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

what is it?

created

used
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

Role Taxonomy

- Formal Role
  - Material Role

is_a relationships:
- Type is_a Type
- Type is_a Material Role
- Material Role is_a Formal Role
- Formal Role is_a Formal Role
Ontology Principles

Type Taxonomy

Role Taxonomy

Type

Formal Role

Material Role

is_a

is_a

associated_with

Associative Relationship
Ontology Principles

Type Taxonomy

Person

is_a

Student

Work without pay

is_a

Teacher

Role Taxonomy

associated_with

Associative Relationship
Ontology Principles

Type Taxonomy

Person

is_a

Student

is_a

Work without pay

is_a

Teacher

associated_with

Adol

Jon

Matt

Mia

NLM staff

Role Taxonomy

Associative Relationship
Formal Definition of Findings

Finding

created

used

what is it?
Formal Definition of Findings

Finding is the result of a Healthcare Activity

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

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

Finding is the result of a Healthcare Activity

- Activity
  - Palpation of Liver
  - CT scan of Abdomen

Finding
  - Enlarged Liver
Formal Definition of Findings

- **Finding** is a measurement of an **Attribute**

```
Finding  measurement_of  Attribute
       |               |          |
Activity | Enlarged Liver | Liver Size |
       |          |            |
Attribute

```
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**

**Diagram**

```
Evidence

is_a

Finding

manifestation_of

Disease
```
Formal Definition of Findings

What is it?
Data, Information, Knowledge

Diagram illustrating the relationship between data, information, and knowledge.
Data, Information, Knowledge

Example - ECG
Formal Definition of Findings

- Finding is **Information** used as **Evidence** of the manifestation of **Disease**

Diagram:

- Information
  - Guilt or Innocence
  - Legal Finding
- Evidence
  - Clinical Finding
  - Disease
Populating the Ontology

Activity
- Ac₁
- Ac₂

Information

Attribute
- At₁
- At₂

Finding
- F₁
- F₂

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

Findings
94182

Training Gold Standard
1000

Testing Gold Standard
1000
Populating the Ontology

◆ FindX
  – 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>Input</th>
</tr>
</thead>
<tbody>
<tr>
<td>Potassium</td>
</tr>
</tbody>
</table>
Lexical Look-up

SPECIALIST Lexicon – Identify Parts of Speech

Potassium: noun

Level: noun

verb

adj

adv

Increased: adj

verb
Resolve Parts of Speech
Identify Noun Phrases
Level Increased Potassium

Map to Metathesaurus – Prefer Multiple Concepts
Potassium Level Increased

- Noun Phrase
  - mod
    - Potassium
      - noun
        - Potassium Measurement
          - Laboratory Procedure
          - Element, Ion, or Isotope
  - head
    - Level
      - noun
        - Spatial Concept
          - Biologically Active Substance
  - mod
    - Increased
      - adj
        - Functional Concept
Level Increased Potassium
Potassium Level Increased

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

Potassium Measurement

Laboratory Procedure
testres

SNOWMED adjectives

value

This is a Finding

Apply Rules
Populating the Ontology

Activity
- Potassium Measurement

Information
- Finding
  - Potassium Level Increased

Evidence
- Disease

Attribute
- ?
FindX

Create New Rules
Populating the Ontology

Activity

Attribute
- Potassium Measurement

Information
- Finding
- Potassium Level Increased

Evidence
- Disease
- Potassium
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 SNOWMED adjectives
  - Faeces Discoloured