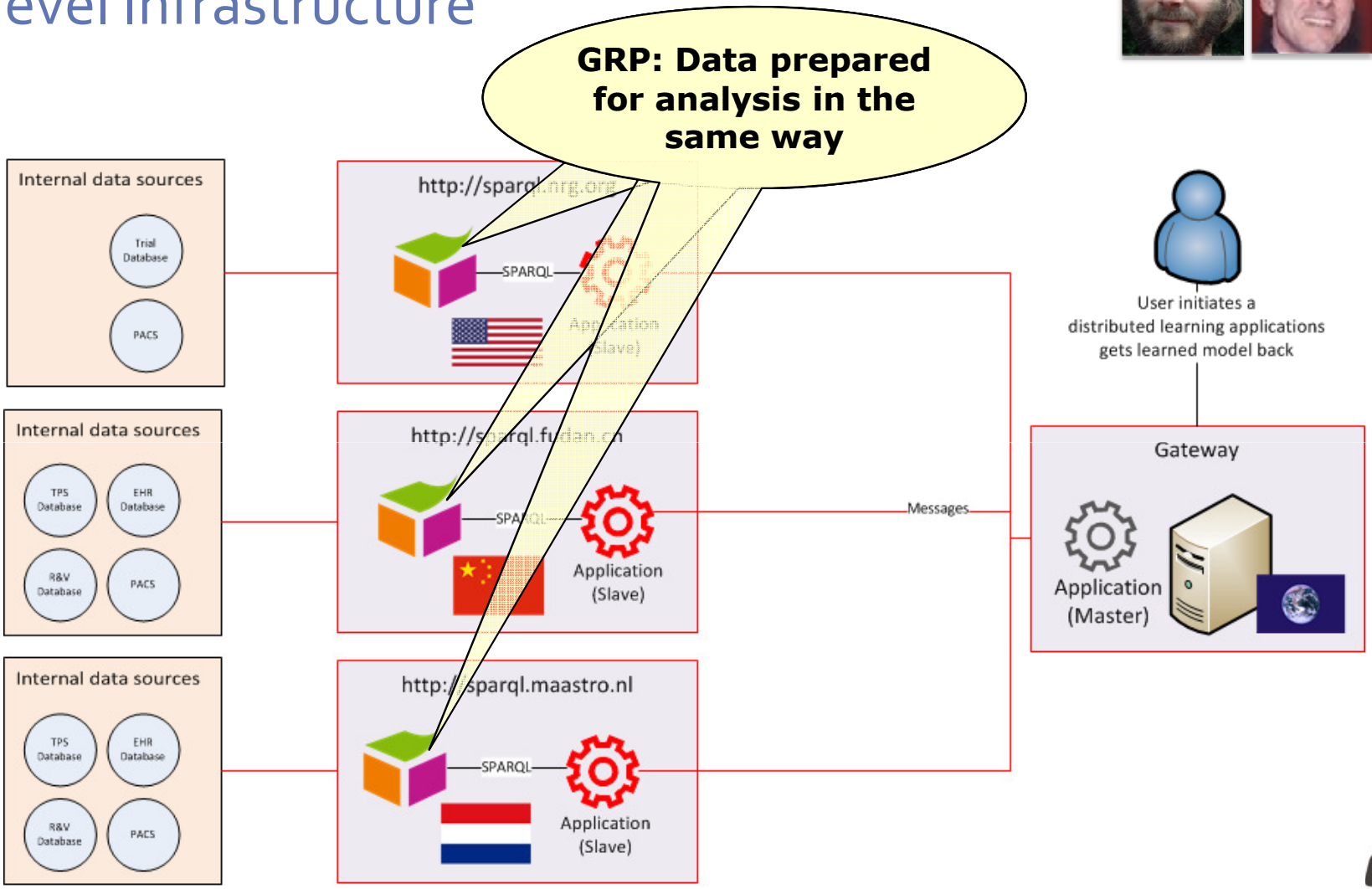
that could not be shared!!!

High level Infrastructure



High level Infrastructure





Scenario: getting data from a personal locker

Special form of FAIR data point

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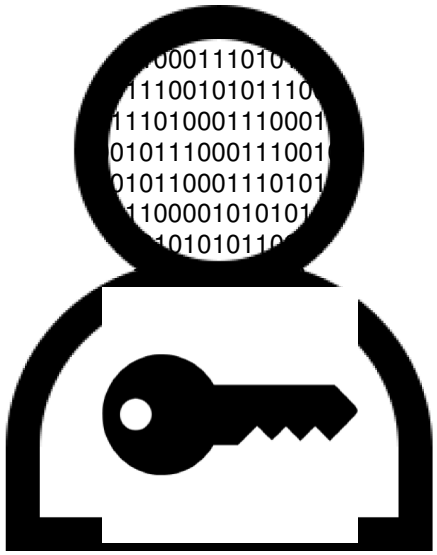
RD Connect

FAIR Compliant

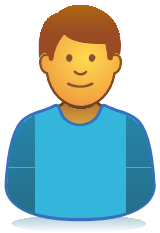
FAIR DATA



FAIR DATA POINT

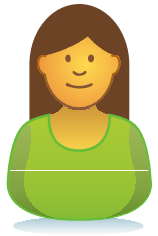
Personal
Data Locker

Data Access



Data Owner

Personal
Data Locker



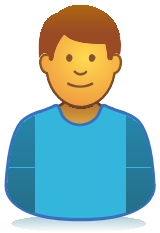
Researcher

Data Access

Data Access

Data Access

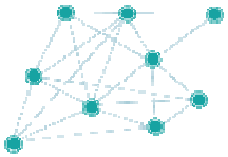
Access to non-sensitive data

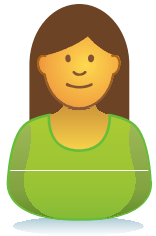


Data Owner

Personal
Data Locker

Non-sensitive data





Researcher

Access to sensitive data

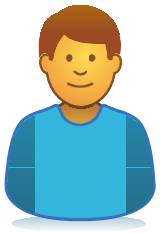
Access to sensitive data

Access to sensitive data

Access to sensitive data

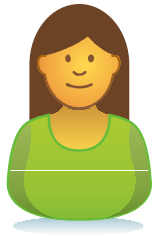
Access to sensitive data

Include user ID
in authorized list



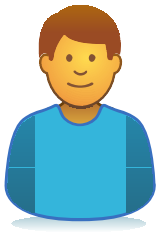
Data Owner

Personal
Data Locker



Researcher

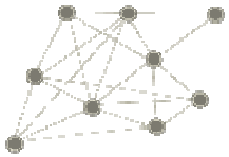
Access to sensitive data

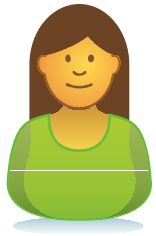


Data Owner

Personal
Data Locker

Sensitive data





Researcher

Thank you…

PLEASE CHECK RD-CONNECT, ELIXIR-EUROPE AND ISS-CNMR WEB SITE FOR UPCOMING RD-CONNECT, E-RARE WORKSHOPS IN MAY, AND INTERNATIONAL SUMMER SCHOOL IN SEPTEMBER !!!

http://rd-connect.eu/events/
http://www.iss.it/cnmr/
http://www.elixir-europe.org/events

FAIR service infrastructure development & deployment in rare disease community

Global initiatives

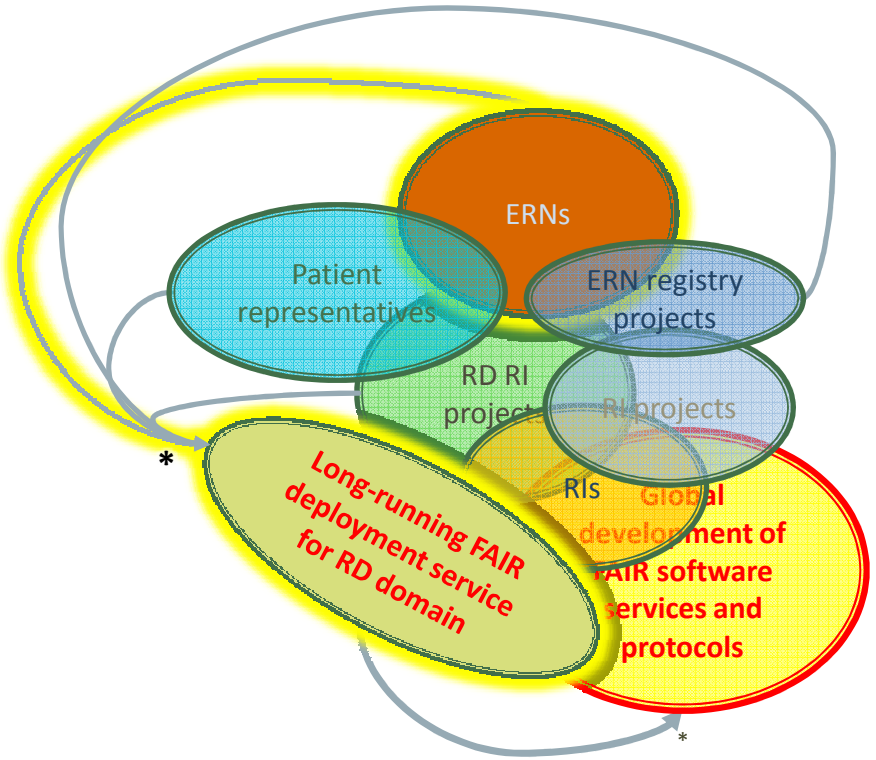
- FAIR tools and protocol developers
- FAIR deployment service

(Research) Infrastructures

- Research infrastructures: ELIXIR, BBMRI
- ERNs
- European Open Science Cloud (and GoFAIR networks)
- NIH Commons

Projects

- ERN registry projects
- RI projects: ELIXIR-EXCELERATE, ELIXIR-CORBEL, BBMRI-ADOPT
- RD RI projects: RD-Connect (→ RD-Bridges)
- Big Data to Knowledge (BD2K) USA

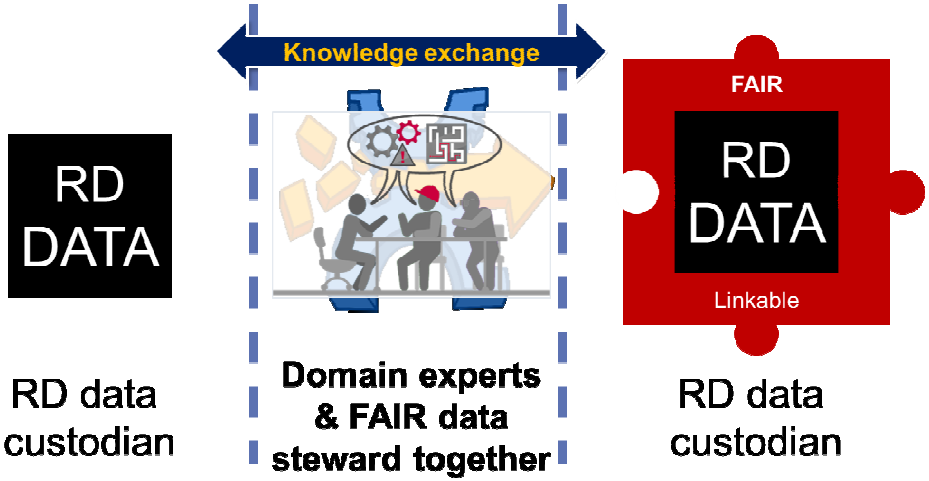


* NB stakeholders/stakeholder projects contribute to the services that they make use of

Starting point: Bring Your Own Data workshop

At BYOD

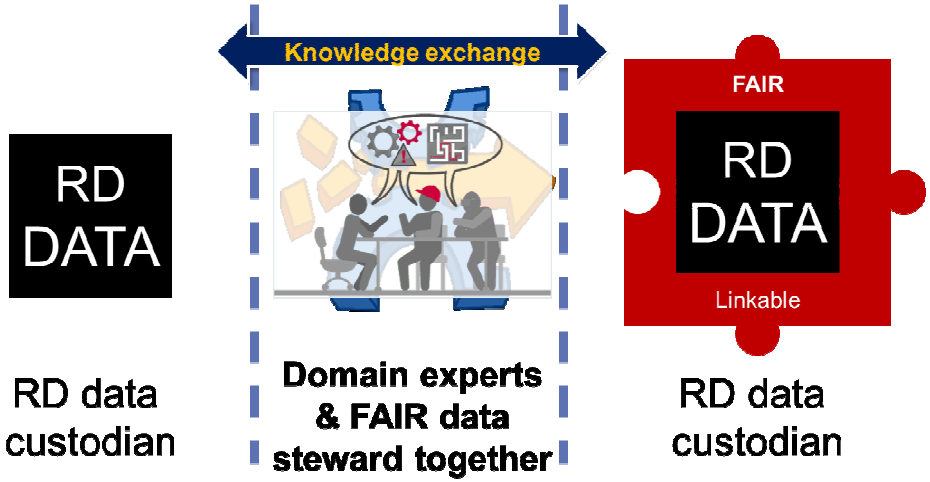
1. Introduce method
2. Refine driving questions with FAIR data experts
3. Scramble data if needed
4. OpenRefine-FAIRifier to convert data
    1. Ontology lookup services (BioPortal/OLS) to find terms from appropriate ontologies
    2. FAIRifier to apply them to the data
5. Share linkable data in linked data cache
6. Answer driving questions with FAIR data experts



Starting point: Bring Your Own Data workshop

Preparation

1. Invite data owners and FAIR data experts
2. Webinar(s) to explain the concept
3. Chase data owners to bring
    1. excerpt from real data
    2. driving questions



FAIR data project blueprint

Tasks

	Task	Period (months)	Associated Milestone Deliverable	Comments
Start of preparation phase	Define driving research questions and target data for FAIRification (use case)	1	M1	
	Technical assessment of requirements for the BYOD	1-2	M2	
	Select BYOD experts	3	M2	<Disease D> experts, <Disease D> database experts, FAIR data experts
	Prepare minimal 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 FDPs for testing
	Design and execute data analysis for testing FAIR Data Points as substrate for knowledge discovery on <Disease D> (proof-of-principle)	9-11	M5	Executed by collaborating research scientists
End of	Release documented	11-12	D1	By <Disease D>

- Timeline
- Effort & Budget
  - 6-10PM
  - currently 50% covered by infrastructure
- Stakeholder benefits

FAIRify rare disease resources

Dataset level

1. Create FAIR data point via metadata editor
2. Incorporate FAIR data point service into local IT infrastructure

Data level

1. Define semantic model for data set & driving integration questions
    1. Ontology lookup services (BioPortal/OLS) to find ontology terms
       Note: terms come from multiple ontologies for a typical data set
    2. Future: apply application ontology tools
2. Add FAIR data point to FAIR data demonstrator

***OR* work with software provider to generate FAIR data**

BYOD for RD data managers

- Apply procedure for data excerpts
- OpenRefine-FAIRifirer to convert
- Cache linkable data
- Answer driving questions
- DIY FAIRification sketch
- Discuss
    - Skills required for FAIRification
    - How to acquire those skills