



Update ILIAD registry

an International Library of Intellectual disability and Anomalies of Development

ITHACA online meeting 11 dec 2020

Tech team: Morris Swertz, Fernanda de Andrade, Dieuwke Prins- Joeri van der Velde, supported by MOLGENIS tech team. Legal/contracts: Gert-Jan van de Geijn. Collaborators from GenIDA, OPBG, RETT. Steering: Alessandra Renieri, Alain Verloes, Klea Vyshka



Genomics coordination
center



OUTLINE

- Project overview (reminder)
- State of play
 - Progress
 - Current work
 - Future steps
- Demo (Fernanda)

Project synopsis

GOAL

ILIAD 'meta' registry:
an International Library of
Intellectual disability and Anomalies
of Development

Linking and making visible patient cohorts at the European level in order to follow the natural course of diseases with sufficient patient data

By creating a **central web-based registry** and a network of satellite/client registries linked to the central registry to form the ITHACA **registry federation** (e.g. linking Genida registry)

And adding relevant F.A.I.R. metadata and interfaces linking to ERDRI (the EU RD platform) and BBMRI (the EU biobank platform), in collaboration with EJP-RD, to **enable cross ERN collaboration**

ILIAD Registry

Core ILIAD registry

Common trunk

Universal core subset

EU fields (mandatory)

1. EUPID pseudonym
2. Personal info (2.1 DOB – 2.2 Sex)
3. Patient status (dead 3.1 / DoD 3.2)
4. Care pathway (date of ascertainment)
- 7.1 Consent for recontact
- 7.2 Consent for reuse
- 7.3 Sample availability

EU fields (optionally filled)

5. Disease history (5.1 onset, 5.2 diagnosis)
- 7.4 Sample biobank
8. Disability

Multi-ERN mandatory extra fields

10. ERN, HCP, MD

EU JRC Fields

ILIAD subregistries

Genetically defined core subset

EU fields

- 6.2 Genetic diagnosis

EU mandatory fields (optionally filled)

- 6.1 ORPHA diagnosis

Phenotypically defined core subset

EU fields

- 6.1 ORPHA diagnosis

EU fields (optionally filled)

- 6.2 Genetic diagnosis

Undiagnosed core subset

EU mandatory fields

- 6.3 Undiagnosed (HPO)

EU fields

- (optionally filled)
6.2 Generic ORPHA diagnosis

Specific subregistries 1... to n

Specific subset

Mandatory fields

Defined by subregistry

Other fields

(optionally filled)
Defined by subregistry

ERN registries

External registries

CPMS

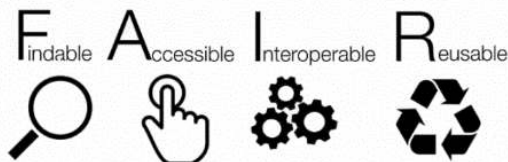
FAIR inter-
operability

Outer
registries

F.A.I.R. data principles

Good data management is the key conduit leading to knowledge discovery and innovation. Applies also to 'non-data'

Co-founder of



To be Findable:

- F1. (meta)data are assigned a globally unique and persistent identifier
- F2. data are described with rich metadata (defined by R1 below)
- F3. metadata clearly and explicitly include the identifier of the data it describes
- F4. (meta)data are registered or indexed in a searchable resource

To be 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

To be Interoperable:

- I1. (meta)data use a formal, accessible, 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

To be 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

Wilkinson et al, 2016, *Scientific Data*: 160018

MOLGENIS PLATFORM

*build using MOLGENIS open
source software platform for
'scientific' data, a web-based
service.*

<https://molgenis.org/>

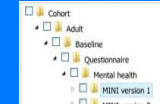
Bulk data

Import your own data and
metadata model
using the EMX
format,
VCF, OBO



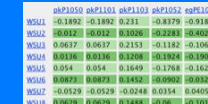
Data request

Find and request (biobank) data
sets and
items



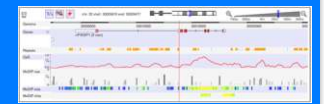
Data explorer

Filter and download for further
analysis



Genome browser

Data sharing and integration DAS
protocol



Model registry

Metadata registry of models for
biobanks and
molecular
data



Annotators

Data integration for diagnostics
and
personalized
medicine



FAIR integration

Using
ontologies to derive
harmonization
rule for data
pooling



R statistics

Use R data api to up/dov
data and integrate
graphics



Pipelines

Large scale computation on
computational
clusters, grids
and clouds



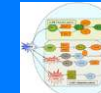
Reports

Tailormade report functionality



App store

Add your own user interfaces and
advanced tools.



Data capture

Questionnaires, forms, importer
to easily
add data to a
running instance



github
SOCIAL CODING



Collaborations

Increase impact by enabling exchange within and between ERNs

By networking 'all' registry developers on FAIR principles

ERN CRANIO



Steering

| | |
|-----------------------|--|
| Morris Swertz (Chair) | Head of Genomics Coordination Center, UMCG |
| Alain Verloes | ERN ITHACA Coordinator, APHP |
| Sylvia Huisman | Department of Paediatrics, Amsterdam UMC |
| Alessandra Renieri | Director of Medical Genetics Unit, AOUS |
| Jill Clayton-Smith | Professor in Medical Genetics, University of Manchester |
| David Koolen | Clinical Geneticist Radboud UMC |
| Carole Herman | ePAG and Patient support |

Progress

CONFIGURED DATA ELEMENTS

Fully Customizable data structure

Model need to be defined can be done in excel(EMX format)

Afterwards changing model with metadata editor

Your Data

| sex | birthyear | disease | consent | since |
|-----|-----------|---------|---------|---------|
| M | 1976 | NA | Y | 1-1-201 |
| F | 1977 | CV | N | 1-1 |
| M | 1985 | COPD | N | 1-1 |
| NA | 1985 | EB | Y | 1-1 |
| F | 1940 | NA | Y | 1-1 |
| F | 2001 | NA | Y | 1-1 |

Your Meta Data

| entity | attribute | dataType | refEntity | required |
|---------|-----------|----------|-----------|----------|
| patient | id | autoid | | true |
| patient | sex | xref | sexValues | |
| patient | birthyear | int | | |
| patient | disease | xref | diseases | |
| patient | consent | boolean | | true |
| patient | since | date | | |

INTER- OPERABILITY

rely on the JRC common data elements, enriched by disease specific elements

| | | | | | |
|--------------------|------|---|--|--|---|
| 5. Disease history | 5.1. | Age at onset | Age at which symptoms/signs first appeared | <ul style="list-style-type: none"> • Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined | |
| | 5.2. | Age at diagnosis | Age at which diagnosis was made | <ul style="list-style-type: none"> • Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined | |
| 6 Diagnosis | 6.1. | Diagnosis of the rare disease | Diagnosis retained by the specialised centre | Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code | http://www.orphadata.org/cgi-bin/inc/product1.inc.php |
| | 6.2. | Genetic diagnosis | Genetic diagnosis retained by the specialised centre | International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code | http://www.hgvs.org |
| | 6.3 | Undiagnosed case | How the undiagnosed case is defined | <ul style="list-style-type: none"> • Phenotype (HPO) • Genotype (HGVS) | |
| 7. Research | 7.1. | Agreement to be contacted for research purposes | Patient's permission exists for being contacted for research purposes | <ul style="list-style-type: none"> • YES • NO | |
| | 7.2. | Consent to the reuse of data | Patient's consent exists for his/her data to be reused for other research purposes | <ul style="list-style-type: none"> • YES • NO | |
| | 7.3. | Biological sample | Patient's biological sample available for research | <ul style="list-style-type: none"> • YES • NO | If YES answer question 7.4 |
| | 7.4. | Link to a biobank | Biological sample stored in a biobank | <ul style="list-style-type: none"> • YES (if appropriate use link) • NO | https://directory.bbmri-eric.eu |
| 8.Disability | 8.1. | Classification of functioning/disability | Patient's disability profile according to International Classification of Functioning and Disability (ICF) | <ul style="list-style-type: none"> • Disability profile / Score | http://www.who.int/classifications/icf/whodasii/en/ |

277 data elements defined



Subjects

description: Collection of all pseudonymized patients

- *Attributes:* **Period of first symptom onset:** categorical
- **Other Remarks:** text
- **Clinical diagnosis:** compound
- **Diagnosis Coding System:** *mref(*ICD-10: International Classification of Diseases for Oncology*)*
- **OMIM Code:** *mref(*OMIM (Online Mendelian Inheritance in Man)*)*
- **Orphanet nomenclature:** *mref(*Orphanet Rare Diseases Database*)*
- **Phenotypic Abnormality (HPO):** *mref(*hpo*)*
- **Biologic diagnosis:** categorical (Biologic diagnosis)
- **Date of first symptoms onset:** date
- **Age at first symptoms onset:** int
- **Period of current diagnosis:** categorical (onset)
- **Date of current diagnosis:** date
- **Age at diagnosis:** int
- **All codes:** string - *Map all codes to this field (field: codes)*
- **Biologic diagnosis:** compound
- **Link to variant:** *mref(*variant*)* - *(same as point 6; G)*
- **Link to Genomic variant:** *mref(*genomeVariant*)*
- **Family History:** compound
- **Other cases in the family?:** categorical (Positive Family History)
- **If Yes – kinship of Carrier:** *xref(*Type of Relative*)*
- **Comorbidities:** compound
- **Other status of diagnosis:** text
- **Diagnosis Coding System:** *mref(*ICD-10: International Classification of Diseases for Oncology*)*
- **International Classification of Diseases for Oncology:** categorical
- **Orphanet nomenclature:** *mref(*Orphanet Rare Diseases Database*)*
- **Other comorbidities:** text
- **Date of diagnosis:** date
- **Status of diagnosis:** categorical (Diagnosis status)
- **Is the patient following any other medication?:** boolean
- **Medication:** compound
- **Is the patient receiving medication for his/her rare disease?:** boolean
- **Medication for Rare Disease Description:** text
- **Active Ingredient (ATC):** *mref(*Anatomical Therapeutic Chemical Classification System*)*
- **If not ATC:** text
- **Start date medication:** date
- **End date medication:** date
- **Active Ingredient (ATC):** *mref(*Anatomical Therapeutic Chemical Classification System*)*
- **If not ATC:** text
- **General comments:** text
- **Consent:** compound
- **Pseudonym:** compound
- **EUPID/ Person ID:** string
- **Patient enrolment:** compound
- **Patient ID:** string
- **Date enrolled:** date
- **Consent for Care:** bool
- **Date of consent:** date
- **Consent for ERN databases/Registries/research:** bool
- **Date of consent:** date
- **Consent to be contacted for research:** bool
- **Date of consent:** date
- **Retraction to be contacted for research:** bool
- **Consent for usage of biobank material:** bool
- **Date of consent:** date
- **Retraction for biobank use:** bool
- **Date of retraction:** date
- **Retraction for reuse:** bool
- **Date of retraction:** date
- **Last name:** string
- **Date of retraction:** date
- **Research:** compound
- **Patient's biological sample available for research:** boolean
- **Other; what patient Data is available:** categorical
- **Biobank:** *mref(*Biobanks*)*
- **Identifying data:** compound
- **First name:** string
- **Record Label:** string - *This Record Label is displayed in the record*
- **Patient Status:** compound
- **Patient's Status:** categorical (Patient status)
- **Date of death:** date
- **Care pathway:** compound
- **First Contact date:** date - *Date first contact with specialist*
- **Consultation Request:** compound

BULK Data ingest

Enable upload of datasets.

1 Upload file

2 Options

3 Packages

4 Validation

5 Result

| Entities | |
|------------------------|------------|
| Name | Importable |
| root_hospital_cities | Yes |
| persons | Yes |
| root_hospital_users | Yes |
| root_hospital_patients | Yes |

| Entity fields | |
|------------------------|--|
| Name | Detected |
| root_hospital_cities | cityName |
| root_hospital_users | userName, active, displayName, firstName, lastName |
| root_hospital_patients | displayName, firstName, lastName, birthdate, birthplace, disease |

← Previous

Restart

Next →

PROGRAM ACCESS

To connect
HCPs

To automate
data federation

Interoperability

Swagger specification

Data API

REST api v1

REST api v2

Files api

Import api

Permission api

[Python-api client](#)

R-api client

Beacon api

FAIR api

RSQL operators

For developers

Developing MOLGENIS

Developing frontend in MOLGENIS

Developing Apps in MOLGENIS

Using an IDE (IntelliJ)

☰ [EDIT THIS PAGE](#)

Python-api client

The MOLGENIS python client API allows you to retrieve, create, update and delete entities from within python.

You can install the Python REST Client as a package with `pip` :

```
pip install molgenis-py-client
```

Now you can create a python script. To get started, you should import the Python client, connect to a molgenis server and login:

```
import molgenis.client
session = molgenis.client.Session("https://molgenis.mydomain.example/api/")
session.login("username", "password")
```

Always put the import and `molgenis.Session` in your script to make the api work.

Overview example

```
import molgenis.client
session = molgenis.client.Session("https://molgenis.mydomain.example/api/")
session.login("username", "password")
my_table = session.get("package_entityName")
print(my_table)
```

<https://molgenis.gitbooks.io/molgenis/>

CONTROLLED ACCESS

In permissions system there are different roles (viewer, editor and data manager).

Plugin Permissions

Package Permissions

Entity Class Permissions

Row-Level Security

RolesUsers

Select Role: NEXT_SAMPLE_REGISTRATION_ED

Package permissions determine the permission on entity types within the package and its child packages. Additionally WRITEMETA permission on a package means that the user can create packages and Entity Types in this package and its children. They do **not** effect permissions on the package metadata itself.

| Package | Edit metadata | Edit | View | Count | View metadata | None |
|--------------------------|-----------------------|----------------------------------|-----------------------|-----------------------|-----------------------|----------------------------------|
| sys_map | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_negotiator | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| next_registration | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_idx | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_job | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_ont | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_mail | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_md | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| next_sample_registration | <input type="radio"/> | <input checked="" type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> |
| sys_sec | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_dec | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_scr | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_set | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |
| sys_sec_oidc | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input type="radio"/> | <input checked="" type="radio"/> |

Current activities

SUITABLE USER INTERFACES TO SEARCH



the international database of dystrophic epidermolysis bullosa patients
and COL7A1 mutations

Home

Patients

Mutations

References

Background

News

Feedback

Search data values

✕

Q

Data item filters

Wizard

Data item selection

Select all

Deselect all

☒ Patient ID

☒ Phenotype

☒ Mutations

☐ PubMed ID

☐ Reference

☐ Material stored?

☒ Phenotypes

☒ Blistering

☒ Location

☒ Hands

☒ Feet

☒ Arms

☒ Legs

Data

| Patient ID | Phenotype | Mutations | Blistering | Location | Hands | Feet | Arms | Legs |
|------------|--------------------------|------------------------|------------|-------------|---------|---------|---------|------|
| P166 | RDEB, generalized other | c.6217-3C>A | yes | localized | unknown | unknown | unknown | unk |
| P167 | RDEB, severe generalized | c.5797C>T | yes | generalized | yes | unknown | unknown | unk |
| P18 | RDEB, severe generalized | c.2387G>A | yes | generalized | unknown | unknown | unknown | unk |
| P214 | RDEB, generalized other | c.448G>A,c.682+1G>A | yes | generalized | yes | unknown | yes | yes |
| P218 | RDEB, severe generalized | c.8226+1del | yes | generalized | unknown | unknown | unknown | unk |
| P219 | RDEB, severe generalized | c.4603G>T,c.8226+1del | yes | generalized | unknown | unknown | unknown | unk |
| P22 | RDEB, generalized other | c.2699G>A,c.7237G>A | | | | | | |
| P223 | RDEB, generalized other | c.846+1G>A,c.7705G>A | | | | | | |
| P224 | RDEB, generalized other | c.682+1G>A,c.6394G>A | | | | | | |
| P225 | RDEB, generalized other | c.682+1G>A,c.2441-2A>G | | | | | | |
| P226 | RDEB, generalized other | c.2587+1G>A,c.5047C>T | | | | | | |
| P227 | RDEB, generalized other | c.425A>G,c.7068+5G>A | | | | | | |
| P228 | RDEB, severe generalized | c.682+1G>A | | | | | | |

Genome Browser

Human GRCh37/hg19 3:48,619,738..48,619,838

Genome

Genes

Repeats

Conservation

Mutations

ThousandGenomes

ExAC

SUITABLE USER INTERFACES FOR DATA ENTRY

Example from [Genturis](#) registry:
Easily create new entries
(subjects)

Create Subjects

EUPID/ Person ID *

Family ID

Search for a Value

+

consent

Consent for registry (Y/N) *

YesNo

Consent for research (Y/N) *

YesNo

Consent for care (Y/N) *

YesNo

Informed consent for other studies (Y/N) *

YesNo

If yes, specify for which study

Demographics

Year of birth *

Number

Sex at birth *

FemaleMaleUnknown

Index status *

Index

consent

Demographics

Disease-related

Clinical symptoms and physical findings (HPO) divided in subcategories

Information for affected patients

Diagnostics

Patient re-contact

Prototype dashboard

Input needed

All registered patients

Craniosynostosis

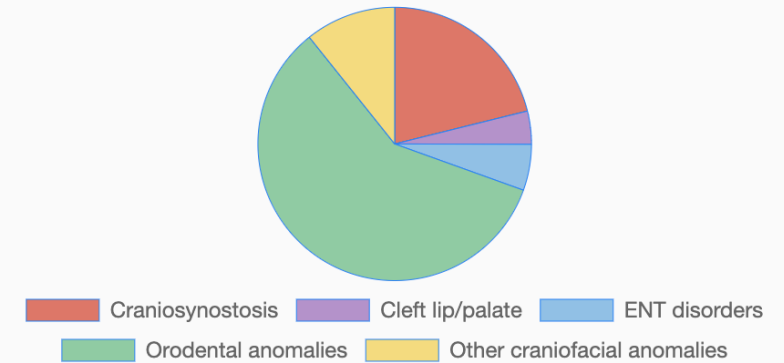
Cleft lip/palate

ENT disorders

Oro dental anomalies

Other craniofacial
anomalies

Registered Patients

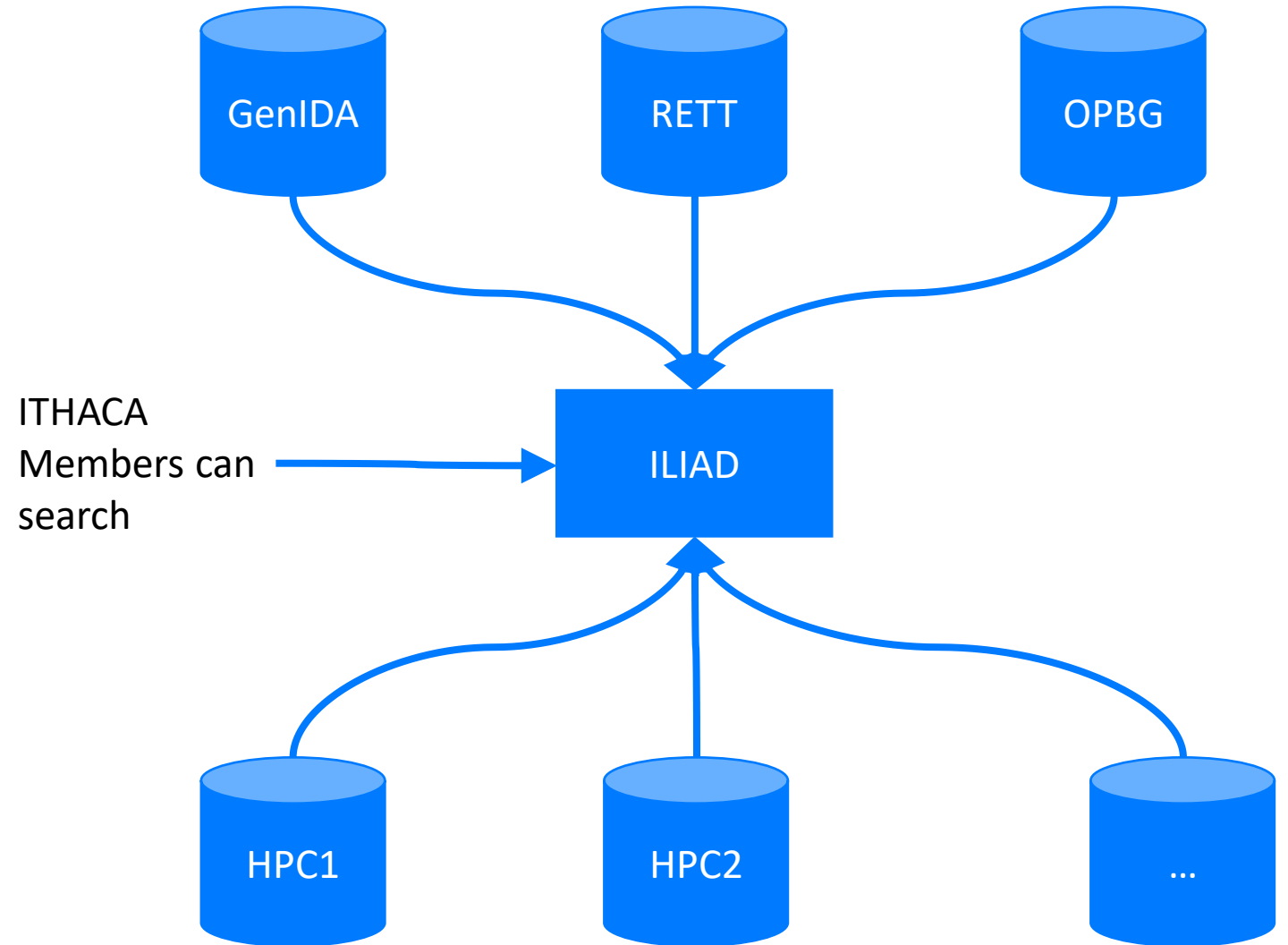


Federating registries and HCPs

Technical connections

Legal connections

Disease specific WGs



EJP FAIR data stewards

Joeri vd Velde from UMCG with 8 others to synergize between ERN registry developers, and with EJP RD developments.



| Tool/ Standard name | Implemented |
|-----------------------------|-------------|
| ORDO | 10 |
| Common Data Elements JRC | 9 |
| ERDRI.dor | 8 |
| ICD-10 | 8 |
| HPO | 8 |
| CSV | 7 |
| HGVS | 6 |
| HGNC | 6 |
| ERDRI.mdr | 5 |
| Excel Reports | 5 |
| OMIM | 4 |
| Molgenis | 4 |
| MOLGENIS metadata structure | 4 |
| EUPID | 3 |
| RDF/XML | 3 |
| RedCap | 3 |
| Own Registry System | 3 |
| Epic EHR | 3 |
| CDE Semantic Model | 2 |
| ICD-11 | 2 |

| Tool/ Standard name | Need expert |
|--------------------------|-------------|
| CDE Semantic Model | 8 |
| EUPID | 7 |
| ERDRI.mdr | 6 |
| Common Data Elements JRC | 3 |
| FAIR Data Point | 3 |
| RML | 3 |
| EJPRD Metadata Model | 2 |
| ORDO | 1 |
| ERDRI.dor | 1 |
| RDF/XML | 1 |
| JSON LD | 1 |
| SPARQL | 1 |
| ATC | 1 |
| iCRF Generator | 1 |
| Turtle | 1 |
| ICD-10 | 0 |
| HPO | 0 |
| CSV | 0 |
| HGVS | 0 |
| HGNC | 0 |

Training

+ Add task

need guidance need expert input

- ☐ GENTURIS, SKIN, CRANIO, ITHACA - needs more information on ERDRI.mdr utility / added value



task is a bit vague need expert input

- ☐ GENTURIS, SKIN, CRANIO, ITHACA - needs more information on EJPRD metadata model



need guidance need expert input

- ☐ GENTURIS, SKIN, CRANIO, ITHACA - what is the use of phenopackets for the registry?
need to know from other ERNs experience, and what is the utility?



need guidance task is a bit vague

- ☐ GUARD-HEART - Query Language
They need to know more about "querying".

Community

+ Add task

waiting for answer

- ☐ ERN GUARD-HEART: how other ERNs deal with Diagnosis and Genetic Diagnosis Elements

They wonder how other ERNs collect information from the two data elements (6.1 Diagnosis and 6.2 Genetic Diagnosis).

need guidance waiting for answer

- ☐ Skin: share ERN tools/standards overview

request for sharing the EJP-RD FAIRification stewards overview of standards and tools chosen to build registries across all ERNs

need better resource

- ☐ Skin: share the database templates other ERNs

request to share any database templates across ERNs, in addition to used standards/tools etc.

need guidance task is a bit vague

- ☐ Skin: learn from others to overcome datasharing issues

need guidance

- ☐ EpiCARE - learn from others

Modelling

+ Add task

need expert input

- ☐ EURACAN - CDE for Rare Cancer

This ERN is focused on rare types of cancer. Since cancer is not a RD, but the types are, they are worried that the CDE (or even other cases) might not fit well for them.

need guidance waiting for answer

need expert input

- ☐ EURACAN - Difference between CDE Model and OMOP-CMD

The ERN is planning to use the OMO-CMD model as common data elements. They want to know what would be the interoperability impact when using EJP-RD CDE Model or OMOP-CMD.

need guidance need better resource

need expert input

- ☐ VASCERN - RADICO - plan to collect treatment info, ATC ontology is not free, need alternative. would like a table with free ontologies options

need guidance need expert input

- ☐ ERN GUARD-HEART - Ontology Recommendation for Diagnosis

For 6.2, which ontology (HVGs, HGNC, OMIM) should ERN choose?

Implementation

+ Add task

need guidance waiting for answer

- ☐ MERITA - Data integration

MERITA registry considers offering an Excel import function. Yet they are awaiting for knowing whether a common template would be made available.

! 20/11

need expert input

- ☐ VASCERN - Radico - EUPID match: how can we see the "similar name" patient that is already registered. how do i know which patient it is similar to?

need expert input

- ☐ VASCERN - RADICO - need EUPID specifications. have 250 patients, need bash implementation or automated.

waiting for answer

- ☐ VASCERN - RADICO - would like erdri.mdr import template.



Bruna DosSantosVieira

waiting for answer

- ☐ VASCERN - RADICO - iCDE connector

Legal

+ Add task

need guidance waiting for answer

- ☐ EURACAN - Data transfer agreement

This ERN works with a set of federated databases, and would like to know how others have been dealing with data transfer policies among databases.

need guidance waiting for answer

- ☐ GENTURIS - Legal issues data sharing

GENTURIS wants to know about legal aspects of data sharing, specifically those of software providers (data consumers) and hospitals/institutes (data producers). ERNs are not legal entities, so difficult. Perhaps

need expert input

- ☐ ENROL - EUPID

Legal questions about policy and licenses issues. Need information from experts, if they can clarify how local registries use EUPIDS, so that they can generate pseudonyms and then provide them to the

MN Michael Nitzlader

need expert input

- ☐ RND - Patient consent

They use their own hospital-patient agreement. Is this allowed?

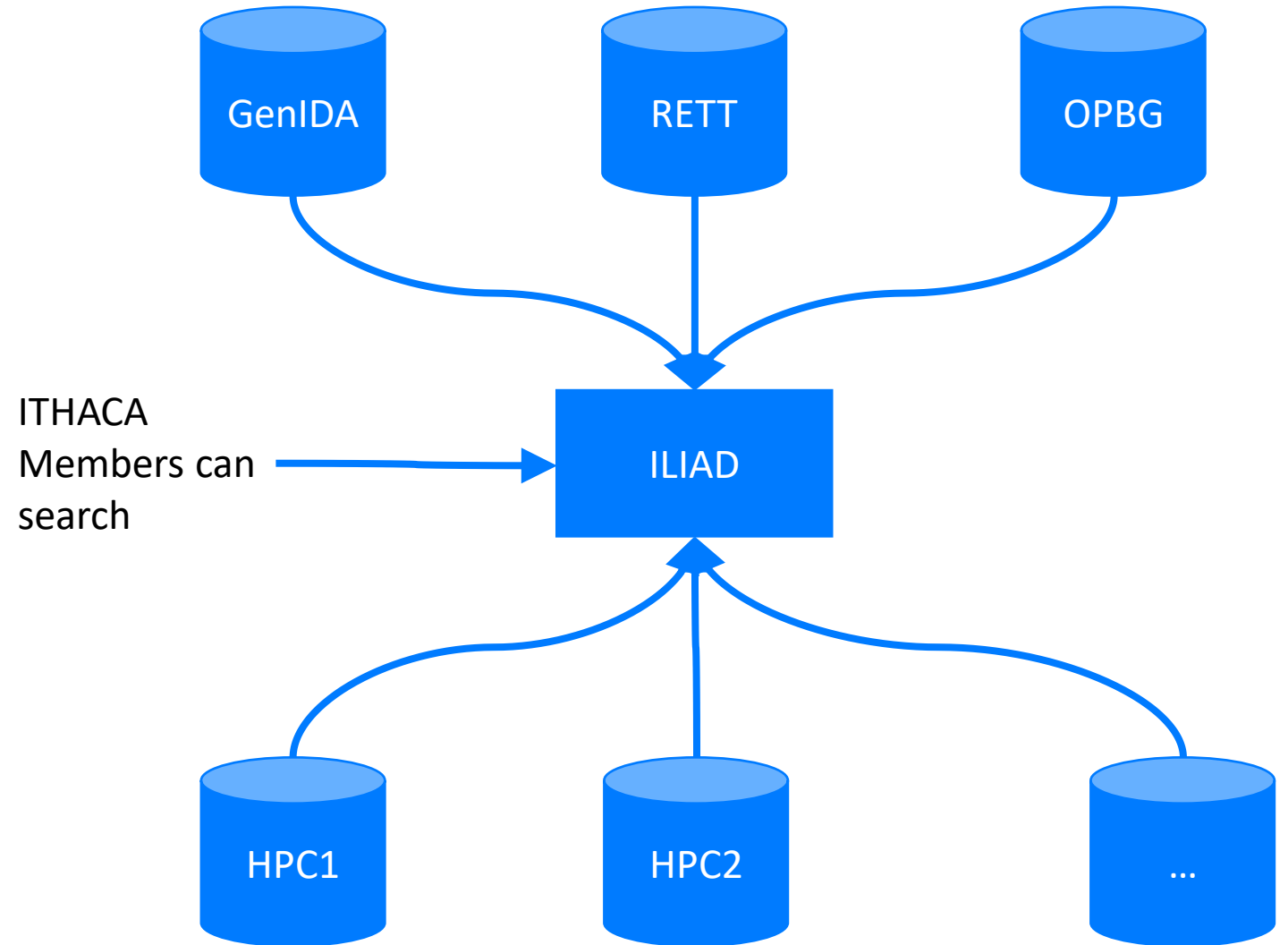
Future steps

Federating registries and HCPs

Technical connections

Legal connections

Disease specific WGs



FINDABILITY

To support findability, the registry will be connected catalogues such as the European Directory of Registries (ERDRI.dor), BBMRI catalogue, RD-connect registry & biobank catalogues.

RDConnect

Sample Catalogue

Register Biobank

Guides

RD-Connect Project

Sample Information

Data

Aggr

Group by

Diagnosis available

Type to search

Materials

Biobank quality marks

Collection quality marks

Collection Types

Data types

Undiagnosed

Healthy subject

Invalid Data

Healthy Control

Ataxia-telangiectasia

Hereditary angioedema type 2

FRAXE intellectual disability

Autosomal dominant spastic paraplegia type 4

Autosomal dominant spastic paraplegia type 17

Female restricted epilepsy with intellectual disability

Northern

Netherlands

European Rare Kidney Disease Registry

In EF

part

Kidney Disease Registry

Go to sample / data negotiation

92 biobanks found

«

<

1

2

3

4

...

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»

ACS Biobank

Collection types: Cohort, Disease specific, Longitudinal

Juridical person: No information

| Collection | Type | Materials | Standards | #Samples |
|--------------------------|--|------------------------------------|-----------|---------------|
| Amsterdam cohort studies | Cohort, Disease specific, Longitudinal | DNA, peripheral blood cells, Serum | | 1000 - 10.000 |
| Amsterdam cohort study 2 | Disease specific, Longitudinal | DNA, peripheral blood cells, Serum | | 1000 - 10.000 |

AGNES Biobank

Collection types: Case-Control, Disease specific

Juridical person: No information

AMC Renal Transplant Biobank

Collection types: Disease specific

Juridical person: No information

| Collection | Type | Materials | Standards | #Samples |
|------------------------------|------------------|---|-----------|------------|
| AMC Renal Transplant Biobank | Disease specific | DNA, Other, Serum, Tissue (paraffin preserved), Urine | | 100 - 1000 |

ARGOS Biobank

Collection types: Disease specific

Juridical person: No information

ARREST Biobank

Collection types: Cohort, Disease specific, Longitudinal

Juridical person: No information

Academic Medical Center Biobank

Collection types: Population-based, Birth cohort, Cohort, Longitudinal, Disease specific, Hospital

Juridical person: AMC

ACCESS

Integrate into existing authentication systems to ensure those accessing have the proper credentials to view sensitive data (e.g. ELIXIR AAI)

Sign in ×

With Google

With SURFconext

OR

Username

Password

Sign in

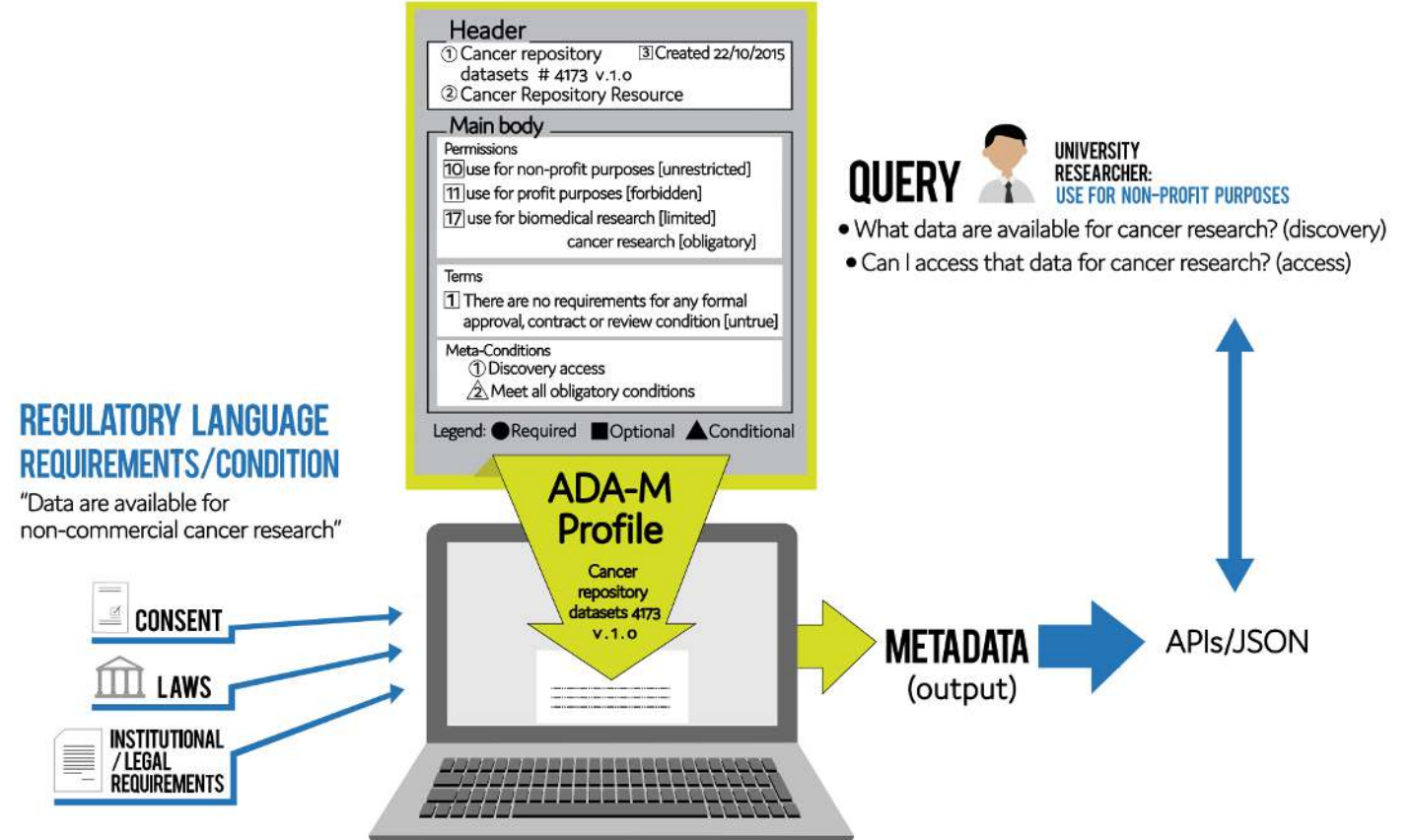
[Forgot password?](#)

Don't have an account? [Sign up](#)

ACCESS (2)

Per project different rules,
different information on access
conditions.

Example of a Basic ADA-M Profile

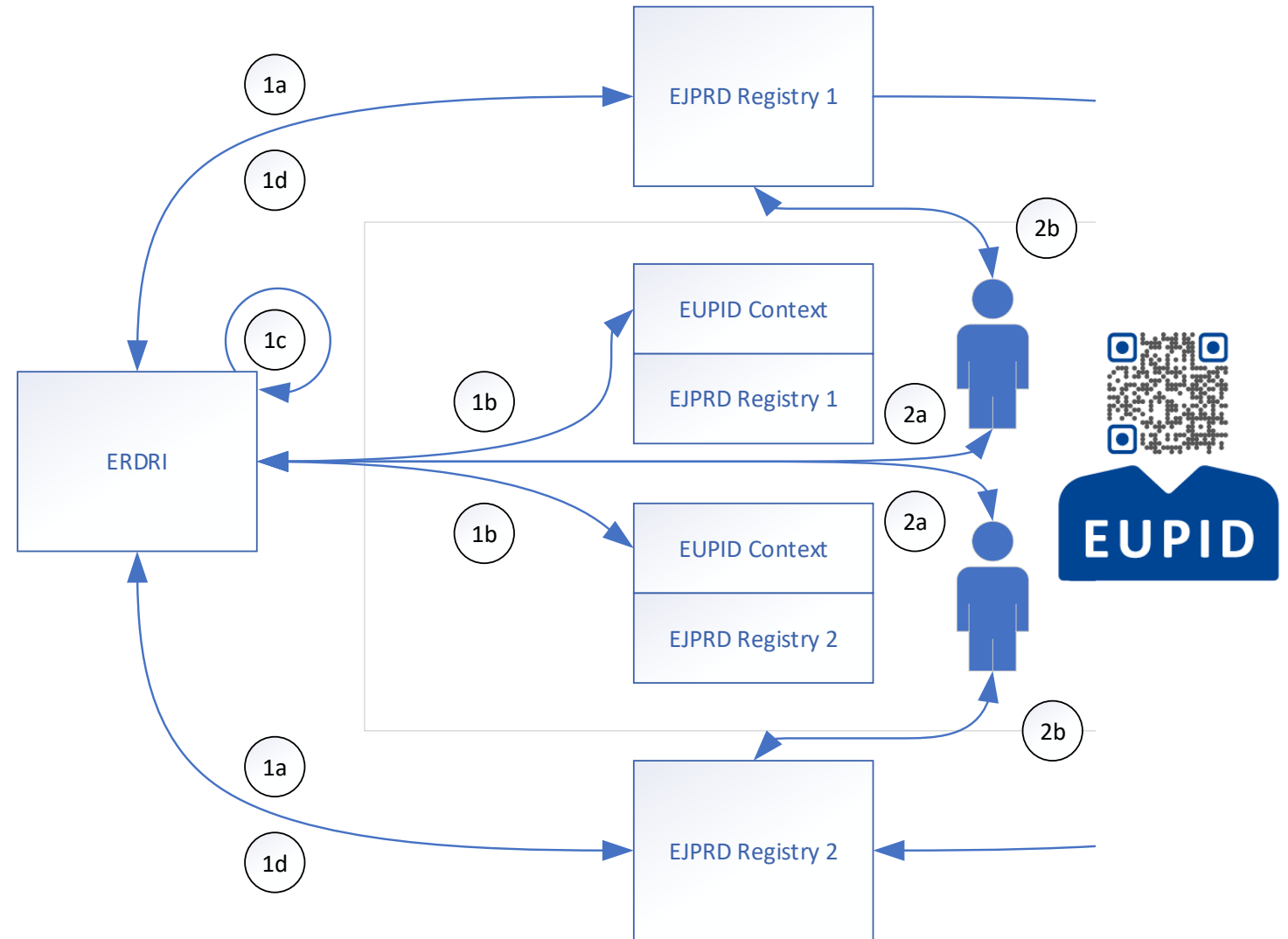


Woolley, J. P., Kirby, E., Leslie, J., Jeanson, F., Cabili, M. N., Rushton, G., ... Brookes, A. J. (2018). Responsible sharing of biomedical data and biospecimens via the "Automatable Discovery and Access Matrix" (ADA-M). *Npj Genomic Medicine*, 3(1), 1–6. <https://doi.org/10.1038/s41525-018-0057-4>

INTER-OPERABILITY

To enable linkage of patients across ERNs we will use the ERDRI Pseudonymisation Tool (EUPID) to allow linking RD patients cohorts

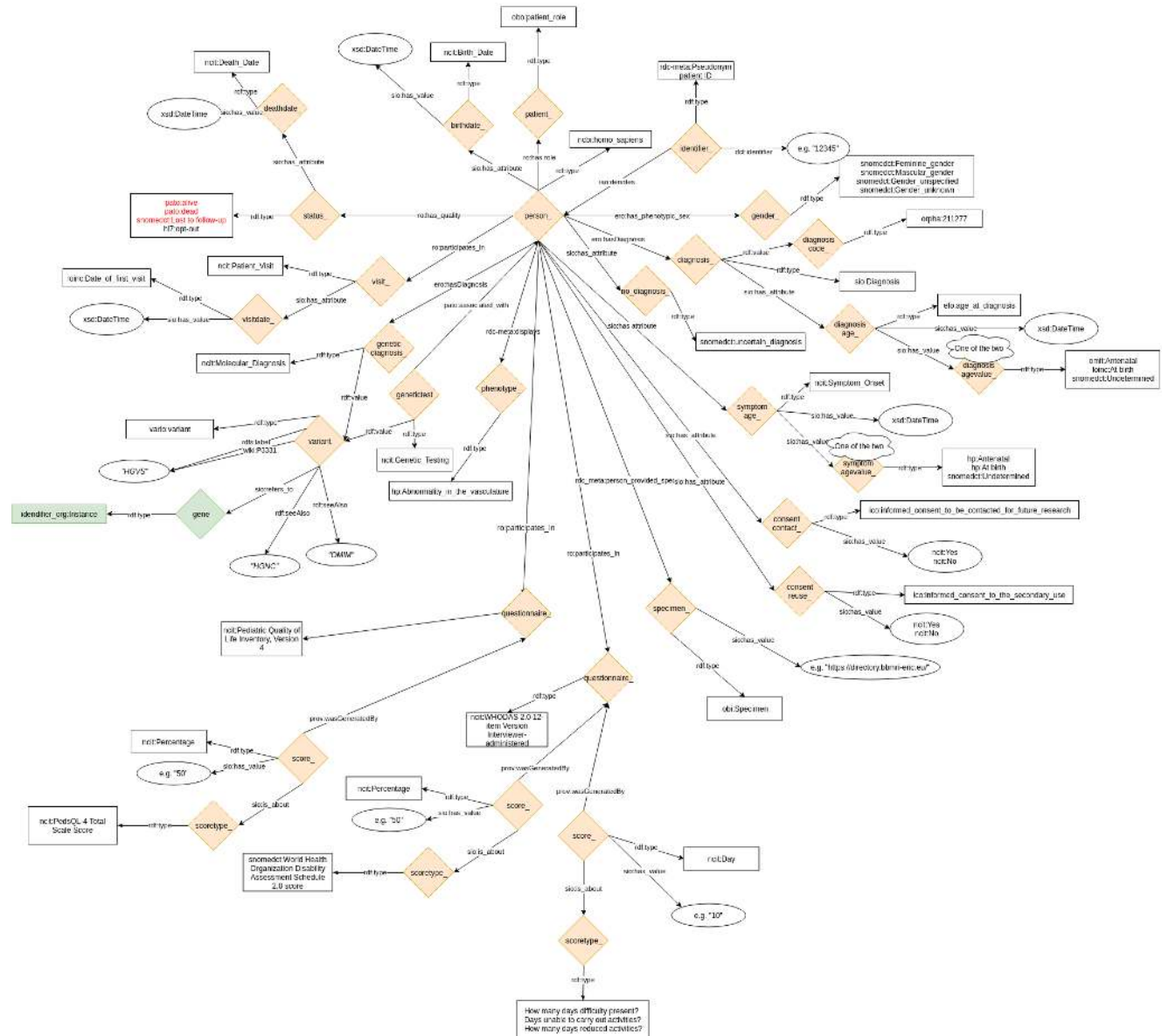
<https://eupid.eu/#/home>



Courtesy of Michael Nitzlnader, AIT, et al

INTER-OPERABILITY

To ease cross search, we will develop extensions together with other registries using EJP RD, GA4GH, BBMRI, ELIXIR, etc, and make the meta-data available in the Central Metadata Repository(ERDRI.mdr)



Courtesy of Annika Jacobsen, Rajaram Kaliyaperumal, Marco Roos, Peter-Bram ' <https://github.com/LUMC-BioSemantics/ERN-common-data-elements>

DEMO

Acknowledgements:

- ERN teams
- MOLGENIS partners past 15y
- GEN2PHEN
- RD-connect
- Solve-RD
- EJP RD
- ELIXIR
- BBMRI

<http://molgenis.org>

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MOLGENIS



umcg

Genomics coordination
center