Why Translational Medicine Depends on Interoperability

03-12-2021

Prof. Dr. med. Sylvia Thun
Core Unit “eHealth & Interoperability”
Berlin Institute of Health (BIH) @ Charité

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Content

- Interoperability
  - ✓ Artificial Intelligence and Big Data
  - ✓ Medical Communication and Connectivity
  - ✓ Research
  - ✓ International Cooperation
- FAIR data – Motivation and Principles
- Standards and Terminologies
- Projects
  - ✓ NFDI4Health;
  - ✓ NFN German Corona Consensus GECCO
  - ✓ Medical Informatics Initiative

Interoperability

“the ability of two or more systems or components to exchange information and
to use the information that has been exchanged” *IEEE Std 610* 1–217 (1991)

https://commons.wikimedia.org/wiki/File:Pieter_Bruegel_the_Elder_-_The_Tower_of_Babel_(Vienna)_-_Google_Art_Project_-_edited.jpg

Motivation

- Enhance **reproducibility** of research
- Improve **reusability of scientific data** across projects
- Harmonize datasets to enable **integration across studies**
- Make data reusable not only for humans, but also **machines**

https://pixabay.com/photos/archive-boxes-documents-folders-1850170

Digital Medicine Depends on Interoperability

AI and Big Data
- provide algorithms with clear data structure and semantics
- ensure validity of analysis results
- create trust in digital technologies

Medical Communication
- enable easy information retrieval
- avoid medical errors caused by communication barriers
- reduce documentation burden
- empower patients

Research
- improve the use of real-world data (e.g. for large-scale observational studies)
- create new research hypotheses (with data mining and AI)
- enable remote development of analysis scripts

International Cooperation
- pool data across organizations (e.g. rare diseases, precision medicine)
- tackle global public health issues (e.g. infection control, epidemics)
- provide global access to new technologies

Lehne M, Sass J, Essenwanger A, Schepers J, Thun S (2019).
Why digital medicine depends on interoperability. *npj Digital Medicine*.

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Communication and Connectivity

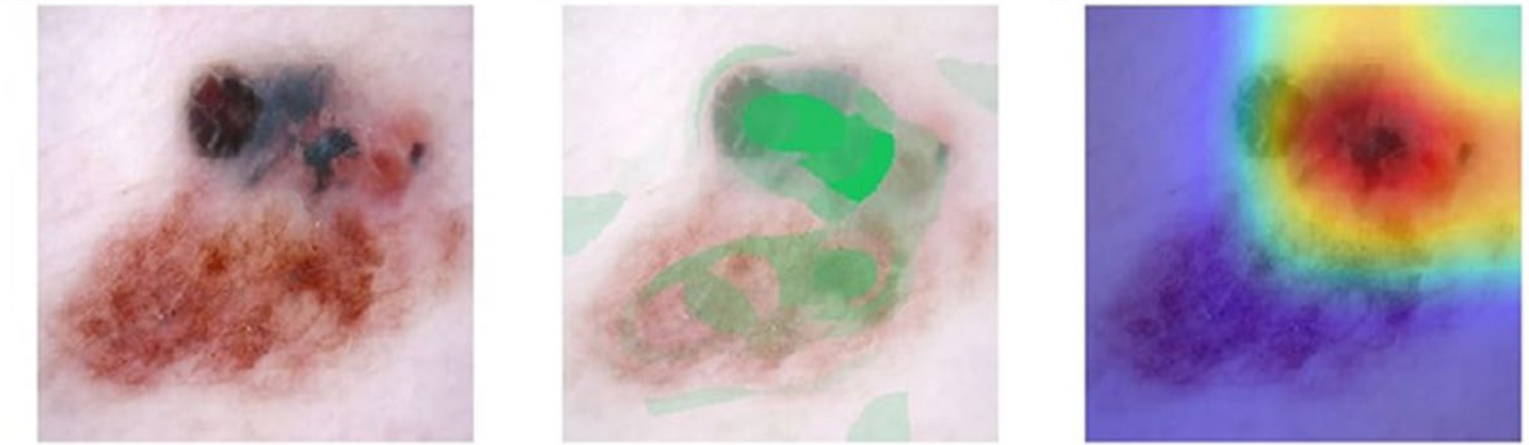
ISO 11073 SDC
HL7 FHIR

- LOINC
- SNOMED
- IDMP
- UCUM

Artificial Intelligence

The Role of DICOM in Artificial Intelligence for Skin Disease

Liam J. Caffery[1,2*], Veronica Rotemberg[3], Jochen Weber[3], H. Peter Soyer[2,4], Josep Malvehy[5] and David Clunie[6]



Research

Health Level Seven® International

For Immediate Release

Contacts:
Andrea Ribick, HL7, andrea@HL7.org, (734) 677-7777
Craig Sachson, OHDSI, sachson@ohdsi.org

HL7 International and OHDSI Announce Collaboration to Provide Single Common Data Model for Sharing Information in Clinical Care and Observational Research

Leading organizations will integrate products to create a single source for the sharing and tracking of data

Ann Arbor, Mich. and New York City, N.Y. – March 1, 2021 – Health Level Seven International (HL7®) and the Observational Health Data Sciences and Informatics (OHDSI) today announced a collaboration to address the sharing and tracking of data in the healthcare and research industries by creating a single common data model. The organizations will integrate HL7 Fast Healthcare Interoperability Resources (FHIR®) and OHDSI's Observational Medical Outcomes Partnership (OMOP) common data model to achieve this goal.

International Cooperation

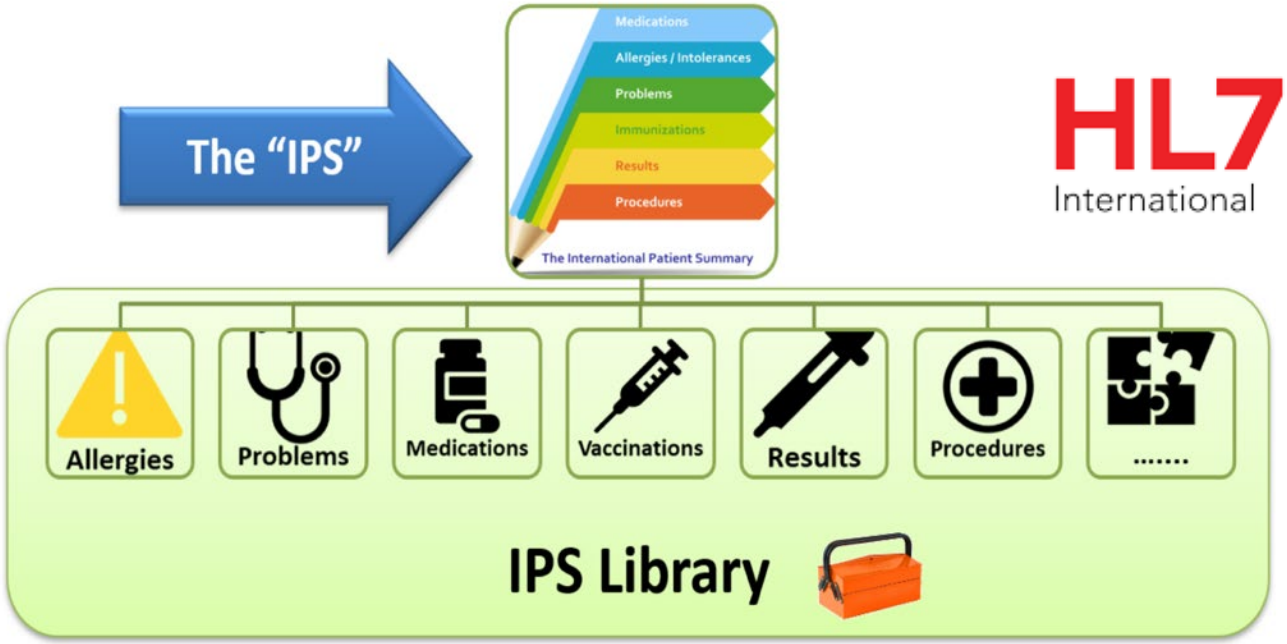
Joint Initiative Council

- Clinical Data Interchange Standards Consortium
- European Committee for Standardisation
- Digital Imaging and Communications in Medicine
- GS1
- Health Level Seven International
- Integrating the Healthcare Enterprise
- International Organisation for Standardisation
- Logical Observation Identifiers Names and Codes
- SNOMED International

Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

International Patient Summary



HL7 International

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Reproducibility crisis

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Scientific Journals

nature research

*"A condition of publication in a Nature Research journal is that **authors are required to make materials, data, code, and associated protocols promptly available to readers without undue qualifications."** –* Nature Research

PLOS ONE

"PLOS journals require authors to make all data necessary to replicate their study's findings publicly available without restriction at the time of publication." – PLOS One

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The FAIR Principles

SCIENTIFIC DATA

Amended: Addendum

OPEN

SUBJECT CATEGORIES

» Research data

» Publication characteristics

Comment: The FAIR Guiding Principles for scientific data management and stewardship

Mark D. Wilkinson *et al.*#

There is an urgent need to improve the infrastructure supporting the reuse of scholarly data. A diverse set of stakeholders—representing academia, industry, funding agencies, and scholarly publishers—have come together to design and jointly endorse a concise and measureable set of principles that we refer to as the FAIR Data Principles. The intent is that these may act as a guideline for those wishing to enhance the reusability of their data holdings. Distinct from peer initiatives that focus on the human scholar, the FAIR Principles put specific emphasis on enhancing the ability of machines to automatically find and use the data, in addition to supporting its reuse by individuals. This Comment is the first formal publication of the FAIR Principles, and includes the rationale behind them, and some exemplar implementations in the community.

Received: 10 December 2015
Accepted: 12 February 2016
Published: 15 March 2016

https://doi.org/10.1038/sdata.2016.18

The FAIR Principles

Box 2 | The FAIR Guiding Principles

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, 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

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

Findability
Accessibility
Interoperability
Reusability

Wilkinson, M., Dumontier, M., Aalbersberg, I. *et al.* The FAIR Guiding Principles for scientific data management and stewardship. *Sci Data* **3,** 160018 (2016). https://doi.org/10.1038/sdata.2016.18

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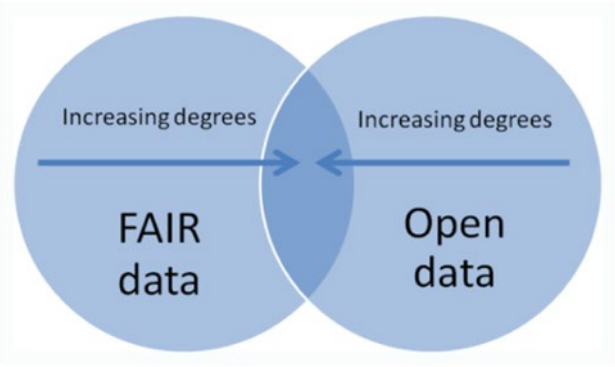
- R1.3. (meta)data meet domain-relevant community standards

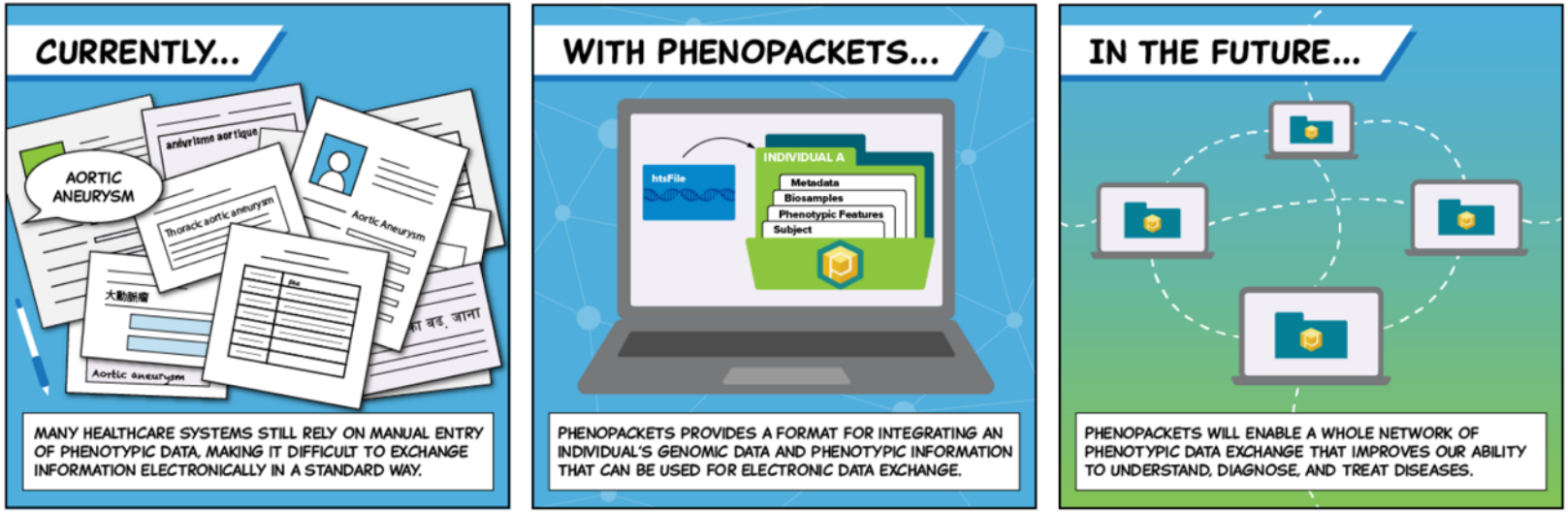
FAIR data vs. Open data

Offene Daten lesbar für Mensch und Maschine. Das ist das Ziel.



Standards and terminologies

Phenopackets: Standardizing and Exchanging Patient Phenotypic Data



Source: GA4GH

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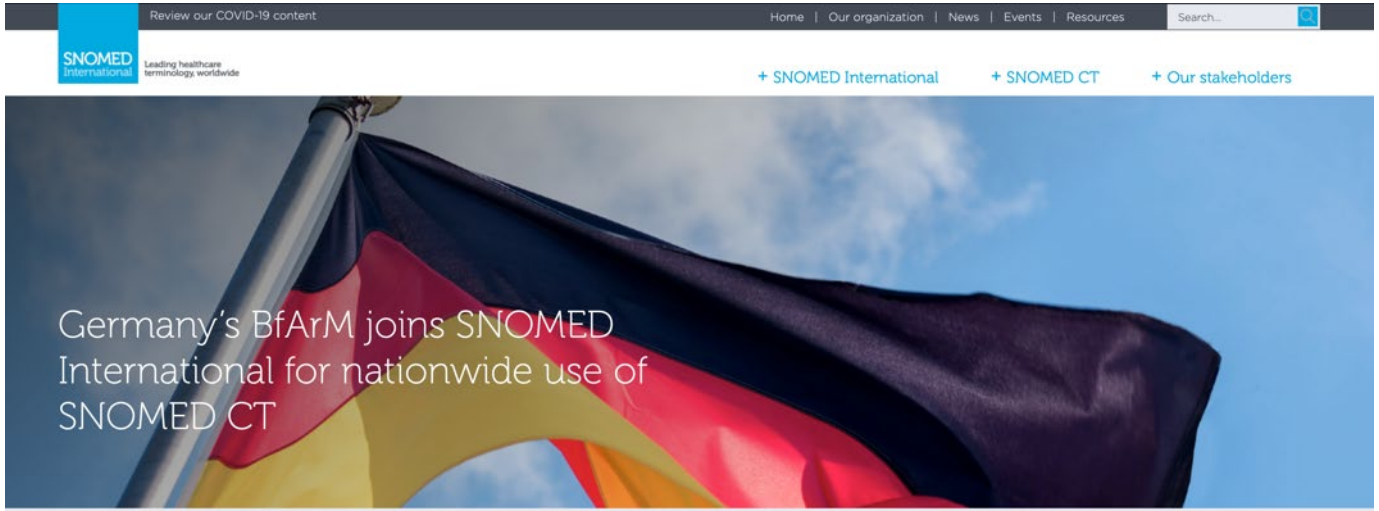
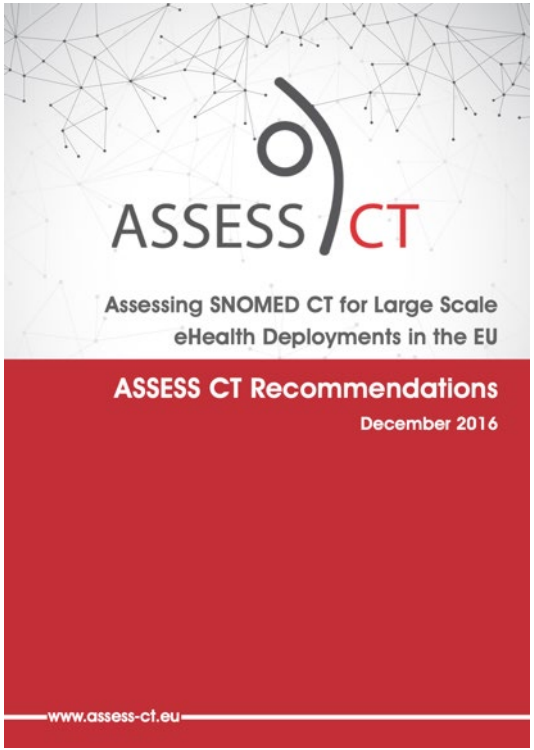
SNOMED - *Systematized Nomenclature of Medicine*



ASSESS CT



Thiel, Thun et al Stud Health Technol Inform 2016
Dewenter, Thun Stud Health Technol Inform 2018

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As well as numerous global experts and speakers at workshops and sessions organised over the course of the project, who shared their knowledge, experience and critical reviews.

Imprint

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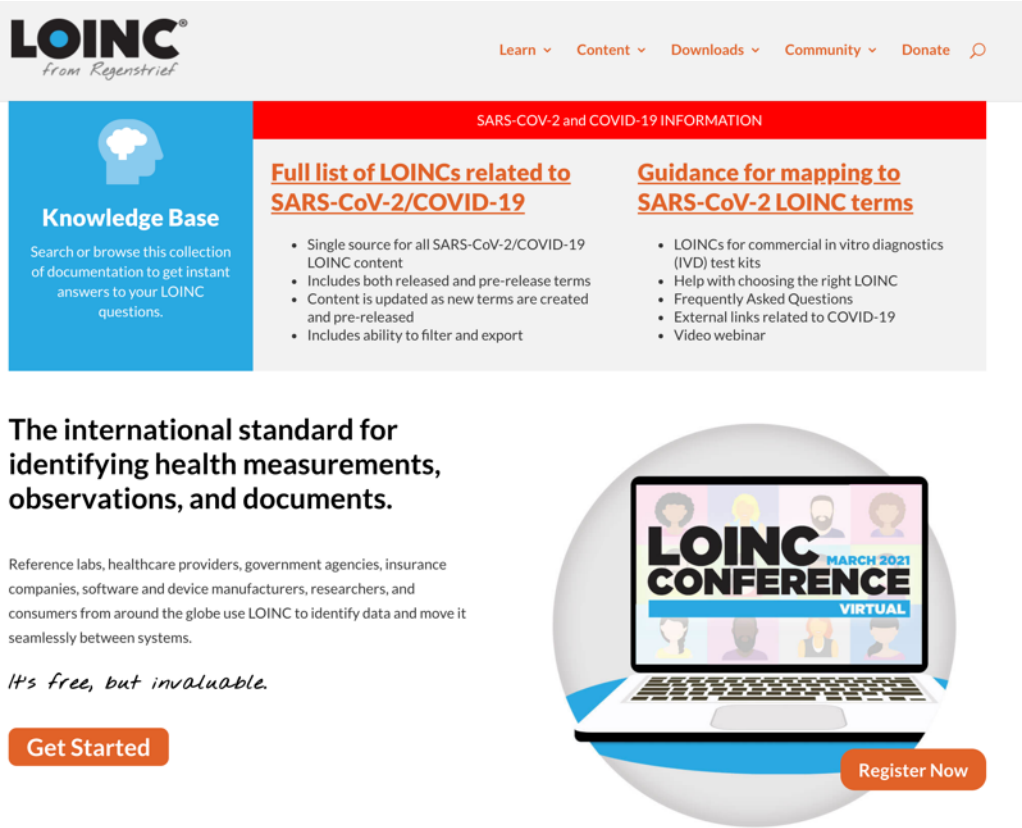
LOINC@BfArM



A survey of licensed Patient Reported Outcome Measures in the process of submission to LOINC

Alexander Bartschke
Berlin Institute of Health, Charité

Naveen Moses Raj Rajkumar
Berlin Institute of Health, Charité

Background: Patient Reported Outcome Measures (PROMs) are Self-report questionnaires that patients complete on their health, prognosis and quality of life. The information collected from PROMs helps in researches, in monitoring patients' progress allowing constant communication between physicians and patients.

Logical Observation Identifiers Names and Codes (LOINC) is a common language (set of identifiers, names and codes) for identifying health measurements, observations and documents. The use of LOINC with PROMs facilitate the collected data to make it further reusable for extended use across health information systems enabling interoperability.

Objective: To investigate the review of 99 Self-report measures included from various sources by Linton et al. for their presence in the LOINC panel. The Self-report measures that are not present in the LOINC panel are categorized into licensed and license free, the copyright owners of the licensed instruments were provided with detailed information about LOINC and asked permission for instruments to be added to LOINC panel. The copyright owners are asked to take part in a survey and to give their reasons for allowing or not allowing the instruments to be added to LOINC panel.

Results: The survey contributes an elaborate explanation on the various reasons for allowing or refusal to add the instruments to LOINC panel by the copyright holders. The reasons for allowing or refusal are categorized on several themes such as intellectual property rights, scoring methods, organizational issues etc.

Conclusion: This report provides the users and researchers on the availability of the instruments in LOINC panel and their reason for their absence. The report provides on the various barriers existing in the usage of licensed instruments particularly to be interoperable for access across systems and aggregation of useful data.

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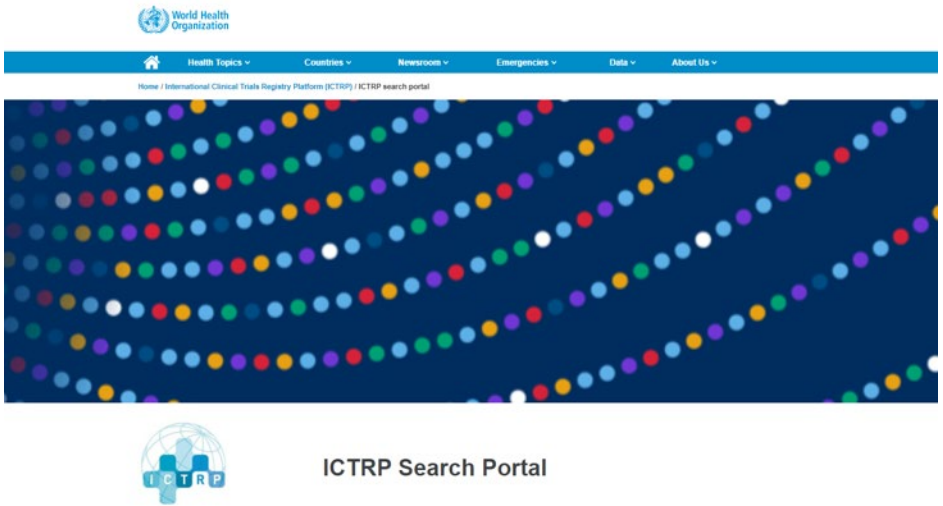
Projects: NFDI4Health

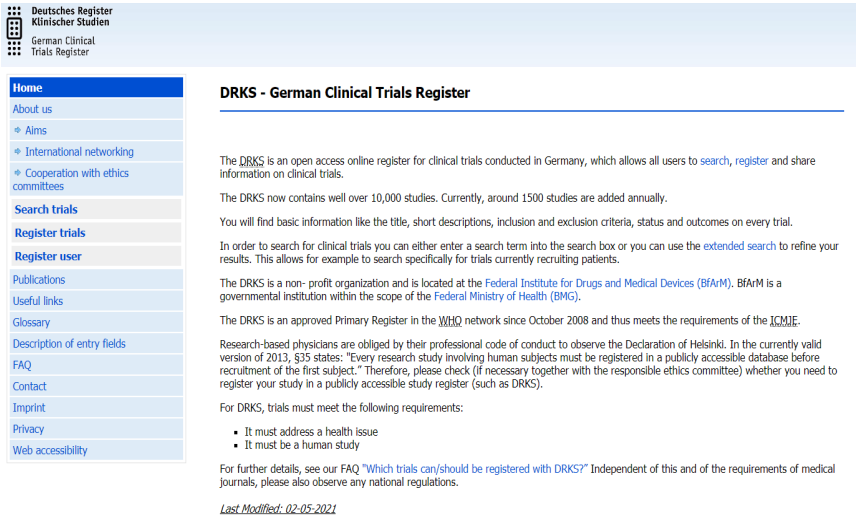
NFDI4Health – Aims

- Creation of the most comprehensive inventory of German epidemiological, public health and clinical trial data to date
- Centralized Data catalogue
  - Search functionalities
  - Sophisticated data access management
  - Data analysis toolbox
- Respecting stringent requirements for privacy concerning personal health data
- High degree of interoperability

Data Registries – Clinical Trials

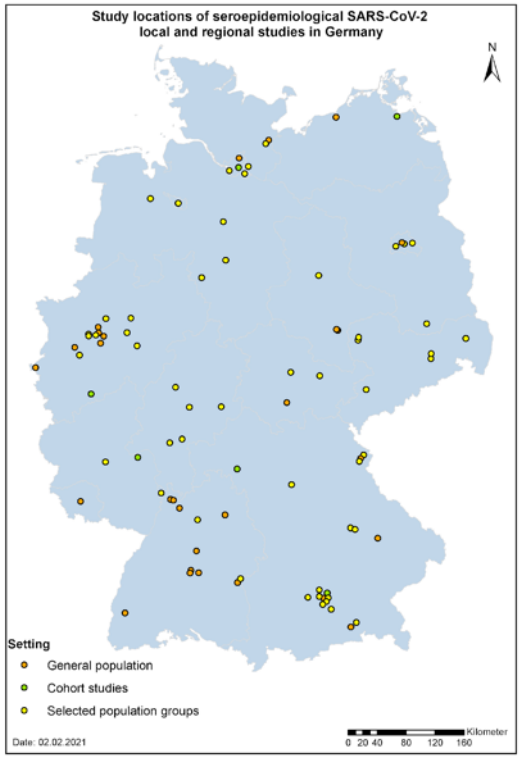


https://www.who.int/clinical-trials-registry-platform/the-ictrp-search-portal

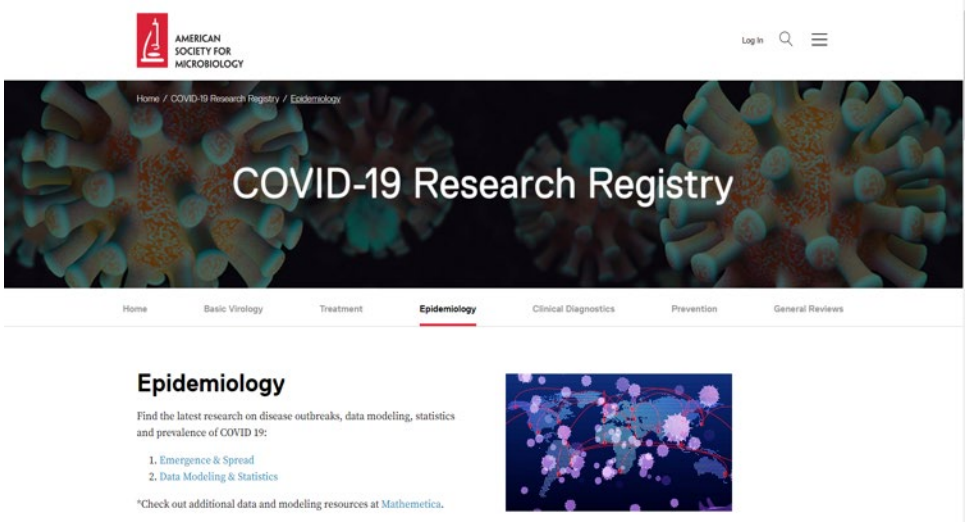


https://www.drks.de/drks_web/navigate.do?navigationId=start

Data Registries – Epidemiological Studies



https://www.rki.de/EN/Content/infections/epidemiology/outbreaks/COVID-19/AK-Studien english/Sero_List.html;jsessionid=3A90E1A2F219ED3E3C6901547E3BB137.internet101



https://asm.org/COVID/COVID-19-Research-Registry/Epidemiology

nfdi 4 health

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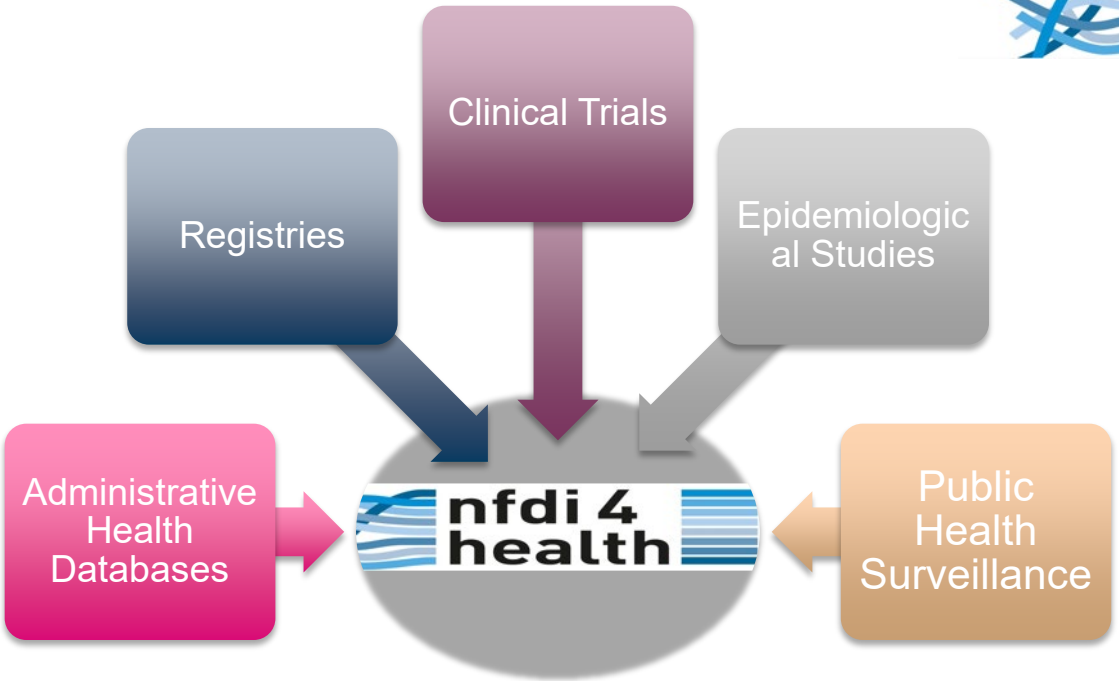
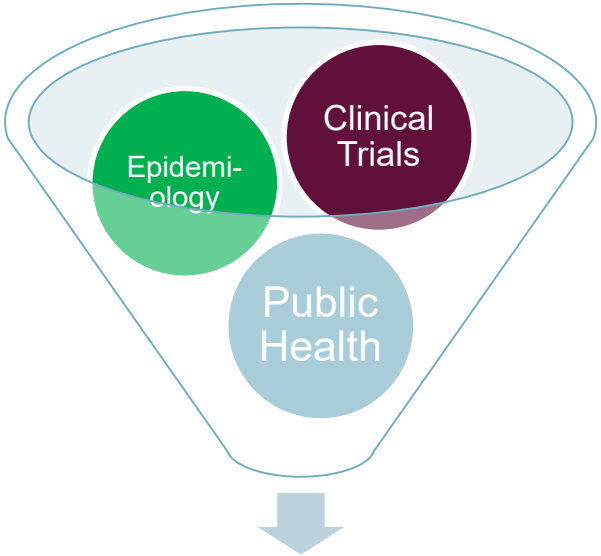
NFDI4Health – Aims



The resulting infrastructure will build bridges between user communities and data holders from epidemiology, public health and clinical trials

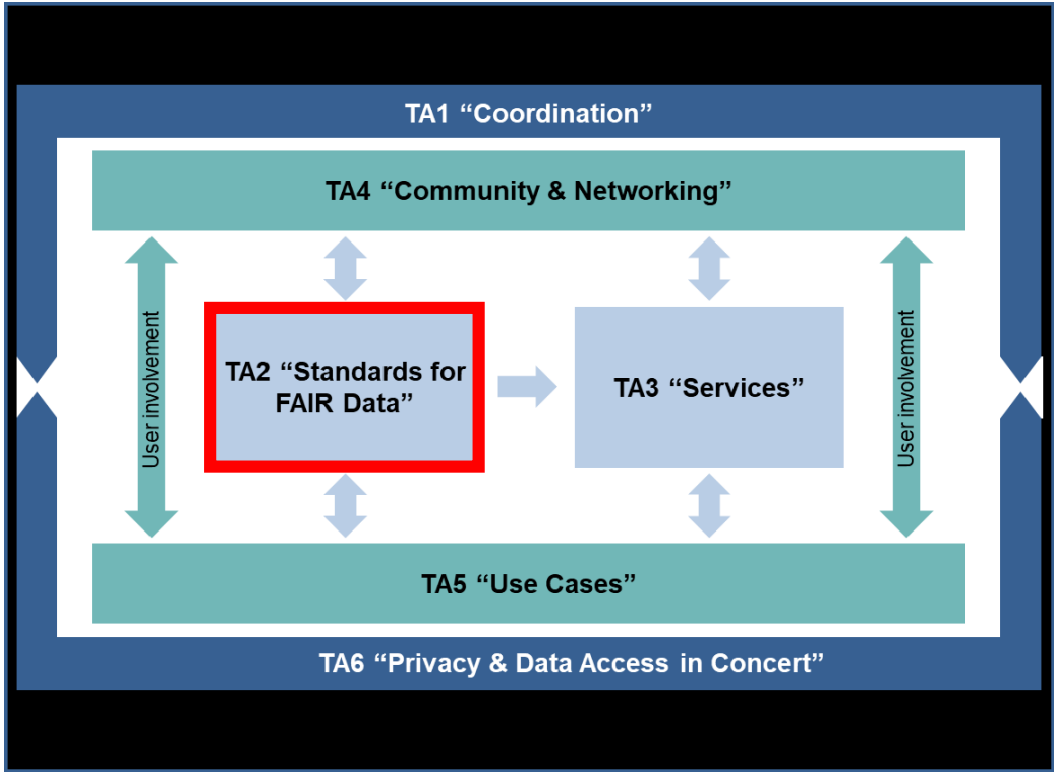
NFDI4Health – Aims



Interoperability

- Cooperation
- Data Harmonization
- Machine-processable (AI)
- Data Sharing
- Record Linkage

Task Area Core-Unit eHealth und Interoperability

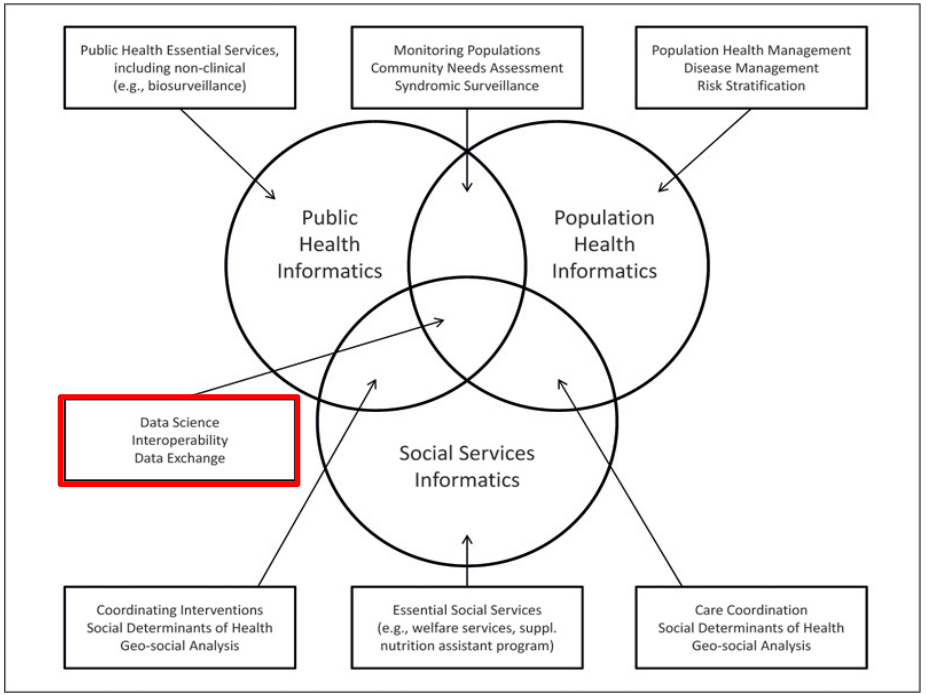


- **Interoperability-Roadmap**
- Identification of relevant **Standards, Terminologies and Ontologies**
- Value Set Implementation
- **USE of SNOMED CT/ LOINC**
- Development of Implementation Guidelines (e.g. HL7 FHIR)
- Development of data models

Scientific Background

Public and Population Health Informatics: The Bridging of Big Data to Benefit Communities

Roland Gamache, Hadi Kharrazi, Jonathan P. Weiner



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Scientific Background: Harmonization

Factors influencing harmonized health data collection, sharing and linkage in Denmark and Switzerland: A systematic review

Lester Darryl Geneviève, Andrea Martani, Maria Christina Mallet, Tenzin Wangmo, Bernice Simone Elger

Barriers		Countries involved in projects		
Cluster	Sub-cluster	Denmark N[a] = 251	Switzerland N = 80	Both countries N = 14
		n[b] (mean no. of barriers per project)	n (mean no. of barriers per project)	n (mean no. of barriers per project)
Ethical	Privacy	6 (0.02)	3 (0.04)	-[c] (N/A)
	Respect for Autonomy	3 (0.01)	3 (0.04)	- (N/A)
	Other	3 (0.01)	1 (0.01)	- (N/A)
Legal	Data Protection Regulations	2 (0.01)	1 (0.01)	1 (0.07)
	Divergence in National Legislations for Data Security and Privacy	2 (0.01)	- (N/A)	2 (0.14)
	Other	5 (0.02)	3 (0.04)	1 (0.07)
Technical	Lack of Data Standards	104 (0.41)	33 (0.41)	14 (1.00)
	Data Quality Issues	181 (0.72)	44 (0.55)	9 (0.64)
	Limited Technical Capabilities	11 (0.04)	9 (0.11)	1(0.07)
	Other	8 (0.03)	2 (0.03)	- (N/A)
Financial	Lack of Funding	4 (0.02)	3 (0.04)	1 (0.07)
	Other	1 (0.00)	- (N/A)	- (N/A)
Political	Mistrust between stakeholders	- (N/A)	3 (0.04)	- (N/A)
	Data Ownership	2 (0.01)	- (N/A)	- (N/A)
	Institutional/constitutional organization issues	2 (0.01)	4 (0.05)	- (N/A)
	Other	- (N/A)	2 (0.03)	- (N/A)
Motivational	Lack of research incentives	6 (0.02)	9 (0.11)	2 (0.14)
	Stakeholder restricts access for re-use of data as deemed unfit for secondary use	2 (0.01)	- (N/A)	- (N/A)
	Stakeholder competing interests	1 (0.00)	1 (0.01)	- (N/A)
	Other	1 (0.00)	3 (0.04)	- (N/A)
Sociocultural	Cultural clash for data collection/sharing/linkage	1 (0.00)	2 (0.03)	- (N/A)
	Other	1 (0.00)	2 (0.03)	- (N/A)

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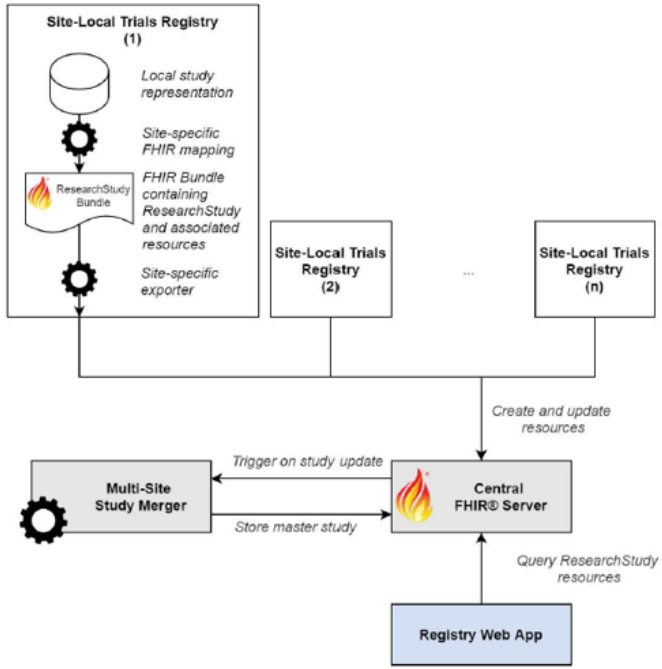
Clinical Trial Registry based on HL7 Standards

HL7 International

Prototypical Clinical Trial Registry Based on Fast Healthcare Interoperability Resources (FHIR): Design and Implementation Study

Christian Gulden[1], MSc; Romina Blasini[2], MSc; Azadeh Nassirian[3], MSc; Alexandra Stein[4], Dipl-Jur; Fatma Betül Altun[5], Dipl-Ing; Melanie Kirchner[6], Dipl-Dokumentarin (FH); Hans-Ulrich Prokosch[1,6], Prof Dr; Martin Boeker[7], Prof Dr



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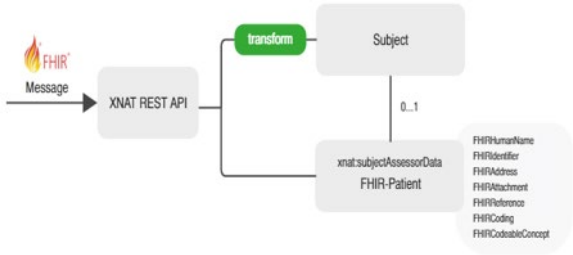
Mapping existing Platforms (XNAT) to Standards

Journal of Medical Systems (2020) 44: 137
https://doi.org/10.1007/s10916-020-01600-y

EDUCATION & TRAINING

Check for updates



Towards Interoperability in Clinical Research - Enabling FHIR on the Open-Source Research Platform XNAT

Maryna Khvastova[1] · Michael Witt[1] · Andrea Essenwanger[2] · Julian Sass[2] · Sylvia Thun[2] · Dagmar Krefting[1,3]

What does XNAT provide?

Full DICOM Integration and Anonymization: Get image data in, and keep PHI out.

Secure Access & Permission Control: You decide who does what with your data.

Integrated Search & Reporting: Report on your image and clinical data together.

Pipeline Processing: Use the power of high-performance computing on your data.

Modular Extensibility: Expand the capabilities of your XNAT to meet your needs.

Developer Community: Benefit from an active and engaged set of XNAT power users.

Source XNAT

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Scientific Background

COVID-19 Questionnaires, surveys, and item-banks: Overview of clinical- and population-based instruments

Carsten O. Schmidt[1,*], Rajini Nagrani[2,*,§], Christina Stange[1], Matthias Löbe[3], Atinkut Zeleke[1] Guillaume Fabre[4], Sofiya Koleva[4], Karine Trudeau[4], Stefan Sauermann[5], Jay Greenfield[6], Claire C Austin[7]; and the RDA-COVID19-WG[8]

- Overview of instruments and resources
- Scoping of content domain on a selection of instruments using the **Maelstrom** taxonomy
- Reuse of existing instruments to make results openly available (machine-readable format)
- Facilitate reuse and maximize comparability of results across studies and countries

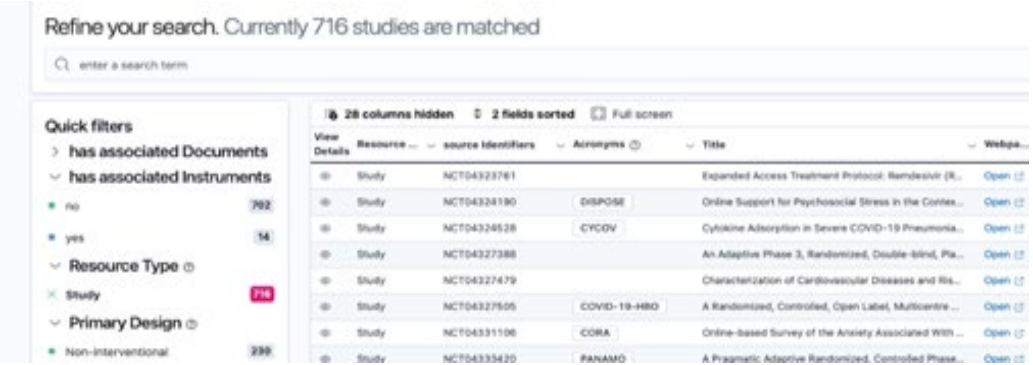
Table 1. Questionnaire instruments: Reference studies

COUNTRY	INSTRUMENTS (Acronym)	ISSUER	TARGET POPULATION	LANGUAGE	COMMENTS
CLINICAL					
Australia	NSW Case questionnaire (NSW)[11]	NSW Government, Australia	Patients	English	
Austria	EMS (EMS)[12]	Federal Ministry of Social Affairs, Health, Care and Consumer Protection	Patients	German	Collects data from doctors and laboratories for national surveillance, feeds into TESSY
Europe	TESSy[13]	European CDC	Patients	English	Surveillance for EU, collects data from surveillance systems in EU member states
Germany	Covid-19 research dataset (GECCO COVID-19)[1]	National Research Network University Medicine, Germany	Patients	German, English	German Corona Consensus (GECCO) item bank of the German National Research Network to study COVID19.
US	Human Infection with 2019 Novel Coronavirus Person Under Investigation (PUI) and Case Report Form (USCDC)[14]	CDC	Patients	English	
Worldwide (Member states of WHO)	Global COVID-19: clinical platform: novel coronavius (COVID-19): rapid version (WHO-CRF)[2]	WHO	Patients	English, French, Russian, Spanish	

NFDI4Health - Task Force COVID-19

Facilitating study and item level browsing for clinical and epidemiological COVID-19 studies

Carsten Oliver SCHMIDT[a,1], Johannes DARMS[b], Aliaksandra SHUTSKO[b], Matthias LÖBE[c], Rajini NAGRANI[d], Bastian SEIFERT[d], Birte LINDSTÄDT[b], Martin GOLEBIEWSKI[e], Sofiya KOLEVA[f], Theresa BENDER[g], Christian Robert BAUER[g], Ulrich SAX[g], Xiaoming HU[e], Michael LIESER[e], Vivien JUNKER[e], Sophie KLOPFENSTEIN[h], Atinkut ZELEKE[a], Dagmar WALTEMATH[a], Iris PIGEOT[d], Juliane FLUCK[b], on behalf of the NFDI4Health Task Force COVID-19



Cohort Browsing

Item Browsing

Access to study resources

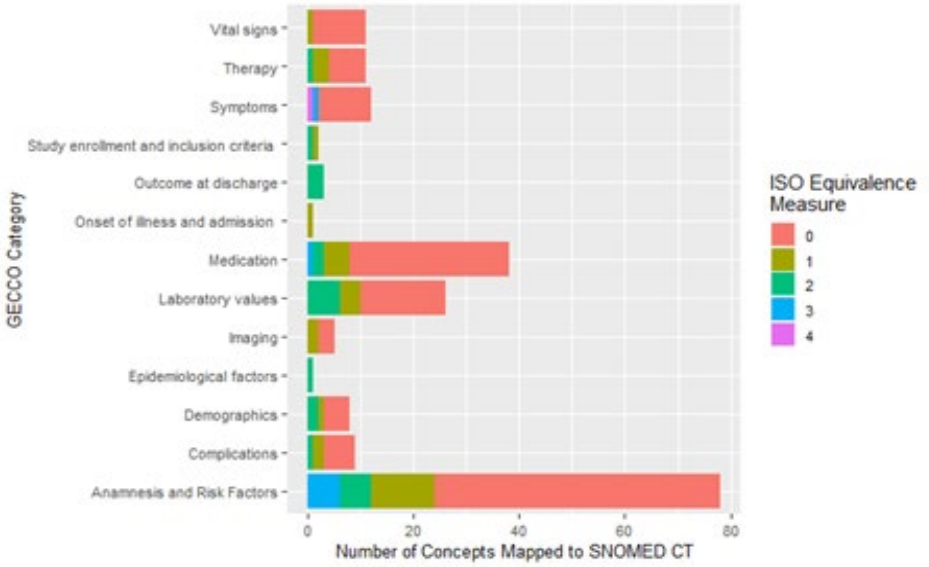
Harmonized COVID-19 research

NFDI4Health - Task Force COVID-19

Evaluating Suitability of SNOMED CT in Structured Searches for COVID-19 Studies

Carina Nina Vorisek[a,1], Sophie Anne Ines Klopfenstein[b], Julian Sass[a], Moritz Lehne[a], Carsten Oliver Schmidt[c], Sylvia Thun[a,d] *on behalf of the NFDI4Health Task Force COVID-19*

Rating	Description	Number of Concepts (%)
0	Equivalent meaning	141 (69)
1	Source is wholly included in target	32 (16)
2	Source is partially included in target	23 (11)
3	Source is mapped however there were many options. Source map is the best comparison rather than an actual correspondence	8 (4)
4	no map possible	1 (0)



Projects: German Corona Consensus Dataset (GECCO)

netzwerk universitäts medizin

TECHNICAL ADVANCE Open Access

Check for updates

The German Corona Consensus Dataset (GECCO): a standardized dataset for COVID-19 research in university medicine and beyond

Julian Sass[1], Alexander Bartschke[2], Moritz Lehne[1], Andrea Essenwanger[1], Eugenia Rinaldi[2], Stefanie Rudolph[2], Kai U. Heitmann[3], Jörg J. Vehreschild[4,5,6], Christof von Kalle[1,2] and Sylvia Thun[1,2,7*]

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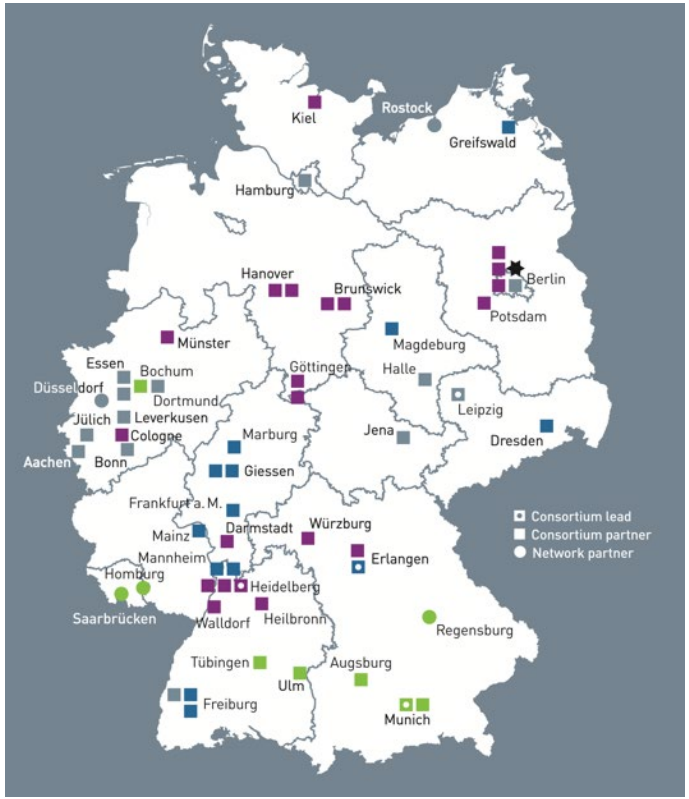
H2020 ORCHESTRA

Connecting European Cohorts

to increase common and effective response to SARS-CoV2 Pandemic

Medical Informatics Initiative



Source: TMFEV

Interoperability working group

The interoperability working group is the platform for agreeing amongst consortia the basis for ensuring interoperability between the proposed data integration centres.

Goals and tasks

The group was established in order to create a platform for coordination and agreement of interoperability between the proposed data integration centres, to plan concrete steps for achieving interoperability, and to agree corresponding minimum requirements.

Activities

The working group members held discussions in physical meetings and conference calls. Several task forces were formed within the working group to produce a number of documents, and to prepare the ground for an agreement within the national steering committee:

- Task force DIC concepts
- Task force core data set
- Task force consent implementation
- Task force process models
- Task force meta data

Results

→ Metadata on data availability, analysis options and collaboration options

→ Core data set

→ Paper summarising key points on interoperability

Working group chairpersons:

- Prof. Dr. Thomas Ganslandt (Universitätsmedizin Mannheim)
- Prof. Dr. Sylvia Thun (Hochschule Niederrhein/BIH)

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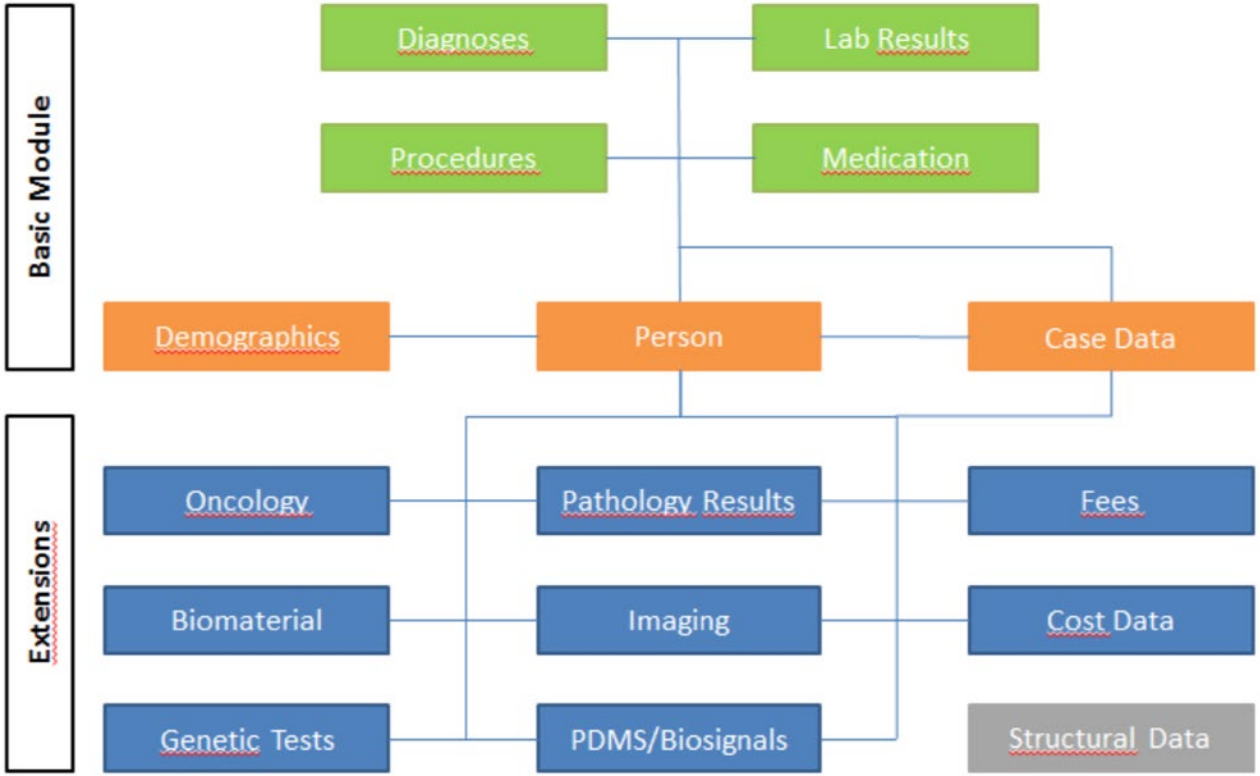
Core Data Set @ International Patient Summary



Source: TMFEV

Genetic Testing Report

GENeALYSE

HL7 FHIR

1+Million Genomes Initiative
Federated, secure, interoperable and privacy-respecting framework and access governance

genomDE
INFRASTRUCTURE + TECHNOLOGY
STANDARDS + QUALITY

HerediVar
Interactive variant database for automated VUS classification

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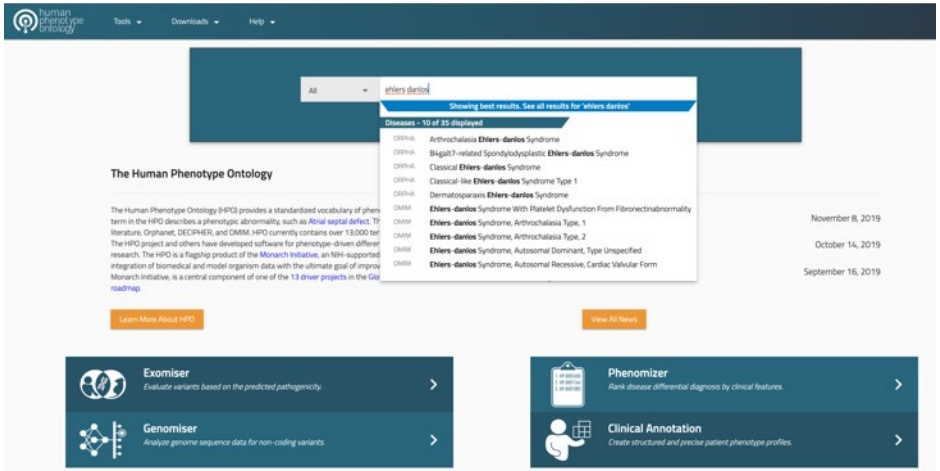
Collaboration on Rare Diseases CORD_MI

Use Case CORD-MI

The Use Case "Collaboration on Rare Diseases" (CORD-MI) is a project involving the four consortia of the Medical Informatics Initiative and involving numerous German university hospitals and partner institutions. The aim is to improve care and research in the field of rare diseases. It builds on the innovation fund projects TRANSLATE-NAMSE and ZSE-DUO as well as the national DIMDI project "Coding of Rare Diseases" and uses the concepts and solutions developed in the Medical Informatics Initiative.



Collaboration on Rare Diseases

It is estimated that approximately four million German citizens are affected by approximately 8,000 known rare diseases. Due to the rarity of each individual disease and the lack of consideration in hospital documentation, no concrete statements on the frequency, distribution and course of the disease are possible to date, which has a negative impact on research, diagnosis and therapy. Based on the National Action Plan for People with Rare Diseases from 2013, various measures have been implemented in Germany to support the coding of rare diseases (DIMDI-SE) and to create better care structures for patients in university hospitals (TRANSLATE-NAMSE, ZSE-Duo). Despite these and other important care and research projects at national, European and international level, it has not yet been possible to establish sustainable structures for a digital data exchange network for rare diseases.

MEDIZIN INFORMATIK INITIATIVE

cord_mi

orphanet

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Dataset HL7 FHIR ‚Condition‘

- **Basismodule**
  - Person
  - Fall
  - Diagnose
    - ICD-10-GM Diagnose kodiert
    - ALPHA-ID kodiert
    - ORPHANET Diagnose kodiert
    - SNOMED Diagnose kodiert
    - Weitere Kodiersysteme
    - Körperstelle
    - Freitextbeschreibung
    - Diagnoseerläuterung
    - Dokumentationsdatum
    - Klinischer Status
    - Klinisch relevanter Zeitraum
    - Feststellungsdatum
  - Prozedur
  - Laborbefund
  - Medikation
- Erweiterungsmodule
- Übergreifende Module
  - POLAR
  - CORD

Condition			0..*	Condition
extension			*0..**	*Extension*
identifier	Σ		0..*	Identifier
clinicalStatus	Σ	?!	1..1	code Binding
verificationStatus	Σ	?!	0..1	code Binding
category			0..1	CodeableConcept Binding
severity			0..1	CodeableConcept Binding
code	Σ		1..1	CodeableConcept
coding	Σ		*0..**	*Coding*
ICD-10-GM	Σ		0..1	Coding Binding
extension			*0..**	*Extension*
system	Σ		1..1	uri Fixed Value
version	Σ		1..1	string
code	Σ		1..1	code
display	Σ		0..1	string
userSelected	Σ		0..1	boolean
text	Σ		0..1	string
bodySite	Σ		0..*	CodeableConcept
subject	Σ		1..1	Reference(Patient)
context	Σ		1..1	Reference(Tumorerkrankun...
onsetDateTime	Σ		0..1	dateTime
abatement[x]			0..1	
assertedDate	Σ		0..1	dateTime
asserter	Σ		0..1	Reference(RelatedPerson | P...
stage			0..1	BackboneElement
summary			0..1	CodeableConcept
assessment	S		*0..**	*Reference(ClinicalImpressio...*
evidence			0..*	BackboneElement
note			0..*	Annotation


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Translational Medicine Depends on Interoperability

FAIR DATA

Conclusion

- Use FAIR principles:
  - **F**indable
  - **A**ccessible
  - **I**nteroperable
  - **R**eusable
- Enhance reusability of scientific data
- Extract maximum benefit from digital data sources
- Allow automatic processing (e.g. AI / machine learning)

This can aid the “democratization” of medicine: making health technologies (globally) accessible, improving healthcare, fostering innovations to enable Translational Medicine

ACKNOWLEDGEMENT

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