The Unified Medical Language System

*What is it and how to use it?*

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Part I
What is the UMLS?

(2) Overview through an example
Addison’s disease

- Addison's disease is a rare endocrine disorder
- Addison's disease occurs when the adrenal glands do not produce enough of the hormone cortisol
- For this reason, the disease is sometimes called chronic adrenal insufficiency, or hypocortisolism
Adrenal insufficiency  Clinical variants

- **Primary / Secondary**
  - Primary: lesion of the adrenal glands themselves
  - Secondary: inadequate secretion of ACTH by the pituitary gland

- **Acute / Chronic**

- **Isolated / Polyendocrine deficiency syndrome**
Addison’s disease: Symptoms

- Fatigue
- Weakness
- Low blood pressure
- Pigmentation of the skin (exposed and non-exposed parts of the body)
- …
AD in medical vocabularies

- **Synonyms: different terms**
  - Addisonian syndrome
  - Bronzed disease
  - Addison melanoderma
  - Asthenia pigmentosa
  - Primary adrenal deficiency
  - Primary adrenal insufficiency
  - Primary adrenocortical insufficiency
  - Chronic adrenocortical insufficiency
- **Contexts: different hierarchies**
  - eponym
  - symptoms
  - clinical
  - variants
Organize terms

- Synonymous terms clustered into a concept
- Preferred term
- Unique identifier (CUI)

<table>
<thead>
<tr>
<th>Term</th>
<th>Source</th>
<th>Identifier</th>
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<tbody>
<tr>
<td>Adrenal gland diseases</td>
<td>MeSH</td>
<td>D000307</td>
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<tr>
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<td>Disorder of adrenal gland</td>
<td>Read</td>
<td>C15z.</td>
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<tr>
<td>Diseases of the adrenal glands</td>
<td>SNOMED</td>
<td>DB-70000</td>
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Adrenal Gland Diseases
Diseases of the endocrine system

Diseases of the Adrenal Glands

Addison’s Disease
Endocrine Diseases

Adrenal Gland Diseases

Adrenal Gland Hypofunction

Addison’s Disease
Endocrine disorder

Disorder of adrenal gland

Hypoadrenalism

Adrenal Hypofunction

Corticoadrenal insufficiency

Addison’s Disease
Primary adrenocortical insufficiency

Other disorders of adrenal gland

Disorders of other endocrine gland

ICD-10
Organize concepts

- Inter-concept relationships: hierarchies from the source vocabularies
- Redundancy: multiple paths
- One graph instead of multiple trees (multiple inheritance)
Adrenal Cortex Diseases

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Addison’s Disease

Endocrine Diseases

Adrenal Gland Diseases

Adrenal Cortex Diseases

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Addison’s Disease

organize concepts

SNOMED
MeSH
AOD
Read Codes

UMLS
Relate to other concepts

- Additional hierarchical relationships
  - link to other trees
  - make relationships explicit
- Non-hierarchical relationships
- Co-occurring concepts
- Mapping relationships
Endocrine Diseases

Adrenal Gland Diseases

Adrenal Cortex Diseases

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Secondary hypocortisolism

Addison’s Disease

Addison’s disease due to autoimmunity

Other disorders of adrenal gland

Disorders of other endocrine gland

Endocrine Diseases

Abdominal organ

Diseases

Adrenal Dysfunction

relate to other concepts
Categorize concepts

- High-level categories (semantic types)
- Assigned by the Metathesaurus editors
- Independently of the hierarchies in which these concepts are located

Diagram:
- Disease or Syndrome
  - Diseases
    - Endocrine Diseases
      - Adrenal Gland Diseases
        - Adrenal Gland Hypofunction
          - Addison’s Disease