WHY FAIR FOR RARE DISEASES?

SPEAKER: MARCO ROOS

Lieze Thielemans

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Imperial College London

Leids Universitair Medisch Centrum

Radboudumc

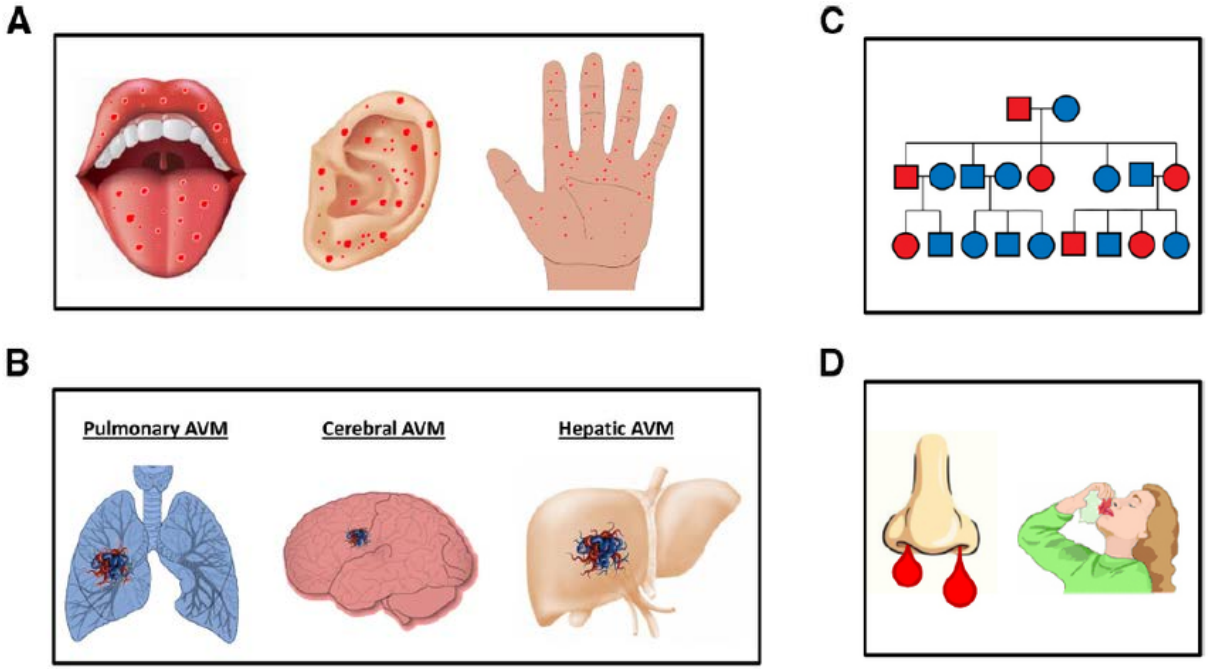
Preliminary evidence of unexpected haemolysis in patients with hereditary haemorrhagic telangiectasia:

Development of FAIR Data Stewardship for Rare Vascular Diseases in Europe

Lieze Thielemans

Supervised by: Prof. Claire Shovlin, Prof. Leo Shultze Kool and Dr Marco Roos

Hereditary haemorrhagic telangiectasia & Iron deficiency anaemia



https://www.researchgate.net/figure/Curacao-clinical-criteria-in-HHT-A-Multiple-telangiectases-at-characteristic-sites_fig4_275047054

Possible mechanisms

1) Excessive bleeding

2) Decreased absorption

3) Destruction of iron stores

Null hypothesis

*"Haemolysis does **not** occur in the HHT cohort, and that there is **no** association with iron infusions or blood transfusions to treat iron deficiency"*

What Lieze did and could not do

Manually extracted data from medical records

→ big effort for small data set

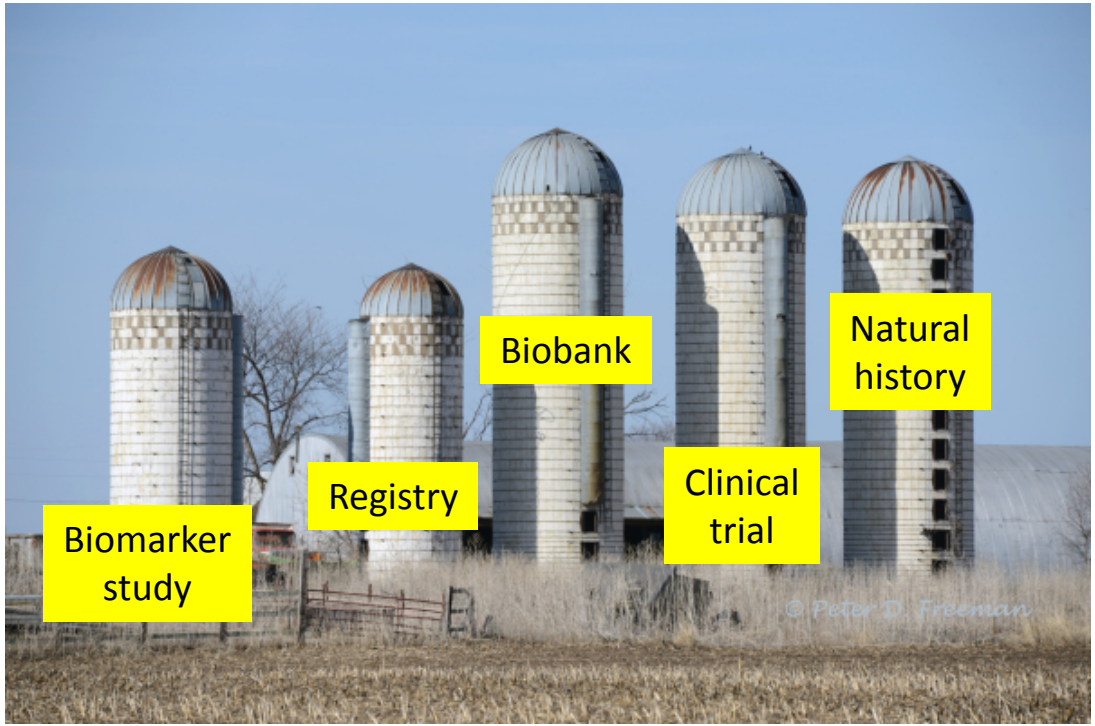
→ choice of statistics driven by this big effort, assumption-driven, not data driven

→ impossible to compare results with data from other patients

Conclusion

Local data and other people's data not readily available for analysis

Multiply this with all researchers investigating biomedical data


Biomarker study
Registry
Biobank
Clinical trial
Natural history
© Peter D. Freeman

Goal

Share data, put data together

Goal

~~Share data, put data together~~

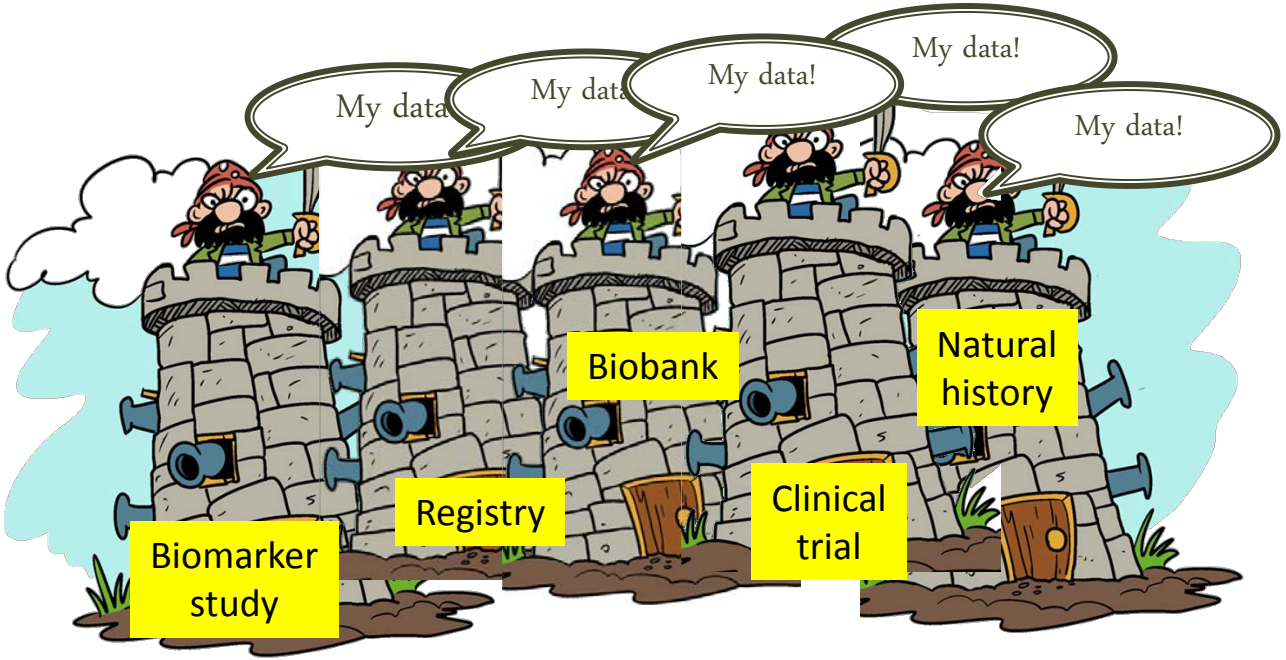
Analyse data, answer questions

Aim: Answer questions efficiently and correctly

photos: MDs, patient representatives, local data stewards involved in FAIRification pilots (DMD, VASCERN, BOND)

- *"What are the differences in age of loss of ambulation as a result of steroid use in DMD patients across EU countries?"*
- *"How do patients with the HHT phenotype respond to radiotherapy in other hospitals; what associations with iron infusions do other patients with iron deficiencies have"*
- *"What is the current information about the course and treatment of vascular malformations and hemangiome in all registries?"*
- *"What number of fractures is the most common in patients carrying mutations on the COL1A1 gene across Europe?"*
- *"What are the associations between genetic variant, disease severity and phenotypes in Rett syndrome?"*
- *"Where can I obtain tissue samples for transcriptome analysis from donors with similar symptoms as my patients?"*

Why is this not easy?



The biggest bottleneck for data sharing

is the need to share data

the *perceived* need

to *physically* share data

Signs

- Data sharing guidelines, embargo's, Data access forms, etc.
- Common data element debates
- Aligning policies between institutes and across borders
- Data warehousing attempts

Absolute need to enable analysis over resources

- Sparse
- Highly distributed
- Heterogeneous
- Different format
- Poorly interoperable
- Sensitive
- ……..

A different approach

The Personal Health Train paradigm

If sharing is hard…

Don’t share

Don’t share, be FAIR

make **your** data

Findable

Don’t share, be FAIR

Accessible

(under well-defined conditions)

Don’t share, be FAIR

Interoperable

Don’t share, be FAIR

Reusable

Don’t share, be FAIR

for humans

and computers

Don’t share, be FAIR

at the source

Improving cancer treatment effect prediction

Courtesy of Andre Dekker and Scott Marshall, Maastro Clinic

Zimbabwe Star
From Zambezi to Limpopo

Doctors Out-Maneuvered By Mathematical Models In Predi
Cancer Patients' Responses T
Trea ent

The Telegraph

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Cancer patients could have treatment chosen by comp
rather than a doctor

Cancer patients may soon have their course of treatment chosen b
than a doctor after scientists devised mathematical formulas t
humans at predicting how sufferers will
hemo

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Vizury

The computers curing cancer:
Software is better than doctors
udging which treatments will

could soon be using computer mod
cide how best to treat cancer
are more successful at
ond to treatmen
an analys
me

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The INDEPENDENT

The computer will see you now... the
cancer prediction software that's better
than a doctor

Mathematical formulas can outperform doctors in forecasting patients' responses, study claims

Steve Connor

Monday, 22 April 2013

eDaily

e latest research news

Web address:
http://www.sciencedaily.com/releases/2013/04/
130420110651.htm

matical Models Out-Perform Doctors in
cting Cancer Patients' Responses to Treatment

Mathematical prediction models are better than doctors at predicting the outcomes and responses of lung
according to new research presented today (Saturday) at the 2nd Forum of the European
(ESTRO).

RD Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Dekker's example

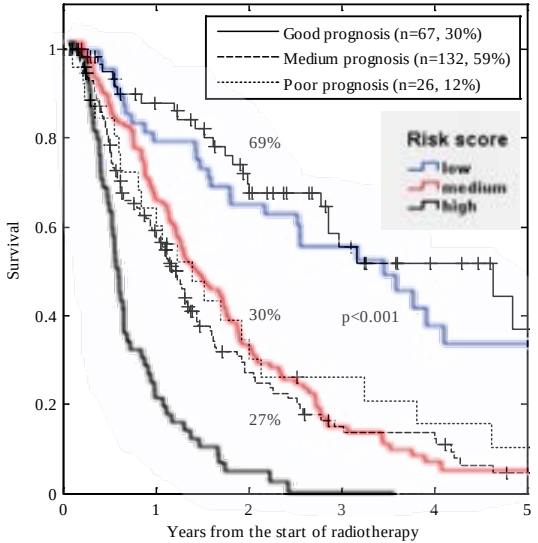
Better prediction of treatment prognosis

Australia

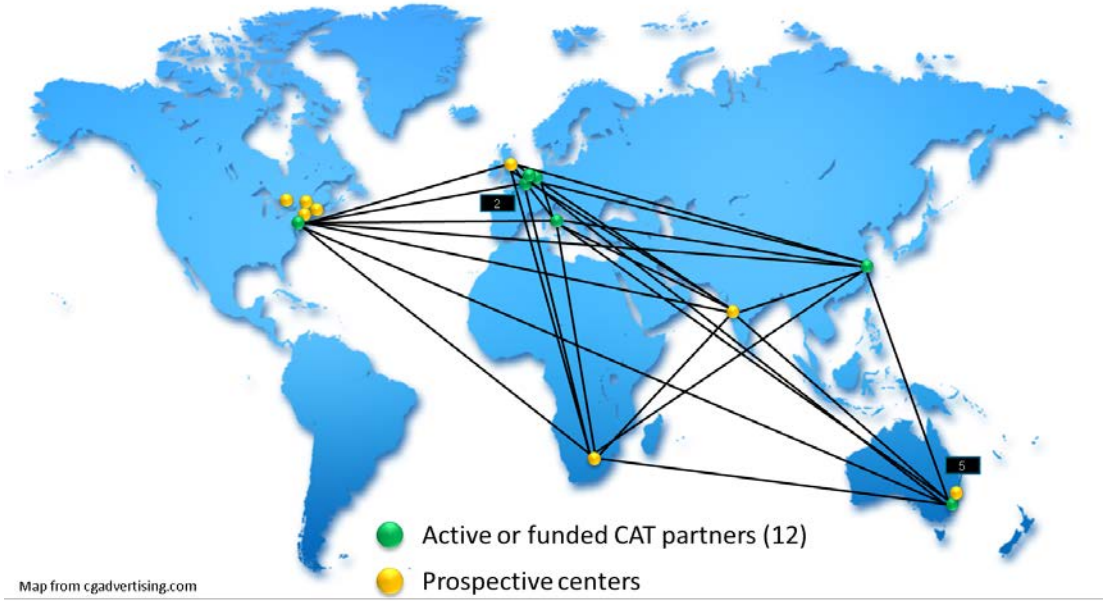
Rethink palliative treatments in good prognosis patients

Netherlands

Rethink curative treatments in poor prognosis patients



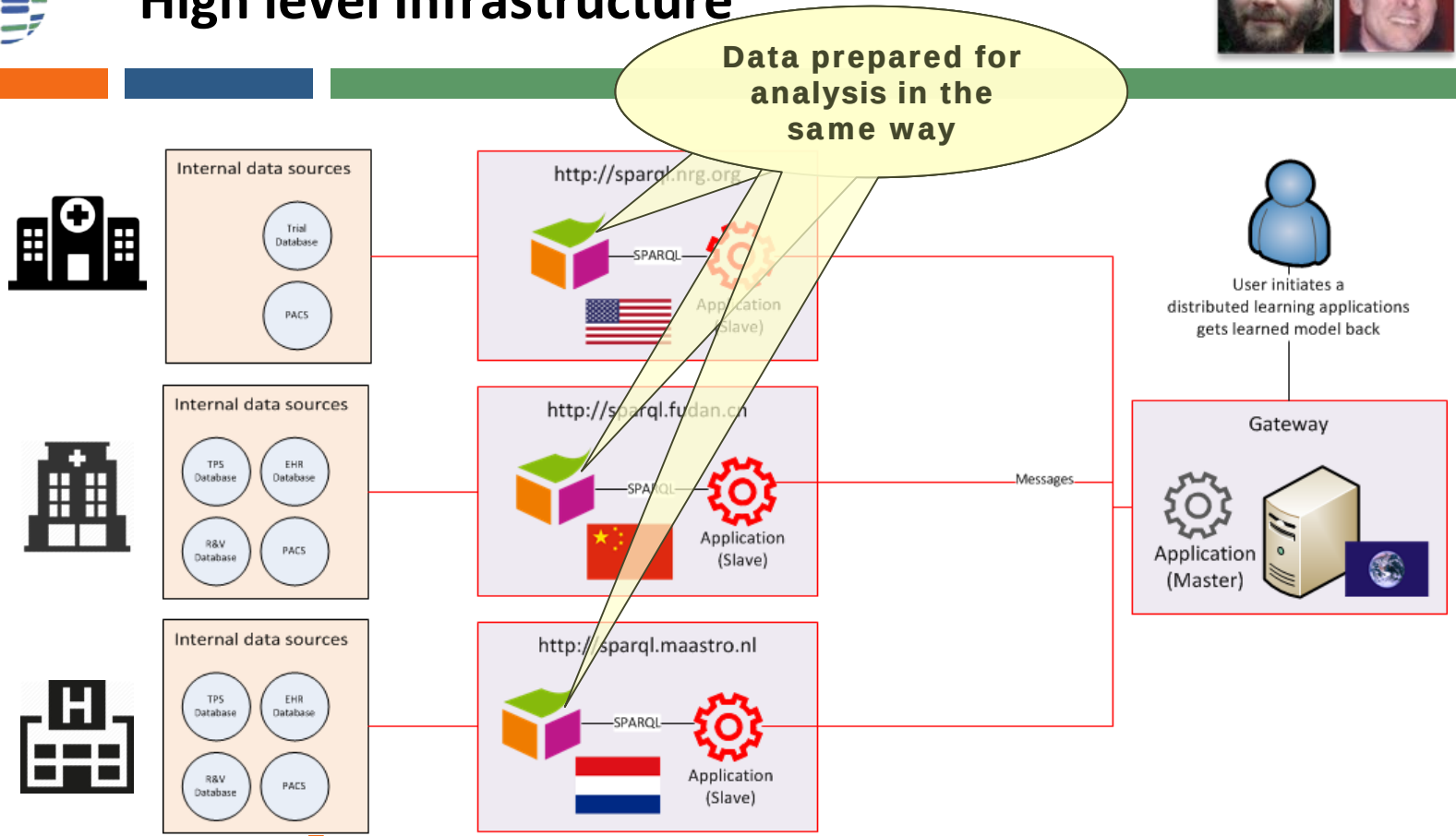
Method



Machine learning algorithm on data from multiple hospitals….

that were never shared and never put together!!

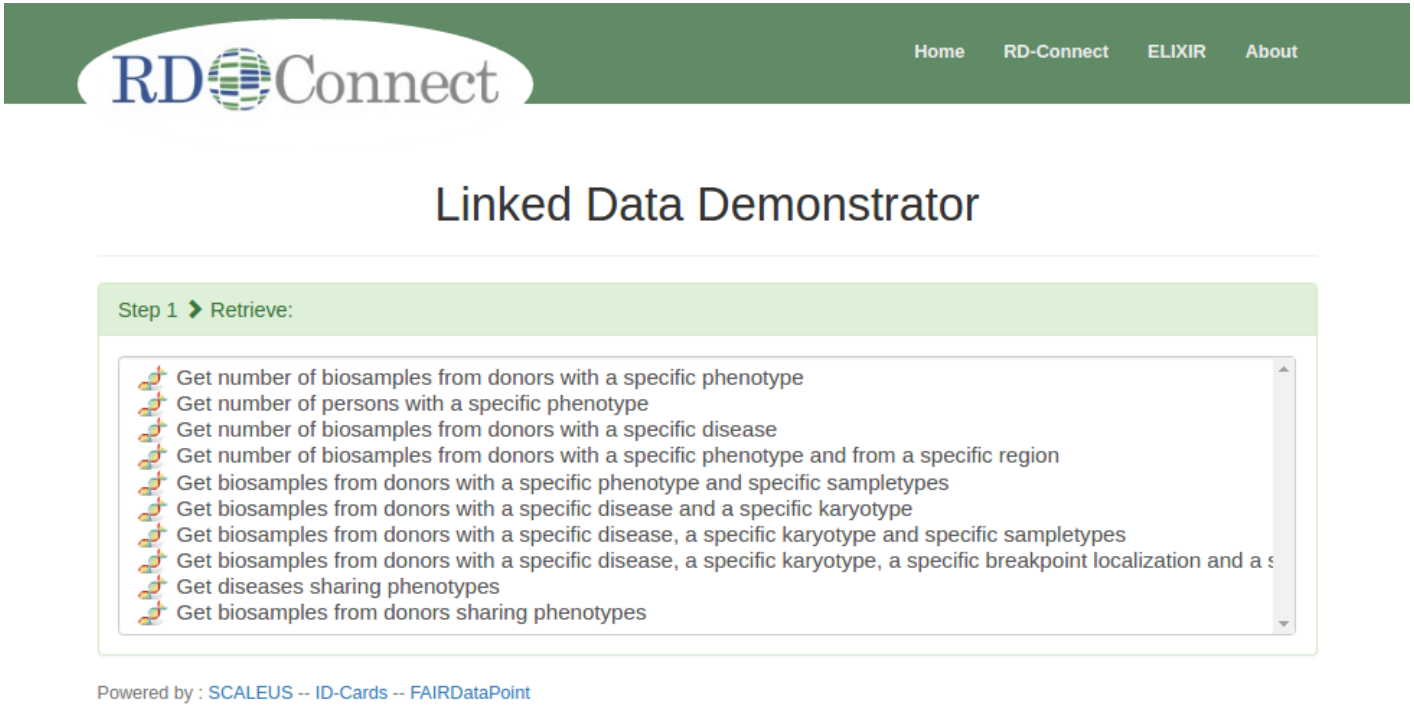
High level Infrastructure



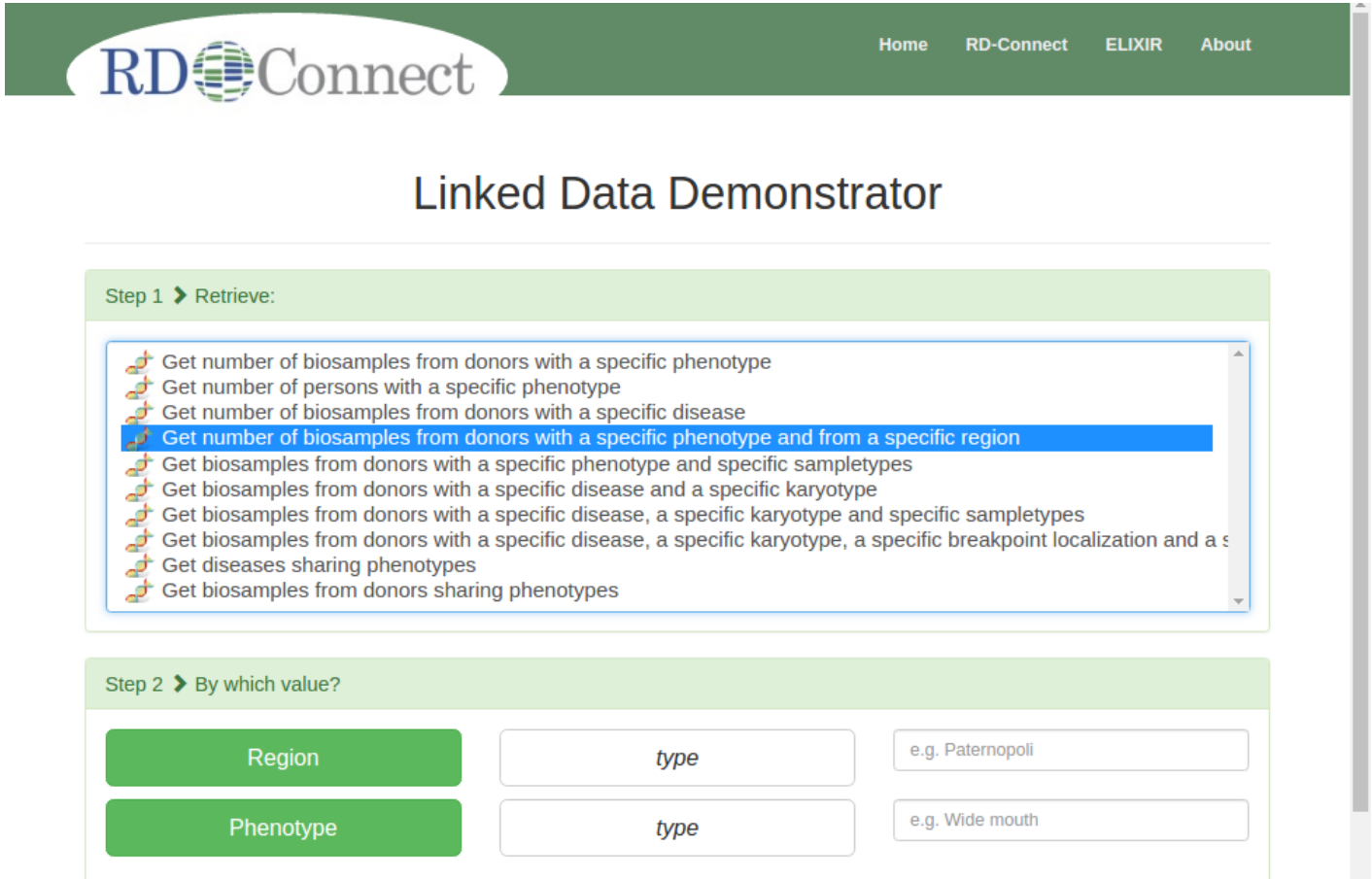
Demonstrator

Where can I obtain **biosamples** of donors with **an abnormality in head or neck**?

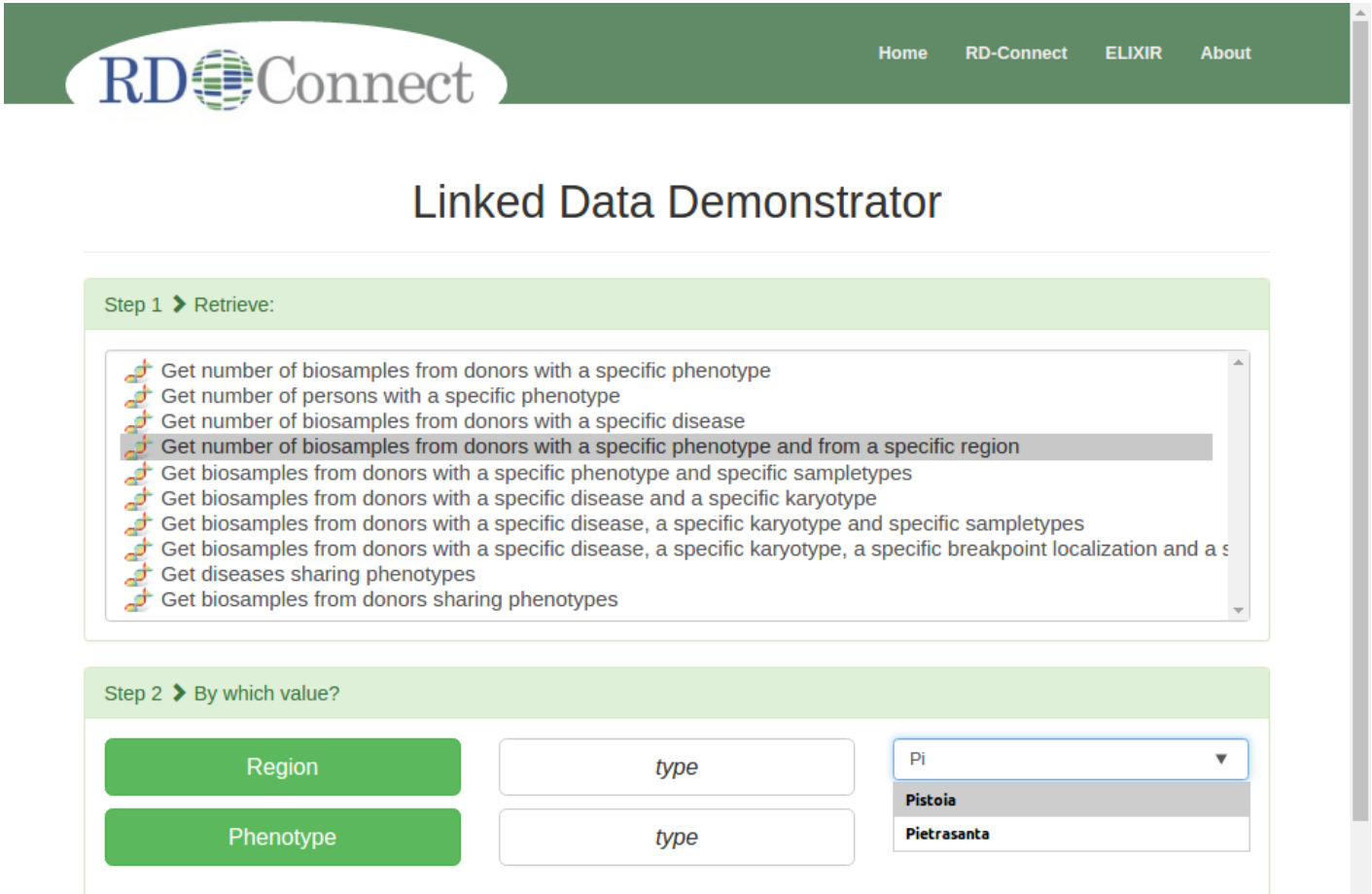
In which **biobanks** can I find these samples?



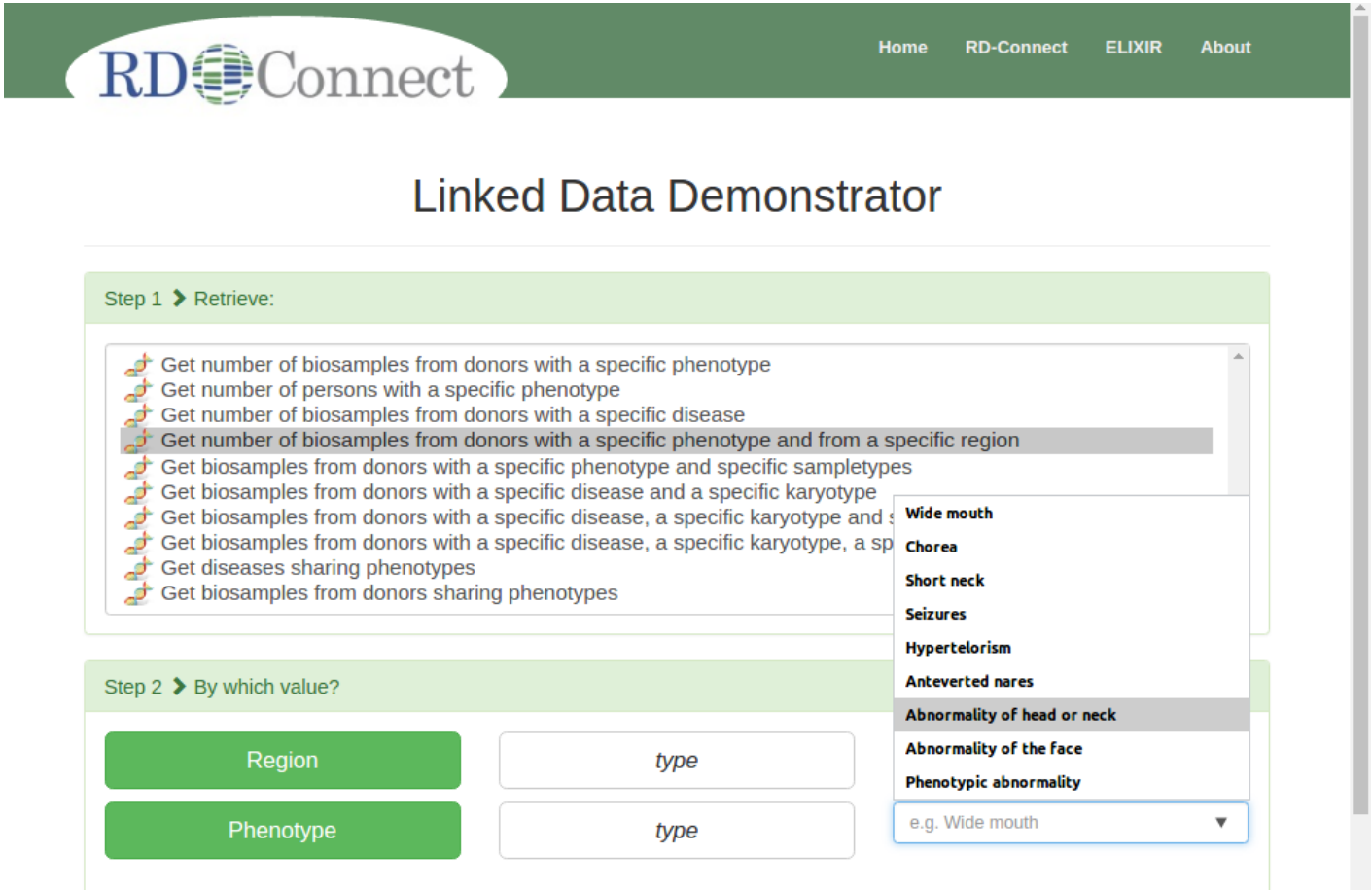
http://semlab1.liacs.nl:8080/fair-data-demo/ 26 September 2018



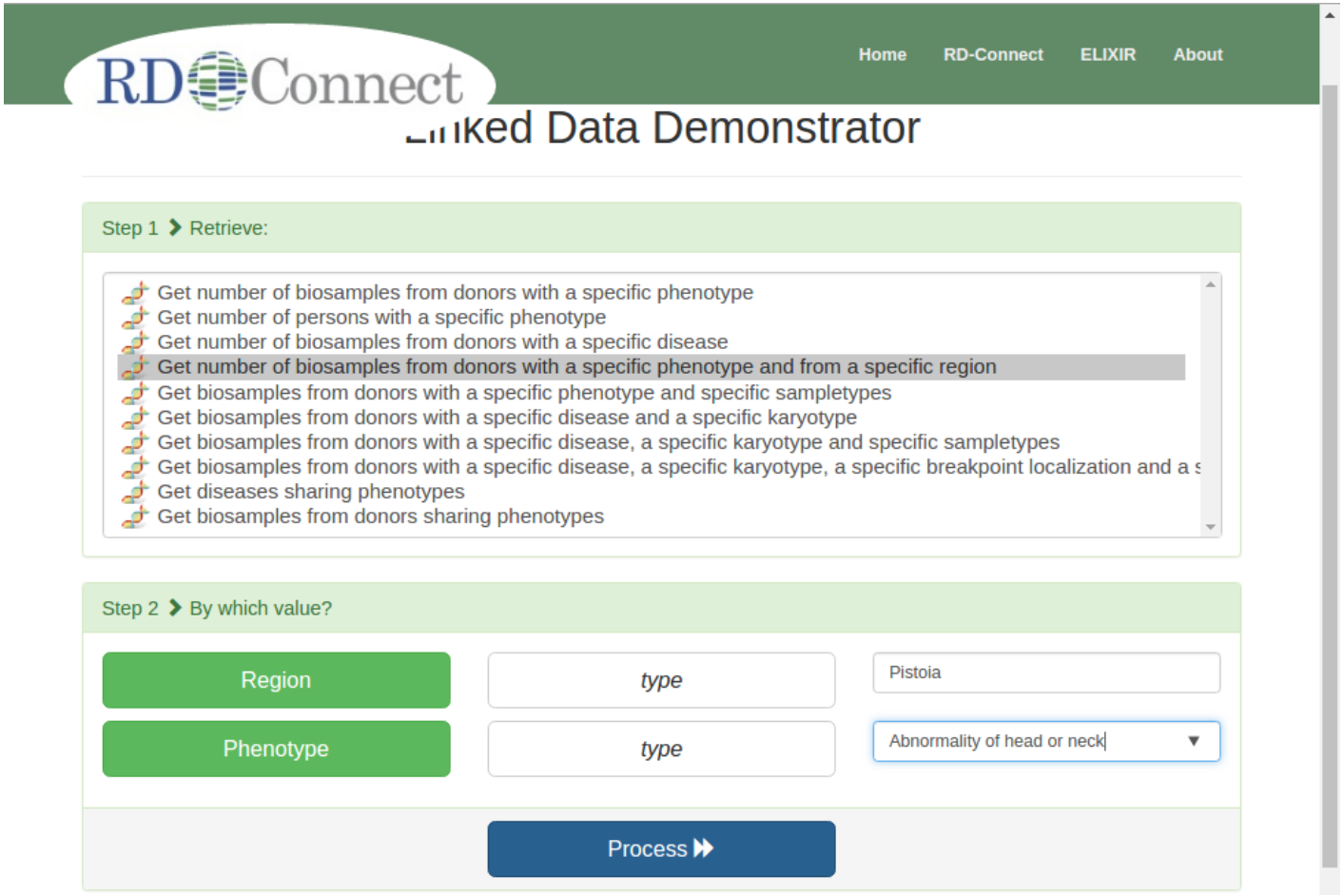
http://semlab1.liacs.nl:8080/fair-data-demo/ 26 September 2018



http://semlab1.liacs.nl:8080/fair-data-demo/ 26 September 2018



http://semlab1.liacs.nl:8080/fair-data-demo/ 26 September 2018


RD Connect
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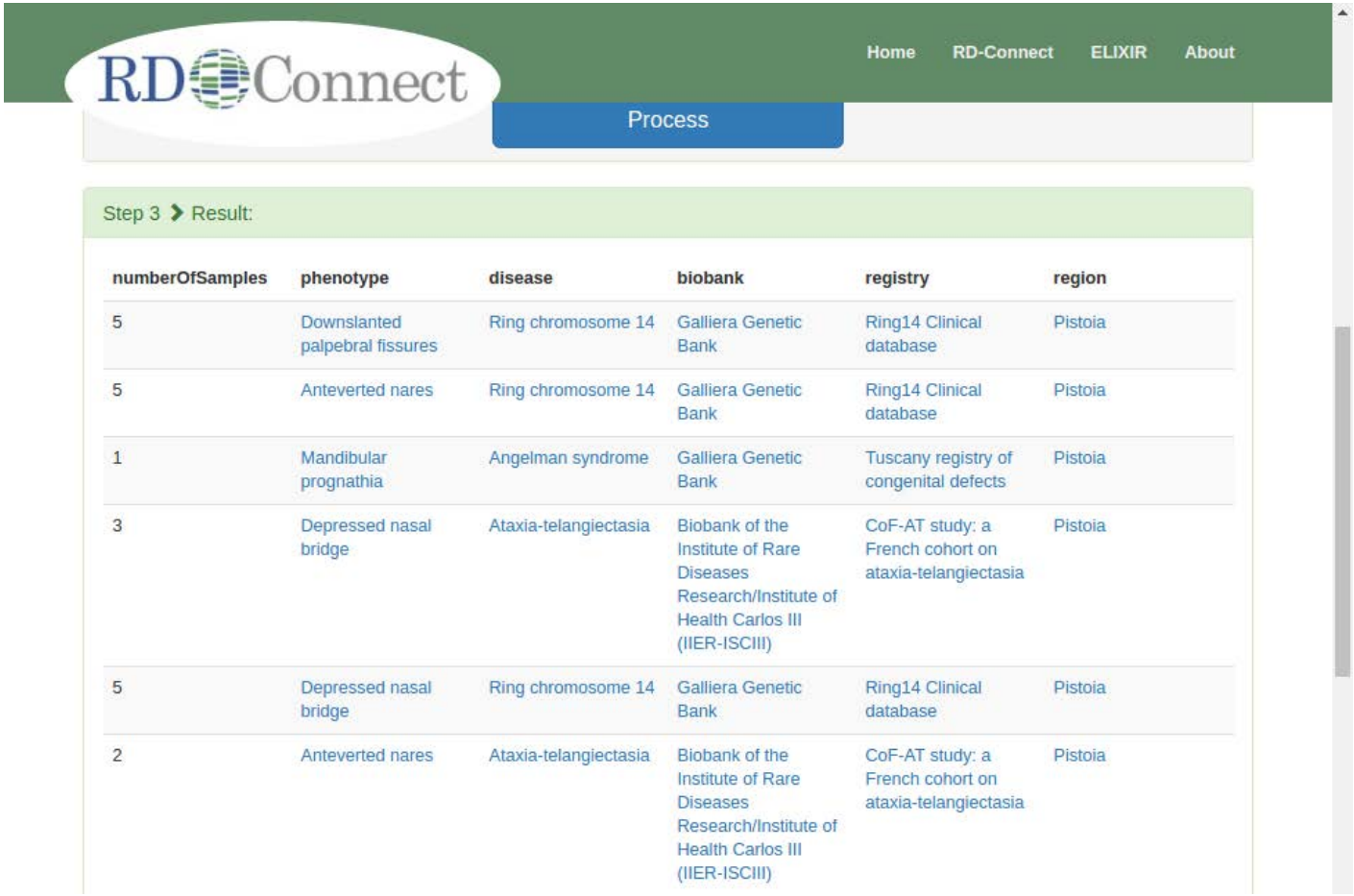
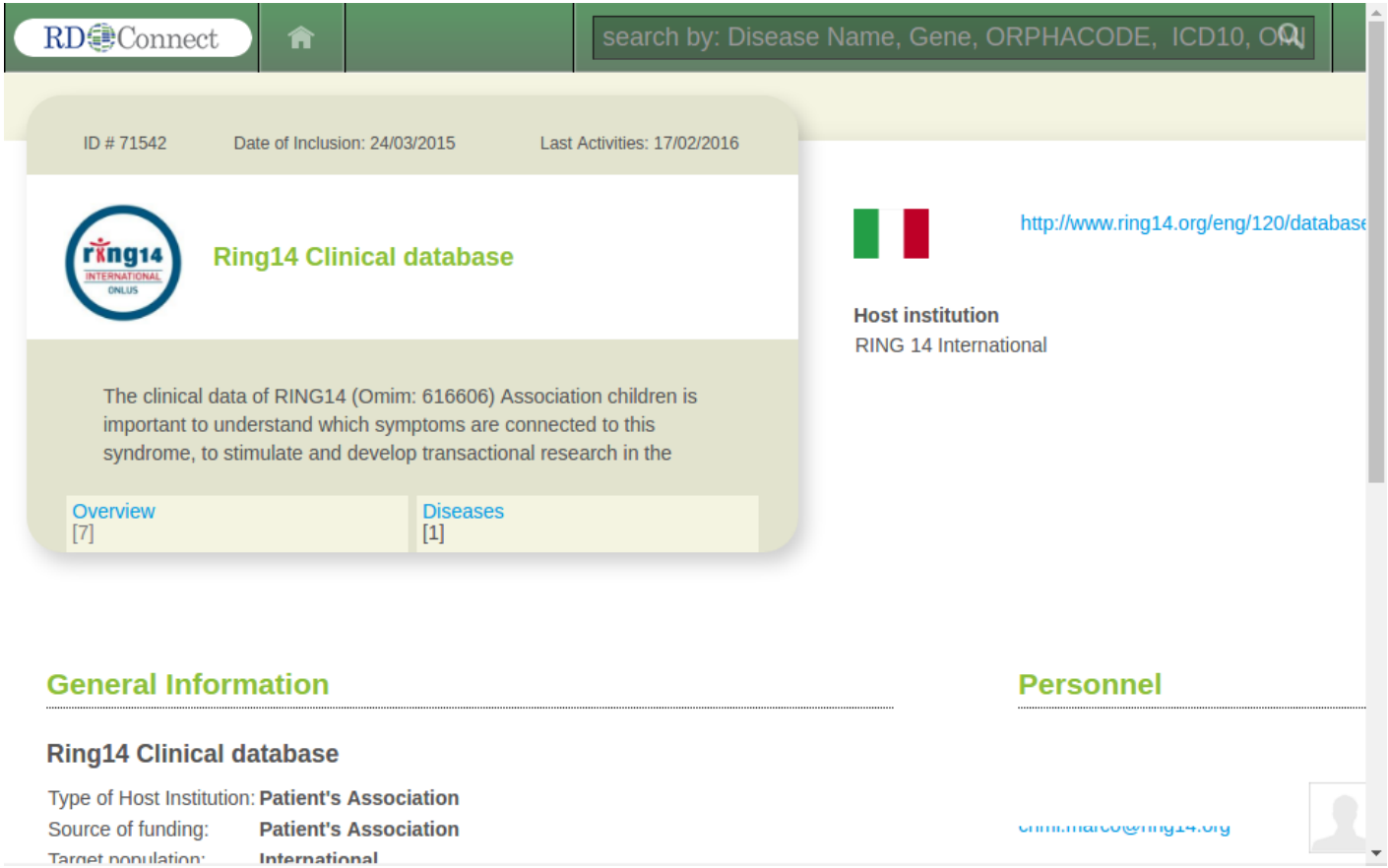
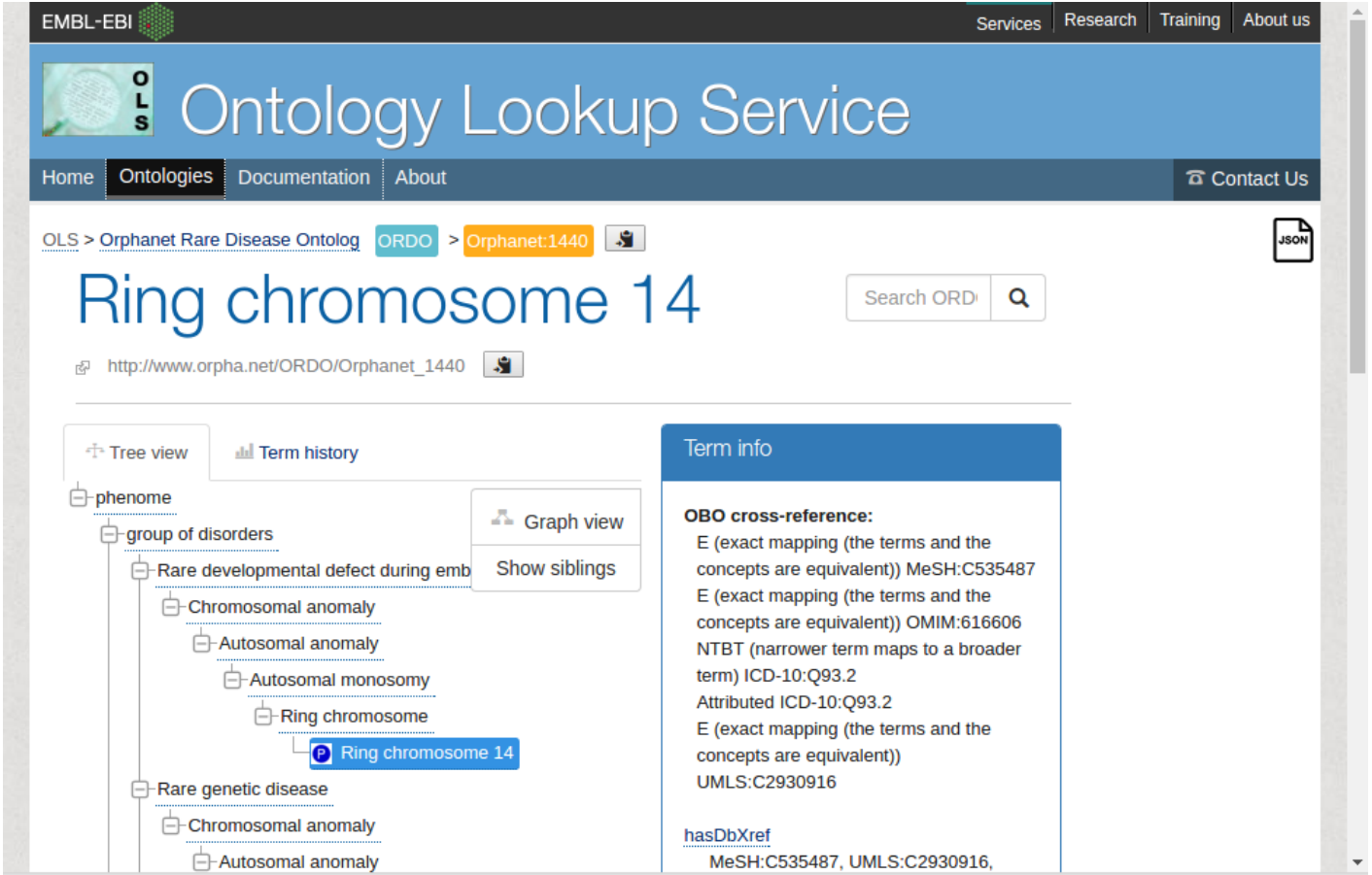
RD-Connect
ELIXIR
About
Linked Data Demonstrator
Step 1 Retrieve:
Get number of biosamples from donors with a specific phenotype
Get number of persons with a specific phenotype
Get number of biosamples from donors with a specific disease
Get number of biosamples from donors with a specific phenotype 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
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



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

http://semlab1.liacs.nl:8080/fair-data-demo/ 26 September 2018


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
http://www.ring14.org/eng/120/database
Host institution
RING 14 International
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]
General Information
Personnel
Ring14 Clinical database
Type of Host Institution: Patient's Association
Source of funding: Patient's Association
Target population: International


EMBL-EBI
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Ontologies
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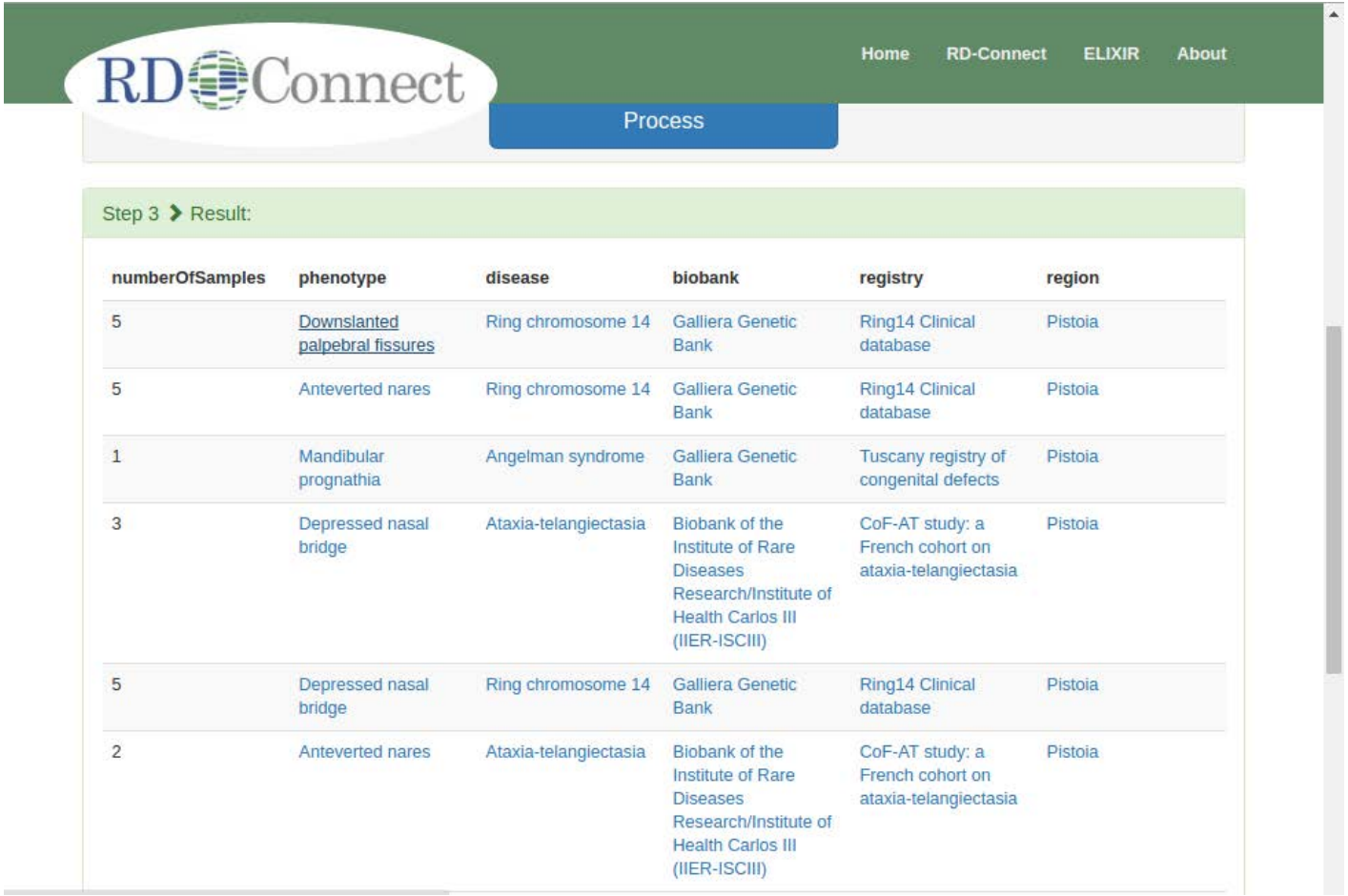
Contact Us
OLS > Orphanet Rare Disease Ontolog
ORDO
> Orphanet:1440
JSON
Ring chromosome 14
Search ORD
http://www.orpha.net/ORDO/Orphanet_1440
Tree view
Term history
phenome
group of disorders
Rare developmental defect during emb
Chromosomal anomaly
Autosomal anomaly
Autosomal monosomy
Ring chromosome
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,


RD Connect
Home
RD-Connect
ELIXIR
About
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
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3
Depressed nasal bridge
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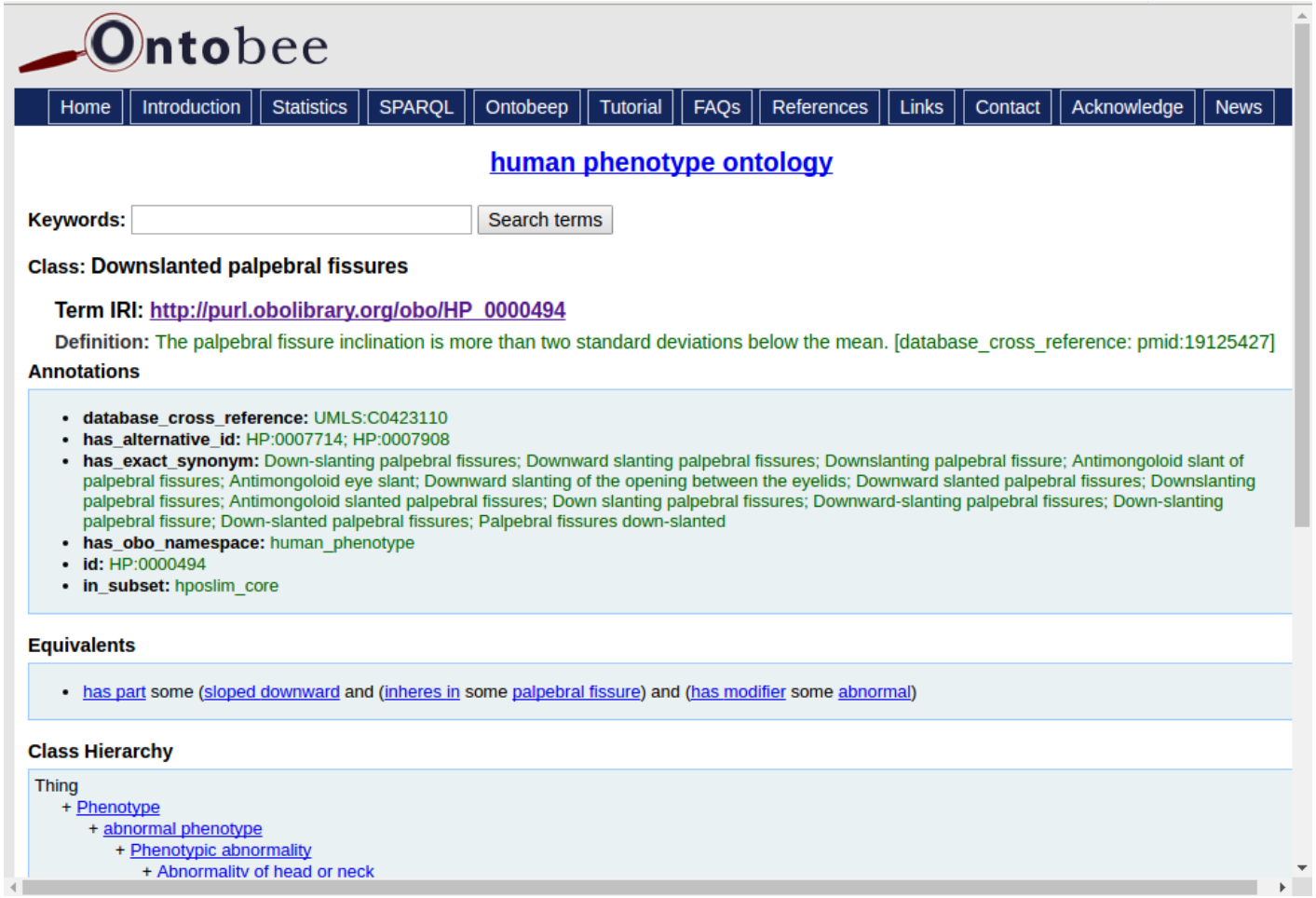
Pistoia
5
Depressed nasal bridge
Ring chromosome 14
Galliera Genetic Bank
Ring14 Clinical database
Pistoia
2
Anteverted nares
Ataxia-telangiectasia
Biobank of the Institute of Rare Diseases Research/Institute of Health Carlos III (IIER-ISCIII)
CoF-AT study: a French cohort on ataxia-telangiectasia
Pistoia

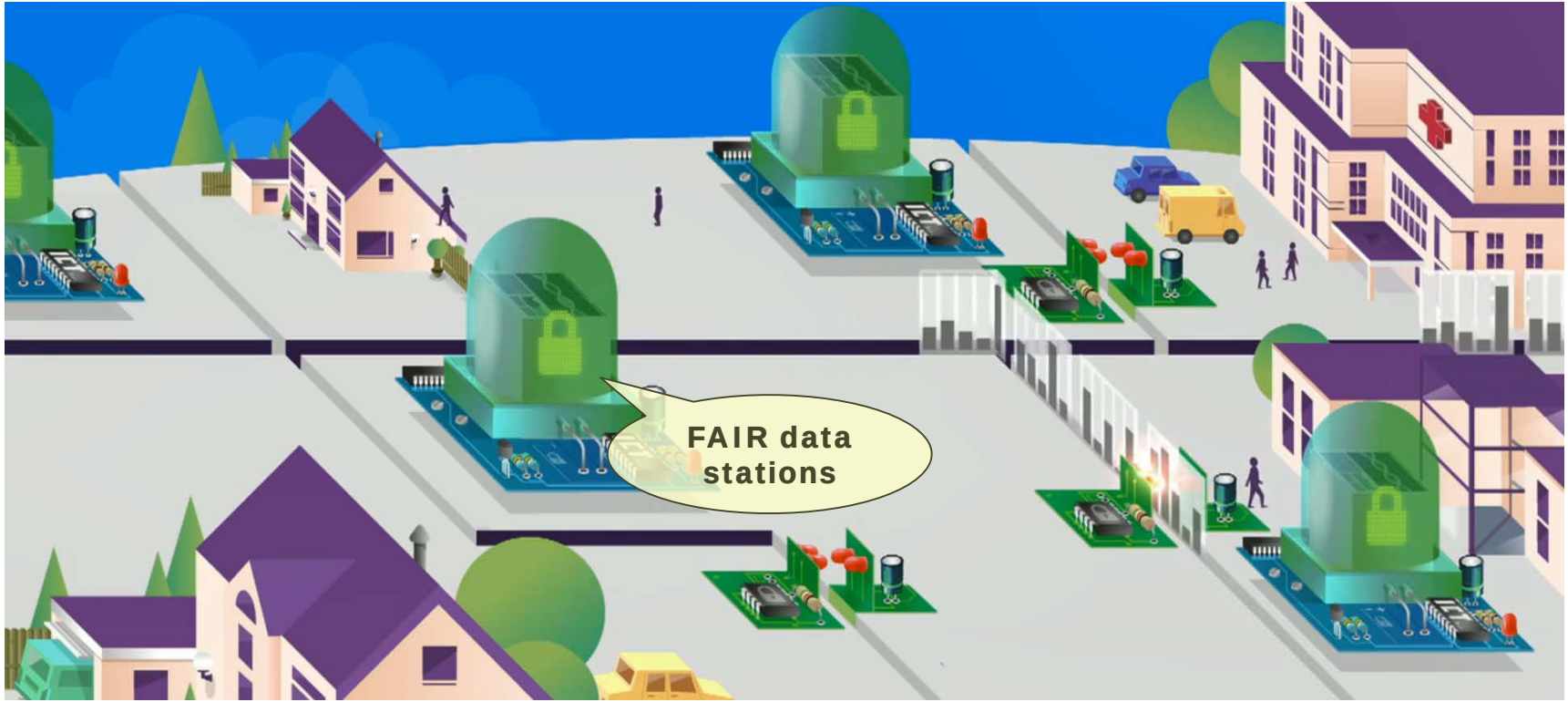

Ontobee
Home
Introduction
Statistics
SPARQL
Ontobeep
Tutorial
FAQs
References
Links
Contact
Acknowledge
News
human phenotype ontology
Keywords:
Search terms
Class: Downslanted palpebral fissures
Term IRI: http://purl.obolibrary.org/obo/HP_0000494
Definition: The palpebral fissure inclination is more 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

Personal Health Train concept

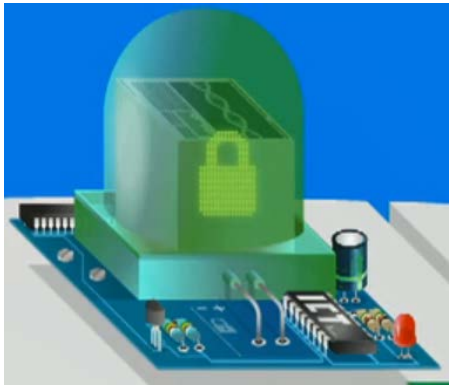
Personal Health Train video:

https://youtu.be/mktAtHmy-FM

Ecosystem of communicating FAIR data stations



FAIR data station



Contains **data** *at the source*

not limited to a common or minimal set

F: Exposes **description** *to be findable*

A: **Controls** *access*

I: Content is **prepared for** *analysis across FAIR data stations*

R: Content is **richly described** for efficient and correct *reuse*

Independently FAIR rare disease data stations

VASCA CDEs

Rett syndrome

DMD pilot
LUMC (Leiden)

DMD pilot UMCL
(Ljubljana)

Hevas

Rizzoli OI pilot

Independently FAIR rare disease data stations


VASCA CDEs


LUMC DMD


LUMC DMD


Hevas


Rett syndrome


orphanet


Open PHACTS

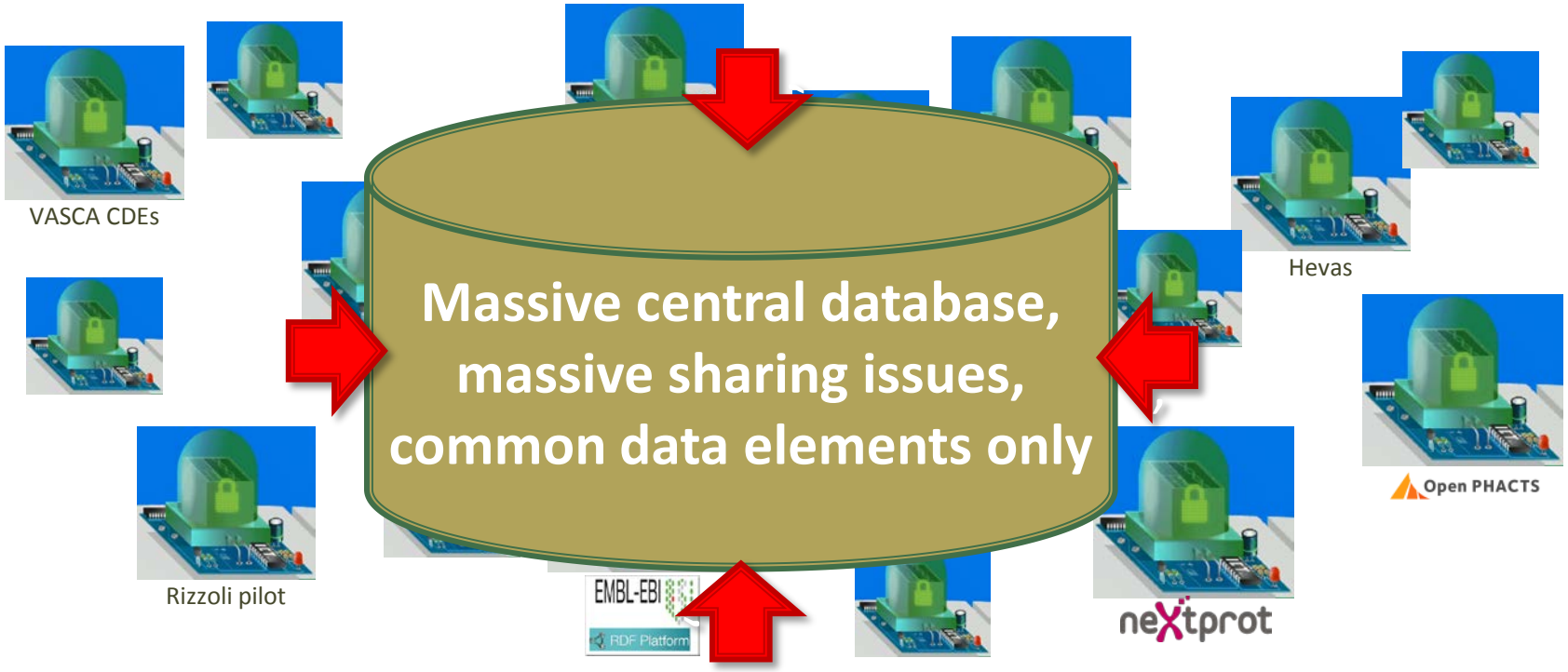

Rizzoli pilot


EMBL-EBI
RDF Platform

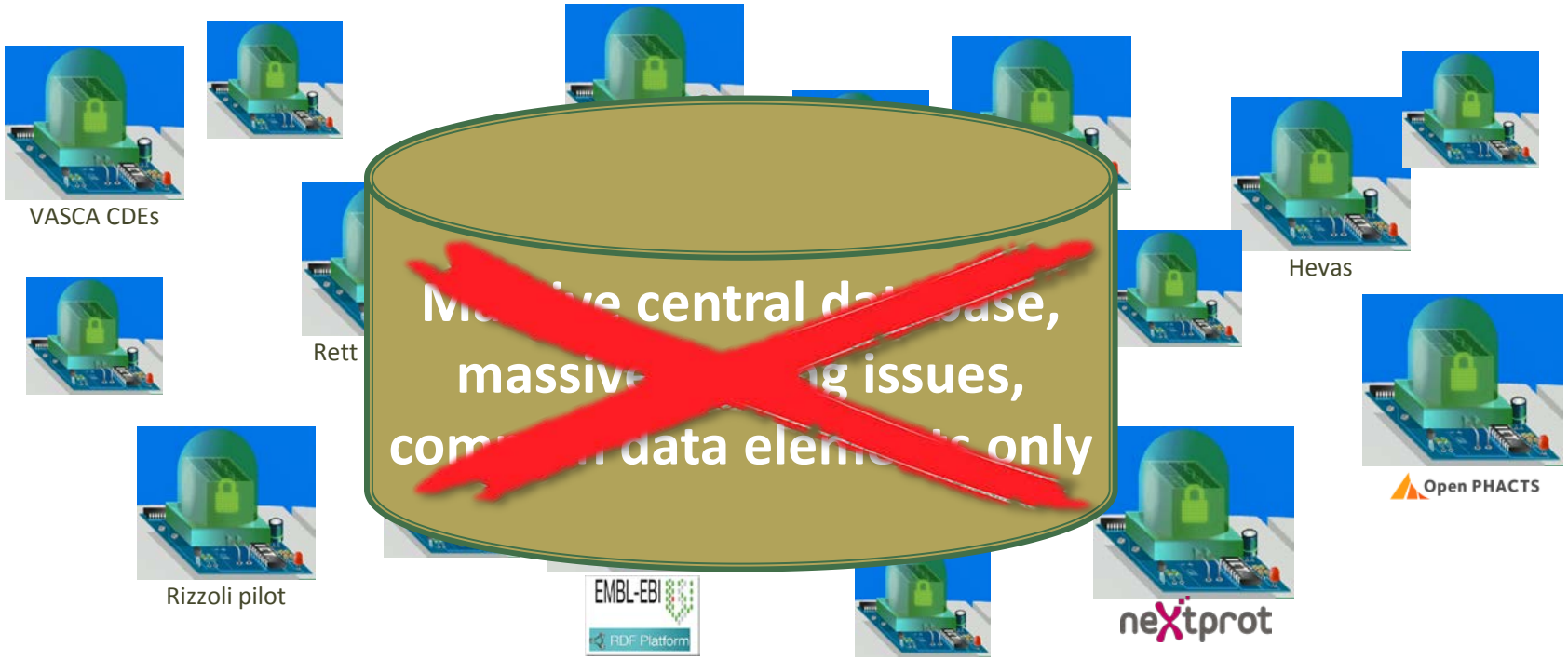

neXtprot

RD-Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Independently FAIR rare disease data stations

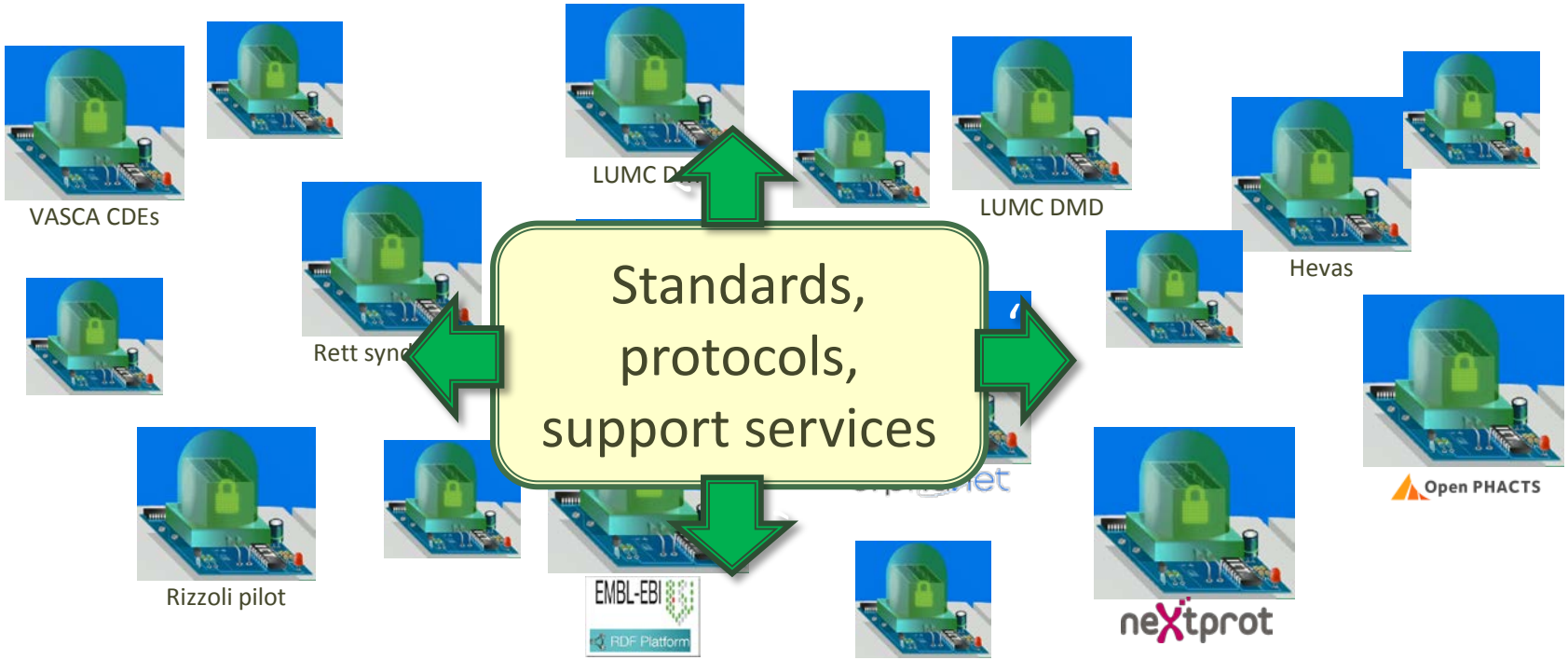


Independently FAIR rare disease data stations

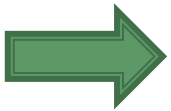


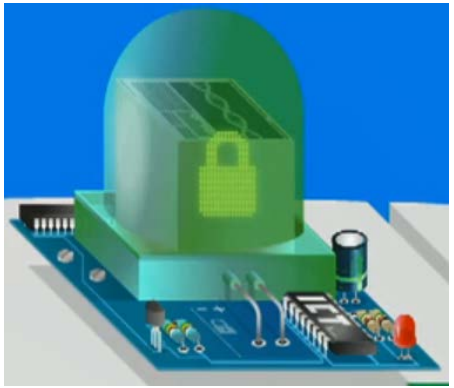
~~Massive central database, massive ... issues, ... data elements only~~

Independently FAIR rare disease data stations



FAIR data station





Your FAIR data station

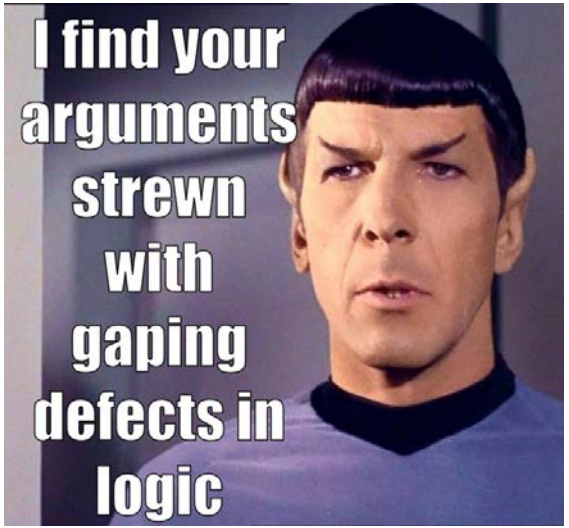
Machine readable?

or how do we help computers to help us?

elixir

RD Connect

What is ‘machine readable’



Mr Spock
Famous Star Trek character

What is ‘machine readable’

Computers are like

Baby Spocks

- know nothing
- can learn a lot
- everything has to be 100% explicit, logical and unambiguous

MutDnaA1	Finding_molecular genetic	nucleotide	c.
c.1532T>C	UBE3Adel	631+1 G>T	386dupC
c.1233_1237del	PAH deletion		2314G>A

Patient (mock)	GenA1	MutDnaA1	GenA2	MutDnaA2
W1	WFS1	c.1532T>C	WFS1	c.1532T>C
		c.1233_1237		c.1233_1237
W2	WFS1	del	WFS1	del

Some text	**Some different text**	**Some word**	**Some text**
Some text	Some text	Some text	Some text
Some other text	Some more text		Some text

Some text	**Some text**	**Some text**	**Some text**	**Some text**
Some text	Some text	Some text	Some text	Some text
Some other text	Some more text	Some more text	Some different text	Some more text

Rare disease data linkage plan

your DATA

Knowledge exchange

Source data steward(s)
FAIR data steward(s)
Domain expert(s)

FAIR

your DATA

Linkable

FAIRification process

1. Define driving user questions(s)

2. Pre-FAIRification analysis

3. Define semantic model

4. Transform data records

5. Define metadata

6. Deploy FAIR data point

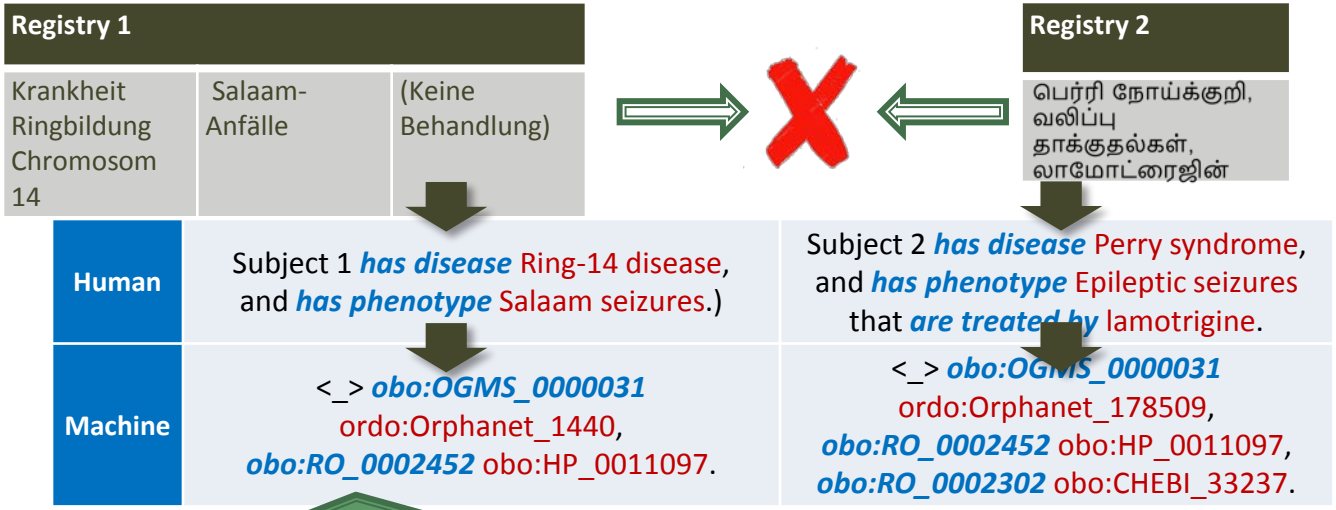
7. Query interface

Making data machine readable

Registry 1		
Krankheit Ringbildung Chromosom 14	Salaam-Anfälle	(Keine Behandlung)

Registry 2
பெர்ரி நோய்க்குறி, வலிப்பு தாக்குதல்கள், லாமோட்ரைஜின்

Making data machine readable



Machine readable data and metadata (data about data)

- Defined in 'ontologies' (for humans and computers)
- **Globally** understood (analogues to 'English' for science)
- Explicit, unambiguous, logical, **linkable**

human phenotype ontology

Keywords: Search terms

Class: Epileptic spasms

Term IRI: http://purl.obolibrary.org/obo/HP_0011097

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. [database_cross_reference: HPO:jalbers][database_cross_reference: HPO:jalbers]

Annotations

- **database_cross_reference:** UMLS:C0037769; UMLS:C1527366; SNOMEDCT_US:28055006; MSH:D013036
- **has_exact_synonym:** Salaam convulsions; Salaam seizures
- **has_obo_namespace:** human_phenotype
- **has_related_synonym:** West syndrome
- **http://www.geneontology.org/formats/oboInOwl#created_by:** peter
- **http://www.geneontology.org/formats/oboInOwl#creation_date:** 2011-05-04T01:56:31Z
- **http://www.geneontology.org/formats/oboInOwl#id:** HP:0011097
- **http://www.w3.org/2000/01/rdf-schema#comment:** The maximum age of onset is between 3 and 12 months, the peak being at 6 months. However, spasms may start from birth, or appear long after the age of 12 months, including into adulthood. Infantile spasms represent a specific type of seizure seen in an epilepsy syndrome of infancy and childhood known as West Syndrome. West Syndrome is characterized by infantile spasms, developmental regression, and hypsarrhythmia (as demonstrated by electroencephalography).

Class Hierarchy

Thing
+ Phenotype
+ abnormal phenotype
+ Phenotypic abnorma
+ Abnormality of th
+ Abnormality o
+ Seizures
+ Status epilepticus
+ Generalized seizures
+ Febrile seizures
+ Focal seizures
+ Symptomatic seizures
+ Dialeptic seizures
- Multifocal seizures
- Nocturnal seizures
- Epileptic spasms

Machine readable

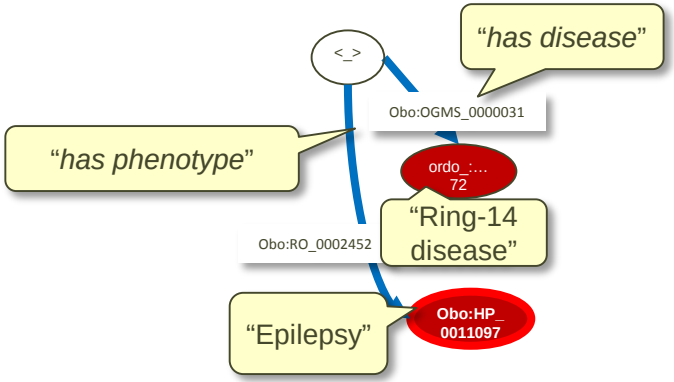
Human readable

Term IRI: http://purl.obolibrary.org/obo/HP_0011097

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

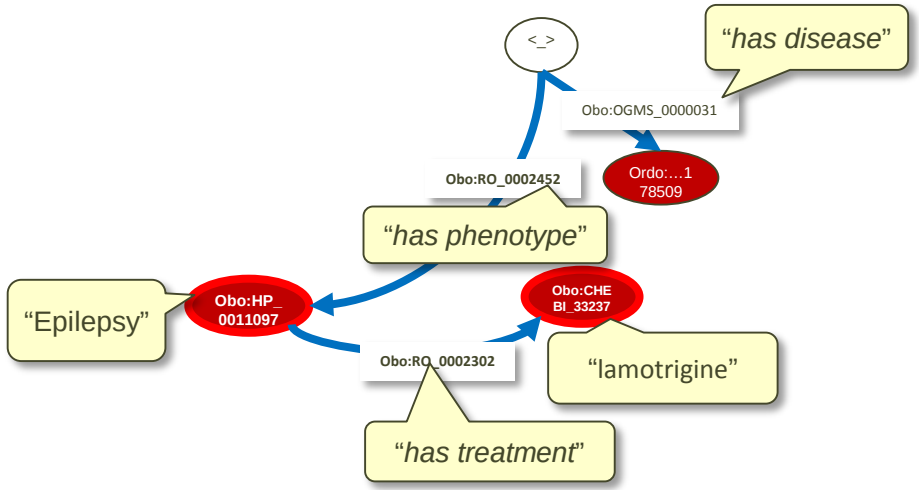
Registry 1		
Krankheit Ringbildung Chromosom 14	Salaam-Anfälle	(Keine Behandlung)

Machine	<_> ***obo:OGMS_0000031*** ordo:Orphanet_1440, ***obo:RO_0002452*** obo:HP_0011097.



"Registration at the source"
+
"Machine readable at the source"

Registry 2	Machine
பெர்ரி நோய்க்குறி, வலிப்பு தாக்குதல்கள், லாமோட்ரைஜின்	<_> ***obo:OGMS_0000031*** ordo:Orphanet_178509, ***obo:RO_0002452*** obo:HP_0011097, ***obo:RO_0002302*** obo:CHEBI_33237.



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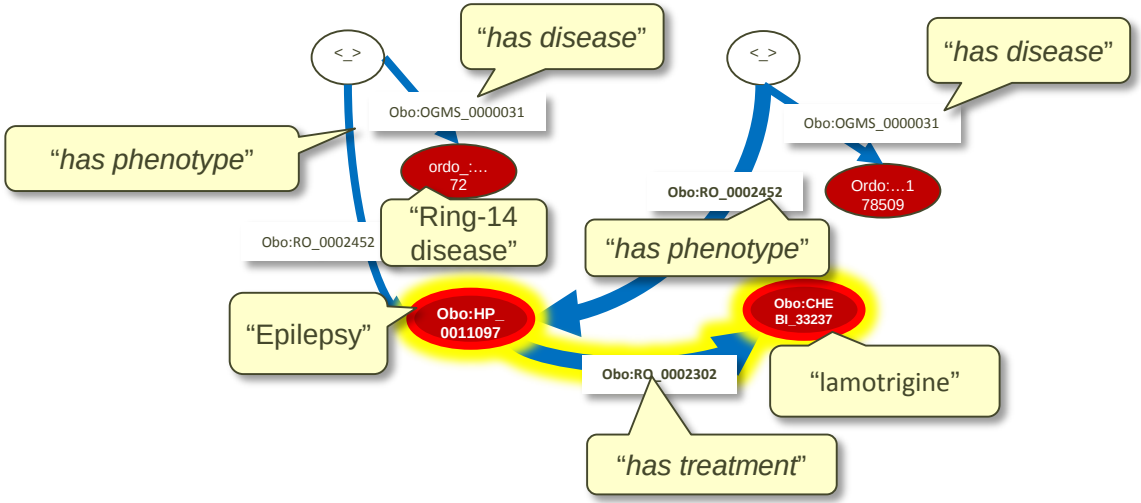


Rare disease data linkage plan for patient/disease registries

Registry software providers

Rare disease FAIR data experts & engineers

Rare disease data stewards

FAIR software engineers

LUMC FAIR data stewardship

LUMC ICT

LUMC librarians and Data Managers

LUMC FAIR data stewards

FAIR software engineers

Conclusion

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

FAIR is

- efficient and correct computer analysis across rare disease data sources
- being clear & explicit about what data elements mean
- encoding data with a globally understood computer language

Conclusion

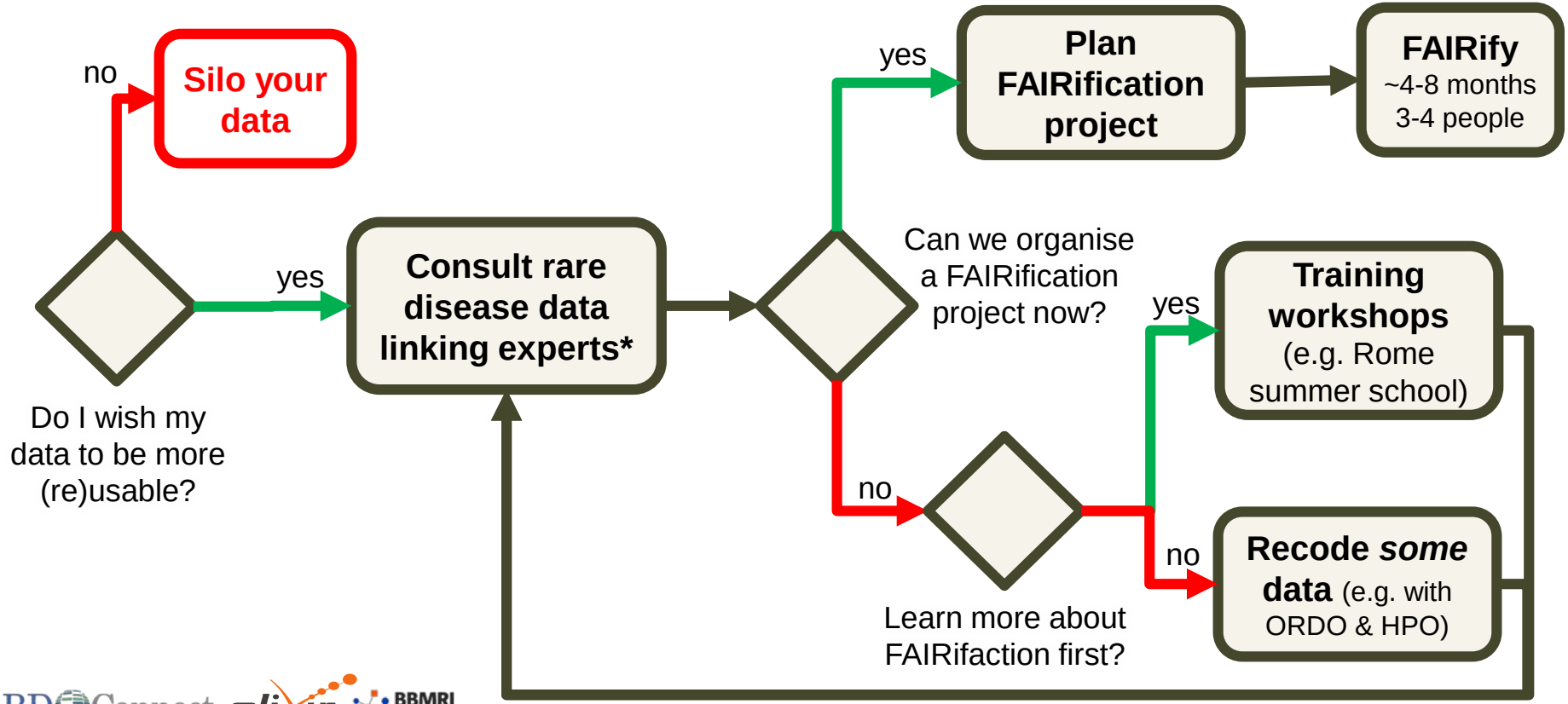
"Now! *That* should clear up a few things around here!"

FAIR is

- making Baby Spock happy

Support for FAIR

Adopting FAIR principles



* Contact: fair-rd-info@elixir-europe.org or the RD-Connect community contact

FAIR 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>

https://goo.gl/bJqH1d

Supporting infrastructure 1: you!

› 300 HOSPITALS

› 900 HEALTHCARE UNITS

THOUSANDS OF PATIENTS HELPED BY 2020

CROSS-BORDER HEALTHCARE DIRECTIVE

ERNs are being set up under the 2011 Directive on patients' rights in cross-border healthcare. This Directive also makes it easier for patients to access information on healthcare and thus increase their treatment options.

EURORDIS RARE DISEASES EUROPE

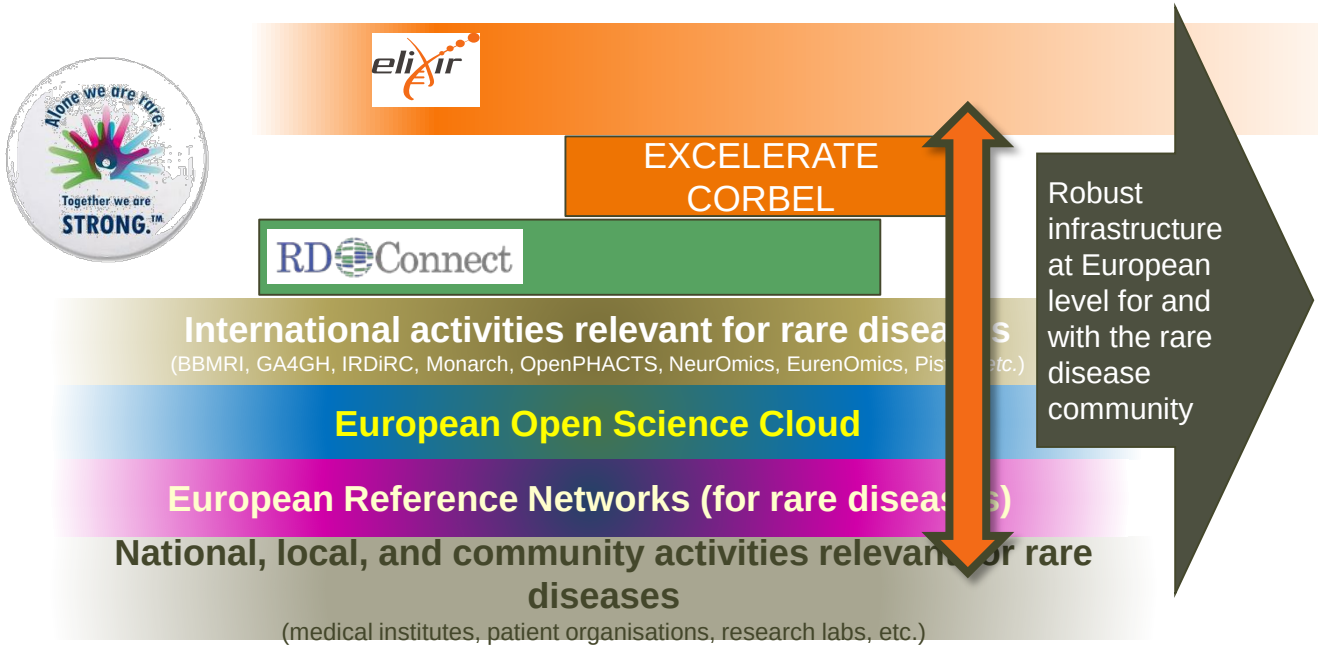
NORD® National Organization for Rare Disorders

CORD Canadian Organization for Rare Disorders

RD Connect

RD Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Infrastructure 2: collaborating infrastructures



RD Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Infrastructure 3: ELIXIR platforms, ELIXIR nodes

Platforms

ELIXIR's activities are divided into five areas called 'Platforms'. These are Data, Tools, Interoperability, Compute and Training. The Platforms are managed by Platform leaders and the work is carried out by groups within the Platforms.

Data

Resources to help the integration and sustainability of life science data

Tools

A discovery portal for data and analysis tools

Compute

Storage, compute and authentication/access services

Interoperability

Resources to aid the discovery, integration and analysis of biological data

Training

A discovery portal for data and analysis tools

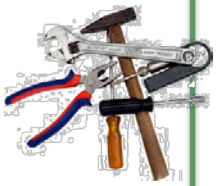
Consultancy: align with cross-national standards

National nodes are service providers

Example: FAIR services by NL

Infrastructure 4: FAIR data services by tools and people

(DTL, UPM, ISS, LUMC, ELIXIR-NL/UK/SL, BBMRI, RD-Connect, FAIR-dICT, …)



Plan

DSW Data Stewardship Wizard

Create

FAIRIFIER

Publish

FAIR DATA POINT

Find

FAIR DATA

Courses

Bring Your Own Data Workshop

FAIR Hackathon

FAIR data projects
(Data stewardship planning)

Rare disease data linkage plan → service

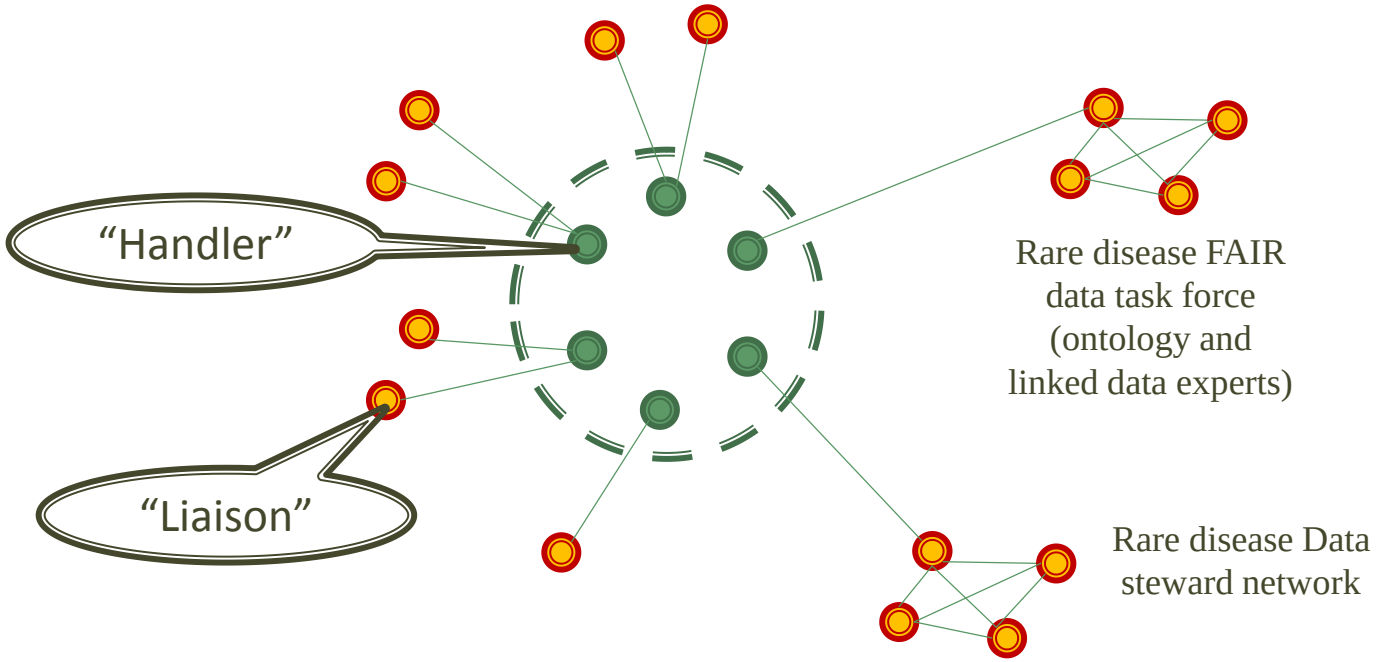
RD-Connect elixir BBMRI Biobanking and Biomolecular Resources Research Infrastructure

Infrastructure 4:
organise our own FAIR service ecosystem

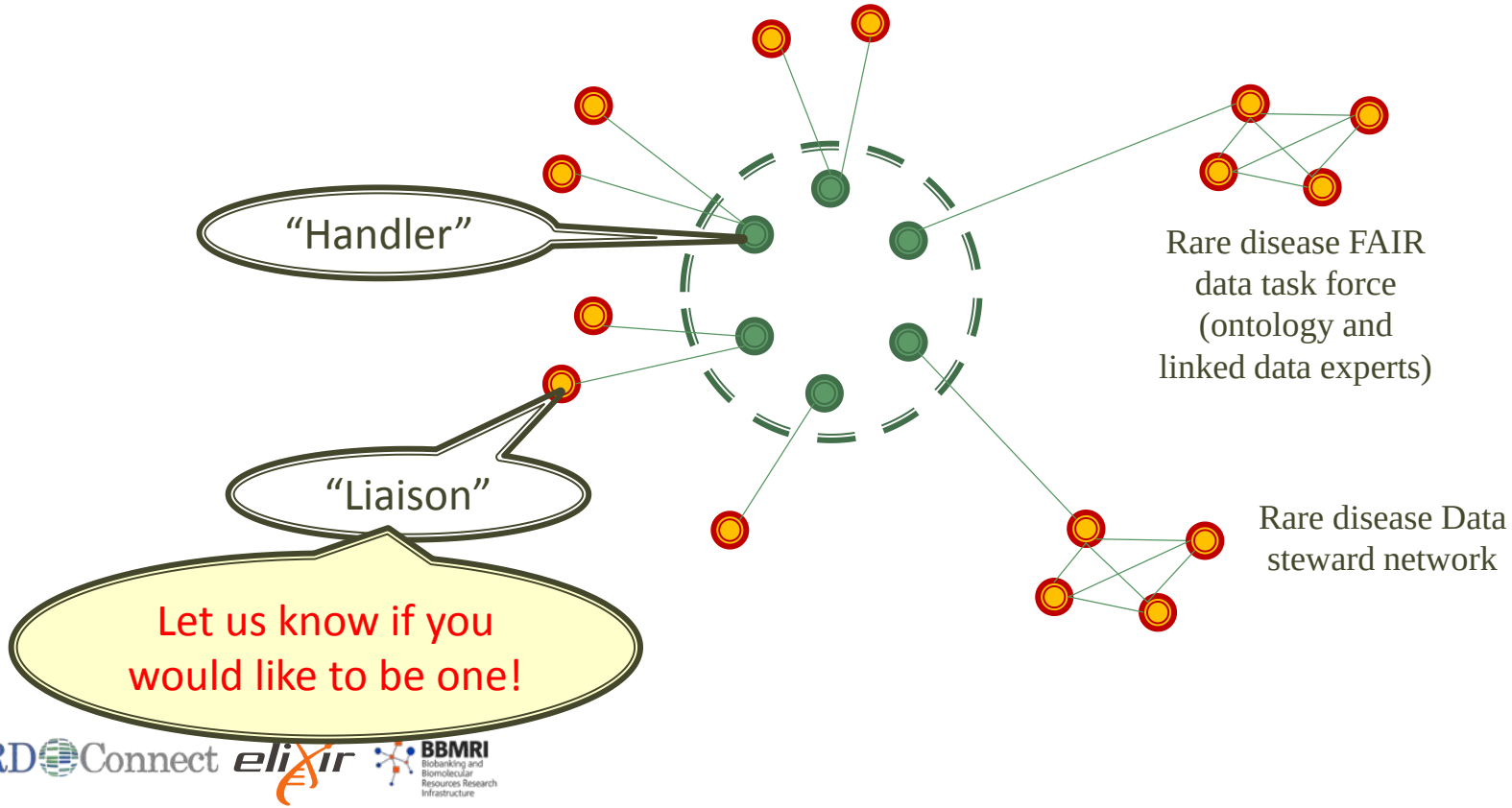
Objective 1

Foster and strategically oversee the expansion of the **adoption of FAIR data principles by the RD community** towards a critical mass of FAIR data, **providing a home for stakeholders** seeking advice on FAIR data stewardship and **guiding the transition** of the rare disease community towards understanding, embracing and refining the FAIR Principles and Goals.

Organisation



Organisation



Future (EJP/ELIXIR/…)

- Address FAIR more fully
  - Metadata
  - FAIR metrics
  - Automating ELSI
  - Filling gaps for analytics
- Scaling up
  - Data steward network
  - ‘more FAIR’ guidelines (Annika Jacobsen *et al.*)
  - Routine tools → FAIR data generating tools (e.g. registry software)
- Approach: real-life cases in interdisciplinary collaboration

SAVE THE DATE

International Summer School on Rare Disease Registries and FAIRification of Data

Mid/End of September 2019

Istituto Superiore di Sanità – Rome (Italy)

Thank you

https://rd-connect.eu/about-rd-connect/become-member/

FAIR data engineering team
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Training material:
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ELIXIR: Chris Evelo and team (Maastricht), Carole Goble and team (Manchester), Ivo Gut and team (CNAG), Helen Parkinson and team (EBI), Olivier Poch and team (Strassbourg), Orphanet, Brane Leskosek and team (Ljubljana)

The BioSemantics group and the Human Genetics Department LUMC