Identifying the Essence of Clinical Findings

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Goals and Methods

◆ Project Goal
  – Create an ontology for Clinical Findings
  – Populate with UMLS concepts

◆ Two Approaches
  – Top-down approach – principle driven
  – Bottom-up approach – data driven
Top-Down Approach

- Ontology Principles
  - Role vs. Type
- Formal Definition of Findings

Finding

created \[\rightsquigarrow\] Finding

used

what is it?
Ontology Principles

Type Taxonomy

- Type
  - is_a
  - Type
    - is_a
      - Type

Role Taxonomy

- Formal Role
  - is_a
  - Formal Role
  - is_a
    - Formal Role
Ontology Principles

Type Taxonomy

- Type
  - Type

Material Role

- Formal Role
  - Formal Role

Formal Role

- Material Role
  - Formal Role

- Material Role
Ontology Principles

Type Taxonomy

- Type
- Material Role
  - is_a

- Formal Role
  - is_a

Role Taxonomy

- Type
  - associated_with

Associative Relationship
Ontology Principles

Type Taxonomy

Person

Student

is_a

Teacher

Role Taxonomy

Work without pay

associated_with

Associate Relationship
Ontology Principles

Type Taxonomy

Person
  is_a
  Student
  is_a
  Work without pay
  is_a
  Teacher

Adol
Jon
Matt
Mia

Role Taxonomy

NLM staff

Associative Relationship
Formal Definition of Findings

Finding

created → Finding → used

what is it?
Formal Definition of Findings

Finding is the result of a Healthcare Activity

Activity \[\xrightarrow{\text{result\_of}}\] Finding

- History Taking
- Physical Exam
- Laboratory Procedures
- Other Test Procedures

- Symptom
- Sign
- Laboratory Results
- Test Results
Formal Definition of Findings

- **Finding** is the result of a Healthcare Activity

Diagram:

```
Activity
  ▼
  Palpation of Liver
  CT scan of Abdomen
  ▼
  Finding
    Enlarged Liver
```
Formal Definition of Findings

◆ Finding is a measurement of an Attribute

Activity

Finding

measurement_of

Attribute

Liver Size

Enlarged Liver
Formal Definition of Findings

- Finding is the result of an Activity that measures an Attribute.
Formal Definition of Findings

- **Finding** is a type of **Evidence of the manifestation of Disease**
Formal Definition of Findings

Finding

what is it?

created

used
Data, Information, Knowledge
Data, Information, Knowledge

Example - ECG
Formal Definition of Findings

Finding is Information used as Evidence of the manifestation of Disease

- Information
- Evidence
- Guilt or Innocence
- Legal Finding
- Clinical Finding
- Disease
Ontology for Clinical Findings

Activity

Attribute

Information

Evidence

Finding

Disease
Populating the Ontology

Activity

Attribute

Information

Finding

Evidence

Disease
Populating the Ontology

Bottom-Up Approach

Data Set

UMLS Metathesaurus Concepts

Rules for extracting Findings, Activities, Attributes

Modified version of NLP program FindX
UMLS Metathesaurus Concepts

776940

Finding
56214

Laboratory or Test Result
4582

Sign or Symptom
11348

Total Findings Concepts
69144

Total Terms
94182
UMLS Metathesaurus Concepts

Findings
94182

Training
Gold
Standard
1000

Testing
Gold
Standard
1000
Populating the Ontology

 participates in

 – Rule Structure
   – Attribute – semantic grouping
   – Value
     – numeric value
     – SNOWMED adjective
Populating the Ontology

◆ FindX
  – Four Rules
    – Anatomy Rule
    – Physiologic Function Rule
    – Test Result Rule
    – Finding Rule
Populating the Ontology

- FindX Mechanics
  - Syntactic Analysis
  - Lexical Look-up
  - Xerox Tagger
  - Parser
  - MetaMap – Semantic Types
  - FindX
    - Semantic Grouping
    - Identify Values
    - Apply Rules
<table>
<thead>
<tr>
<th>Potassium</th>
<th>Level</th>
<th>Increased</th>
</tr>
</thead>
</table>

**Input**
Lexical Look-up

Potassium (noun)
Level (noun
verb
adj
adv)
Increased (adj
verb)

SPECIALIST Lexicon – Identify Parts of Speech
Potassium
n
Level
n
Increased
adj

Resolve Parts of Speech
Identify Noun Phrases
Level Increased Potassium

- mod
- head
- mod

Potassium
- noun
- noun
- adj

Potassium Measurement
- Potassium
- Level
- Increased

Map to Metathesaurus – Prefer Multiple Concepts
Semantic Grouping
FindX

Noun Phrase

mod
Potassium
noun

head
Level
noun

mod
Increased
adj

Potassium Measurement
Laboratory Procedure
testres

SNOWMED adjectives

value

Identify Values
FindX

Noun Phrase

mod
Potassium
noun

head
Level
noun

mod
Increased
adj

SNOWMED
adjectives

value

This is a Finding

Apply Rules

Potassium Measurement

Laboratory Procedure
testres
Populating the Ontology

Activity
  - Potassium Measurement

Information

Attribute

Evidence
  - Disease
    - Potassium Level Increased

Finding
  - ?
Populating the Ontology

- Activity
  - Potassium Measurement

- Attribute
  - Potassium

- Information

- Evidence
  - Disease
    - Potassium Level Increased
New FindX Rules

- Biological Substance Rule
- Pathogen Rule
- Therapy Rule
- Abnormality Rule
- Modified Abnormality Rule
- Regional Abnormality Rule
Performance

- Recall and Precision
Special FindX Modifications

- Clean-up ambiguity designators
  - Arterial Pressure <2>

- Inflectional variation of SNOMED adjectives
  - Faeces Discoloured