

Why Translational Medicine Depends on Interoperability

03-12-2021

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

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](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

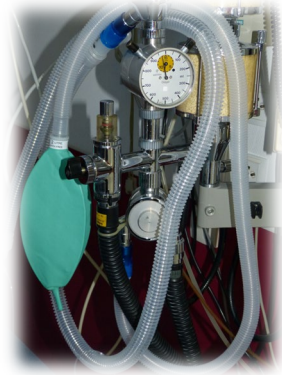
- improve the use of real-world data (e.g. for large-scale observational studies)
- create new research hypotheses (with data mining and AI)
- enable remote development of analysis scripts

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*.

Communication and Connectivity



ISO 11073 SDC

HL7 FHIR

- LOINC
- SNOMED
- IDMP
- UCUM

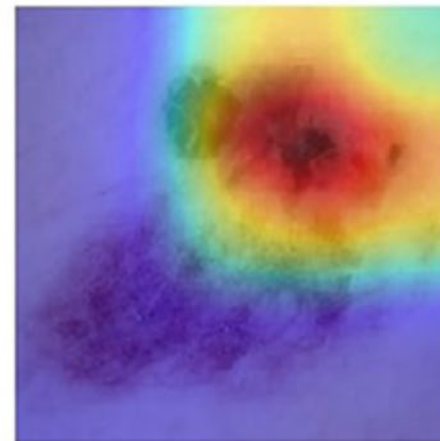
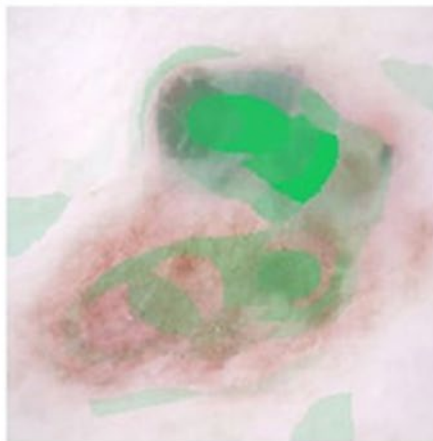


Artificial Intelligence

The Role of DICOM in Artificial Intelligence for Skin Disease

 Liam J. Caffery^{1,2*},  Veronica Rotemberg³,  Jochen Weber³,
 H. Peter Soyer^{2,4},  Josep Malvehy⁵ and  David Clunie⁶

A.



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



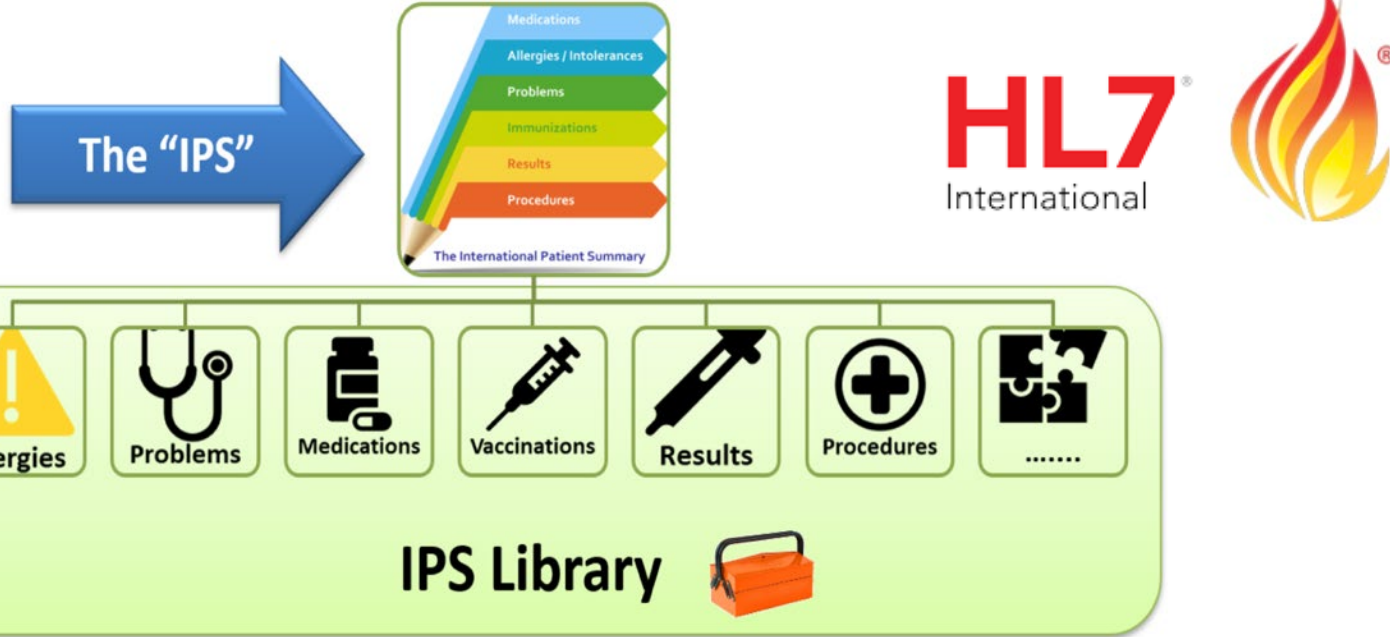
SNOMED
International



Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

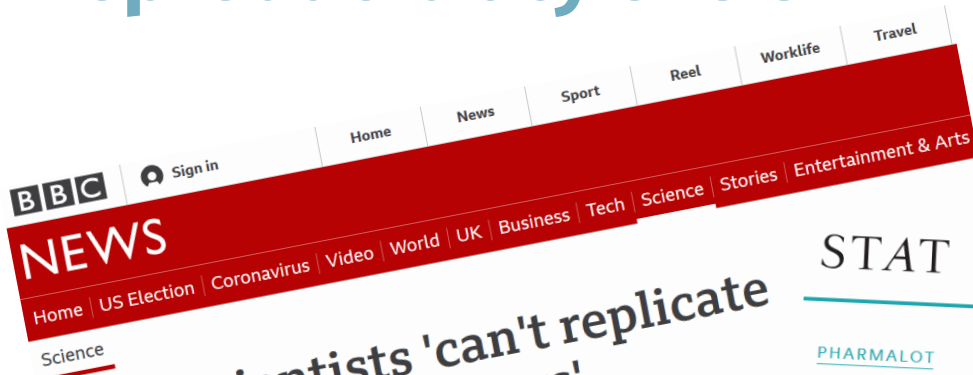
International Patient Summary



HL7[®]
International



Reproducibility crisis



Most scientists 'can't replicate studies by their peers'

By Tom Feilden
Science correspondent, Today programme



[PHARMALOT](#)

Most scientists believe there is a 'crisis' reproducing experiments

By ED SILVERMAN [@Pharmalot](#) / JUNE 1, 2016

[Reprints](#)

Is There a Reproducibility Crisis in Science?

By Nature Video on May 28, 2016

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

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 measurable 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>

FAIR: Findability



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FAIR: Interoperability



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FAIR: Reusability



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

FAIR data vs. Open data



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Open Data: Berlin Kitas

In this notebook I want to explore some data I found on the Berlin Open Data portal. The data source contains information of Kitas (Kindertagesstätte, kindergartens) in Berlin. This is a big topic as finding a spot in a Kita in Berlin is extremely difficult. We first provide an initial exploratory data analysis of the data set, then we merge it with population data to create some geo-location maps.

claimer: In our way we do not expect to answer on how to find a Kita, but rather answer questions about their location, distribution and sizes.

Open Data Notebook

```
import numpy as np
import pandas as pd
import geopandas as gpd
import matplotlib.pyplot as plt
```

Analyse zum Berliner Kita-Datensatz

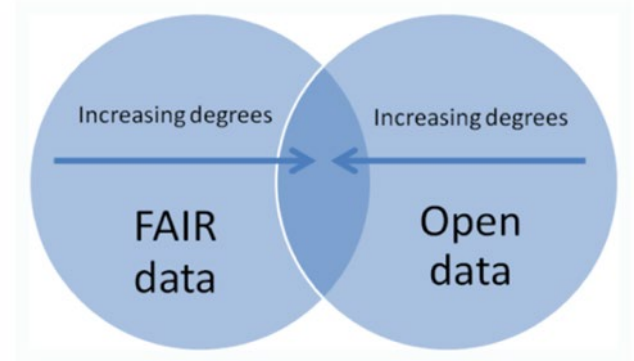
Eine interaktive Analyse der Kitaliste mit Diagrammen, Karten und Quellcode

[» Weitere Informationen](#)

[Analyse zu Kitas in Berlin](#) | [Suchbegriffe des Datenportals](#)

[Bildwechsel anhalten](#)

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



Standards and terminologies



Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.



European Committee for Standardization

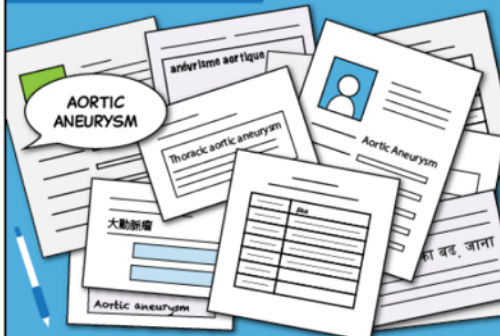


DICOM[®]
Digital Imaging and Communications in Medicine




Phenopackets: Standardizing and Exchanging Patient Phenotypic Data

CURRENTLY...



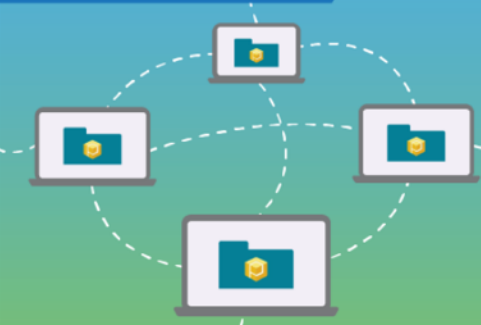
MANY HEALTHCARE SYSTEMS STILL RELY ON MANUAL ENTRY OF PHENOTYPIC DATA, MAKING IT DIFFICULT TO EXCHANGE INFORMATION ELECTRONICALLY IN A STANDARD WAY.

WITH PHENOPACKETS...



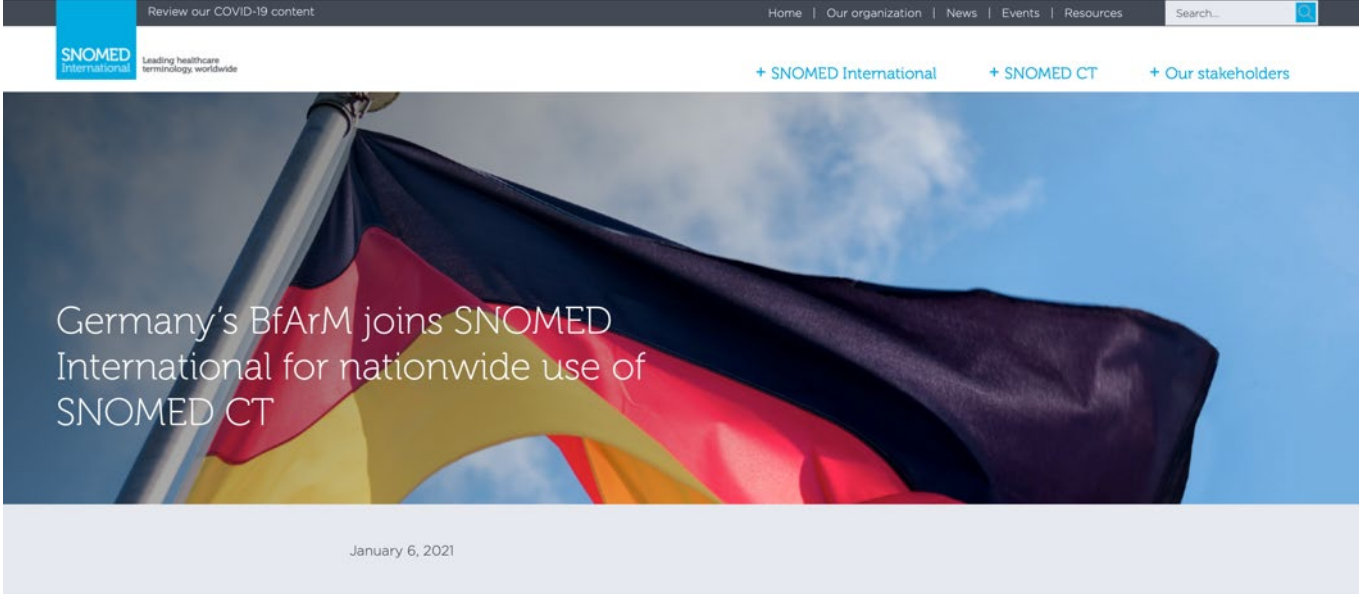
PHENOPACKETS PROVIDES A FORMAT FOR INTEGRATING AN INDIVIDUAL'S GENOMIC DATA AND PHENOTYPIC INFORMATION THAT CAN BE USED FOR ELECTRONIC DATA EXCHANGE.

IN THE FUTURE...



PHENOPACKETS WILL ENABLE A WHOLE NETWORK OF PHENOTYPIC DATA EXCHANGE THAT IMPROVES OUR ABILITY TO UNDERSTAND, DIAGNOSE, AND TREAT DISEASES.

SNOMED - *Systematized Nomenclature of Medicine*



Review our COVID-19 content

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SNOMED International
Leading healthcare terminology worldwide

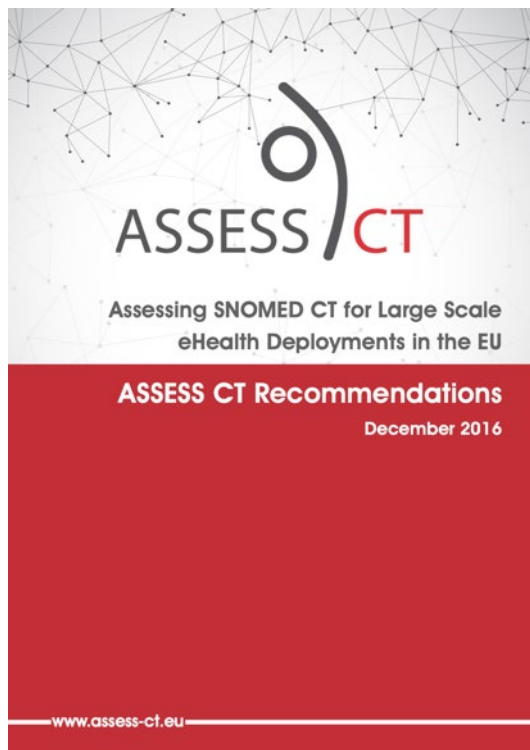
+ SNOMED International + SNOMED CT + Our stakeholders

Germany's BfArM joins SNOMED International for nationwide use of SNOMED CT

January 6, 2021

London, United Kingdom, January 6, 2021 (GLOBE NEWSWIRE) -- Germany's Federal Institute for Drugs and Medical Devices (BfArM) has announced their membership in SNOMED International for national use of SNOMED CT from the beginning of January 2021.

ASSESS CT



Authors / Contributors

Authors - core team

Dipak Kaira, Stefan Schulz, Daniel Karlsson, Robert Vander Stichele, Ronald Cornet, Kirstine Rosenbeck Gøeg, Giorgio Cangioli, Catherine Chronaki, Rainer Thiel, Sylvia Thun, Veli Stroetmann

Editor

Veli Stroetmann

Scientific Coordinator

Sylvia Thun

ASSESS CT Consortium

*University of Applied Science
Hochschule Niederrhein
(Krefeld, Germany)*

- Sylvia Thun
- Heike Dewenter
- Reza Fathollah-Nejad

*empirica Gesellschaft
für Kommunikations- und
Technologieforschung mbH
(Bonn, Germany)*

- Veli Stroetmann
- Rainer Thiel
- Strahil Birov
- Klaus Piesche

*The European Institute for Health
Records (EuroRec)
(Paris, France)*

- Dipak Kaira
- Robert Vander Stichele

*Medical University of Graz
(Graz, Austria)*

- Stefan Schulz
- Jose Antonio Miñarro-Giménez
- Catalina Martínez-Costa

*HL7 International Foundation
(Brussels, Belgium)*

- Giorgio Cangioli
- Catherine Chronaki

*Academisch Medisch Centrum
bij de Universiteit van Amsterdam
(Amsterdam, The Netherlands)*

- Ronald Cornet

*Niizt National IT Institute for Health-
care in the Netherlands
(The Hague, The Netherlands)*

- Pim Volkert

*Linköping University
(Linköping, Sweden)*

- Daniel Karlsson

*Croatian Health Insurance Fund
(Zagreb, Croatia)*

- Vesna Kronstein Kufirin
- Zlatko Boni
- Tatjana Pavešković
- Martina Oresković

*Regione Lombardia, General
Directorate for Health
(Milan, Italy)*

- Marcello Melgara

*INSERM Laboratoire d'Informatique
Médicale et d'Ingénierie des
Connaissances en e-Santé
(Paris, France)*

- Marie-Christine Jaulent

*National Institute for Health and
Welfare
(Helsinki, Finland)*

- Päivi Hämäläinen
- Virpi Kalliokuusi
- Riika Vuokko

*Aalborg University
(Aalborg, Denmark)*

- Kirstine Rosenbeck Gøeg

*Averbis
(Freiburg, Germany)*

- Kornél Markó

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

Layout: Klaus Piesche, Meropi Papageorghe, Strahil Birov (empirica GmbH)

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

LOINC@BfArM

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Knowledge Base

Search or browse this collection of documentation to get instant answers to your LOINC questions.

SARS-COV-2 and COVID-19 INFORMATION

[Full list of LOINC terms related to SARS-CoV-2/COVID-19](#)

- Single source for all SARS-CoV-2/COVID-19 LOINC content
- Includes both released and pre-release terms
- Content is updated as new terms are created and pre-released
- Includes ability to filter and export

[Guidance for mapping to SARS-CoV-2 LOINC terms](#)

- LOINC terms for commercial in vitro diagnostics (IVD) test kits
- Help with choosing the right LOINC
- Frequently Asked Questions
- External links related to COVID-19
- Video webinar

The international standard for identifying health measurements, observations, and documents.

Reference labs, healthcare providers, government agencies, insurance companies, software and device manufacturers, researchers, and consumers from around the globe use LOINC to identify data and move it seamlessly between systems.

It's free, but invaluable.

[Get Started](#)



[Register Now](#)

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.

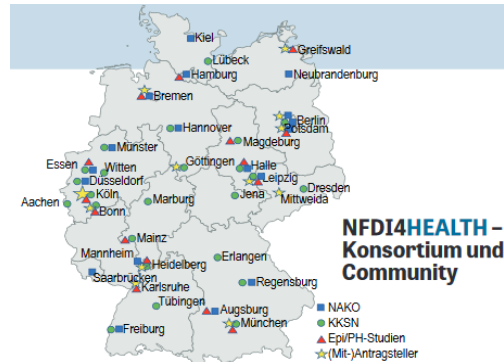
Logical Observation Identifiers Names and Codes (LOINC) is a common language (set of identifiers, names and codes) for identifying health measurements, observations and documents. The use of LOINC with PROMs facilitate the collected data to make it further reusable for extended use across health information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

Projects: NFDI4Health

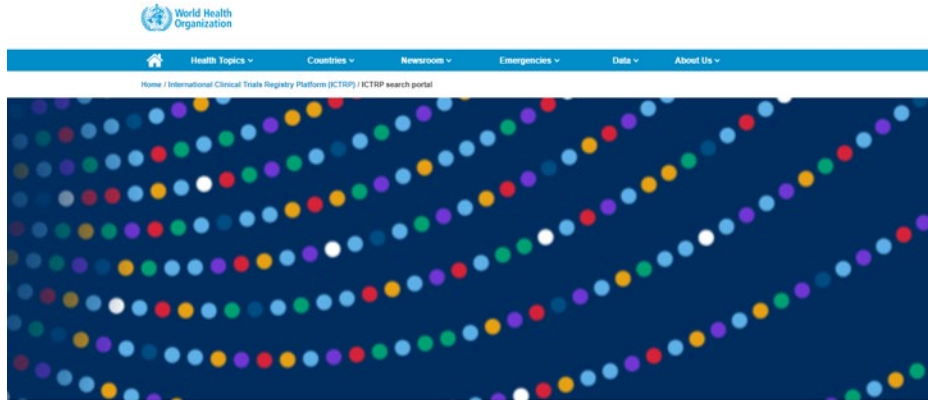


NFDI4Health – Aims



- 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



ICTRP Search Portal

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

Deutsches Register
Klinischer Studien
German Clinical
Trials Register

Home

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• Cooperation with ethics committees

Search trials

Register trials

Register user

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Description of entry fields

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Privacy

Web accessibility

DRKS - German Clinical Trials Register

The DRKS is an open access online register for clinical trials conducted in Germany, which allows all users to [search](#), [register](#) and share information on clinical trials.

The DRKS now contains well over 10,000 studies. Currently, around 1500 studies are added annually.

You will find basic information like the title, short descriptions, inclusion and exclusion criteria, status and outcomes on every trial.

In order to search for clinical trials you can either enter a search term into the search box or you can use the [extended search](#) to refine your results. This allows for example to search specifically for trials currently recruiting patients.

The DRKS is a non-profit organization and is located at the [Federal Institute for Drugs and Medical Devices \(BfArM\)](#). BfArM is a governmental institution within the scope of the [Federal Ministry of Health \(BMG\)](#).

The DRKS is an approved Primary Register in the [WHO](#) network since October 2008 and thus meets the requirements of the [ICMJE](#).

Research-based physicians are obliged by their professional code of conduct to observe the Declaration of Helsinki. In the currently valid version of 2013, §35 states: "Every research study involving human subjects must be registered in a publicly accessible database before recruitment of the first subject." Therefore, please check (if necessary together with the responsible ethics committee) whether you need to register your study in a publicly accessible study register (such as DRKS).

For DRKS, trials must meet the following requirements:

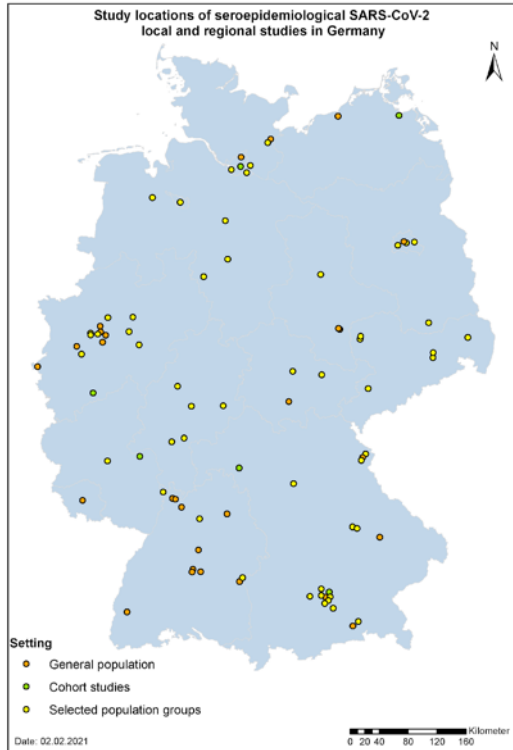
- It must address a health issue
- It must be a human study

For further details, see our FAQ "[Which trials can/should be registered with DRKS?](#)" Independent of this and of the requirements of medical journals, please also observe any national regulations.

Last Modified: 02-05-2021

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

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COVID-19 Research Registry

Home Basic Virology Treatment **Epidemiology** Clinical Diagnostics Prevention General Reviews

Epidemiology

Find the latest research on disease outbreaks, data modeling, statistics and prevalence of COVID 19:

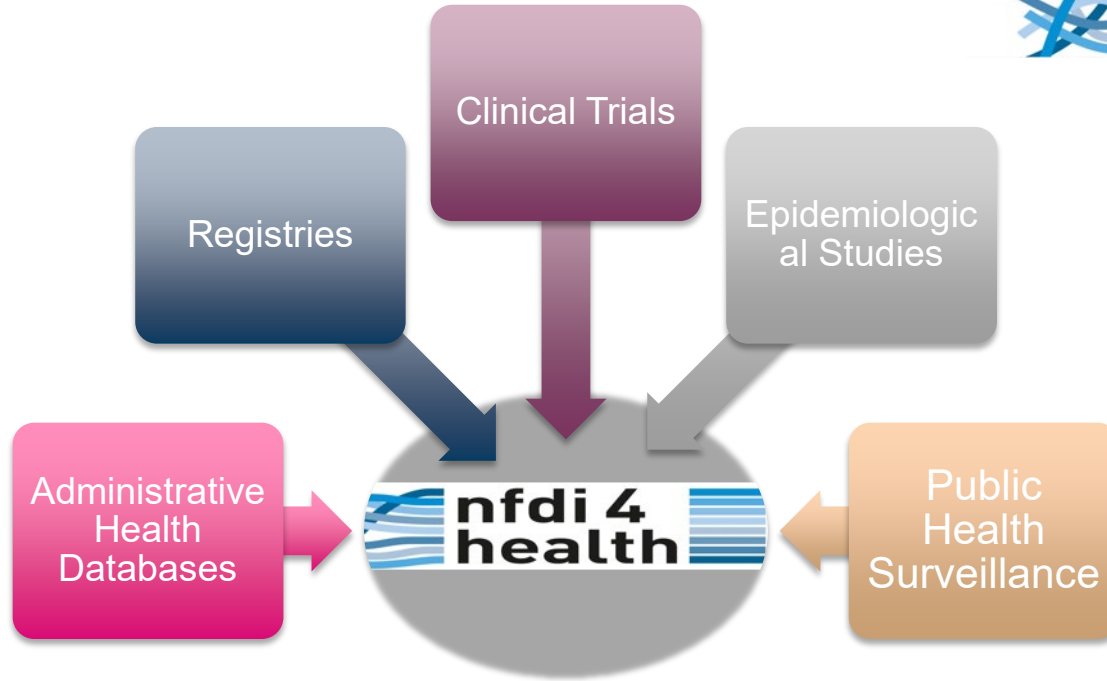
1. Emergence & Spread
2. Data Modeling & Statistics

*Check out additional data and modeling resources at [Mathematica](#).



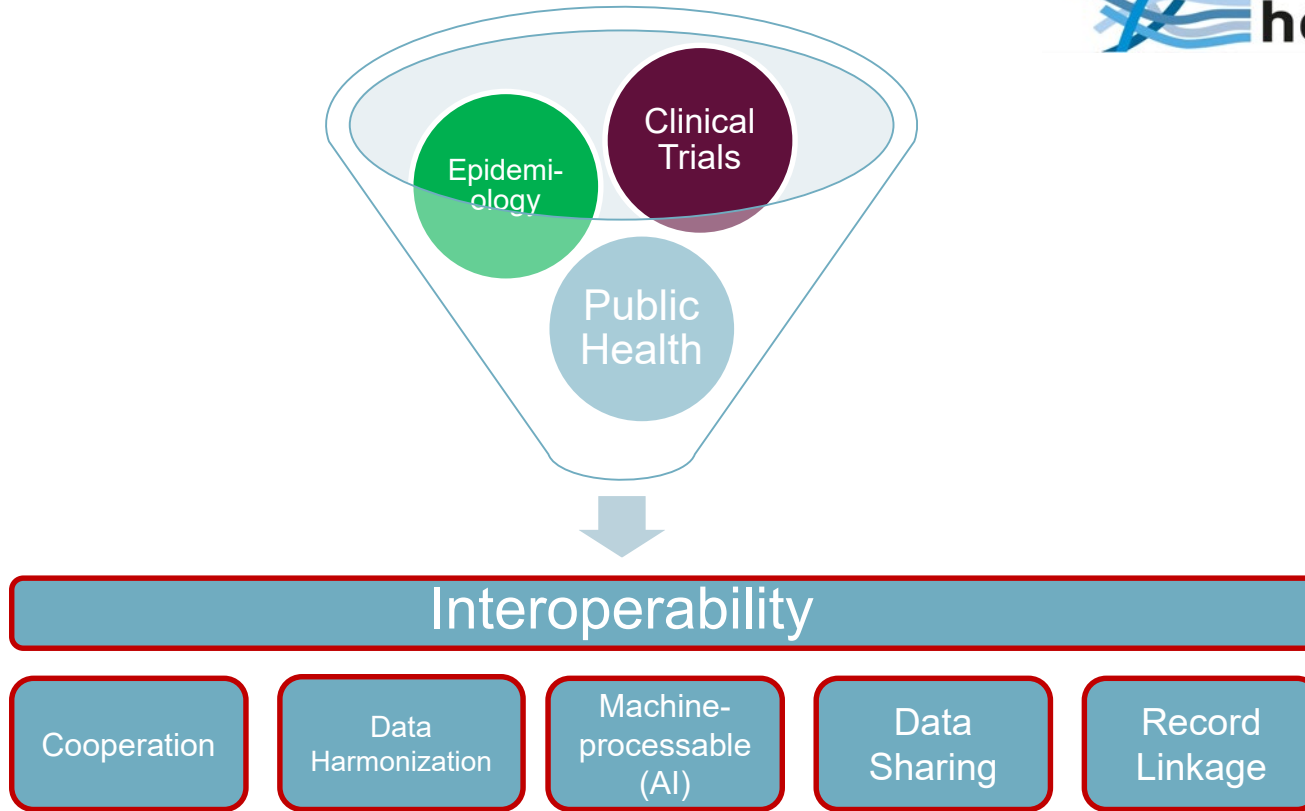
<https://asm.org/COVID/COVID-19-Research-Registry/Epidemiology>

NFDI4Health – Aims

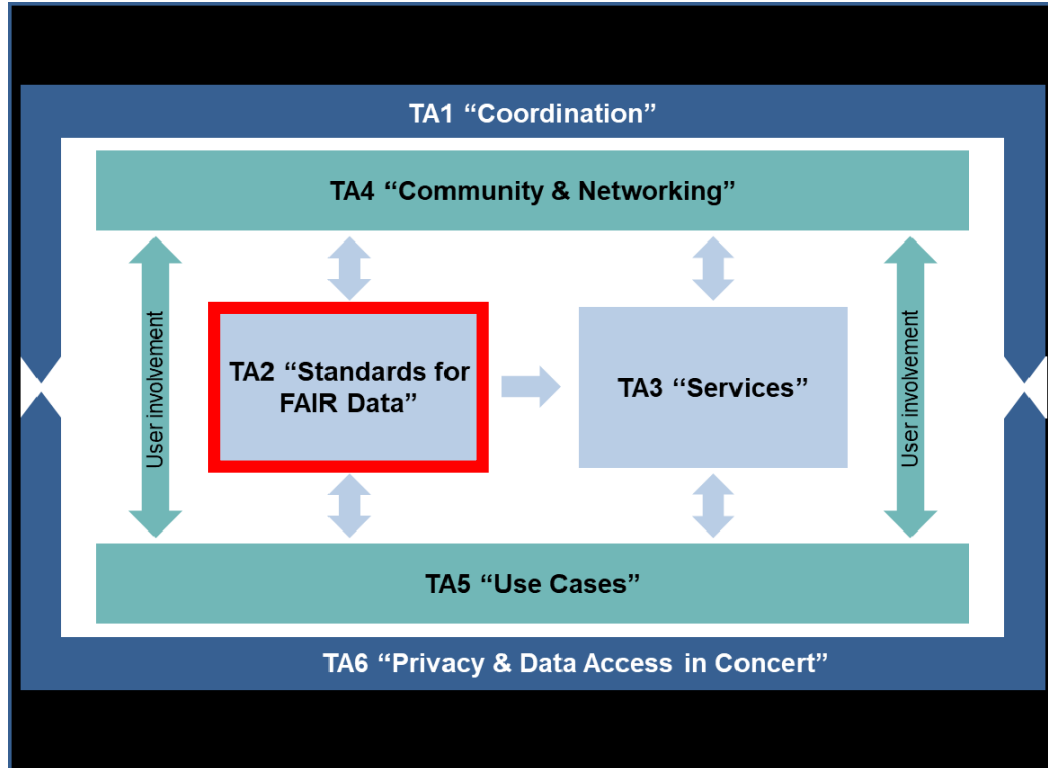


The resulting infrastructure will build bridges between user communities and data holders from epidemiology, public health and clinical trials

NFDI4Health – Aims



Task Area Core-Unit eHealth und Interoperability

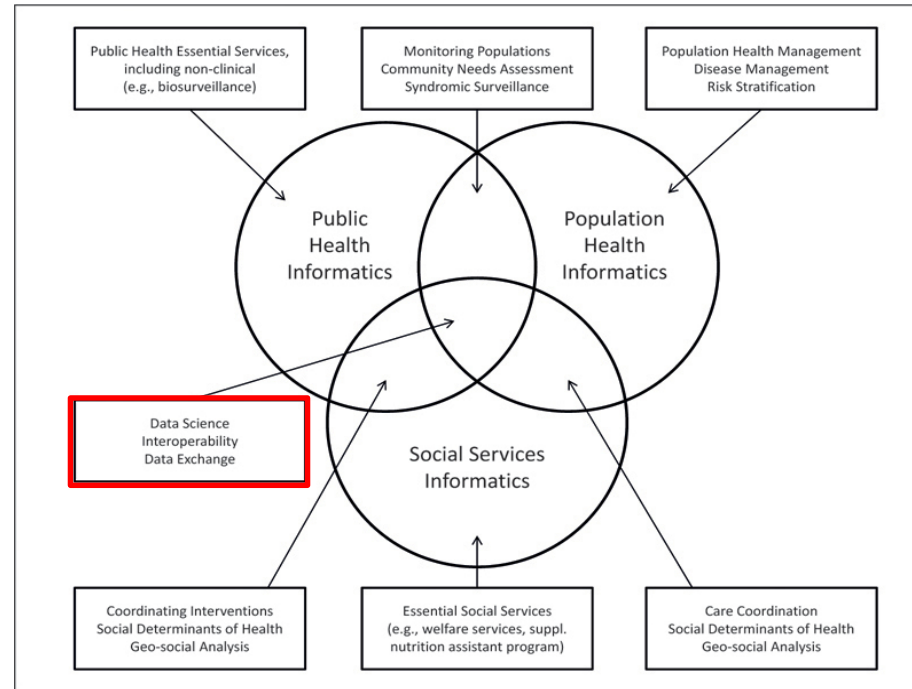


- **Interoperability-Roadmap**
- Identification of relevant **Standards, Terminologies and Ontologies**
- Value Set Implementation
- **USE of SNOMED CT/ LOINC**
- Development of Implementation Guidelines (e.g. HL7 FHIR)
- Development of data models

Scientific Background






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

Roland Gamache, Hadi Kharrazi, Jonathan P. Weiner



Scientific Background: Harmonization

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

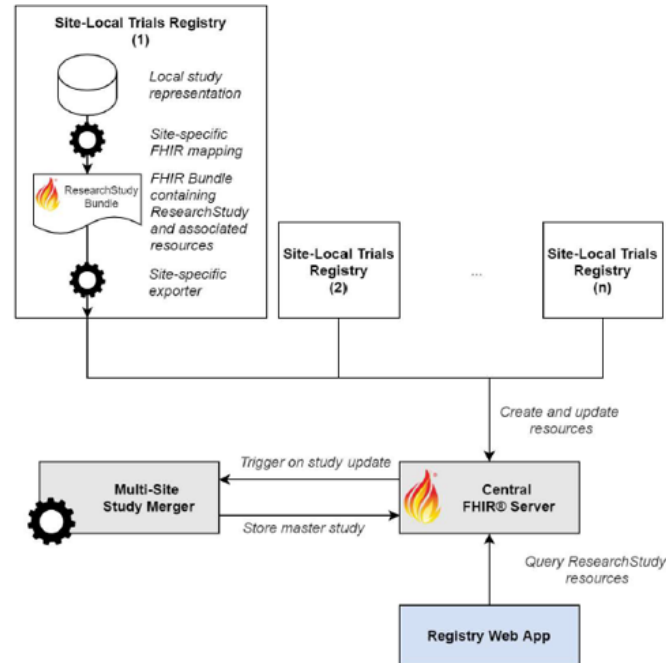
Lester Darryl Geneviève  , Andrea Martani , Maria Christina Mallet, Tenzin Wangmo , Bernice Simone Elger 

Cluster	Sub-cluster	Countries involved in projects		
		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)

Clinical Trial Registry based on HL7 Standards

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

Christian Gulden¹, MSc; Romina Blasini², MSc; Azadeh Nassirian³, MSc; Alexandra Stein⁴, Dipl-Jur; Fatma Betül Altun⁵, Dipl-Ing; Melanie Kirchner⁶, Dipl-Dokumentarin (FH); Hans-Ulrich Prokosch^{1,6}, Prof Dr; Martin Boeker⁷, Prof Dr



Mapping existing Platforms (XNAT) to Standards

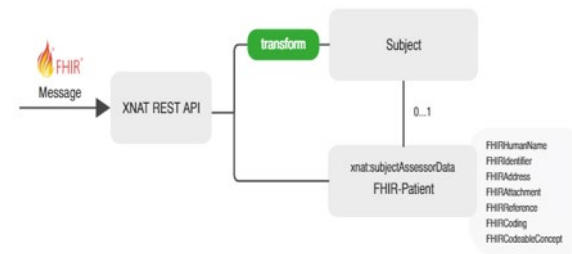
Journal of Medical Systems (2020) 44: 137
<https://doi.org/10.1007/s10916-020-01600-y>

EDUCATION & TRAINING



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

Maryna Khvastova¹ · Michael Witt¹ · Andrea Essenwanger² · Julian Sass² · Sylvia Thun² · Dagmar Krefting^{1,3}



What does XNAT provide?



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



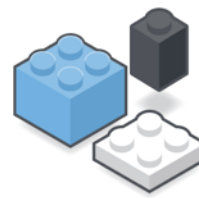
Secure Access & Permission Control: You decide who does what with your data.



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

Scientific Background

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

Carsten O. Schmidt^{1,*}, Rajini Nagrani^{2,*§}, Christina Stange¹, Matthias Löbe³, Atinkut Zeleke¹ Guillaume Fabre⁴, Sofiya Koleva⁴, Karine Trudeau⁴, Stefan Sauermann⁵, Jay Greenfield⁶, Claire C Austin⁷; and the RDA-COVID19-WG⁸

- 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 coronavirus (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

Refine your search. Currently 716 studies are matched

Quick filters

- has associated Documents
- has associated Instruments
 - no: 702
 - yes: 14
- Resource Type
 - Study: 716
 - Primary Design
 - Non-interventional: 239

View	Resource	source identifiers	Aliases	Title	Website
Study	NCT04323761			Expanded Access Treatment Protocol: Remdesivir (R...	Open
Study	NCT04324190	DISPOSE		Online Support for Psychosocial Stress in the Contex...	Open
Study	NCT04326528	CYCOV		Cytokine Adsorption in Severe COVID-19 Pneumonia...	Open
Study	NCT04327388			An Adaptive Phase 3, Randomized, Double-blind, Pla...	Open
Study	NCT04327479			Characterization of Cardiovascular Diseases and Ris...	Open
Study	NCT04327505	COVID-19-HBO		A Randomized, Controlled, Open Label, Multicentre ...	Open
Study	NCT04331106	CORA		Online-based Survey of the Anxiety Associated With ...	Open
Study	NCT04335420	PANAMO		A Pragmatic Adaptive Randomized, Controlled Phase...	Open

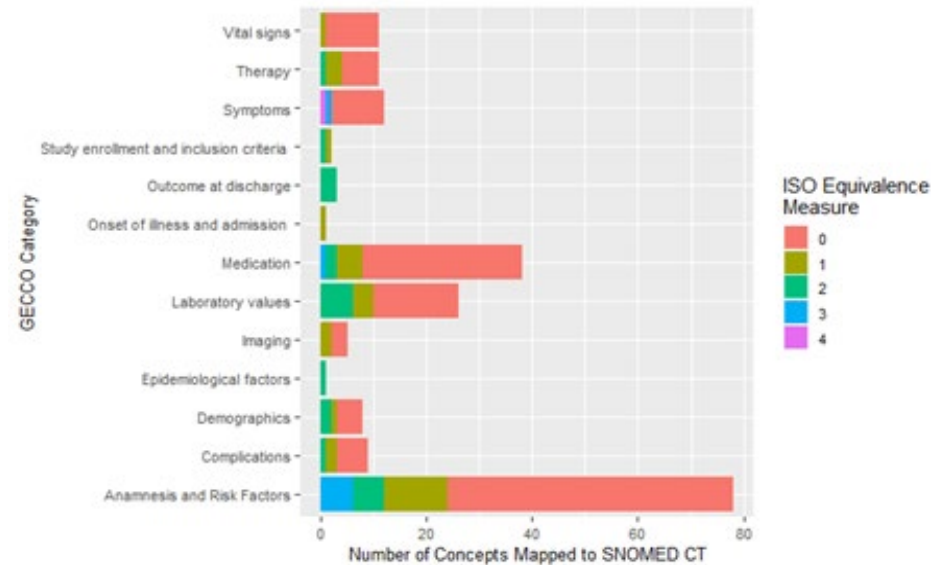
NFDI4Health - Task Force COVID-19



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,4} *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)

TECHNICAL ADVANCE

Open Access

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



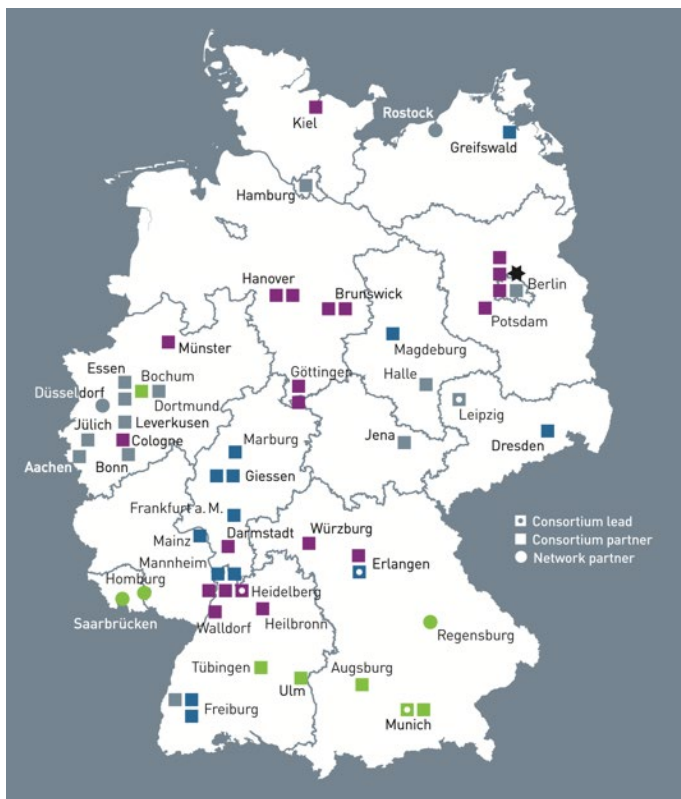
Julian Sass¹, Alexander Bartschke², Moritz Lehne¹, Andrea Essenwanger¹, Eugenia Rinaldi², Stefanie Rudolph², Kai U. Heitmann³, Jörg J. Vehreschild^{4,5,6}, Christof von Kalle^{1,2,7*} and Sylvia Thun^{1,2,7*}



H2020 ORCHESTRA



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

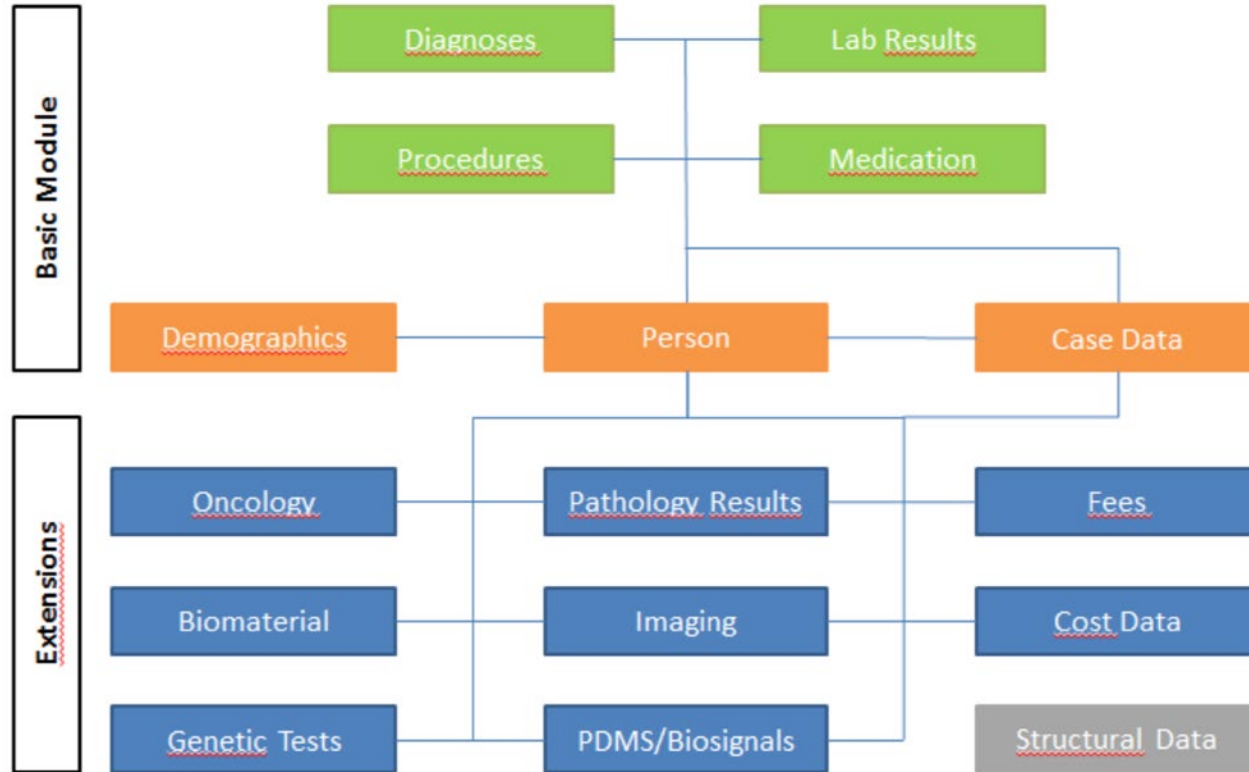
Results

- Metadata on data availability, analysis options and collaboration options
- Core data set
- Paper summarising key points on interoperability

Working group chairpersons:

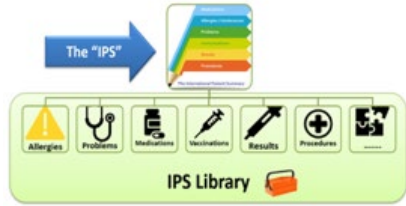
- Prof. Dr. Thomas Ganslandt (Universitätsmedizin Mannheim)
- Prof. Dr. Sylvia Thun (Hochschule Niederrhein/BIH)

Core Data Set @ International Patient Summary



Source: TMFEV

Genetic Testing Report



Declaration for delivering cross-border access to genomic database

- 1 million **genomes accessible** in the EU by 2022
- Linking access** to existing and future genomic database across the EU
- Providing a sufficient scale for **new clinically impactful** associations in research

genomDE:
Nationale
und europäische
Genominitiativen

GERMAN CONSORTIUM
Hereditary Breast
and Ovarian Cancer

established by German Cancer Aid

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

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



cord_mi

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.

The screenshot shows the Human Phenotype Ontology (HPO) website interface. At the top, there is a navigation bar with 'Tools', 'Downloads', and 'Help' menus. A search bar contains the text 'ehlers danlos', and a dropdown menu displays a list of 10 related HPO terms, including 'Arthrochalasia Ehlers-Danlos Syndrome', 'Biglycan-related Spondyloepiphyseal Ehlers-Danlos Syndrome', and 'Classical Ehlers-Danlos Syndrome'. Below the search results, there is a section titled 'The Human Phenotype Ontology' with a brief description of the HPO project. On the right side, there are dates for updates: 'November 8, 2019', 'October 14, 2019', and 'September 16, 2019'. At the bottom, there are four tool cards: 'Exomiser' (Evaluate variants based on the predicted pathogenicity), 'Genomiser' (Analyze genome sequence data for non-coding variants), 'Phenomizer' (Rank disease differential diagnosis by clinical features), and 'Clinical Annotation' (Create structured and precise patient phenotype profiles).

orphanet

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	0..*	Reference(ClinicalImpressio...)
evidence	0..*	BackboneElement
note	0..*	Annotation

```

    "url": "http://fhir.de/StructureDefinition/icd-10-gm-haupt-kreuz",
    "valueCoding": {
      "system": "http://fhir.de/CodeSystem/dimdi/icd-10-gm",
      "version": "2019",
      "code": "E10.30"
    }
  },
  {
    "url": "http://fhir.de/StructureDefinition/icd-10-gm-stern",
    "valueCoding": {
      "system": "http://fhir.de/CodeSystem/dimdi/icd-10-gm",
      "version": "2019",
      "code": "H36.0"
    }
  }
],
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"version": "2019",
"code": "E10.30† H36.0*",
"display": "Diabetische Retinopathie"

```

Translational Medicine Depends on Interoperability



FAIR DATA



Conclusion

- Use FAIR principles:
 - Findable
 - Accessible
 - Interoperable
 - Reusable
- 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

Team of the Core Unit eHealth and Interoperability



Prof. Dr. Sylvia Thun
Director of eHealth and interoperability

Phone: +49 30 450 5430 71
E-mail: sylvia.thun@bihealth.de



Alexander Bartschke
Medical Information Manager

E-mail: alexander.bartschke@charite.de



Andrea Essenwanger
Medical Informatics

E-mail: andrea.essenwanger@bihealth.de



Dr. Moritz Lehne
Health Data Scientist /
Wissenschaftlicher Mitarbeiter

E-mail: moritz.lehne@bihealth.de



Eugenia Rinaldi
Highmed

E-mail: eugenia.rinaldi@charite.de



Julian Saß
Semantic and Syntactic Interoperability

E-mail: julian.sass@bihealth.de



Dr. Josef Schepers
Medical Data Scientist /
Coordinator Medical Informatics

E-mail: josef.schepers@bihealth.de



Dr. Judith Albrecht
Scientific Project Manager
for CORD-MI

E-mail: judith.albrecht@charite.de



Anna Trelinska-Finger
Research Assistant for
EUCANcan

E-mail: anna.trelinska-finger@charite.de

Sophie Klopfenstein
Research Assistant for
NFDI4Health Task-Force
Covid-19

E-mail: sophie.klopfenstein@charite.de

Michael Rusongoza Muzoora
Research Assistant for NUM-
COMPASS

E-mail: michael.muzoora@charite.de

Naveen Moses Raj Rajkumar
Research Assistant for
AIQNET

E-mail: naveen.rajkumar@bihealth.de

Nabil El-Badawi
Research Assistant for NUM-
COMPASS

E-mail: nabil.el-badawi@charite.de

Rasim Atakan Poyraz
Research Assistant for NUM-
COMPASS

E-mail: rasim-atakan.poyraz@charite.de

Alonso Essenwanger
Student Assistant

E-mail: alonso.essenwanger@charite.de

Caroline Stellmach
Research Assistant for
CODEX

E-mail: caroline.stellmach@charite.de

Dr. Carina Vorisek
Research Assistant for
NFDI4Health

E-mail: carina-nina.vorisek@charite.de

Steffen Sander
Research Assistant for
CORD-MI

E-mail: steffen.sander@charite.de

Thomas Haese
Research Assistant for
NAPKON

E-mail: thomas.haese@charite.de

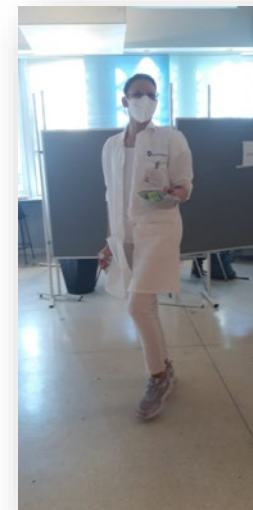
Hassan Sami Adnan
Research Assistant for
NAPKON

E-mail: hassan.adnan@charite.de

Sally Lucie Brose
Research Assistant for
NAPKON

E-mail: sally-lucie.brose@charite.de

Sophie Albrecht
Student Assistant



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

Berlin Institute of Health at Charité – Universitätsmedizin
Berlin, Charitéplatz 1
10117 Berlin, Germany
sylvia.thun@charite.de

www.bihealth.org

