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Bringing Communities Together: Mapping the Investigation-Study- Assay-Model to Fast Healthcare Interoperability Resources

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Introduction – (Meta-)data of health datasets

- Publication as objective of the European Commission and the Federal Government of Germany
- Need of common data infrastructures
- Fulfilment of FAIR principles



National Research Data Infrastructure for Personal Health Data (NFDI4Health):

- Collaboration between health related communities
- Infrastructure based on existing platforms, and provision of services
- Creation of a metadata schema compatible with different registries



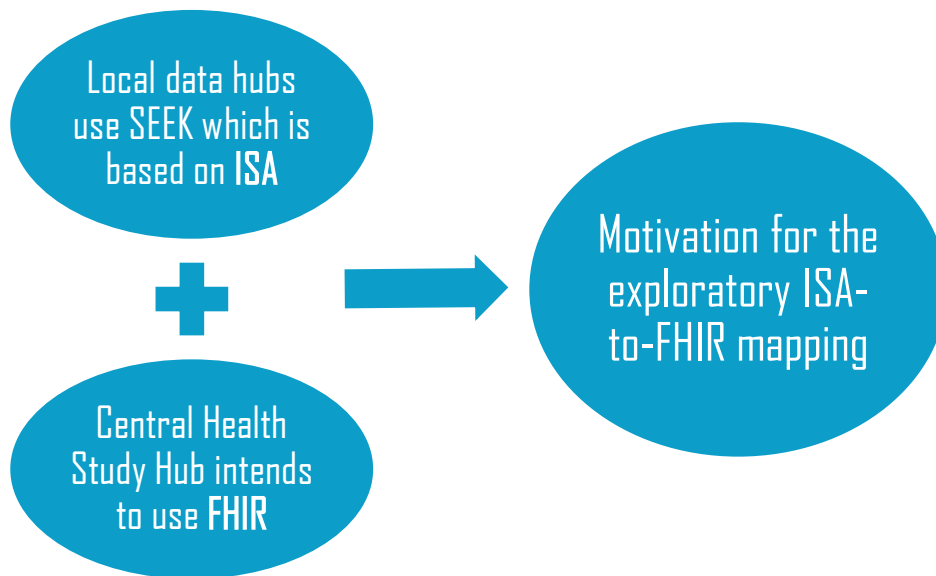
Introduction – Standards and Models

- Diverse, sometimes domain-specific standards, terminologies, minimum reporting guidelines, information models...

Models	ISA – Investigation, Study, Assay	FHIR – Fast Healthcare Interoperability Resources
Creator & Communities	ISA Commons community ; Biosciences	Health Level 7 ; Health(care)
Structure	Metadata framework composed of the ISA model and a suite of tools	Modular information technology model with standard information blocks (resources), extension and referencing mechanisms
Formats	Tab or JSON	XML, JSON, or Turtle



Introduction – Motivation





Methods

Analysis

- ISA Model and Serialization Specifications (Version 1.0, 2016.10.28)
- ISA Example data

Inclusion

- All elements grouped as „properties“ (except „additional properties = false“) in the ISA Model and Serialization Specification

Addition

- *Where available:* Information regarding data types, formats, references, and descriptions
- Cardinalities derived from ISA JSON schemas based on JSON schema specification

Creation

- Overview of the ISA JSON Schemas as FHIR Logical Models (LM) to ease the ISA-to-FHIR mapping

Mapping

- **Source:** Previously created LM with ISA properties prioritized 1) that could be covered by standard FHIR elements or 2) that are necessary for referencing purpose and **Target:** FHIR® Release 4 (R4; v4.0.1 generated on November 11, 2019)

Discussion

- Internal discussions regarding correctness and conciseness, disagreements solved based on votes
- Qualitative analysis



Results

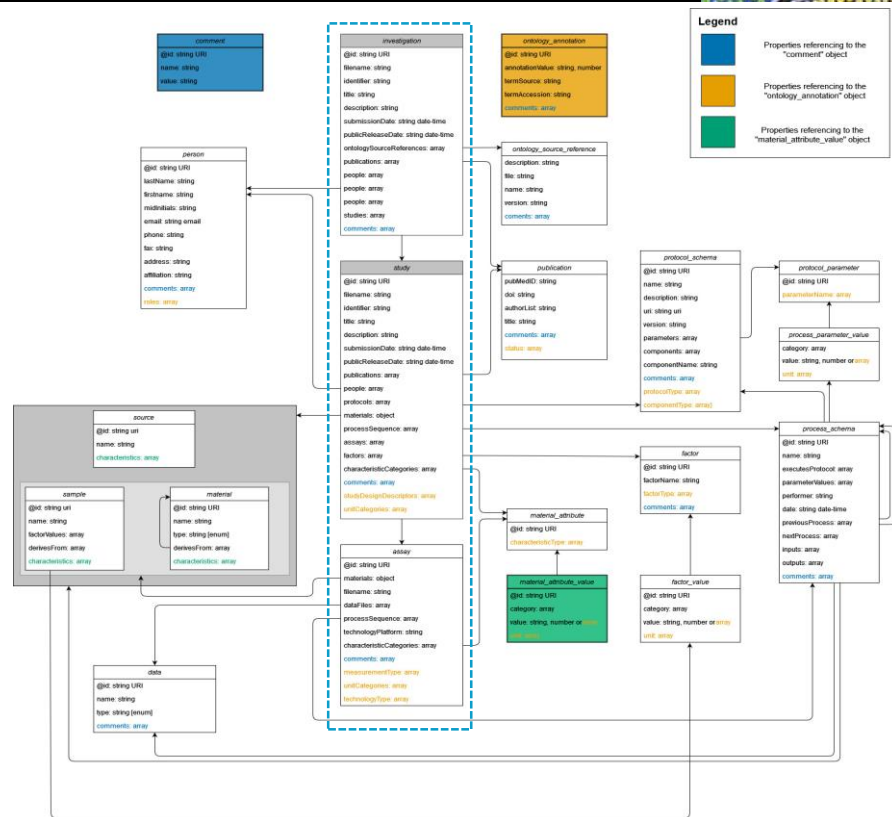
- Creation of an Implementation guide (available on Simplifier)
 - Schematic overview of the JSON schemas
 - ISA FHIR Logical Models
 - Mappings to FHIR
- Qualitative analysis and description of challenges





Results - Overview

- 20 ISA JSON schemas in total – Investigation, Study and Assay as central schemas
- Some ISA JSON schemas are referenced by properties in other schemas (e.g., comment)





Results – Snapshot of a Logical Model

This schema implements Study from the ISA Abstract Model.

isa-lm-study	isa-lm-study
Study	0..* BackboneElement
@id	0..1 uri
filename	0..1 string
identifier	0..1 string
title	0..1 string
description	0..1 string
submissionDate	0..1 dateTime
publicReleaseDate	0..1 dateTime
publications	0..* Reference(ISA LM Publication)
people	0..* Reference(ISA LM Person)
studyDesignDescriptors	0..* Reference(ISA LM Ontology Annotation)
protocols	0..* Reference(ISA LM Protocol)
materials	0..1 BackboneElement
processSequence	0..* Reference(ISA LM Process)
assays	0..* Reference(ISA LM Assay)
factors	0..* Reference(ISA LM Factor)
characteristicCategories	0..* Reference(ISA LM Material Attribute)
unitCategories	0..* Reference(ISA LM Ontology Annotation)
comments	0..* Reference(ISA LM Comment)

135 ISA properties in total
in the LMs

Element Id

isa-lm-study.Study.protocols

Short description

protocols

Definition

protocols

Data Type

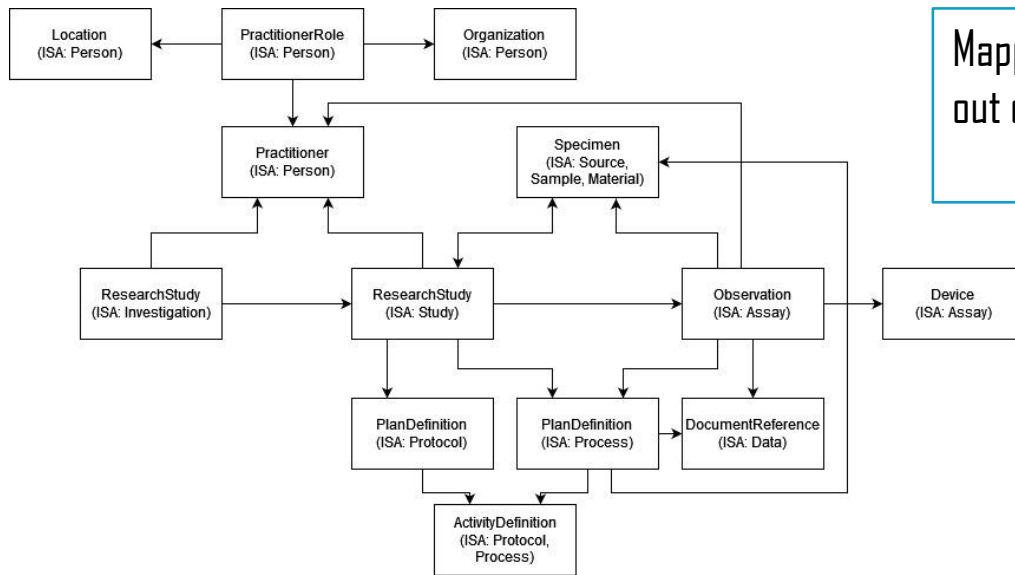
Reference(ISA LM Protocol)

Mappings

- FHIR:ResearchStudy.protocol



Results - Network of FHIR resources



Mapping possible for 102 out of 135 properties



Results - Challenges

- **Documentation of the ISA model**
 - Inconsistencies between abstract model and specifications
 - Different documentation depending on the ISA specification
 - Sparse documentation of cardinalities and descriptions in ISA JSON specification
- **Data types and cardinalities in ISA v.s. FHIR**
 - Generic data types in ISA
 - Cardinalities derived from ISA JSON schemas based on the JSON schema specification



Results - Challenges

- **Modelling approaches in ISA v.s. FHIR**
 - ISA: Information stored in generic schemas referencing to each other
 - FHIR: same information might be included in a single resource
- **Representation of studies and related activities in FHIR**
 - Study processes and associated measures are not well represented in R4
 - “Observation” might not be suitable in studies
 - Other resources to be considered depending on methods / procedures



Discussion

- **First ISA-to-FHIR Mapping:**
 - **102/135 ISA properties could be mapped to FHIR**
 - more than one third with extensions (\neq 80:20 rule of FHIR)
 - ISA properties not mapped were either too generic or “not necessary” in FHIR
 - Eight additional elements would be required in FHIR when profiling
 - Tried to stick to **ISA’s modularity while mapping precisely**



Discussion

- **ISA's genericity** allows to store metadata from different communities but impedes a precise mapping to FHIR
 - Solving open questions, and further analyses planned
- **Emerging use of FHIR in research**
 - Refinements of ResearchStudy, Observation and definition resources, or creation of new resources suggested
 - Modifications in R5 seem to improve the ISA-to-FHIR mapping



Conclusion and Outlook

- Mapping ISA to FHIR is **mostly possible**.
 - **Refinement** necessary after **clarification** with the **ISA commons group** and analyses of further example data.
- Findings of this study will be implemented in NFDI4Health' FHIR profiling activities of its metadata schema
- An **ISA-to-FHIR conversion tool** could help fostering (meta-)data exchange and export between scientific domains



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Thank you for your attention!

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