Ontological Spring
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What needs to be represented in a biomedical ontology?

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Outline

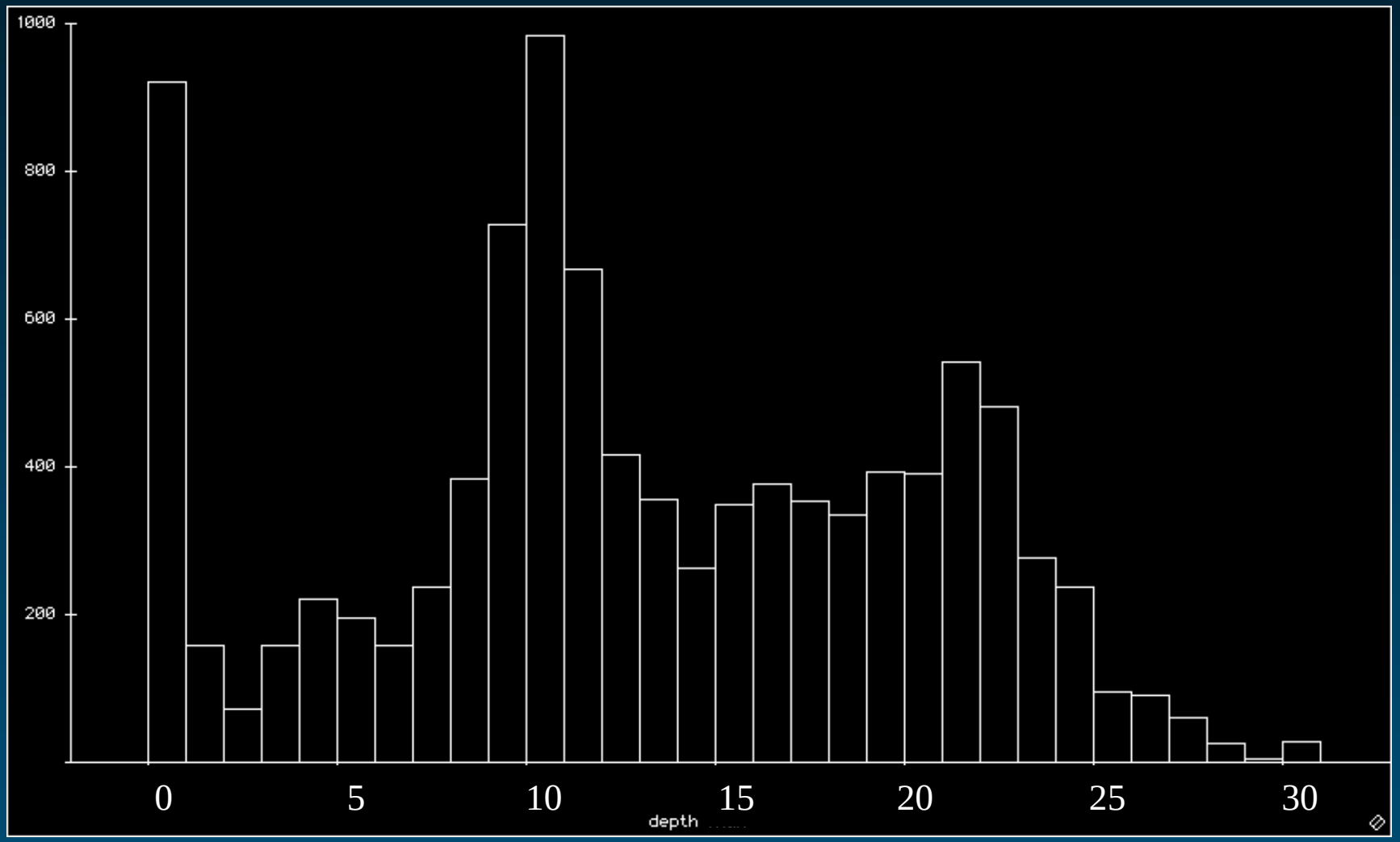
- Granularity issues
- Types vs. instances
- Ontology vs. information model

Granularity issues

Granularity

- No theoretical limit
- No ideal granularity
- Arbitrary limitation
  - Number of digits in code
  - By design (purpose driven)
- Increased
  - When several perspectives are combined (UMLS / individual vocabularies)
  - When “hierarchy” is loosely defined

Granularity in UMLS

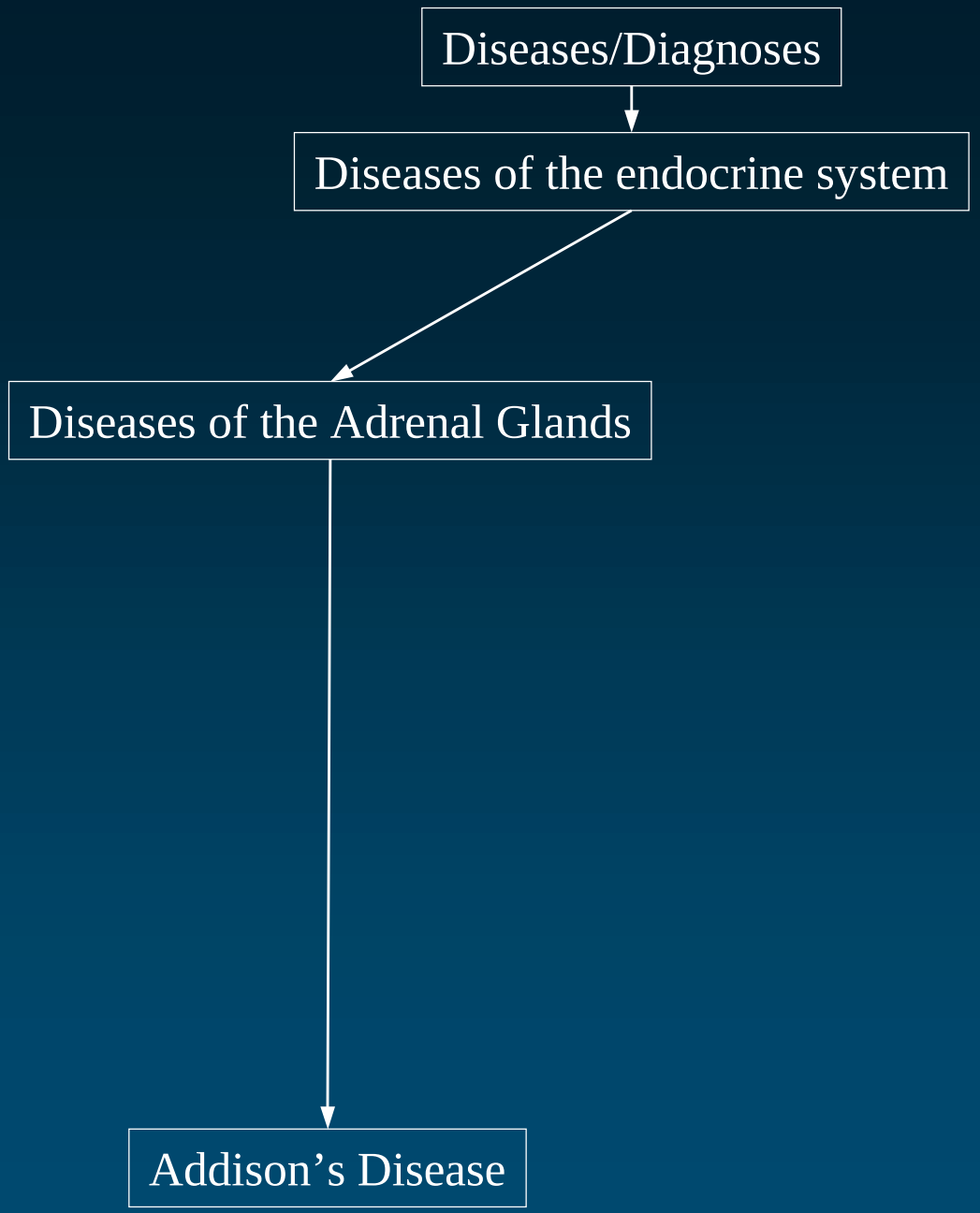


Distribution of depth max for 10,000 randomly selected UMLS concepts (2001)

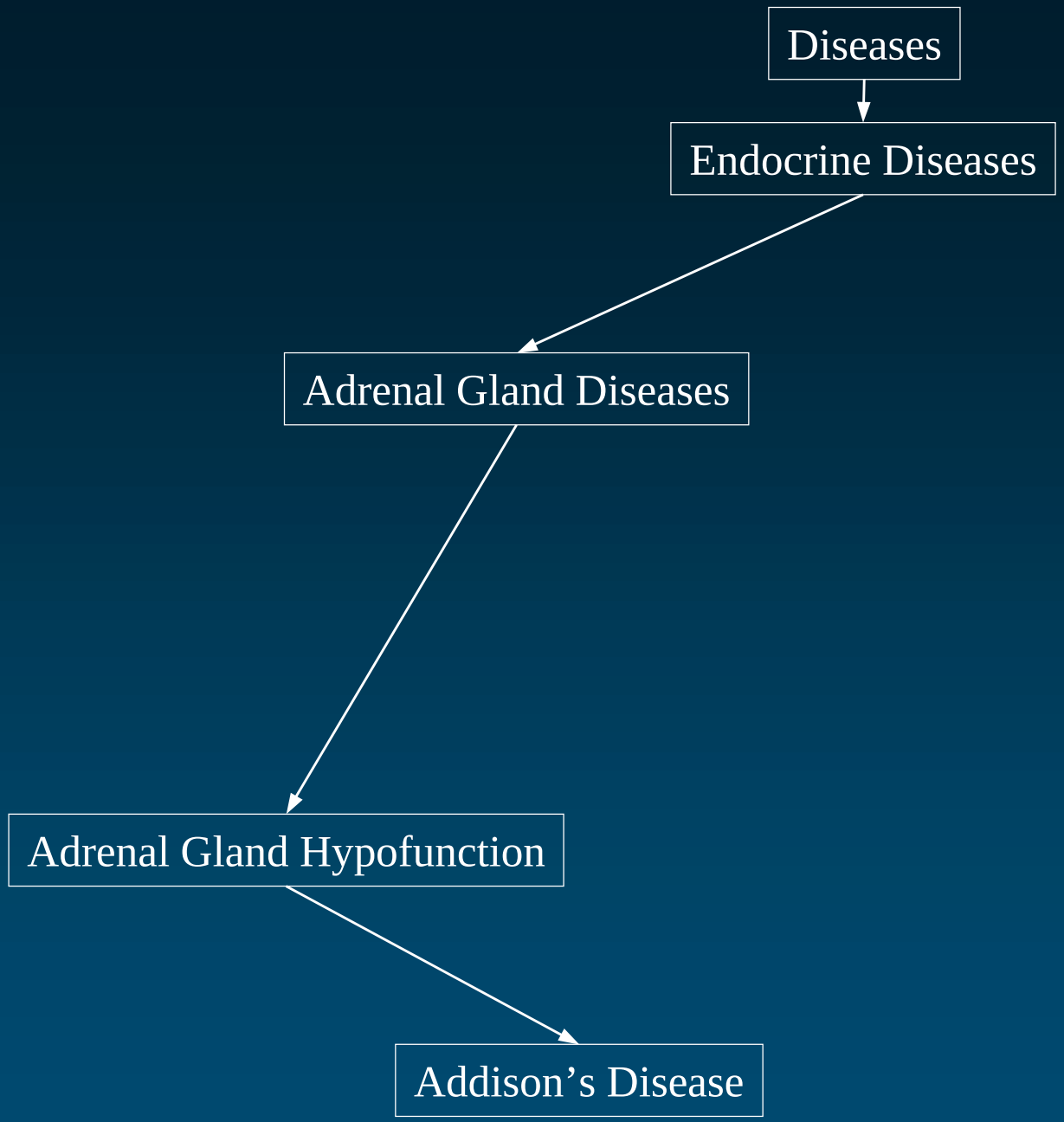
Example Addison's disease

- Representation in several medical vocabularies
  - SNOMED International
  - Medical Subject Headings (MeSH)
  - Alcohol and Other Drugs Thesaurus
  - Read Codes (CTV3)
  - International Classification of Diseases
- Combined representation in the UMLS Metathesaurus

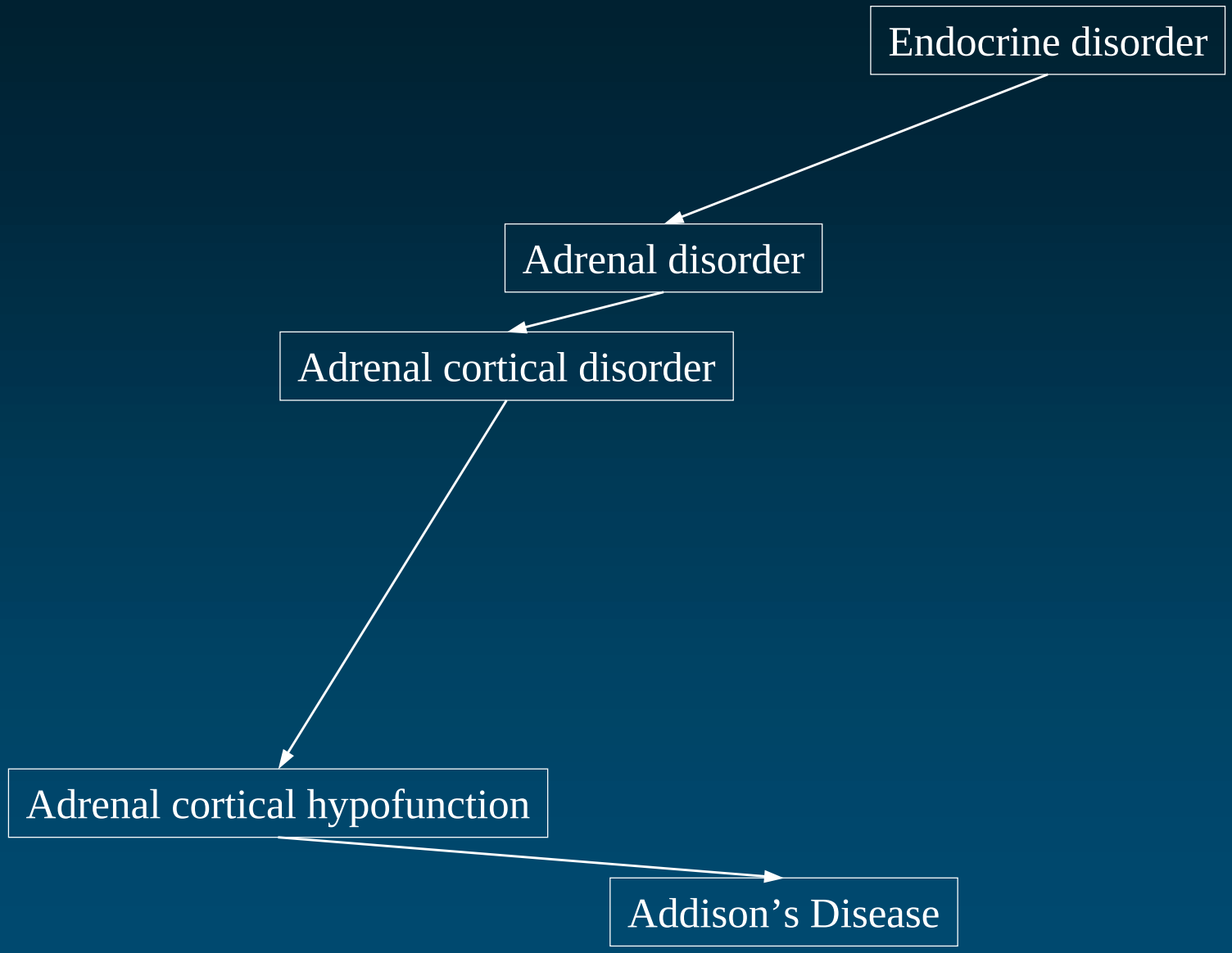
SNOMED International



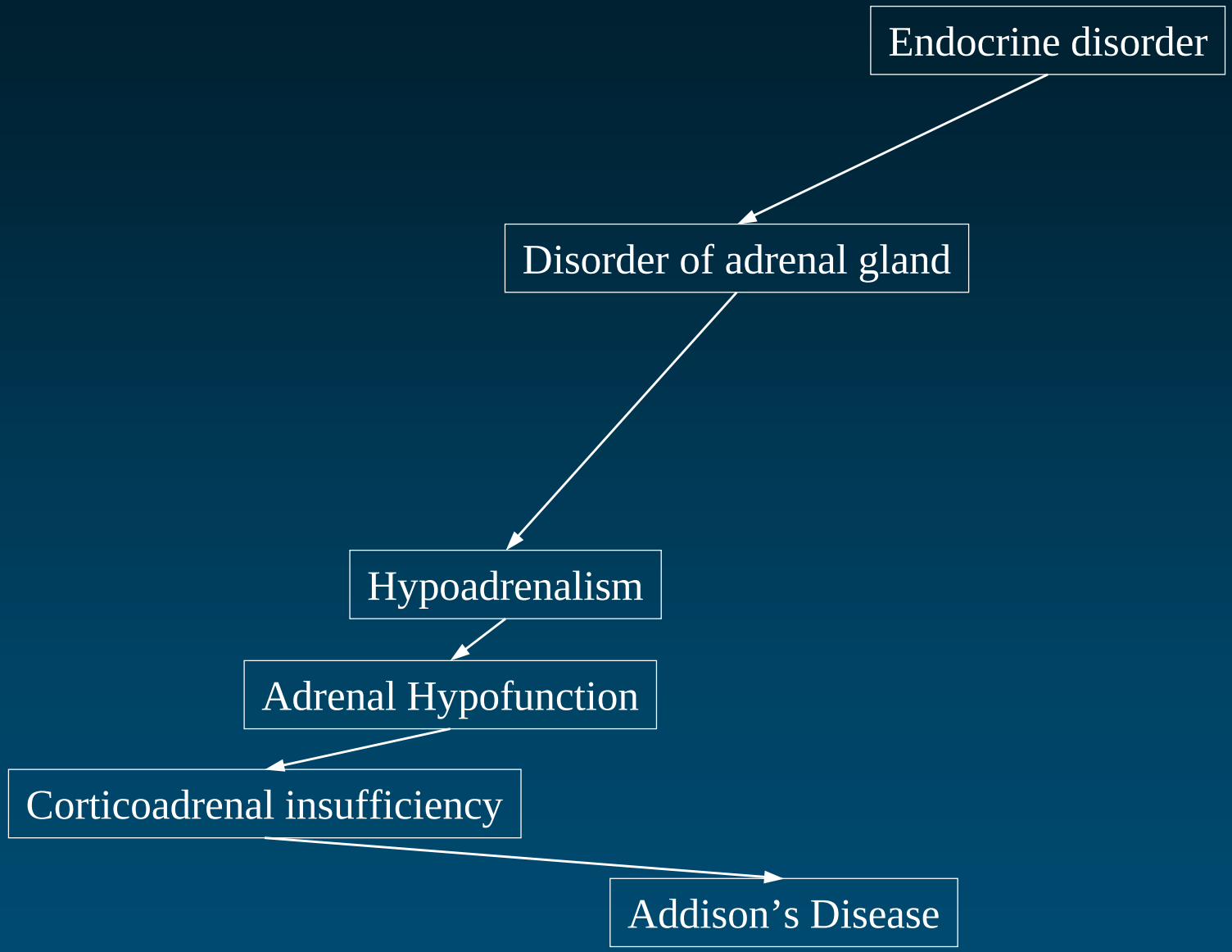
MeSH


Diseases
Endocrine Diseases
Adrenal Gland Diseases
Adrenal Gland Hypofunction
Addison’s Disease

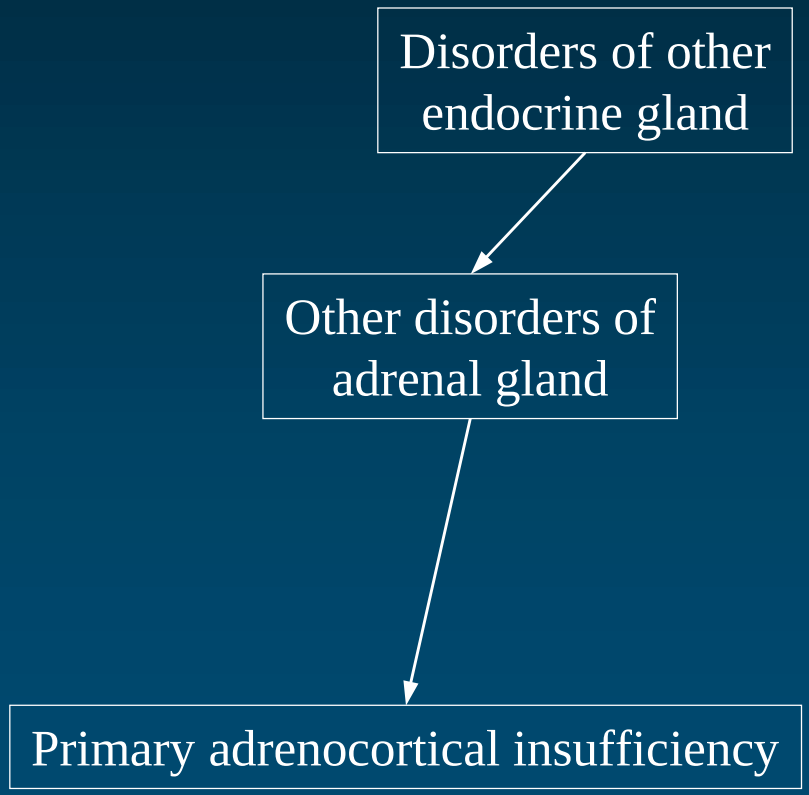
AOD


Endocrine disorder
Adrenal disorder
Adrenal cortical disorder
Adrenal cortical hypofunction
Addison’s Disease

Read Codes


Endocrine disorder
Disorder of adrenal gland
Hypoadrenalism
Adrenal Hypofunction
Corticoadrenal insufficiency
Addison’s Disease

ICD-10


Disorders of other endocrine gland
Other disorders of adrenal gland
Primary adrenocortical insufficiency

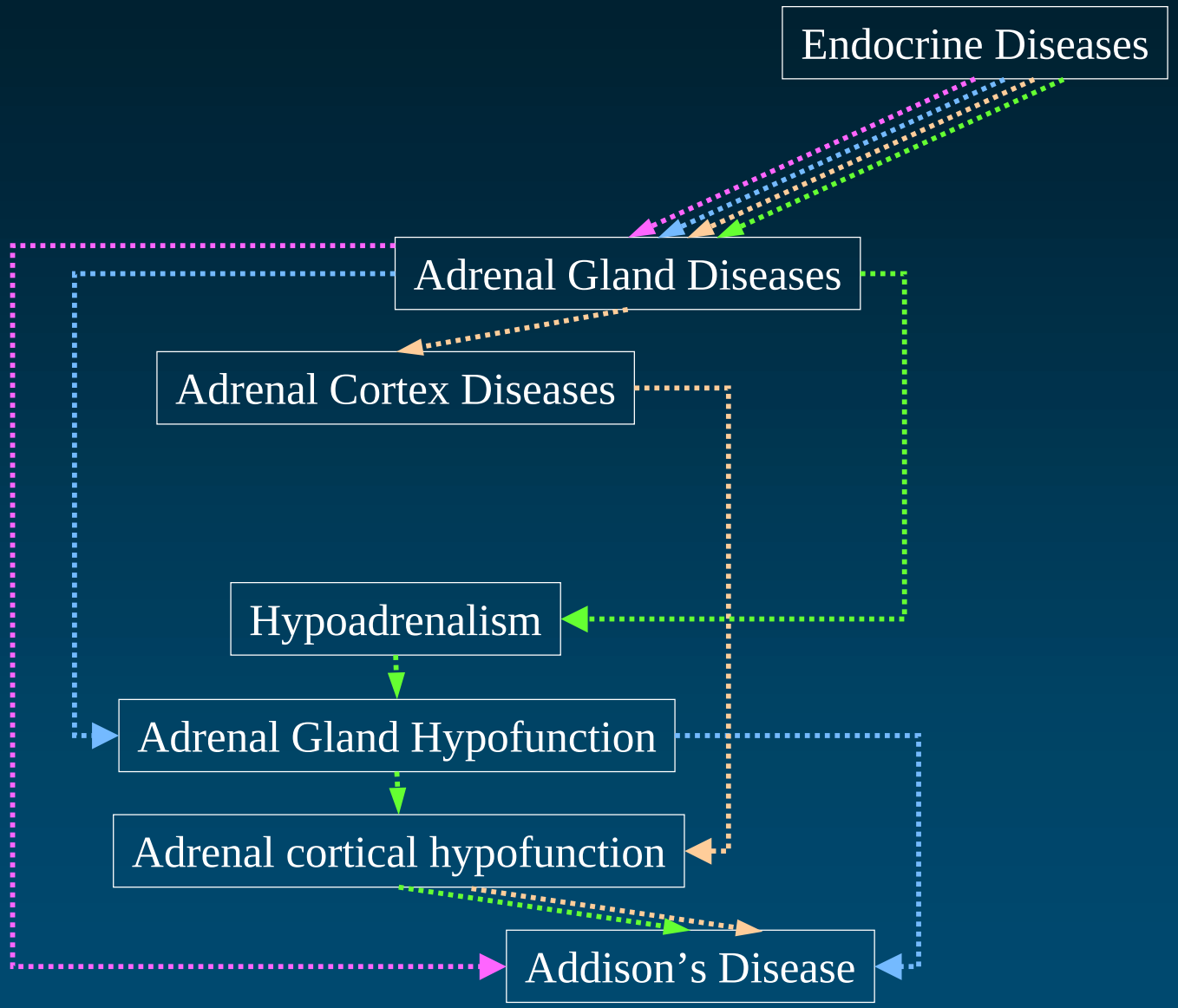
organize concepts

SNOMED

MeSH

AOD

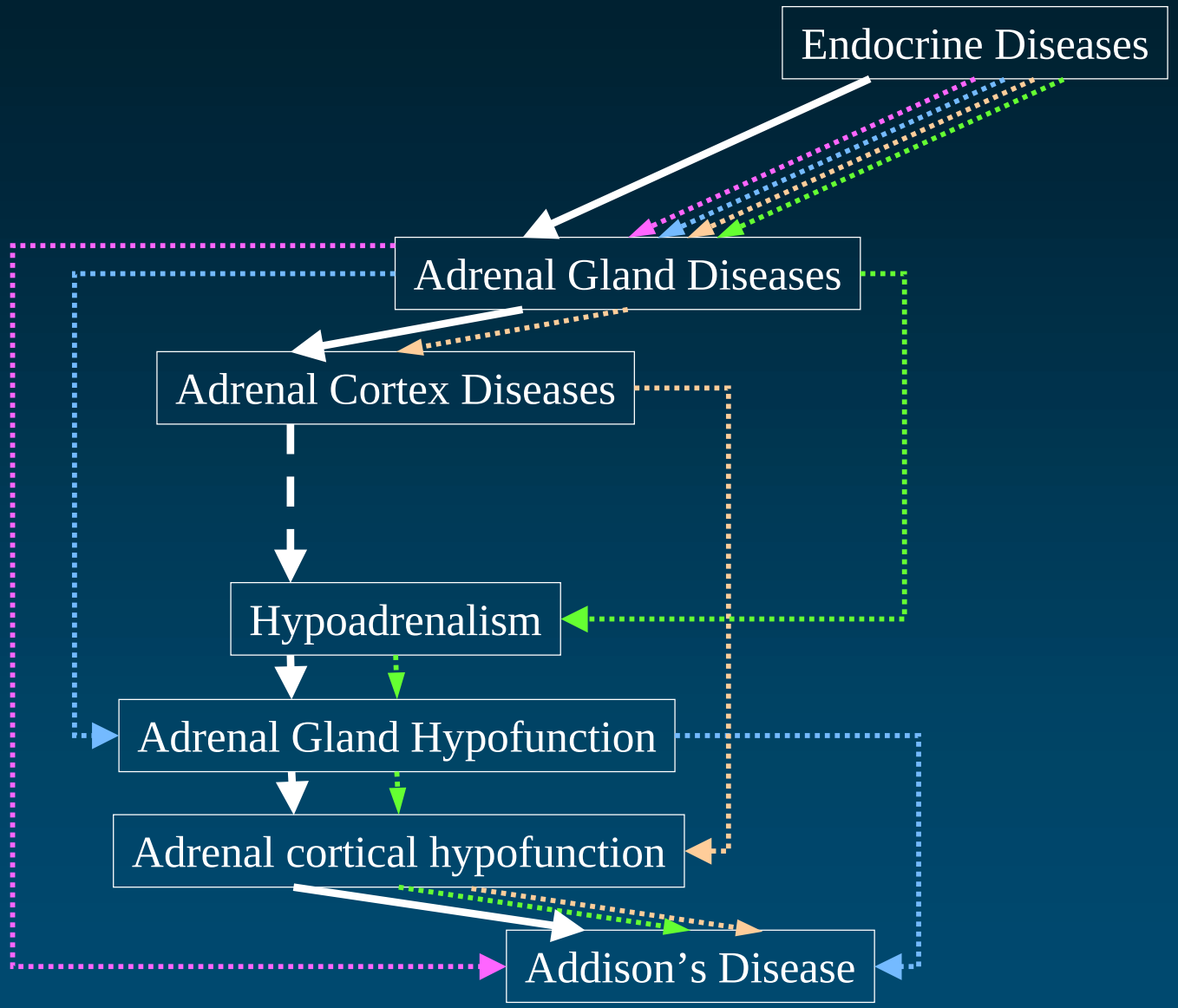
Read Codes

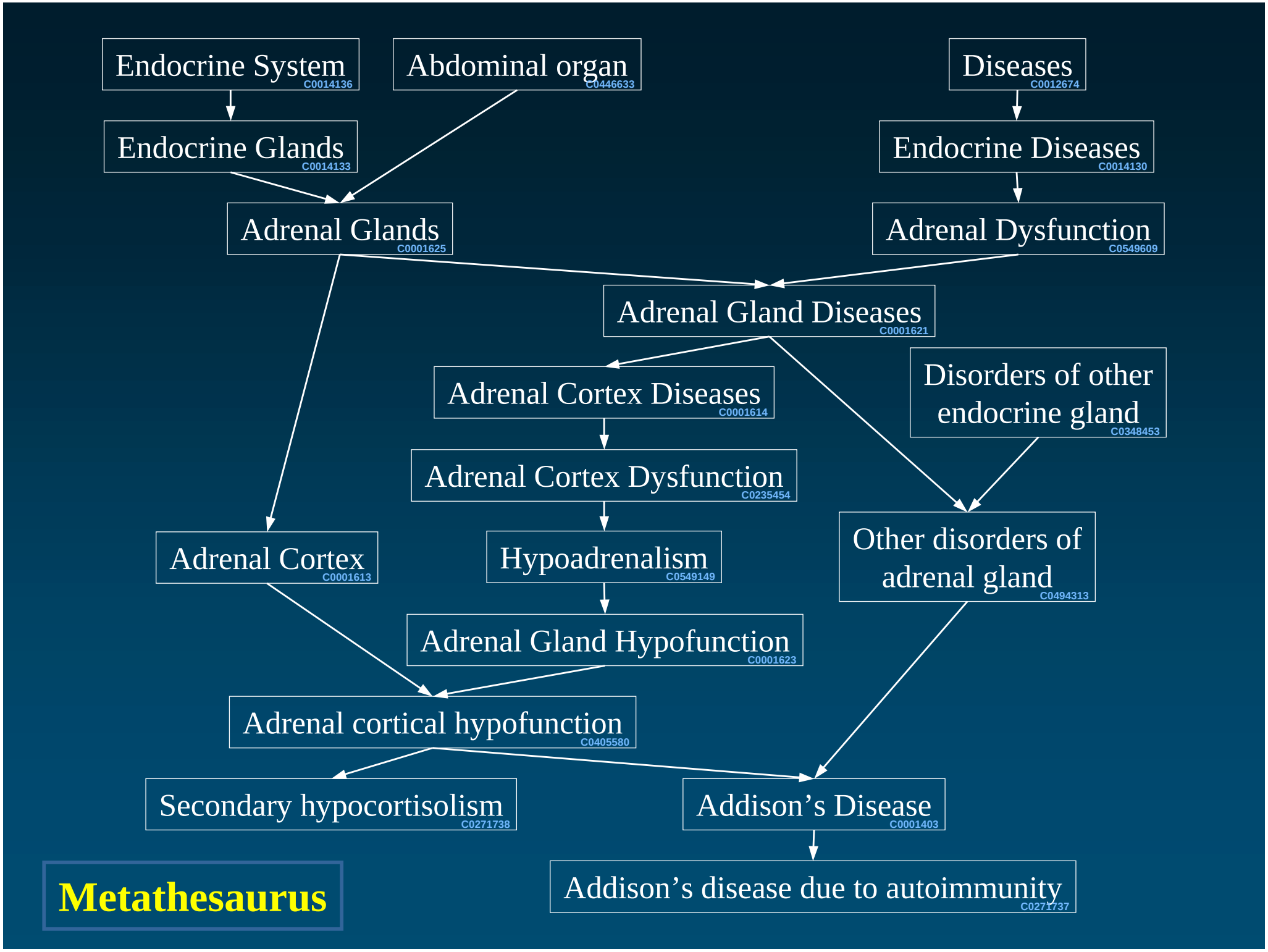


organize concepts

SNOMED
MeSH
AOD
Read Codes

UMLS

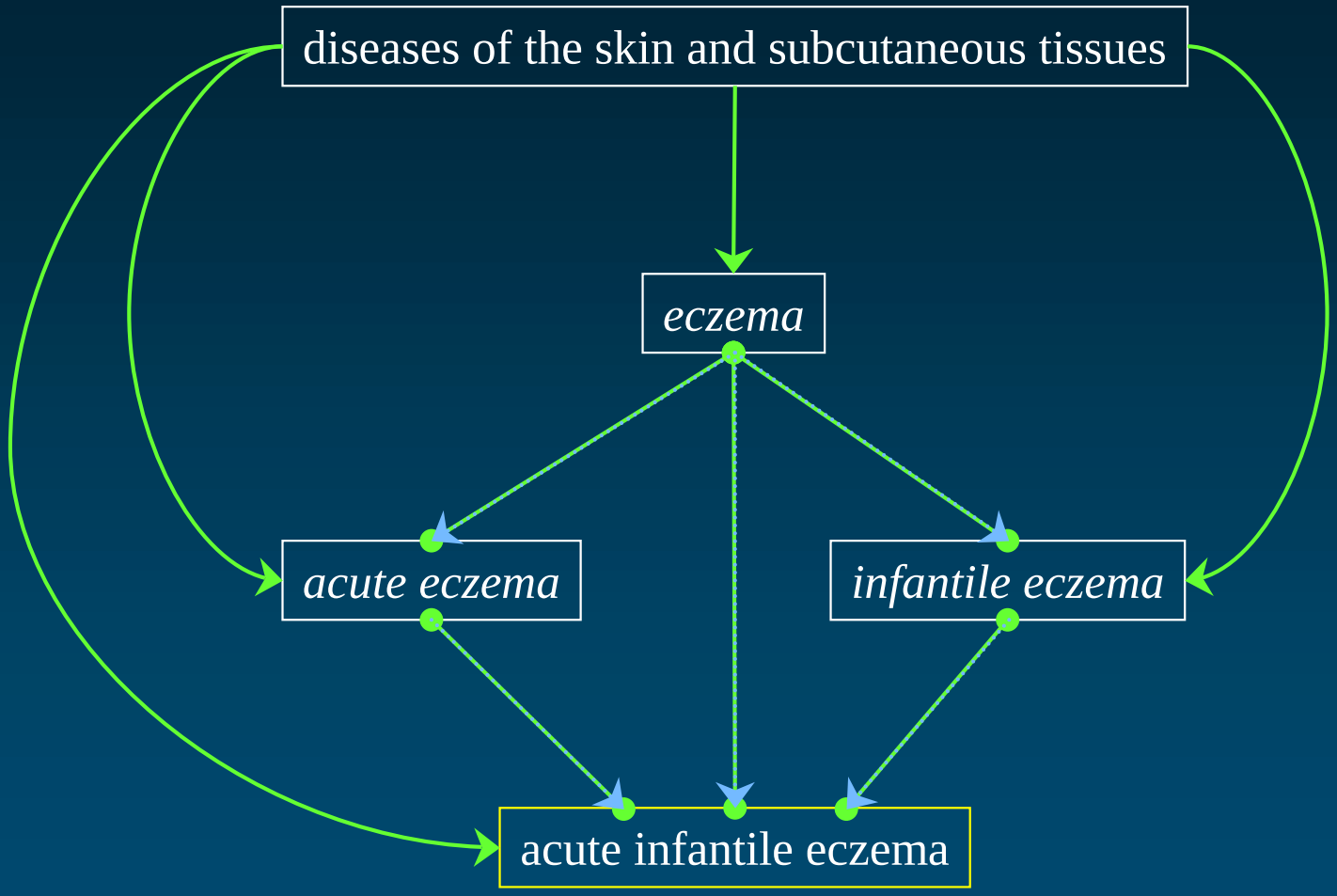



Endocrine System
C0014136
Abdominal organ
C0446633
Diseases
C0012674
Endocrine Glands
C0014133
Endocrine Diseases
C0014130
Adrenal Glands
C0001625
Adrenal Dysfunction
C0549609
Adrenal Gland Diseases
C0001621
Adrenal Cortex Diseases
C0001614
Disorders of other endocrine gland
C0348453
Adrenal Cortex Dysfunction
C0235454
Adrenal Cortex
C0001613
Hypoadrenalism
C0549149
Other disorders of adrenal gland
C0494313
Adrenal Gland Hypofunction
C0001623
Adrenal cortical hypofunction
C0405580
Secondary hypocortisolism
C0271738
Addison’s Disease
C0001403
Addison’s disease due to autoimmunity
C0271737
Metathesaurus

Granularity and synonymy

- Granularity may be limited in order to represent only significant differences among concepts (linguistic synonymy vs. clinical synonymy)
- When granularity is limited by design, it may not be possible to represent some hierarchical relationships
  Example: Acute infantile eczema in SNOMED

Lack of structure within a source



Granularity and redundancy

- "Core" concepts
  - Present in more than one vocabulary
  - Essentially eliminates leaf nodes (structural equivalent)
  - Also reduces the density of the graph
  - Reduces the number of concepts dramatically (> 80%)

Types vs. Instances

Types and instances Examples

- Types
  - Liver *is a kind of* Organ
  - Cirrhotic liver *is a kind of* Liver
- Instances
  - Leipzig *is an instance* of City
  - Barry Smith *is an instance* of Philosopher
  - My liver *is an instance* of Liver
  - This aspirin tablet *is an instance* of Clinical drug

Nothing can be *a kind of*

- Leipzig
- Barry Smith
- My liver
- This aspirin tablet

Types and instances Biomedical domain

- Types
  - Terminologies
  - Ontologies
- Instances
  - Medical records
  - Patient databases

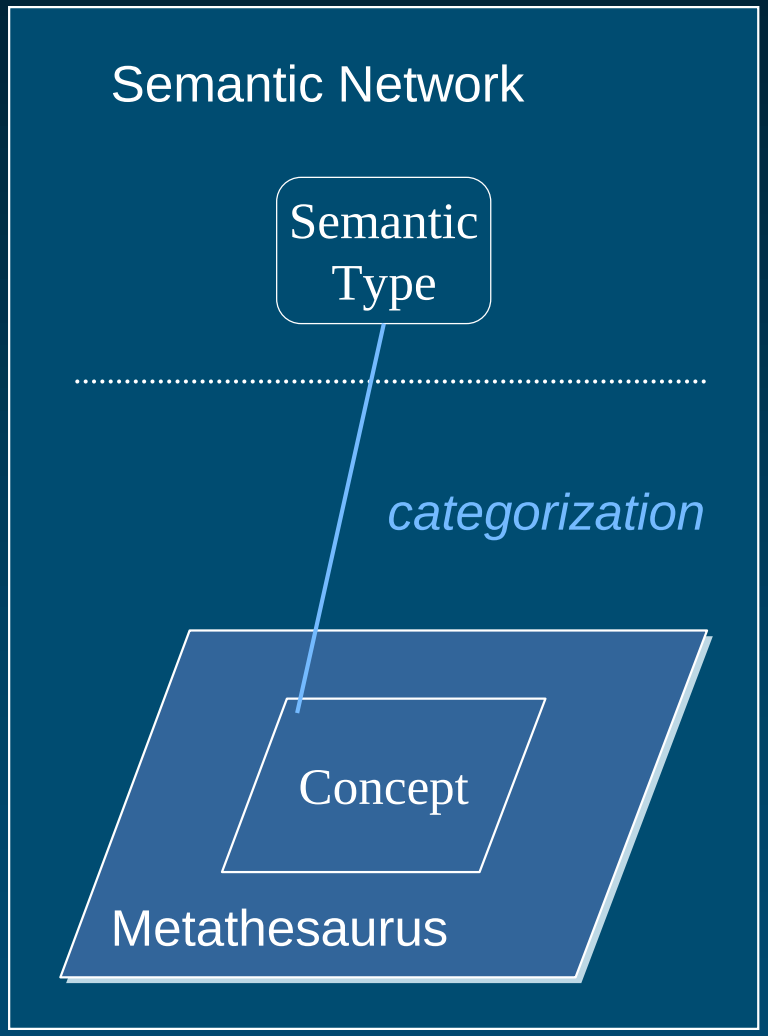
used to
abstract away from
or reason about

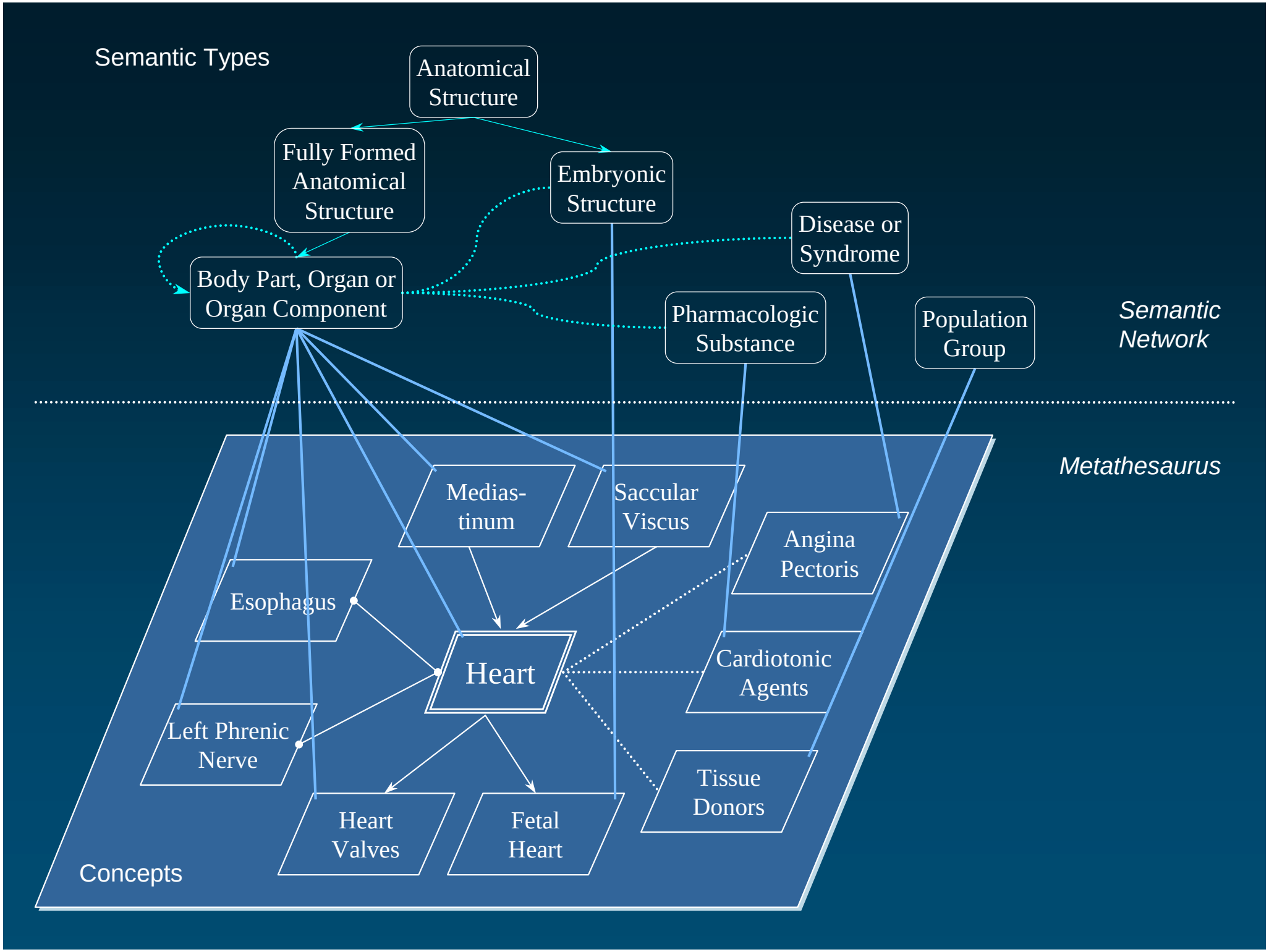
- Classes
  - Taxonomy
  - Classification

Class:	Schizomycetes
Order:	Eubacteriales
Family:	Bacillaceae
Genus:	Clostridium
Species:	Clostridium botulimun

Types and instances UMLS

- Two-level structure
  - Semantic Network
    - 134 Semantic Types (STs)
    - Relationships among STs
  - Metathesaurus
    - 800,000 concepts
    - Inter-concept relationships
  - Link = categorization
    - Often isa
    - Rarely is an instance of




Semantic Types
Anatomical Structure
Fully Formed Anatomical Structure
Embryonic Structure
Disease or Syndrome
Body Part, Organ or Organ Component
Pharmacologic Substance
Population Group
Semantic Network
Metathesaurus
Medias-tinum
Saccular Viscus
Angina Pectoris
Esophagus
Heart
Cardiotonic Agents
Left Phrenic Nerve
Tissue Donors
Heart Valves
Fetal Heart
Concepts

Types and instances UMLS

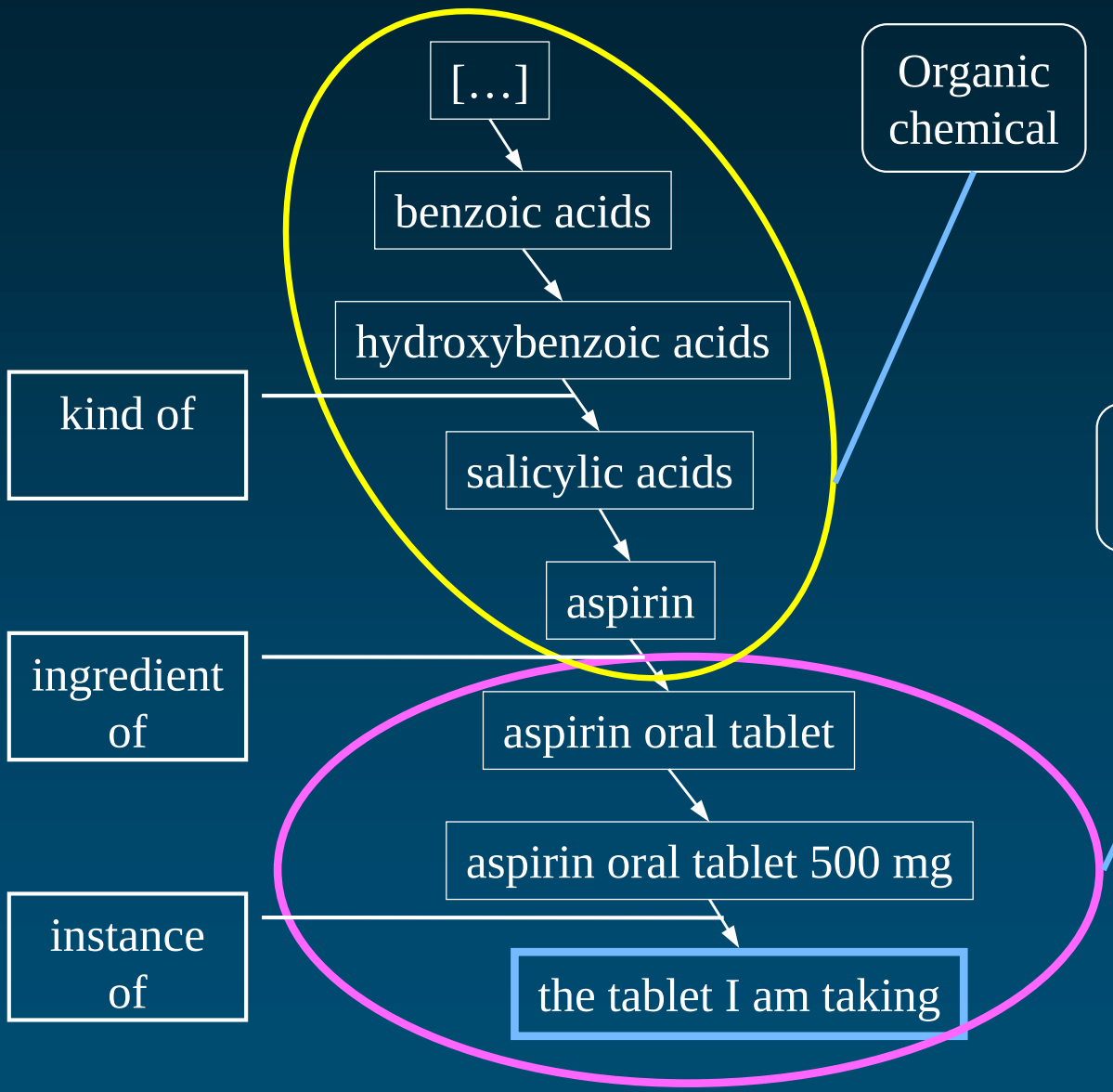
- Essentially all types
  - Semantic types
  - Concepts
- Exceptions
  - Named geographic areas
    Germany, Europe
  - Named laws
    National Health Planning and Resources Development Act of 1974
  - Named intellectual products
    Finnish translation of the Medical Subject Headings
- No explicit distinction
  between *is a kind of* and *is an instance of*

Types and instances Identification

[E. Alfonseca, GWA 2002]

- Features of instances
  - Structural: often leaf nodes
  - Morphologic: often capitalized
  - Syntactic: usually not preceded by a determiner
- Applications
  - Named entity recognition

Types, instances and granularity



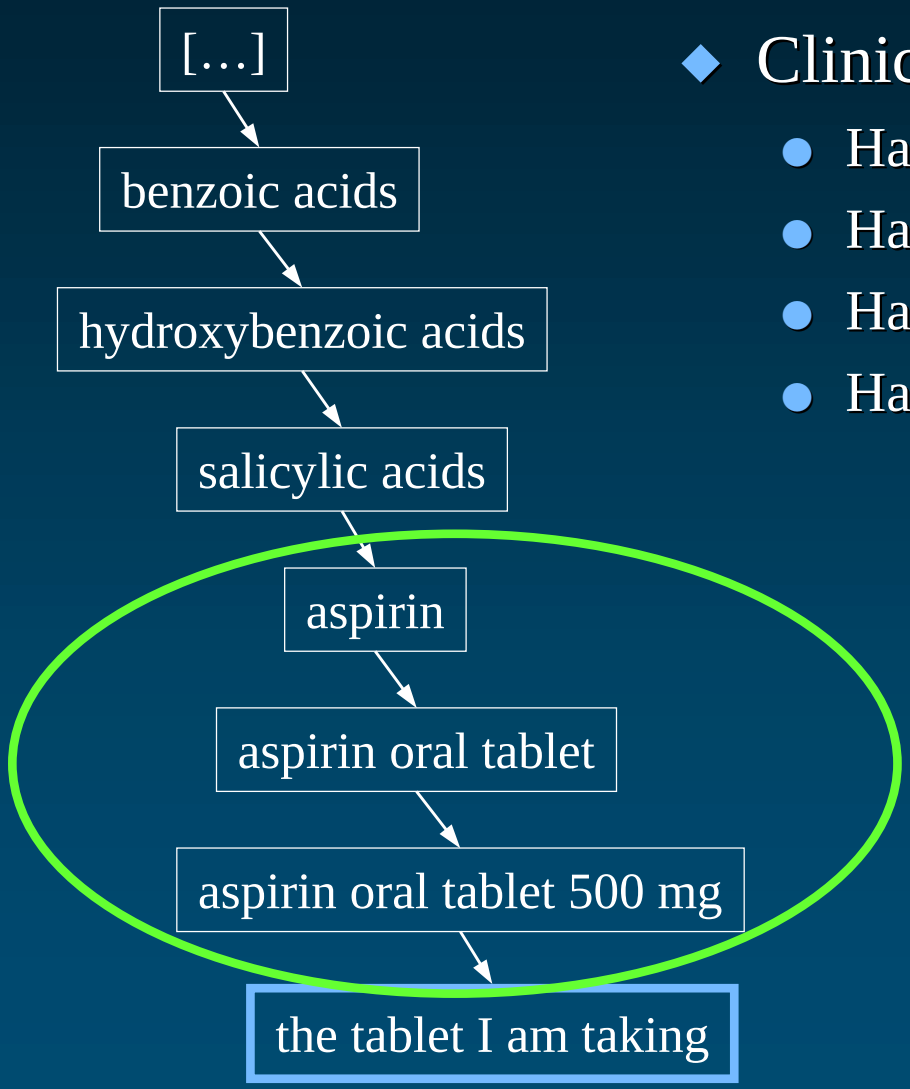
- Is there a gradient between type and instance?
- Is the hierarchy well-formed?

Types and instances Summary

- Biomedical ontology
  - Essentially types
  - Some classes (taxonomy)
  - Hardly any instances
- Instanciation: medical records
- Relationships: must distinguish between
  - Is an kind of (type-type)
  - Is an instance of (instance-class, type-class)

Ontology vs. Information model

Aspirin revisited



- Clinical drug
  - Has ingredient(s) aspirin
  - Has dosage 500 mg
  - Has route oral
  - Has form tablet

[RTM Drug Model, VA]

Other examples Medical procedures

- Medical procedure
  - Action
  - Anatomic site
  - Instruments
  - Approach
- Appendicectomy
  - Remove
  - Appendix
  - Surgical instruments
  - Open surgery

[MAOUSSC, GALEN-IN-USE, SNOMED-RT]

Other examples Lab/clinical results

- Lab test
  - Component
  - Property
  - Time aspect
  - System/Sample
  - Scale
  - Method
- Sodium measurement
  - Sodium
  - Serum concentration
  - Point in time
  - Serum/Plasma
  - Quantitative
  - N/A

LOINC “terms”: SODIUM:SCNC:PT:SER/PLAS:QN

[LOINC, Regenstrief Institute]

Ontology vs. Information model

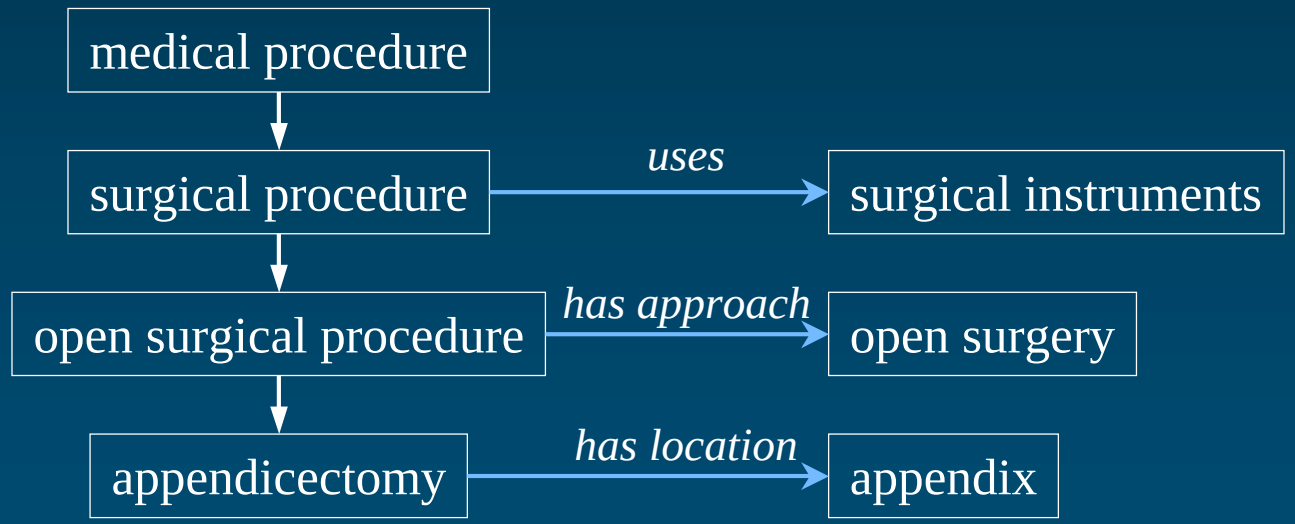
- Ontology
  - What: Meaning
  - How: hierarchies, frames, description logics
  - How big: often very large (hundreds of thousands of concepts)
  - Access: through browsers
- Information model
  - What: Structure
  - How: UML diagrams
  - How big: the model is often limited in size
  - Access: “readable” diagram
  - Populated with concepts from the ontology

[A. Rector, MEDINFO 2001]

Although O. and I.M. may be equivalent

- Appendicectomy
  - Remove
  - Appendix
  - Surgical instruments
  - Open surgery

(simplified representation)



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