Semantic Web Representation of LOINC: an Ontological Perspective

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Abstract
The Logical Observation Identifiers Names and Codes terminology (LOINC) [1] has been proposed as a nomenclature for clinical laboratory tests. We present a formal representation of LOINC using a Semantic Web-based ontology that defines LOINC concepts in terms of the six main LOINC axes and their relationships with UMLS Semantic Types and the UMLS Metathesarus. This representation may enable automated information integration and decision support in public health surveillance.

Introduction
Each LOINC code corresponds to a single test or panel result and is uniquely identifiable by combining the following axes: 1) Component or analyte, (e.g. potassium); 2) Property measured (e.g. a mass concentration); 3) Timing (point in time, or integrated over duration of time); 4) Type of sample (system) (e.g. blood); 5) Type of scale (quantitative, ordinal, nominal or narrative); and 6) Method used to produce the result (where relevant). Our objective was to represent LOINC as a formal ontology using the Semantic Web framework to enable automated information integration, exchange, and processing.

Method
A LOINC concept can be identified in two ways: by a unique LOINC code, and by the unique composition of the six axes representing the meaning of the code in a clinical laboratory setting. The formal representation of the LOINC concept should enable its definition from both perspectives.
We first constructed a simple hierarchic (taxonomic) representation of LOINC codes as they appear in UMLS using Simple Knowledge Organization System (SKOS) [2]. In this representation a LOINC concept is defined as a SKOS:Concept having one and only one *LOINC Code*. LOINC codes are defined in the representation as a functional property that uniquely identifies a LOINC concept. To define a LOINC code based on its six axes, the axes first need to be defined and represented formally (Table 1).
All concepts in the first axis (component or analyte) were further decomposed into SKOS:broader and SKOS:narrower relationships to represent any meaningful superclass/subclass hierarchy between component classes, known to the UMLS-KS.

```
Class (LOINC:LNC_896-1 partial
            annotation(LOINC:Synonymn "Ab Scn SerPl Ql Cold Abs")
            annotation(LOINC:LOINCID "896-1")
            annotation(LOINC:CUI "C0368676")
            annotation(LOINC:Synonymn "ANTIBODY
SCREEN:ACNC…")
Class (LOINC:LNC_896-1 complete
  intersectionOf(
    restriction(LOINC:hasScale value (LOINC:CUI_C1442114))
    restriction(LOINC:hasProperty value (LOINC:CUI_C1264649))
    restriction(LOINC:hasAnalyte value (LOINC:CUI_C0368676))
    restriction(LOINC:hasSystem value (LOINC:CUI_C1442362))
    restriction(LOINC:hasTimeAspy value (LOINC:Point_in_Time))
    restriction(LOINC:hasMethod value (LOINC:CUI_C1264649))))
```

Table 1. LOINC Axiomatic Expressions

All other LOINC axes and their relevant concepts, except the time axis, were mapped to a unique UMLS concept and its corresponding semantic type. The time axis was modeled using the Time ontology introduced by Feng et al [3].
Each LOINC concept defined in the previous steps was then further defined as an equivalent concept to the intersection classes of the concepts representing their six LOINC axes (Table 1).

Results and Discussion
We have presented a formal ontology to represent clinical laboratory tests for automated classification of test results and decision support (e.g., automated notification and alert systems) using heterogeneous data sets. Our ontology is also mapped to the CDC Nationally Notifiable Diseases Surveillance System (NNDSS) [4] and creates an automated infrastructure to cross map between different coding schemas used in disparate IT implementations

References
1. McDonald C. LOINC, a universal Standard for Identifying Laboratory Observations. Clinical Chemistry 2001;49(4):624-633.
2. Simple Knowledge Organisation System (SKOS) Core Guide, 2nd W3C Public Working Draft 2 November 2005.
3. Feng Pan and J. R. Hobbs. Time in OWL-S. In 1st International Semantic Web Services Symposium, March 2004.
4. Nationally Notifiable Disease. [DB] 2002 http://www.cdc.gov/epo/dphsi/nndsshis.htm.