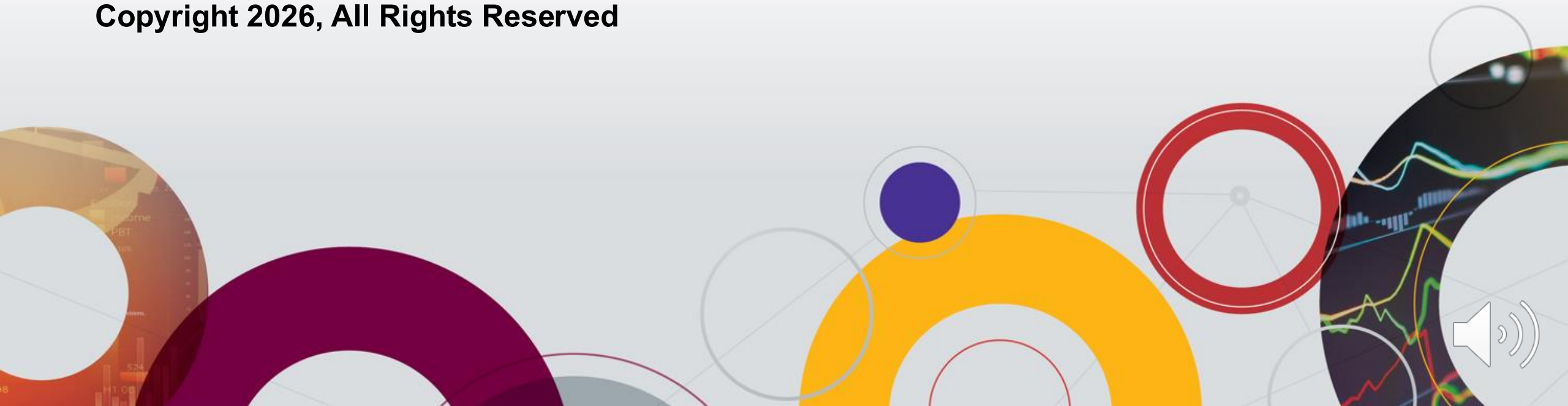


4.4c Terminology Standards

10x10 – Introduction to Biomedical Informatics & Artificial Intelligence
William Hersh, MD, FACMI, FAMIA, FIAHSI
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SNOMED Clinical Terms (SNOMED CT)

Systematized Nomenclature of Medicine (SNOMED)

Originally developed by College of American Pathologists (CAP)

- Originally a classification for pathologists (SNOP) but extended to all of medicine as SNOMED in 1980s (Cote, 1993)
- Merged with English Clinical Terms Project to form SNOMED CT in 2000 (Spackman, 2000)

In 2007, ownership transferred to International Health Terminology Standards Development Organisation (IHTSDO), which is now called [SNOMED International](#)

- Licensed by many countries for national use

Multilingual – currently available in US English, UK English, Spanish, Danish and Swedish; being translated to others

Survey of best practices for terminology binding (Rossander, 2021)

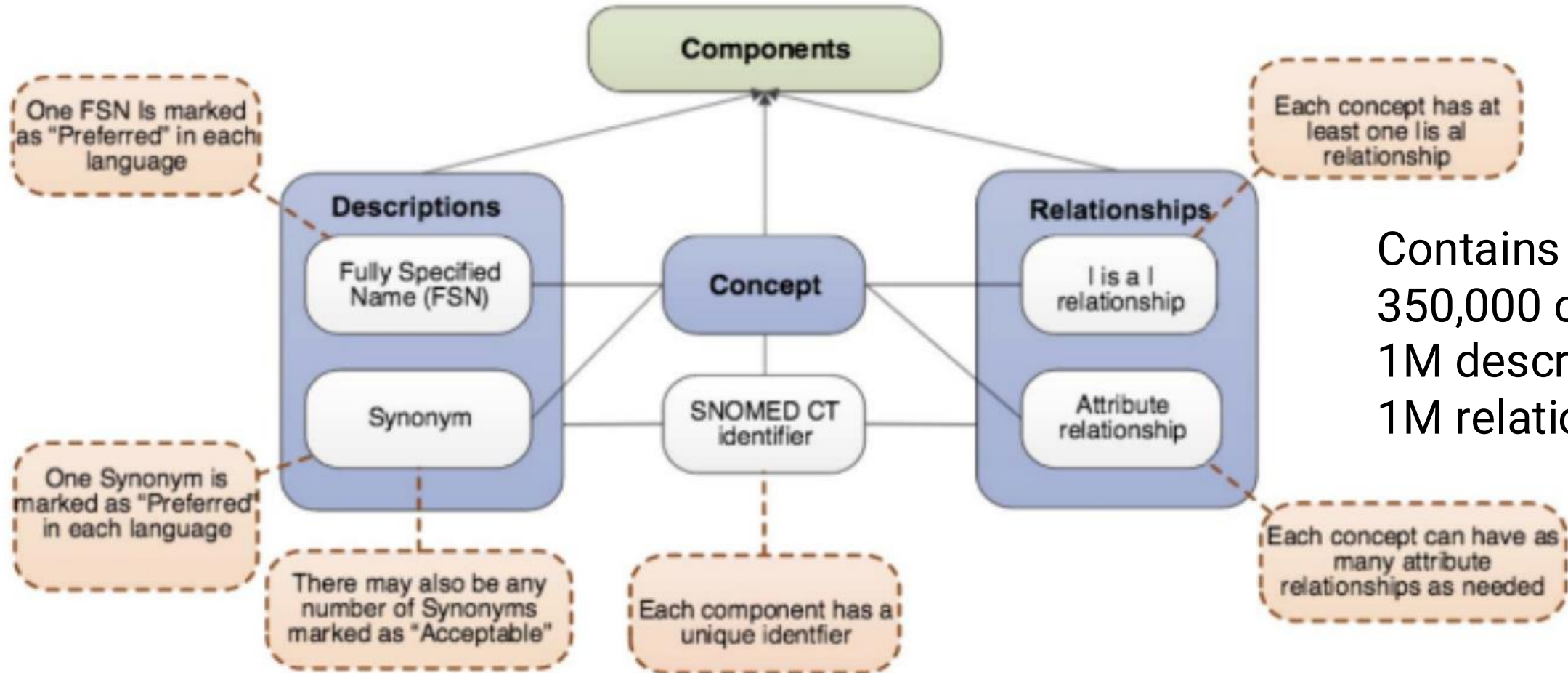
SNOMED CT

Web site provides introduction, documentation, and more

- [Overview](#)
- [Starter guide](#)
- [Educational offerings](#)
- [Browser](#)

Key feature is compositional approach (formerly called multi-axial) that allows complex terms to be built from simpler ones (aka, postcoordination)

Structure of SNOMED CT



Context of SNOMED CT (Bodenreider, 2018)

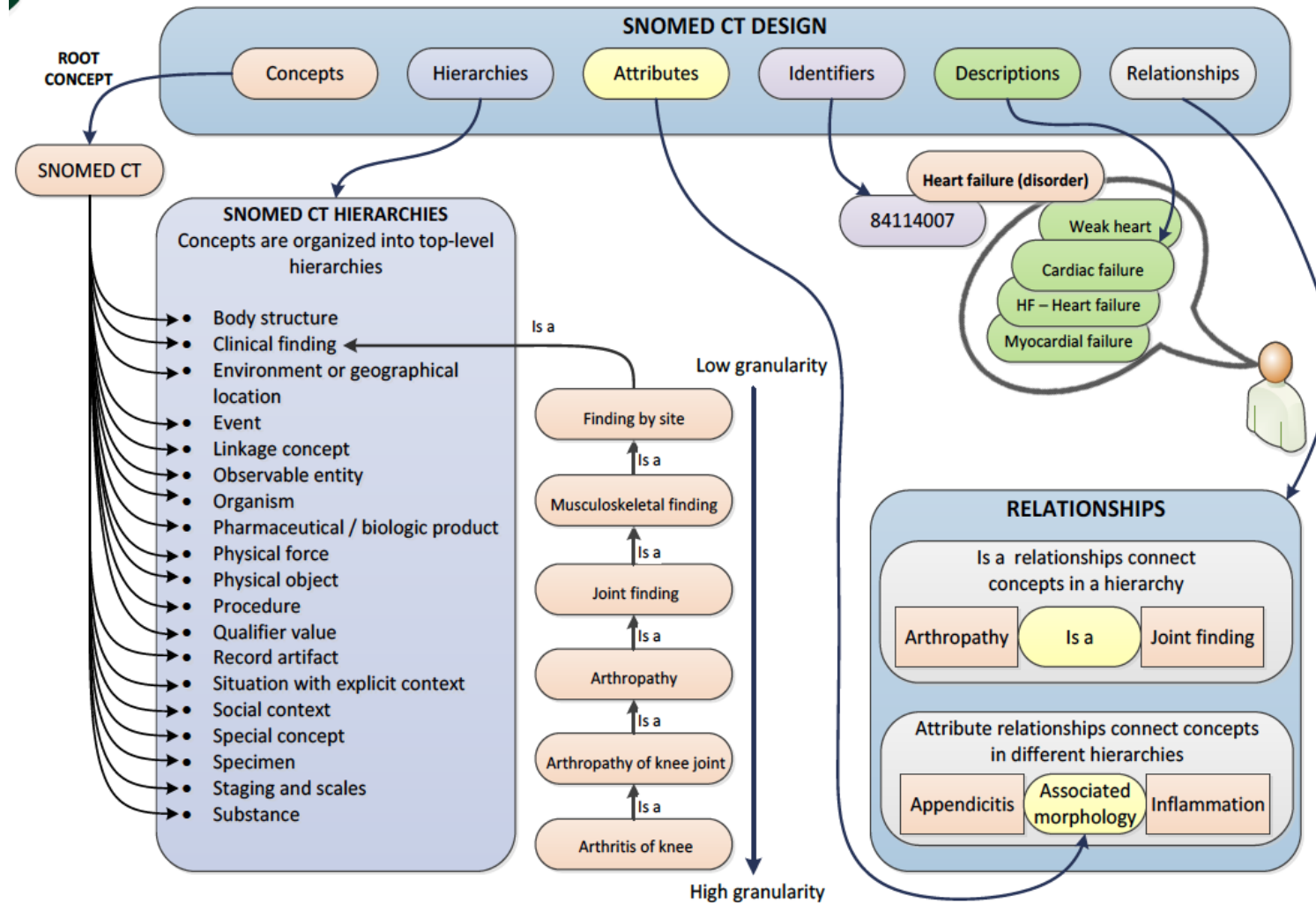
Overview of major three clinical terminologies and their integration

- SNOMED CT
- LOINC
- RxNorm

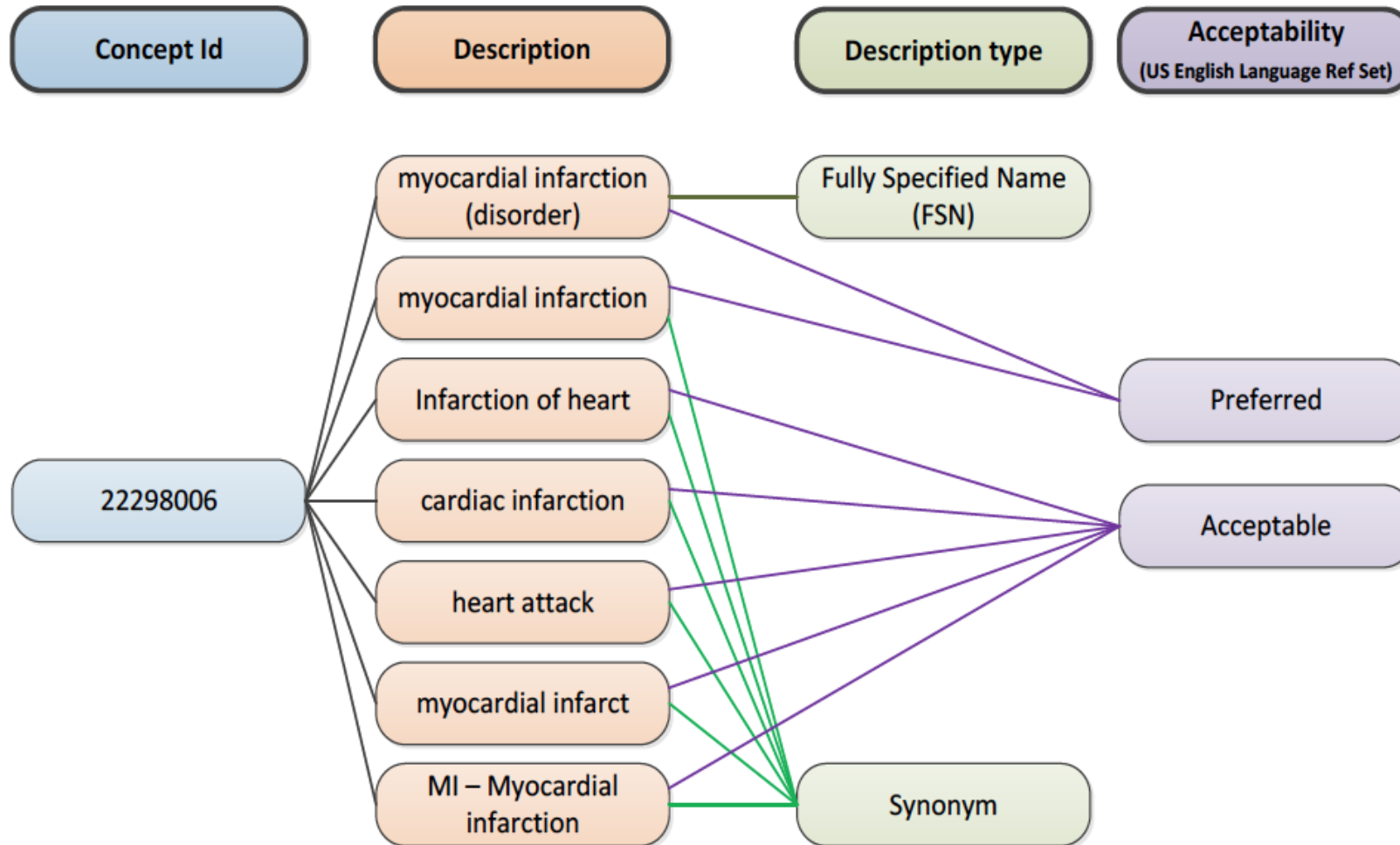
One view: LOINC contains the question and SNOMED CT contains the answer

Or should SNOMED be the *lingua franca* for all?

SNOMED CT design

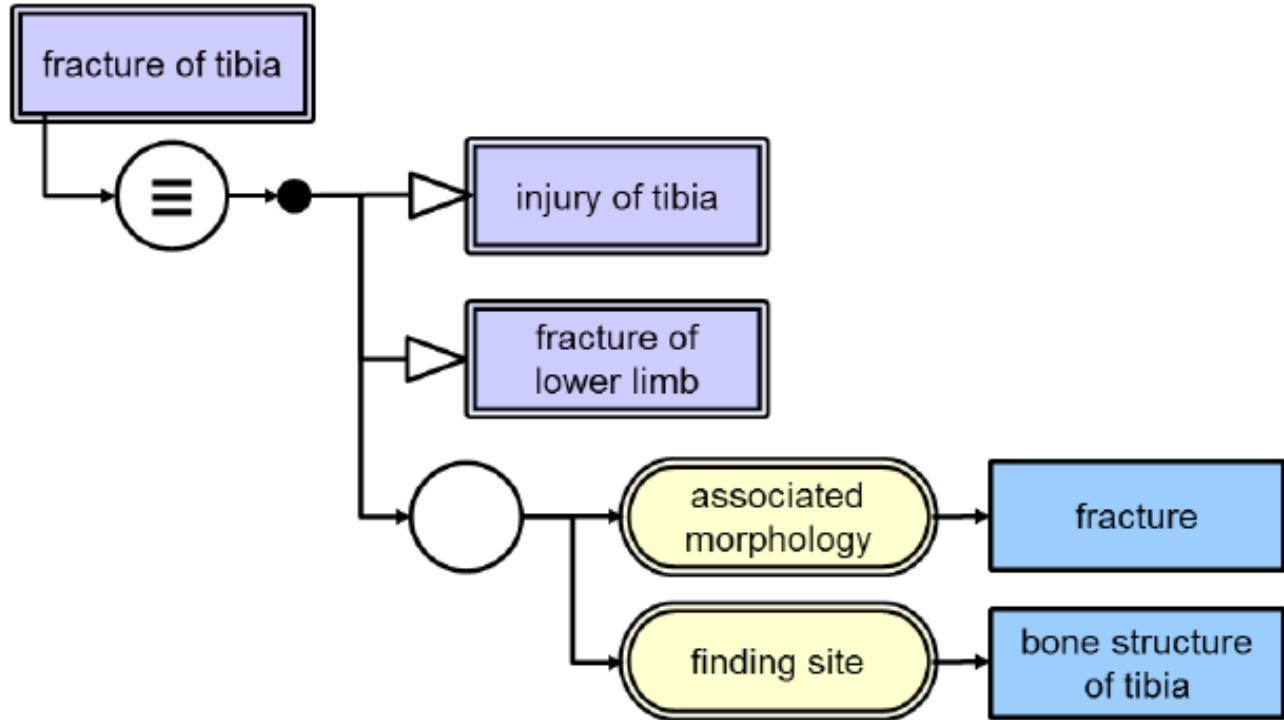


SNOMED CT descriptions



SNOMED CT expressions – some are pre-coordinated

Precoordinated expression representing fracture of tibia	Identifier only	31978002
	With display term	31978002 fracture of tibia

Graphic view of the defining relationships of the concept fracture of tibia	 The diagram illustrates the defining relationships for the SNOMED CT concept 'fracture of tibia'. It starts with a box labeled 'fracture of tibia' on the left. An arrow points from this box to a circle containing three horizontal lines (≡). From this circle, an arrow points to a black dot (junction). From the junction, three arrows branch out: one to a box labeled 'injury of tibia', one to a box labeled 'fracture of lower limb', and one to a circle. From this circle, two arrows branch out: one to a yellow oval labeled 'associated morphology' and one to a yellow oval labeled 'finding site'. The 'associated morphology' oval points to a blue box labeled 'fracture'. The 'finding site' oval points to a blue box labeled 'bone structure of tibia'.
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Nursing vocabularies

Critical for capturing and assessing diagnosis, interventions, and outcomes of nursing care (Welton, 2016)

Many vocabularies over years, with

- Interface terminologies with irreconcilable information models
- Terms not always the way clinicians express themselves
- Tedious to use in patient documentation
- Varying licensing models
- Lack of mapping to reference terminologies, such as LOINC and SNOMED CT

Recent efforts in nursing informatics community to reconcile and map to major other terminologies (Westra, 2015; Matney, 2017; Monsen, 2023)

Nursing terminologies landscape (ONC, 2017)

Interface Terminologies	Minimum Data Sets
1. Clinical Care Classification (CCC) System 2. International Classification for Nursing Practice (ICNP) 3. North American Nursing Diagnosis Association International (NANDA-I) 4. Nursing Interventions Classification System (NIC) 5. Nursing Outcomes Classification (NOC) 6. Omaha System 7. Perioperative Nursing Data Set (PNDS) 8. ABC Codes	1. Nursing Minimum Data Set (NMDS) 2. Nursing Management Minimum Data Set (NMMDS)
	Reference Terminologies
	1. Logical Observation Identifiers Names and Codes (LOINC) 2. SNOMED Clinical Terms (SNOMED CT)

Most approved by the ANA and included in the UMLS Metathesaurus
Coverage still incomplete for nursing problem lists (Kim, 2019)

Efforts at reconciliation of vocabularies

[Unified Medical Language System \(UMLS\)](#) attempt at reconciliation
(Humphreys, 1998; Humphreys, 2020)

Consists of three components

- Metathesaurus – thesaurus across vocabularies
- Semantic network – generic relationships between semantic types of concepts, e.g., diseases and treatment
- Specialist lexicon – based on Metathesaurus words and terms, designed to assist in natural language processing applications

UMLS Metathesaurus

“Meta”-thesaurus across vocabularies

- Only terms from source vocabularies become terms in Metathesaurus

Goal is to link vocabularies, not create new one

Some had hoped/advocated Metathesaurus would become the standard medical vocabulary, but a single vocabulary is probably not feasible

- Most uses for text processing and mapping/linking terminologies (Amos, 2020)

Structure of UMLS Metathesaurus

All terms from all vocabularies representing same notion are grouped as a concept

- Linked by Concept Unique Identifier (CUI)

All source terms of similar form (i.e., differing only in lexical variation) are grouped as terms

- Linked by Term Unique Identifier (LUI)

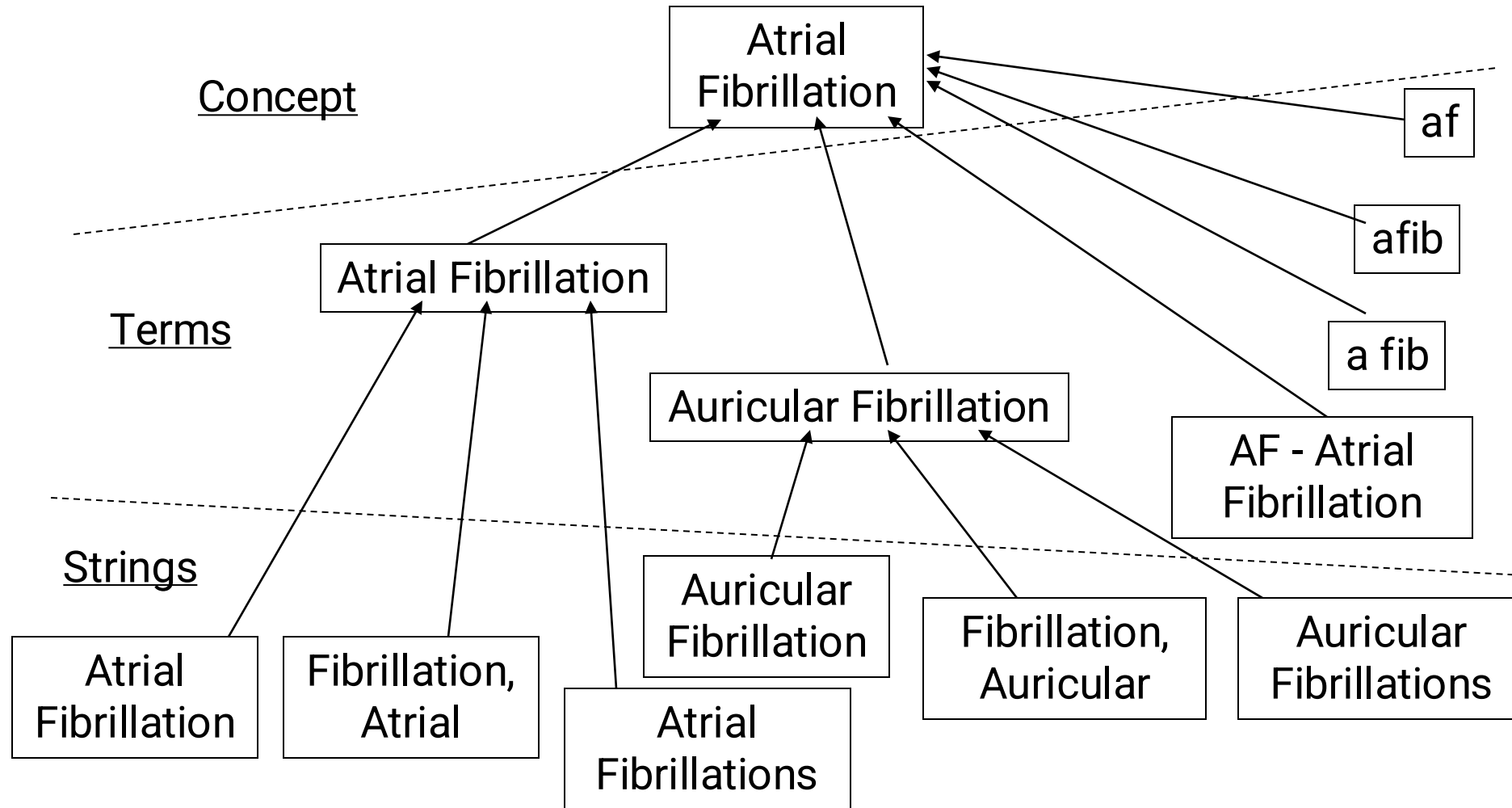
Within each term, lexical variants are strings

- Linked by String Unique Identifier (SUI)

Each string is an atom from its source

- Linked by Atomic Unique Identifier (AUI)

Example Metathesaurus concept: Atrial Fibrillation



Limitations and use of UMLS Metathesaurus

Limitations

- Only one-to-one relationships are mapped
- Only terms from source vocabularies present; no new terms added
 - Challenge for concepts not covered well in any terminologies, such as patient outcomes (Newbury, 2023)
- No unifying hierarchy is present, only those that exist in source vocabularies
- Not extensible (i.e., in the SNOMED sense)

Use

- Modest at this point
- More of a “repository” for vocabularies

Some other healthcare vocabularies (Giannangelo, 2025)

Common Dental Terminology (CDT)

Medical Subject Headings (MeSH) (discussed in information retrieval unit)

Universal Medical Device Nomenclature (UMD)

Diagnostic and Statistical Manual of Mental Disorders (DSM) – has its controversies (Kupfer, 2013)

International Classification of Functioning, Disability, and Health (ICF)

International Classification of Primary Care (ICPC)

Unified Codes for Units of Measure (UCUM)



Common data elements (CDEs)

Common data elements for research studies

Many developed by US National Institutes of Health (NIH)

- [Repository](#)
- Examples
 - [Patient-Reported Outcome Measurement System \(PROMIS\)](#)
 - [National Institute of Neurological Disorders and Stroke Common Data Elements Project](#)
 - [Global Rare Diseases Registry \(GRDR\)](#)
 - [Consensus Measures for Phenotypes and Exposures \(PhenX\)](#)

minimal Common Oncology Data Elements (mCODE) aims to “learn from everyone with cancer” based on EHR data encoded in appropriate terminology standards and using FHIR (Schnitzer, 2023; Osterman, 2023)

Some commercial terminology efforts

[Intelligent Medical Objects \(IMO\)](#) – provides mapping, updates, and access to terminology

[West Coast Informatics](#) – open-source tools for terminology use and development

[Medcin](#) (Medicomp) – focused on documentation at point of care in EHR

[HDD Access](#) – terminology system developed by 3M, moved to open-source model

Value sets (Gold, 2018)

“Collection of codes or terms from controlled medical vocabularies that are treated as equivalent for use in a clinical query or analytic task”

Many challenges in implementation (Zahn, 2025)

- Content analysis found
 - Missing codes – 42%
 - Extraneous codes – 14%
 - Misinterpretations of value set intent – 40%
- Resulting in altering clinical quality measurement output by 3-30 fold

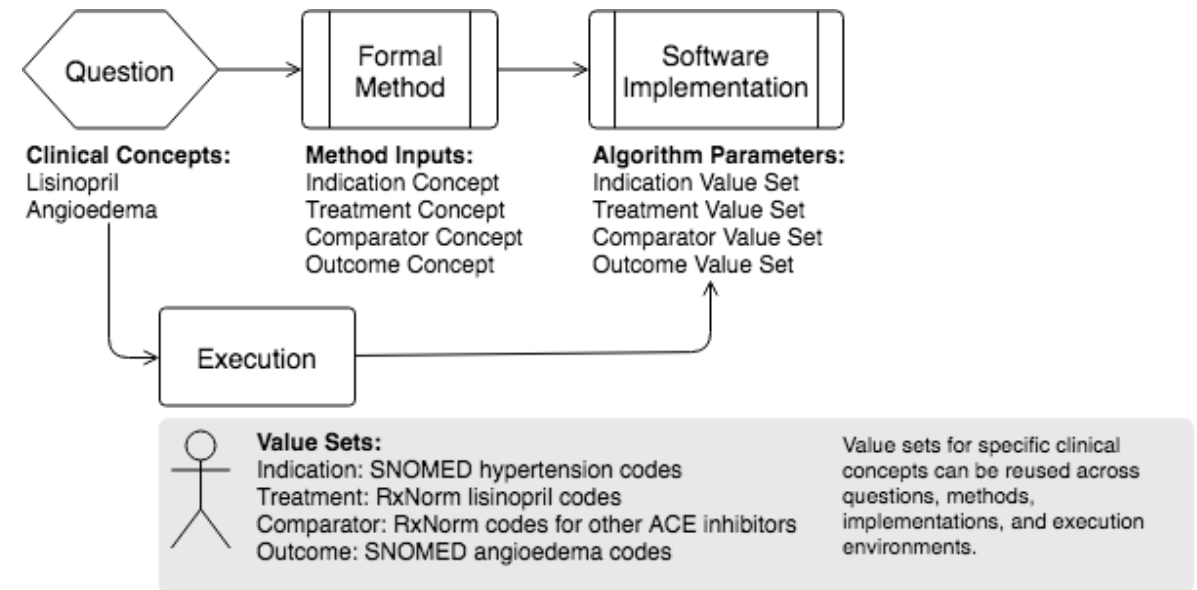


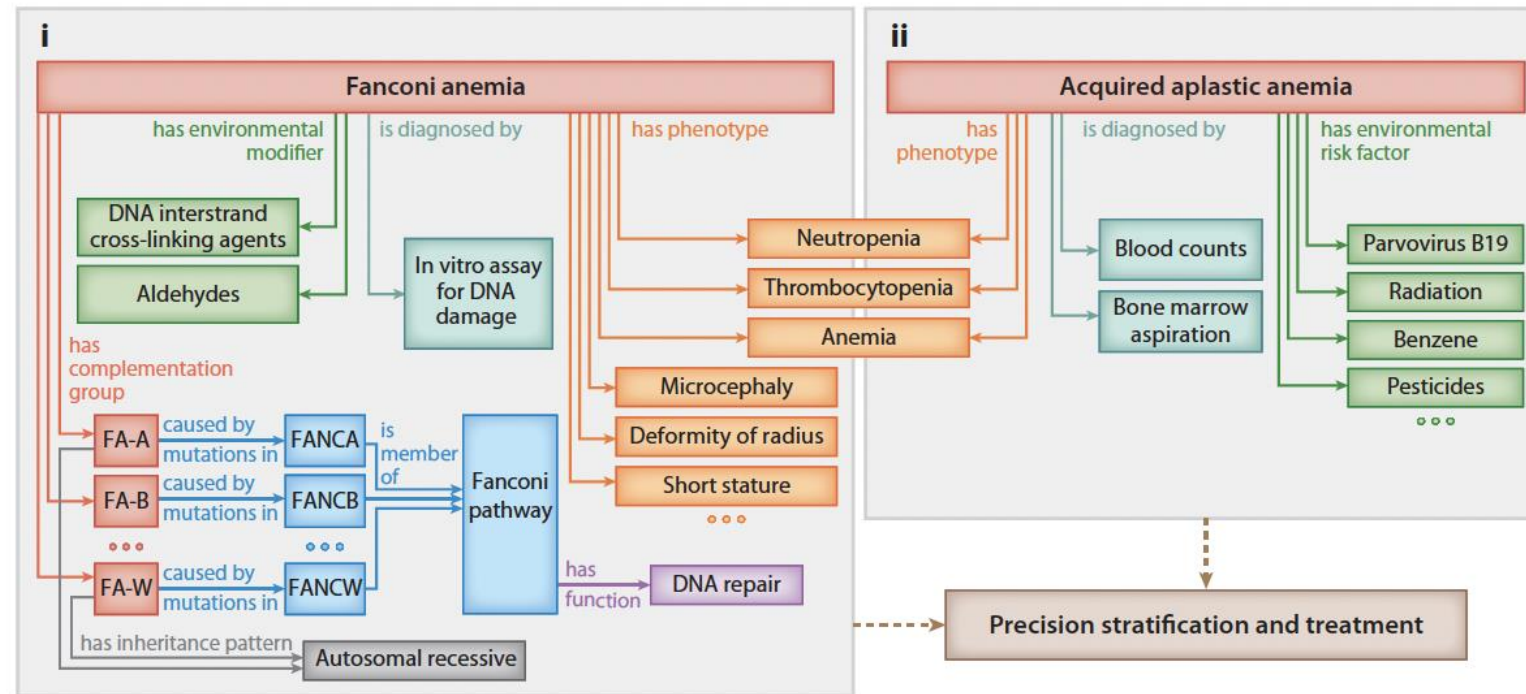
Figure 2. Value sets represent question concepts as parameters for method execution.

- Analyst, as researcher, asks, "Does lisinopril cause angioedema?"
- As biostatistician, chooses a formal effect estimation method.
- As data scientist or programmer, chooses or constructs implementation that require method input be specified as value sets.
- As informaticist or end user, chooses or constructs value sets to represent clinical concepts for execution.

Beyond lists and taxonomies: ontologies (Haendel, 2018)



e Knowledge graphs of Fanconi anemia (i) and acquired aplastic anemia (ii)



Bringing it all together

Toward semantic interoperability – “computer utterance” in one system has same effect in any other (Dolin, 2011)

Needs platforms for both major types of data

- Documents – IHE specifications, CCDA
- Discrete – structured lab results, diagnoses, etc. using LOINC, SNOMED CT, etc.