

Consolidated Health Informatics Initiative Final Recommendation Information Sheet¹

Domain Title and Team Lead:

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Scope:

Anatomy

Used to describe anatomical locations for the following purposes:

- Clinical
 - Site of a procedure such as:
 - Source of culture specimen
 - Surgical site
 - Location of blood pressure, temperature, other measurement
 - Etc.
 - Location of an observation such as:
 - Site of fracture
 - Site of injury
 - Etc.
- Surgical:
 - Precise anatomical structure involved in procedure
- Pathology:
 - Detailed gross description of item observed
 - Cellular description of item observed
- Research:
 - Uses many clinical terms
 - Subcellular components

and having the following requirements:

- Is-a hierarchy
- Part-of hierarchy
- Laterality
- Synonyms
- Virtual locators (Concepts added to the terminology that may not physically exist but are added for representational purposes. An example might be liver as a physical object that is the concept used when referring to the entire liver and liver structure when describing the relationships of the various parts

¹ Information Sheet designed specifically to facilitate communication between CHI and NCVHS Subcommittee on Standards and Security resulting from May 20, 2003 testimony. CHI may seek assistance to help further define scope, alternatives to be considered and/or issues to be included in evaluation process.

of the liver such as left lobe. SNOMED CT uses concepts similar to those just described.)

- Modifiers of basic terms such as “necrotic”
- Compatibility with animal models

Physiology

Used to describe or infer human physiology at least at the organ system, cellular, and biochemical levels. Physiology terminology includes tests that are used to infer the physiological state at any of the levels noted. Terminology that infers cellular physiology by direct inspection of cells is also included. The terminology must include concepts for both normal and abnormal physiology.

Domain/Sub-domain	In-Scope (Y/N)
Anatomical location of a procedure	Y
Anatomical location of an injury	Y
Anatomical description of specimen	Y
Subcellular anatomy	Y
Physiology of patient	N
Measured or inferred physiology of organ or organ system	Y
Measured or inferred physiology of cell	Y
Morphology	Y

Alternatives Identified

1. MESH (Medical Subject Headings)
2. National Cancer Institute (NCI) Anatomical Terminology
3. SNOMED CT
4. Clinical LOINC
5. Foundational Model of Anatomy (University of Washington)
6. HL 7 (Site Table)
7. Veteran’s Administration NDF-RT Physiology Effects Hierarchy

Final Recommendations:

Anatomy:

1. **SNOMED CT (Systematized Nomenclature of Medicine Clinical Terms)**

The specific locations in the SNOMED CT hierarchy that form the basis of our recommendation are:

Body structure:acquired body structure

Body structure:acquired body structure:post-surgical anatomy

Body structure:anatomical concepts:combined site

Body structure:anatomical concepts:physical anatomical entity:anatomical spatial entity

Body structure:anatomical concepts:physical anatomical entity:anatomical structure

Body structure:anatomical concepts:physical surface topography

Body structure:morphologically altered structure

For modifier terms not pre-coordinated above:

Qualifier Value:Additional Values

Qualifier Value:Modifier and/or Qualifier

2. HL7 Site table

The Workgroup determined that the HL7 Site table would provide for a more simple anatomy terminology for use in the general practice of healthcare. While a subset of SNOMED-CT would serve this purpose, the HL7 Site table is recommended to fill this role.

3. NCI Thesaurus

To support its research programs and international based clinical trials, the National Cancer Institute is revising the anatomy component of its widely use Thesaurus (<http://www.nci.nih.gov/cancerinfo/terminologyresources>). This work extends present anatomy terminologies into sub-cellular structures that are required for research and is primarily recommended for that purpose. Additionally, the remaining terminology appears well ordered and complete. The two terminologies can relate through mapping.

*Mapping is an essential requirement of the anatomy domain. It is the workgroup's recommendation that these mappings be developed, maintained, validated and distributed through the UMLS.

Physiology: No Recommendation

Cellular physiology is a basic medical concept that is not widely used at the clinical level and has diverse requirements at the research level. It is not surprising that a terminology was not found to meet this need. We note the potential need for terminology at this level to serve as a reference terminology that would link other terminologies that use physiology concepts. We recommend that the NLM investigate funding such a development, perhaps using the VA NDF-RT medication physiologic effect axis as a basis.

Clinical physiology, which we defined as the identification of tests and their results to infer the underlying cellular physiology, is an area that requires good terminology. We observed that both candidates, SNOMED-CT and Clinical

LOINC approached this area differently. We also felt that the approaches did not fully meet the needs of the area from a content or organization viewpoint.

Content Coverage:

The range of coverage for SNOMED-CT and corresponding UMLS Category 0 terms appears adequate for use now, containing approximately 3-5,000 concepts (synonyms can expand the number by a factor of 10), for expressing general descriptive clinical and anatomical concepts. No large gaps in coverage in this area were noted. It is noted; however, that the coverage is weak in the sub-cellular structures required for research, hence the augmentation with the NCI Thesaurus. The NCI Thesaurus covers vocabulary for clinical care, translational and basic research, and public information and administrative activities. The NCI Thesaurus provides definitions, synonyms, and other information on more than 7000 cancers and related diseases, 5500 single agents and combination therapies, and a wide range of other cancer-related topics. The HL7 site table, even in expanded form, is envisioned to contain approximately 100-200 terms.

Acquisition:

An in-principal agreement has been reached that provides, in the US, SNOMED CT as one of the Category 0 codesets, essentially allowing free distribution and use in the US.

Standards and associated terminology are available from HL7. HL7 asserts and retains copyright in all works contributed by members and non-members relating to all versions of the Health Level Seven standards and related materials unless other arrangements are specifically agreed upon in writing. No use restrictions are applied.

The NCI Thesaurus is covered by an open content license. The license allows free distribution and modification of the NCI Thesaurus content. Modification of NCI Thesaurus, including development of extensions, may be made using either Protégé available from Stanford University (<http://protege.stanford.edu/>) or DTS/TDE terminology development/distribution environment available from Apelon, Inc (www.apelon.com). Developers of extensions are encouraged to share their extensions.

Conditions:

Temporal Condition: The current HL7 site table is incomplete and requires addition of more general anatomy terms before it will be completely ready for use. HL7 has a mechanism to facilitate the addition of these terms through their Vocabulary Technical Committee. It is further recommended that the present and added terms to the site table be closely coordinated with the corresponding SNOMED-CT terms.

The NCI Thesaurus anatomy terminology is, at the time of this report, not released and is a work in progress. A development version was shared with the Workgroup for review. It is anticipated that the anatomy terminology will be included in the NCI Thesaurus by Fall, 2003. Hence this recommendation is conditional upon completion of the work. Review should be made in six to 12 months.