Table 8. Potentially suitable standards for NFDI4Health’ use cases – current state

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| --- | --- | --- | --- |
| **Name** | **Interoperability Layer and/or Type of Standard** | **Description** | **Use Case** |
| Fast Healthcare Interoperability Resources (FHIR) | Syntactic ; enables semantic interoperability | Standard to exchange information generated in healthcare (processes) using standard resources and extensions.  Besides the resources ResearchStudy, DocumentReference and Questionnaire used to map NFDI TF 19’s MDS, FHIR can be used e.g. for imaging (ImagingStudy), to document consent (Consent[[1]](#endnote-1)) or provenance (Provenance[[2]](#endnote-2)), or to control the quality of measures (Measure[[3]](#endnote-3)). | All |
| Systematized Nomenclature of Medicine - Clinical Terms (SNOMED CT) | Semantic ; Terminology | Extensive comprehensive health(care) terminology available in several languages, used in more than eighty countries and with mappings to other international standards. SNOMED CT is included in several information models or other standards such as OMOP CDM or FHIR[[4]](#endnote-4).  For T5.1 e.g. the collaboration between the Academy of Nutrition and Dietetics with the goal to include the Nutrition Care Process Terminology (NCPT) in SNOMED CT’s International Edition might be of interest[[5]](#endnote-5). | All |
| LOINC | Semantic ; Nomenclature | Identifies health measurements, observations, and documents.  LOINC is included in several information models or other standards such as OMOP CDM or FHIR. | All but T5.3 |
| PICO Ontology | Semantic ; Ontology | Model established by Cochrane to draft research questions, search strategies, and to describe studies or meta-analyses by gathering information on “Patient, Population or Problem”, “Intervention”, “Comparison” and “outcome”[[6]](#endnote-6). Different vocabularies and terminologies have been specified to annotate the content[[7]](#endnote-7). | T5.1, T5.2 |
| ISO 8000 | Standard for Data Quality | Is the global standard for “Data Quality and Enterprise Master Data” used to describe features and specify requirements to exchange in a standardized way master data among business partners. Can also be used in healthcare. | All |
| ISO/IEC 11179 | Standard for Metadata Registries | Is the global standard for “Metadata Registry (MDR)” and specifies the standardization, representation and registration of metadata in order to enable the exchange of (meta-)data. | All |
| ISO 13119:2012 | Standard for Metadata | Is the standard “Health informatics — Clinical knowledge resources — Metadata” and defines some metadata elements needed to describe resources including medical knowledge, either digital or paper documents. | All |
| ISO 21526:2019 | Standard for Requirements of Metadata Repositories in Healthcare | Is the standard defining the requirements to collect metadata (of data elements, models and datasets) in healthcare.  The standard is an extension and/or clarification of the ISO/IEC 11179 series[[8]](#endnote-8). | All |
| ISO/TS 14265:2011 | Standard for the Definition of Purposes to Process Personal Health Information | Is the standard “Classification of purposes for processing personal health information” and relates mainly to data that is not de-identified[[9]](#endnote-9). | All |
| ISO/TS 25237 | Standard for Pseudonymization in Health Informatics | Is the standard “Health informatics – Pseudonymization" and defines concepts and requirements for pseudonymization services with the aim of personal health information’ protection[[10]](#endnote-10). | Mainly T5.3, (all) |
| ISO/TS 22220:2011 | Standard for the Identification of subjects of care in Health Informatics | Is the standard “Health informatics – Identification of subjects of care” and defines suitable data elements and structure for sensitive identification of persons in health care – either in a face-to-face setting or through interaction between IT systems[[11]](#endnote-11). | Mainly T5.3, (all) |
| PROV | Standards for Data provenance | The PROV Family of Documents comprises a. o. of an ontology and data model, supporting definitions and corresponding serializations to allow the interoperable sharing of provenance information[[12]](#endnote-12).  A mapping from Dublin Core to PROV exists[[13]](#endnote-13). | All |
| Dublin CoreTM Metadata Initiative (DCMI) | Standard for Data provenance | Supports innovation in meta design and foster best practices. DCMI provides core specifications and schemas. Information about the provenance of a resource is partially included in Dublin Core vocabulary[[14]](#endnote-14).  A mapping from Dublin Core to PROV exists. | All |
| Data Documentation Initiative (DDI) | Semantic ; (meta-)data standard | Fosters the description of data arising from surveys and observational methods in various domains. Offers controlled vocabularies and a Lifecycle to improve the documentation and data management across the entire processes[[15]](#endnote-15). | All |
| Study Cohort Ontology (SCO) | Semantic ; Ontology | Characterizes the structure and patterns of cohorts' variables and groups in clinical trials and observational case studies; offers a. o. a Diabetes related terminology[[16]](#endnote-16). | T5.1, T5.2 |
| Clinical Trial Ontology or Core Ontology of Clinical Trials (CTO) | Semantic ; Ontology | Ontology created collaboratively by serval institutions to support the representation and inclusion of all concepts and terms utilized to describe and register clinical trials. Extensions of CTO can be used to annotate variables in study documents[[17]](#endnote-17),[[18]](#endnote-18). | T5.4 ; maybe T5.1 + T5.2 + T5.6 |
| Ontology of Clinical Research (OCRE) | Semantic ; Ontology | Enables study elements and studies to be systematically describes and supports interoperable queries[[19]](#endnote-19). | T5.4 ; maybe T5.1 + T5.2 + T5.6 |
| Ontology for Biomedical Investigations (OBI) | Semantic ; Ontology | Supports the unambiguous communication about scientific investigations with a list of more than 2500 terms to describe e.g. devices, assays, objectives as well as protocols, intrumentation and materials used, etc.[[20]](#endnote-20). | All but T5.3 |
| CARRE Risk Factor ontology | Semantic ; Ontology | Represents risk factors, evidence and observables with links to widely recognized standards such as ICD, SNOMED CT, LOINC, ICD10PCS, UMLS, W3C[[21]](#endnote-21). | All but T5.3 |
| Standards from ISO/TC 34 Food Products | Different types of standards | Develops of standards related to food and the food chain including a. o. “terminology, sampling, methods of test and analysis, product specifications, food and feed safety and quality management and requirements for packaging, storage and transportation”[[22]](#endnote-22). | T5.1 (T5.2) |
| FoodEx2 | Semantic ; Classification / Description system | Supports the description of food in data collections[[23]](#endnote-23).  *Evaluated as well fitting by T5.1.* | T5.1 (T5.2) |
| Ontology for Nutritional Epidemiology (ONE) | Semantic | Combines different ontologies such as FoodOn and FoodEx, standards for publication such as STROBE or standards for data analysis and fits well for epidemiological studies[[24]](#endnote-24). ONE allows to describe the nutritional epidemiological output. | T5.1 (T5.2) |
| Ontology for Nutritional Studies (ONS) | Semantic | Focuses rather on interventional and observational studies than on epidemiological studies and includes also other food ontologies (but no standards for results publication)[[25]](#endnote-25) | T5.1 (T5.2) |
| FoodON | Semantic | Fosters data annotation while observing FAIR principles and is included e.g. in ONE and ONS[[26]](#endnote-26). | T5.1 (T5.2) |
| Bionutrition Ontology (BNO) | Semantic | Enables to describe nutritional concepts[[27]](#endnote-27). | T5.1 (T5.2) |
| Langua alimentaria (LanguaL) | Semantic ; Thesaurus | Enables both the description and the capture/retrieval of food data[[28]](#endnote-28). | T5.1 (T5.2) |
| Strengthening the Reporting of observational studies in epidemiology (STROBE) | Publication guidelines | Fosters the adequate reporting and dissemination of epidemiological findings[[29]](#endnote-29).  Further guidelines for specific use cases, such as STROBE-nut for nutrition[[30]](#endnote-30). | T5.1, T5.2, maybe T5.3 |
| European Nutritional Phenotype Assessment and Data Sharing Initiative (ENPADASI) | “Minimal information guidelines” and semantic | Collects and standardizes study metadata and phenotypic from nutritional interventional, mechanistic and epidemiological studies. Develops templates for data collection as well as minimal requirements to share nutritional data and study information[[31]](#endnote-31). | T5.1 (T5.2) |
| Core Outcome Set (COS) of the Core Outcome Measures in Effectiveness Trials Initiative (COMET) | Core set of outcomes for effectiveness trials | The COMET initiative provides a. o. a handbook and guidelines how to develop a core outcome set (for effectiveness trials)[[32]](#endnote-32).  *Possibilities of adaption of the guidelines to other types of clinical trials or epidemiological research should be evaluated.* | T5.1, T5.2, T5.4 |
| National Cancer Institute’s Thesaurus (NCIT) | Semantic ; Thesaurus/Ontology | Enables the description of concepts in the cancer domain[[33]](#endnote-33). | T5.1, T5.2 |
| EpiJson | Data format | Specifies a standard handling computer primitive data-types in an unambiguous way in order to share and transmit data from epidemiology while respecting data fidelity[[34]](#endnote-34). | T5.1, T5.2, maybe T5.3 |

1. HL7.org. HL7® FHIR® Release 4. Resource Consent - Content. [Internet]. 2019. [Cited 2021 Nov 08]. Available from: <https://hl7.org/FHIR/consent.html> [↑](#endnote-ref-1)
2. HL7.org. HL7® FHIR® Release 4. Resource Provenance - Content. [Internet]. 2019. [Cited 2021 Nov 08]. Available from: <https://hl7.org/FHIR/provenance.html> [↑](#endnote-ref-2)
3. HL7.org. HL7® FHIR® Release 4. Resource Measure - Content. [Internet]. 2019. [Cited 2021 Nov 08]. Available from: <https://hl7.org/FHIR/measure.html> [↑](#endnote-ref-3)
4. SNOMED International. 5-Step briefing. [Internet]. 2021. [Cited 2021 Nov 08]. Available from: <https://www.snomed.org/snomed-ct/five-step-briefing> [↑](#endnote-ref-4)
5. SNOMED International. Collaboration agreement formalized with the Academy of Nutrition and Dietetics for nutrition content in SNOMED CT. [Internet]. 2021. [Cited 2021 Nov 08]. Available from: <https://www.snomed.org/news-and-events/articles/snomed-academy-nutrition-dietetics-agreement> [↑](#endnote-ref-5)
6. Mavergames C, Everett J, Becker L, Wilton P, Oliver S. Cochrane PICO Ontology. Revision: 4.0.1. [↑](#endnote-ref-6)
7. Cochrane Linked Data. Metadata and vocabularies. [Internet]. 2021. [Cited 2021 Nov 08]. Available from: [Metadata and vocabularies | Cochrane Linked Data](https://linkeddata.cochrane.org/linked-data-project/metadata-and-vocabularies) [↑](#endnote-ref-7)
8. International Standardization Organization (ISO). ISO/TS 21526:2019(en) Health informatics - Metadata repository requirements (MetaRep) [↑](#endnote-ref-8)
9. International Standardization Organization (ISO). ISO/TS 14265:2011(en) Health Informatics – Classification for processing personal health information. [↑](#endnote-ref-9)
10. International Standard Organization (ISO). ISO 25237:2017 Health informatics - Pseudonymization [↑](#endnote-ref-10)
11. International Standard Organization (ISO). ISO/TS 22220:2011 Health informatics – Identification of subjects of healthcare. [↑](#endnote-ref-11)
12. W3C Working Group Note. PROV-Overview. [Internet]. 2013. [Cited 2021 Nov 08]. Available from: https://www.w3.org/TR/prov-overview/ [↑](#endnote-ref-12)
13. W3C Working Group Note. Dublin Core to PROV Mapping. [Internet]. 2013. [Cited 2021 Nov 08]. Available from: https://www.w3.org/TR/prov-dc/ [↑](#endnote-ref-13)
14. Dublin CoreTM Metadata Initiative. Home. [Internet]. 2021. [Cited 2021 Nov 08]. Available from: http://dublincore.org/ [↑](#endnote-ref-14)
15. Data Documentation Initiative (DDI). Document, Discover and Interoperate. [Internet]. 2021. [Cited 2021 Nov 08]. Available from: <https://ddialliance.org/> [↑](#endnote-ref-15)
16. Chari S, Qi M, Agu N, Seneviratne O, McCusker J, Bennett K, Das A, Mcguinness D. Making Study Populations Visible Through Knowledge Graphs. The Semantic Web - ISWC 2019. 2019 Oct;53-68. doi: 10.1007/978-3-030-30796-7\_4. [↑](#endnote-ref-16)
17. Lin AY, Gebel S, Li Q, Madan S, Darms J, Bolton EE, Smith B, Hofmann-Apitius M, He YO, Kodamullil AT. CTO: A Community-Based Clinical Trial Ontology and Its Applications in PubChemRDF and SCAIView. ICBO/ODLS. 2020. [↑](#endnote-ref-17)
18. GitHub. ClinicalTrialOntology/CTO. [Internet]. 2021. [Cited 2021 Nov 08]. Available from: [GitHub - ClinicalTrialOntology/CTO: CTO: Core Ontology of Clinical Trials](https://github.com/ClinicalTrialOntology/CTO/) [↑](#endnote-ref-18)
19. Sim I, Tu SW, Carini S, Lehmann HP, Pollock BH, Peleg M, Wittkowski KM. The Ontology of Clinical Research (OCRe): an informatics foundation for the science of clinical research. J Biomed Inform. 2014 Dec;52:78-91. doi: 10.1016/j.jbi.2013.11.002. Epub 2013 Nov 13. PMID: 24239612; PMCID: PMC4019723. [↑](#endnote-ref-19)
20. Bandrowski A, Brinkman R, Brochhausen M, Brush MH, Bug B, Chibucos MC, Clancy K, Courtot M, Derom D, Dumontier M, Fan L, Fostel J, Fragoso G, Gibson F, Gonzalez-Beltran A, Haendel MA, He Y, Heiskanen M, Hernandez-Boussard T, Jensen M, Lin Y, Lister AL, Lord P, Malone J, Manduchi E, McGee M, Morrison N, Overton JA, Parkinson H, Peters B, Rocca-Serra P, Ruttenberg A, Sansone SA, Scheuermann RH, Schober D, Smith B, Soldatova LN, Stoeckert CJ Jr, Taylor CF, Torniai C, Turner JA, Vita R, Whetzel PL, Zheng J. The Ontology for Biomedical Investigations. PLoS One. 2016 Apr 29;11(4):e0154556. doi: 10.1371/journal.pone.0154556. PMID: 27128319; PMCID: PMC4851331. [↑](#endnote-ref-20)
21. Carre Project. What is Carre Risk Factor Ontology? [Internet]. 2015. [Cited 2021 Nov 08]. Available from: https://www.carre-project.eu/innovation/risk-factor-ontology/ [↑](#endnote-ref-21)
22. International Standardization Organization (ISO). ISO/TC 34 Food products. [Internet]. [Cited 2021 Nov. 08]. Available from: https://www.iso.org/committee/47858.html [↑](#endnote-ref-22)
23. Eftimov T, Korošec P, Koroušić Seljak B. StandFood: Standardization of Foods Using a Semi-Automatic System for Classifying and Describing Foods According to FoodEx2. Nutrients. 2017 May 26;9(6):542. doi: 10.3390/nu9060542. PMID: 28587103; PMCID: PMC5490521. [↑](#endnote-ref-23)
24. Yang C, Ambayo H, Baets B, Kolsteren P, Thanintorn N, Hawwash D, Bouwman J, Bronselaer A, Pattyn F, Lachat C. An Ontology to Standardize Research Output of Nutritional Epidemiology: From Paper-Based Standards to Linked Content. Nutrients. 2019 Jun 8;11(6):1300. doi: 10.3390/nu11061300. PMID: 31181762; PMCID: PMC6628051. [↑](#endnote-ref-24)
25. Vitali F, Lombardo R, Rivero D, Mattivi F, Franceschi P, Bordoni A, Trimigno A, Capozzi F, Felici G, Taglino F, Miglietta F, De Cock N, Lachat C, De Baets B, De Tré G, Pinart M, Nimptsch K, Pischon T, Bouwman J, Cavalieri D; ENPADASI consortium. ONS: an ontology for a standardized description of interventions and observational studies in nutrition. Genes Nutr. 2018 Apr 30;13:12. doi: 10.1186/s12263-018-0601-y. PMID: 29736190; PMCID: PMC5928560. [↑](#endnote-ref-25)
26. Dooley DM, Griffiths EJ, Gosal GS, Buttigieg PL, Hoehndorf R, Lange MC, Schriml LM, Brinkman FSL, Hsiao WWL. FoodOn: a harmonized food ontology to increase global food traceability, quality control and data integration. NPJ Sci Food. 2018 Dec 18;2:23. doi: 10.1038/s41538-018-0032-6. PMID: 31304272; PMCID: PMC6550238. [↑](#endnote-ref-26)
27. Coffran C., Meehan D., Ronning A. Bionutrition Ontology. [Internet]. 2013. [Cited 2021 Nov 08]. Available from: https://bioportal.bioontology.org/ontologies/BNO?p=classes&conceptid=root [↑](#endnote-ref-27)
28. Danish Food Informatics. LanguaL—The International Framework for Food Description. [Cited 2021 Nov 08]. Available online: <http://www.langual.org/> [↑](#endnote-ref-28)
29. von Elm E, Altman DG, Egger M, Pocock SJ, Gøtzsche PC, Vandenbroucke JP; STROBE Initiative. The Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) Statement: guidelines for reporting observational studies. Int J Surg. 2014 Dec;12(12):1495-9. doi: 10.1016/j.ijsu.2014.07.013. Epub 2014 Jul 18. PMID: 25046131. [↑](#endnote-ref-29)
30. Lachat C, Hawwash D, Ocké MC, Berg C, Forsum E, Hörnell A, Larsson C, Sonestedt E, Wirfält E, Åkesson A, Kolsteren P, Byrnes G, De Keyzer W, Van Camp J, Cade JE, Slimani N, Cevallos M, Egger M, Huybrechts I. Strengthening the Reporting of Observational Studies in Epidemiology-Nutritional Epidemiology (STROBE-nut): An Extension of the STROBE Statement. PLoS Med. 2016 Jun 7;13(6):e1002036. doi: 10.1371/journal.pmed.1002036. PMID: 27270749; PMCID: PMC4896435. [↑](#endnote-ref-30)
31. Pinart M, Nimptsch K, Bouwman J, Dragsted LO, Yang C, De Cock N, Lachat C, Perozzi G, Canali R, Lombardo R, D'Archivio M, Guillaume M, Donneau AF, Jeran S, Linseisen J, Kleiser C, Nöthlings U, Barbaresko J, Boeing H, Stelmach-Mardas M, Heuer T, Laird E, Walton J, Gasparini P, Robino A, Castaño L, Rojo-Martínez G, Merino J, Masana L, Standl M, Schulz H, Biagi E, Nurk E, Matthys C, Gobbetti M, de Angelis M, Windler E, Zyriax BC, Tafforeau J, Pischon T. Joint Data Analysis in Nutritional Epidemiology: Identification of Observational Studies and Minimal Requirements. J Nutr. 2018 Feb 1;148(2):285-297. doi: 10.1093/jn/nxx037. PMID: 29490094. [↑](#endnote-ref-31)
32. Williamson PR, Altman DG, Bagley H, Barnes KL, Blazeby JM, Brookes ST, Clarke M, Gargon E, Gorst S, Harman N, Kirkham JJ, McNair A, Prinsen CAC, Schmitt J, Terwee CB, Young B. The COMET Handbook: version 1.0. Trials. 2017 Jun 20;18(Suppl 3):280. doi: 10.1186/s13063-017-1978-4. PMID: 28681707; PMCID: PMC5499094. [↑](#endnote-ref-32)
33. 1. Golbeck J, Fragoso G, Hartel F, Hendler J, Oberthaler J, Parsia B. The National Cancer Institute's Thesaurus and Ontology. SSRN Electronic Journal. 2003. doi: 10.2139/ssrn.3199007 [↑](#endnote-ref-33)
34. Finnie TJ, South A, Bento A, Sherrard-Smith E, Jombart T. EpiJSON: A unified data-format for epidemiology. Epidemics. 2016 Jun;15:20-6. doi: 10.1016/j.epidem.2015.12.002. Epub 2015 Dec 29. PMID: 27266846; PMCID: PMC7104924. [↑](#endnote-ref-34)