RareLink: scalable REDCap-based framework linking international registries to FHIR and Phenopackets

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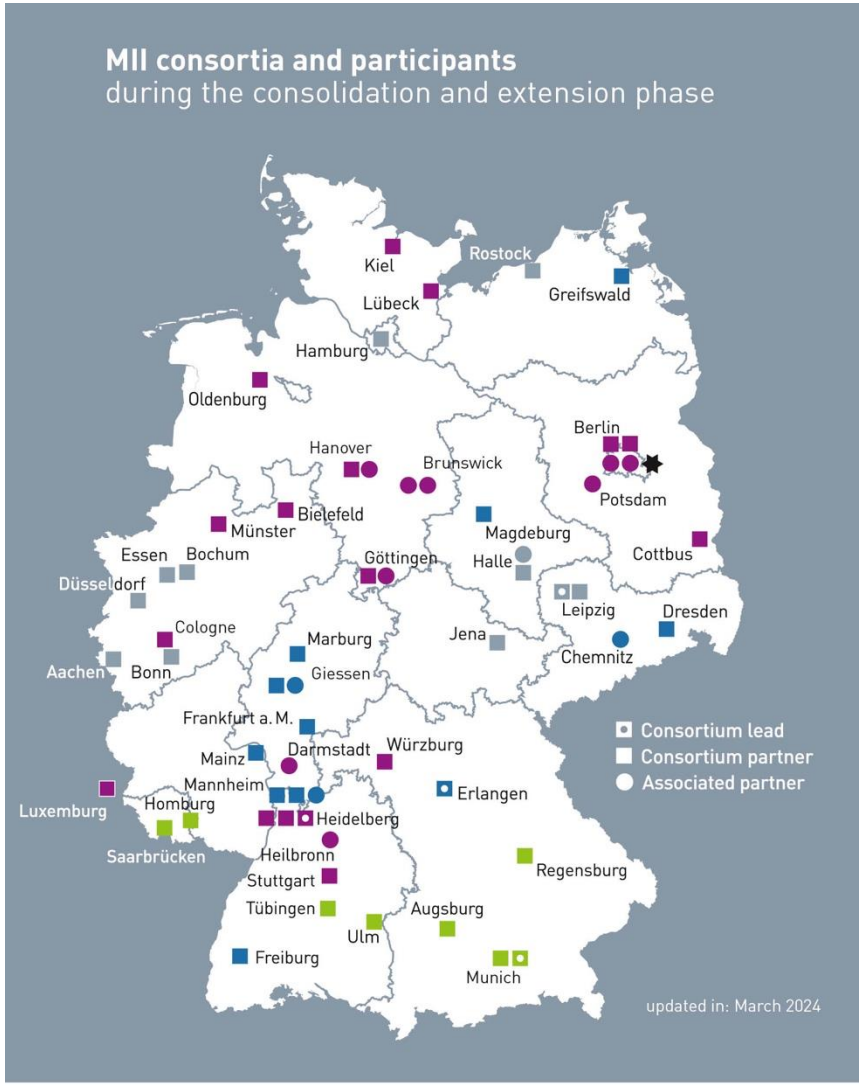

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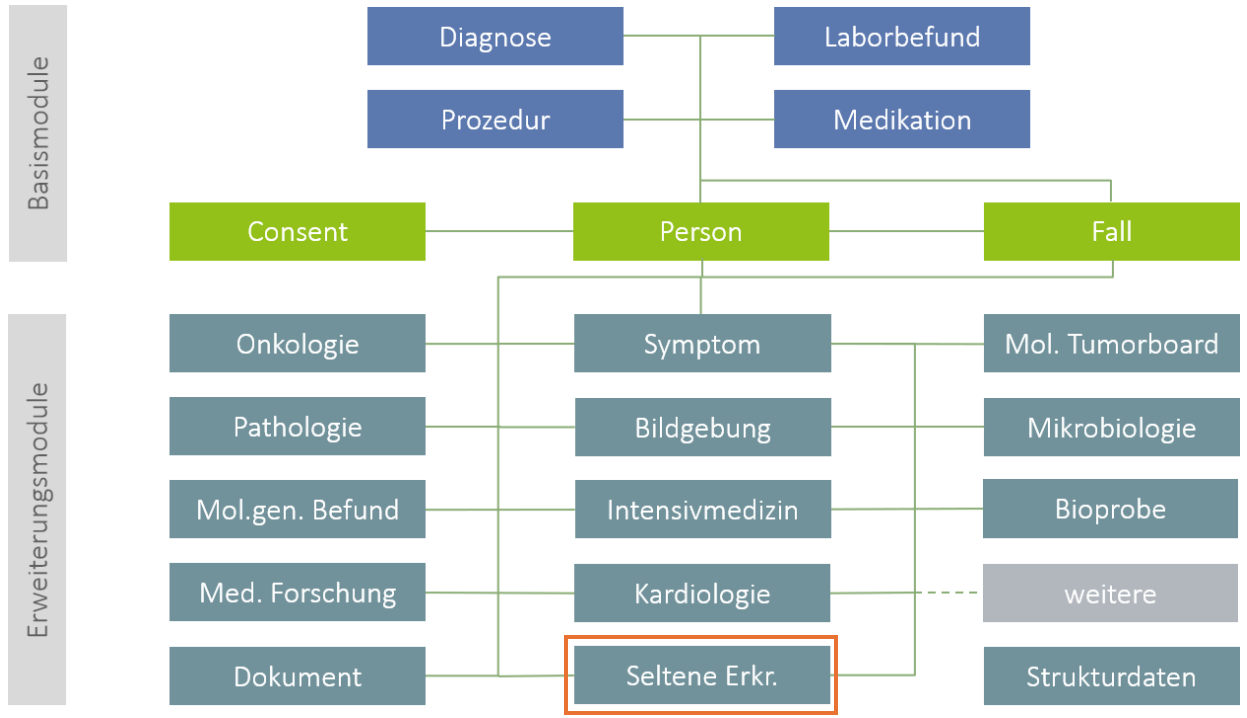

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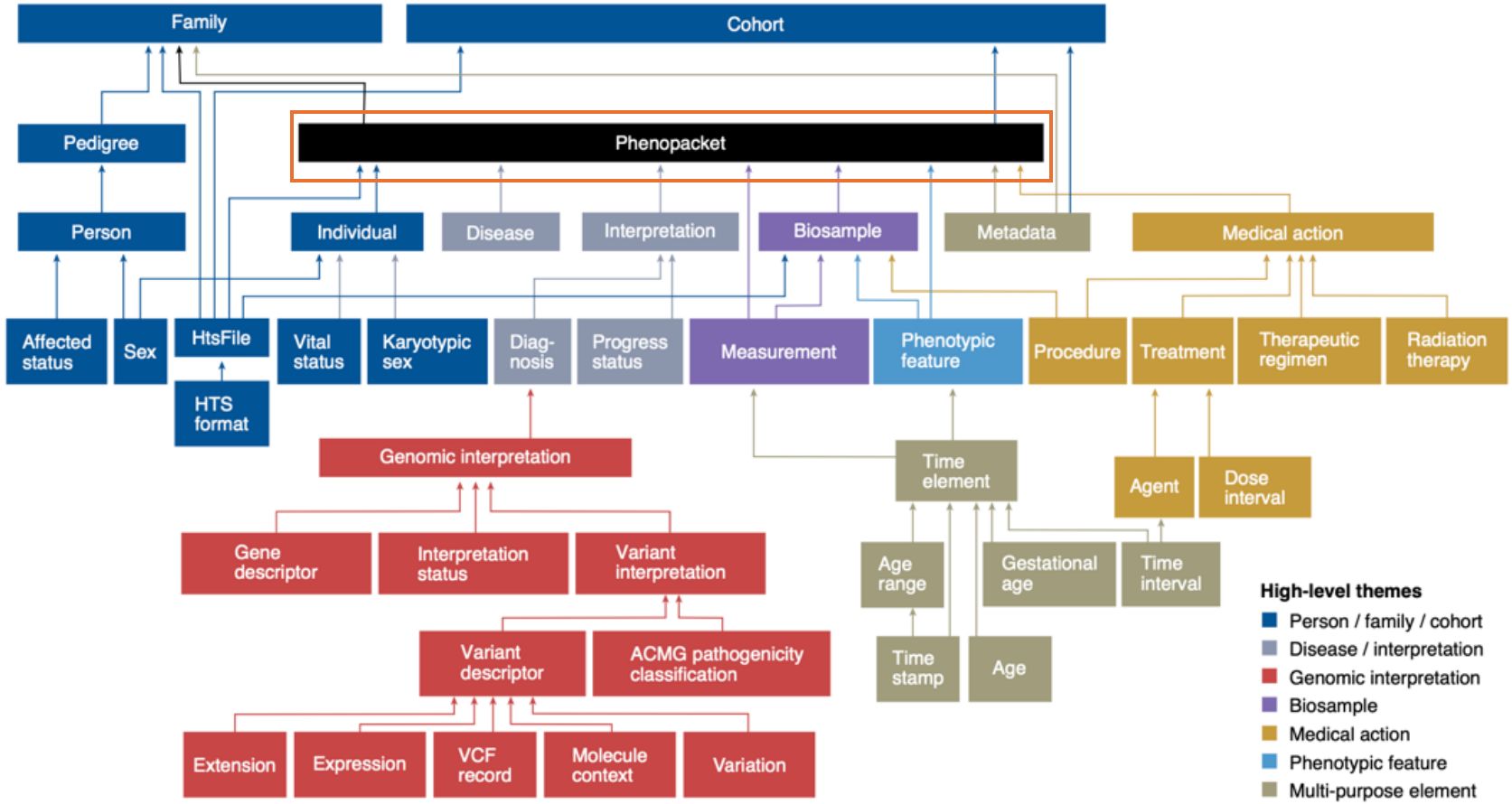
phenopackets v2.0



nature biotechnology

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The GA4GH Phenopacket schema defines a computable representation of clinical data

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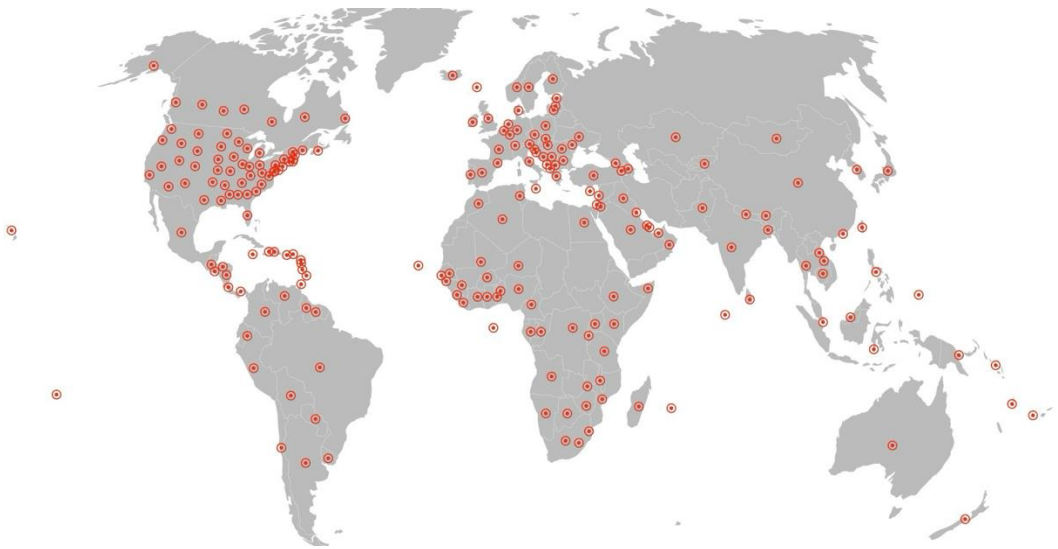
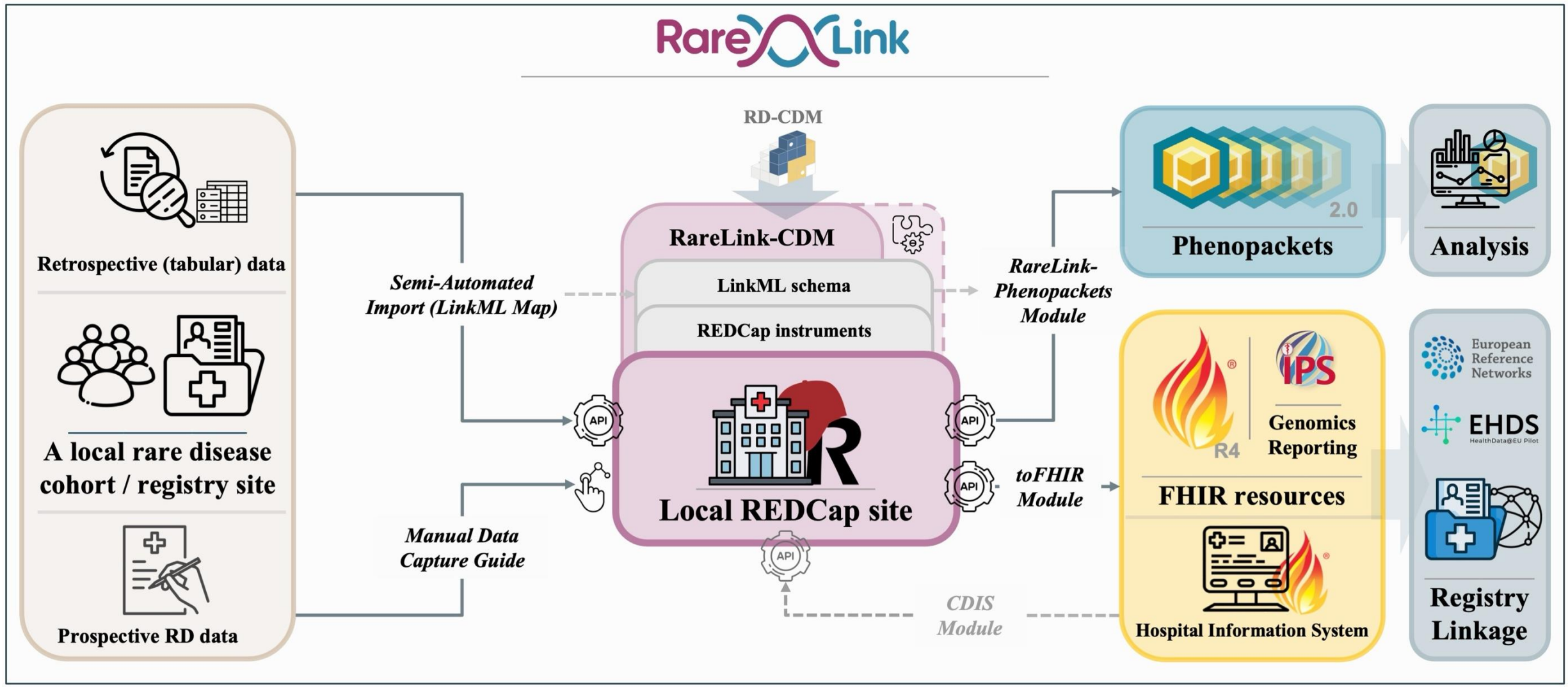
REDCap

Research Electronic Data Capture

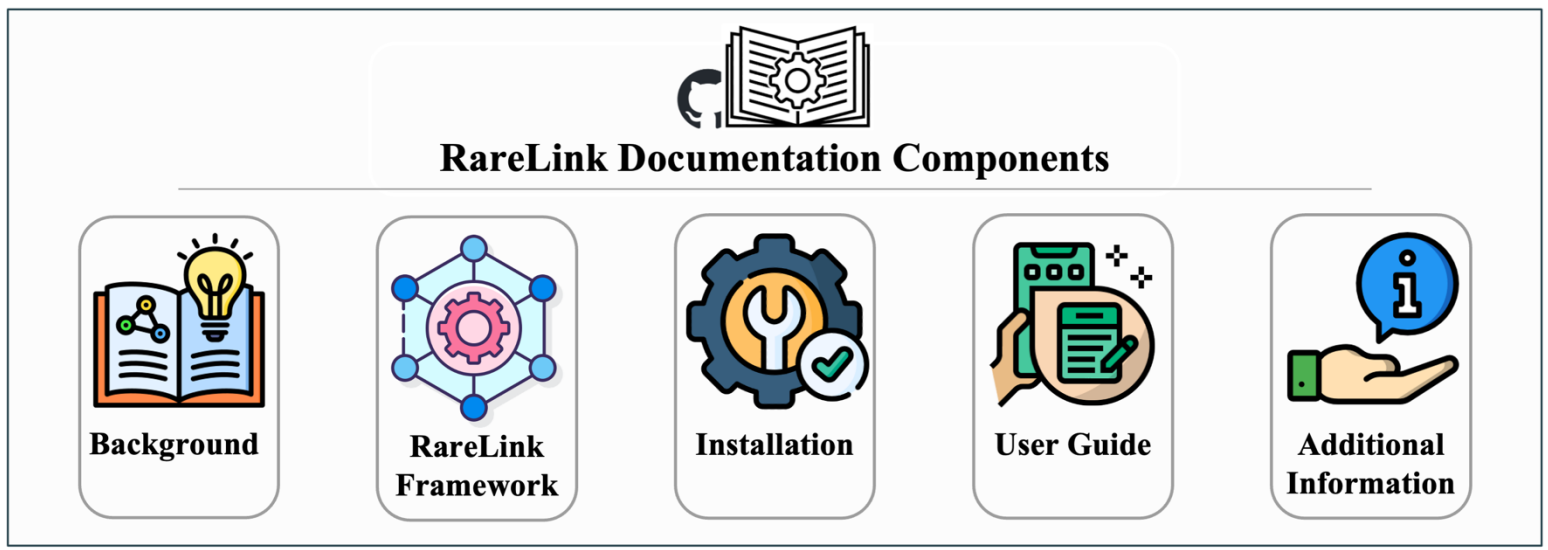


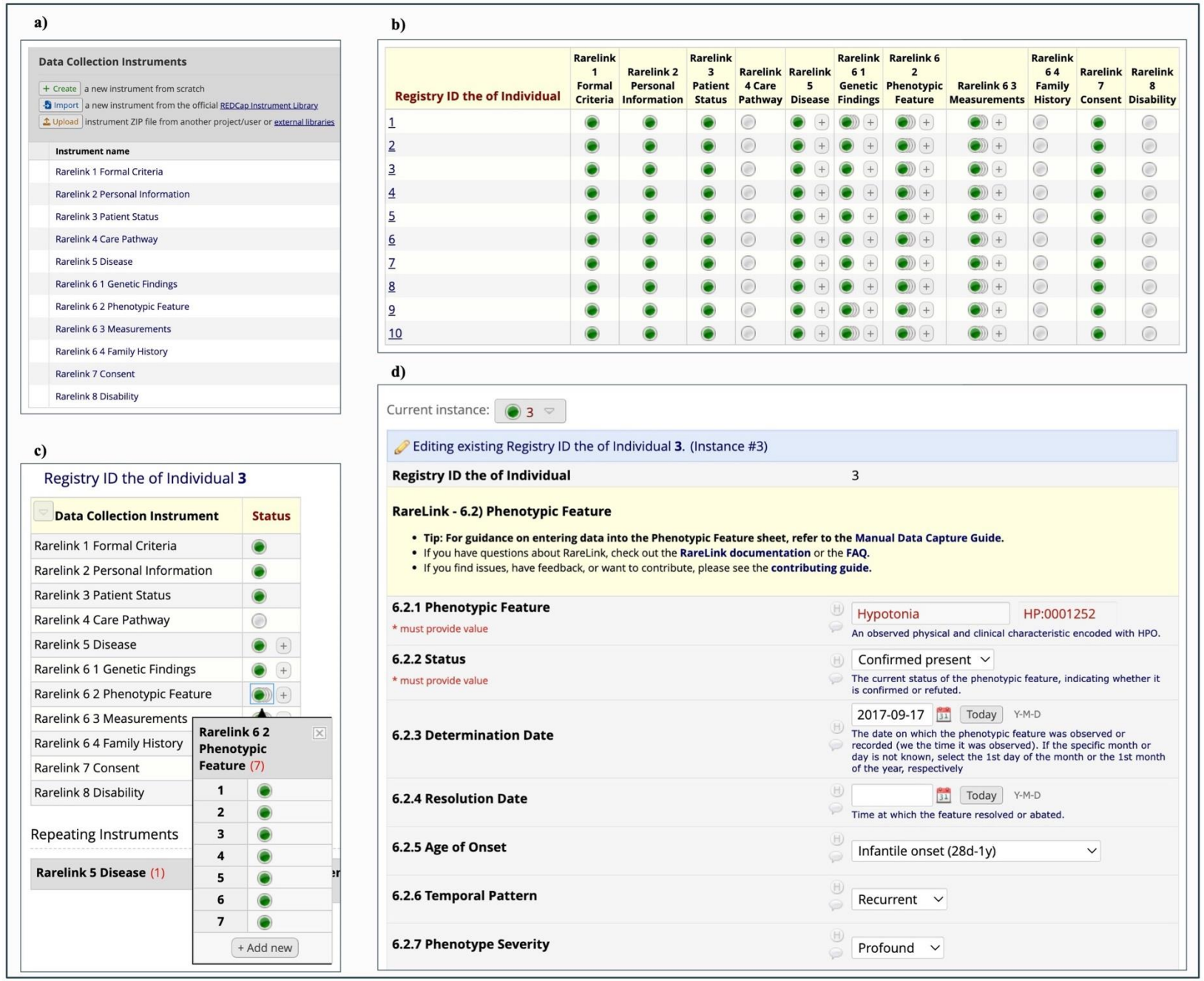

RareLink
RD-CDM
Retrospective (tabular) data
A local rare disease cohort / registry site
Prospective RD data
Semi-Automated Import (LinkML Map)
Manual Data Capture Guide
RareLink-CDM
LinkML schema
REDCap instruments
Local REDCap site
API
CDIS Module
RareLink-Phenopackets Module
toFHIR Module
Phenopackets
2.0
Analysis
R4
IPS
Genomics Reporting
FHIR resources
Hospital Information System
European Reference Networks
EHDS
HealthData@EU Pilot
Registry Linkage

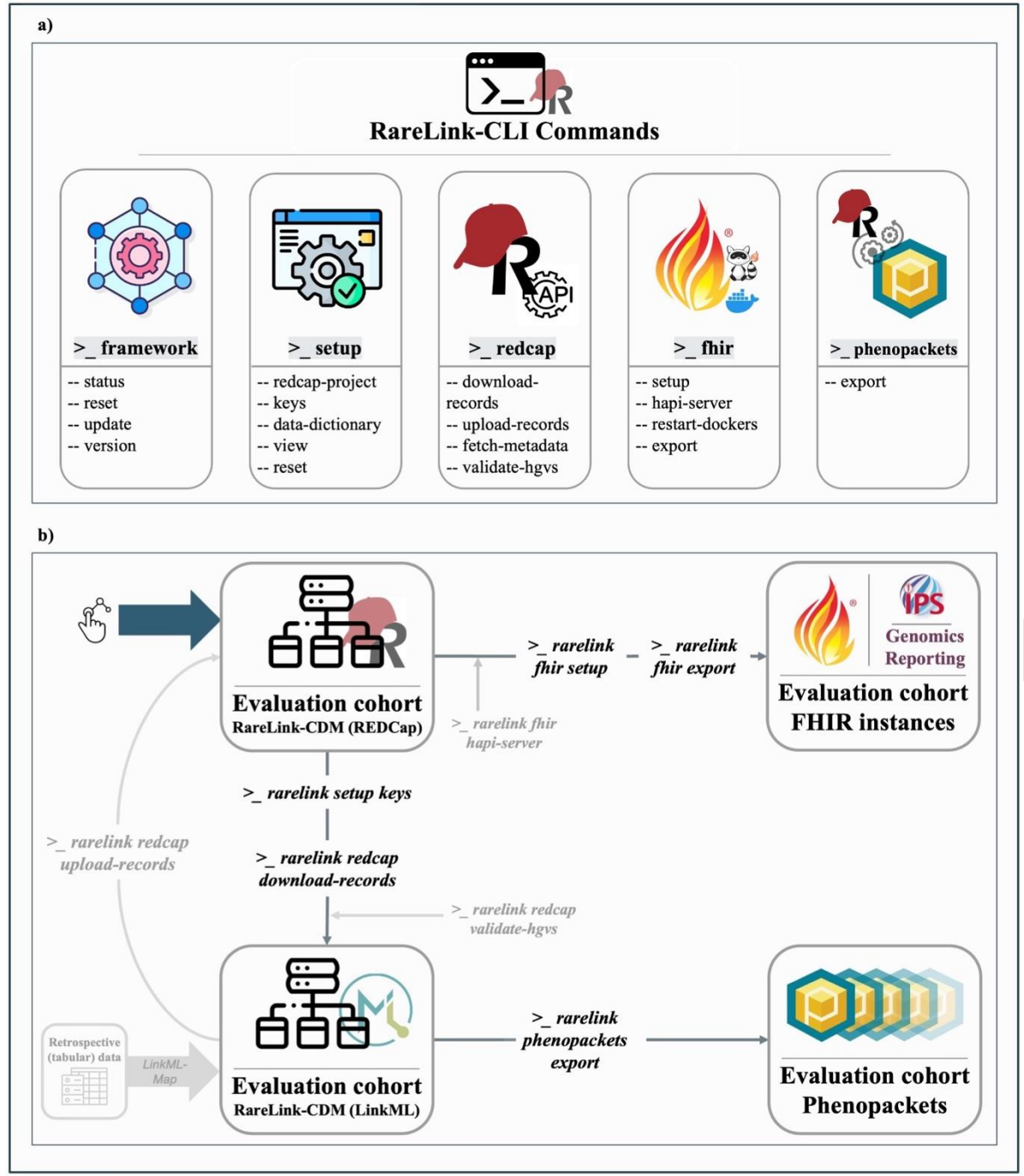
Welcome to the RareLink REDCap Documentation!

docs passing

RareLink is a novel framework designed for managing and processing rare disease (RD) data within the REDCap. Rare diseases affect over 260 million individuals worldwide, yet data quality and scarcity pose significant challenges in research and clinical care. RareLink aims to standardize and streamline RD data management around REDCap by providing a structured project setup that ensures consistency across data collection instruments, variables, and data dictionaries. RareLink has integrated the ontology-based rare disease common data model (RD-CDM) into its core, allowing its linkage and preconifgured export to data the standards HL7 FHIR and the GA4GH Phenopacket Schema. In the following you will find detailed information on the RareLink framework, including its background, components, installation instructions, user guide, and full examples.




a)
Data Collection Instruments
+ Create a new instrument from scratch
Import a new instrument from the official REDCap Instrument Library
Upload instrument ZIP file from another project/user or external libraries
Instrument name
Rarelink 1 Formal Criteria
Rarelink 2 Personal Information
Rarelink 3 Patient Status
Rarelink 4 Care Pathway
Rarelink 5 Disease
Rarelink 6 1 Genetic Findings
Rarelink 6 2 Phenotypic Feature
Rarelink 6 3 Measurements
Rarelink 6 4 Family History
Rarelink 7 Consent
Rarelink 8 Disability
b)
Registry ID the of Individual
Rarelink 1 Formal Criteria
Rarelink 2 Personal Information
Rarelink 3 Patient Status
Rarelink 4 Care Pathway
Rarelink 5 Disease
Rarelink 6 1 Genetic Findings
Rarelink 6 2 Phenotypic Feature
Rarelink 6 3 Measurements
Rarelink 6 4 Family History
Rarelink 7 Consent
Rarelink 8 Disability
1
2
3
4
5
6
7
8
9
10
c)
Registry ID the of Individual 3
Data Collection Instrument
Status
Rarelink 1 Formal Criteria
Rarelink 2 Personal Information
Rarelink 3 Patient Status
Rarelink 4 Care Pathway
Rarelink 5 Disease
Rarelink 6 1 Genetic Findings
Rarelink 6 2 Phenotypic Feature
Rarelink 6 3 Measurements
Rarelink 6 4 Family History
Rarelink 7 Consent
Rarelink 8 Disability
Repeating Instruments
Rarelink 5 Disease (1)
Rarelink 6 2 Phenotypic Feature (7)
1
2
3
4
5
6
7
+ Add new
d)
Current instance: 3
Editing existing Registry ID the of Individual 3. (Instance #3)
Registry ID the of Individual 3
RareLink - 6.2) Phenotypic Feature
Tip: For guidance on entering data into the Phenotypic Feature sheet, refer to the Manual Data Capture Guide.
If you have questions about RareLink, check out the RareLink documentation or the FAQ.
If you find issues, have feedback, or want to contribute, please see the contributing guide.
6.2.1 Phenotypic Feature
* must provide value
Hypotonia
HP:0001252
An observed physical and clinical characteristic encoded with HPO.
6.2.2 Status
* must provide value
Confirmed present
The current status of the phenotypic feature, indicating whether it is confirmed or refuted.
6.2.3 Determination Date
2017-09-17
Today
Y-M-D
The date on which the phenotypic feature was observed or recorded (we the time it was observed). If the specific month or day is not known, select the 1st day of the month or the 1st month of the year, respectively
6.2.4 Resolution Date
Today
Y-M-D
Time at which the feature resolved or abated.
6.2.5 Age of Onset
Infantile onset (28d-1y)
6.2.6 Temporal Pattern
Recurrent
6.2.7 Phenotype Severity
Profound



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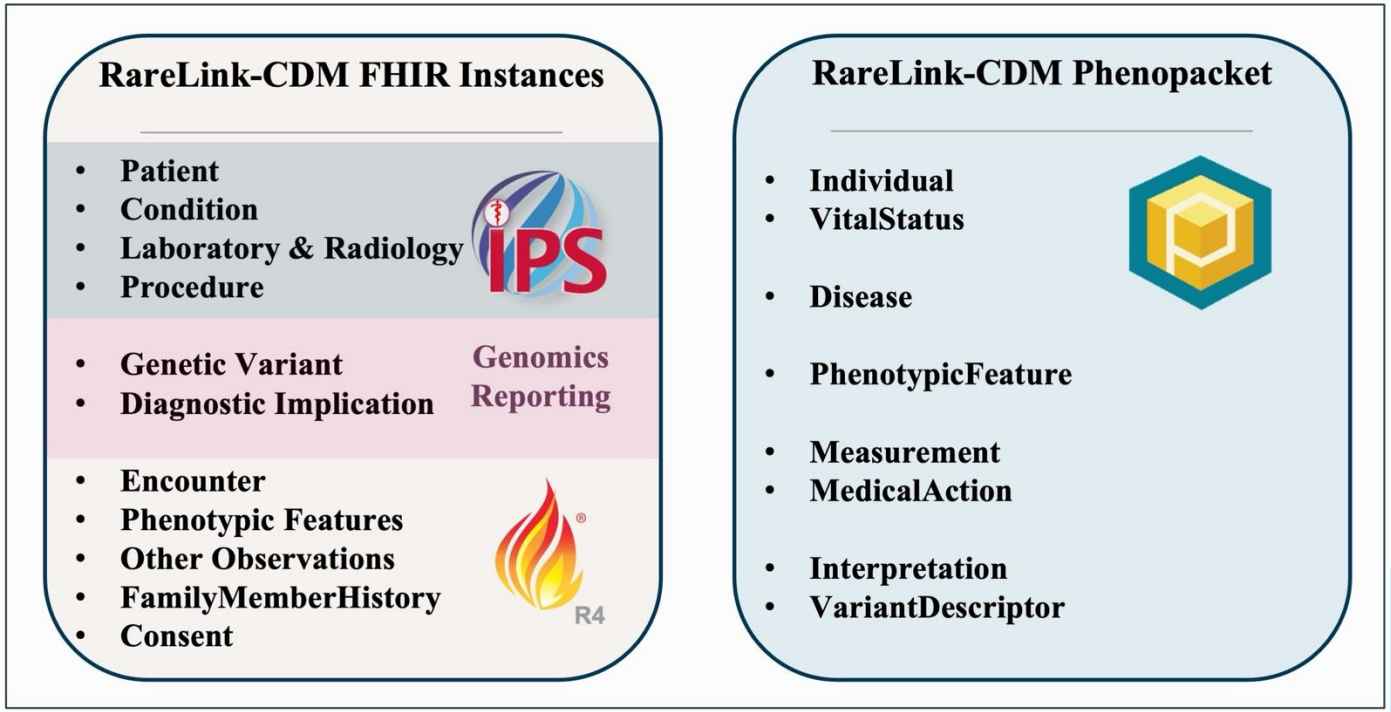

RareLink Implementation Guide
2.0.0 - 2.0.0

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Official URL: **https://rarelink.bih-charite.de/fhir/ImplementationGuide/rarelink-ig**	*Version*: **2.0.0**
Draft as of 2025-09-25	*Computable Name*: RareLinkImplementationGuide

Welcome to the RareLink Implementation Guide

Welcome to the RareLink Implementation Guide – a non-balloted extension of the European Rare Disease Infrastructure Common Data Set (ERDRI-CDS). RareLink is a novel, open-source, and REDCap-based framework linking international registries to HL7 FHIR R4 and GA4GH Phenopackets to enable rare disease data interoperability.

- RareLink repository
- RareLink documentation
- RareLink paper prepint

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Article

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RareLink: scalable REDCap-based framework for rare disease interoperability linking international registries to FHIR and Phenopackets

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https://www.nature.com/articles/s41525-025-00534-z