Ontology of Cancer Diagnosis in the OMOP Vocabulary

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INTRODUCTION:

- Observational research in cancer requires substantially more detail to represent conditions than most other therapeutic areas.
- Cancer attributes are covered in many terminology systems and data collection standards including ICD-O, SNOMED CT, LOINC, AJCC, NCIt, NAACCR, CAP
- For the first version of the OMOP CDM Oncology Module a new vocabulary "Cancer Modifiers" was developed based on the content of LOINC, NCIt, NAACCR, and CAP.
- In the new version of the OMOP Oncology Module, we addressed the problem of missing ontological relationships and mappings between the source and the Cancer Modifier vocabulary.

Methods

Each cancer modifier concept is a result of pre-coordination of two or more attributes. For example, *Prostate Cancer* by AJCC 7th edition Stage 4 is precoordinated from the dimensions of Stage, Staging System, and diagnostic Schema (Table 1). Concepts in the Cancer Modifier vocabulary are grouped into concept classes each covering certain diagnostic aspects (e.g. Staging/Grading, Metastasis). Each class of cancer modifiers has a set of defined dimensions. Cancer modifiers are recorded in the measurement table and can be explicitly connected with the cancer diagnosis through modifier_of_event_id modifier_of_field_concept_id represent a comprehensive patient's cancer diagnosis. 1

We extended the Cancer Modifier vocabulary with relationships to their dimensions. respective These dimensions serve as nodes in the concept hierarchy. A complete set of dimensions for each cancer modifier class is depicted in Table 1.

relationships Adding between base diagnosis and Staging/Grading modifiers via diagnostic schemas

modifier attributes Adding enable aggregation and hierarchical queries.

Building mappings from NAACCR and vocabularies to Cancer Modifier vocabulary

Note, this work is in progress, please join the Oncology Working group if you want to contribute





Table 1. Cancer modifier concepts and their attributes

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Concept Class	Attribute types	Example	Resulting Concept
Staging and Grading	Stage	Stage IV	Prostate Cancer by AJCC 7th edition Stage 4
	Staging system	AJCC v7	
	Schema	Prostate Cancer	
Metastasis	Fact of metastasis	Metastasis to	Metastasis to the Body of Stomach
	Metastatic site	Body of Stomach	
Nodes	Type of involvement	Macrometastasis	Paraaortic Lymph Nodes with Macrometastasis
	Group of lymph nodes involved	Paraaortic Lymph Nodes	
Dimension	Measurement type	Greatest Dimension	Greatest Dimension of viable tumor
	Measured entity	Viable tumor	
Margin	Margin type	Distal Margin	Distal Margin Involved by High-Grade squamous dysplasia
	Tumor growth	High-Grade squamous dysplasia	
Extension/Invasion	Fact of invasion of extension	Microscopic invasion	Microscopic Lymphovascular Invasion (LVI) into the Lymphatics
	Target structure of invasion	Lymphatics	
Histopattern	Histological finding	Nodular Growth Pattern	Nodular Growth Pattern
Topography	Tumor location	10 o'clock	10 o'clock

Results

- Added relationships between base diagnosis and Staging/Grading modifiers via diagnostic schemas enable post-coordination of base diagnosis with stage and grade when these links are not available in the source data. This enhancement is critical for precise definition of cancer diagnosis and supports high specificity in cohort and phenotype building.
- Added modifier attributes enable aggregation and hierarchical queries.
- Mappings from NAACCR and CAP vocabularies to Cancer Modifier vocabulary serve two purposes. One as a crosswalk from these representations to OMOP. Another as a deduplication of highly redundant concepts in NAACCR and CAP. These mappings enable vocabulary-driven data conversion to OMOP from the two major sources of US cancer data, tumor registries and synoptic pathology reports.

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