

LOINC and SNOMED CT

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INDIANA UNIVERSITY
DEPARTMENT OF MEDICINE
School of Medicine



Regenstrief Institute
Medical Informatics

A deep space photograph showing a vast field of stars. In the center, there is a bright, glowing nebula or galaxy core, possibly the core of the Milky Way, with a prominent bright white star. The background is filled with numerous smaller, distant stars of varying colors and brightness.

Why am I here?

COOPERATION AGREEMENT

dated July 2013

Between

**The International Health Terminology Standards
Development Organisation
(IHTSDO)**

and

**The Regenstrief Institute, Incorporated
(RII)**

Shared vision.
Cooperation, not duplication.



Happily Ever After

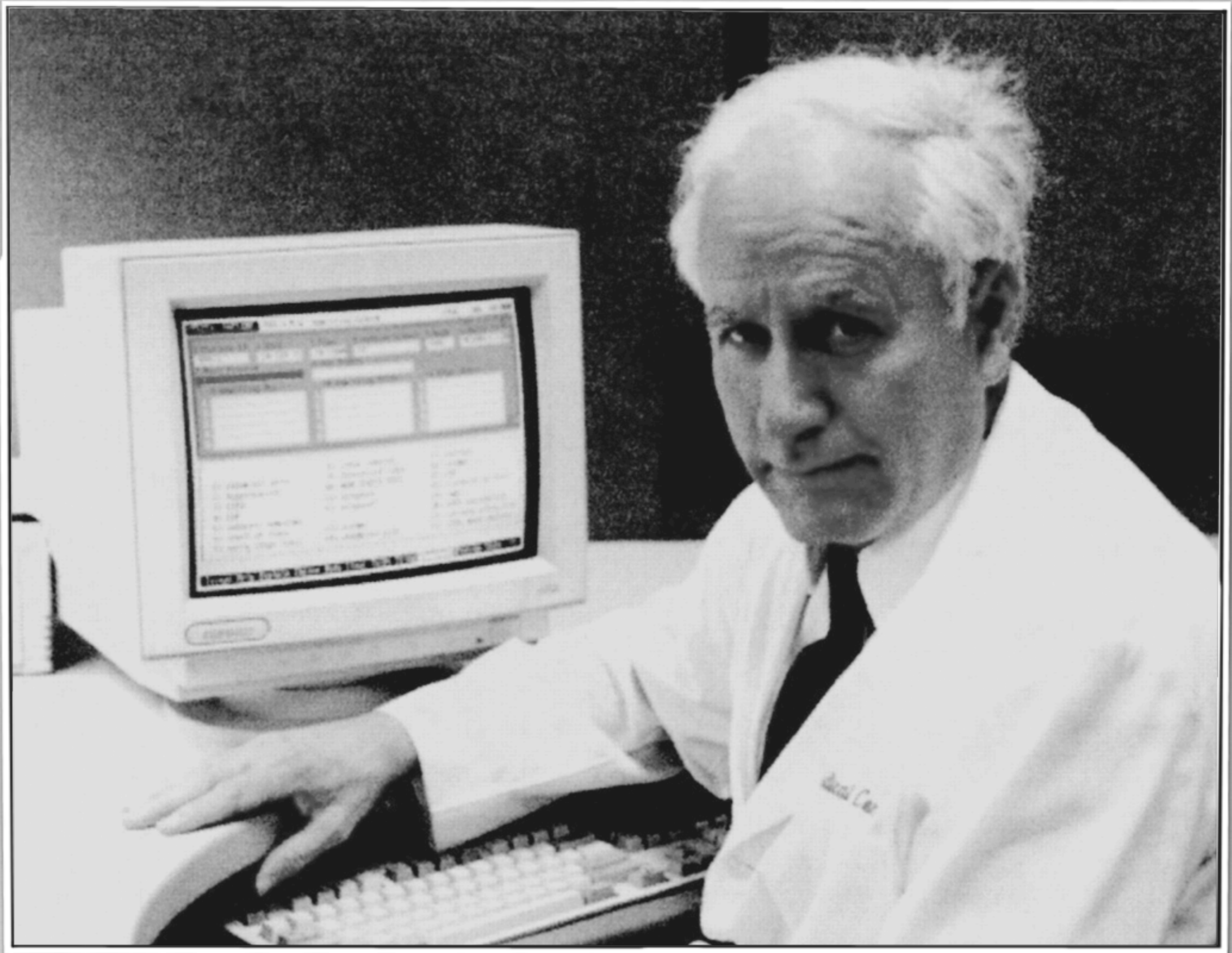
Starts Here



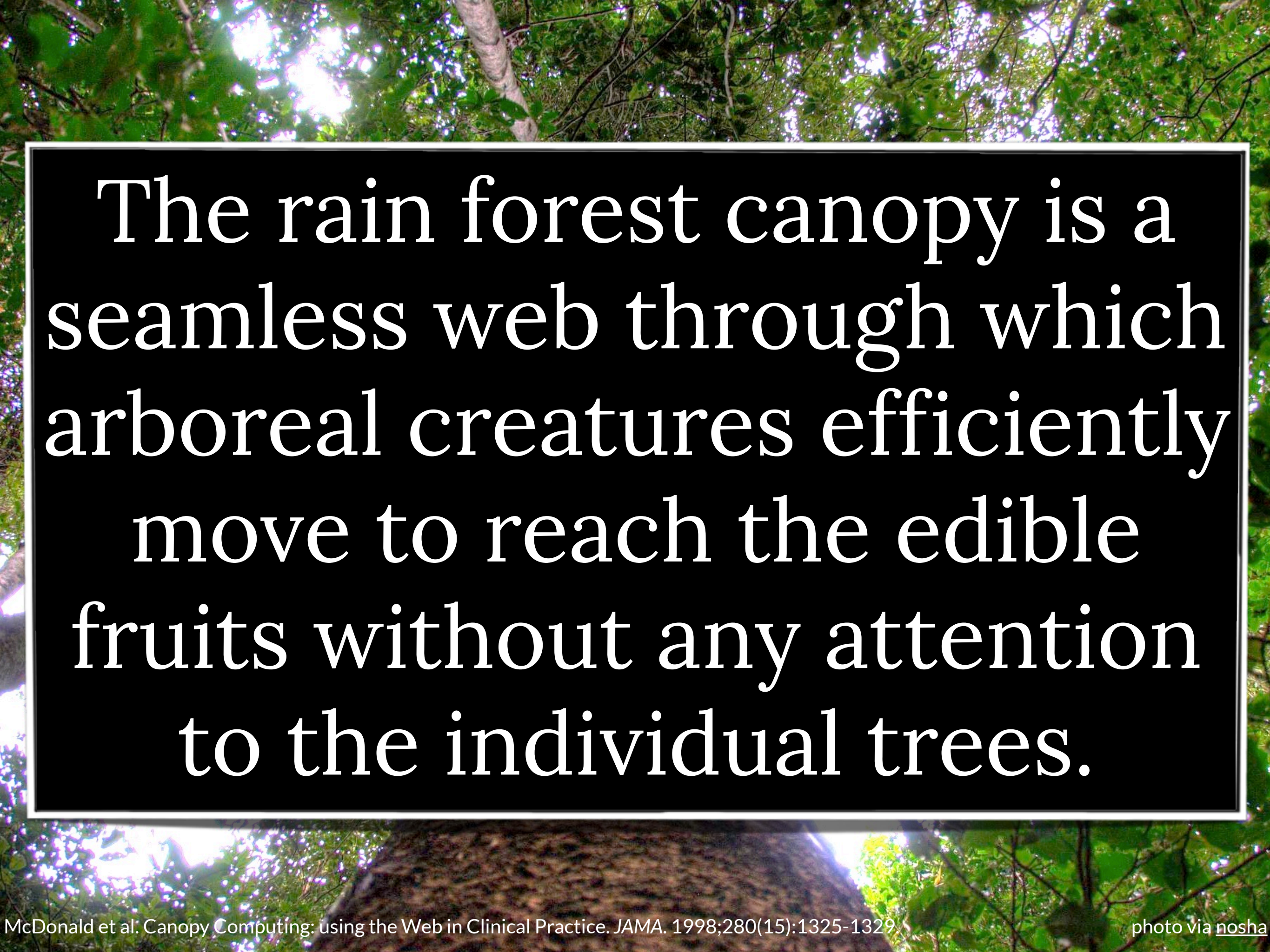


Origins of LOINC

A universal catalog of laboratory and clinical observations







The rain forest canopy is a seamless web through which arboreal creatures efficiently move to reach the edible fruits without any attention to the individual trees.

Fundamental challenge:

Local systems have different ways of identifying the same concept. And, things that look alike aren't the same.

Same or Different?

What you see in the test catalog

Lab A

Test Name: Lyme Disease Serology

Lab B

Test Name: Lyme Disease Antibody

Same or Different?

What you see in the test catalog

Lab A

Test Name: Lyme Disease Serology

Measures: *B. burgdorferi* Ab IgG

Method: ELISA

Scale: quantitative

e.g.: Titer 1:40

LOINC Code = 5062-5

Lab B

Test Name: Lyme Disease Antibody

Measures: *B. burgdorferi* Ab IgM

Method: Immune blot

Scale: qualitative

e.g.: Positive

LOINC Code = 6321-4

MORE THAN MEETS THE EYE





**Only way to corral the
infinite indiosyncracies is
with standards**

Logical Observation Identifiers Names and Codes

**A universal code system that facilitates exchange,
pooling, and processing of results**



PRESS
ME

MEASUREMENTS

“Я”

US

Anatomy of a LOINC Term

CD3+CD4+ (T4 helper) cells [# / volume] in Blood

24467-3:Cells.CD3+CD4+:NCnc:Pt:Bld:Qn:

Anatomy of a LOINC Term

CD3+CD4+ (T4 helper) cells [# / volume] in Blood

24467-3:Cells.CD3+CD4+:NCnc:Pt:Bld:Qn:

24467-3

LOINC Code

Cells.CD3+CD4+

Component

NCnc

Property Measured

Pt

Timing

Bld

System

Qn

Scale

Method

Anatomy of a LOINC Term

CD3+CD4+ (T4 helper) cells [# / volume] in Blood

24467-3:Cells.CD3+CD4+:NCnc:Pt:Bld:Qn:

24467-3

LOINC Code

Cells.CD3+CD4+

NCnc

Pt

Bld

Qn

Component

Property Measured

Timing

System

Scale

Method

There are six major LOINC axes

Anatomy of a LOINC Term

CD3+CD4+ (T4 helper) cells [# / volume] in Blood

24467-3:Cells.CD3+CD4+:NCnc:Pt:Bld:Qn:

24467-3

LOINC Code

Cells.CD3+CD4+

NCnc

Pt

Bld

Qn

Component
Property Measured
Timing
System
Scale
Method



There are six major LOINC axes

Born in 1994 by Regenstrief Institute.

Worldwide distribution at no cost.

Copyrighted, but open license.

Released twice per year.



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LOINC[®]
from Regenstrief

Born in 1994 by Regenstrief Institute.

It's free, but invaluable.

Worldwide distribution at **no cost.**

Just don't use it to make another standard!

Copyrighted, but **open license.**

Released twice per year.

LOINC[®]
from Regenstrief

*If an observation is a question and
the observation value is
an answer...*

LOINC provides codes for
questions

Where needed, other
vocabularies provide codes for
answers

Laboratory LOINC

challenge chemistry tests

Chemistry

Blood Bank

Hematology and Cell counts

Microbiology

Serology

Cell Markers

Antibiotic Susceptibilities

Coagulation

Allergy Testing

Pathology

Urinalysis

HLA Antigens

Drug Doses

Mutations

Drug toxicology

Clinical LOINC

PhenX

Radiology

MDS

Vital Signs

General Health Survey

OASIS

Specific Hemodynamics

General Health Survey panels

Home Health Care Classification

NEURO-Qol

Physical Exam

PROMIS

Dental

History and Physical

Clinical Report Document

OMAHA

End Stage Renal Disease variables

EKG

OB Ultrasound

Respiratory

Cardiac Ultrasound

Tumor Registry

Clinical report attachments



LOINC Collections

Panels, forms, surveys, and other
patient assessments

Standardized Assessments and Collections

Representing Patient Assessments in LOINC®

Daniel J. Vreeman, PT, DPT, MSc^a, Clement J. McDonald, MD^b, Stanley M. Huff, MD^c

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^bLister Hill Center, National Library of Medicine, Washington DC; ^cUniversity of Utah and Intermountain Healthcare, Salt Lake City, UT

ABSTRACT

Without being included in accepted vocabulary standards, the results of completed patient assessment instruments cannot be easily shared in health information exchanges. To address this important barrier, we have developed a robust model to represent assessments in LOINC through iterative refinement and collaborative development. To capture the essential aspects of the assessment, the LOINC model represents the hierarchical panel structure, global item attributes, panel-specific item attributes, and structured answer lists. All assessments are available in a uniform format within the freely available LOINC distribution. We have successfully added many assessments to LOINC in this model, including several federally required assessments that contain functioning and disability content. We continue adding to this "master question file" to further enable interoperable exchange, storage, and processing of assessment data.

INTRODUCTION

Despite progress on many fronts, interoperable health information exchange continues to be hampered by the plethora of idiosyncratic conventions for representing clinical concepts in different electronic systems. Many times, the lack of interoperable connections between systems means that valuable results are unavailable to clinicians when they need it.¹ LOINC® (Logical Observation Identifiers Names and Codes) is a universal code system for identifying

representation of assessments since its early development when it included codes for standardized scales such as the Glasgow Coma Score and the Apgar Score. Prior work^{5,6} has demonstrated the capability of LOINC's semantic model to represent many assessments with only modest extensions.

Over time, we have both significantly refined LOINC's model for patient assessments and added much new content. Here we present a summary of this progress. Specifically, the purpose of this paper is to describe LOINC's model for assessments, the methods and rationale by which this model was developed, the current assessment content, and some of the lessons learned in the process.

BACKGROUND

Fully specified LOINC names are constructed on six main axes (Component, Property, Timing, System, Scale, and Method) containing sufficient information to distinguish among similar observations.² Different LOINC codes are assigned to observations that measure the same attribute but have different clinical meanings. The LOINC codes, names, and other attributes are distributed in the main LOINC database made available at no cost in regular releases on the LOINC website (<http://loinc.org>). In addition to the LOINC database, Regenstrief develops and distributes at no cost a software program called RELMA that provides tools for searching the LOINC database, viewing detailed accessory content, and for mapping local terminology to LOINC terms.

LOINC®: a universal catalogue of individual clinical observations and uniform representation of enumerated collections

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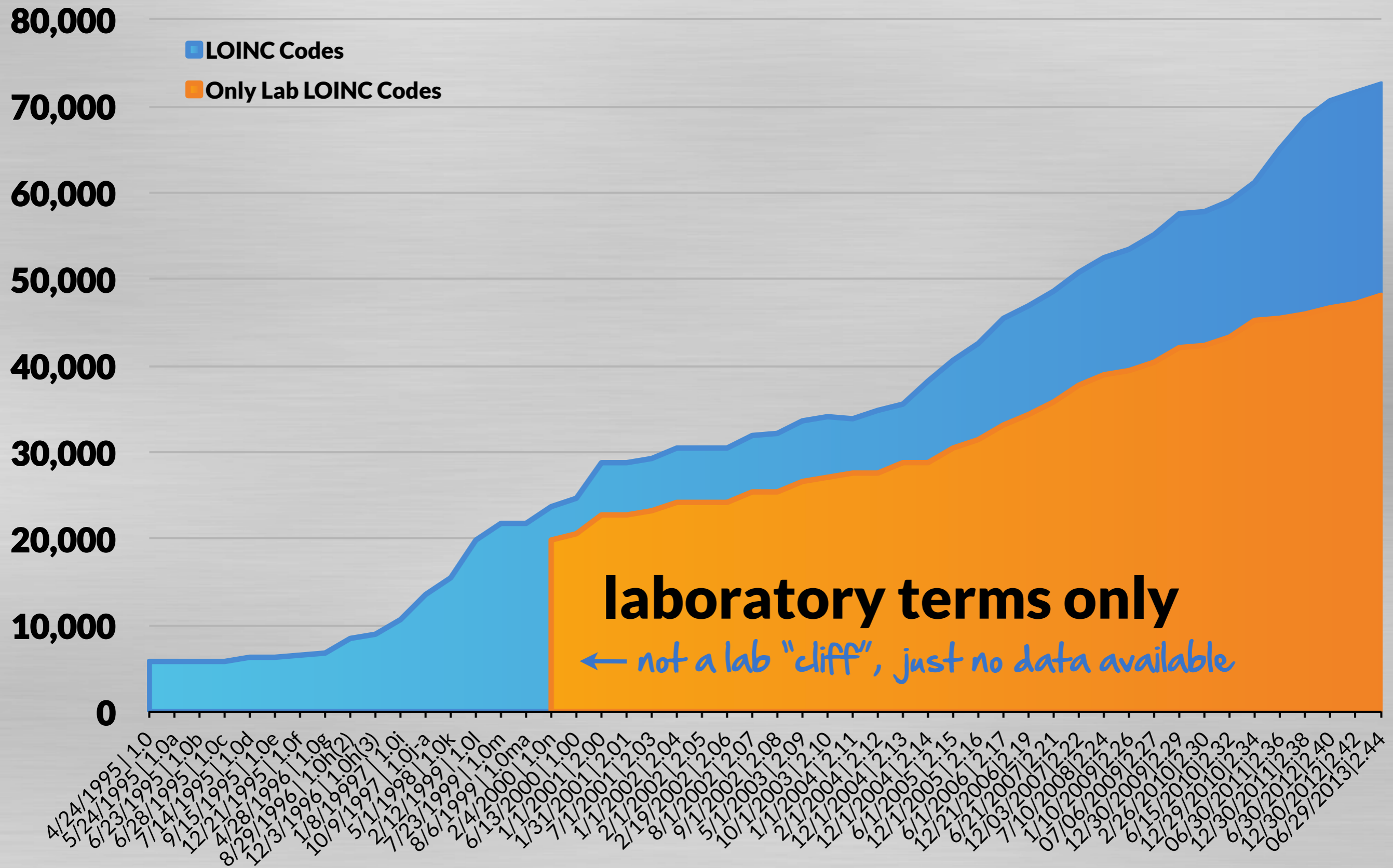
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Abstract: In many areas of practice and research, clinical observations are recorded on data collection forms by asking and answering questions, yet without being represented in accepted terminology standards these results cannot be easily shared among clinical care and research systems. LOINC contains a well-developed model for representing variables, answer lists and the collections that contain them. We have successfully added many assessments and other collections of variables to LOINC in this model. By creating a uniform representation and distributing it worldwide at no cost, LOINC aims to lower the barriers to interoperability among systems and make this valuable data available across settings when and where it is needed.

Keywords: clinical observations; framework; health information technology; patient data; patient assessments; data sets; public health; research; standards; terminology.

LOINC Codes Over Time by Release





Lots Cooking...

Always more lab tests

Genetic reporting

Lots of survey instruments, forms, and assessments

More radiology reports

Structured document titles

...

A black and white photograph of two public payphones mounted on a brick wall. The payphones are dark-colored with a handset on top and a coin slot on the front. The background is a light-colored brick wall with some vertical pipes or conduits. The text is overlaid on the image in a large, bold, black font.

Data standards are like telephones.

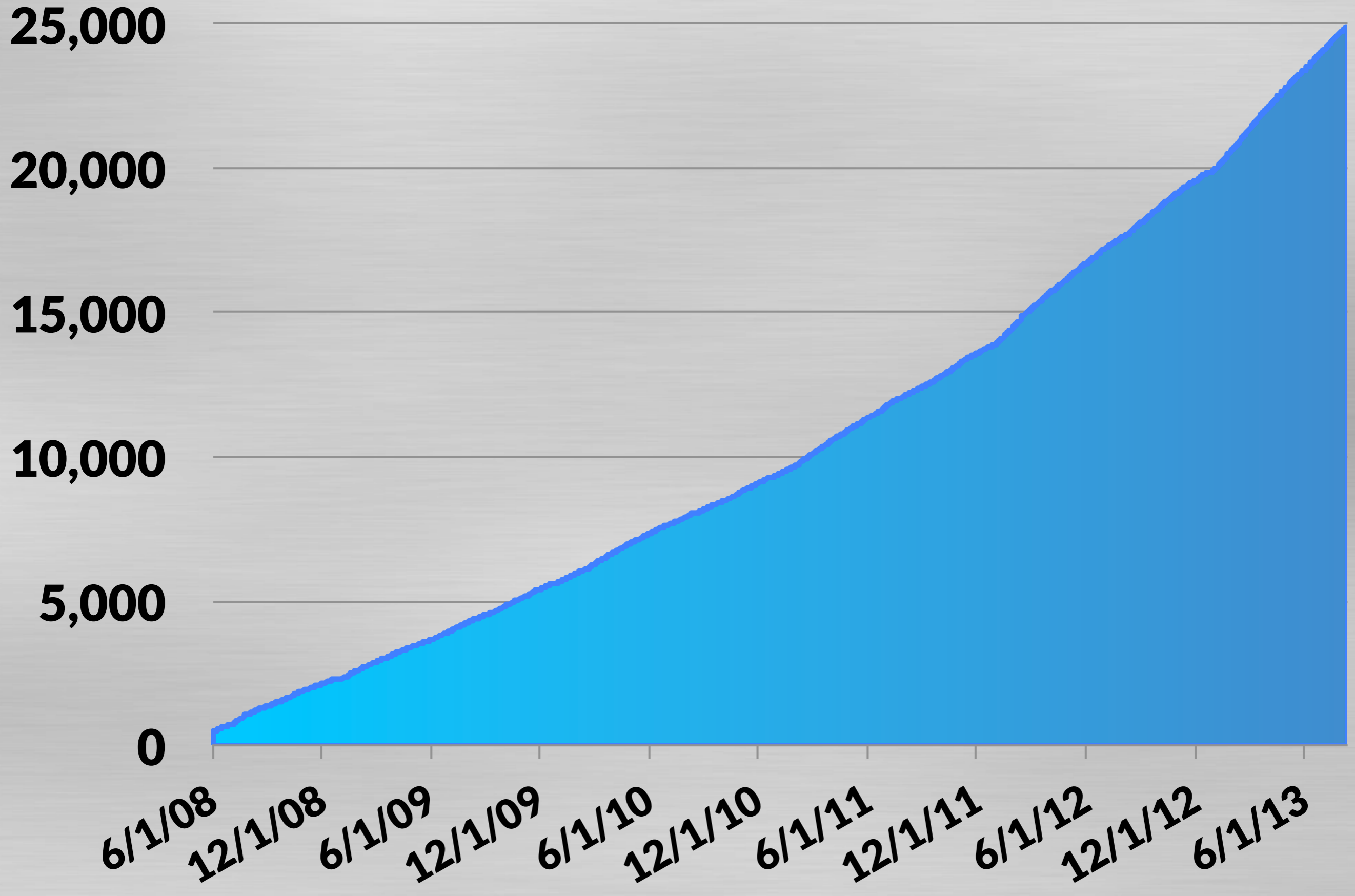
**They require a critical mass of users
before they become useful.**

- Clem McDonald, MD 1998

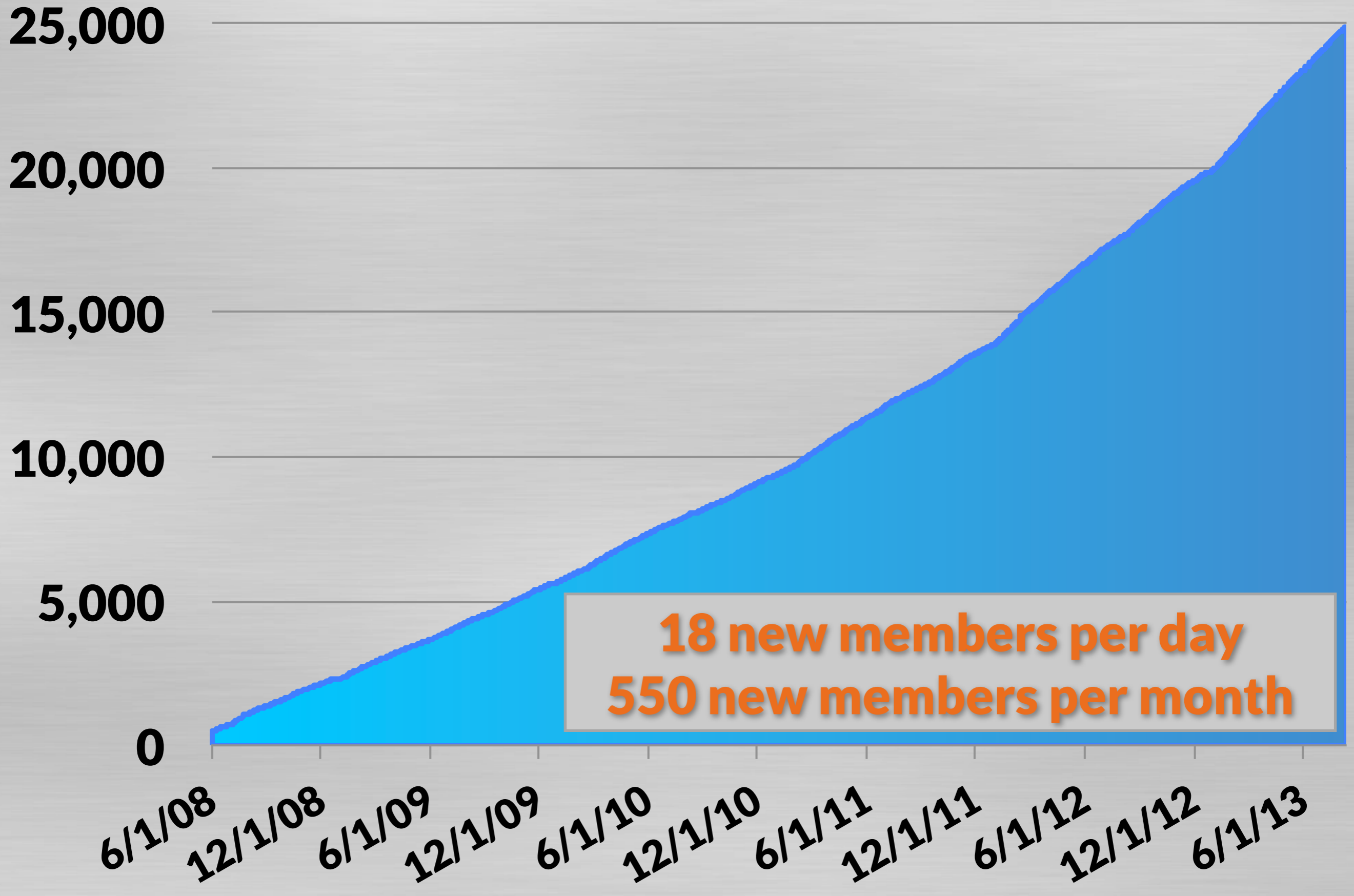
26,000+ users in 155 countries



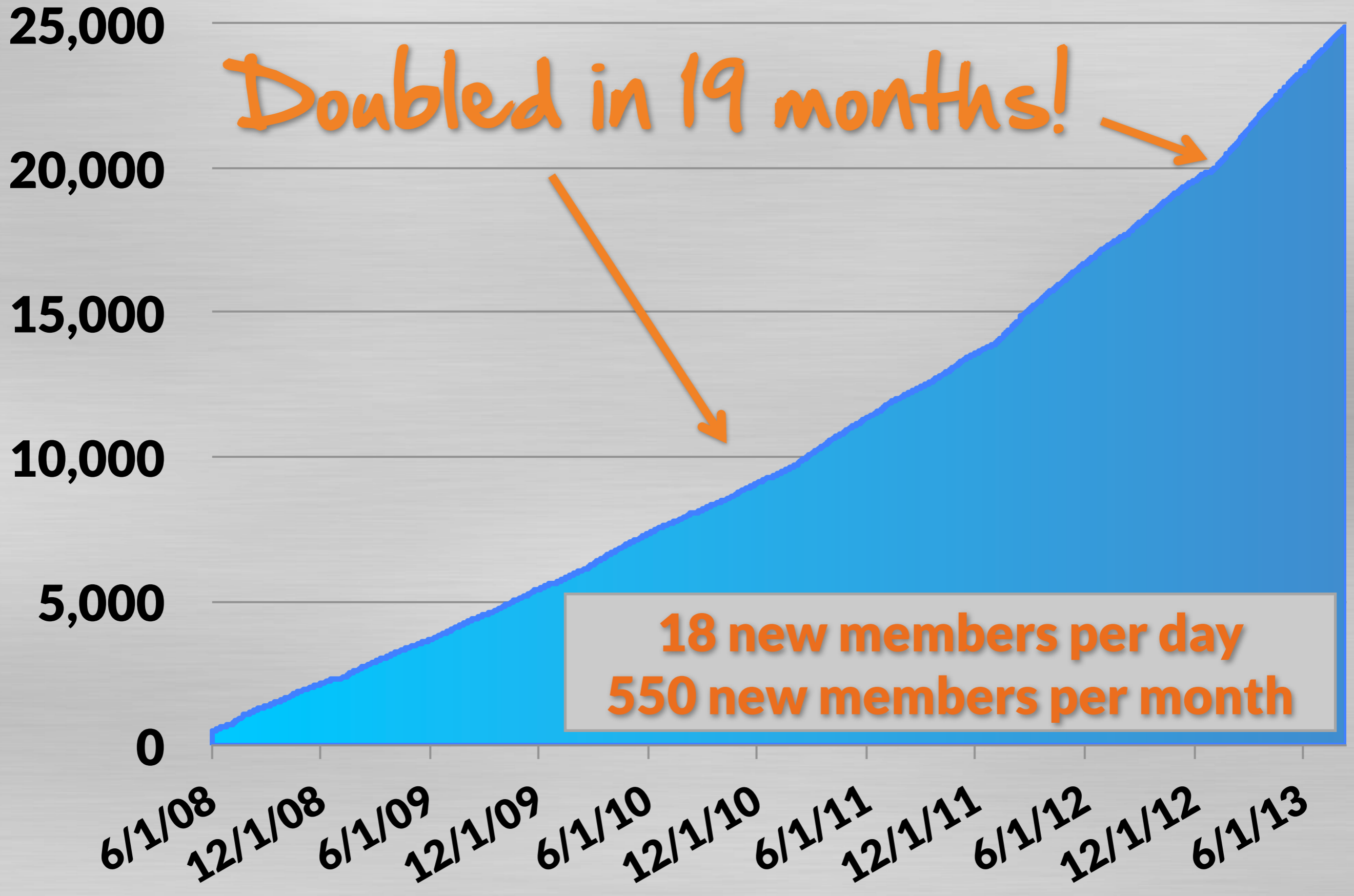
loinc.org members



loinc.org members



loinc.org members



LOINC Translators



22 organizations.

Currently translations into 17 variants of 11 languages

How do *you* say glucose?

Glucose

葡萄糖

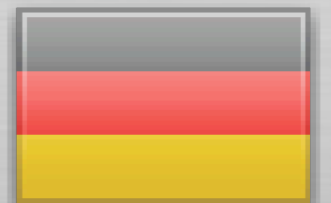
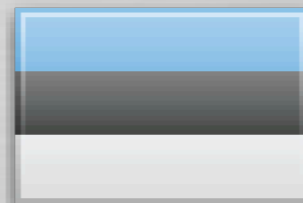
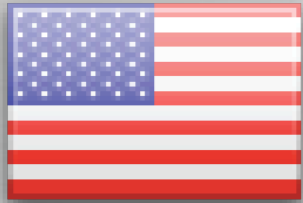
Glükoos

Glucose

Glucose

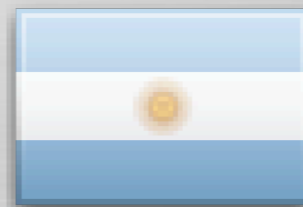
Gluc|kose

Glukose



LOINC

the *lingua franca* of clinical observation exchange



Γλυκόζη

Glucosio

포도당

Glicose

Glucosa

Glucosa

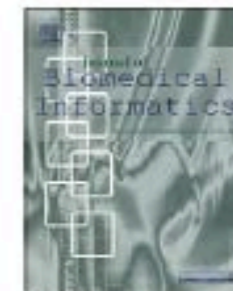
Glucosa



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Enabling international adoption of LOINC through translation

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controlled

Multilingualism

Translating

Clinical laboratory information

systems/standards

Medical records systems

Computerized/standards

ABSTRACT

Interoperable health information exchange depends on adoption of terminology standards, but international use of such standards can be challenging because of language differences between local concept names and the standard terminology. To address this important barrier, we describe the evolution of an efficient process for constructing translations of LOINC terms names, the foreign language functions in RELMA, and the current state of translations in LOINC. We also present the development of the Italian translation to illustrate how translation is enabling adoption in international contexts. We built a tool that finds the unique list of LOINC Parts that make up a given set of LOINC terms. This list enables translation of smaller pieces like the core component “hepatitis c virus” separately from all the suffixes that could appear with it, such “Ab.IgG”, “DNA”, and “RNA”. We built another tool that generates a translation of a full LOINC name from all of these atomic pieces. As of version 2.36 (June 2011), LOINC terms have been translated into nine languages from 15 linguistic variants other than its native English. The five largest linguistic variants have all used the Part-based translation mechanism. However, even with efficient tools and processes, translation of standard terminology is a complex undertaking. Two of the prominent linguistic challenges that translators have faced include: the approach to handling acronyms and abbreviations, and the differences in linguistic syntax (e.g. word order) between languages. LOINC’s open and customizable approach has enabled many different groups to create translations that met their needs and matched their resources. Distributing the standard and its many language translations at no cost worldwide accelerates LOINC adoption globally, and is an important enabler of interoperable health information exchange.

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Adopted as National Standard



Australia

Austria

Belgium

Brazil

Canada

Cyprus

Estonia

Finland

France

Germany

Iceland

Mexico

Mongolia

The Netherlands

New Zealand

Philippines

Portugal

Rwanda

Slovakia

Slovenia

Spain

Thailand

Turkey

United States

Large Implementations

SIGA Saúde project

7+ Provincial systems in Canada

15+ Regional Health Information Exchanges in Spain

ePSOS

Assistance publique - Hôpitaux de Paris

Hong Kong Hospital Authority

Philippine Health Insurance Corporation

InFSE Project in 5 regions in Italy (LOINC Italia)

Many more...

The background of the slide is a stylized, semi-transparent American flag with its characteristic stars and stripes. The text is overlaid on this background.

**In USA, major driver of
eHealth standards adoption is**

EHR Incentive Program

a.k.a. “*Meaningful Use*”

Stage 2

starting 2014

**DEPARTMENT OF HEALTH AND
HUMAN SERVICES**

Office of the Secretary

45 CFR Part 170

RIN 0991-AB82

**Health Information Technology:
Standards, Implementation
Specifications, and Certification
Criteria for Electronic Health Record
Technology, 2014 Edition; Revisions to
the Permanent Certification Program
for Health Information Technology**

AGENCY: Office of the National
Coordinator for Health Information
Technology (ONC), Department of
Health and Human Services.

ACTION: Final rule.

SUMMARY: With this final rule, the
Secretary of Health and Human Services
adopts certification criteria that
establish the technical capabilities and
specify the related standards and
implementation specifications that
Certified Electronic Health Record
(EHR) Technology will need to include
to, at a minimum, support the
achievement of meaningful use by
eligible professionals, eligible hospitals,

CQM Clinical Quality Measure
CY Calendar Year
EH Eligible Hospital
EHR Electronic Health Record
EP Eligible Professional
FY Fiscal Year
HHS Department of Health and Human
Services
HIPAA Health Insurance Portability and
Accountability Act of 1996
HIT Health Information Technology
HITECH Health Information Technology for
Economic and Clinical Health
HITPC HIT Policy Committee
HITSC HIT Standards Committee
HL7 Health Level Seven
ICD-9-CM International Classification of
Diseases, 9th Revision, Clinical
Modification
ICD-10 International Classification of
Diseases, 10th Revision
ICD-10-CM International Classification of
Diseases, 10th Revision, Clinical
Modification
ICD-10-PCS International Classification of
Diseases, 10th Revision, Procedure Coding
System
IHE Integrating the Healthcare Enterprise®
LOINC® Logical Observation Identifiers
Names and Codes
MU Meaningful Use
ONC Office of the National Coordinator of
Health Information Technology
NCPDP National Council for Prescription
Drug Programs
NIST National Institute of Standards and
Technology

a. Ambulatory and Inpatient Setting
b. Ambulatory Setting
c. Inpatient Setting
10. Revised Certification Criteria
a. Ambulatory and Inpatient Setting
b. Ambulatory Setting
c. Inpatient Setting
11. Unchanged Certification Criteria
a. Refinements to Unchanged Certification
Criteria
b. Unchanged Certification Criteria
Without Refinements
12. Gap Certification
13. "Disability" Status
B. Redefining Certified EHR Technology
and Related Terms
1. Certified EHR Technology (CEHRT)
Definition
2. Base EHR Definition
3. Complete EHR Definition
4. Certifications Issued for Complete EHRs
and EHR Modules
5. Adaptations of Certified Complete EHRs
or Certified EHR Modules
IV. Provisions of the Final Rule Affecting the
Permanent Certification Program for HIT
("ONC HIT Certification Program")
A. Program Name Change
B. "Minimum Standards" Code Sets
C. Revisions to EHR Module Certification
Requirements
1. Privacy and Security Certification
2. Certification to Certain New Certification
Criteria
D. ONC-ACB Reporting Requirements
E. Continuation and Representation of
Certified Status

LOINC adopted for:

1. View, download, transmit data to third party
2. Cancer case reporting to state registry
3. Send/receive electronic lab results in ambulatory settings
4. Provide a care summary at care transition
5. Provide clinical summaries for patients
6. Submit reportable lab results to public health

(EHR) Technology will need to include to, at a minimum, support the achievement of meaningful use by eligible professionals, eligible hospitals,

Health Information Technology
NCPDP National Council for Prescription Drug Programs
NIST National Institute of Standards and Technology

2. Certification to Certain New Certification Criteria
D. ONC-ACB Reporting Requirements
E. Continuation and Representation of Certified Status

Use in Quality Measures



Health IT Policy Committee

A Public Advisory Body on Health Information Technology to the National Coordinator for Health IT

September 9, 2011

Farzad Mostashari, MD, ScM
National Coordinator for Health Information Technology
Department of Health and Human Services
200 Independence Avenue, SW
Washington, DC 20201

Dear Dr. Mostashari:

The HIT Standards Committee's (HITSC) Clinical Quality Measures Workgroup (CQM WG) and Vocabulary Task Force (VTF) jointly developed recommendations on the assignment of code sets to clinical concepts [data elements] for use in quality measures.

The CQM WG and VTF held a series of joint meetings to develop the set of recommendations. This letter transmits the recommendations to the Department of Health and Human Services (HHS) on the assignment of code sets to clinical concepts for use in quality measures. On August 17, 2011, the CQM WG and VTF reported on and discussed their findings with the HITSC, which were subsequently approved as outlined below.

LOINC Adopted For

Patient Characteristics

[Non-lab] Diagnostics studies

Patient experience

Family history

Functional status

Health record component

Interventions (that produce an assessment or measured results)

Laboratory tests

Physical exam

Patient preference

Risk evaluation

System resources (healthcare resources)

You too can fall for LOINC





**Agreement paves the way
for new shared benefits
(down the road)**

4 Main Deliverables

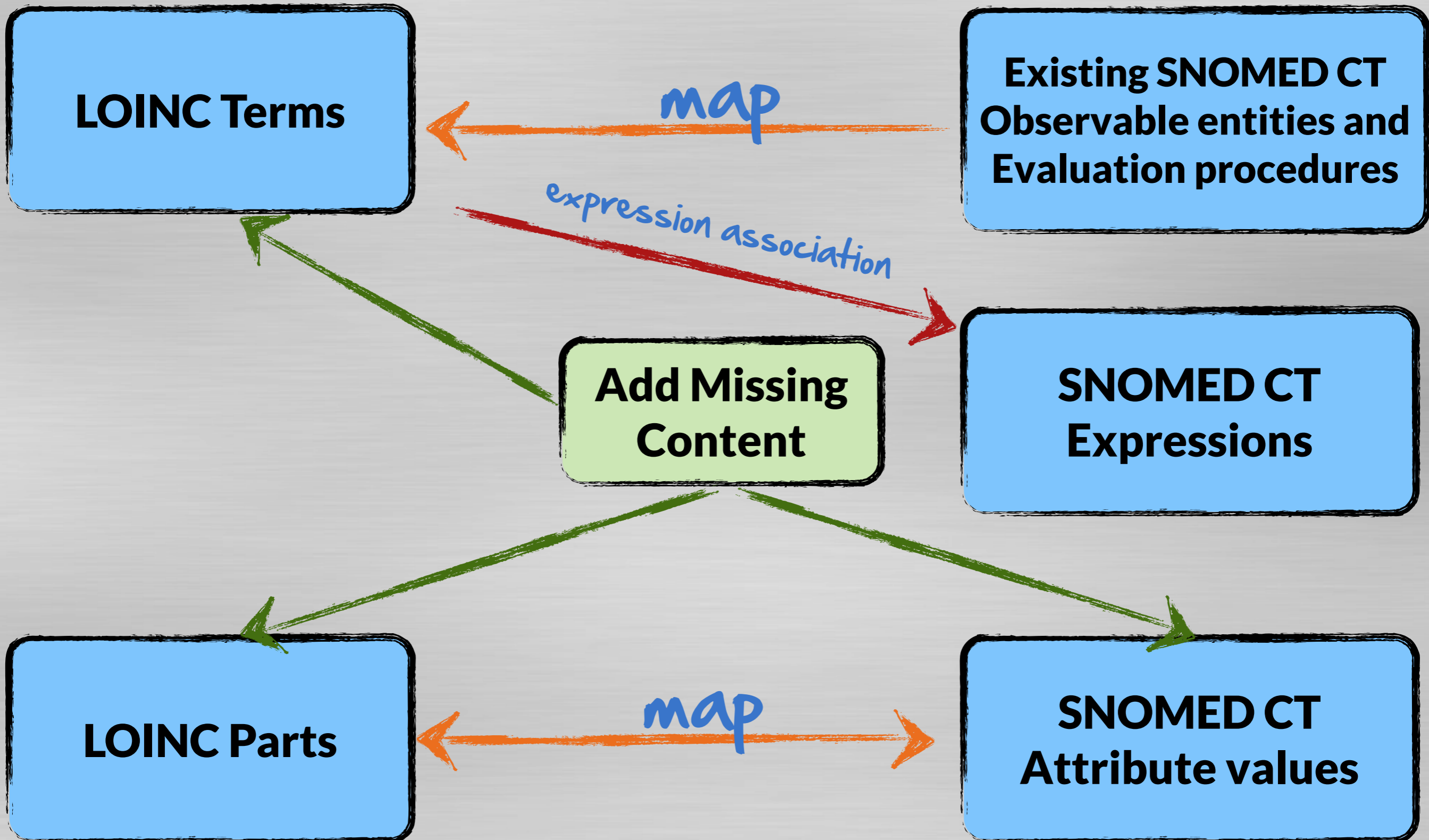
Mapping LOINC Parts to SNOMED CT concepts

Mapping existing SNOMED CT Observables to LOINC terms

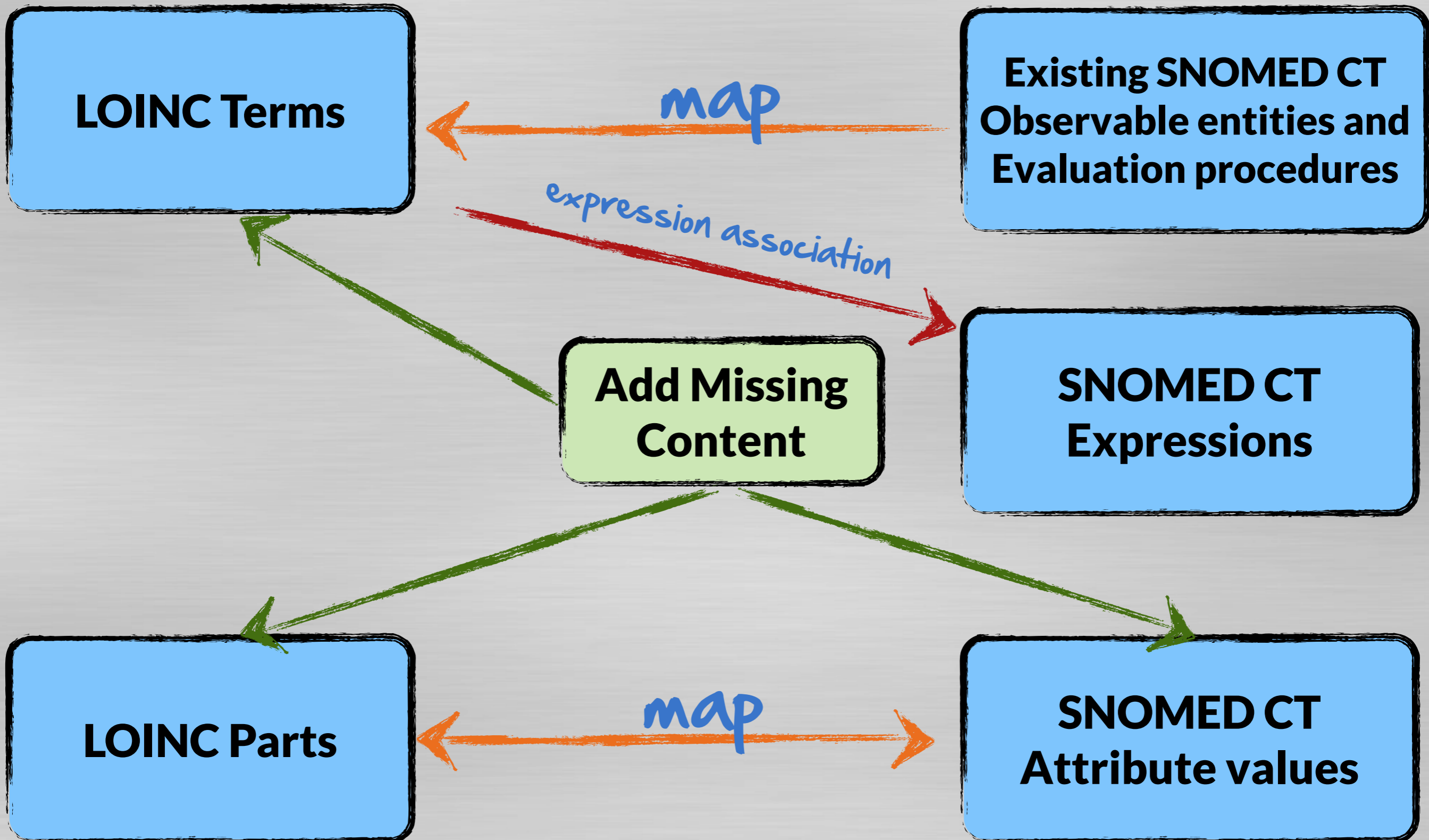
LOINC terms associated with post-coordinated expressions

LOINC terms with categorical answer values linked to SNOMED CT answers

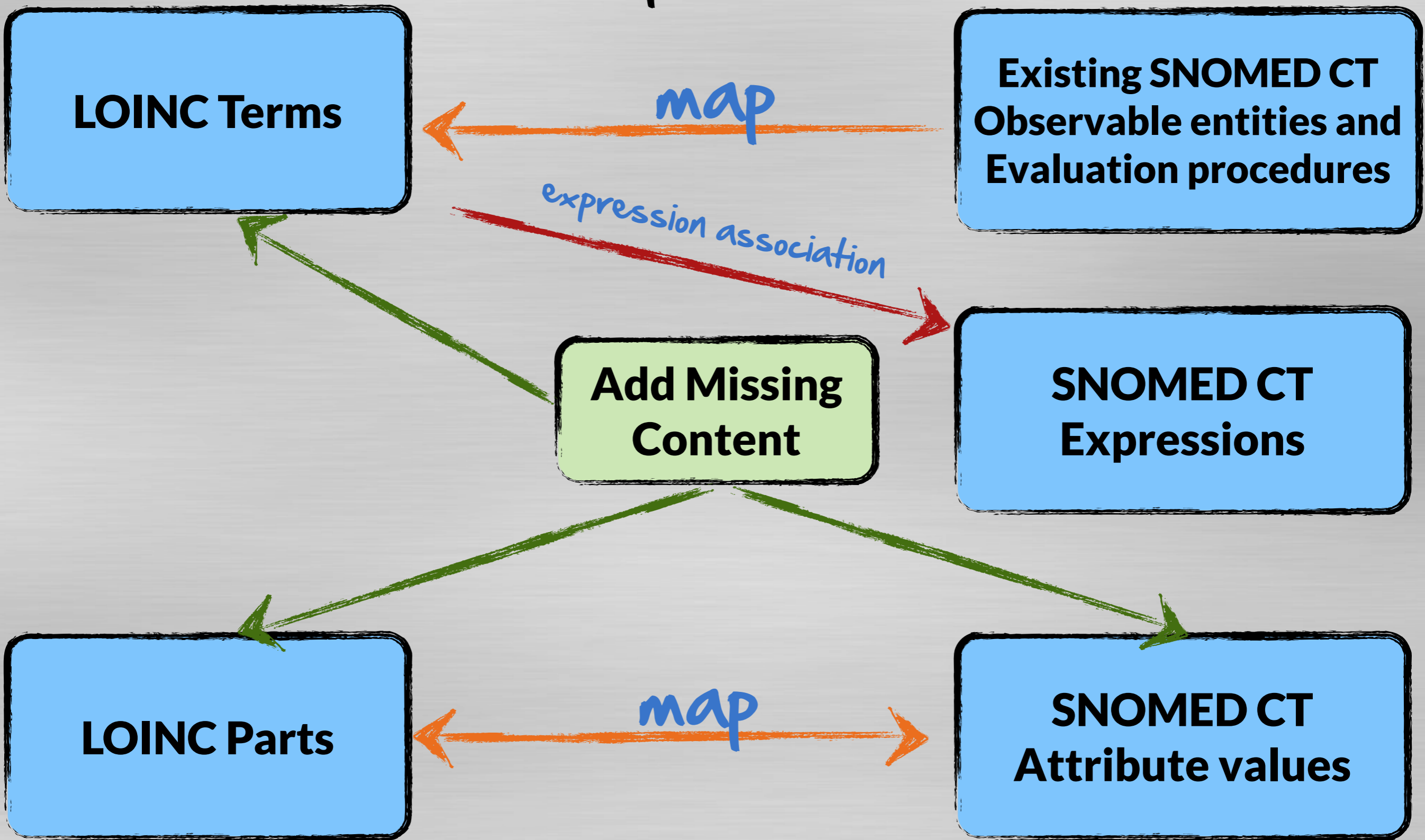
Approach



Maps between LOINC Parts and SNOMED CT - add stuff where needed



LOINC Terms represented in SNOMED CT as "expressions"



How We'll Proceed

Pragmatically, not perfectly

Starting with these domain areas

Laboratory

Including discrete orders and observations and panel names for orders (excluding panel structure)

Anthropomorphic measurements and evaluations

Vital signs and physiological measurements

We'll need some
patience from
users



Using LOINC and SNOMED CT Together



Electronic Lab Reporting to Public Health

Lab to Public Health Reporting

V251_IG_LB_LABRPTPH_R1_INFORM_2010FEB



HL7 Version 2.5.1 Implementation Guide:
Electronic Laboratory Reporting to Public Health,
Release 1 (US Realm)

HL7 Version 2.5.1: ORU^R01

HL7 Informative Document

February, 2010

Sponsored by:

Public Health / Emergency Response Work Group

RCMT

Reportable Condition Mapping Table

Links reportable conditions to associated LOINC laboratory tests and SNOMED results

Acute poliomyelitis (disorder)

SCTID: 398102009

Lab Test Name (from LOINC)

40709-8	Polio virus Ab [Units/volume] in Cerebral spinal fluid
16284-2	Polio virus Ab [Units/volume] in Serum
42980-3	Polio virus Ab [Titer] in Serum
27261-7	Polio virus Ab [Titer] in Serum by Complement fixation
60546-9	Polio virus identified [Type] in Isolate by Organism specific culture
53645-8	Polio virus identified in Stool by Organism specific culture
...lots more...	

Lab Test Result (from SNOMED CT)

44172002	Human poliovirus (organism)
22580008	Human poliovirus 1 (organism)
55174004	Human poliovirus 2 (organism)
16362001	Human poliovirus 3 (organism)

Electronic Laboratory Reporting: Barriers, Solutions and Findings

J. Marc Overhage, Jeffrey Suico, and Clement J. McDonald

Electronic laboratory reporting can improve surveillance for notifiable conditions. Building on standards for message structure and content, we have implemented an electronic laboratory reporting system by building on the infrastructure created for the Indiana Network for Patient Care (INPC). The system has proven reliable in delivering results and scalable to multiple laboratories over 36 months of use. In April 2000, the system identified over 1,000 cases of notifiable conditions from the laboratories at four different laboratories. Our experience in developing the system has highlighted the need for improved compliance with HL7 result message formats by the laboratory information systems and more structured reporting of results for tests such as microbiology including consistent use of the abnormal flag.

Key words: *disease notification, electronic laboratory reporting, Indiana, population surveillance, public health*

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J Public Health Management Practice, 2001, 7(6), 60-66
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Introduction

Public health traditionally has relied on laboratory and hospital staff, physicians, and other relevant sources to take the initiative to provide data to health departments, where public health officials analyze and interpret the information as it comes in. Previous evaluations of notifiable condition surveillance relying on spontaneous reporting have found that sources often submit reports late and there is substantial underreporting.^{1,2} Most condition reports received by health departments originate from clinical laboratories.³⁻⁵ Active surveillance, in which public health officials contact sources and inquire about potential cases, produces more complete information than passive surveillance but it requires more time and money.

Background

One form of active surveillance is electronic laboratory-based reporting (ELR). Clinical laboratories using ELR send data indicating cases of notifiable

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Clement J. McDonald, MD, is Director and Distinguished Professor of Medicine, Regenstrief Institute for Health Care and Division of General Internal Medicine, Geriatrics, and International Medicine, Department of Medicine, Indiana University School of Medicine.

The authors performed this work at the Regenstrief Institute for Health Care and the National Library of Medicine supported us with contract number N01-LM-4-3510 and N01-LM-6-3546.

Electronic Laboratory Reporting: Barriers, Solutions and Findings

J. Marc Overhage, Jeffrey Suico, and Clement J. McDonald

4X greater detection rate
than MD-based reporting.

Same day versus 2-5 day
reporting.

Electronic laboratory reporting can...
...implemented an electronic
laboratory reporting system by
building of the infrastructure created
for the Indiana Network for Patient
Safety (INPS). The system has proven
reliable in delivering results and
scalable to multiple laboratories over
36 months of use. In April 2000, the
system identified over 1,000 cases of
notifiable conditions from the
laboratories at four different
laboratories. Our experience in
developing the system has highlighted
the need for improved compliance
with HL7 result message formats by
the laboratory information systems
and more structured reporting of
results for such as microbiology
including...
...norm...

Key words: disease notification, electronic laboratory reporting, Indiana, population surveillance, public health

Address correspondence to J. Marc Overhage, MD, PhD, Regenstrief Institute for Health Care, 5th Floor, Regenstrief Health Center, 1001 W. 10th St., Indianapolis, IN 46202; Telephone: 317-630-8883; fax: 317-630-8962; e-mail: jovernag@iupui.edu.

J Public Health Management Practice, 2001, 7(6), 60-66
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Introduction

Public health notification has relied on laboratory and hospital staff, physicians, and other relevant sources to identify and report cases of notifiable conditions, where public health officials analyze and interpret the information as it comes in. Previous evaluations of notifiable condition surveillance relying on spontaneous reporting have found that on-site submission of reports to public health departments and reporting to public health departments from clinical laboratories.¹⁻³ Active surveillance, in which public health officials contact sources and inquire about potential cases, produces more complete information than passive surveillance but it requires more time and money.

Background

One form of active surveillance is electronic laboratory-based reporting (ELR). Clinical laboratories using ELR send data indicating cases of notifiable

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The authors performed this work at the Regenstrief Institute for Health Care and the National Library of Medicine supported us with contract number N01-LM-4-3510 and N01-LM-6-3546.

Electronic Laboratory Reporting: Barriers, Solutions and Findings

J. Marc Overhage, Jeffrey Suico, and Clement J. McDonald

Electronic laboratory reporting can improve surveillance for notifiable conditions. Building on standards for message structure and content, we have implemented an electronic laboratory reporting system by building on the infrastructure created for the Indiana Network for Patient Care (INPC). The system has proven reliable in delivering results and scalable to multiple laboratories over 36 months of use. In April 2000, the system identified over 1,000 cases of notifiable conditions from the laboratories at four different laboratories. Our experience in developing the system has highlighted the need for improved compliance with HL7 result message formats by the laboratory information systems and more structured reporting of results for tests such as microbiology including consistent use of the abnormal flag.

Key words: *disease notification, electronic laboratory reporting, Indiana, population surveillance, public health*

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Introduction

Public health traditionally has relied on laboratory and hospital staff, physicians, and other relevant sources to take the initiative to provide data to health departments, where public health officials analyze and interpret the information as it comes in. Previous evaluations of notifiable condition surveillance relying on spontaneous reporting have found that sources often submit reports late and there is substantial underreporting.^{1,2} Most condition reports received by health departments originate from clinical laboratories.³⁻⁵ Active surveillance, in which public health officials contact sources and inquire about potential cases, produces more complete information than passive surveillance but it requires more time and money.

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Newborn Screening



U.S. National Library of Medicine

Newborn Screening Coding and Terminology Guide

Data Standards for Electronic Reporting

[Home](#) | [Views](#) | [Downloads](#) | [HL7](#) | [Resources](#) | [Code Standards](#) | [About](#) | [Updates](#) | [Contact Us](#)

The goal of the Newborn Screening Coding and Terminology Guide is to promote and facilitate the use of electronic health data standards in recording and transmitting newborn screening test results. The Web site includes standard codes and terminology for newborn tests and the conditions for which they screen, and links to other related sites. The codes and vocabulary standards are provided in a series of tables that you can view on the Web and/or download for your own use. These tables cover conditions recommended for screening by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) or by a state within the U.S.

Use of these standards can speed the delivery of newborn screening reports, facilitate the care and follow-up of infants with positive test results, enable the use (and comparison) of data from different laboratories, and support the development of strategies for improving the newborn screening process.

This Web site also includes [draft guidance for creating an HL7 version 2.x message using these codes](#) with examples. If you would like us to notify you about updates to this guidance and other new content, please subscribe to the [RSS feed for Updates](#), or join the [NBS-Announcements](#) e-mail list from the U.S. National Library of Medicine.

You can reach these various resources by picking a choice below.

Views: Generate customized Web views from the tables of conditions and analytes/measurements maintained by the U.S. National Library of Medicine (NLM®).

- **Conditions** — Conditions that are targeted by newborn screening
- **Analytes/Measurements** — Tests that are used as markers for newborn screening conditions
- **Tailored Views** — Specify subsets, or see relationships between conditions and analytes/measurements

Downloads: Download the tables of newborn screening conditions, of markers for these conditions and/or of mappings between conditions and their markers.

newbornscreeningcodes.nlm.nih.gov

PKU - Phenylketonuria - Condition Details

▸ [Overview](#) ▸ [Names and Codes](#) ▸ [Affected Protein Names and Codes](#) ▸ [Analytes or Measurements](#) ▸ [More Information](#)

Phenylketonuria is an inherited disorder that increases levels of the amino acid phenylalanine in the blood. Infants with classic PKU appear normal until they are a few months old. The signs and symptoms of PKU vary from mild to severe, including seizures, delayed development, behavioral problems, and psychiatric disorders. Permanent intellectual disability can be prevented with a special low-phenylalanine diet. Phenylketonuria is caused by mutations in the PAH gene; it has an autosomal recessive pattern of inheritance.

Names and Codes

Condition: ¹	Phenylketonuria
Abbreviation: ¹	PKU
SACHDNC Category: ²	Core
SNOMED CT Code: ³	7573000 — Classical phenylketonuria UMLS CUI: ⁴ C0751434
ICD-9-CM Code: ⁵	270.1 — Phenylketonuria [PKU]
ICD-10-CM Code: ⁶	E70.0 — Classical phenylketonuria

Affected Protein Names and Codes

Enzyme Commission Number: ⁷	1.14.16.1  — Phenylalanine 4-monooxygenase
UniProt Number: ⁸	P00439  — Phenylalanine-4-hydroxylase

Analytes or Measurements

These measurements are associated with the condition:

LOINC Long Common Name ⁹	Analyte Short Name ¹⁰	LOINC Number ¹¹	Units ¹²
Phenylalanine [Moles/volume] in Dried blood spot	PHE	29573-3	mmol/L
Phenylalanine/Tyrosine [Molar ratio] in Dried blood spot	PHE / TYR	35572-7	{ratio}

57131-5 Newborn conditions with positive markers [Identifier] in Dried blood spot

NAME

Fully-Specified Name:	Component	Property	Time	System	Scale	Method
	Newborn conditions with positive markers	Prid	Pt	Bld.dot	Nom	

TERM DEFINITION/DESCRIPTION(S)

This variable list the conditions that that the markers suggest may be present. It is a coded result intended for easy access by decision support systems to identify the cases that need special attention. The LOINC code will include an answer list that covers all of the conditions screened for by any state. States would only make statements about the conditions they screen for. (This item is still under discussion by the NBS community and subject to change)

Source: Regenstrief LOINC

BASIC ATTRIBUTES

Class/Type:	CHEM/Lab
Last Updated:	2011/04/05
Order vs. Obs.:	Observation
Status:	Active

NORMATIVE ANSWER LIST (LL835-0)

SEQ#	Answer	Global ID	Global ID Code System	Code	Answer ID
0	None				LA137-2
1	HEAR	15188001	SCT		LA12463-8
2	2M3HBA	791000124107	SCT		LA12464-6
3	2MBG	445596006	SCT		LA12465-3
4	3-MCC	13144005	SCT		LA12466-1
5	3-MCC (mat)	206001006	SCT		LA12467-9
6	3MGA	297235006	SCT		LA12468-7
7	5-OXO	39112005	SCT		LA12469-5
8	ARG	23501004	SCT		LA12470-3
9	ASA	41013004	SCT		LA12471-1
10	BIOPT-BS	237914002	SCT		LA12472-9
11	BIOPT-REG	58256000	SCT		LA12473-7
12	BKT	237953006	SCT		LA12474-5
13	CACT	238003000	SCT		LA12475-2
14	CBL A	73843004	SCT		LA12476-0
15	CBL B	82245003	SCT		LA12477-8
16	CBL C	74653006	SCT		LA12478-6
17	CBL D	31220004	SCT		LA12479-4
18	CBL E	360373000	SCT		LA12480-2
19	CBL G	237938003	SCT		LA12481-0

The Future



Great Scott!

Maps and Expressions

CD3+CD4+ (T4 helper) cells [# / volume] in Blood

24467-3:Cells.CD3+CD4+:NCnc:Pt:Bld:Qn:

LOINC

SNOMED CT

Cells.CD3+CD4+



T lymphocyte positive for both CD3 antigen and CD4

NCnc



Number concentration (qualifier value)

Pt



Single point in time (qualifier value)

Bld



Blood specimen (specimen)

Qn



Quantitative (qualifier value)

Expression Associations

SNOMED Observable Model for Qualities

Incoming LOINC Code 24467-3 means...

Property Type: 118550005

Inheres in: 119297000

Cells.CD3+CD4+:NCnc:Pt:Bld:Qn

Process duration: 123029007

Towards: 115396002

Scale: 30766002

Now, you can use the SNOMED CT hierarchy for queries, etc...

SNOMED CT Concept

Body structure

Anatomical or acquired body structure

Anatomical structure

Body system structure

Structure of hematological system

Blood cell

Leukocyte

Peripheral blood mononuclear cell

Lymphocyte

T lymphocyte

T lymphocyte positive for CD4 antigen

T lymphocyte positive for both CD3 antigen and CD4 antigen



Common Questions

When will all this be done?

Remember the request for patience?

Starting first with the most common lab tests (LOINC Top 2000) and components from Microbiology and Chemistry

Alpha (~ March 2014)

Preview release (~ July 2014)

Initial release (~Jan 2015)

Should I stop using LOINC?

Of course not!

IHTSDO endorses the use of LOINC Codes for representation of orders and observations in countries where LOINC has been adopted.

More reasons now than ever!

IHTSDO and RII both endorse the statement that, LOINC provides codes that represent the names of information items (e.g. questions) and SNOMED CT provides codes that may represent nominal and ordinal values (e.g. answers) for these named information items.

**Will IHTSDO keep making
SNOMED CT Observables?**

Yes. But like we said, LOINC has >70,000 of them and we're committed to minimizing duplication.

Yes. But like we said, LOINC has >70,000 of them and we're committed to minimizing duplication.

Within the scope of this Agreement, IHTSDO will not add new SNOMED CT Concepts that are subtypes of “Observable Entity” or “Evaluation Procedure” within the Cooperative Areas, except where one of the following conditions applies:

Condition 1

A specific requirement for such an addition has been formally submitted to the IHTSDO by two or more IHTSDO Members, and the additional concept has been modeled and reviewed in line with SNOMED CT editorial guidelines.

Requests for additions derived from lists of LOINC Terms, will not be accepted without special permission from RII.

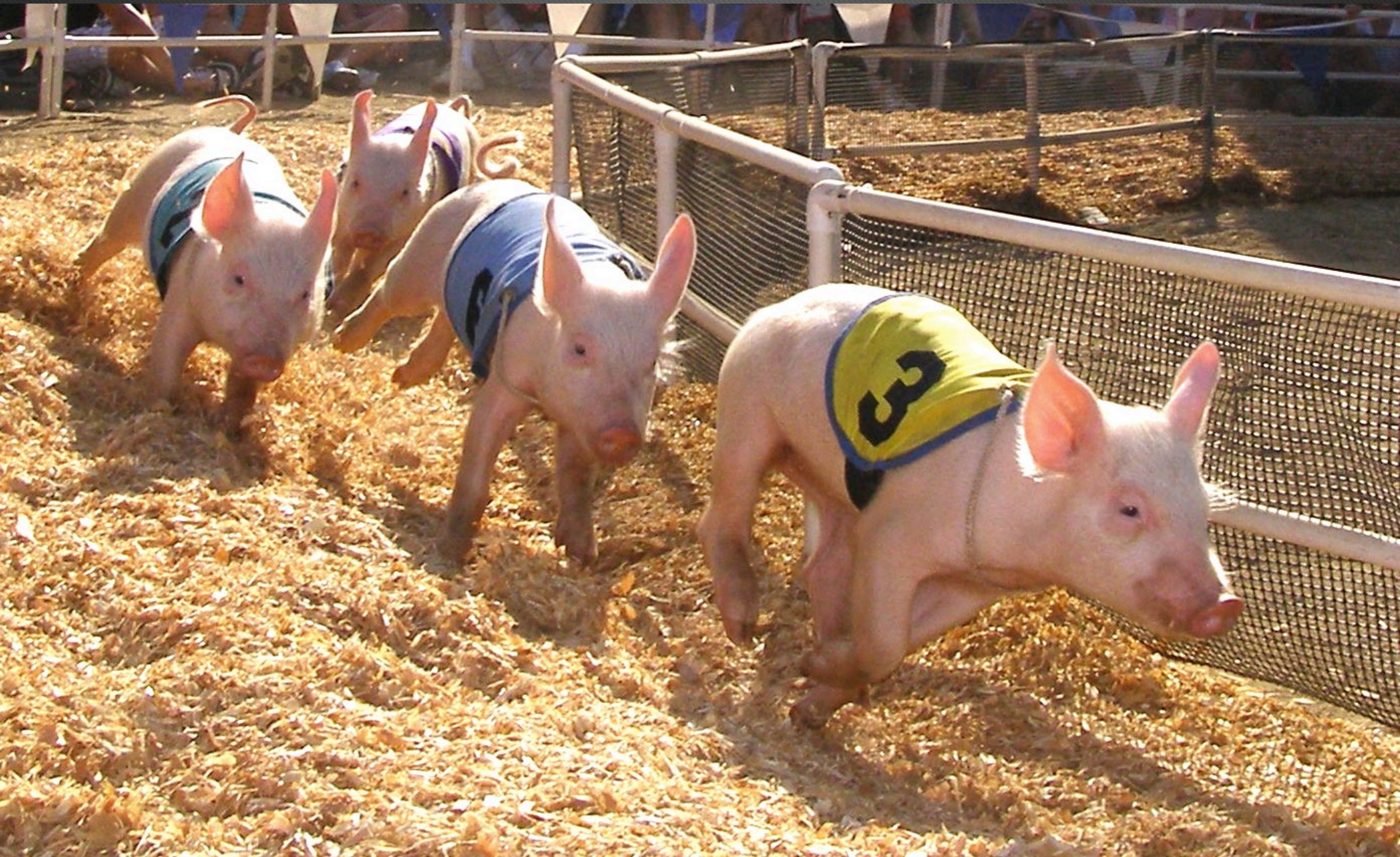
Or

Condition 2

A LOINC Term cannot be sufficiently specified by a post-coordinated expression based on the SNOMED CT Concept model, and the additional concept has been modeled and reviewed in line with SNOMED CT editorial guidelines.

IHTSDO will seek to modify the SNOMED Observable Model to accommodate Observable Entities that measure the sum of 2 or more analyses and other changes to minimize the need to invoke this clause.

The Race is On!



Happy LOINCing!

photo via [ryarwood](#)

