

# LOINC Ontology PREVIEW Release Documentation

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# 1 Introduction

### 1.1 Background

A new cooperative agreement [1] was signed between the Regenstrief Institute (RII) and SNOMED International (IHTSDO) in October 2022. The organizations agreed to work together to minimize duplication between LOINC [2,8] and SNOMED CT [9,10] and to enhance the ability of stakeholders to use the two terminologies together. A primary aim of the agreement is to create an extension of SNOMED CT for the LOINC terminology. This effort builds on strengths of both terminologies by increasing the computability of LOINC and making the more granular observables in LOINC available to be used in a SNOMED format integrated with SNOMED CT concepts.

### 1.2 Purpose

- This document is intended to give a brief description, background context and explanatory notes on the first preview of the LOINC Ontology.
- This is not a detailed technical document about the release file formats, use cases, or on implementing LOINC and SNOMED CT together.
- Information about a production release of the file will be available in the future.

### 1.3 Audience

The intended audience for this document includes National Release Centers, vendors of electronic health record software, and terminology developers / managers who wish to partake in the review of the first preview of the LOINC Ontology of SNOMED CT.



# 2 Content Development Activity

### 2.1 Overview

The preview release is RF2 compliant and contains a full release file and a snapshot file. The Production content is released under the 11010000107 |LOINC Ontology module (core metadata concept)| with concept identifiers from the 20231001 version of the International release of SNOMED CT and Version 2.76 of LOINC released September 2023. This preview release includes the LOINC Extension of SNOMED CT plus the full international release of SNOMED CT.

This preview includes approximately 23840 SNOMED CT concepts representing active laboratory LOINC Terms. Areas of content specifically excluded from this preview include:

- LOINC Terms that include divisors, such as Protein/Creatinine [Ratio] in Urine
- LOINC Terms considered a calculated or derived observable, such as Anion gap in Blood
- Panels
- Veterinary specific LOINC Terms
- LOINC Terms that have a status of "Discouraged," "Trial," and "Deprecated"
- All other domains of LOINC including clinical, documents, surveys, etc.

Note: The "LOINC Term to SNOMED CT Expression Reference Set" was released in 2017 under the 715515008 |LOINC - SNOMED CT Cooperation Project module (core metadata concept). As this reference set is no longer maintained or released, 715515008 | LOINC - SNOMED CT Cooperation Project module (core metadata) will be inactivated in 2024

### 2.2 Content usage

The main purposes of the extension are to (a) create SNOMED CT concepts for LOINC terms using the SNOMED CT concept model to provide a consistent representation of similar concepts, (b) limit duplication of effort related to overlapping areas, and (c) focus resources on common and collaborative effort. In practical terms there are a variety of ways to deploy and benefit from the resulting extension product.

This preview contains an extension of SNOMED CT for laboratory LOINC Terms which have been evaluated thus far in the project. The LOINC Terms are represented with a SNOMED CT concept identifier, SNOMED CT attributes and values, as well as the LOINC Code identifier. The LOINC concepts can be used on their own, or in the context of the SNOMED CT International release. When used on their own, a hierarchy is created amongst the LOINC concepts. When used in the context of



the SNOMED CT International release, a hierarchy is created amongst the LOINC concepts and between LOINC concepts and defined SNOMED CT observable entity concepts.

### 2.3 Content detail

#### 2.3.1 SNOMED CT concept modeling for LOINC Terms

- This preview contains 23835 Laboratory LOINC Terms modeled using the SNOMED CT observable entity model [3,4,5]. This preview release includes quality observables only. For a list of attributes and description formats for the Observables model used in this work, see the templates developed thus far for this project [3,4].
- All of the LOINC concepts in the preview are subclasses of 363787002 | Observable entity (observable entity) in SNOMED CT
- See Figure 1 for an example of a LOINC Term represented in the SNOMED CT structure
- The majority of the LOINC concepts (~80%) included are fully defined.
- Section "Modeling Guidelines" contains general rules on the modeling of LOINC concepts in the extension

Parents				
> Observable entity (observable entity)				
<ul> <li>Presence of</li></ul>	$\begin{array}{l} \mbox{Property} \rightarrow \mbox{Presence (property)} \\ \mbox{Component} \rightarrow \mbox{Calcium} \\ \mbox{Time aspect} \rightarrow \mbox{Single point in time} \\ \mbox{Direct site} \rightarrow \mbox{Blood specimen} \\ \mbox{Scale type} \rightarrow \mbox{Ordinal value} \end{array}$			

**Figure 1**. Example of a concept in the preview of the LOINC Extension of SNOMED CT. This shows a LOINC term "Calcium:PrThr:Pt:Bld:Ord" (LOINC code 27182-5) rendered as a SNOMED CT observable entity concept. The fully specified name (FSN) and preferred term (PT) of the concept is in a SNOMED compliant format. Additional descriptions are included for the LOINC fully specified



name (Calcium:PrThr:Pt:Bld:Ord) and the LOINC long common name (Calcium [Presence] in Blood). Note the included LOINC code ID is a new feature in SNOMED CT. This concept is designated as sufficiently defined in SNOMED CT - indicated here by the three black bars in the yellow oval..

# 3 Modeling Guidelines

## 3.1 General Guidelines

- LOINC Terms are defined using the Observable entity model and a set of templates developed for this phase of the project
- A script was developed that utilizes a map from ~6000 LOINC Parts to SNOMED CT concepts, an additional LOINC detail file which further specifies LOINC Parts, and a set of editorial rules to create the concepts in a SNOMED CT compliant format
- Primitive grouper only concepts are not included as the SNOMED CT Editorial Guide discourages the use of intermediated primitive groupers [6].
- SNOMED CT Expression Constraint Language (ECL) is the preferred method for querying the LOINC concepts

## 3.2 LOINC Part to SNOMED CT Concept and Attribute

## Representation

While a LOINC Part to SNOMED CT Concept map is not listed as part of the formal agreement, such a map is used as the basis for the modeling and terming of the LOINC Term as a SNOMED CT concept. The map currently contains over 6000 LOINC Parts. Table 1 below shows how each LOINC axis is represented by a SNOMED CT attribute in the preview release. Note the table includes the representation for quality observables, the only type of observables included in this preview. Process observables have a different model in SNOMED CT.

LOINC axis	SNOMED CT attribute	Note
Component	246093002   Component (attribute) OR 704319004   Inheres in (attribute)	-The Property of the LOINC Term determines if the LOINC Component is modeled with 246093002   Component (attribute) or 704319004   Inheres in (attribute), e.g., a



	704326004   Precondition (attribute) (included when the LOINC Component includes a challenge)	term with property of mass concentration is modeled using 246093002   Component (attribute) whereas a term representing an inherent part of component such as type is modeled using 704319004   Inheres in (attribute). -When the LOINC Component includes an adjustment and/or a count, this additional information is included in the terming of the LOINC concept but not by an attribute in the model.
Property	370130000  Property (attribute)	
Scale	370132008   Scale type (attribute)	
System	704327008   Direct site (attribute) OR 704319004   Inheres in (attribute)	704319004   Inheres in (attribute) is used when the LOINC Term has a property type of Prid, Type, Number of cells, ID
Time	370134009   Time aspect (attribute)	
Method	246501002   Technique (attribute)	

**Table 1**. Representation of the 6 LOINC axes by a SNOMED CT attribute when modeling quality observables in the preview release.

## 3.4 LOINC Term represented as a SNOMED CT concept

• After the above attributes and values are applied to the LOINC concept, a concept definition status (sufficiently defined or primitive) is applied to each concept. Our goal is to have the



majority of the concepts designated as sufficiently defined. Where this is not possible, the concept is designated as primitive. Some of the concepts will remain as primitive in future releases and some will change to fully defined as the work progresses.

- For example, LOINC Terms that include "unidentified", "other", "NOS," "unk sub," "unknown," "unspecified," etc. are designated as primitive since this type of information is out of scope of SNOMED CT and thus these concepts are expected to remain as primitive.
- Some concepts are designated as primitive in this preview because a map from the needed LOINC Part to SNOMED CT concept does not yet exist, but is expected to be created in the future, e.g., the SNOMED CT concept for LOINC Term 25398-9 Echinococcus sp Ab [Units/volume] in Serum by Counterimmunoelectrophoresis (CIE) is marked as primitive because a SNOMED CT technique concept for "Counterimmunoelectrophoresis (qualifier value)" has not been created yet. Once the technique concept is created and the LOINC concept is updated, this concept can be marked as sufficiently defined.
- Automated terming rules are applied to the LOINC concepts according to the editorial guidelines developed during the project. This enables creation of a fully specified name and preferred term for each concept that is compliant with SNOMED CT editorial guidelines.
- When the LOINC concepts are classified using the reasoner in SNOMED CT, more specific LOINC concepts are subsumed by more general LOINC Terms. Some examples include:
  - LOINC concepts with a more specific substance are subsumed by concepts with a more generic substance
  - LOINC concepts with a technique are subsumed by concepts with a more generic method or methodless expressions. See Figure 2 for an example.
  - LOINC concepts containing adjustments are subsumed by generic or unadjusted concepts
  - LOINC concepts containing challenges are subsumed by generic concepts
  - LOINC concepts with a more specific specimen are subsumed by more generic concepts, e.g. Arterial blood is necessarily a kind of Blood
- ✓ Number areic of leucine crystal in urine sediment at point in time (observable entity)

■ ■ Number areic of leucine crystal in urine sediment at point in time by high power field light microscopy (observable entity)

- (=) Number areic of leucine crystal in urine sediment at point in time by low power field light microscopy (observable entity)

**Figure 2**. Example of the hierarchy created in LOINC concepts after classification with the reasoner in SNOMED CT. Two LOINC concepts with specific methods are children of a LOINC concept which does not specify a method.



- SNOMED CT Expression Constraint Language (ECL) [7] is used to query the LOINC concepts and return a set of concepts that satisfies the query. ECL is expected to be used in place of primitive grouper concepts for retrieval. Appendix A contains a list of sample ECL queries which can be executed in the preview browser for example purposes. A brief example of a simple ECL query is shown below.
  - The following ECL expression returns a list of all LOINC Terms which include some type of antibody as value of the Component attribute:
    - << 363787002 |Observable entity (observable entity)| : 246093002</li>
       |Component (attribute)| = << 68498002 |Antibody (substance)|</li>
    - This query produces over 4000 results when performed on the extension preview including 239191010000108 |Presence of cryoglobulin in serum at point in time by 5D cold incubation (observable entity)| (LOINC term 12205-1)

## 3.5 Known issues

- Some terming in this preview needs to be improved, e.g.:
  - Presence of Cryptosporidium DNA in stool at point in time by non-probe.amp.tar (observable entity) for LOINC term 82205-6 Cryptosporidium sp DNA [Presence] in Stool by NAA with non-probe detection. Phrases like "non-prob.amp.tar" are not compliant with rules for proper descriptions in SNOMED CT.
  - Mass concentration of C reactive protein in blood at point in time by high sensitivity (observable entity) for LOINC term 71426-1 C reactive protein [Mass/volume] in Blood by High sensitivity method. The term "...by high sensitivity" in the concept FSN and PT might be updated to "...by high sensitivity method."
  - The FSN and PT of the LOINC extension concepts are automatically created based on the terming of the SNOMED CT concepts used in the modeling of the LOINC extension concepts. Therefore, issues or inconsistencies in the terming of the SNOMED CT content will appear in the FSN and PT of the LOINC extension concepts. This legacy SNOMED CT content that does not adhere to current editorial guidelines will be updated as resources allow.
- Some Terms defined by LOINC Parts require additional discussion before finalizing the modeling and terming, e.g., LOINC Terms defined by LOINC Part "Antibiotic XXX" ('other antibiotic').
- Inclusion of Great Britain (GB) language refset descriptions containing American English terming, e.g., hemoglobin.

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## 4 Future steps

Future steps in the project include:

- Resolve known issues.
- Analyze feedback received on the preview
- Decide on modeling for calculated observables
- Decide on modeling for high use laboratory LOINC terms not included in the preview
- Include LOINC Term Category status of "Order only," "Observation only," or "Both order and observation" for each LOINC concept using the annotation functionality once it is available. Note: 10071010000104 |Has concept categorization status (attribute)| was originally for this purpose. It might be replaced or renamed when used as an annotation.
- Expand the LOINC Part to SNOMED CT concept map so that additional LOINC Terms can be included in the work and more LOINC concepts can be marked as sufficiently defined, which will improve classification and querying of the content.

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## **5** References

- 1. https://www.snomed.org/news/new-collaboration-agreement-between-snomedinternational-and-loinc%C2%AE-from-regenstrief
- 2. Logical Observation Identifiers Names and Codes (LOINC): <u>http://loinc.org/</u>
- 3. SNOMED CT Observables Model template for Quality observable with Component for LOINC (observable entity) v1.0: <u>https://confluence.ihtsdotools.org/x/zleaBw</u>
- 4. SNOMED CT Observables Model template for Quality observable with Inheres in for LOINC (observable entity) v1.0: https://confluence.ihtsdotools.org/x/-4BpCg
- 5. Observable Entity Attributes Summary in the SNOMED CT Editorial Guide: https://confluence.ihtsdotools.org/x/JpFpCg
- 6. SNOMED CT Editorial Guide section on "Grouper Concepts:". <u>https://confluence.ihtsdotools.org/x/ZpVpCg</u>
- 7. SNOMED CT Expression Constraint Language Specification and Guide: https://confluence.ihtsdotools.org/x/xo2ZAQ
- 8. "LOINC Users' Guide" available here: <u>https://loinc.org/download/loinc-users-guide/</u>
- 9. SNOMED International (formerly and also known as The International Health Terminology Standards Organization IHTSDO): <u>http://www.snomed.org/</u>
- 10. SNOMED CT Editorial Guide: https://confluence.ihtsdotools.org/x/3Y9pCg



# Appendix A - Example ECL Queries

The table below lists example ECL queries which can be performed to retrieve a set of concepts that matches the criteria delineated in the ECL expression.

The image below shows how an ECL expression can be placed into the preview browser to return a set of concepts.

		Perspec	tive: Full - Feedback	About 👻 📕	SNOMED Leading heat
Ι	Concept Details	Expression Constraint Queries			
	Enter an ECL	QUOIY (ECL Version: 2.0)		c	lear Help
J	<< 363787002  0 (substance)  AND	bservable entity (observable entity)  704327008  Direct site (attribute)  =	: 246093002  Component (atti 122592007  Acellular blood (s	ibute)  = << 684980 erum or plasma) sp	102  Antibody ecimen (specimen)
	ECL Builder	Execute			
	Enter additional s	earch filter (optional)	8		
	Description type:	✓ Language Refsets ✓ M	lodules 🕶		
	Results: Four	id 245 concepts	Preferred Term		Id
	Presence of blo plasma at point entity)	od group antibody in serum or in time by elution (observable	Presence of blood group and plasma at point in time by el	ibody in serum or ution	277051010000108

ECL query	Text description of query	Example results:
< 363787002  Observable entity (observable entity)  : 246501002  Technique (attribute)  = *	Query for all observable entity concepts which have a 246501002  Technique (attribute)	>7000 concepts including 154831010000100  Substance concentration of pro-brain natriuretic peptide in serum or plasma at point in time by immunoassay (observable entity)
<< 363787002  Observable entity (observable entity)  : 246093002  Component (attribute)  = << 68498002  Antibody (substance)  AND 704327008  Direct site (attribute)  = << 122592007	Query for all observable entity concepts where 246093002  Component (attribute)  is a or is a subtype of 68498002  Antibody (substance)  and 704327008  Direct site (attribute)  is a or is a subtype of 122592007  Acellular blood (serum or plasma) specimen (specimen)	>3000 concepts including 204371010000103  Presence of Aspergillus fumigatus serotype 3 antibody in serum at point in time (observable entity)



Acellular blood (serum or plasma) specimen (specimen)		
<< 363787002  Observable entity (observable entity)  : 246093002  Component (attribute)  = << 68498002  Antibody (substance)  AND 704327008  Direct site (attribute)  = 122592007  Acellular blood (serum or plasma) specimen (specimen)	Query for all observable entity concepts where 246093002  Component (attribute)  is a or is a subtype of 68498002  Antibody (substance)  and 704327008  Direct site (attribute)  is 122592007  Acellular blood (serum or plasma) specimen (specimen)	>200 concepts including 59001010000108  Mass concentration of complement C3 nephritic factor in serum or plasma at point in time (observable entity)
> 80151010000108  Mass concentration of monoclonal IgG in serum at point in time (observable entity)	Query for all ancestors of 80151010000108  Mass concentration of monoclonal IgG in serum at point in time (observable entity)	3 concepts including 132201010000103  Mass concentration of immunoglobulin G in serum or plasma at point in time (observable entity)
>! 176671010000107  Arbitrary concentration of Afipia felis IgG in serum at point in time (observable entity)	Query for parent of 176671010000107  Arbitrary concentration of Afipia felis IgG in serum at point in time (observable entity)	253191010000109  Arbitrary concentration of immunoglobulin G in serum at point in time (observable entity)
< 96371010000104  Arbitrary concentration of alpha- fetoprotein in body fluid at point in time (observable entity)	Query for all descendants of 96371010000104  Arbitrary concentration of alpha- fetoprotein in body fluid at point in time (observable entity)	4 concepts including 137271010000105  Arbitrary concentration of alpha- fetoprotein in pleural fluid at point in time (observable entity)
<< 363787002  Observable entity (observable entity)  : 370130000  Property (attribute)  = << 705057003  Presence (property) (qualifier value)	Query for all observable entity concepts where 370130000  Property (attribute)  is a or is a subtype of 118584009 705057003  Presence (property) (qualifier value)	>1000 concepts including 157591010000108  Presence of Influenza A virus RNA in microbial isolate at point in time by probe with target amplification (observable entity)
< 363787002  Observable entity (observable entity)  {{ C definitionStatus = defined }}	Query for all sufficiently defined observable entity concepts	>18000 concepts including 219541010000104  Presence of hepatitis B virus rRNA in



		specimen at point in time by nucleic acid hybridization probe (observable entity)
< 363787002  Observable entity (observable entity)  {{ C definitionStatusId = 900000000000074008  Primitive  }} : 370130000  Property (attribute)  = << 705057003  Presence (property) (qualifier value)	Query for all primitive observable entity concepts where 370130000  Property (attribute)  is a or is a subtype of 118584009 705057003  Presence (property) (qualifier value)	>1000 concepts including 192351010000101  Presence of opioid receptor agonist in unk sub at point in time (observable entity)
<< 363787002  Observable entity (observable entity)  : 246093002  Component (attribute)  = << 29246005  Immunoglobulin G (substance)  OR 246093002  Component (attribute)  = << 74889000  Immunoglobulin M (substance)	Query for all observable entity concepts where 246093002  Component (attribute)  is a or a subtype of 29246005  Immunoglobulin G (substance)  OR where 246093002  Component (attribute)  is a or is a subtype of 74889000  Immunoglobulin M (substance)	>1500 concepts including 171561010000105  Presence of Borrelia burgdorferi 30kD IgG in serum at point in time by immunoblot assay (observable entity)   and 155551010000107  Arbitrary concentration of Rotavirus IgM in serum at point in time by immunoassay (observable entity)
<< 363787002  Observable entity (observable entity)  : 246093002  Component (attribute)  = << 29246005  Immunoglobulin G (substance)  AND 246093002  Component (attribute)  != << 712606001  Monoclonal immunoglobulin G (substance)	Query for all observable entity concepts where the 246093002  Component (attribute)  is a or is a subtype of 29246005  Immunoglobulin G (substance)  AND where the 246093002  Component (attribute)  is not a or subtype of 712606001  Monoclonal immunoglobulin G (substance)	>900 concepts including 14370101000100  Presence of Hepatitis C virus IgG in plasma or serum or whole blood at point in time by rapid immunoassay (observable entity)