PHUSE EU CONNECT, NOV 2023

A Dive Into the World of **Biomedical Concepts**

Content

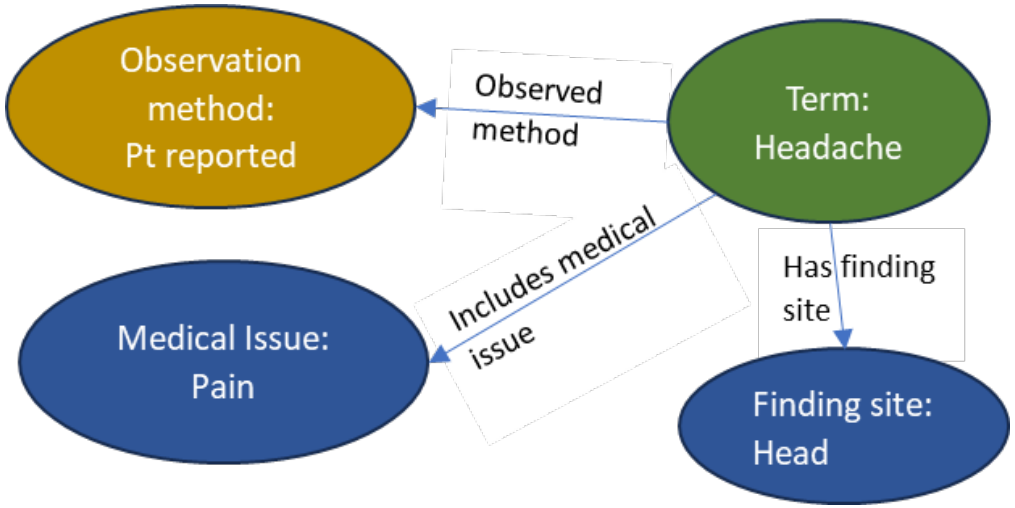
- What are Biomedical Concepts?
- How are Biomedical Concepts represented?
- How to map Biomedical Concepts
- Issues in mapping
- Conclusion

What are biomedical concepts

- Observations or measurements that can be obtained from a biological species
- Other definitions:
  - J. Ulander, K. Walther, PharmaSUG 2021: a generic description of something that can be measured on a subject or other information such as events happened or interventions made in relation to a subject's health records.
  - Nurocor: Identifies a discrete unit of knowledge in any of the biomedical information sciences.
  - …

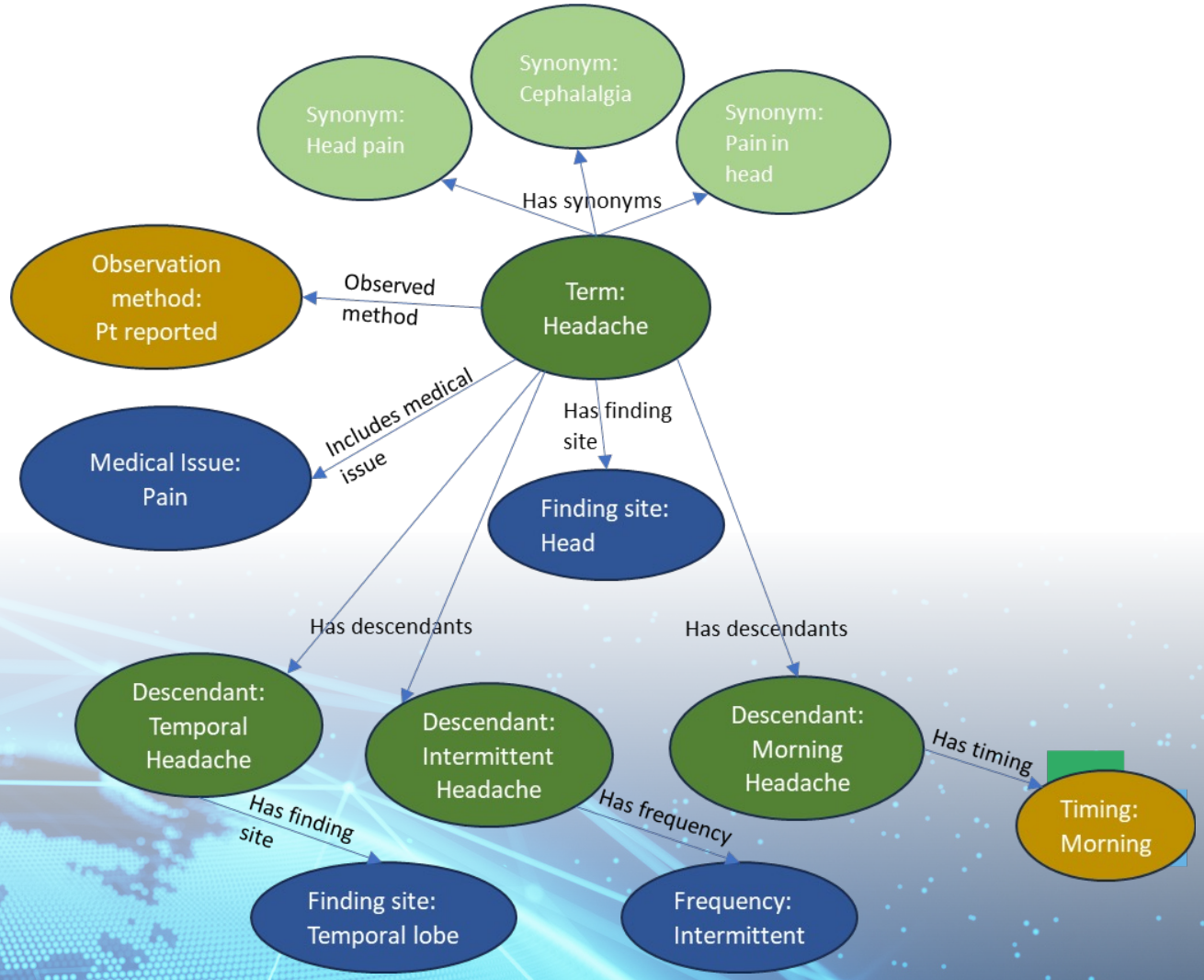
Breaking down biomedical concepts (BCs) to specific elements

- Define all elements that represent a specific feature of the BC
- As small and detailed as possible!
- Can be anything that links to the specific observation or measurement
  - How it was measured/observed
  - Where it was measured/observed
  - When it was measured/observed
  - Frequency
  - Units
  - etc



Synonyms, descendants and ancestors

- Synonyms
  - Exactly the same elements
- Descendants / children
  - Additional elements
- Ancestors / parents
  - Subset of elements



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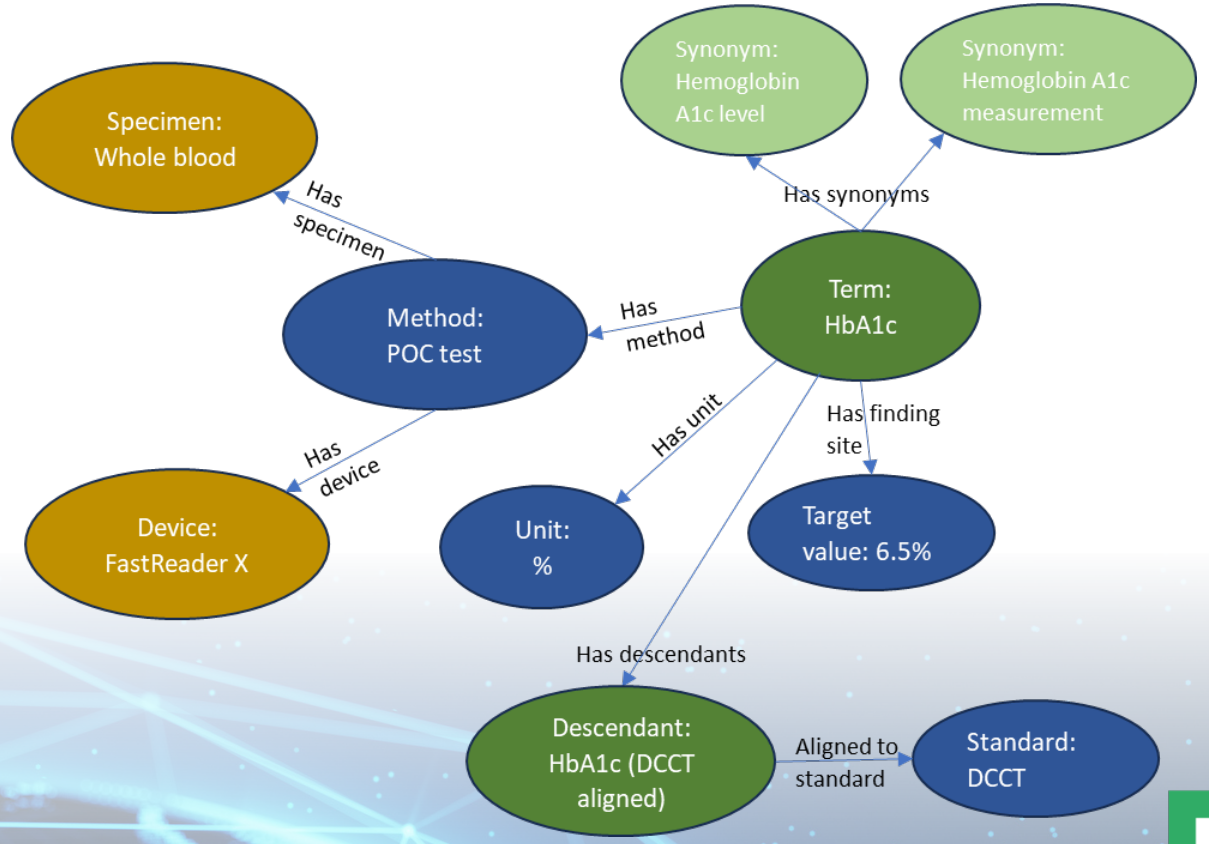
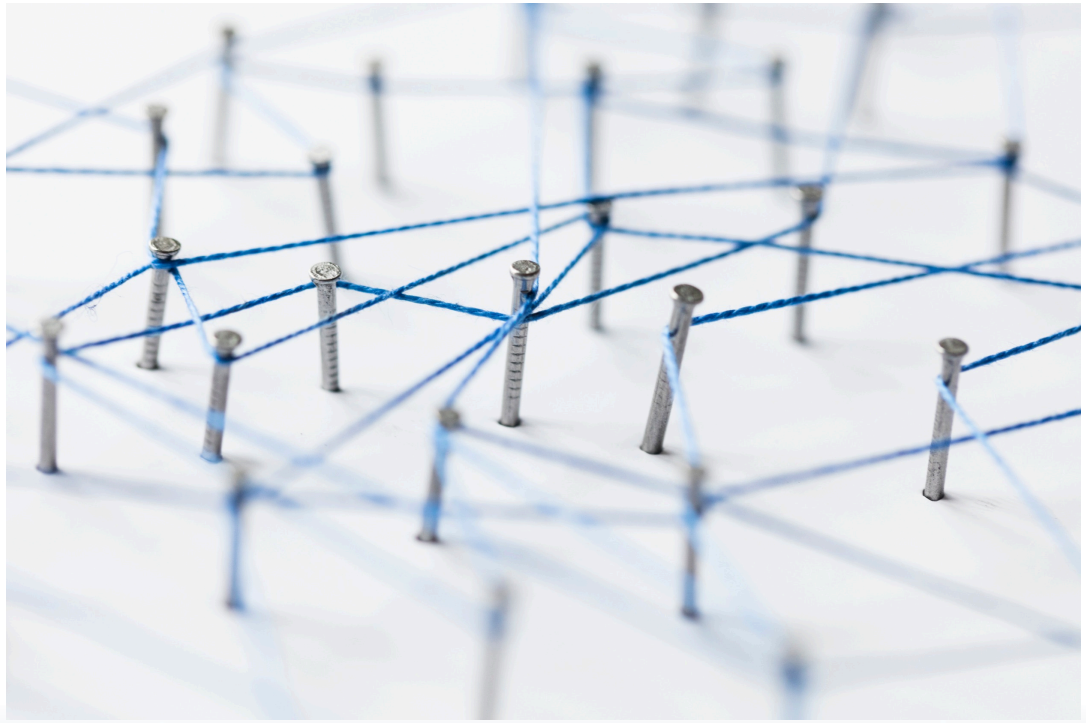


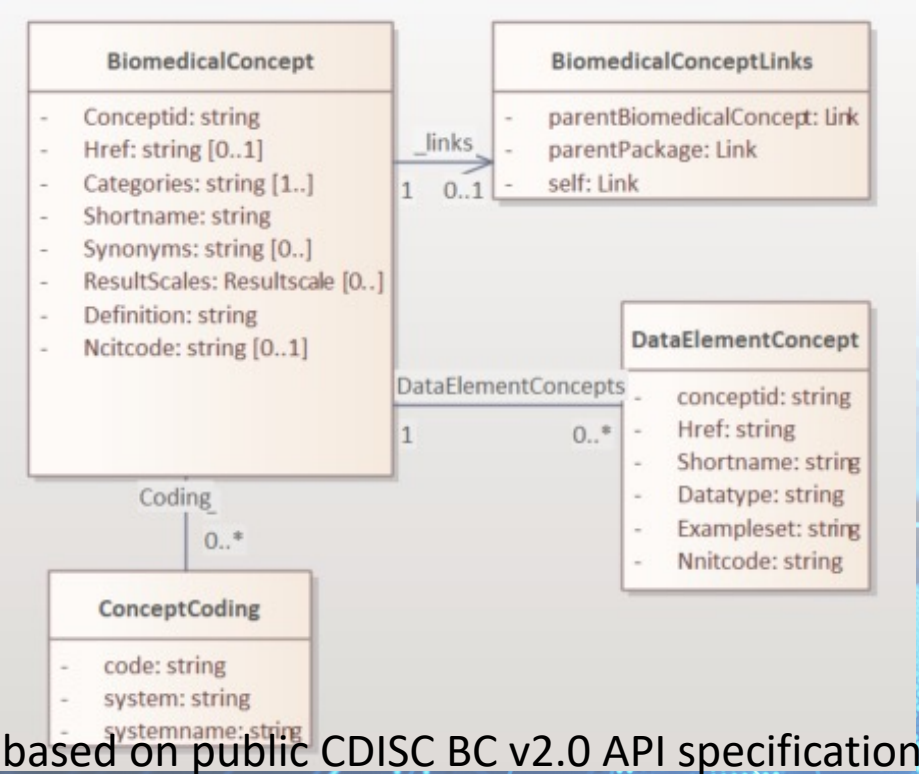
Presented as



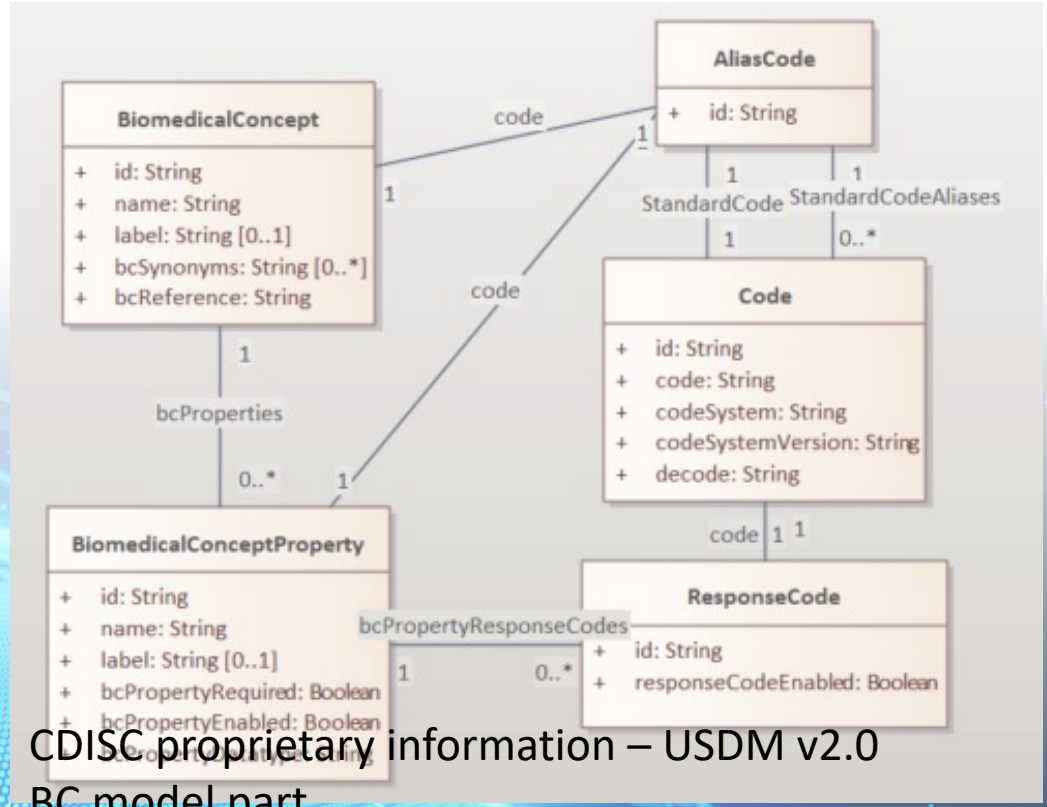
- Technical:
  - Datasets / Relational Databases
  - Document Databases
  - Graph Databases
- Include specifications for:
  - Concepts
  - Properties
  - Relationships
    - Synonyms
    - Ancestors
    - Descendants

CDISC Biomedical Concepts

CDISC kicked off the Conceptual and Operational Standards Metadata Services (COSMoS) project in 2022, taking a pragmatic, iterative approach to creating biomedical concepts and representing them in the Foundational Standards as dataset specializations with Value Level Metadata definitions. Biomedical Concepts fill gaps in the current standards by adding semantics, variable relationships, and the detailed metadata needed to generate CRFs or Define-XML.

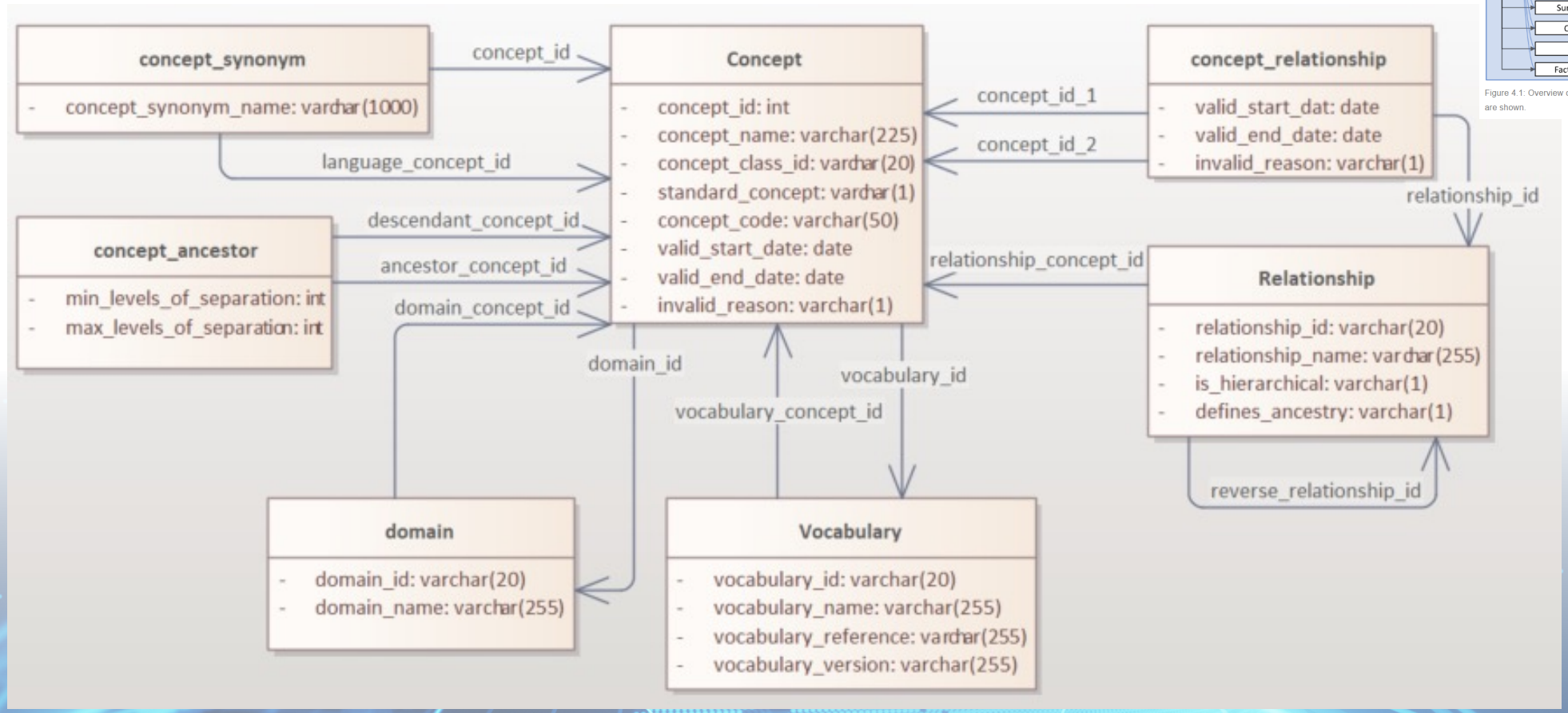


based on public CDISC BC v2.0 API specifications



CDISC proprietary information – USDM v2.0 BC model part

OMOP – Athena dictionary



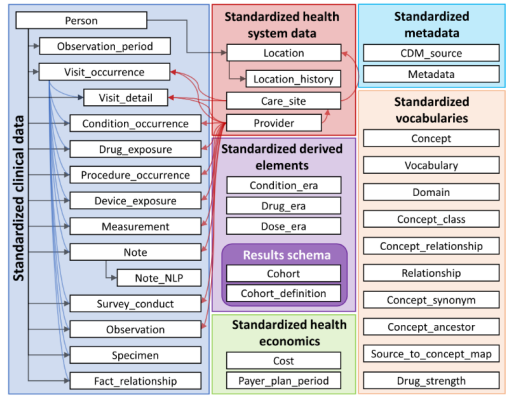


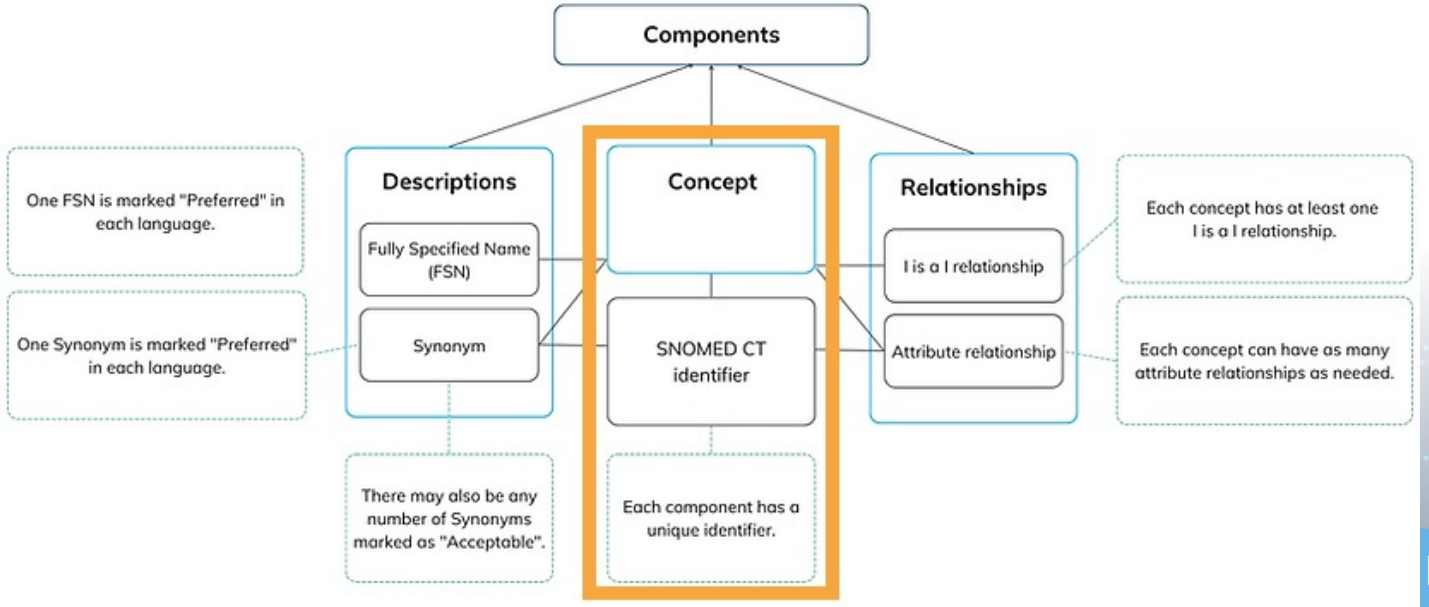
Figure 4.1: Overview of all tables in the CDM version 6.0. Note that not all relationships between tables are shown.

SNOMED

"The SNOMED CT clinical terminology has unmatched depth, enabling clinicians to record data with enhanced accuracy and consistency."

Concepts

Every **concept** represents a unique clinical meaning, which is referenced using a unique, numeric and machine readable SNOMED CT identifier. The **identifier** provides an unambiguous unique reference to each concept and does not have any ascribed human interpretable meaning.



Source: www.SNOMED.org/five-step-brieving

LOINC

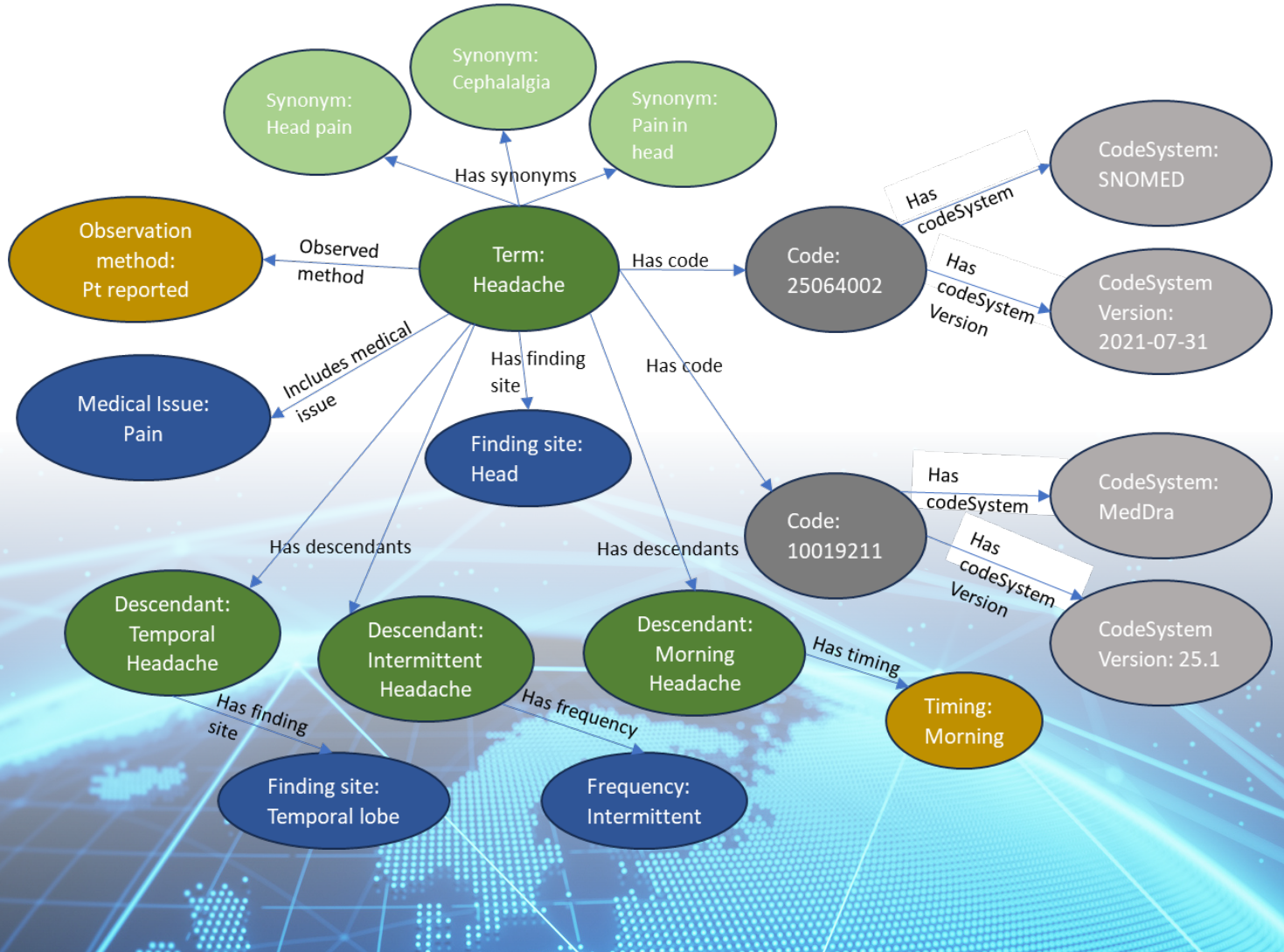
Table 1: Hierarchical Structure of Fully Specified *Analyte* Name

Export table to CSV

Subpart Name	Section
Component/Analyte	2.2
Name and modifier	2.2.1
Component/Analyte name	2.2.1.1
Component/Analyte subname	2.2.1.2
Information about the *Challenge* (e.g., 1H post 100 gm PO challenge)	2.2.2
Adjustments/corrections	2.2.3
Kind of *Property* (mass concentration, mass)	2.3
Time Aspect (point or moment in time vs. time interval)	2.4
System/sample type (urine, serum)	2.5
"Super System" (patient, donor, blood product unit)	2.5.2
Type of *Scale* (nominal, ordinal, quantitative)	2.6
Method Type	2.7

https://loinc.org/kb/users-guide/major-parts-of-a-loinc-term/

Identifying Codes



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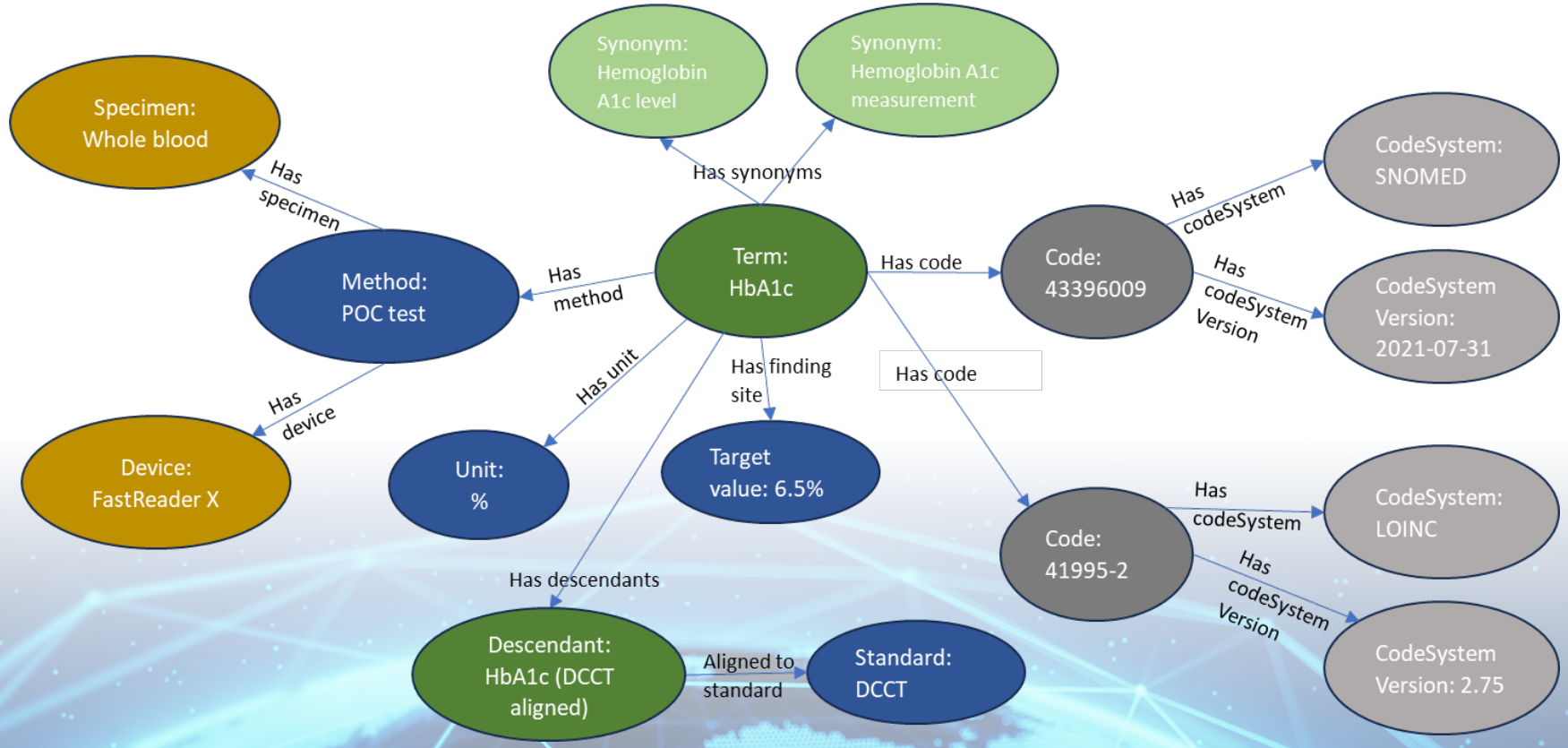

Issues in mapping

- Other data structure
- Other representation
- Multiplicity
- Non-exact or no matches
- Dictionary versions
- .. ? ..

How to map?

1. Try exact map
2. If not possible: Look for additional information to map exactly
3. Generalize, if exact match is not possible

Documentation

- Store the complete mapping trail
- Keep original information as reference
  - like OMOP including
    - Standardized concept_id
    - Source concept_id
    - Source value

Automation

- Exact matches – no review
- Non-exact matches
  - Parents / Suggestions
  - Manual authentication

Conclusions

- Different structure and focus of dictionaries makes it complex to map
- Create/Use standard mapping for frequently used BCs
- Manual mapping and medical review for non-frequent BCs
- Provenance and traceability of mapping is key to be able to prove the reliability of your data

Questions and contact

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