



SemanticMining

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Towards an ontology for laboratory medicine

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Summary

In reports for medical diagnosis and treatment it is necessary to ensure connectivity between laboratory production databases, medical health records and request and report systems. A prerequisite to enable this connectivity is the ability to represent examined properties in a systematic standardized way.

To this end a structure for reporting of results in clinical chemistry was given in detail by IFCC and IUPAC already in 1966. The recommendation has over the years been the basis for much of the work in representation of kinds of property in laboratory medicine. A more general concept of property (in relation to quantity) was later introduced and a thorough terminological analysis has been performed.

To create a coding scheme and a reference terminology for laboratory medicine which can be used as the basis for coding the representations for and kind-of-properties as part of a clinical laboratory messages for transmission between different locations and which contains sufficient information to allow the message to be translated from and to the required "local dialect" at each end must be seen as an important issue for the EU health care philosophy. The coding scheme should be based on a standardized representation structure and deliver as a CEN (or ISO) standard.

Using such a standard the risks of misunderstanding will be diminished when the "professional distance" between communicating parties increases, e.g. laboratory to health administrator rather than laboratory to clinician. Work internationally has not yielded a common strategy to overcome the problem of representing the same kind throughout the world although consensus regarding representation should be achievable. These facts underlay the work in WP25 and an ontologically correct system of representations has been the goal of the WP. With the publication of EN1614 this is achieved.



1 Overview

1.1 Objectives

| <i>Objectives</i> | <i>Progress towards achieving objectives</i> |
|--|--|
| To work out concept systems for representation of properties in laboratory medicine that can also be used in electronic health care records. | CEN EN 1614 accepted as a CEN standard. |

1.2 Milestones

| <i>Milestone</i> | <i>Planned date</i> | <i>Actual date</i> | <i>Comments</i> |
|---|---------------------|--------------------|--|
| Document on “dedicated-kind-of-property” published.(Forsum U, Karlsson D, Terminology, categories and representation of examinations in laboratory medicine. Clin. Chem. Lab. Med. 2005;43:344-345.) | | 2005 | Key ontology issues for Laboratory Medicine explored and given a theoretical sound bases. |
| CEN circulates the EN1614 standard, prepared as a main outcome of WP25, for final vote and the standard is accepted by the EU countries. | | 2006 | The standard puts reporting of properties examined in medical laboratories on a ontologically and metrologicalsound basis. |

1.3 Project meetings

| <i>Milestone</i> | <i>Planned date</i> | <i>Actual date</i> | <i>Comments</i> |
|--------------------------------|---------------------|-----------------------|--|
| Tihany Hungary | | 2004-06-27 – 07-03 | Key interactions with ontology experts within the NoE |
| Copenhagen Denmark 2005-02-02. | | 2005-02-02 | Meeting with René Dybkaer to discuss and work out ways |



| | | | |
|---------------------|--|-----------------------|--|
| | | | of representing the concept “dedicated kind-of-property”. |
| Tihany Hungary | | 2004-06-27 – 07-03 | Key interactions with ontology experts within the NoE |
| Saarbrücken Germany | | 2005-12-01 – 12-02 | Verification of key issues developed within the WP on the ontology of representing properties. |

1.4 Deviations from Plan

| <i>Causes and Description</i> | <i>Corrective actions</i> |
|-------------------------------|---------------------------|
| | None |



2 Main Results

See final standard text below.



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Foreword

This document (EN 1614:2006) has been prepared by Technical Committee CEN/TC 251 "Health informatics", the secretariat of which is held by NEN.

This European Standard shall be given the status of a national standard, either by publication of an identical text or by endorsement, at the latest by March 2007, and conflicting national standards shall be withdrawn at the latest by March 2007.

This document supersedes ENV 1614:1995.

The major technical changes are that issues relating to the distinction between kinds and instances of property have been resolved and that normative references to IUPAC-IFCC C-NPU have been removed.

According to the CEN/CENELEC Internal Regulations, the national standards organizations of the following countries are bound to implement this European Standard: Austria, Belgium, Cyprus, Czech Republic, Denmark, Estonia, Finland, France, Germany, Greece, Hungary, Iceland, Ireland, Italy, Latvia, Lithuania, Luxembourg, Malta, Netherlands, Norway, Poland, Portugal, Romania, Slovakia, Slovenia, Spain, Sweden, Switzerland and United Kingdom.



Introduction

This European Standard provides a model for the representation of dedicated kinds of property in laboratory medicine.

The need for this work stems from the increasing use of computerized clinical laboratory information systems, and the increasing need for reliable communication between laboratory information systems and between laboratory and other health care information systems (HCIS).

Potential users of this European Standard are:

- international and national organizations responsible for development, maintenance or registration of nomenclatures, classifications and coding systems;
- designers and developers of HCIS, e.g. laboratory information systems (LIS);
- persons responsible for acquisition of HCIS and checking compliance with standards;
- designers and developers of computerized diagnostic devices and data acquisition systems;
- developers of communication standards.

The degree to which a message (such as a clinical laboratory report) needs to be expressed in a formal, systematic language depends on the geographical, linguistic, social or professional distance between the communicating parties. The greater the distance, the greater the risk of misunderstanding.

Within any one clinical laboratory, local jargon terms may be used which are usually well understood between colleagues (Local Dialect A in Figure 1), but which would not be sufficiently widely known for communication with the outside world.

Likewise, a laboratory and its local community of users, such as hospital or community physicians, may use a "local dialect" of the language of clinical laboratories which is well understood by all concerned; but if communication possibilities are wider, even transnational, risks of serious misunderstanding arise.

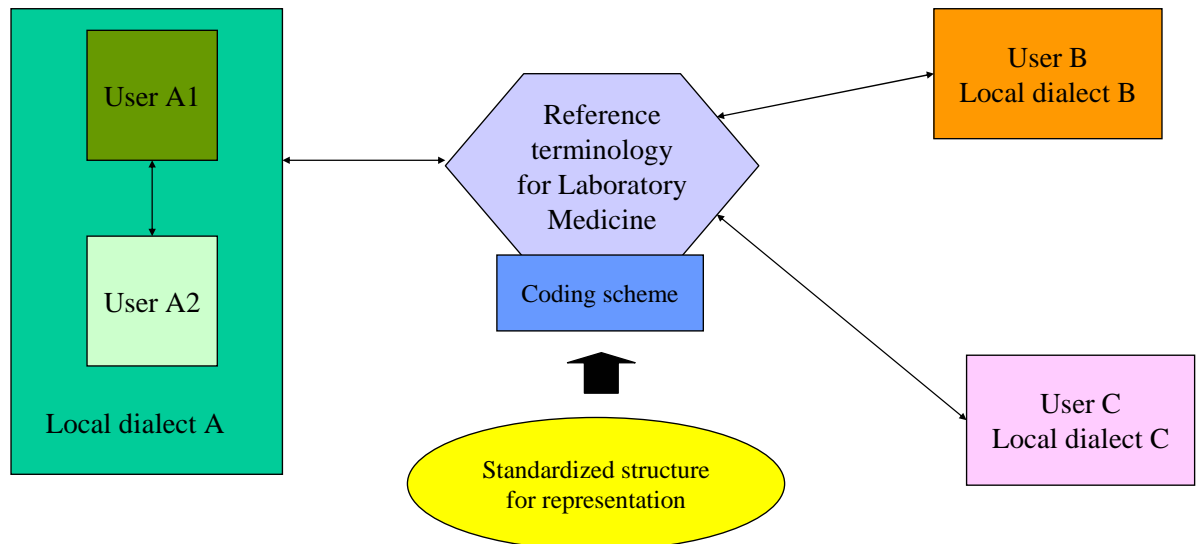


Figure 1 — Reference terminology as the bridge between local dialects

Risks of misunderstanding also increase when the "professional distance" between communicating parties increases, e.g. laboratory to health administrator rather than laboratory to clinician.

Two approaches to reducing this risk are:

1. To standardize the technical language used by clinical laboratory workers, users of the service, and other interested parties throughout the whole area in which communications may take place by eliminating all "local dialects".

This is obviously impracticable. Laboratory workers and clinicians would object to any such attempt from medical informatics. In the long run, agreement between professional bodies, with the cooperation of educational institutions, may lead to a greater degree of uniformity in the language of clinical laboratories, but this will not happen quickly and cannot be forced.

2. To create a coding scheme and a reference terminology for laboratory medicine which can be used as the basis for coding the dedicated kind-of-property part of a clinical laboratory messages for transmission between different locations and which contains sufficient information to allow the message to be translated from and to the required "local dialect" at each end. The coding scheme should be based on a standardized representation structure.

This is the more practical approach.

Scope

Purpose

This European Standard provides a structure aiding the representation, e.g. systematic terms or coding systems, of dedicated kinds of property, including dedicated kinds of



quantity, in laboratory medicine. The structure for representation is intended to facilitate the unambiguous communication of messages containing information about properties.

Field of application

This European Standard is applicable to all branches of laboratory medicine and other bodies offering laboratory analytic services. Examinations performed in the physician's office, at the bedside, or in the home are considered to be part of the laboratory medicine domain and thus this European Standard applies.

Uses

This structure for representation constitutes the essential basis for development of nomenclatures and coding systems intended for use in unambiguous and fully informative communication about properties, which fall within the field of application. Every such communication, including requests to and reports from clinical laboratories, and information retrieval for management reporting, research and reimbursement, will require additional information which is outside the scope of this European Standard.

Limitations

It should be emphasized that it is not the purpose of this European Standard to standardize the language used by health care practitioners in requesting or reporting clinical laboratory data. It may, however, be used as a guide by those who wish to adopt systematic terms for routine requesting and reporting of laboratory data. The syntax used for representing dedicated kinds-of-property is outside the scope of this European Standard, as are syntactic rules for the construction of codes in coding schemes.

The purpose is not to standardize the presentation of properties or kinds-of-property in user interfaces of computer systems nor the presentation in printed documents.

Normative references

Not applicable.

Terms and definitions

For the purposes of this document, the following terms and definitions apply.

3.1

system

part or phenomenon of the perceivable or conceivable world consisting of a demarcated arrangement of a set of elements and a set of relationships or processes between these elements [1]

EXAMPLE A given human being; a given portion of urine; the blood of a given person.

NOTE A system is, with the exception of the universe, a part of at least one more comprehensive super system and can itself contain one or several subsystems.



3.2

component

part of a **system** (3.1)

EXAMPLE Body of a given human being; glucose in a given portion of urine; the process of coagulation of the blood of a given person.

NOTE 1 Systems are open, i.e. transport occurs across their borders, both as input and output. Such transported entities may be conveniently regarded as components of the system.

NOTE 2 Components may be complex in that they may be aggregates of other components.

3.3

property

inherent state- or process-descriptive feature of a **system** (3.1) including any pertinent **component** (3.2) being determined

EXAMPLE Mass of the body of a given person at a given point in time; amount-of-substance concentration of glucose in a portion of urine at a given point in time.

3.4

quantity

attribute of a phenomenon, body or substance that may be distinguished qualitatively and determined quantitatively [VIM]

EXAMPLE Mass of a given object at a given point in time.

NOTE 1 **Quantity** is more specific in relation to **property** (3.3).

NOTE 2 The adjectives "measurable" and "physical" are used in VIM and in ISO 31, respectively, when required to point out that the word "quantity" is used in its metrological sense. In general, these adjectives can be omitted.

3.5

kind-of-property

common defining aspect of mutually comparable **properties** (3.3) [1]

EXAMPLE Colour; mass; amount-of-substance concentration.

NOTE The hyphens are used to clarify that the modifier "kind" should be seen as part of a connected whole.

3.6

dedicated kind-of-property

kind-of-property (3.5) with a given kind of **system** (3.1) and a given kind of **component** (3.2) subject for determination

EXAMPLE Mass of the body of a non-specified human being; amount-of-substance concentration of glucose in urine.

3.7

procedure

specified way to carry out an activity or a process [9]

3.8

measurement scale

ordered set of values of quantities of a given kind, continuous or discrete, used in arranging quantities of the same kind by magnitude [12]



3.9 unit

scalar quantity, defined and adopted by convention, with which other quantities of the same kind are compared in order to express their magnitudes [12]

Requirements

Representation of dedicated kind-of-property

The following elements stemming from the ontology of property shall be used for the representation of **dedicated kinds-of-property** (3.6) in laboratory medicine:

- kind of **system** (3.1);
- kind of **component** (3.2);
- kind-of-property** (3.5).

Representations of additional entities shall be appended to any or all of the elements when this is necessary to further specify the **dedicated kind-of-property** (3.6) to the degree required for a given purpose. While the three elements above represent aspects of the property examined, other aspects related to the examination of the property or the representation of the value of the property may need to be specified. Thus, a representation of a **dedicated kind-of-property** (3.6) may be further specified by:

- procedure** (3.7);
- measurement scale** (3.8);
- unit** (3.9).

EXAMPLE Specification to the system plasma, may be the super-system venous blood; to the component chromium, the specification Stock notation IV; to the kind-of-property mass concentration, the specification may be the measuring scale. Procedure specifications often apply to the whole dedicated kind-of-property.

NOTE Components may be complex, i.e. consist of aggregates of other components, e.g. in the case of sums or ratios.



Annex A (informative)

Representation of dedicated kinds-of-property

In reports for medical diagnosis and treatment it is necessary to ensure connectivity between laboratory production databases, medical health records and request and report systems. A prerequisite to enable this connectivity is the ability to represent examined properties in a systematic standardized way. This European Standard aims to provide such a systematic standardized way of representing the part of laboratory messages pertaining to the kind-of-property examined.

To illustrate how a property can be represented in a laboratory message, the following diagram is given. The example chosen is the amount-of-substance concentration of glucose in plasma from venous blood of a fasting patient examined in a sample from the patient at a given point in time. The laboratory message representing the examined property contains, among other things, the spatiotemporal specification, i.e. the subject of care and the time of examination, and the dedicated kind-of-property examined. In a practical context more complex message formats are of course needed. The message refers to a representation of a dedicated kind-of-property, usually using a code value obtained from a laboratory reference terminology, which in turn uses a representation structure according to this European Standard [2].

The representation of the dedicated kind-of-property consists of representations for the kind of system, the kind of component, and the kind-of-property. In the example, the kind of system plasma is specified by stating that the plasma is taken from venous blood from a fasting patient, where venous blood and fasting patient are specifications to the kind of system plasma. In terms of Aristotelian definition, the system is the genus and the specifications are the differentiae. The kind of component and the kind-of-property as well as the dedicated kind-of-property can be specified similarly.

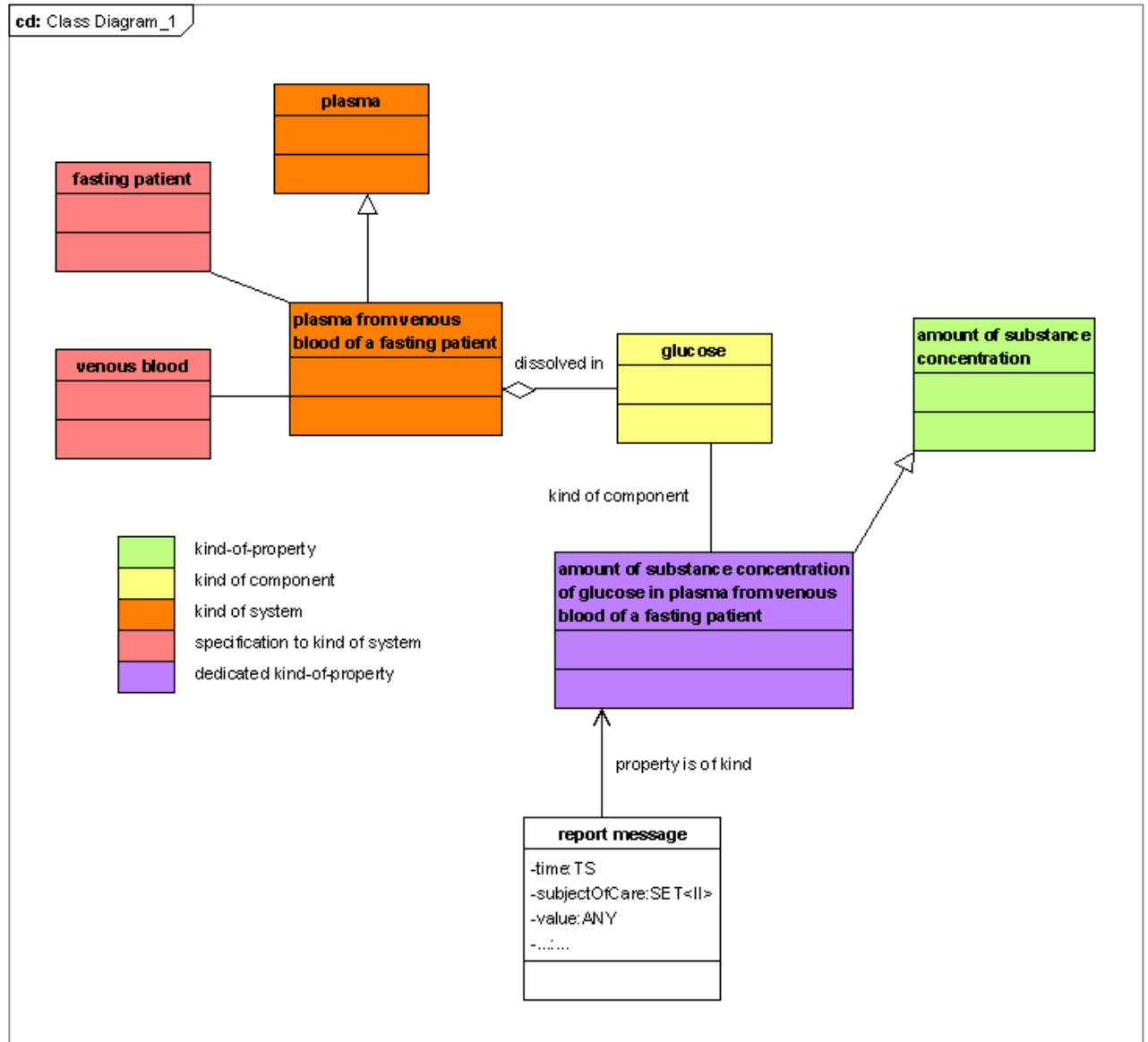


Figure A.1 — Example of a representation of a dedicated kind-of-property

Two important distinctions when reasoning about properties are the instance-kind distinction and the semiotic level distinction. Consider the following example: There are two properties examined, the amount of substance concentration of glucose in plasma from venous blood of two distinct fasting patients at two different points in time. These two properties are two instances of one kind. Considering that these two properties have been examined in the real world in some real patients, these two properties can exist on a conceptual level in the mind of observers of the examination

* "dissolved in" here means that instances and not kinds are dissolved.



and on a representational level, e.g. in report messages or in laboratory information system (LIS) databases. The scope of this European Standard is solely the representation of kinds, specifically of dedicated kinds-of-property. The standard does not aim to standardize representation of instances.



| Property | | |
|-----------------|--|---|
| | Instance | Kind (dedicated) |
| Representation | E.g. LIS database record or laboratory message | Systematic term for dedicated kind-of-property (e.g. according to C-NPU [2] or LOINC [3]) |
| Concepts | Individual concept | General concept |
| Reality | Object | Universal [4] |



Annex B (informative)

C-NPU

IFCC / Committee on Nomenclature, Properties and Units **IUPAC / Subcommittee on Nomenclature, Properties and Units** **Properties and Units in the Clinical Laboratory Sciences**

A structure for reporting of results in clinical chemistry was given in detail by IFCC and IUPAC in "Recommendation 1966" [5]. The recommendation has been the basis for much of the work in representation of kinds of property in laboratory medicine [6]. Over the years, the model of representation of "Recommendation 1966" has been developed to encompass all examinations in laboratory medicine. The more general concept of property (in relation to quantity) was introduced and a thorough terminological analysis has been performed.

The C-NPU syntax states the following requirements regarding the format of the systematic term for a dedicated kind-of-property:

System(specification)-Component(specification); Kind-of-property(specification).

The element "(specification)" shall be empty when no specification is required. The em dash following the name of the system and the semicolon following the name of the component are part of this syntax and these characters shall not be used within terms for kind of system, kind of component nor kind-of-property.

To use the C-NPU coding scheme for representation of properties, the examination shall be specified in space and time, i.e. a patient id and date and time shall be given.



Annex C (informative)

LOINC

LOINC Committee, The Regenstrief Institute, Indianapolis, Indiana, USA Logical Observation Identifier Names and Codes

The LOINC database provides a set of universal names and ID codes for identifying laboratory and clinical test results [7, 8]. Currently, many US laboratories are using ASTM 1238 or its sister standard HL7, (Health Level Seven, <http://www.hl7.org>), to send laboratory results electronically from producer laboratories to clinical care systems in hospitals. A HL7 message carries one record for each separate test observation. Within this record is one field that identifies the test, and another that reports its value. In HL7 nomenclature, the field that carries the observation identifier is called OBX-3, and the field that carries the observation value is called OBX-5. The LOINC database provides universal identifiers for observations in HL7 messages. The scope of the LOINC includes the codes that identify the test observation per se, not the codes that might be reported in the values of some test observations. If observations are seen as a question and the observation values as answers, LOINC provides codes for the questions. Other code systems provide codes for the answers. LOINC names are defined in terms of six major, and up to four minor, axes. The formal LOINC name shall include entries for the first six major axes. The method axis is included only when the method distinction makes an important difference to the clinical interpretation of the result.

Syntax of LOINC names:

<analyte/component>:<kind of property of observation or measurement>:<time aspect>:

<system (sample)>:<scale>:<method>

The minor axes include challenge information, adjustments, super-system and time operators. The challenge axis is the most complex and includes amount, route, and timing.

The LOINC code used to report a single observation can also be used to order that observation. The LOINC database includes codes for some test packages (panels) but only the most common and standardized ones. Each record carries the formal six-part LOINC name; the LOINC code, a number with a check digit, the observation class, related names, and other attributes. For most classes of laboratory observations, the database also includes a "short" report name that is <30 characters.



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²⁾ To be revised.