

Using SNOMED CT with the UMLS



IHTSDO October Conference 2011
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Sydney, Australia

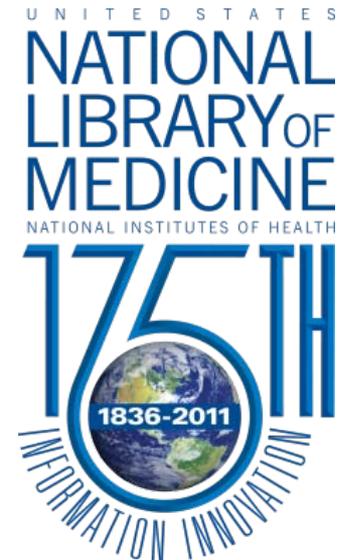
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Are there handouts?

- A copy of these slides is available at <http://mor.nlm.nih.gov/pubs/pres/20111014-IHTSDO-tutorial.pdf>



Outline

- Overview of the UMLS
- SNOMED CT integration into UMLS (principles and editorial choices)
- Representing SNOMED CT in the UMLS domain model
- Applying UMLS lexical tools to SNOMED CT descriptions
- Applying UMLS quality assurance processes to SNOMED CT content
- Browsing SNOMED CT with the UTS browser
- Accessing SNOMED CT content with the UTS API
- Finding correspondences to other terminologies through UMLS
- Finding terms in other languages through UMLS
- Use case: The role of UMLS in the NLM US SNOMED CT Content Request System (USCRS)

Dr. Olivier Bodenreider

Overview of the UMLS

Overview of the UMLS

Introduction



What does UMLS stand for?

- Unified
- Medical
- Language
- System



UMLS®

Unified Medical Language System®

UMLS Metathesaurus®

Motivation

- Started in 1986
- National Library of Medicine
- “Long-term R&D project”

«[...] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

- The first is **the variety of ways the same concepts are expressed** in different machine-readable sources and by different people.
- The second is the **distribution** of useful information among many disparate databases and systems.»

The UMLS in practice

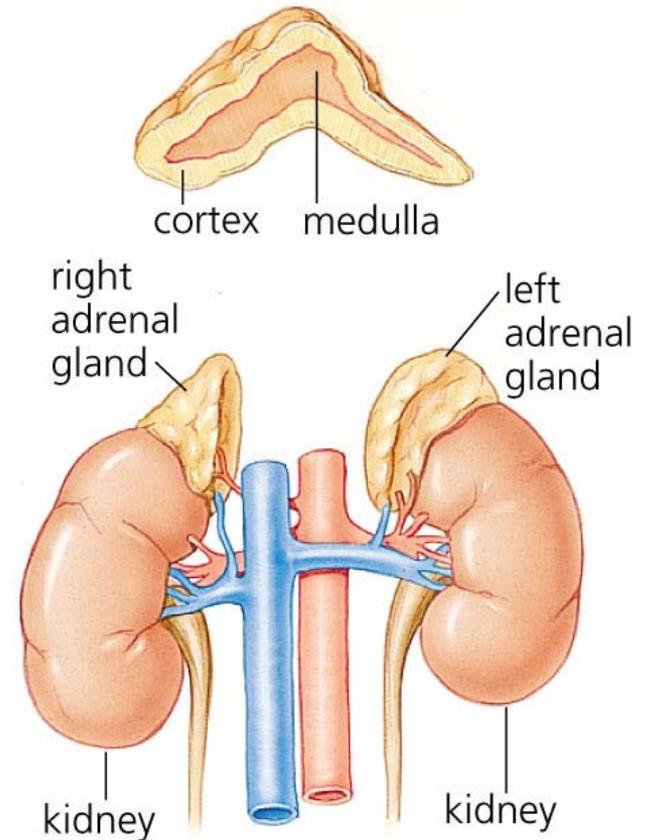
- Database
 - Series of relational files
- Interfaces
 - Web interface: UMLS Terminology Services (UTS)
 - Application programming interfaces (Java and web services)
- Applications
 - lvg (lexical programs)
 - MetamorphoSys (installation and customization)
 - RRF browser (browsing subsets)
- **The UMLS is *not* an end-user application**

Overview of the UMLS

Overview through an example

Addison's disease

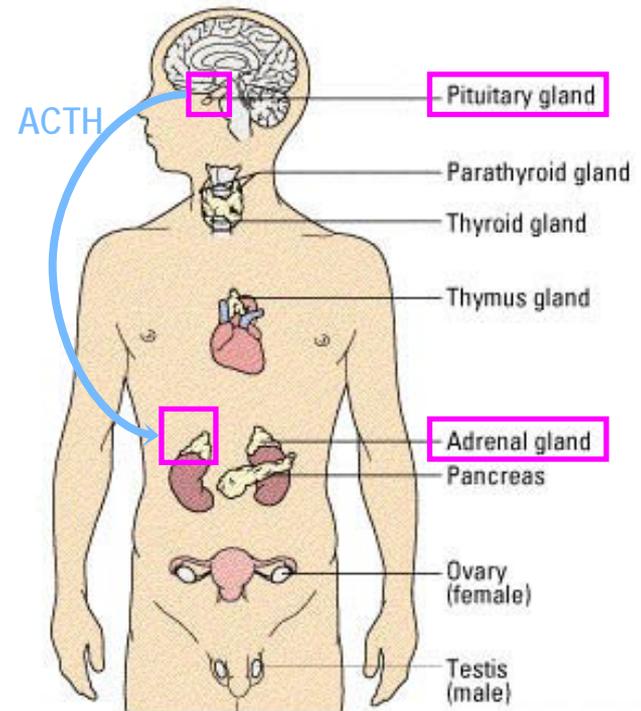
- Addison's disease is a rare **endocrine disorder**
- Addison's disease occurs when the **adrenal glands** do not produce enough of the hormone **cortisol**
- For this reason, the disease is sometimes called **chronic adrenal insufficiency**, or **hypocortisolism**



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

The Endocrine System
Glands which release chemicals directly into the blood stream.



adam.com

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

- Melasma addisonii

- Asthenia pigmentosa

- Primary adrenal deficiency

- Primary adrenal insufficiency

- Primary adrenocortical insufficiency

- Chronic adrenocortical insufficiency

} eponym

} symptoms

} clinical variants

- Contexts: **different hierarchies**

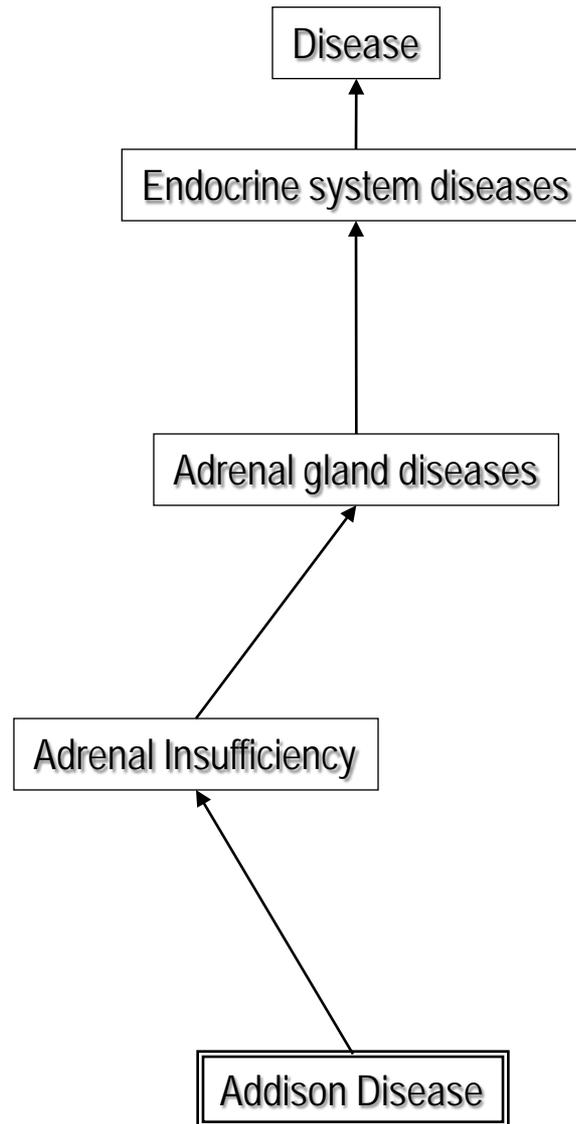
Organize terms

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

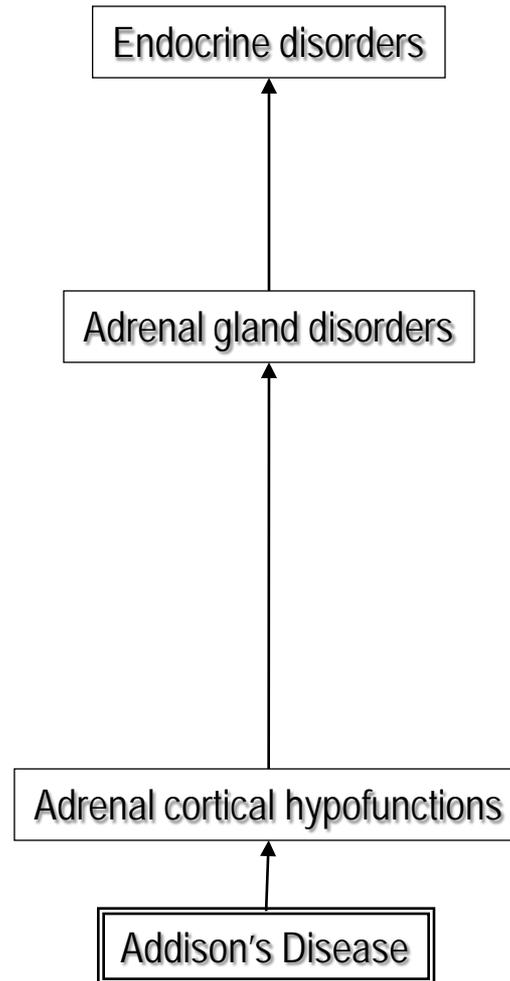
Addison Disease	MeSH	D000224
Primary hypoadrenalism	MedDRA	10036696
Primary adrenocortical insufficiency	ICD-10	E27.1
Addison's disease (disorder)	SNOMED CT	363732003

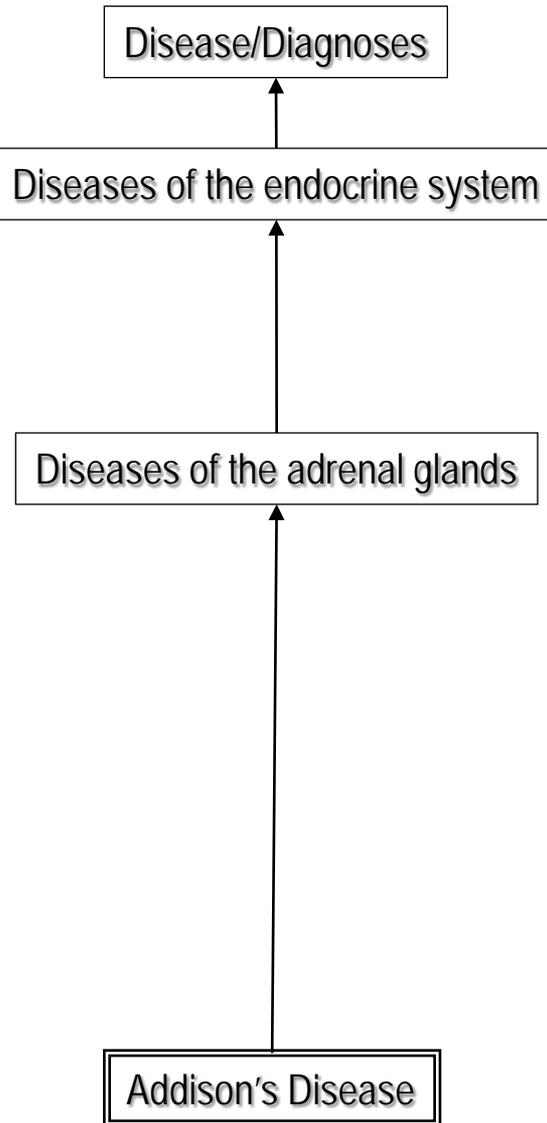
C0001403

Addison's disease

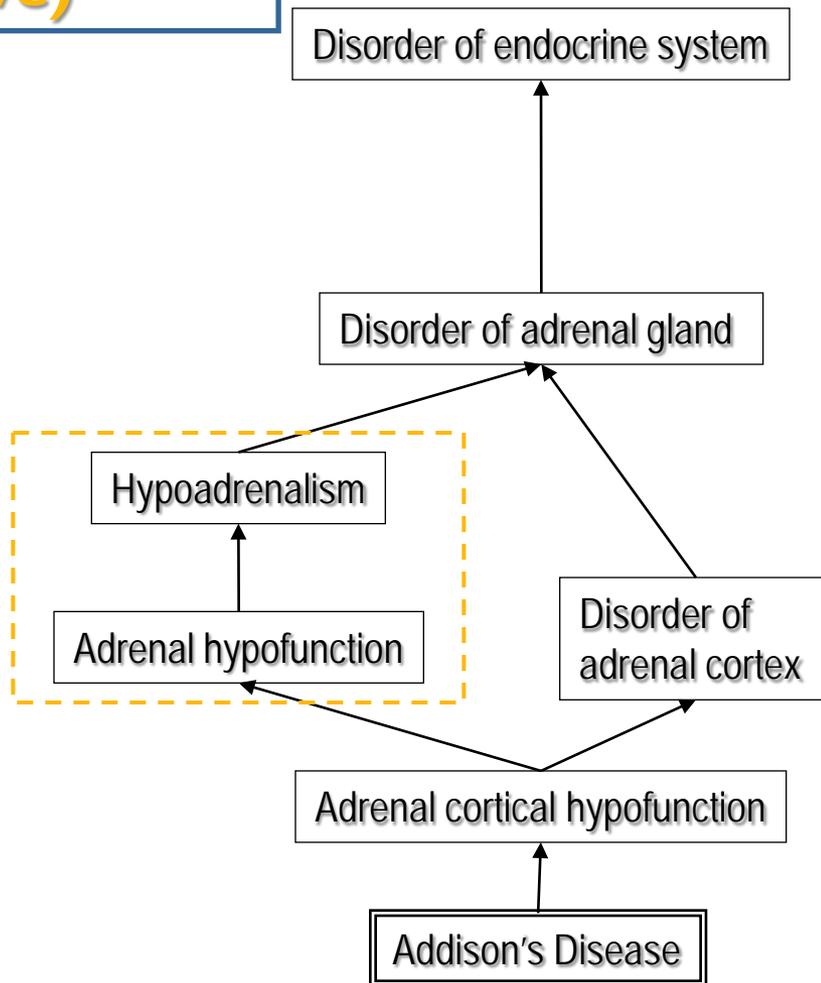


MedDRA

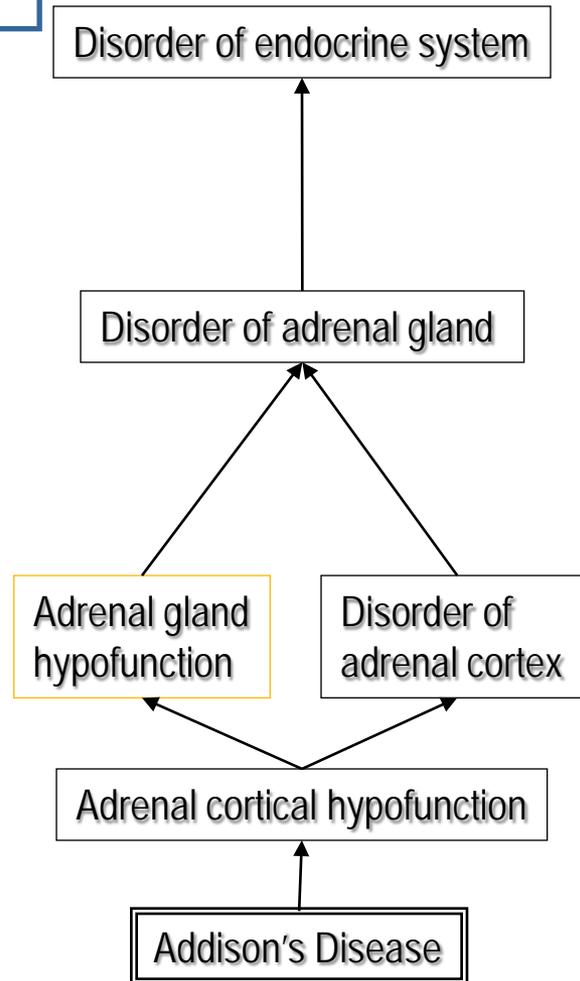




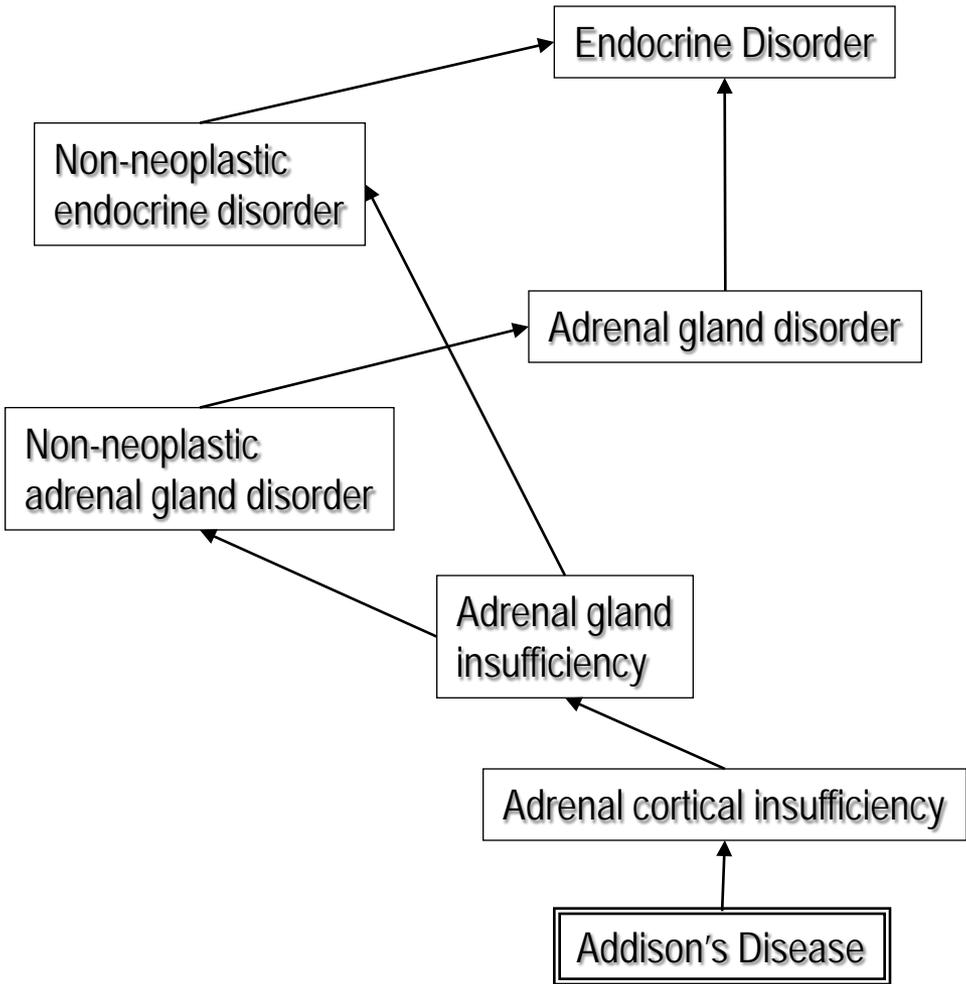
SNOMED CT (native)



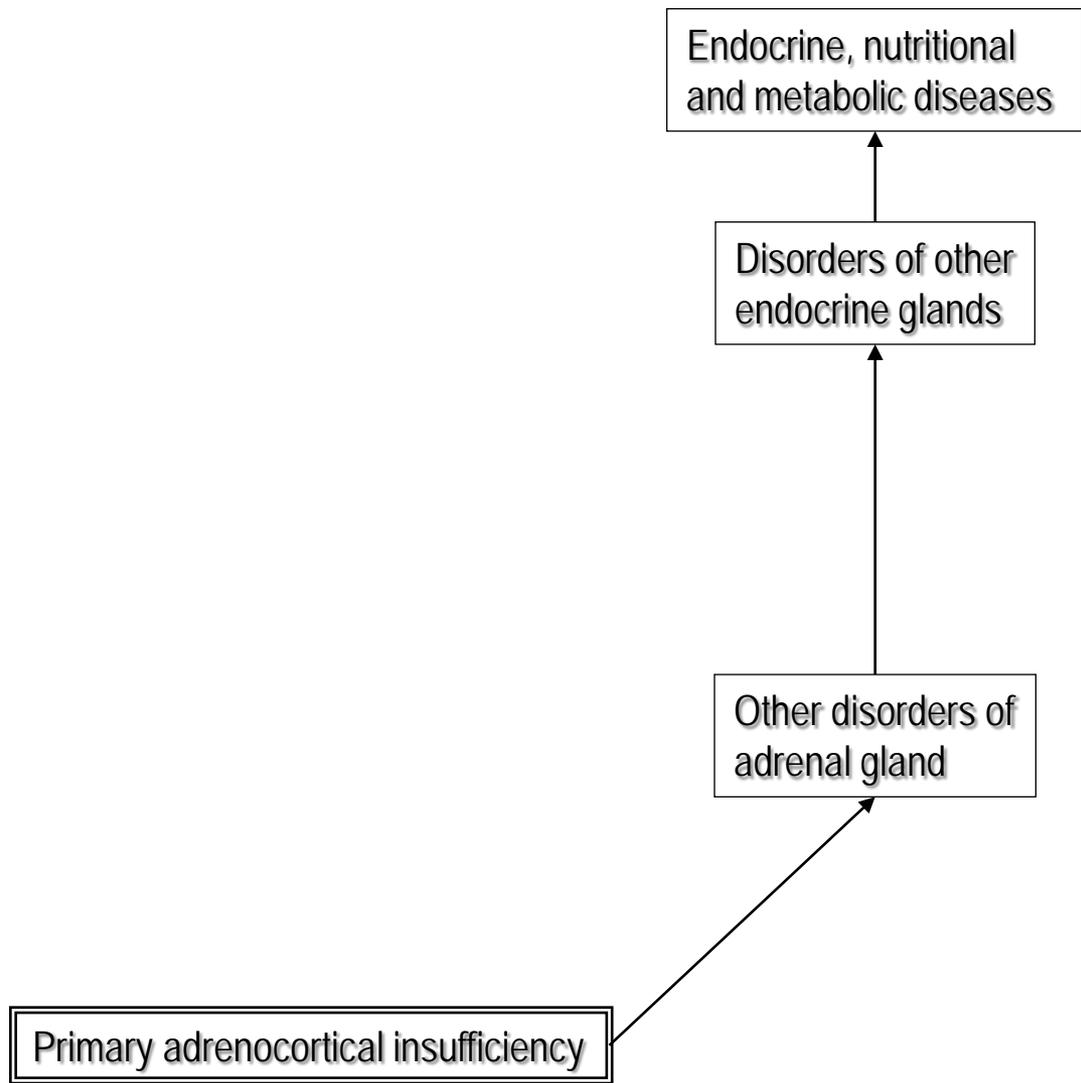
SNOMED CT (UMLS view)



NCI Thesaurus

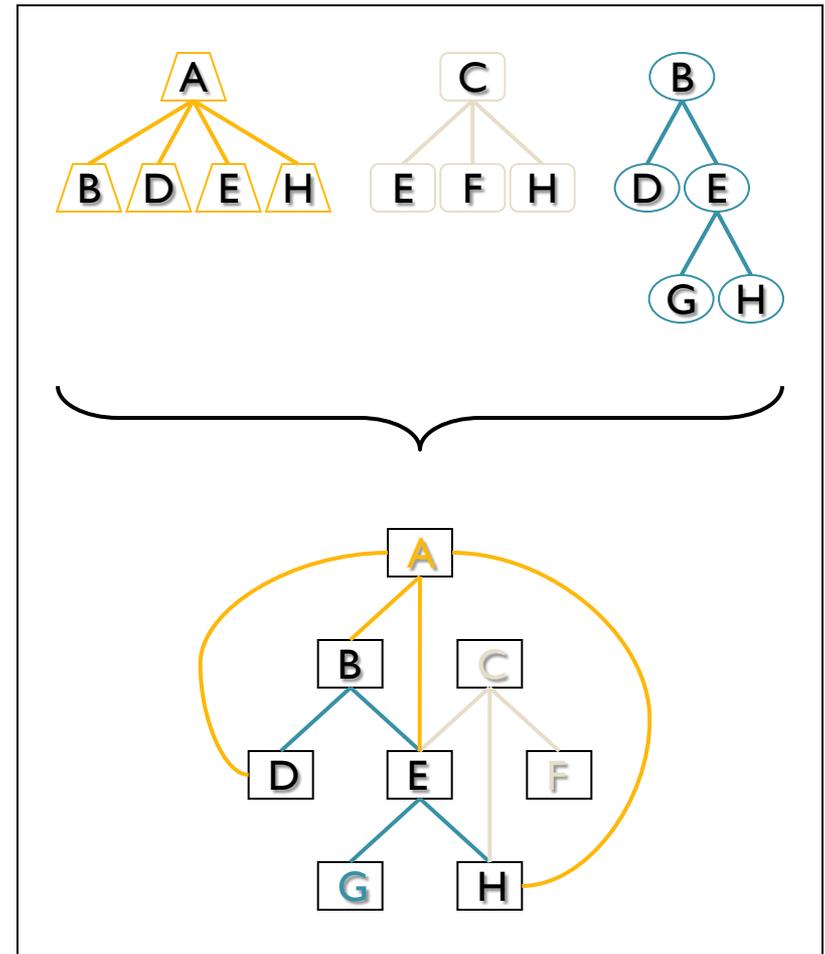


ICD-10

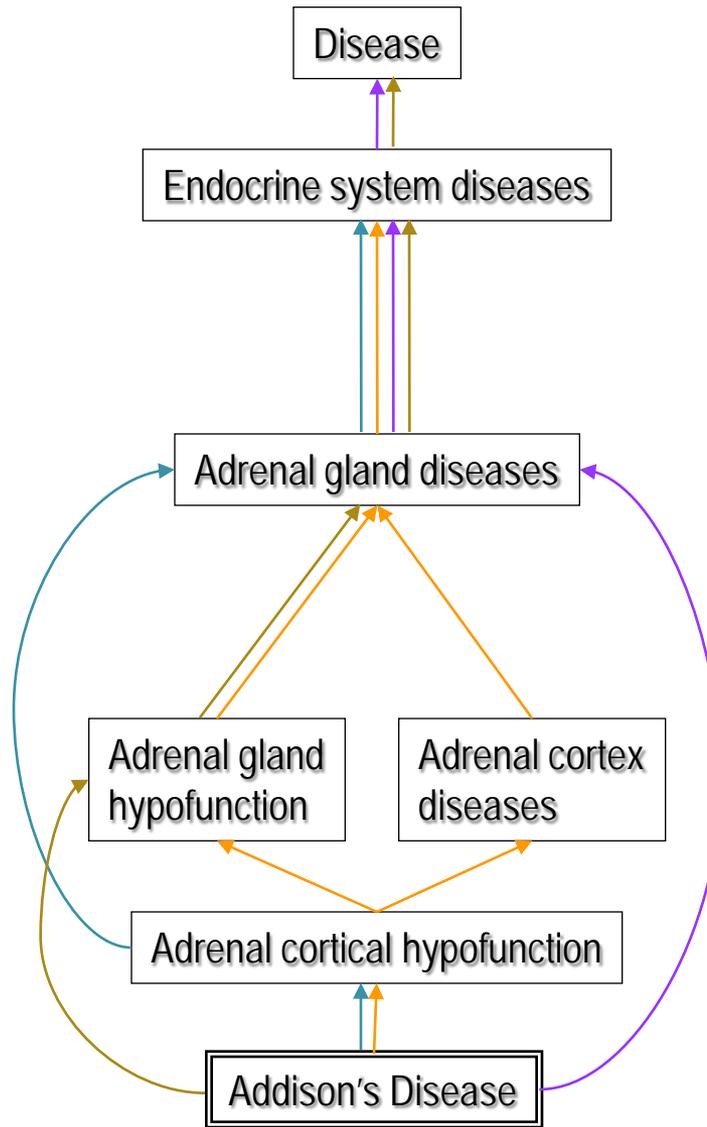


Organize concepts

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

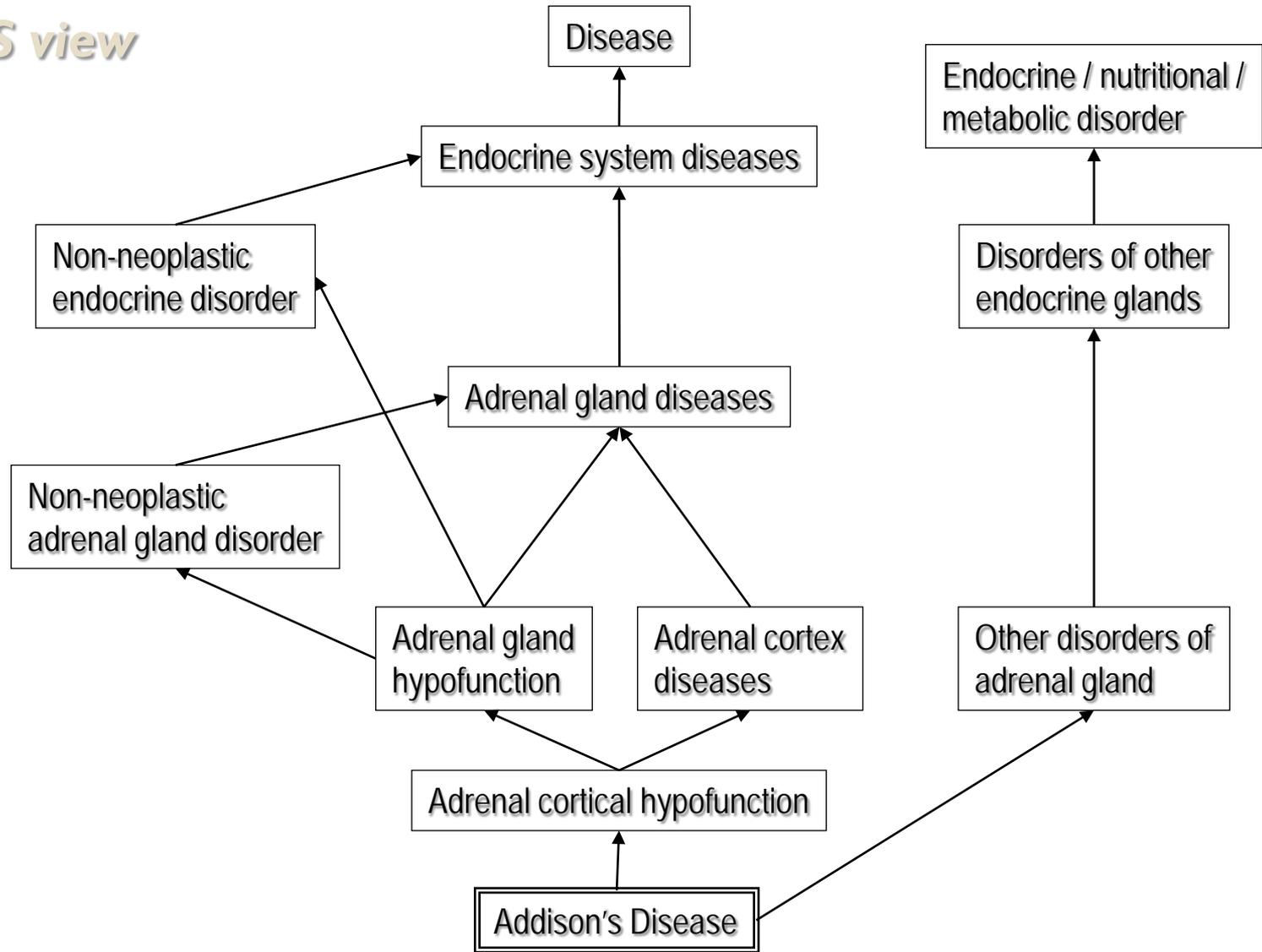


organize concepts

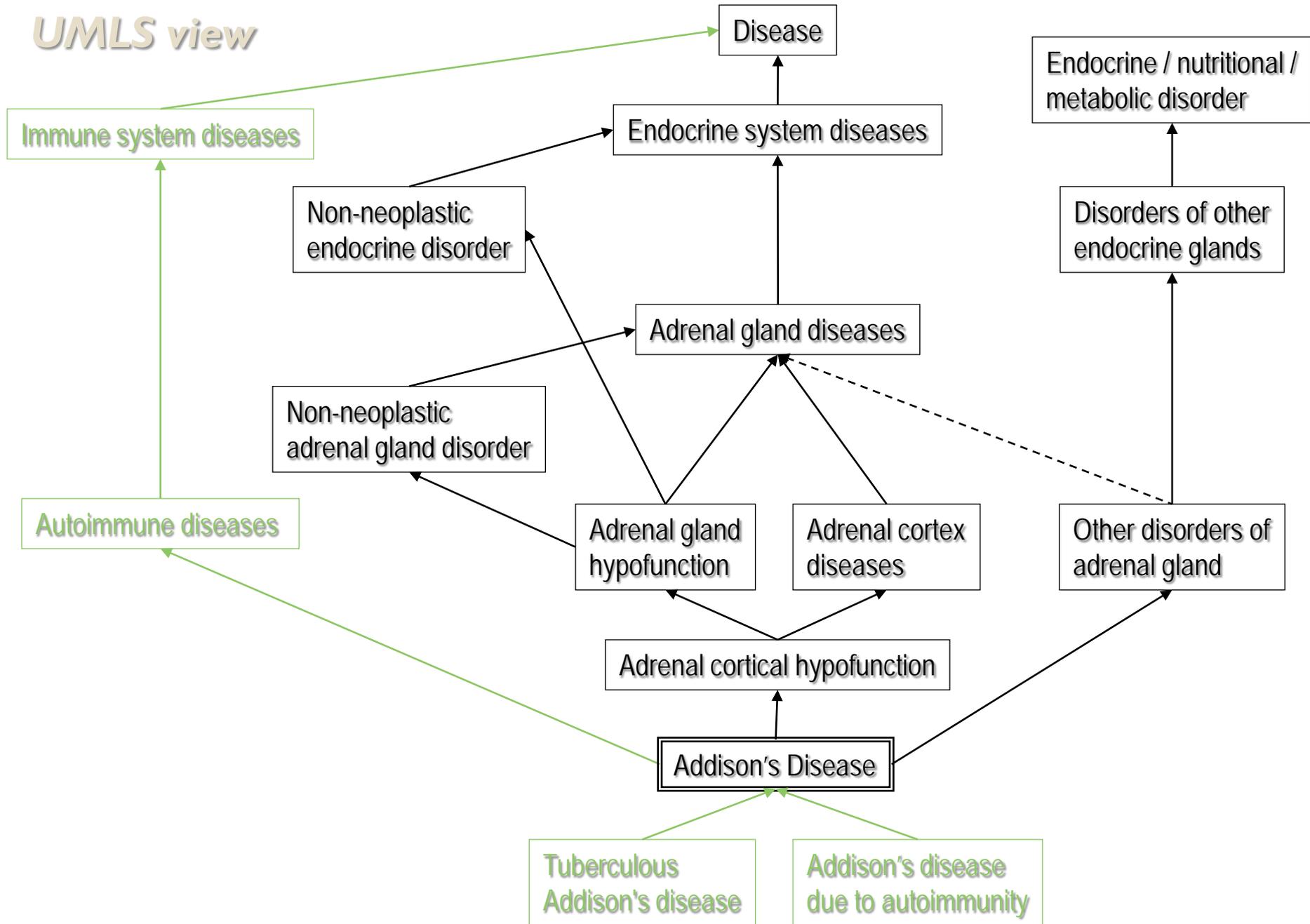


SNOMED CT
SNOMED Intl
MeSH
MedDRA

UMLS view



UMLS view



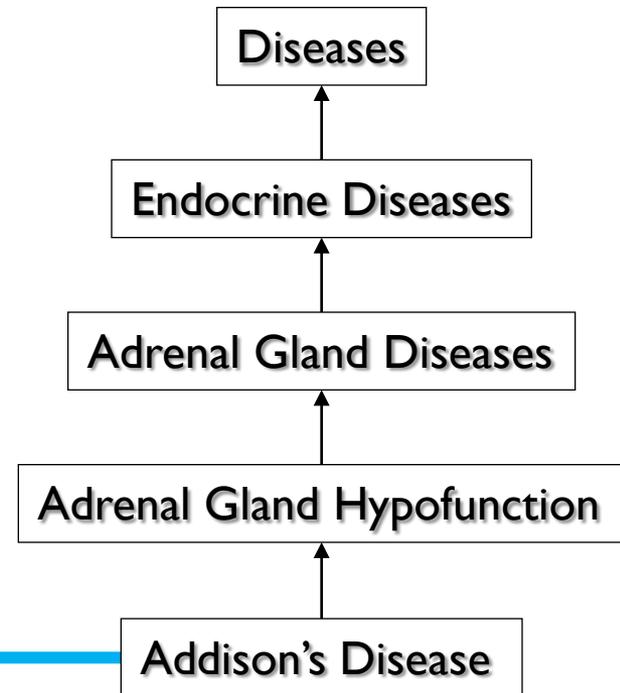
Relate to other concepts

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

Categorize concepts

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

Disease or Syndrome



How do they do that?

- Lexical knowledge
- Semantic pre-processing
- UMLS editors

Lexical knowledge

Adrenal gland diseases

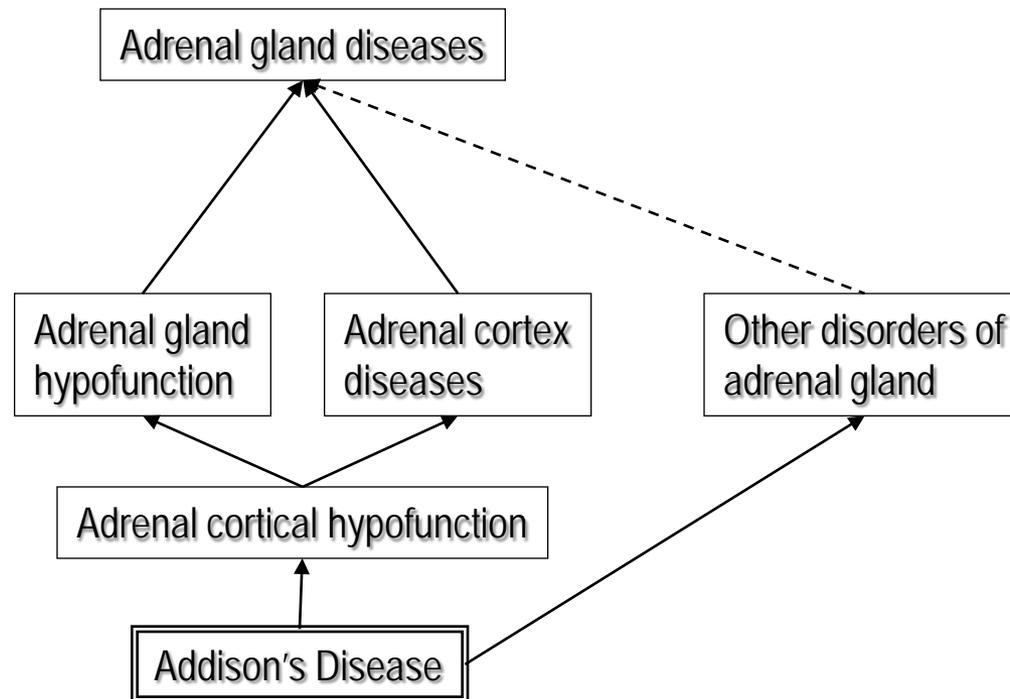
Diseases of the adrenal glands

C0001621

Semantic pre-processing

- Metadata in the source vocabularies
- Tentative categorization
- Positive (or negative) evidence for tentative synonymy relations based on lexical features

Additional knowledge: UMLS editors



UMLS Summary

- Synonymous terms clustered into concepts
- Unique identifier
- Finer granularity
- Broader scope
- Additional hierarchical relationships
- Semantic categorization

Overview of the UMLS

UMLS Knowledge Sources



UMLS 3 components



- Metathesaurus
 - Concepts
 - Inter-concept relationships
- Semantic Network
 - Semantic types
 - Semantic network relationships
- Lexical resources
 - SPECIALIST Lexicon
 - Lexical tools

UMLS Metathesaurus



Metathesaurus Basic organization

- Concepts
 - Synonymous terms are clustered into a concept
 - Properties are attached to concepts, e.g.,
 - Unique identifier
 - Definition
- Relations
 - Concepts are related to other concepts
 - Properties are attached to relations, e.g.,
 - Type of relationship
 - Source

Source Vocabularies

(2011AA)

- 160 source vocabularies
- 21 languages
- Broad coverage of biomedicine
 - 8M names (normalized)
 - 2.4M concepts
 - >10M relations
- Common presentation

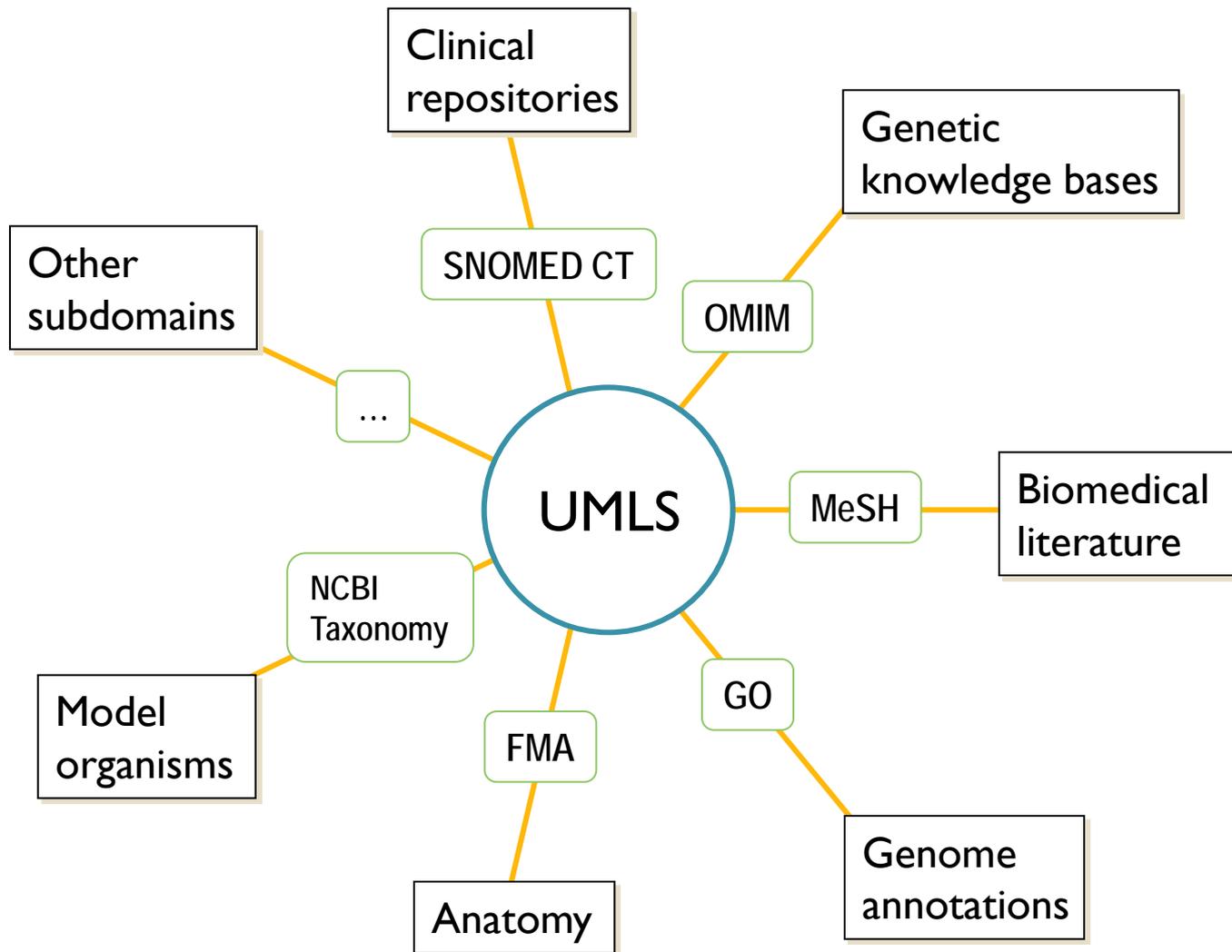
Biomedical terminologies

- General vocabularies
 - anatomy (FMA, Neuronames)
 - drugs (RxNorm, First DataBank, Micromedex)
 - medical devices (UMD, SPN)
- Several perspectives
 - clinical terms (SNOMED CT)
 - information sciences (MeSH)
 - administrative terminologies (ICD-9-CM, CPT-4)
 - data exchange terminologies (HL7, LOINC)

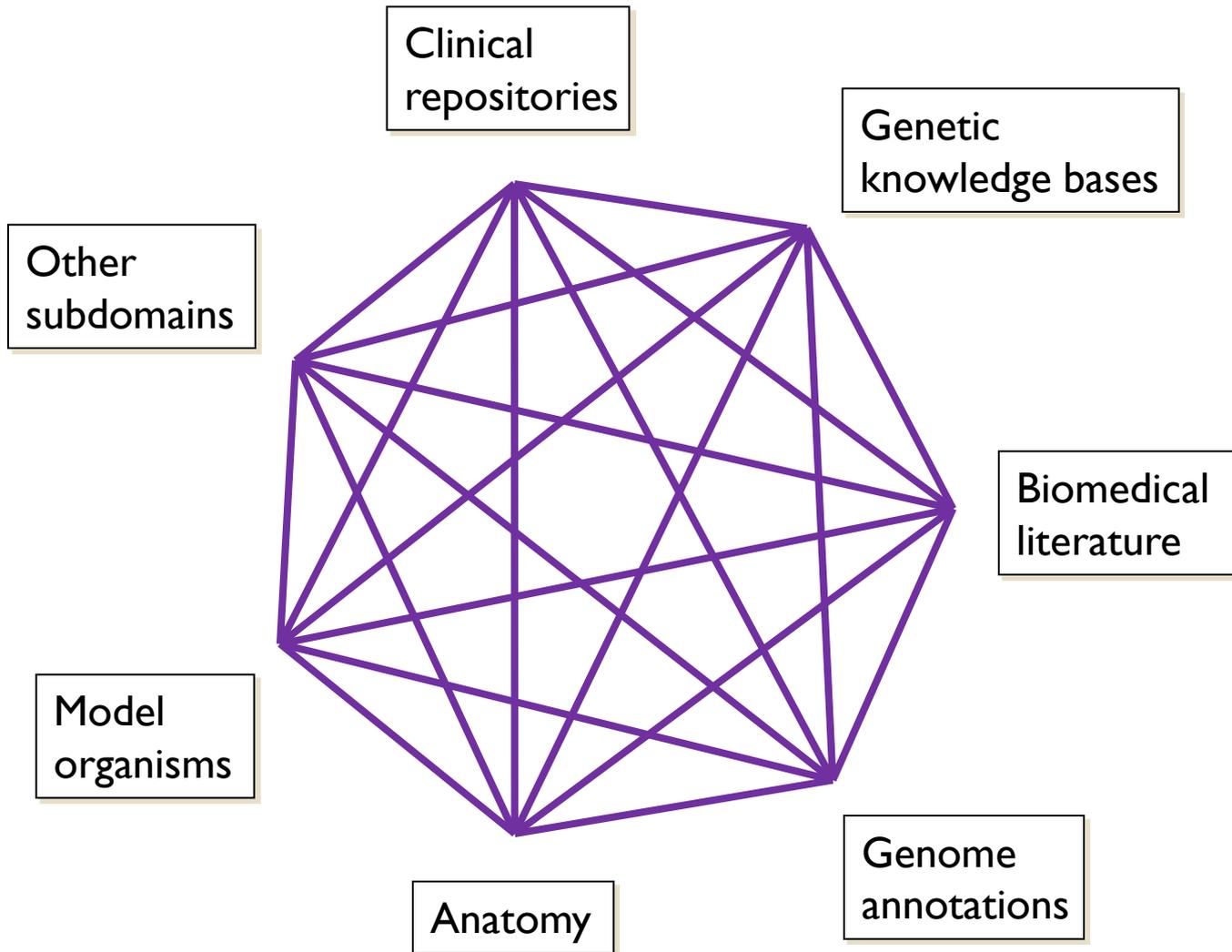
Biomedical terminologies (cont'd)

- Specialized vocabularies
 - nursing (NIC, NOC, NANDA, Omaha, ICNP)
 - dentistry (CDT)
 - oncology (PDQ)
 - psychiatry (DSM, APA)
 - adverse reactions (MedDRA, WHO ART)
 - primary care (ICPC)
- Terminology of knowledge bases (AI/Rheum, DXplain, QMR)
- **The UMLS serves as a vehicle for the regulatory standards (HIPAA, HITSP, Meaningful Use)**

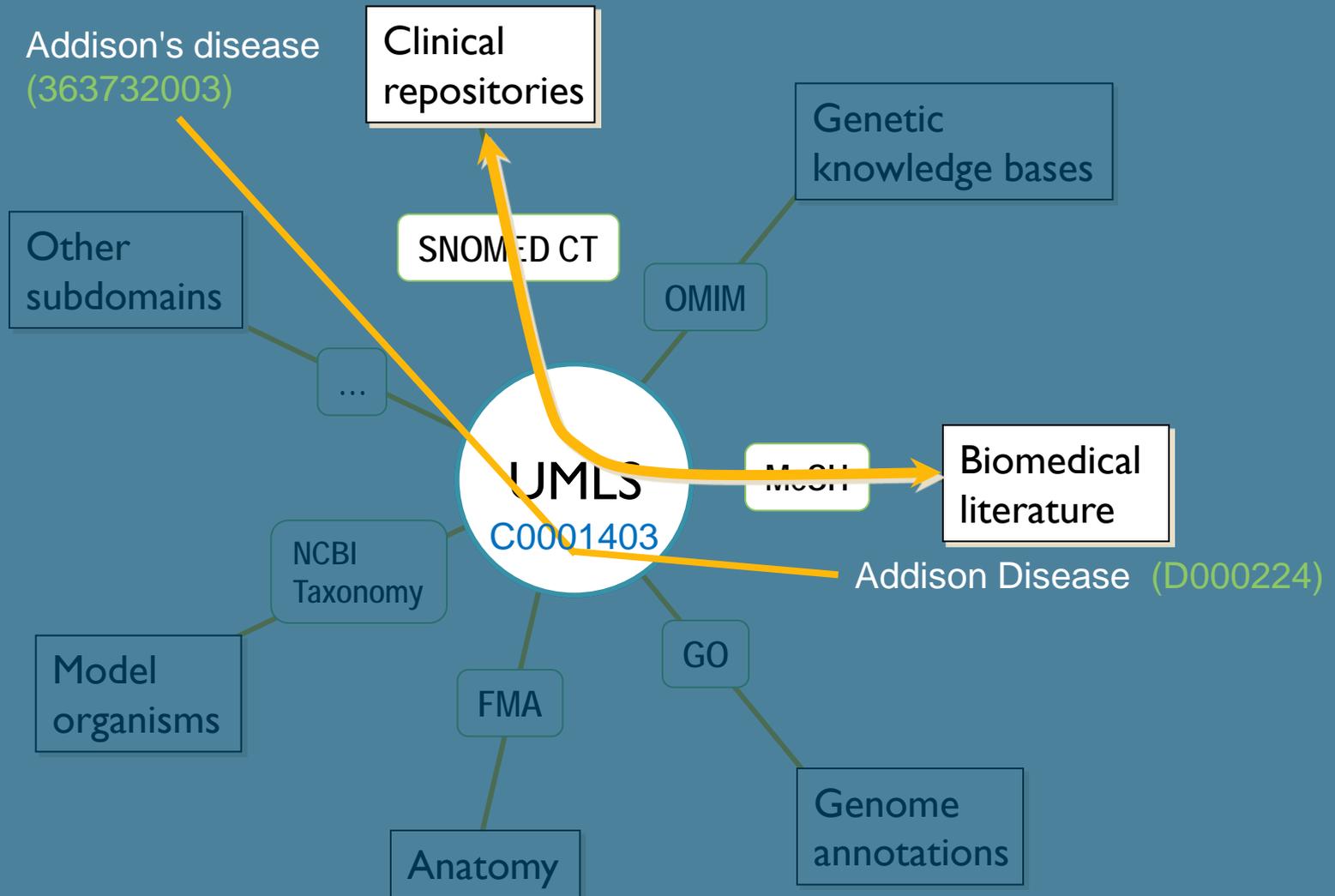
Integrating subdomains



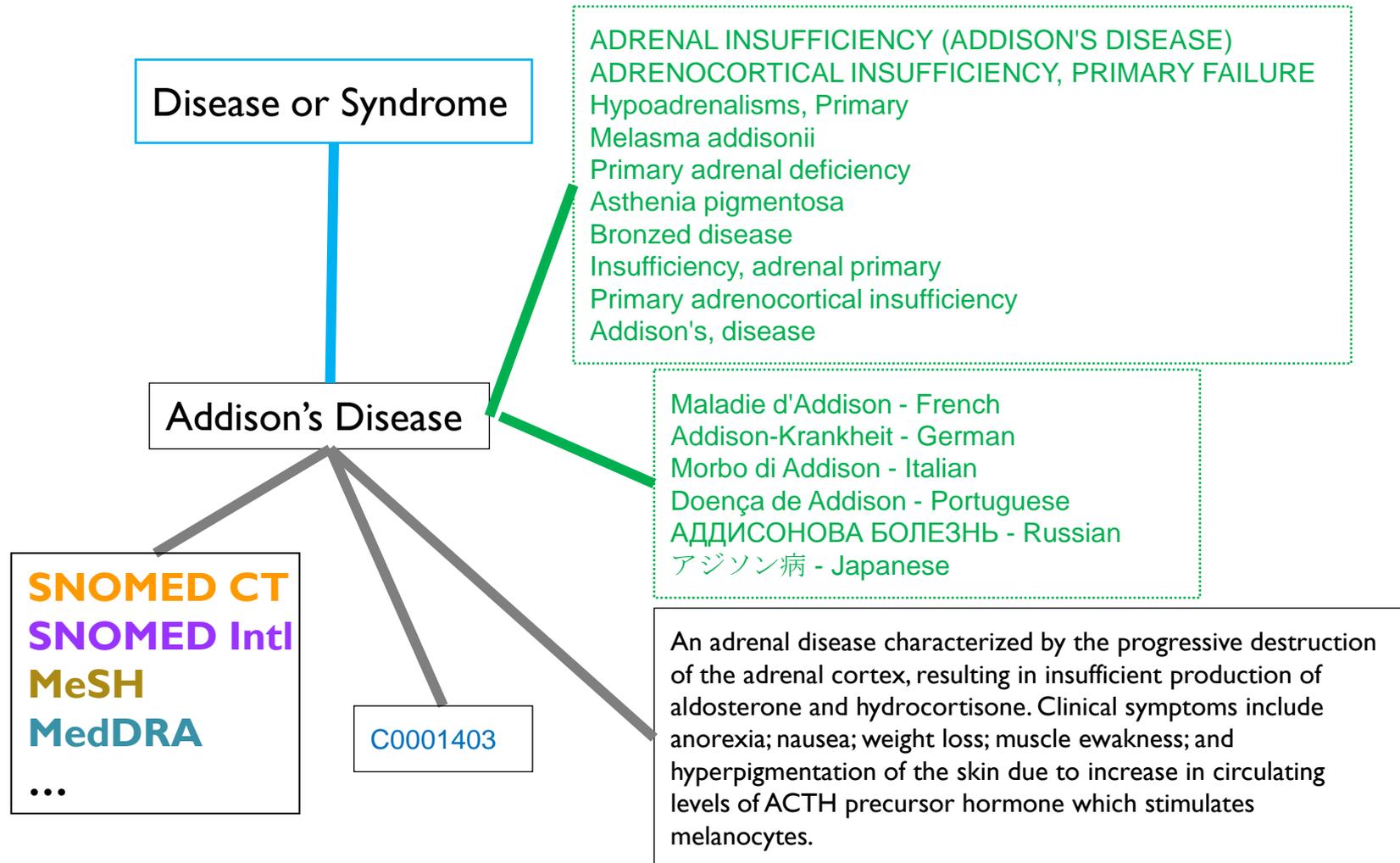
Integrating subdomains



Trans-namespace integration



Addison's Disease: Concept



Metathesaurus Concepts

(2011AA)

- Concept (2.6M) **CUI**
 - Set of synonymous concept names
- Term (7.9M) **LUI**
 - Set of normalized names
- String (8.9M) **SUI**
 - Distinct concept name
- Atom (10.6M) **AUI**
 - Concept name in a given source

A0066000	Headache	(MeSH)
A0065992	Headache	(ICD-10)
S0046854		

A0066007	Headaches	(MedDRA)
A12003304	Headaches	(OMIM)
S0046855		

L0018681

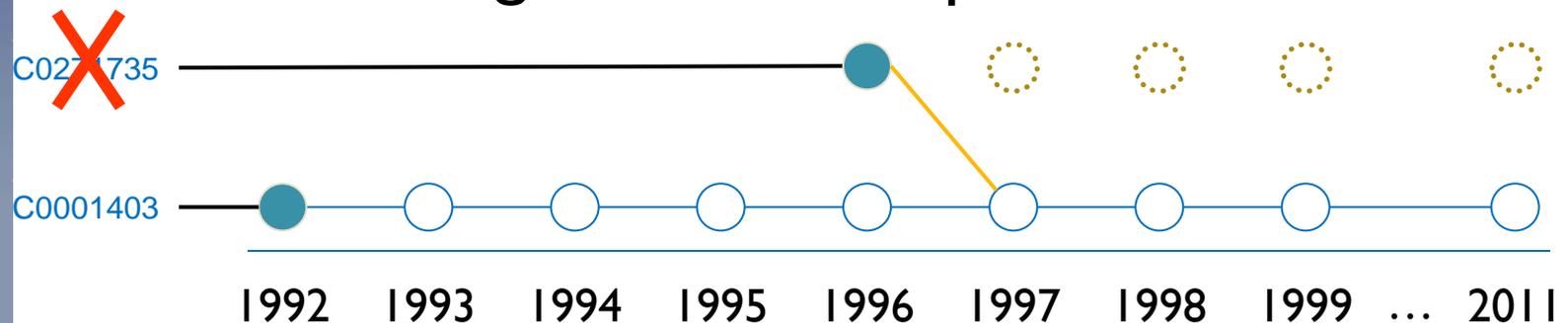
A0540936	Cephalodynia	(MeSH)
S0475647		

L0380797

C0018681

Metathesaurus Evolution over time

- Concepts never die (in principle)
 - CUIs are permanent identifiers
- What happens when they do die (in reality)?
 - Concepts can merge or split
 - Resulting in new concepts and deletions



Metathesaurus Relations

- Symbolic relations: ~8 M pairs of concepts
- Statistical relations: ~6 M pairs of concepts (co-occurring concepts)
- Mapping relations: ~150,000

-
- Categorization: Relationships between concepts and semantic types from the Semantic Network

Symbolic relations

- Relation
 - Pair of “atom” identifiers
 - Type
 - Attribute (if any)
 - List of sources (for type and attribute)
- Semantics of the relationship:
defined by its **type** [and **attribute**]

Source transparency: the information is recorded at the “atom” level

Mapping relations

- Simple mappings
 - <atom 1> mapped_to <atom 2>
 - e.g.,
 - SNOMED CT to ICD-9-CM
- Complex mappings
 - <atom 1> mapped_to <boolean expression>
 - e.g.,
 - ICD-9-CM to MeSH (search strategies)

NB: partially redundant with relations in MRREL

Everything else

- Co-occurrence information (MRCOC)
 - Co- occurrence of MeSH descriptors in MEDLINE for the most part
- Source-specific attributes (MRSAT)
 - Legacy identifiers, external cross-references
 - SNOMED International legacy codes (SNOMED CT)
 - RxNorm to NDC
 - Concept status in a particular source (SNOMED CT)
 - Frequency of occurrence in MEDLINE (MeSH)
 - MedlinePlus URL (MeSH)
 - ...

UMLS Semantic Network

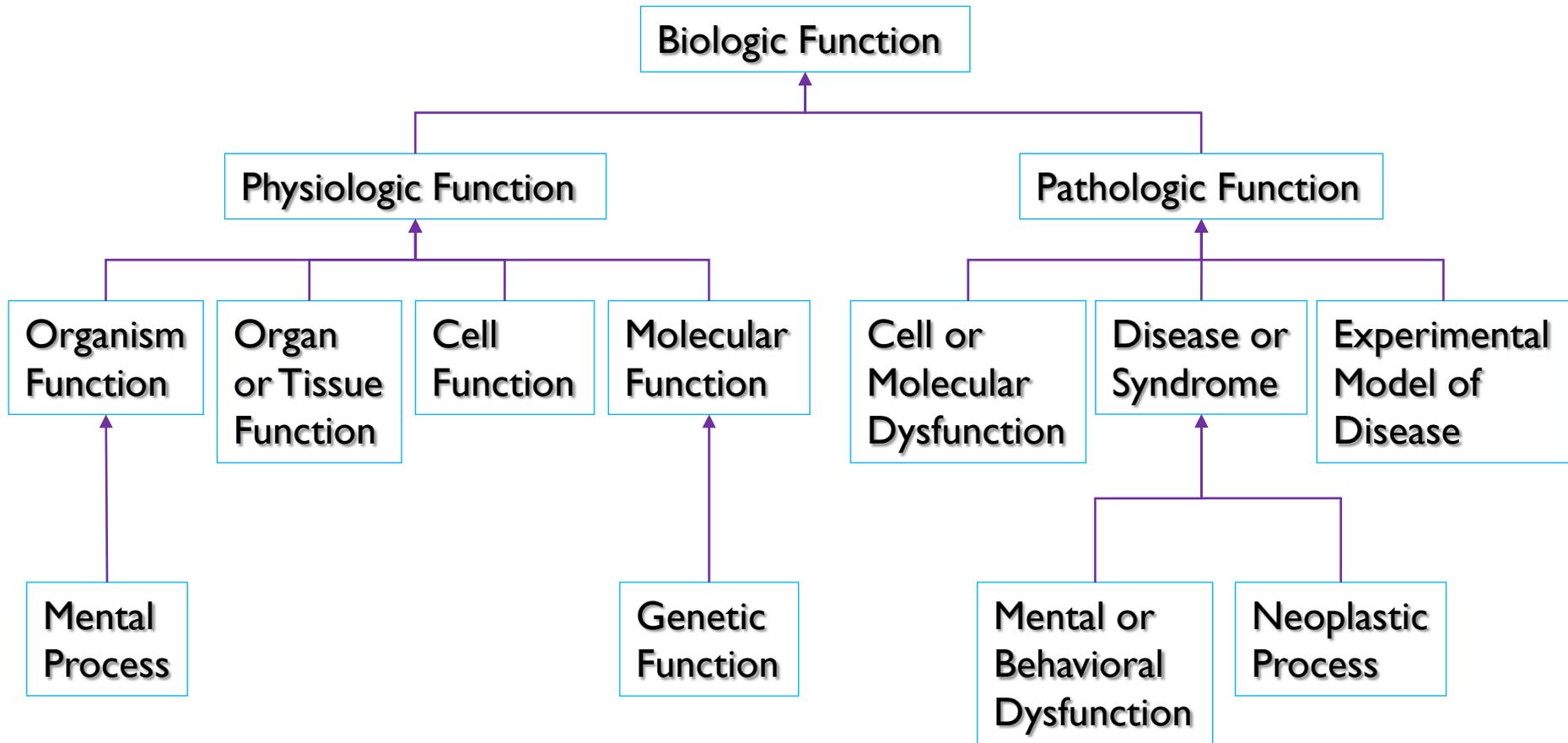
Semantic Network

- Semantic types (133)
 - tree structure
 - 2 major hierarchies
 - Entity
 - Physical Object
 - Conceptual Entity
 - Event
 - Activity
 - Phenomenon or Process

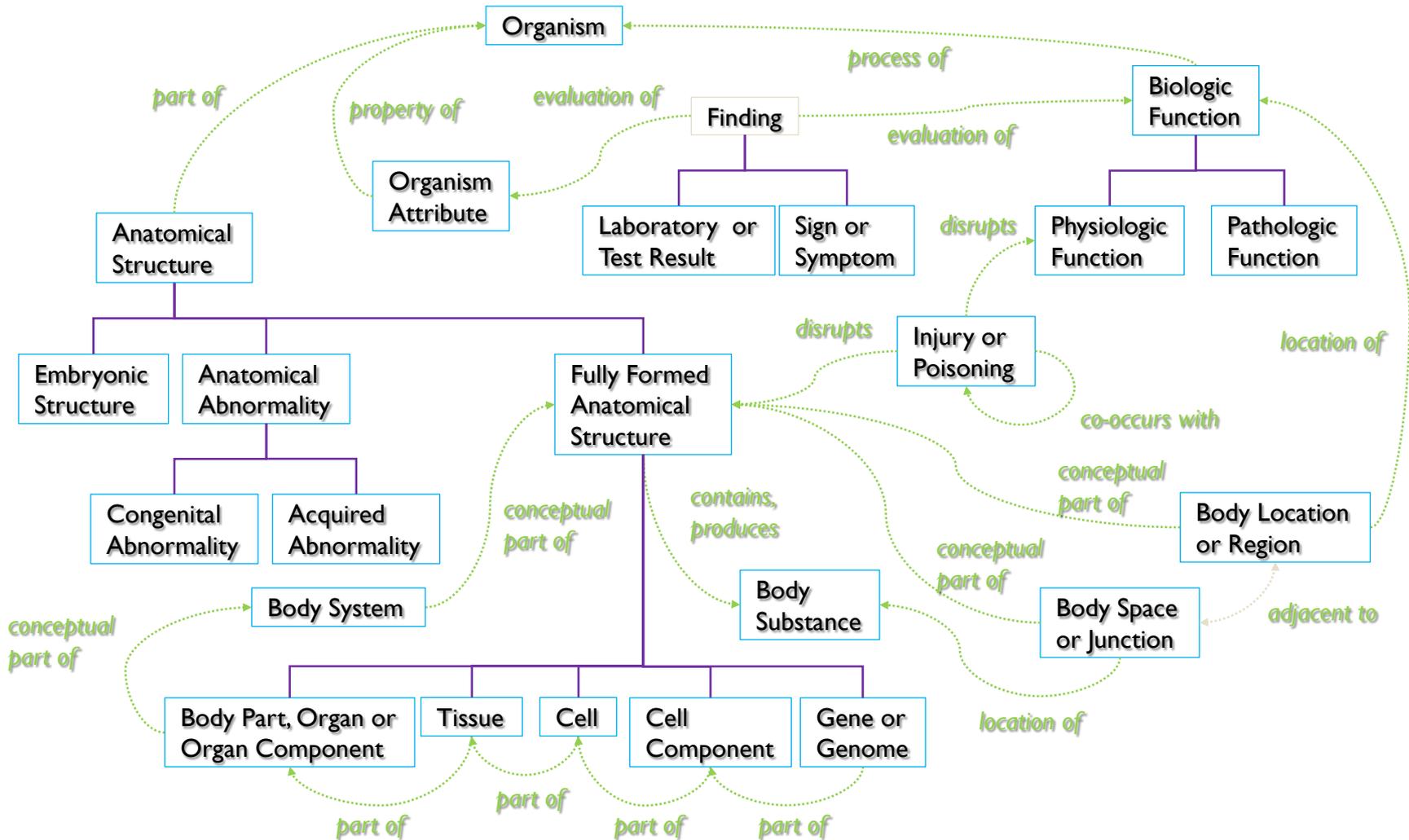
Semantic Network

- Semantic network
 - 54 relationships
 - 603 asserted relations
 - 6101 inferred relations
- Asserted semantic network relations (603)
 - hierarchical (isa = is a kind of)
 - among types (133)
 - **Animal** *isa* **Organism**
 - **Enzyme** *isa* **Biologically Active Substance**
 - among relations (54)
 - *treats isa affects*
 - non-hierarchical (416)
 - **Sign or Symptom** *diagnoses* **Pathologic Function**
 - **Pharmacologic Substance** *treats* **Pathologic Function**

“Biologic Function” hierarchy (isa)



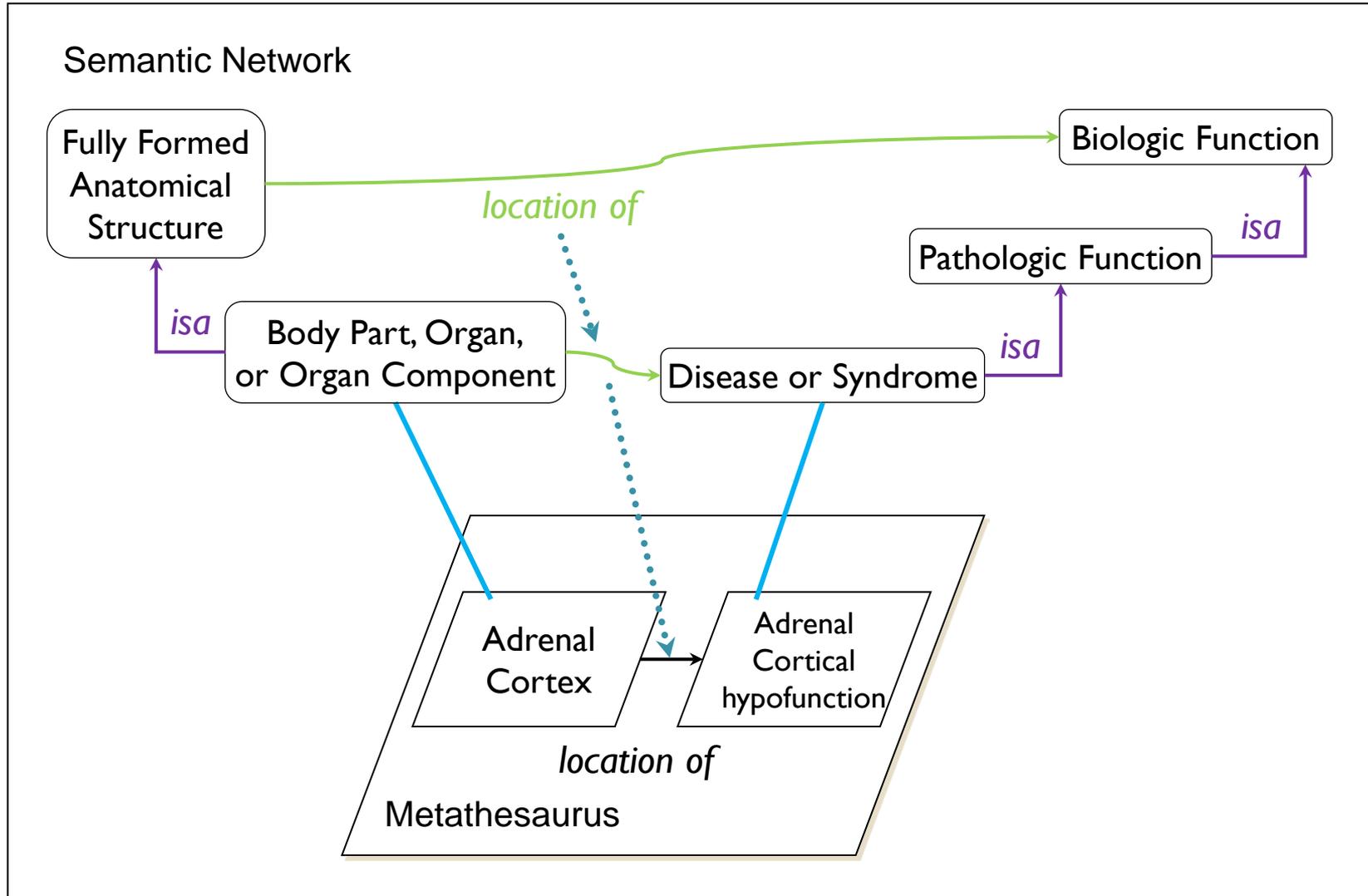
Associative (non-isa) relationships



Why a semantic network?

- Semantic Types serve as high level categories assigned to Metathesaurus concepts, *independently of their position in a hierarchy*
- A relationship between 2 Semantic Types (ST) is a possible link between 2 concepts that have been assigned to those STs
 - The relationship may or may not hold at the concept level
 - Other relationships may apply at the concept level

Relationships *may* inherit semantics



SPECIALIST Lexicon and lexical tools

SPECIALIST Lexicon

- Content
 - English lexicon
 - Many words from the biomedical domain
- 450,000 lexical items
- Word properties
 - morphology
 - orthography
 - syntax
- Used by the lexical tools

Morphology

- Inflection

- Noun nucleus, nuclei
- Verb cauterize, cauterizes, cauterized, cauterizing
- Adjective red, redder, reddest

- Derivation

- Verb-noun cauterize -- cauterization
- Adjective-noun red -- redness

Orthography

- Spelling variants

- oe/e oesophagus - esophagus
- ae/e anaemia - anemia
- ise/ize cauterise - cauterize
- genitive mark Addison's disease
 Addison disease
 Addisons disease

Syntax

- Complementation

- verbs

- intransitive I'll **treat**

- transitive He **treated** **the patient**

- ditransitive He **treated** **the patient** **with a drug**

- nouns

- prepositional phrase

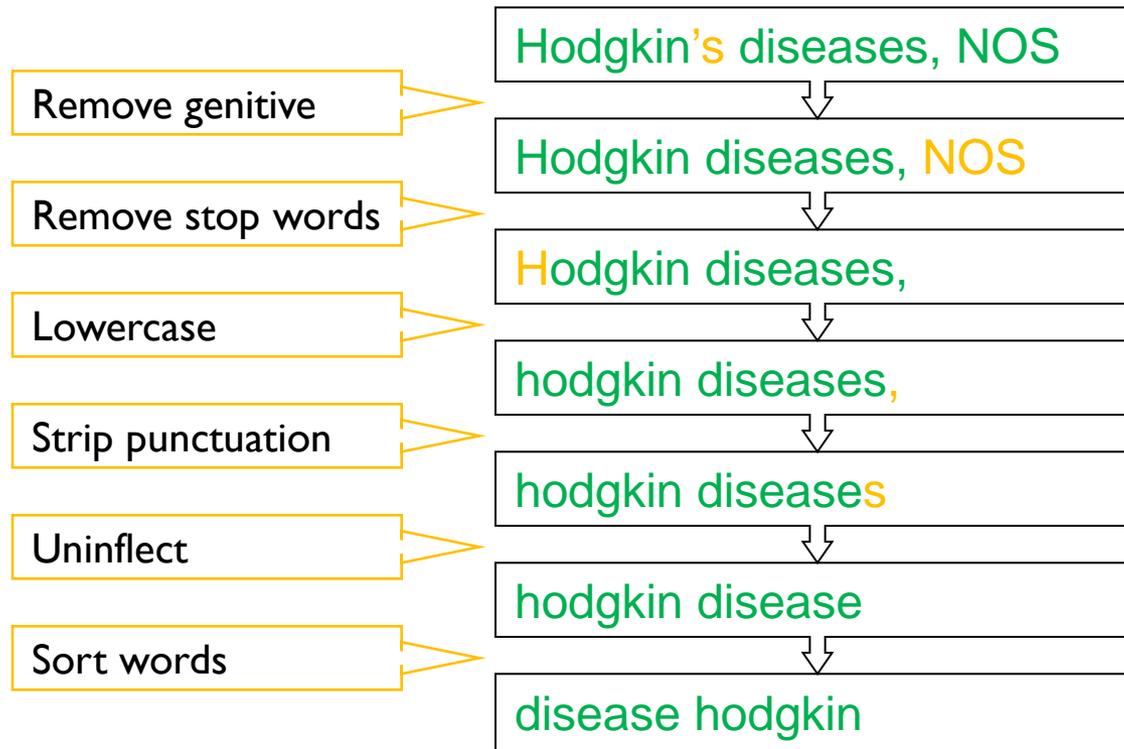
- Valve** **of** **coronary sinus**

- Position for adjectives

Lexical tools

- To manage lexical variation in biomedical terminologies
- Major tools
 - Normalization
 - Indexes
 - Lexical Variant Generation program (lvg)
- Based on the SPECIALIST Lexicon
- Used by noun phrase extractors, search engines

Normalization



Normalization Example

Hodgkin Disease
HODGKINS DISEASE
Hodgkin's Disease
Disease, Hodgkin's
Hodgkin's, disease
HODGKIN'S DISEASE
Hodgkin's disease
Hodgkins Disease
Hodgkin's disease NOS
Hodgkin's disease, NOS
Disease, Hodgkins
Diseases, Hodgkins
Hodgkins Diseases
Hodgkins disease
hodgkin's disease
Disease, Hodgkin

normalize

disease hodgkin

Normalization Applications

- Model for lexical resemblance
- Help find lexical variants for a term
 - Terms that normalize the same usually share the same LUI
- Help find candidates to synonymy among terms
- Help map input terms to UMLS concepts

Indexes

- Word index
 - word to Metathesaurus strings
 - one word index per language
- Normalized word index
 - normalized word to Metathesaurus strings
 - English only
- Normalized string index
 - normalized term to Metathesaurus strings
 - English only

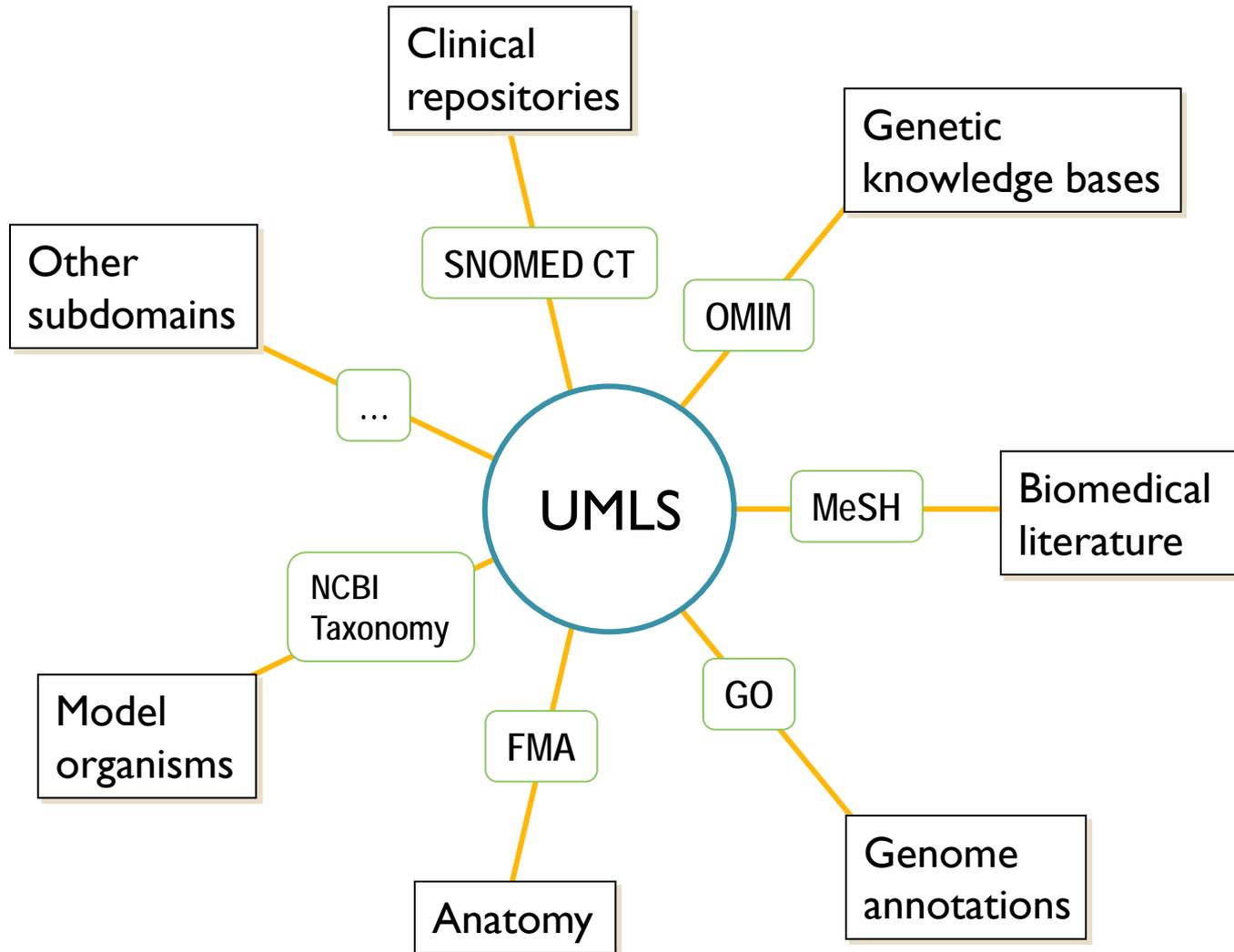
Lexical Variant Generation program

- Tool for specialists (linguists)
- Performs atomic lexical transformations
 - generating inflectional variants
 - lowercase
 - ...
- Performs sequences of atomic transformations
 - a specialized sequence of transformations provides the normalized form of a term (the *norm* program)

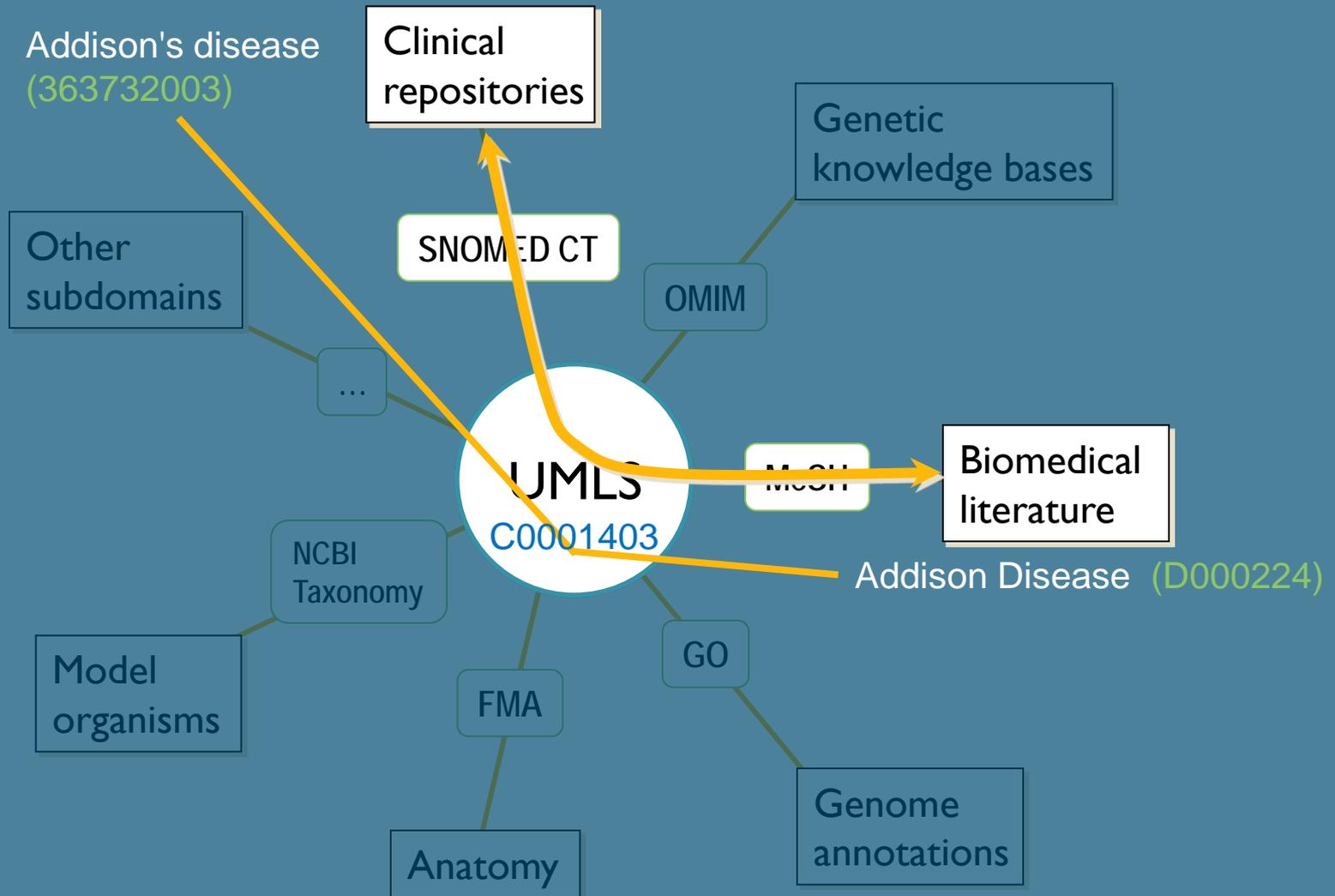
Overview of the UMLS

Conclusions

Integrating subdomains



Trans-namespace integration



Other things you would need to know

- UMLS license agreement
 - <https://uts.nlm.nih.gov/help/license/LicenseAgreement.pdf>
- MetamorphoSys
 - http://www.nlm.nih.gov/research/umls/implementation_resources/metamorphosys/index.html
- UMLS Terminology Services (UTS)
 - <https://uts.nlm.nih.gov/>

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SNOMED CT integration into UMLS

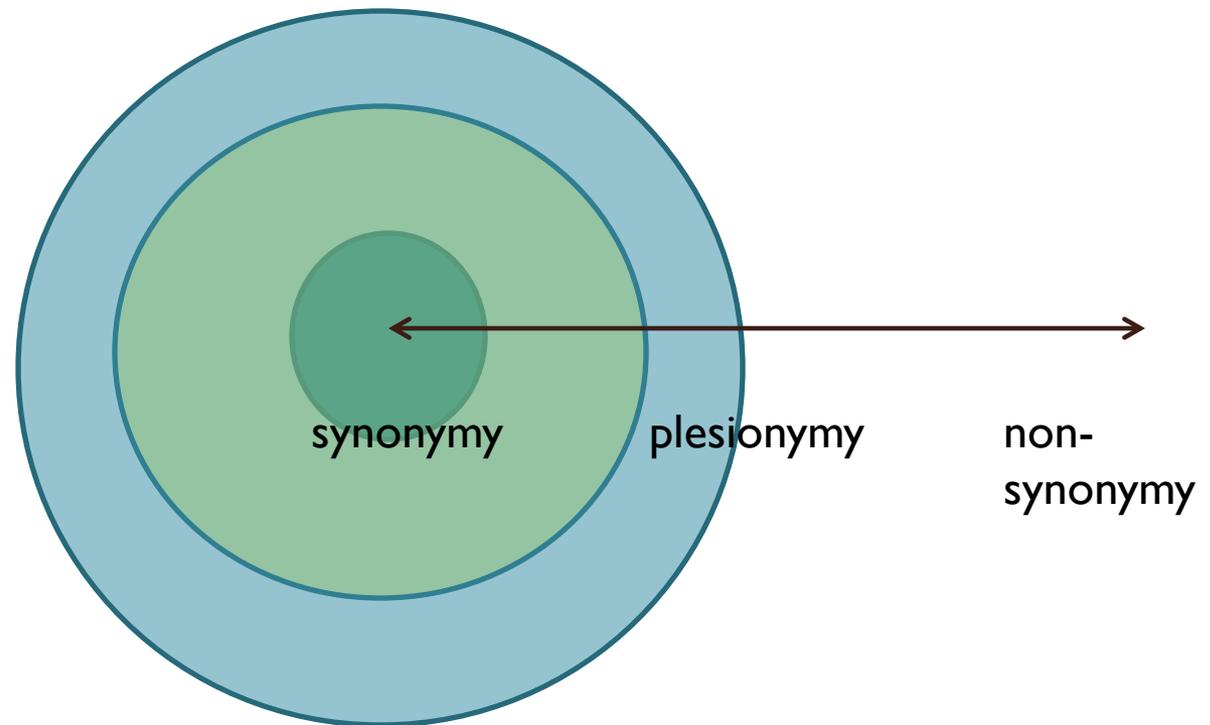


Concept-based terminologies

- SNOMED CT and UMLS are both ‘concept-based’ terminology systems
- Synonymous names (terms) are grouped into the same concept
- Different terminologies have different ‘views of synonymy’
- What constitutes ‘synonymy’?
 - In linguistics - X and Y are synonyms if any sentence S1 containing X is equivalent to another sentence S2, which is identical to S1 except that X is replaced by Y

Different views of synonymy

- In practice, synonymy is a fuzzier notion



Different terminologies have different 'semantic spans' depending on their purposes and organizing principles

Examples of fuzzy synonymy

- Tobacco dependency syndrome vs. tobacco abuse
- Muscle weakness vs. incomplete paralysis
- Malaise vs. ill-defined experience
- Congenital disease vs. fetal developmental abnormality

UMLS editing process

- Analysis of incoming terminology: domain, file structure, concept-oriented or not, hierarchies, term types etc.
- The smallest units (an atom – usually an individual string associated with a code) are inserted in the editing environment
- The atoms are algorithmically assigned into existing or new concepts based on lexical matching with existing terms
- Potential problems (e.g. conflicts) are flagged for review by UMLS editors

SNOMED CT asserted synonyms

- Generally, the assertion of synonymy within a SNOMED CT concept is respected
 - the fully-specified name and preferred name of a SNOMED CT concept always stay together in the same UMLS concept
 - The synonyms may be split out if they conflict with the views of synonymy of other terminologies, depending on the judgment of UMLS editors

Examples of split SNOMED CT synonyms

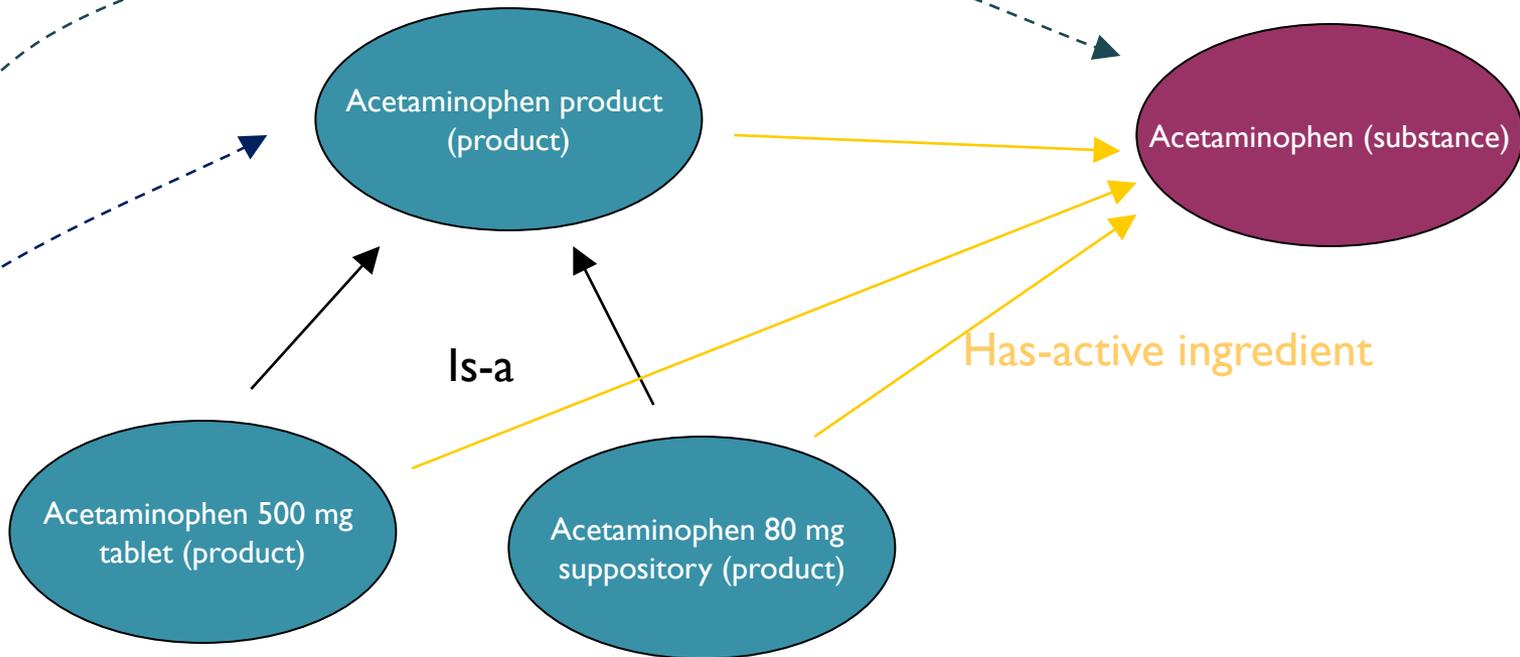
Fully-specified name	Synonym
Adolescence (finding)	Youth
Blinking (finding)	Winking
Candidiasis (disorder)	Thrush
Motor vehicle accident victim (finding)	Motor vehicle accident
Eczema (disorder)	Dermatitis

- In SNOMED CT, the assertion of synonyms tend to be more accommodating, based on common usage in clinical exchange rather than strict meaning
- However, failure to distinguish between the different meanings in the UMLS may cause problems with some applications e.g. natural language processing

Merging of SNOMED CT concepts

- SNOMED CT sometimes make ontological distinction between classes of concepts which are created to support Description Logic
- The UMLS does not necessarily distinguish between SNOMED CT hierarchies, if the meanings of the concepts are not usually distinct in the clinical setting
- This is necessary to prevent ‘unnecessary’ proliferation of concepts in the UMLS and to facilitate data integration

SNOMED CT Concept View

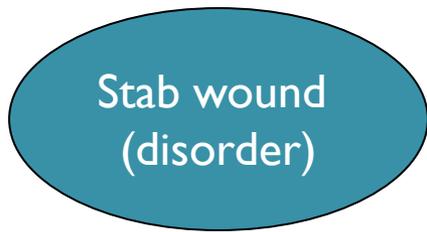


'This patient is on acetaminophen.'

'Tylenol contains acetaminophen.'

~~Common clinical distinction?~~

SNOMED CT Concept View



Has-associated-
morphology



‘The patient was admitted for a stab wound sustained during a fight.’

‘On physical examination, there was a 3 cm stab wound in the right upper quadrant of the abdomen.’



Common types of cross-hierarchy merging

- Product and substance
 - Acetaminophen (substance), acetaminophen product (product)
- Disorder and morphologic abnormality
 - Stab wound (disorder), stab wound (morphologic abnormality)
- Observable entity and finding
 - Body temperature (observable entity), Body temperature finding (finding)
- Qualifier value and procedure
 - Drainage - action (qualifier value), Drainage procedure (procedure)

Within hierarchy merging

- Product
 - Vitamin B₁₂ preparation (product), Cyanocobalamin preparation (product)
 - Gentamicin 60mg/mL injection solution 2mL ampule (product), Gentamicin 60mg/mL injection solution 1mL ampule (product)
- Disorder/Finding
 - Bleeding from nose (finding), Epistaxis (disorder)
- Procedure
 - Abdominal paracentesis (procedure), Percutaneous drainage of ascites (procedure)

Extent of disagreement

- Splitting of synonyms
 - 2,702 (1% of current) concepts and 3,958 (3% of synonyms)
- Merging of concepts
 - 20811 (7% of current) concepts
 - Within hierarchy merges > across hierarchy merges
- Trend of less disagreement over time (14% concepts in 2004)

UMLS- and SNOMED CT- centric views

- Even though the views of synonymy between SNOMED CT and UMLS sometimes differ, both views are preserved in the UMLS
 - If you want to see the UMLS view – organize things by their CUI (concept unique identifier)
 - If you want to see the SNOMED CT view – organize things by their SCUI (source concept unique identifier)

Dr. Kin Wah Fung

Representing SNOMED CT in the UMLS domain model

Source transparency principle: *It's all there!*

- There is no loss of information in the process of insertion of a source terminology. Every element of information contained in a source is included in the release files, even though they may be organized differently
- Proof of source transparency in insertion of SNOMED CT:
 - source-derived files extracted from the original SNOMED CT files
 - UMLS-derived files extracted from the UMLS release files in the same format as the source-derived files
 - row-by-row comparison proved that they are identical

SNOMED CT is more complex than other terminologies

- **Concept-oriented:**
 - each concept contains multiple descriptions (concept names)
 - concepts and descriptions have their own unique identifiers and sets of attributes (e.g. `ConceptStatus`, `DescriptionType`)
- **Relationships:**
 - defined at the concept level
 - relationships have their own unique identifiers and set of attributes (e.g. `RelationshipGroup`)
- **Mappings to ICD9CM:**
 - in three linked tables (`Cross Map Sets`, `Cross Maps`, `Cross Map Target`)
 - different sets of attributes at each level
- The original release format of the UMLS Metathesaurus cannot represent all the above information

Original Release Format (ORF) of UMLS Metathesaurus

- Represents information by the **Metathesaurus-Concept-Centric view**
- Main function: as a bridge to bring together various biomedical vocabularies with explicit concept-based connection between terms in one vocabulary and equivalent or related terms in another
- A degree of abstraction and simplification – most of the information is represented at the Concept (CUI) level
- In order to accommodate the level of granularity of SNOMED CT, a new format of the release files is required

Rich Release Format (RRF)

- Also supports a new **Source-Centric View** that allows users to retrieve the original information contained in a source vocabulary (source transparency)
- Information is represented at the Atom (AUI) level – an atom is a unit of meaning (usually a term identified by a code) in a source
- New fields to capture source identifiers for various entities (e.g. SCUI, SAUI, SRUI)
- New files to capture new information and provide new functionalities e.g. MRMAP, MRHIER, MRDOC

Representation of SNOMED CT data elements in the UMLS

- Information contained in every field of SNOMED CT tables is represented in the UMLS Metathesaurus
- For the 3 main SNOMED CT tables: Concepts, Descriptions and Relationships; corresponding fields can be found in 3 RRF files: MRCONSO, MRSAT and MRREL in UMLS (some fields will not be available in ORF files)
- Detailed documentation and sample SQL statements available at:
 - http://www.nlm.nih.gov/research/umls/Snomed/snomed_represented.html

SNOMED CT Concepts

SNOMED CT Concepts Table:

ConceptId|ConceptStatus|FullySpecifiedName|CTV3ID|SNOMEDID|IsPrimitive|
27|737000|0|Anemia (disorder)|XM05A|DC-10009||

MRCONSO.RRF:

CUI|...|AUI|...|SCUI|...|SAB|TTY|...|STR|...|
C000287|A3597593|27|737000|SNOMEDCT|FN|Anemia (disorder)|
C000287|A2878480|27|737000|SNOMEDCT|PT|Anemia|
C000287|A2952250|27|737000|SNOMEDCT|SY|Absolute anemia|
C000287|A3095181|27|737000|SNOMEDCT|PTGB|Anaemia|
C000287|A3089808|27|737000|SNOMEDCT|SYGB|Absolute anaemia|

MRSAT.RRF:

CUI|...|STYPE|CODE|...|ATN|SAB|ATV|...|
C000287|SCUI|27|737000|CONCEPTSTATUS|SNOMEDCT|0|
C000287|SCUI|27|737000|CTV3ID|SNOMEDCT|XM05A|
C000287|SCUI|27|737000|SNOMEDID|SNOMEDCT|DC-10009|
C000287|SCUI|27|737000|ISPRIMITIVE|SNOMEDCT||

SNOMED CT Descriptions

SNOMED CT Descriptions Table:

DescriptionId|DescriptionStatus|ConceptId|Term|InitialCapitalStatus|DescriptionType|
LanguageCode|

4066360|3|0|27|737000|Anemia|0||en-US|

MRCONSO.RRF:

CUI|...|AUI|SAUI|SCUI|...|SAB|TTY|...|STR|...|

C000287|1|A2878480|4066360|3|27|737000|SNOMEDCT|PT|Anemia|

MRSAT.RRF:

CUI|...|METAUI|STYPE|CODE|...|ATN|SAB|ATV|...|

C000287|1|A2878480|SAUI|27|737000|DESCRIPTIONSTATUS|SNOMEDCT|0|

C000287|1|A2878480|SAUI|27|737000|INITIALCAPITALSTATUS|SNOMEDCT|0|

C000287|1|A2878480|SAUI|27|737000|DESCRIPTIONTYPE|SNOMEDCT|1|

C000287|1|A2878480|SAUI|27|737000|LANGUAGECODE|SNOMEDCT|en-US|



SNOMED CT Relationships

SNOMED CT Relationships Table

RelationshipId|ConceptId1|RelationshipType|ConceptId2|CharacteristicType|Refinability|RelationshipGroup|

85555020|263245004|I|16680003|15574005|0|0|0|

MRCONSO

CUI|...|AUI|SCUI|...|SAB|TTY|...|STR|...|

C0281851|A3134233|263245004|SNOMEDCT|PT|Fracture of tarsal bone|

C1292718|A3524752|16680003|SNOMEDCT|PT|Is a|

C0272774|A3467699|15574005|SNOMEDCT|PT|Fracture of foot|

MRREL

|AUI1|STYPE1|REL|...|AUI2|STYPE2|RELA|RUI|SRUI|SAB|...|RG|DIR|...|

|A3467699|SCUI|CHD|A3134233|SCUI|isa|R20132044|85555020|SNOMEDCT|0|Y|

|A3134233|SCUI|PAR|A3467699|SCUI|inverse_isa|R20453736|85555020|SNOMEDCT|0|N|

MRSAT

|METAUI|STYPE|...|ATN|SAB|ATV|...|

|R20132044|RUI|CHARACTERICTYPE|SNOMEDCT|0|

|R20132044|RUI|REFINABILITY|SNOMEDCT|0|

New SNOMED CT release format

- RF2 (Release Format 2) released since July 2011
- Three kinds of releases: Full, Snapshot, Delta releases
- New files (e.g. definition refset) and data fields (e.g. effectiveTime, moduleId)
- History tracking mechanism
- We're working on the best way to represent RF2 data in the UMLS

Dr. Olivier Bodenreider

Applying UMLS lexical tools to SNOMED CT descriptions

Normalization

- Model of lexical resemblance
 - Linguistically-motivated
 - Abstracts away from minor lexical variation
 - Tuned for clinical terminology
 - Can be adapted to specialized vocabularies (e.g., names of clinical drugs)
- Applications
 - Mapping across terminologies
 - Identify candidates for synonymy
 - Search beyond exact matches

Beta-blocker in SNOMED CT

The screenshot displays the SNOMED CT browser interface for the concept 'beta-Adrenergic blocking agent' (Concept Id: 33252009, Description Id: 55491010). The interface is divided into several sections:

- Header:** Shows the Concept Id (33252009) and Description Id (55491010). The concept name is 'beta-Adrenergic blocking agent' with a type of 'pharmaceutical / biologic product'.
- Search:** A search bar contains 'beta adrenergic blocking agent', and a list of search results is shown below it, with 'beta-Adrenergic blocking agent' selected.
- Properties:** A toolbar with various icons and a dropdown menu set to 'Words - any order' with a value of 7.
- Concept Status:** Indicated as 'current'.
- Descriptions:** A list of descriptions in English (en-US), including 'beta-Blocking agent (product)', 'beta-Blocking agent', 'beta-blocker', 'beta-Adrenergic blocking agent' (highlighted), and 'beta-adrenoceptor blocking drug'.
- Definition:** Labeled as 'Primitive', with a definition 'is a hypotensive agent'.
- Codes:** Lists the 'Original SNOMED Id' as C-80135 and the 'Read Code (Ctv3Id)' as Xa2E9.
- Hierarchy:** Set to 'Subtype hierarchy', showing a tree structure starting with 'hypotensive agent' and 'beta-Blocking agent' (highlighted) as its child. Other children include 'Beta 1 blocking agent' and 'non-selective beta-blocking agent'.

Normalization

Agents, beta-Adrenergic Blocking
beta-adrenergic blocking agent
beta-Adrenergic blocking agent
Beta-Adrenergic Blocking Agent
beta-Adrenergic blocking agent, NOS
beta adrenergic blocking agents
beta-adrenergic blocking agents
beta Adrenergic Blocking Agents
beta-Adrenergic Blocking Agents
Beta-adrenergic Blocking Agents
Blocking Agents, beta-Adrenergic



adrenergic agent beta blocking

Finding variants in other sources

Source	Synonym
CHV	beta-adrenergic blocking agents
CHV	beta adrenergic blocking agents
CHV	beta-adrenergic blocking agent
MEDCIN	beta adrenergic blocking agents
MeSH	beta-Adrenergic Blocking Agents
MeSH	beta Adrenergic Blocking Agents
MeSH	Agents, beta-Adrenergic Blocking
MeSH	Blocking Agents, beta-Adrenergic
NCI Thesaurus	Beta-Adrenergic Blocking Agent
SNOMED Intl	beta-Adrenergic blocking agent, NOS
<i>SNOMED CT</i>	<i>beta-Adrenergic blocking agent</i>
USPMG	Beta-adrenergic Blocking Agents
USPMG	Beta-adrenergic Blocking Agents

Retrieving similar variants

beta-Adrenergic Blocking Agents



adrenergic agent beta blocking



Agents, beta-Adrenergic Blocking
beta-adrenergic blocking agent
beta-Adrenergic blocking agent
Beta-Adrenergic Blocking Agent
beta-Adrenergic blocking agent, NOS
beta adrenergic blocking agents
beta-adrenergic blocking agents
beta Adrenergic Blocking Agents
beta-Adrenergic Blocking Agents
Beta-adrenergic Blocking Agents
Blocking Agents, beta-Adrenergic

Ms. Janice H. Willis

Applying UMLS quality assurance processes to SNOMED CT content



SNOMED CT QA

What we learn from maintaining the UMLS

- Analyze each SNOMED CT International release
 - Convert SNOMED CT native → UMLS format
 - Critical use of files, documentation
 - Build on previous experience and specifications
- Add converted data to Metathesaurus
 - Automated QA generates stats, comparisons (before/after, and new/previous releases)
 - Tooling assists review searching, browsing, sampling
- Editing
 - Editors ensure synonymy and assign Semantic Types
 - Editors note other issues

SNOMED CT QA

What we learn from maintaining the UMLS 2

- UMLS Release QA - 2nd round of review, counts, comparisons
 - Individual sources (SNOMED CT) and all sources integrated into Metathesaurus concept structure
- Update Web-based documentation for each UMLS terminology
 - Stats, sample data, conversion specs and details of native and UMLS format
- User feedback (customer service, LISTSERV)
- Recent example:
 - Error in SNOMED CT-ICD-9-CM Cross Map 2012

Ms. Janice H. Willis

Browsing SNOMED CT with THE UTS browser



UTS browser

- <https://uts.nlm.nih.gov/>





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The UMLS Terminology Services (UTS) allows you to:

- Request a UMLS Metathesaurus License
- Search and display content from UTS Applications
 - Metathesaurus Browser
 - Semantic Network Browser
 - SNOMED CT Browser
- Download data files including:
 - UMLS Knowledge Sources
 - RxNorm weekly and monthly updates
 - SNOMED CT
 - CORE Problem List and Route of Action
 - SNOMED CT
- Query data remotely via Web Services (see [Web Services](#))
- Complete UMLS Annual Report and SNOMED CT

UMLS Terminology Services (UTS) provide both as Web Services to search and retrieve UMLS

We welcome you to [contact us](#) with your comments to improve the UTS.

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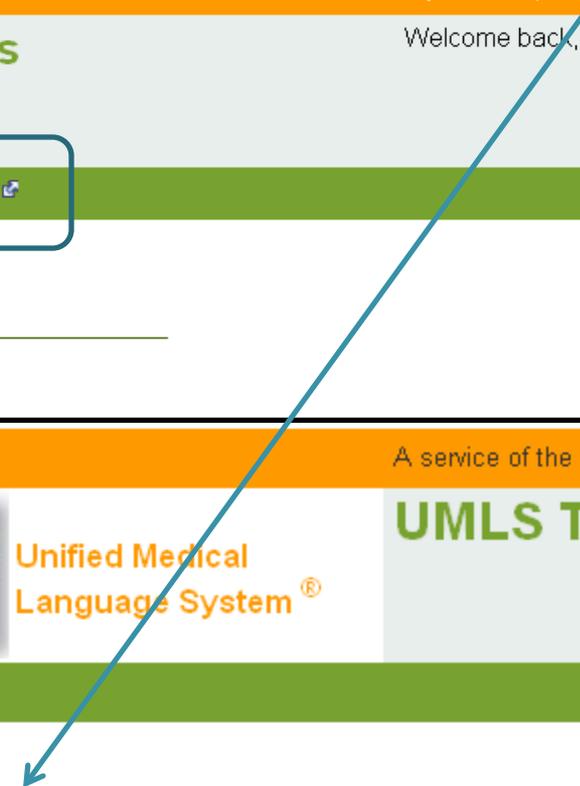
Username:

Password:

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- News/Announcements
- Training

- SNOMED CT at NLM
- International Release
- US Extension to SNOMED CT
- Convergent Medical Terminology
- SNOMED CT Browser
- Subsets

UTS) allows you to:

- license and create a UITS account
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- Route of Administration Subset
- Nursing Problem List Subset

- Download data files including:
 - UMLS Knowledge Sources
 - RxNorm weekly and monthly updates
 - SNOMED CT
 - CORE Problem List and Route of Administration Subsets of SNOMED CT
- Query data remotely via Web Services (see API Documentation)
- Complete UMLS Annual Report and SNOMED CT® Affiliate Reports

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Search | Tree | Recent Searches | Basic View

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Term CUI Code

Go

Release: 2011AA

Search Type: Word

Source: All Sources, AIR, ALT, AOD, AOT

Search | Tree | Recent Searches

Term CUI Code

addison's disease | Go

Release: 2011AA

Search Type: Word

Source: All Sources, AIR, ALT, AOD, AOT

Search options include limiting by one or more sources

Search Tree Recent Searches

Term CUI Code

Release:

Search Type:

Source:

- AIR
- ALT
- AOD
- AOT

Search Results (1)
[C0001403](#) Addison Disease

3 report views – basic, report (detailed) and raw Metathesaurus data

Basic View Report View Raw View

+ Concept: [C0001403] Addison Disease

- Semantic Types
[Disease or Syndrome](#) [T047]

+ Definitions

- Atoms (231) string [AUI / RSAB / TTY / Code]

- + Addison's disease [A0388276/AOD/DE/0000006012]
- + ADDISON DISEASE [A0385542/CCPSS/PT/0022753]
- + addison disease [A18626845/CHV/SY/0000000703]
- + addison's disease [A18682447/CHV/PT/0000000703]
- + primary adrenal insufficiency [A18645441/CHV/SY/0000000703]
- ADRENAL INSUFFICIENCY (ADDISON'S DISEASE) [A0385630/COSTAR/PT/U0000]
- + Addison's disease [A0388277/CSP/PT/0060-3321]
- + ADDISON'S DISEASE [A0385544/CST/GT/ADREN INSUFFIC]
- + DISEASE ADDISON'S [A0404749/CST/GT/ADREN INSUFFIC]
- Primaere Nebennierenrindeninsuffizienz [A1480496/DMDICD10/PT/E27.1]
- ADDISON DISEASE [A0385543/DXP/SY/NOCODE]
- ADRENOCORTICAL INSUFFICIENCY, PRIMARY FAILURE [A0385641/DXP/SY/NOC]
- + Primary adrenocortical insufficiency [A0778564/ICD10/PT/E27.1]
- + Primary adrenocortical insufficiency [A0778565/ICD10AM/PT/E27.1]
- + Addison's disease [A17799651/ICD10CM/ET/E27.1]
- + Primary adrenocortical insufficiency [A17786892/ICD10CM/PT/E27.1]

Search Tree Recent Searches

Release: 2011AA

- ⊕ (SNM) Systematized Nomenclature of Medicine. 2nd ed
- ⊕ (SNMI) SNOMED International
- ⊖ (SNOMEDCT) SNOMED CT Concept
 - ⊕ Body structure
 - ⊖ Clinical finding
 - ⊕ Administrative statuses
- ⊕ Adverse incident outcome categories
- ⊕ Bleeding
- ⊕ Calculus finding
- ⊕ Clinical history a
- ⊕ Clinical stage fir
- ⊕ Cyanosis
- ⊕ Deformity
- ⊖ Disease
 - ⊕ AIDS-associated disorder
 - ⊕ Acute disease
 - ⊕ Angioedema and/or urticaria
 - Biphasic disease
 - ⊕ Chromosomal disorder
 - ⊕ Chronic disease
 - ⊕ Communication disorder
 - ⊕ Complication
 - ⊕ Congenital disease
 - ⊕ Cyst
 - ⊕ Degenerative disorder
 - ⊕ Developmental disorder
 - ⊕ Diabetic complication

Metathesaurus Browser Tree search to expand sources organized by hierarchies

Basic View Report View Raw View

- ⊕ **Concept: [C0008679] Chronic disease**
- ⊕ **Semantic Types**
- ⊕ **Definitions**
- ⊖ **Atoms (67)** string [AUI / RSAB / TTY / Code]
 - ⊕ chronic disease [A0476171/AOD/DE/0000004195]
 - ⊕ chronic disease [A18646393/CHV/PT/0000002973]
 - ⊕ chronic diseases [A18627734/CHV/SY/0000002973]
 - ⊕ chronic disorder [A18646394/CHV/SY/0000002973]
 - ⊕ chronic disorders [A18627735/CHV/SY/0000002973]
 - ⊕ chronic illness [A18683389/CHV/SY/0000002973]
 - ⊕ chronic illnesses [A18571921/CHV/SY/0000002973]
 - ⊕ chronic disease [A0476172/CSP/ET/0944-4942]
 - ⊕ chronic disease/disorder [A1883108/CSP/PT/0944-4942]
 - ⊕ chronic disorder [A1304788/CSP/ET/0944-4942]
 - Chronic diseases [A0015542/LCH/PT/U001015]
 - ⊕ **CHRONIC DIS [A12079202/MSH/DEV/D002908]**
 - ⊕ Chronic Disease [A0039239/MSH/MH/D002908]
 - ⊕ Chronic Diseases [A0401105/MSH/PM/D002908]
 - ⊕ Chronic Illness [A0039250/MSH/EP/D002908]
 - ⊕ Chronic Illnesses [A0039252/MSH/PM/D002908]
 - ⊕ Disease, Chronic [A0049681/MSH/PM/D002908]
 - ⊕ Diseases, Chronic [A0049961/MSH/PM/D002908]
 - ⊕ Illness, Chronic [A0072124/MSH/PM/D002908]
 - ⊕ Illnesses, Chronic [A0072127/MSH/PM/D002908]
 - ⊕ chronická nemoc [A13045053/MSHCZE/MH/D002908]
 - Chronische ziekte [A8185237/MSHDUT/MH/D002908]
 - Ziekte, chronische [A8212634/MSHDUT/MH/D002908]



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SNOMED CT Browser

Welcome back, testuser

Search Tree Recent Searches

Report View

SNOMED CT Version: 2011_01_31

Term ConceptID DescriptionID

addison

Active concepts only:

Restrict results to:

- Search Results (15)**
- [363732003](#) Addison's disease (disorder)
 - [84027009](#) Pernicious anemia (disorder)
 - [201048007](#) Localized morphea (disorder)
 - [186118888](#) Addison melanoderma (disorder)
 - [83728000](#) Polyglandular autoimmune syndrome, type 2
 - [34253008](#) Myopathy in Addison's disease (disorder)
 - [237780008](#) Addison's disease with adrenoleucodystrophy
 - [186270000](#) Tuberculous Addison's disease (disorder)
 - [11244009](#) Polyglandular autoimmune syndrome, type 1
 - [65389002](#) Adrenoleukodystrophy (disorder)
 - [91003006](#) Salt-losing nephropathy (disorder)
 - [76715008](#) Addison's disease due to autoimmunity (disor
 - [37495007](#) Familial adrenocortical hypoplasia (disorder)
 - [12427005](#) Congenital primary adrenocortical hypofunctio
 - [403252006](#) Buccal pigmentation due to Addison's disease

NLM SNOMED CT Browser

- Search against entire Metathesaurus
- Display SNOMED CT content in SNOMED CT concept format

SNOMED CT-centric display
 Click CUI to display
 Metathesaurus Browser view
 of concept.

Search Tree Recent Searches

SNOMED CT Version: 2011_01_31

Term ConceptID DescriptionID

addisons disease

Active concepts only:
 Restrict results to:

- Search Results (2)**
- [373662000](#) Primary adrenocortical insufficiency (disorder)
 - [111562000](#) Addison's disease [Ambiguous]

Report View

Concept: [373662000] Primary adrenocortical insufficiency (disorder)

UMLS information
 CUI: [\[C0001403\]](#) Addison Disease
 Semantic Types: [Disease or Syndrome](#) [T047]

ConceptStatus	IsPrimitive	SnomedId	CTV3Id
Current (0)	1	DB-70608	XUWM8

Descriptions (3)

Id	Description	Type	Status
1198962018	Primary adrenocortical insufficiency (disorder)	FullySpecifiedName (3)	Current (0)
1212124016	Primary adrenocortical insufficiency	Preferred (1)	Current (0)
1490869013	Primary hypoadrenalism	Synonym (2)	Current (0)

Parents (1)

Adrenal cortical hypofunction [\[386584007\]](#)

Relationships from *this* concept (5)

- Primary adrenocortical insufficiency | [Is a](#) | [Adrenal cortical hypofunction](#) (Defining)
- Primary adrenocortical insufficiency | [Finding site](#) | [Adrenal cortex structure](#) (Defining)
- Primary adrenocortical insufficiency | [Clinical course](#) | [Courses](#) (Qualifier)
- Primary adrenocortical insufficiency | [Episodicity](#) | [Episodicities](#) (Qualifier)
- Primary adrenocortical insufficiency | [Severity](#) | [Severities](#) (Qualifier)

Relationships to *this* concept (3)

- [Adrenal cortical hypofunction](#) | [MAY BE A](#) | Primary adrenocortical insufficiency (Historical)
- [Corticoadrenal insufficiency \(& Addison's \[disease\] or \[crisis\]\)](#) | [MAY BE A](#) | Primary adrenocortical insufficiency (Historical)
- [Corticoadrenal insufficiency \(& Addison's \[disease\] or \[crisis\]\)](#) | [MAY BE A](#) | Primary adrenocortical insufficiency (Historical)

Tree Positions (20)

- Primary adrenocortical insufficiency [Context 1]
- Primary adrenocortical insufficiency [Context 2]
- Primary adrenocortical insufficiency [Context 3]



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SNOMED CT Browser

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Full SNOMED CT concept information

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Search

Tree

Recent Searches

SNOMED CT Version: 2011_01_31

Term ConceptID DescriptionID

addison

Go

Active concepts only:

Restrict results to:

–None–

Search Results (15)

- [363732003](#) Addison's disease (disorder)
- [84027009](#) Pernicious anemia (disorder)
- [201048007](#) Localized morphea (disorder)
- [186118006](#) Addison melanoderma (disorder)
- [83728000](#) Polyglandular autoimmune syndrome, type 2
- [34253008](#) Myopathy in Addison's disease (disorder)
- [237760008](#) Addison's disease with adrenoleucodystrophy
- [186270000](#) Tuberculous Addison's disease (disorder)
- [11244009](#) Polyglandular autoimmune syndrome, type 1
- [65389002](#) Adrenoleucodystrophy (disorder)
- [91003006](#) Salt-losing nephropathy (disorder)
- [78715008](#) Addison's disease due to autoimmunity (disorder)
- [37495007](#) Familial adrenocortical hypoplasia (disorder)
- [12427005](#) Congenital primary adrenocortical hypofunction
- [403252006](#) Buccal pigmentation due to Addison's disease

Report View

Tuberculous Addison's disease [[186270000](#)]

Relationships from *this* concept (5)

- Addison's disease | [Is a](#) | [Adrenal cortical hypofunction](#) (Defining)
- Addison's disease | [Finding site](#) | [Adrenal cortex structure](#) (Defining)
- Addison's disease | [Clinical course](#) | [Courses](#) (Qualifier)
- Addison's disease | [Episodicity](#) | [Episodicities](#) (Qualifier)
- Addison's disease | [Severity](#) | [Severities](#) (Qualifier)

Relationships to *this* concept (9)

- [Addison's disease due to autoimmunity](#) | [Is a](#) | Addison's disease (Defining)
- [Addison's disease with adrenoleucodystrophy](#) | [Is a](#) | Addison's disease (Defining)
- [Polyglandular autoimmune syndrome, type 1](#) | [Is a](#) | Addison's disease (Defining)
- [Tuberculous Addison's disease](#) | [Is a](#) | Addison's disease (Defining)
- [Addison melanoderma](#) | [Due to](#) | Addison's disease (Defining)
- [Buccal pigmentation due to Addison's disease](#) | [Due to](#) | Addison's disease (Defining)
- [Addison's disease](#) | [MAY BE A](#) | Addison's disease (Historical)
- [Corticoadrenal insufficiency \(& Addison's \[disease\] or \[crisis\]\)](#) | [MAY BE A](#) | Addison's disease (Historical)
- [Corticoadrenal insufficiency \(& Addison's \[disease\] or \[crisis\]\)](#) | [MAY BE A](#) | Addison's disease (Historical)

Tree Positions (20)

- Addison's disease [Context: 1]
 - SNOMED CT Concept
 - Clinical finding
 - Disease
 - Disorder by body site
 - Disorder of body system
 - Disorder of endocrine system
 - Disorder of adrenal gland
 - Hypoadrenalism
 - Adrenal hypofunction
 - Adrenal cortical hypofunction

Search Tree Recent Searches

- [-] SNOMED CT Concept
 - [-] Body structure
 - [-] Anatomical or acquired body structure
 - [+] Acquired body structure
 - [-] Anatomical structure
 - [-] Body organ structure
 - [-] Blood vessel structure
 - [-] Arterial structure
 - [-] Arterial part
 - [-] Aorta part
 - Aortic tunica adventitia
 - [+] Aortic tunica intima structure
 - Aortic tunica media
 - [+] Descending aorta structure
 - [+] Thoracic aorta structure
 - [+] Coronary artery part
 - External elastic membrane of arte
 - [+] Structure of anulus fibrosus of aor
 - [+] Structure of anulus fibrosus of pul
 - [+] Structure of carotid sinus
 - [+] Structure of cavernous portion of
 - [+] Structure of cerebral portion of int
 - [+] Structure of cervical portion of inte
 - [+] Structure of petrous portion of inte
 - [+] Suprapulmonic valve area structu
 - Tunica adventitia of artery
 - [+] Tunica intima of artery
 - [+] Tunica media vasorum

Metathesaurus Browser Tree search display. Click to expand the nodes in the hierarchies.

UMLS Source documentation

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 - Metathesaurus Browser
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 - SNOMED CT
 - CORE Problem List and Route of Administration Subsets of SNOMED CT
- Query data remotely via Web Services (see API Documentation)
- Complete UMLS Annual Report and SNOMED CT® Affiliate Reports

UMLS Terminology Services (UTS) provide both web interfaces as well as Web Services to search and retrieve UMLS data.

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General ▶

Technical ▶

- [UMLS Reference Manual](#)
- [UMLS Source Documentation](#)
- [RxNorm](#)
- [SNOMED CT](#)
- [US Extension to SNOMED CT](#)



2011AA SNOMED CT Source Information

Web-based documentation
for each source in the UMLS
Metathesaurus

Synopsis MRSAB.RRF **Statistics and Sample Data** Representation
Terms and term types Attributes Relationships Semantic Types Source Overlap

Notes:

- These reports provide sample data and statistics for new and updated sources in the UMLS Metathesaurus. They are designed to help users better understand the content of individual sources, and how these sources are represented in the Metathesaurus.
- These reports may also be useful to help users to create customized subsets when using MetamorphoSys to:
 - Select appropriate sources
 - Apply term type, relationship, and attribute filters, and
 - Enable options on the Output, Suppressibility, and Precedence tabs.
- The counts and data in customized subsets may differ from the default counts and sample data displayed in these reports.
- Sample data are extracted from Metathesaurus files; complete rows are not displayed.
- Send comments and suggestions about the Source Release Documentation to [NLM Customer Service](#).



2011AA SNOMED CT Source Information

- Synopsis
- MRSAB.RRF
- Statistics and Sample Data
- Representation
- Terms and term types
- Attributes
- Relationships
- Semantic Types
- Source Overlap

Counts (skip to: [notes](#) [samples](#))

| Term Type | Description | Count (MRCONSO.RRF) |
|----------------------|-------------------------------|---------------------|
| FN | Full form of descriptor | 293768 |
| PT | Designated preferred name | 293768 |
| OF | Obsolete fully specified name | 207950 |
| SY | Designated synonym | 149107 |
| OP | Obsolete preferred term | 105880 |
| IS | Obsolete Synonym | 88053 |
| PTGB | British preferred term | 20054 |
| SYGB | British synonym | 8462 |

- [MTH FN](#)
- [MTH PT](#)
- [MTH SY](#)
- [MTH IS](#)
- [MTH OF](#)
- [MTH OP](#)
- [MTH PT](#)

[see more samples](#) (Click and hold to drag this window)

Sample Data (FN)

| CUI | AUI | LUI | SUI | SAUI | SCUI | CODE | STR |
|----------|----------|----------|----------|-----------|----------|----------|---|
| C0269048 | A3562033 | L3023011 | S3415273 | 742167018 | 15363000 | 15363000 | Metritis (disorder) |
| C0273049 | A3327522 | L2919310 | S3197039 | 729778010 | 12589008 | 12589008 | Brain stem laceration with open intracranial wound (disorder) |

| | | |
|--------------------------|--|----|
| OB | British preferred term | 64 |
| MTH_SYGB | Obsolete term | 12 |
| SB | Metathesaurus-supplied form of British synonym | 2 |
| XM | Named subset of a source | 1 |
| | Cross mapping set | 1 |

Dr. Olivier Bodenreider
(courtesy of Dr. Bastien Rance, NLM)

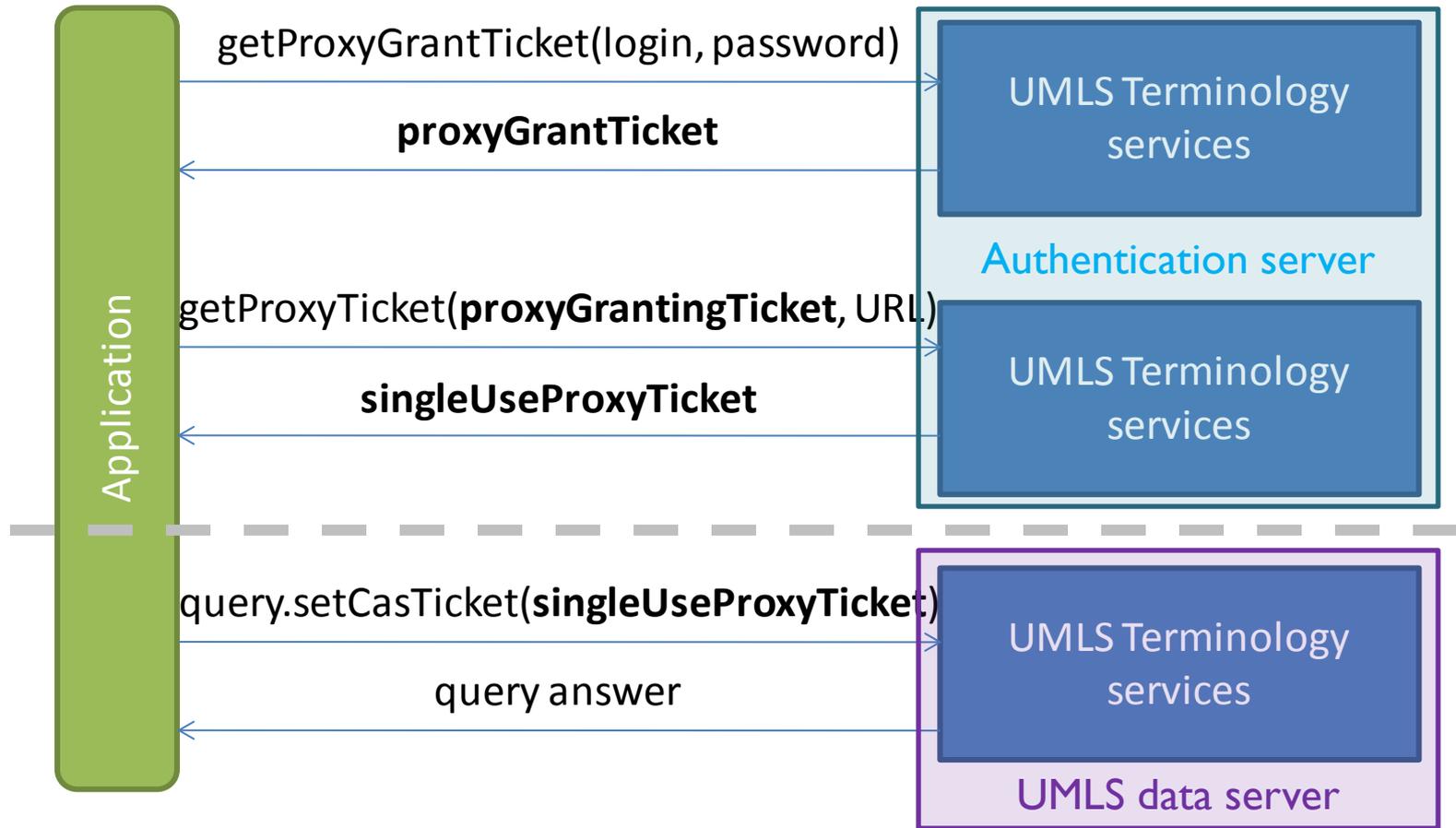
Accessing SNOMED CT content with the UTS API



Getting started with the UTS API

- Useful resources
 - UTS Website: signing up for a UMLS license, browsing the UMLS, access to technical documentation
<https://uts.nlm.nih.gov/>
 - UMLS API JavaDoc
<https://uts.nlm.nih.gov/doc/devGuide/javadocs/index.html>
 - UMLS API documentation
<https://uts.nlm.nih.gov/doc/devGuide/index.html>

Communication architecture



Getting a proxy granting ticket

```
private String _login;
private String _password;
public UMLSConnection(String login, String password) {
    _login = login;
    _password = password;
    try {
        // Locate the authentication web service
        URL authURL = new URL(
            "https://uts-ws.nlm.nih.gov/authorization/services/AuthorizationPort");
        _authPortType = new AuthorizationPortTypeServiceLocator().getAuthorizationPort(authURL);
        // Obtain a proxy granting ticket
        _proxyGrantingTicket = _authPortType.getProxyGrantTicket(login, password);
    } catch(Exception e) {
        System.err.println("ProxyGrantTicket");
        e.printStackTrace();
    }
}
```

Getting a single-use proxy ticket

```
private AuthorizationPortType _authPortType;

private String getSingleUseProxyTicket() {
    // Obtain a proxy granting ticket
    try {
        return _authPortType.getProxyTicket(_proxyGrantingTicket, "http://umlsks.nlm.nih.gov");
    } catch (RemoteException e) {
        System.err.println("SingleUseProxyTicket");
        e.printStackTrace();
        System.exit(2);
    }
    return null;
}
```

Getting a single-use proxy ticket

```
private String _kshost = "https://uts-ws.nlm.nih.gov";
private String _ksURI = _kshost + "/UMLSKS/services/UMLSKSService";
private String _UMLSVersion = "2010AB";

public List<ProvenanceInformation> getCUI(String searchString) {
    try {
        // Locate the UMLSTS web service
        URL ksURL = new URL(_ksURI);
        UMLSKSServicePortType uml sksService
            = new UMLSKSServiceLocator(). getUMLSKSServicePort (ksURL);

        // Build exact match request object
        ConceptIdExactRequest exactRequest = new ConceptIdExactRequest ();
        exactRequest. setCasTicket (this. getSingleUseProxyTicket ());
        exactRequest. setRelease(_UMLSVersion);
        exactRequest. setSearchString(searchString);
        exactRequest. setLanguage(_language);

        ConceptIdGroup exactRequestResults = uml sksService. findCUIByExact (exactRequest);
        List<String> cuis = new ArrayList<ProvenanceInformation>();
        for(Object o : exactRequestResults. getContents()) {
            results. add((ConceptId) o). getCUI);
        }
        return results;
    }
    [...]
    return null;
}
```

Dr. Kin Wah Fung

Finding correspondences to other terminologies through UMLS



Mapping

- ‘Mapping’ generally refers to 2 kinds of activity
 - Mapping a term to a terminology
 - Mapping between terminologies
- UMLS can help in both

The UMLS as a resource for terminology mapping

- Over 100 source vocabularies in the UMLS
- content organized according to meaning (concept-based organization) – an interlingua between terminologies
- rich network of relations between concepts – hierarchical relations and associative relations
- various lexical resources and lexical tools

Mapping terms to a terminology

- Generally, it works like this:
 - Find a match the incoming term against all terms in the UMLS
 - Normalization will increase yield
 - Synonymous word/phrase substitution
 - Ranking of degree of match
 - Restrict the match to UMLS concepts containing terms from the target terminology

The UMLS as a source of synonyms

▫ Descriptions

▫ Lang: en-US

- F antimalarial agent (product)
- P antimalarial agent
- S antimalarial



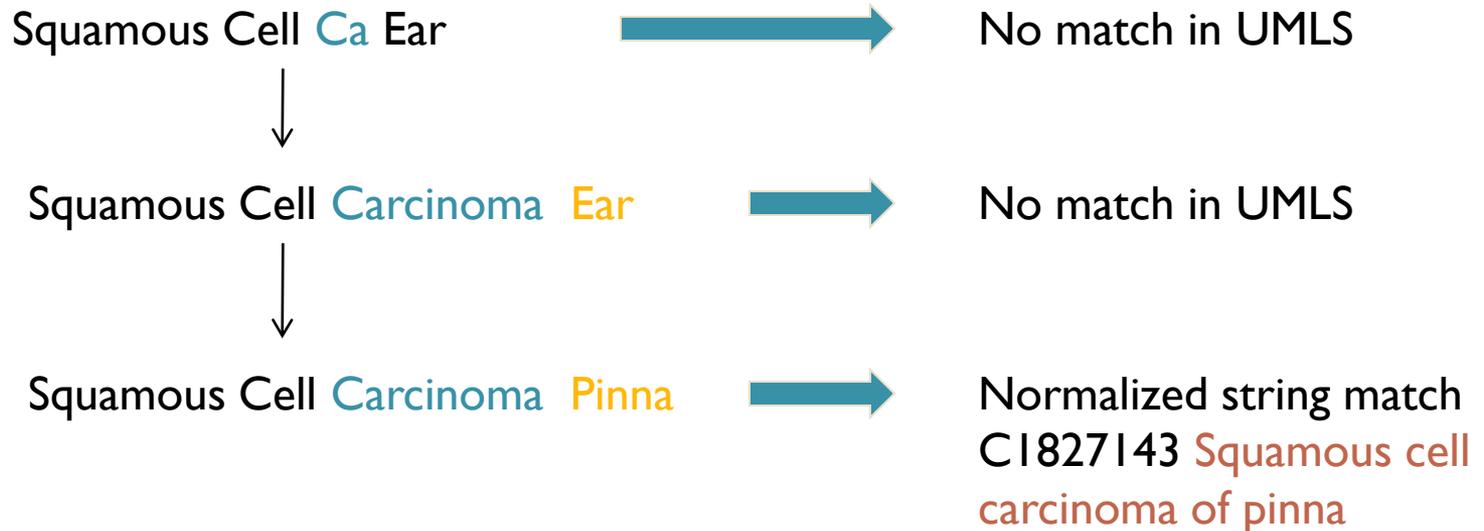
C0003374

MeSH
NCI Thesaurus
NDF-RT
SNOMED CT
[...]

Anti Malarials
anti-malarials
antimalarial
antimalarial agent
Antimalarial agent (product)
Antimalarial agent (substance)
Antimalarial agent, NOS
antimalarial agents
ANTIMALARIAL AGENTS
antimalarial drug
antimalarial drugs
Antimalarial [EPC]
antimalarials
ANTIMALARIALS
antimalarials (medication)
Drugs, Antimalarial
[AP101] ANTIMALARIALS
[...]

Example of synonym substitution

- In the research leading to the publication of the CORE Problem List Subset, we mapped all local problem list terms by exact and normalized string matches to all English strings in the UMLS
- Using a synonyms table, re-matching after synonymous word/phrase substitution (up to 2 substitutions)



MetaMap

- Developed by NLM
<http://metamap.nlm.nih.gov/>
- To map biomedical text to UMLS concepts
- Target can be restricted to specific vocabularies
- Mapping algorithm
 - parsing – noun phrases identified
 - variant generation
 - candidate retrieval – retrieve all Metathesaurus strings containing at least one of the variants
 - candidate evaluation – based on centrality, variation, coverage and cohesiveness
 - mapping construction – complete mappings constructed by combining candidates

Inter-terminology mapping

- Make use of the concept structure of the UMLS to identify equivalence between terminologies
- Utilize the relationships between UMLS concepts to improve yield

The IntraMap algorithm (I)

- Making use of the concept structure and relations in the UMLS, the algorithm sequentially goes through a series of steps until a target term is found

I. A target term is in the same concept as the source term

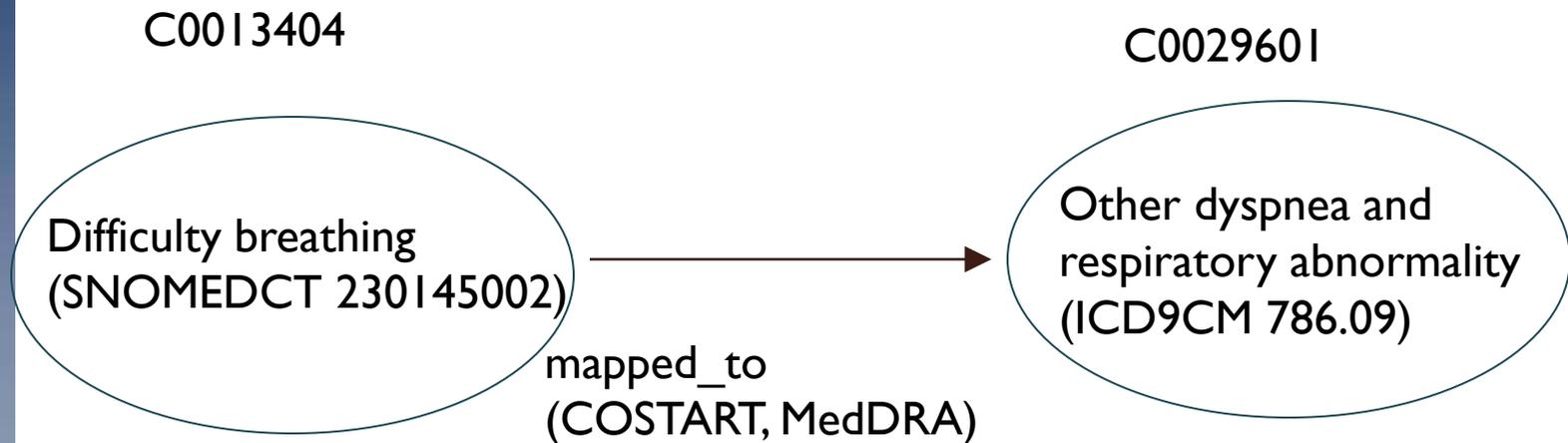
C0341350

Acute appendicitis with
peritonitis (SNOMEDCT
196781001)

Acute appendicitis with
peritonitis (ICD9CM 540.0)

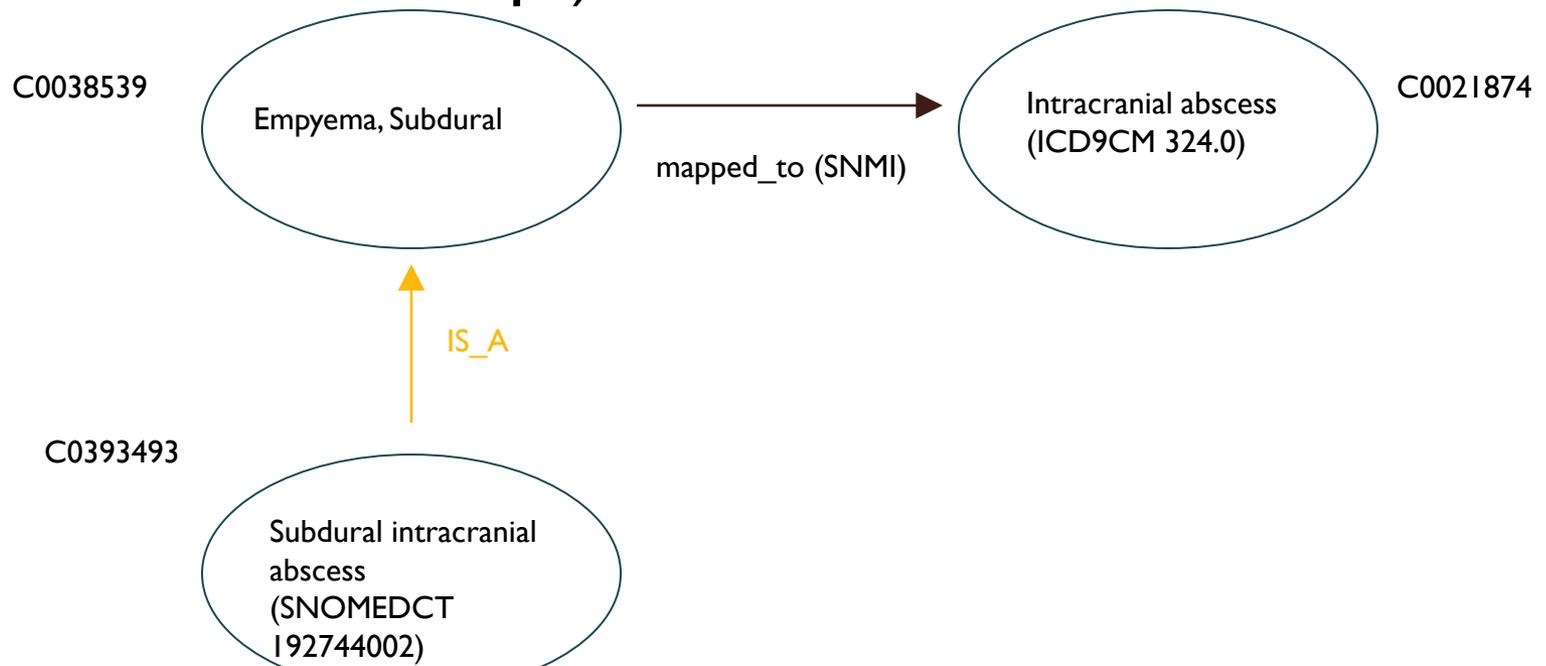
The IntraMap algorithm (2)

2. A target term is in a target concept linked to the source concept by an explicit mapping relation



The IntraMap algorithm (3)

3. A target term is found through any of the ancestors of the source concept (ancestors must have semantic type related to that of source concept)



Use of MetaMap for inter-terminology mapping

- Input:
 - terms from the source terminology, with ‘term processing’ option turned on to bypass parsing into component phrases
- Output:
 - restricted to UMLS concepts containing terms from the target terminology
 - further restriction can be done by semantic types
 - MetaMap score used to rank mappings

Ms. Janice H. Willis

Finding terms in other languages through UMLS



Motivation

- Translation
 - Seeding the translation
 - Checking translated terms against other terminologies

Languages in the UMLS Metathesaurus

| Language | Name Count | % of Metathesaurus |
|----------|------------|--------------------|
| ENG | 7258337 | 68.12% |
| SPA | 1857522 | 17.43% |
| JPN | 285857 | 2.68% |
| DUT | 220186 | 2.07% |
| FRE | 197500 | 1.85% |
| GER | 185254 | 1.74% |
| POR | 157124 | 1.47% |
| ITA | 136103 | 1.28% |
| CZE | 131367 | 1.23% |
| RUS | 106860 | 1% |
| POL | 42127 | 0.4% |
| SWE | 26311 | 0.25% |
| FIN | 25489 | 0.24% |
| KOR | 11372 | 0.11% |
| SCR | 8844 | 0.08% |
| LAV | 1406 | 0.01% |
| DAN | 723 | 0.01% |
| NOR | 722 | 0.01% |
| HUN | 718 | 0.01% |
| BAQ | 695 | 0.01% |
| HEB | 485 | 0% |

Multi-lingual vocabularies in the UMLS

(2011AA)

- SNOMED CT (SPA)
- MeSH
 - CZE, DUT, FIN, FRE, GER, ITA, JPN, LAV, POL, POR, RUS, SCR, SPA, SWE
- ICPC / ICPC2
 - BAQ, DAN, DUT, FIN, FRE, GER, HEB, HUN, ITA, NOR, POR, SPA, SWE
- ICD10
 - GER, DUT
- MedDRA
 - CZE, DUT, FRE, GER, ITA, JPN, POR, SPA
- UMDNS
 - GER
- Minimal Standard Terminology Digestive Endoscopy
 - FRE, ITA
- WHO Adverse Drug Reaction Terminology (WHOART)
 - FRE, GER, POR, SPA
- Korean Standard Classification of Disease
 - KOR

Multi-lingual vocabularies in the UMLS

| Language | MeSH | ICPC | MedDRA |
|----------|------|------|--------|
| BAQ | | X | |
| CZE | X | | X |
| DAN | | X | |
| DUT | X | X | X |
| ENG | X | X | X |
| FIN | X | X | |
| FRE | X | X | X |
| GER | X | X | X |
| HEB | | X | |
| HUN | | X | |
| ITA | X | X | X |
| JPN | X | | X |
| KOR | | | |
| LAV | X | | |
| NOR | | X | |
| POL | X | | |
| POR | X | X | X |
| RUS | X | | |
| SCR | X | | |
| SPA | X | X | X |
| SWE | X | X | |

Finding terms in other languages

- UTS
 - Search SNOMED CT term in English (or SNOMED CT ID)
 - Find UMLS CUI
 - Explore source vocabularies / languages for this concept

Example

- SNOMED CT concept
Addison's disease [363732003]

The screenshot displays the SNOMED CT browser interface for the concept 'Addison's disease' (ID: 363732003). The interface is divided into several sections:

- Header:** Shows the Concept ID (363732003) and Description ID (485624014). The concept name is 'Addison's disease' (Type: clinical finding).
- Search:** A search bar contains 'addison's disease', and a list of 14 results is shown. The top result is 'Addison's disease' (P).
- Hierarchy:** A 'Subtype hierarchy' view shows 'Addison's disease' as a subtype of 'adrenal cortical hypofunction'.
- Details:** A right-hand pane provides detailed information:
 - Concept Status:** current
 - Descriptions:** Includes 'Addison's disease (disorder)', 'Addison's disease', and 'Addison disease'.
 - Definition:** Primitive. It is defined as 'is a' of 'adrenal cortical hypofunction' and 'finding site' of 'adrenal cortex structure'.
 - Qualifiers:** Includes 'severity' (severities), 'episodicity' (episodicities), and 'clinical course' (courses).
 - Codes:** Original SNOMED ID: DB-70620; Read Code (Ctv3Id): C1541.

Addison's disease in UMLS

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Term CUI Code

363732003

Release: 2011AA

Search Type: EXACT_MATCH

Source: All Sources

- AIR
- ALT
- AOD
- AOT

Search Results (1)

[C0001403](#) Addison Disease

[Basic View](#) | **[Report View](#)** | [Raw View](#)

Concept: [C0001403] Addison Disease

Semantic Types

- [Disease or Syndrome](#) [T047]

Definitions

CSP/PT | disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin; due to tuberculosis or autoimmune induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol.

MEDLINEPLUS/PT |

Your adrenal glands are just above your kidneys. The outside layer of these glands makes hormones that help your body respond to stress and regulate your blood pressure and water and salt balance. Addison's disease occurs if the adrenal glands don't make enough of these hormones.

A problem with your immune system usually causes Addison's disease. The immune system mistakenly attacks your own tissues, damaging your adrenal glands.

Symptoms include

- Weight loss
- Muscle weakness
- Fatigue that gets worse over time
- Low blood pressure

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Languages for C0001403

| Language | Name |
|----------|-----------------------|
| CZE | Addisonova choroba |
| DUT | Addison, ziekte van |
| FIN | Addisonin tauti |
| FRE | Maladie d'Addison |
| GER | Addisonsche Krankheit |
| ITA | Malattia di Addison |
| JPN | アジソン病 |
| KOR | 원발성 부신피질 기능부전 |
| POL | Choroba Addisona |
| POR | Doença de Addison |
| RUS | АДДИСОНОВА БОЛЕЗНЬ |
| SCR | ADDISONOVA BOLEST |
| SPA | Enfermedad de Addison |
| SWE | Addisons sjukdom |

C0001403 in Dutch

| Source | Term type | Code | Name |
|--------|-----------|----------|--------------------------------------|
| ICD10 | PT | E27.1 | Primaire bijnierschorsinsufficiëntie |
| MedDRA | PT | I0001130 | Addison, ziekte van |
| MedDRA | PT | I0052381 | primaire bijnierinsufficiëntie |
| MeSH | MH | D000224 | Ziekte van Addison |
| MeSH | SY | D000224 | Addison, syndroom van |
| MeSH | SY | D000224 | Addison, ziekte van |
| MeSH | SY | D000224 | Primaire bijnierschorsinsufficiëntie |

C0001403 in French

| Source | Term type | Code | Name |
|---------|-----------|----------|---|
| MedDRA | PT | I0001130 | Maladie d'Addison |
| MedDRA | PT | I0052381 | Insuffisance surrénalienne primaire |
| MeSH | EP | D000224 | Insuffisance corticosurrénalienne primitive |
| MeSH | EP | D000224 | Insuffisance surrénale lente |
| MeSH | EP | D000224 | Insuffisance surrénale primitive |
| MeSH | EP | D000224 | Insuffisance surrénalienne primitive |
| MeSH | MH | D000224 | Maladie d'Addison |
| WHO ART | IT | 410 | MALADIE D'ADDISON |

Dr. James T. Case

**Use case: The role of UMLS in
the NLM US SNOMED CT
Content Request SYSTEM
(USCRS)**



Rationale for USCRS

- IHTSDO requires submissions to the International release to go through the member National Release Center
 - Existing request system being phased out
 - No new accounts accepted
 - Older users “grandfathered – in”
- Needed support for U.S. users who need to add content to SNOMED CT
- Support users who need content with stable (maintained) identifiers before next release
- Must support functions defined by IHTSDO
- Triage and handling by NLM

Access to the USCRS

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- [US SNOMED CT Content Request System](#)
- [Convergent Medical Terminology](#)
- [SNOMED CT Browser](#)
- [Subsets](#)

As you to:

and create a UTS

ations including:

- Semantic Network Browser
- SNOMED CT Browser
- Download data files including:
 - UMLS Knowledge Sources
 - RxNorm weekly and monthly updates
 - SNOMED CT
 - CORE Problem List and Route of Administration Subsets of SNOMED CT
- Query data remotely via Web Services (see API Documentation)
- Complete UMLS Annual Report and SNOMED CT® Affiliate Reports

UMLS Terminology Services (UTS) provide both web interfaces as well as Web Services to search and retrieve UMLS data.



What's New

Welcome to the beta version of the new U.S. SNOMED CT® Content Request System (USCRS). This system allows users to request basic changes to SNOMED CT.

NLM values your input to assist us in improving this system. Enhancements to its functionality and documentation are already in the works and will be implemented in the coming months.

Please send your suggestions, comments and questions to NLM Customer Service with the subject line "U.S. SNOMED CT Content Request System".

Recent Requests

Request 1 .. 10 of 100

| Batch Id | Request Id | Topic | Request Type | Status | Submitted By | Submitted On | Last Modified | Summary | Actions |
|--------------------------|----------------------------|-----------------------|------------------------------|------------------------|------------------------------|------------------------------|-------------------------------|---|-------------------------|
| 8673 | 9081 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Bacterial Isolate Specimen (specimen) | |
| 8673 | 9082 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Lesion Swab (specimen) | |
| 8673 | 9083 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Nasopharyngeal and Rectal and Eye swab (specimen) | |
| 8673 | 9086 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Tracheal Swab (specimen) | |
| 8673 | 9085 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Vaginal and Rectal Swab (specimen) | |
| 8673 | 9084 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Throat and Rectal Swab (specimen) | |
| 8673 | 9088 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Vesicle Swab (specimen) | |
| 8673 | 9087 | specimen terms | New Concept | Accepted | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Stool Swab (specimen) | |
| 8673 | 9089 | specimen terms | New Concept | Rejected | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Muscle Specimen (specimen) | |
| 8673 | 9090 | specimen terms | New Concept | Under Appeal | Riki Merrick | Sep 28, 2011 | Sep 29, 2011 | Cardiac Muscle Specimen (specimen) | |

Request 1 .. 10 of 100

Show requests per page

[Previous](#) [Next](#)

Summary

Total Requests: 2416
My Requests:
[Submitted: 1795](#)
[Accepted: 723](#)
[Approved: 21](#)
[Draft: 0](#)

Important Dates

September 15, 2011
Submit requests for next US Extension of SNOMED CT

November 2011
CORE Problem List Subset of SNOMED CT

January 31, 2012
SNOMED CT International release

March 2012
U.S. Extension to SNOMED CT next release (est.)



Request Type:

- Add Parent
- Change Description
- Change Parent
- Change Relationship
- New Concept**
- New Relationship
- New Synonym
- Retire Concept
- Retire Description
- Retire Relationship

New request allows a number of request types



Search Text:

Request Type:

Status:

Submitted on: from to

Last modified: from to

Restrict to My Requests Include Draft Requests

Search allows you to search all current requests from any user



- Home**
- New Requests**
- My Requests**
- Search Requests**
- Resources**
- Admin**

Submitter:

Request Type:

Topic:*

Source Terminology:*

Local Code:

Local Term:

Fully Specified Name:*

Semantic Tag:*

Preferred Term:*

Synonym:

Parent Concept Id:*

UMLS CUI:

Definition:*

Proposed Use:

Justification:*

Note:

*indicates required information

-
-
-

USCRS Status

- Currently “Post-beta”
- General availability began late September 2011
- Future phases
 - Access to complete history of request changes
 - Manager alerts (priority todo)
 - Notes enhancements: links & attachments
 - Enhanced reporting capabilities (e.g. report of submitter activity)
 - Duplicate request identification and review
 - Extended request types – ref sets, content areas, etc.
 - Mobile app interface
- <http://uscrs.nlm.nih.gov/>



Why integrate UMLS with a SCT browser?

- Leverages the UMLS to find SCT concepts
 - Allows location of SNOMED CT concepts using UMLS descriptions
- As part of UTS, will link directly to other UTS services



UMLS Terminology Services

SNOMED CT Browser

Search Tree Recent Searches

SNOMED CT Version: 2011_0

Term ConceptID Description

sclerosis

Active concepts only:

Restrict results to: --disorder

Search Results (101)
[: 1 - 25 :]

- [230380005](#) Balo concentric sclerosis (disorder)
- [49692006](#) Schilder's disease (disorder)
- [53741008](#) Coronary arteriosclerosis (disorder)
- [34643004](#) Diaphyseal dysplasia (disorder)
- [24700007](#) Multiple sclerosis (disorder)
- [7199000](#) Tuberos sclerosis syndrome (disorder)
- [60576007](#) Subacute combined degeneration of spinal
- [53889007](#) Nuclear cataract (disorder)
- [48124008](#) Fibrosis of bile duct (disorder)
- [398716006](#) Endomyocardial fibrosis (disorder)
- [67754003](#) Aortic valve sclerosis (disorder)
- [89155008](#) Systemic sclerosis (disorder)
- [86352002](#) Sclerosis of the skin (disorder)
- [235899008](#) Hepatic sclerosis (disorder)
- [95426000](#) Apophyseal sclerosis (disorder)
- [417094009](#) Valvular sclerosis (disorder)
- [102028003](#) Generalized multiple sclerosis (disorder)

Can limit to active concepts only

Restrict to a specific top level concept

Results pared down with additional search terms

Full information about concept available

Report View

CUI: [\[C0007795\]](#) Diffuse Cerebral Sclerosis of Schilder

Semantic Types: [Disease or Syndrome \[T047\]](#)

| Id | Description | Type | Status |
|------------|----------------------------------|------------------------|-------------|
| 787398016 | Schilder's disease (disorder) | FullySpecifiedName (3) | Current (0) |
| 82759010 | Schilder's disease | Preferred (1) | Current (0) |
| 1230760015 | Diffuse sclerosis | Synonym (2) | Current (0) |
| 82760017 | Encephalitis periaxialis diffusa | Synonym (2) | Current (0) |
| 1230759013 | Schilder disease | Synonym (2) | Current (0) |

Parents (1)

Relationships from *this* concept (6)

Relationships to *this* concept (3)

No SNOMED CT Description?

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SNOMED CT Version: 2011_01_31

Term ConceptID DescriptionID

mesial temporal sclerosis

Go

Active concepts only:

Restrict results to: --None--

There were no matches for your query in SNOMED CT with the restrictors you have chosen.

However, there are matching concepts in the UMLS Metathesaurus.

Follow [this link](#) to query the UMLS.

Informs you whether there is a UMLS concept matching your search term

Harnessing the UMLS

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Search term of interest

[Search](#) [Tree](#)

[View](#)

SNOMED CT Version: 2011

Term Concept DescriptionID

hypotension

Active concepts only:

Restrict results to: --None--

Search Results (31)

[: 1 - 25 :]

- [45007003](#) Low blood pressure (disorder)
- [28651003](#) Orthostatic hypotension (disorder)
- [19721008](#) Hypotony of eye (disorder)
- [77545000](#) Chronic hypotension (disorder)
- [195506001](#) Idiopathic hypotension (disorder)
- [408668005](#) Iatrogenic hypotension (disorder)
- [241727003](#) Induced hypotension (procedure)
- [266695002](#) Hypothermia with hypotension (procedure)
- [276519002](#) Neonatal hypotension (disorder)
- [429561008](#) Exertional hypotension (disorder)
- [408667000](#) Hemodialysis-associated hypotension (disorder)
- [31695008](#) Hypotension, induction and maintenance (procedure)
- [75181005](#) Chronic orthostatic hypotension (disorder)

Concept: [45007003] Low blood pressure

UMLS information

CUI: [\[C0020649\]](#) Hypotension

Semantic Types: [Finding](#) [T033]

| ConceptStatus | IsPrimitive | SnomedId | CTV3Id |
|---------------|-------------|----------|--------|
| Current (0) | 1 | D3-04000 | G87.. |

Descriptions (7)

| Id | Description | Type | Status |
|-----------|-------------------------------|------------------------|-----------------|
| 782199011 | Low blood pressure (disorder) | FullySpecifiedName (3) | Current (0) |
| 494085018 | Low blood pressure | Preferred (1) | Current (0) |
| 75075016 | Arterial hypotension | Synonym (2) | Current (0) |
| 75073011 | Hypopiesis | Synonym (2) | Current (0) |
| 75074017 | Arterial hypotension, NOS | Synonym (2) | Non-Current (1) |
| 75071013 | Hypotension | Synonym (2) | Non-Current (1) |
| 75072018 | Hypotension, NOS | Synonym (2) | Non-Current (1) |

Parents (2)

Children (11)

Relationships from *this* concept (6)

Relationships to *this* concept (16)

Tree Positions (12)

Matching SNOMED CT
concept

Not one of the active
descriptions

UMLS Concept

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UMLS Terminology Services Metathesaurus Browser

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Term CUI Code

mesial temporal sclerosis

Go

Release: 2011AA

Search Type: Word

Source: All Sources

AIR

ALT

AOD

AOT

[C2062593](#) mesial temporal sclerosis

[C2104602](#) mesial temporal sclerosis with intractable ep

Basic View | [Report View](#) | [Raw View](#)

Concept: [C2062593] mesial temporal sclerosis

Semantic Types

[Disease or Syndrome](#) [T047]

Atoms (4) string [AUI / RSAB / TTY / Code]

Mesial temporal sclerosis [A17787307/ICD10CM/ET/G93.81]

mesial temporal sclerosis [A13982327/MEDCIN/PT/31984]

mesial temporal sclerosis (diagnosis) [A13794850/MEDCIN/FN/31984]

Mesial temporal sclerosis [A16982626/MTHICD9/ET/348.81]

Contexts (1)

Concept exists in ICD9;
ICD10CM and
MEDCIN

References

References: UMLS home page

- UMLS home page
 - <http://www.nlm.nih.gov/research/umls/>
- UMLS documentation
 - Reference manual
<http://www.ncbi.nlm.nih.gov/books/NBK9676/>
 - Source documentation
<http://www.nlm.nih.gov/research/umls/sourcereleasedocs/index.html>
- UMLS online tutorials
 - http://www.nlm.nih.gov/research/umls/user_education/index.html

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