Using SNOMED CT with the UMLS

Unified Medical Language System®

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INFORMATION INNOVATION

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

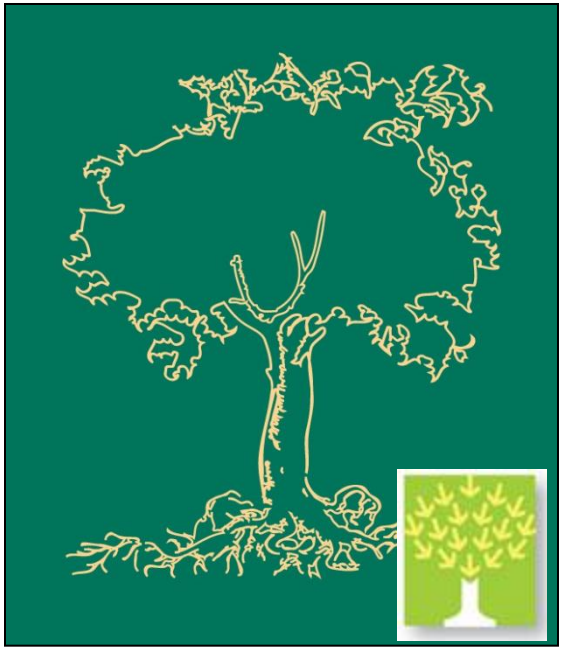
Overview of the UMLS

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

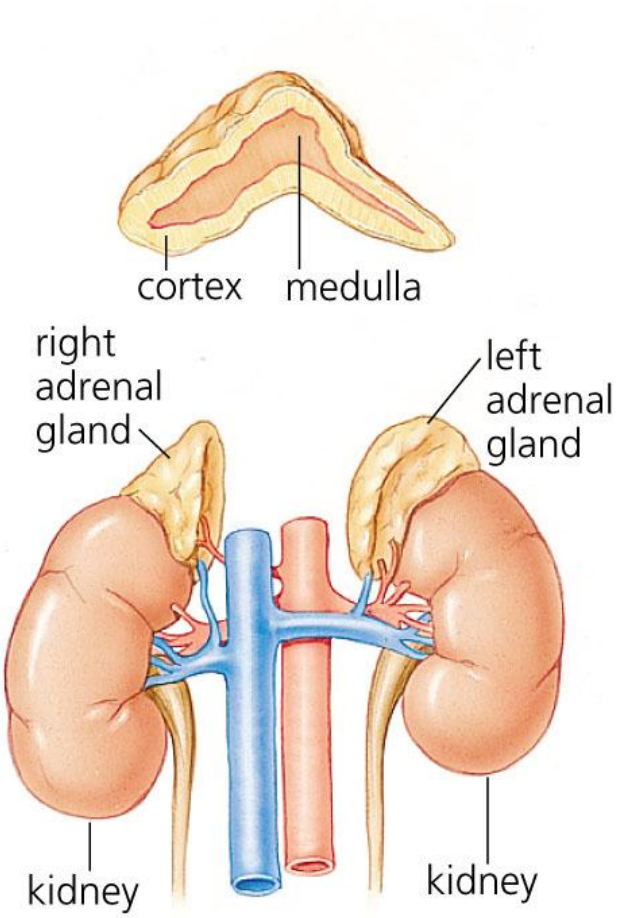
- Database
  - Series of relational files
- Interfaces
  - Web interface: Knowledge Source Server (UMLSKS)
  - Application programming interfaces (Java and web services)
- Applications
  - lvg (lexical programs)
  - MetamorphoSys (installation and customization)
  - RRF browser (browsing subsets)
- The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

Overview of the UMLS

Overview through an example

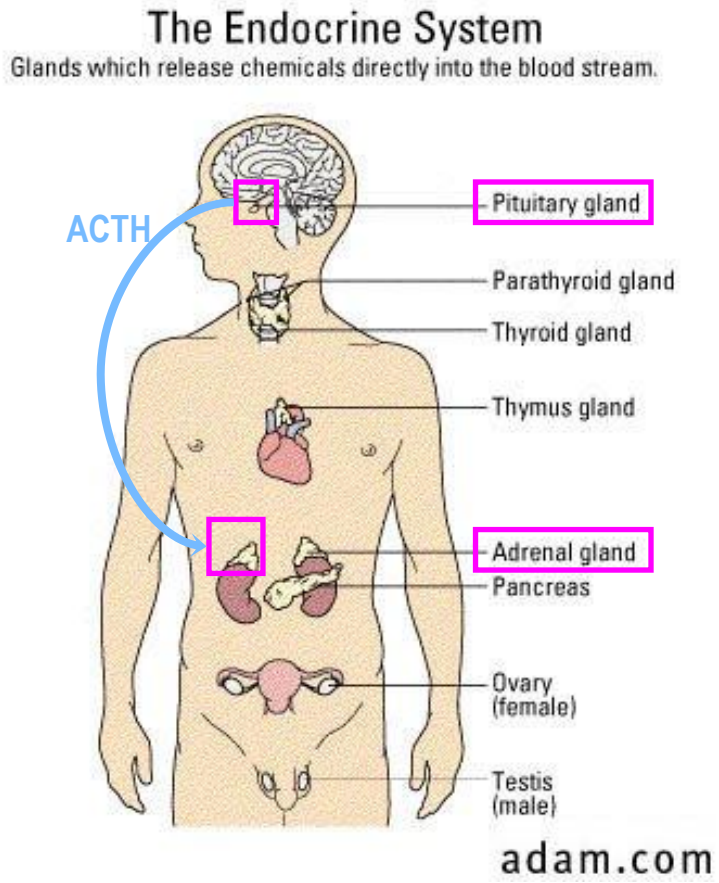
Addison's disease

- 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



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
- Contexts: different hierarchies

eponym

symptoms

clinical variants

Organize terms

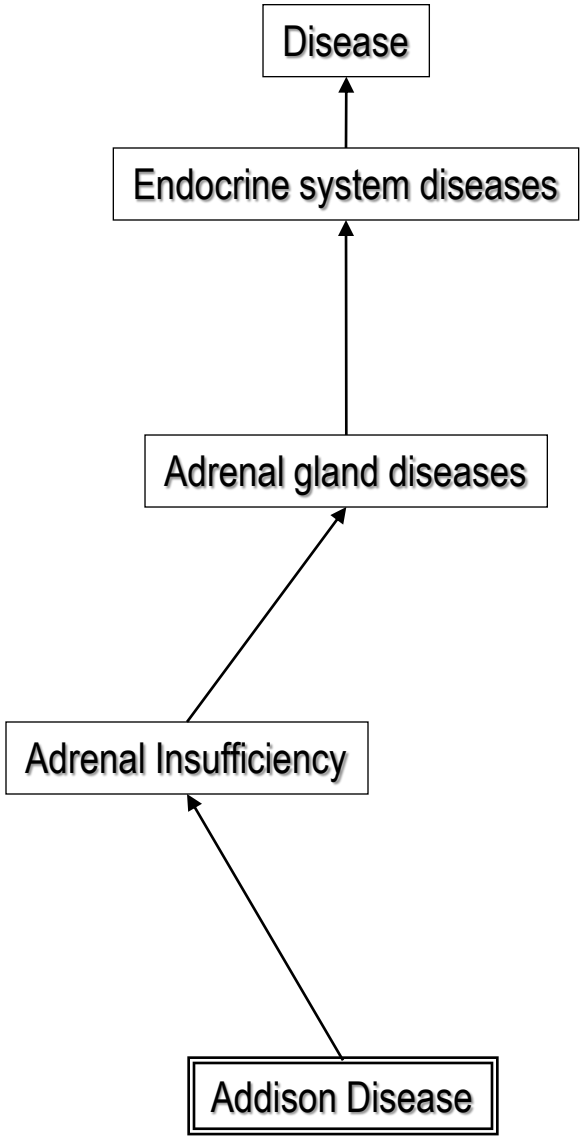
- Synonymous terms clustered into a concept
- Preferred term
- Unique identifier (CUI)

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

MeSH



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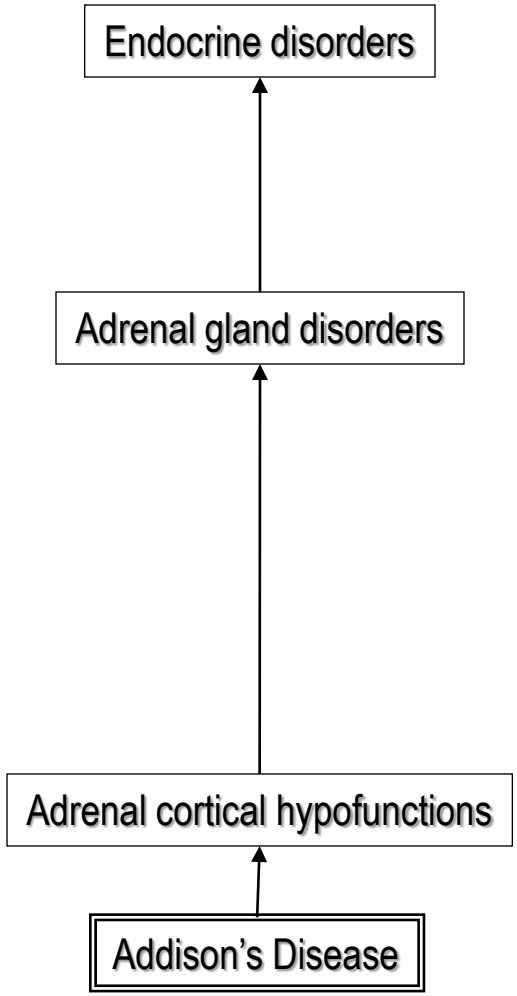
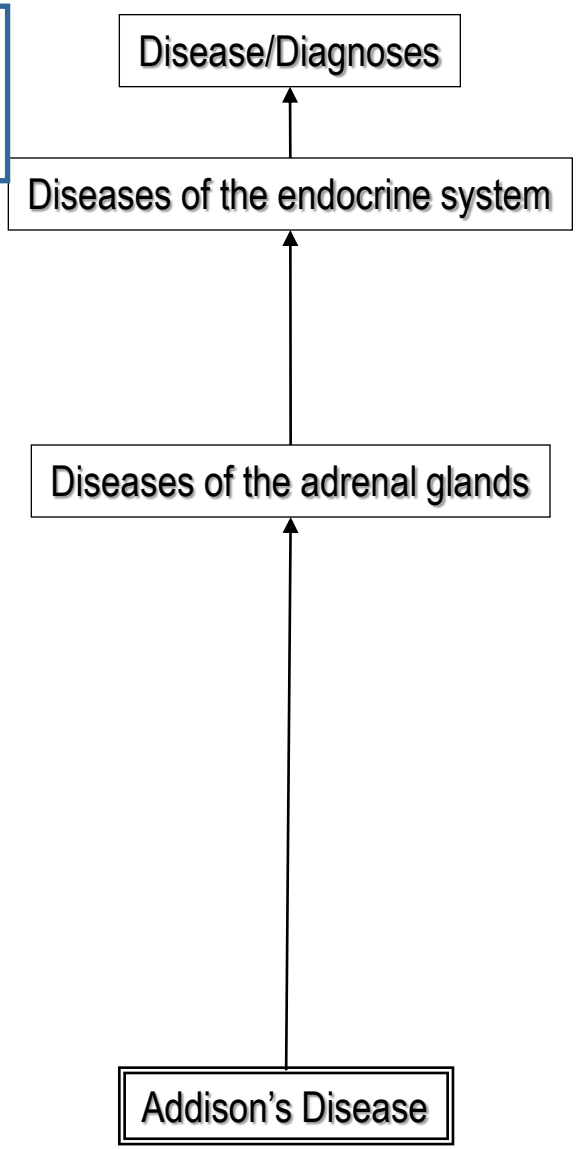
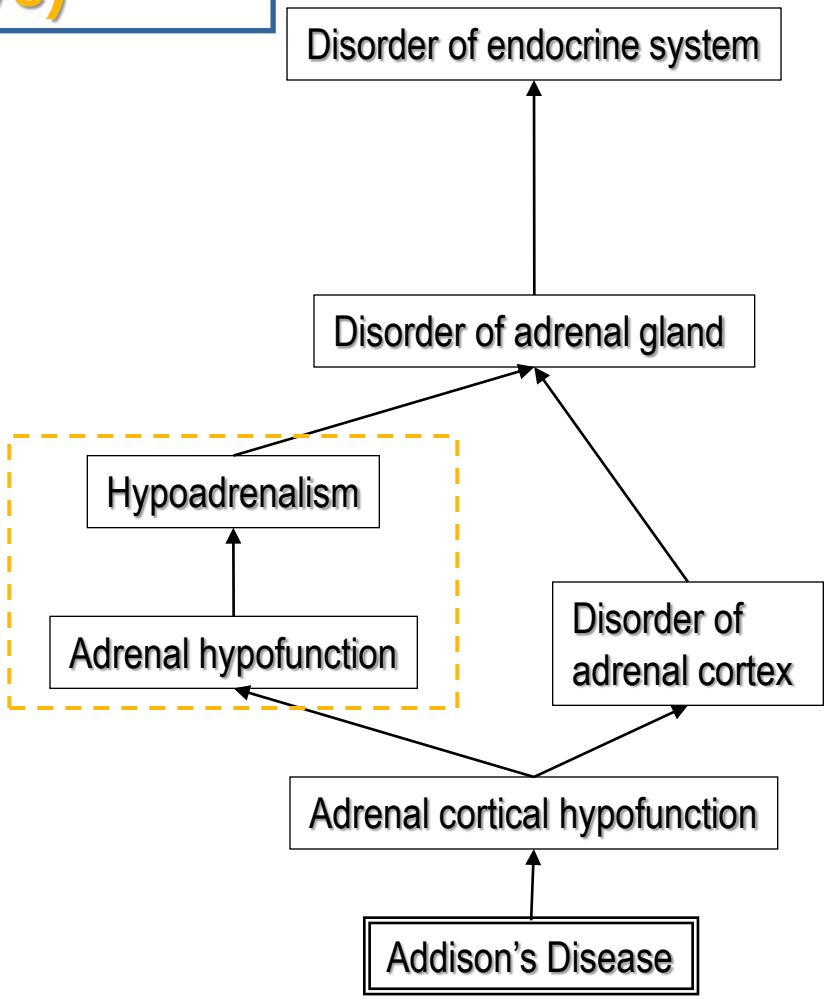
MedDRA


Endocrine disorders
Adrenal gland disorders
Adrenal cortical hypofunctions
Addison's Disease

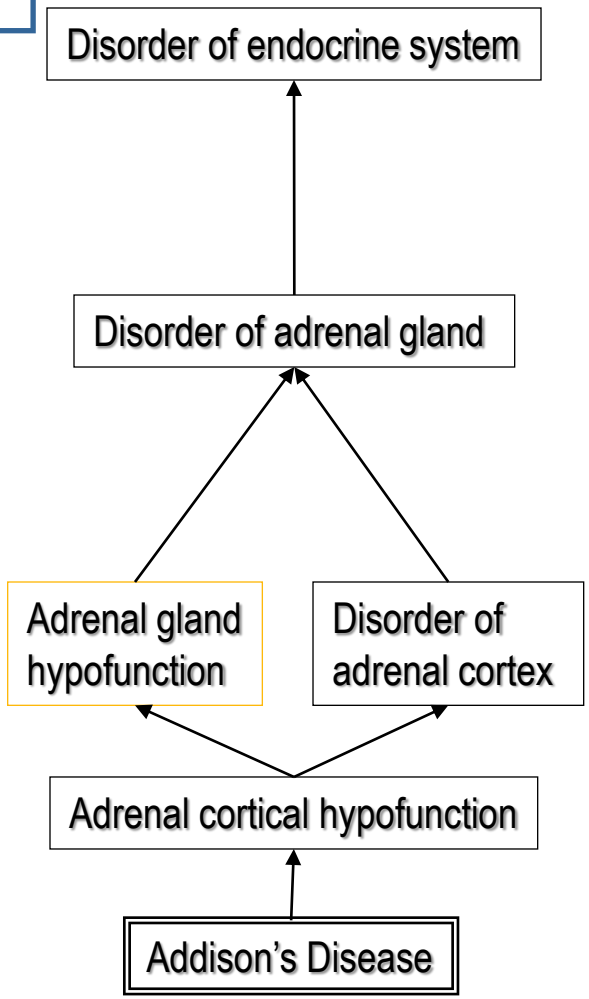
SNOMED International



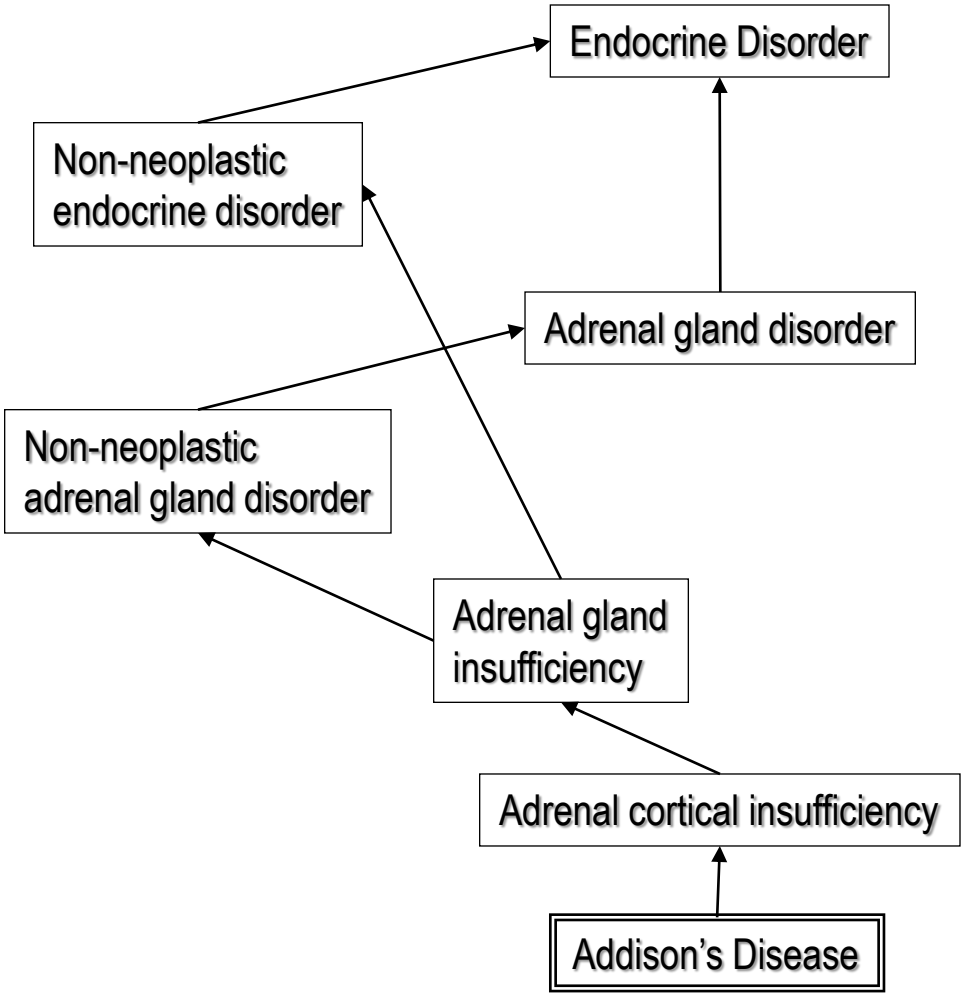
SNOMED CT (native)



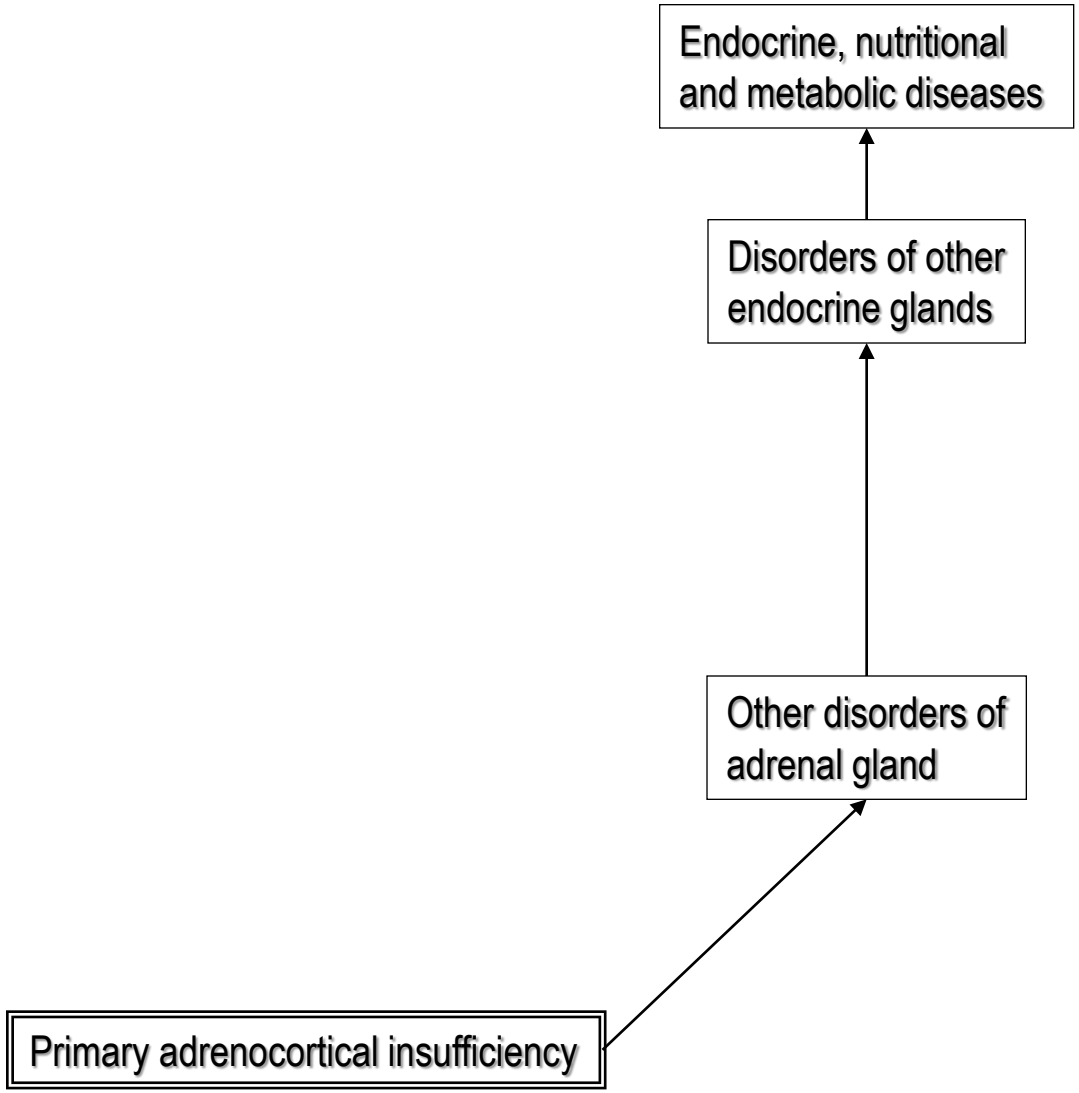
SNOMED CT (UMLS view)



NCI Thesaurus

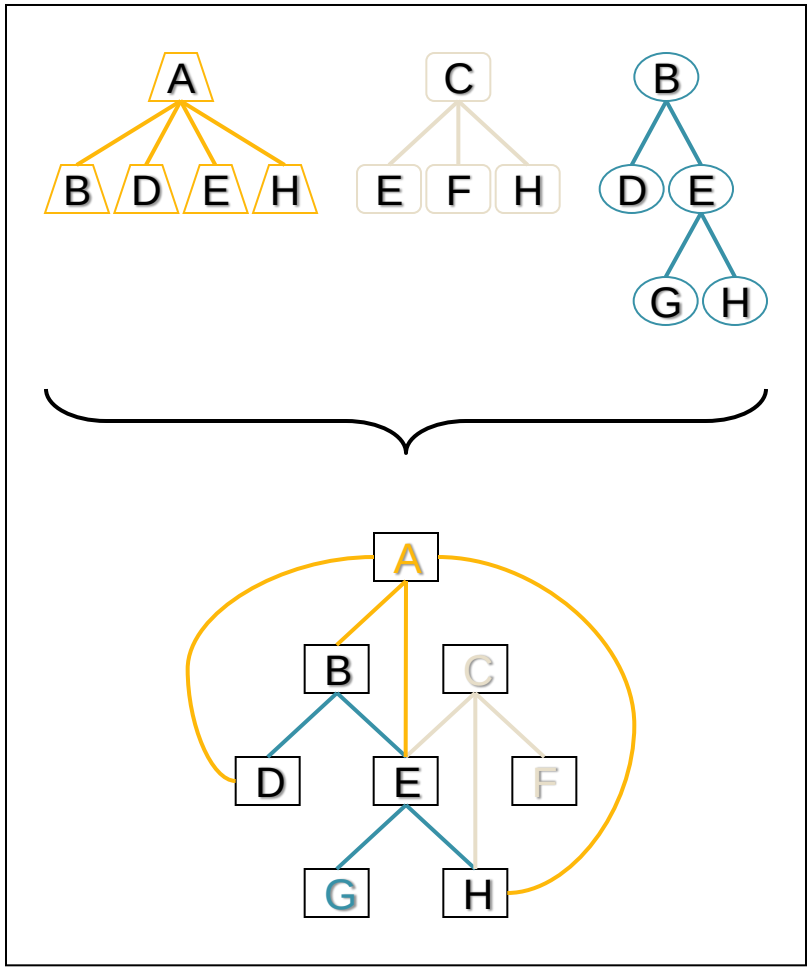


ICD-10

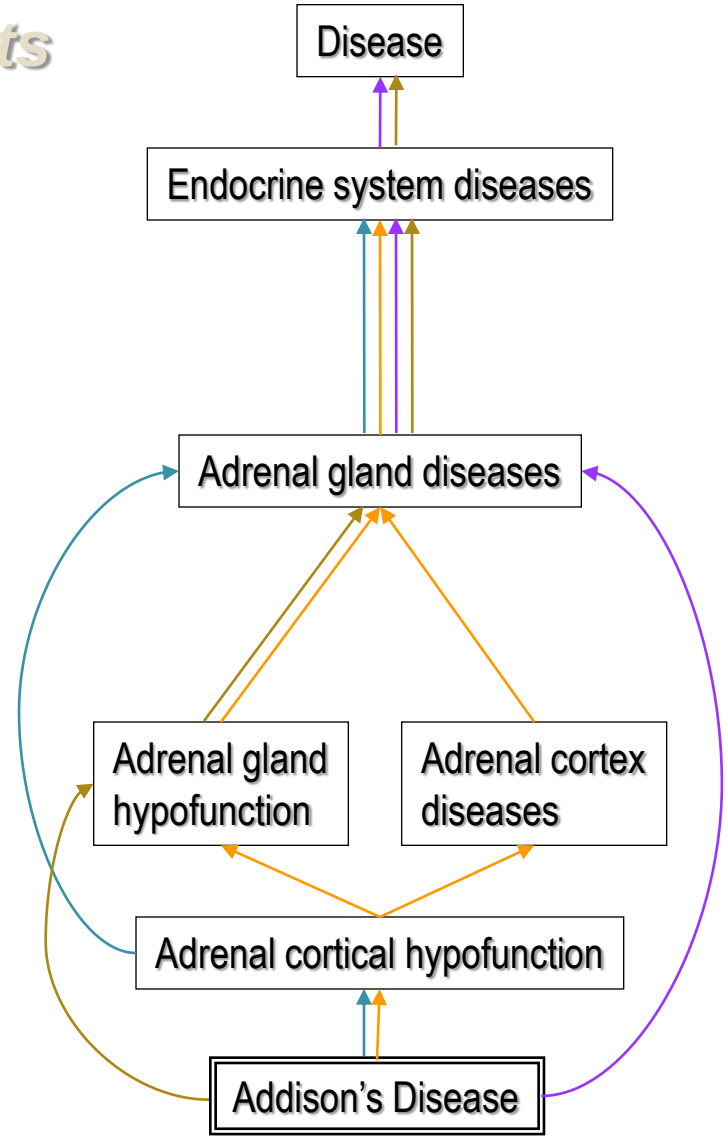

Endocrine, nutritional and metabolic diseases
Disorders of other endocrine glands
Other disorders of adrenal gland
Primary adrenocortical insufficiency

Organize concepts

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

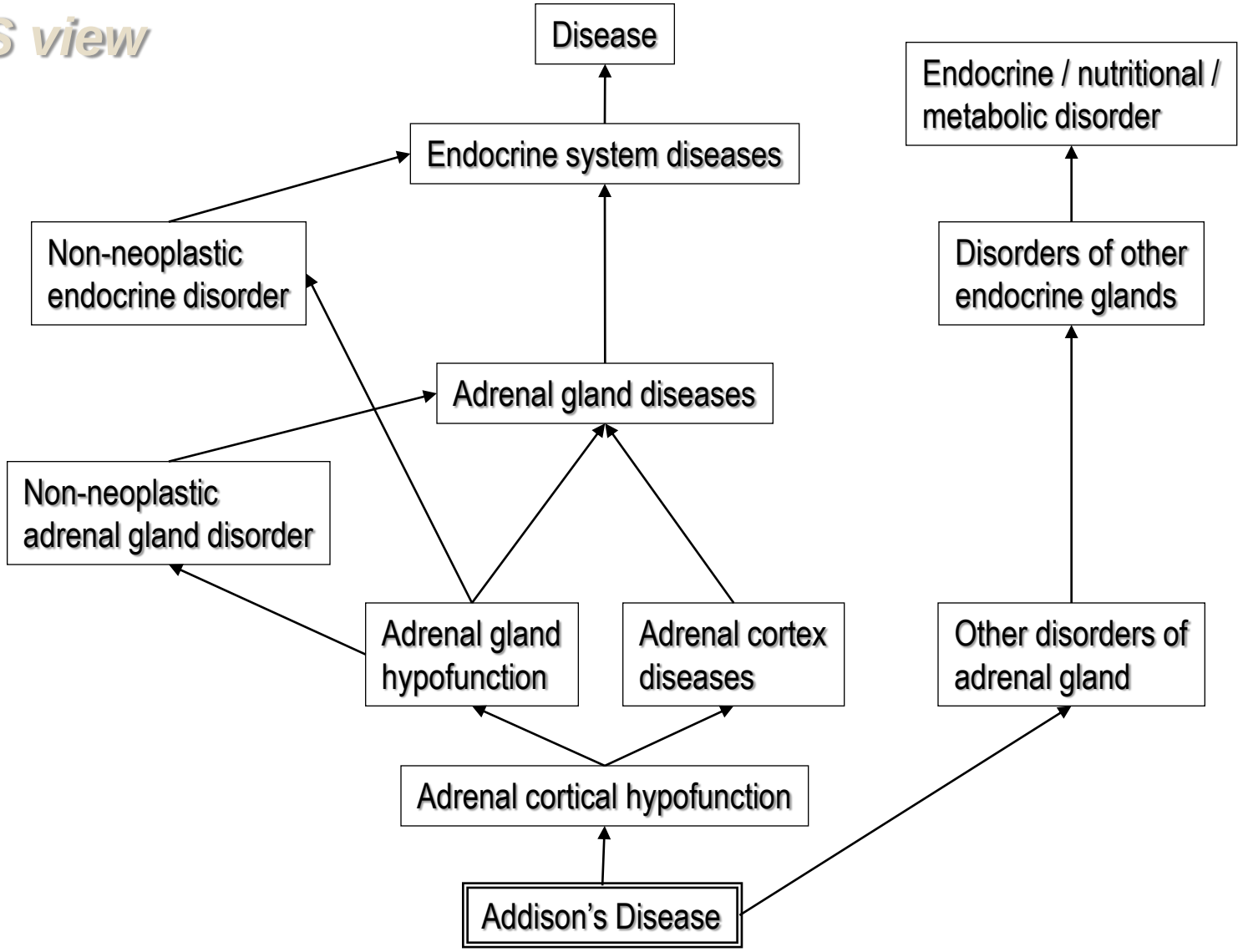


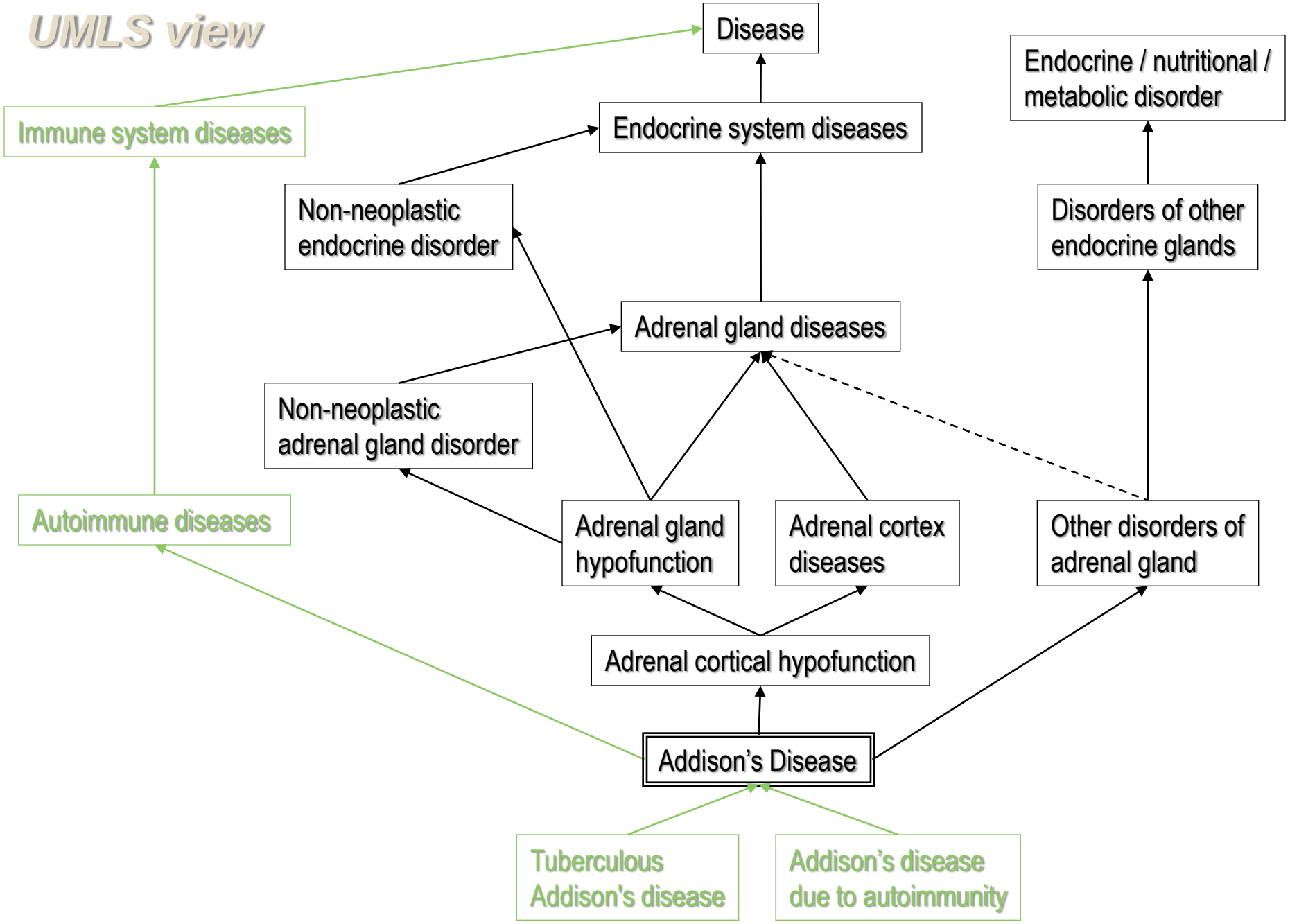
organize concepts



SNOMED CT
SNOMED Intl
MeSH
MedDRA

UMLS view


Disease
Endocrine / nutritional / metabolic disorder
Endocrine system diseases
Non-neoplastic endocrine disorder
Disorders of other endocrine glands
Adrenal gland diseases
Non-neoplastic adrenal gland disorder
Adrenal gland hypofunction
Adrenal cortex diseases
Other disorders of adrenal gland
Adrenal cortical hypofunction
Addison's Disease

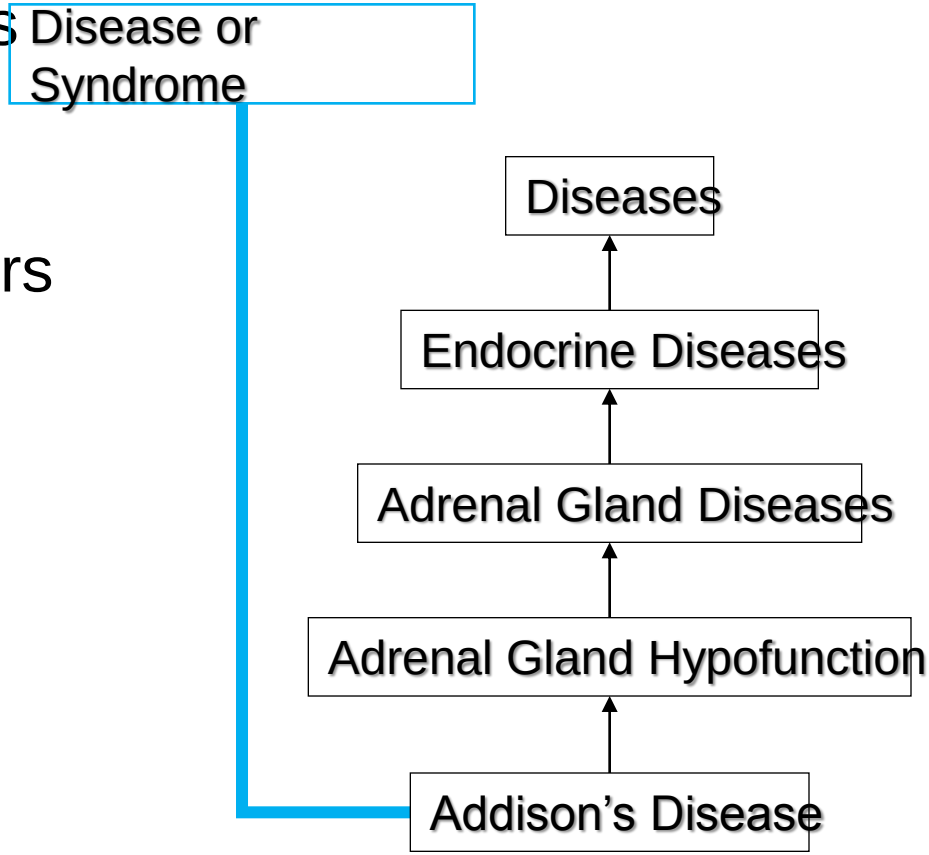

UMLS view
Disease
Endocrine / nutritional / metabolic disorder
Immune system diseases
Endocrine system diseases
Non-neoplastic endocrine disorder
Disorders of other endocrine glands
Adrenal gland diseases
Non-neoplastic adrenal gland disorder
Autoimmune diseases
Adrenal gland hypofunction
Adrenal cortex diseases
Other disorders of adrenal gland
Adrenal cortical hypofunction
Addison's Disease
Tuberculous Addison's disease
Addison's disease due to autoimmunity

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



How do they do that?

- Lexical knowledge
- Semantic pre-processing
- UMLS editors

Lexical knowledge

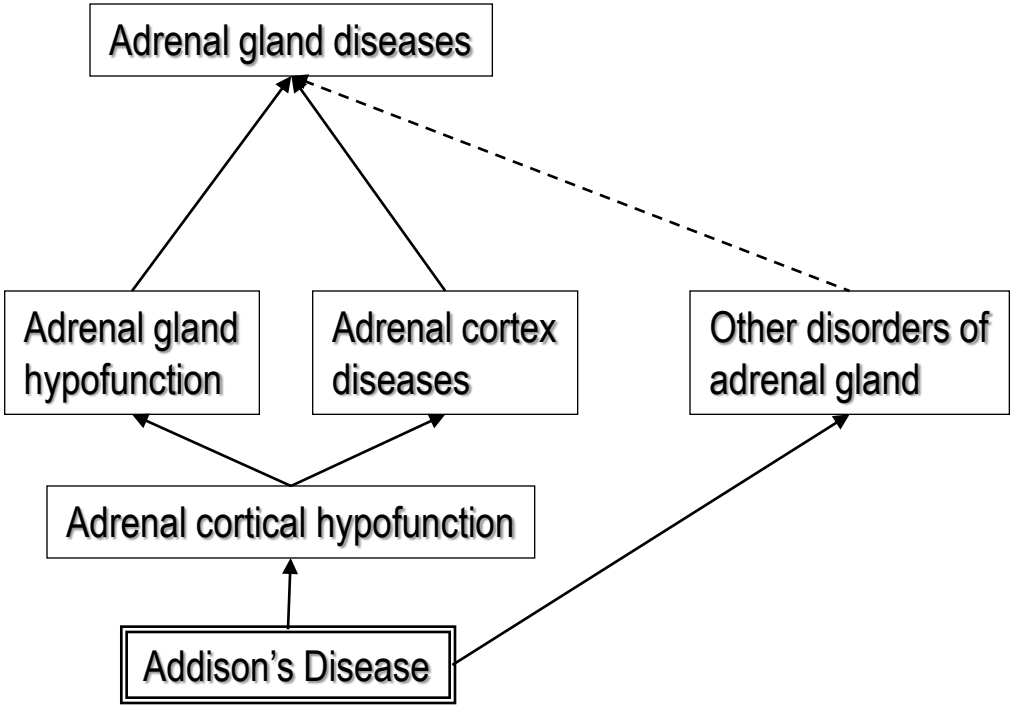
C0001621

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

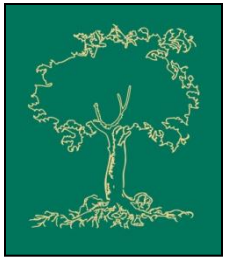
- Synonymous terms clustered into concepts
- Unique identifier

- 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

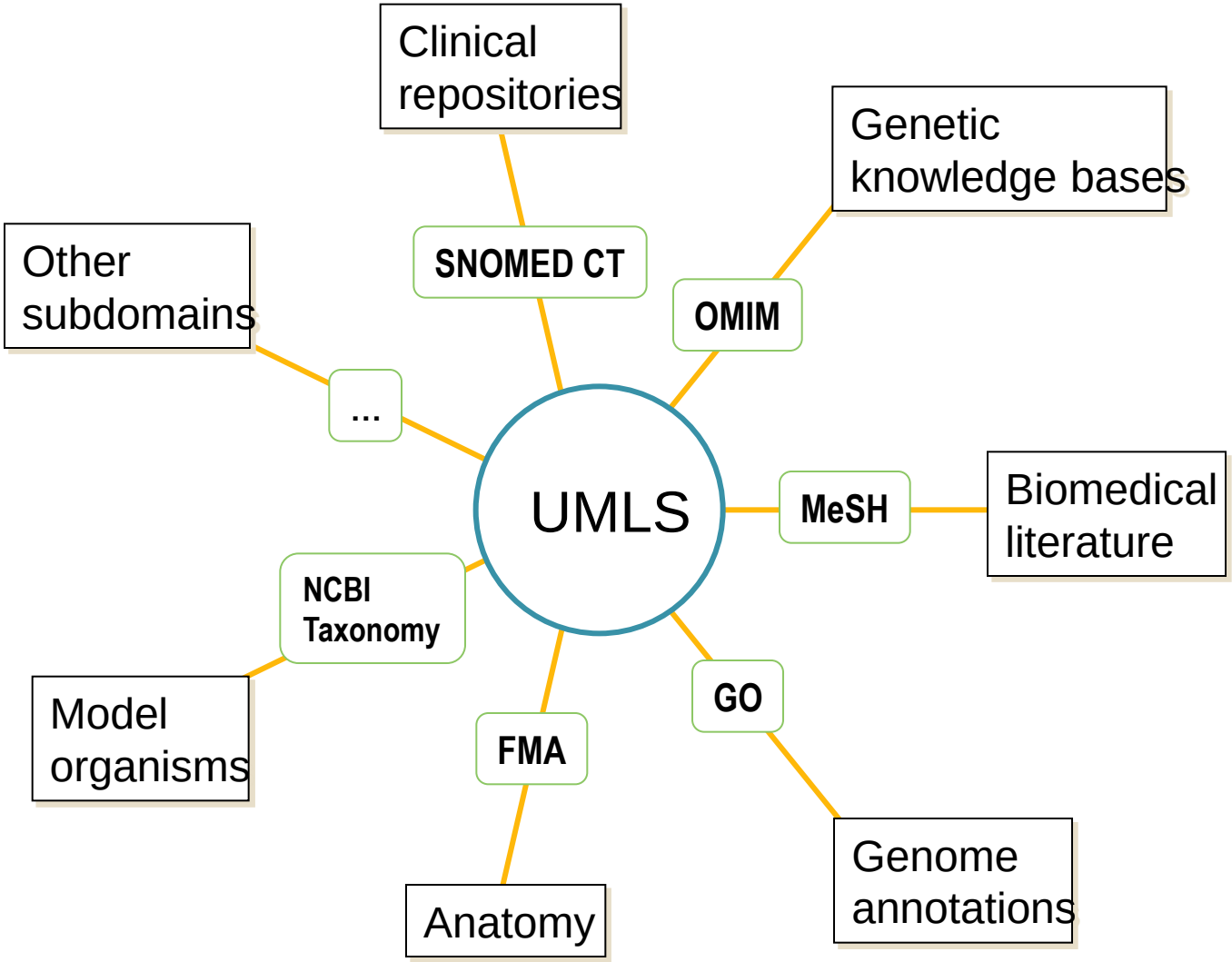
Biomedical terminologies

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

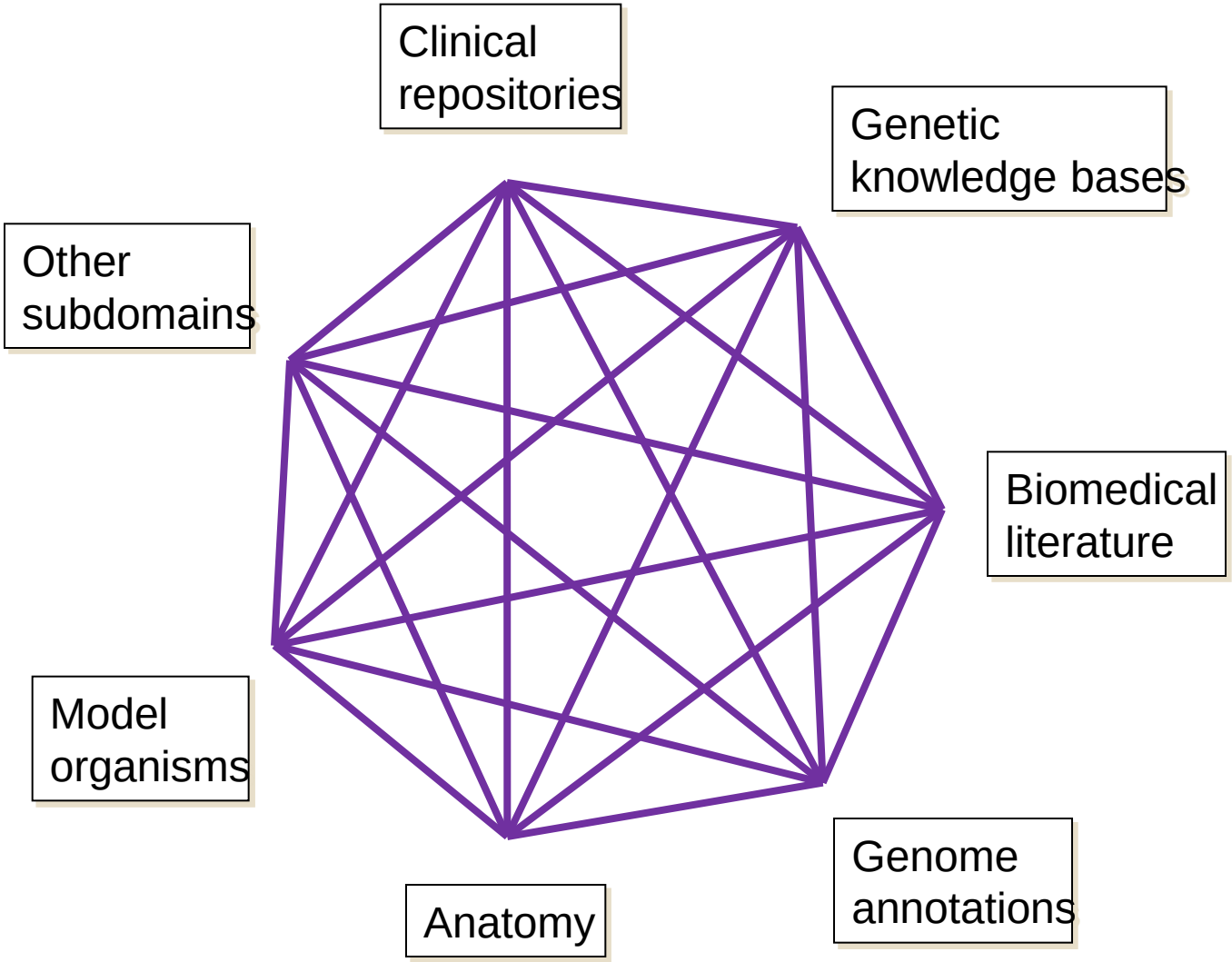
Biomedical terminologies (cont'd)

- Specialized vocabularies
  - nursing (NIC, NOC, NANDA, Omaha, PCDS)
  - 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)

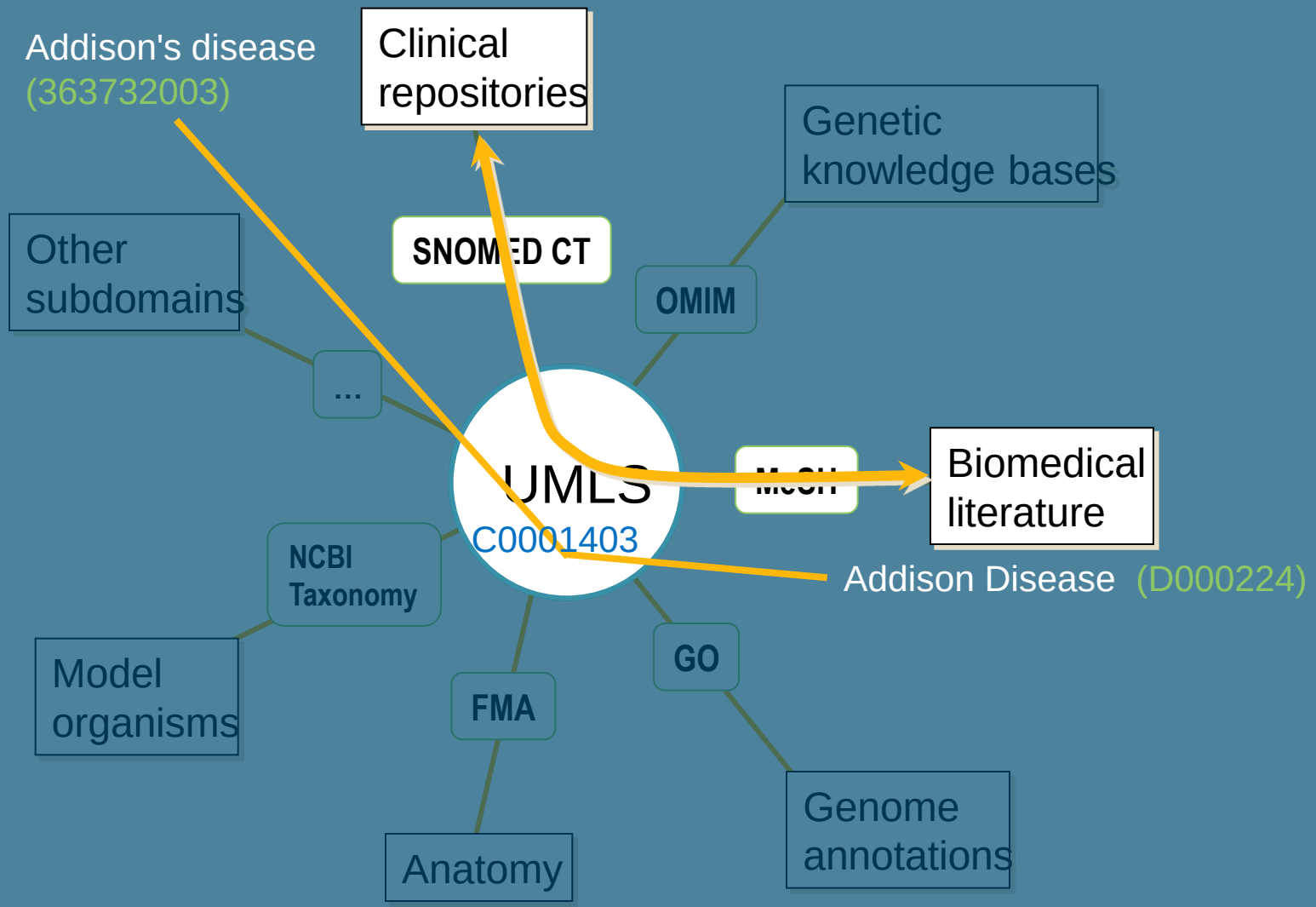
Integrating subdomains



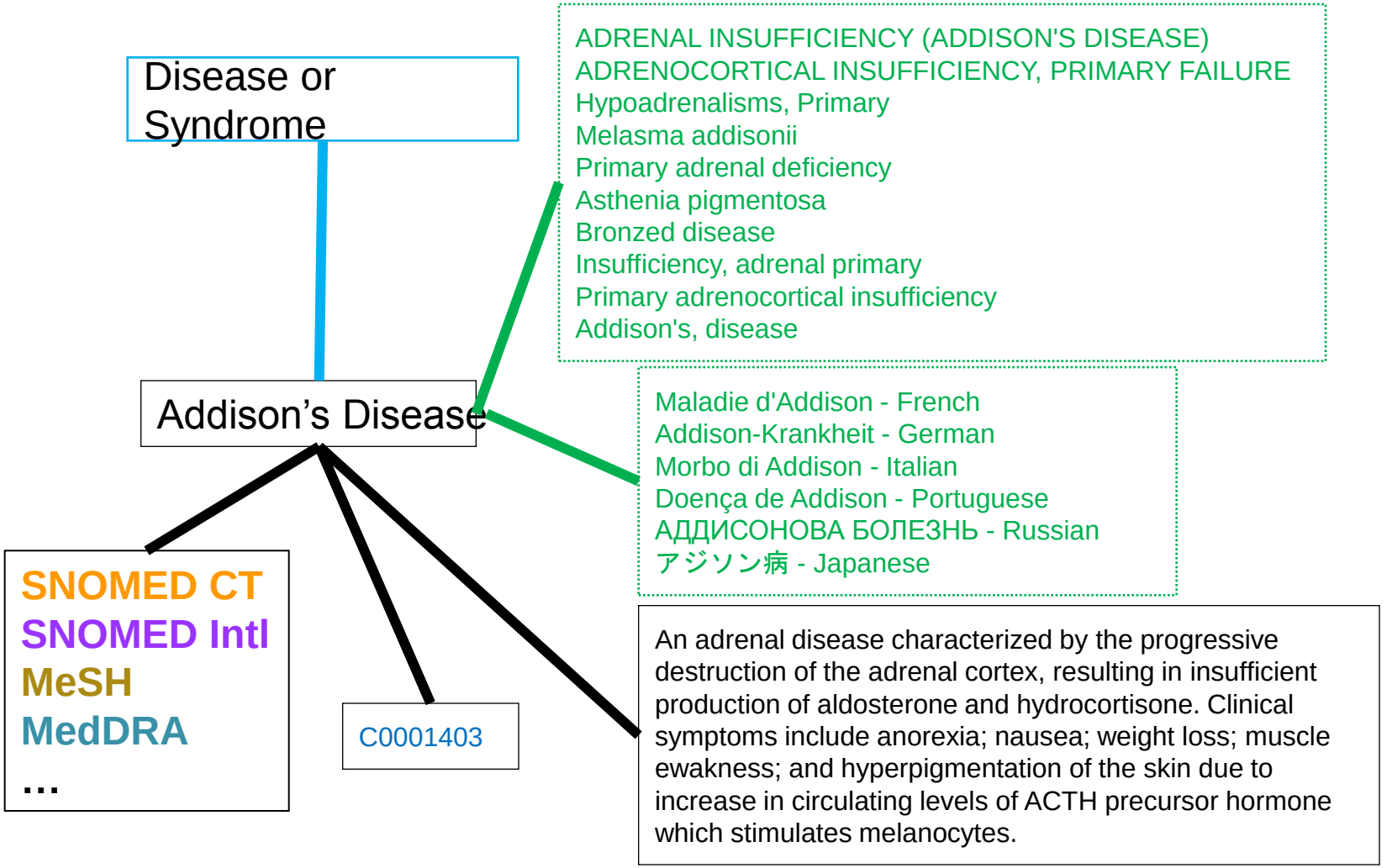
Integrating subdomains



Trans-namespace integration

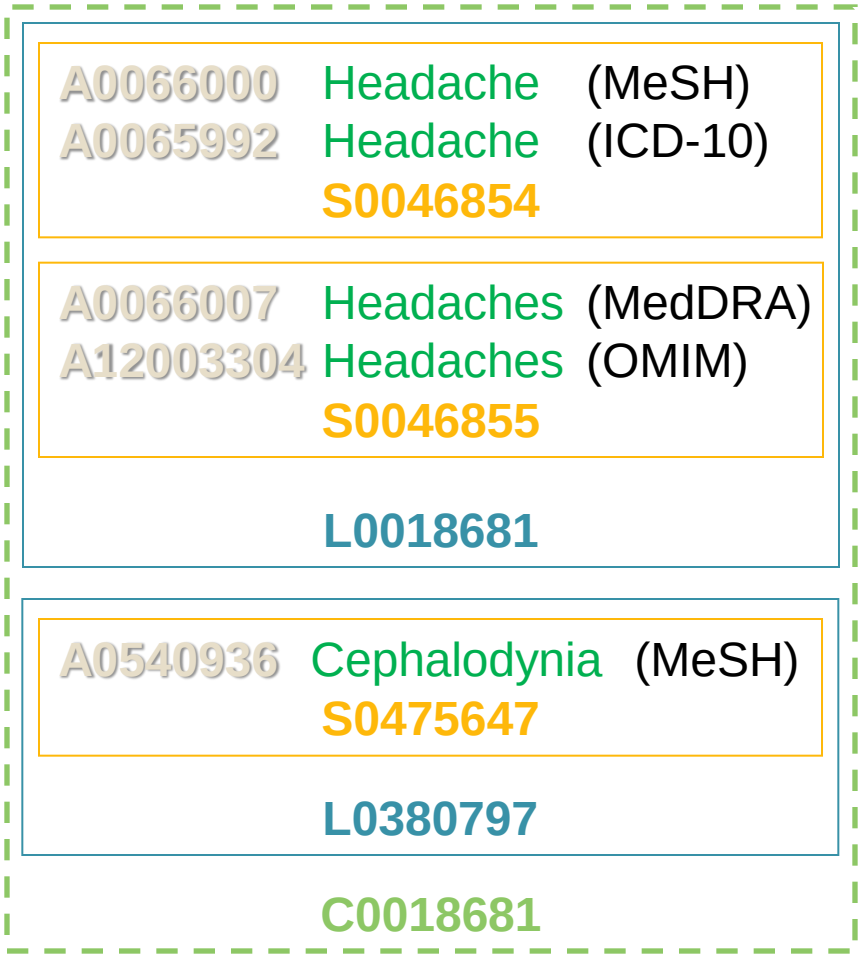


Addison's Disease: Concept



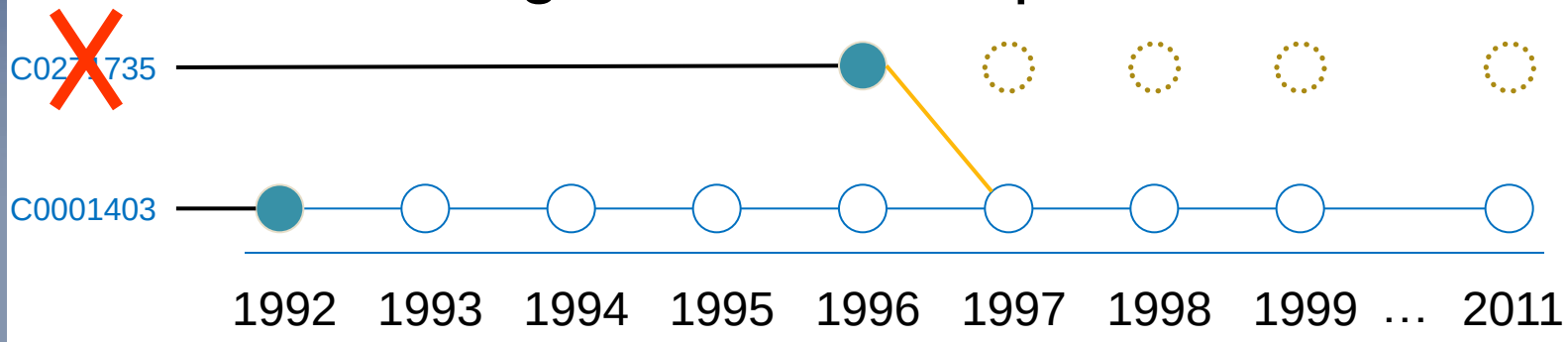
Metathesaurus Concepts (2011AA)

- 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



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

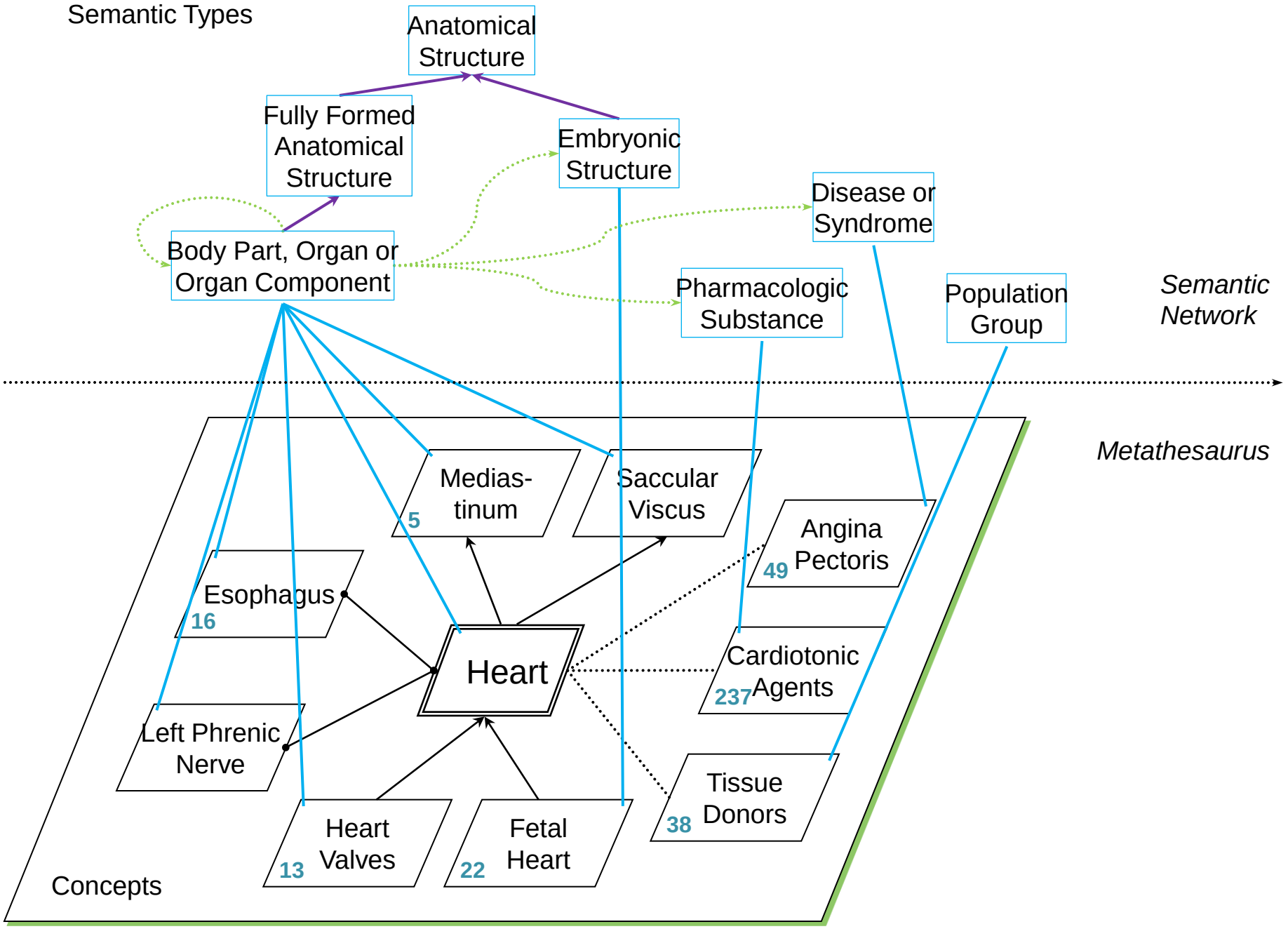
Mapping relations

- 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)
  - …


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
5
Saccular Viscus
Angina Pectoris
49
Esophagus
16
Heart
Cardiotonic Agents
237
Left Phrenic Nerve
Tissue Donors
38
Heart Valves
13
Fetal Heart
22
Concepts

UMLS Semantic Network

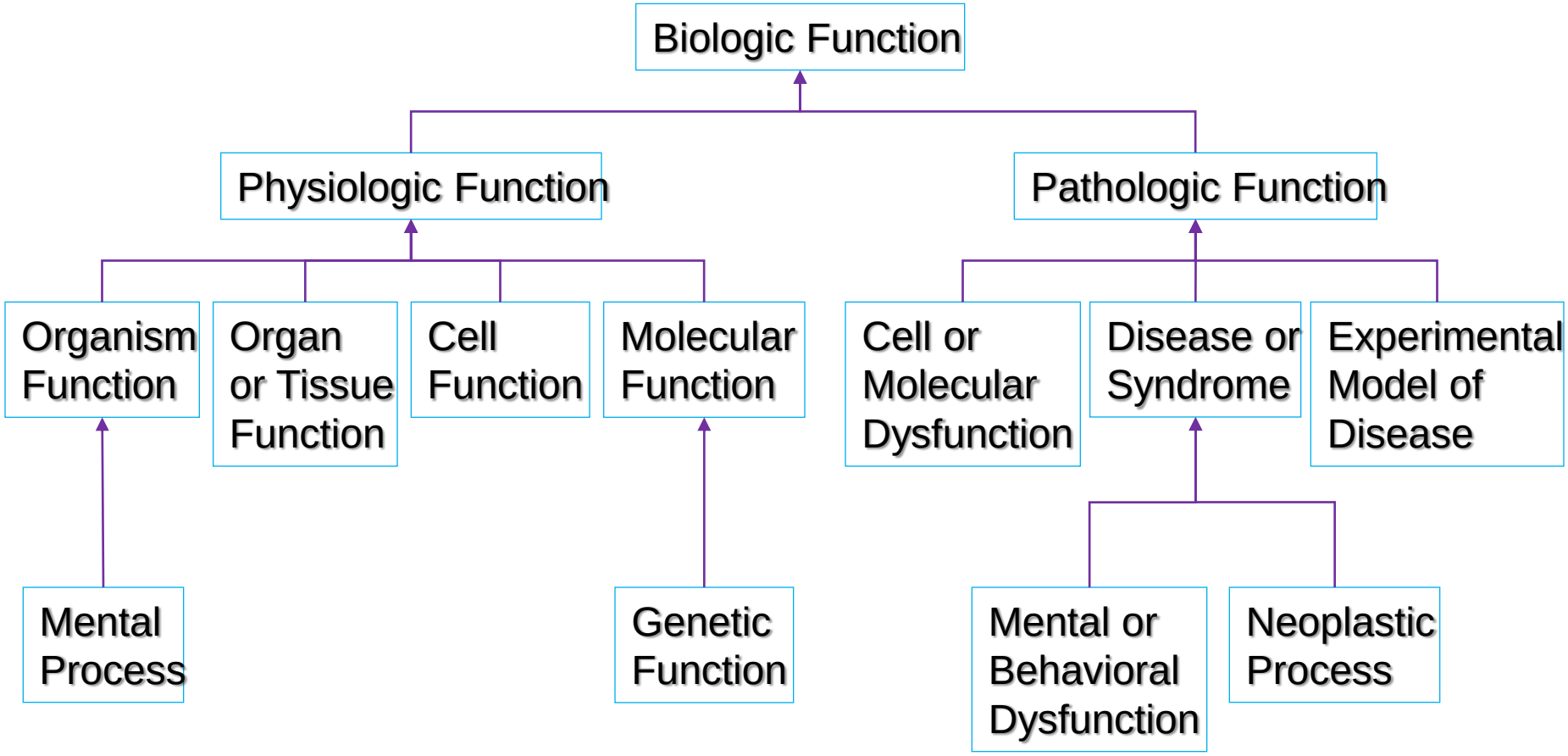
Semantic Network

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

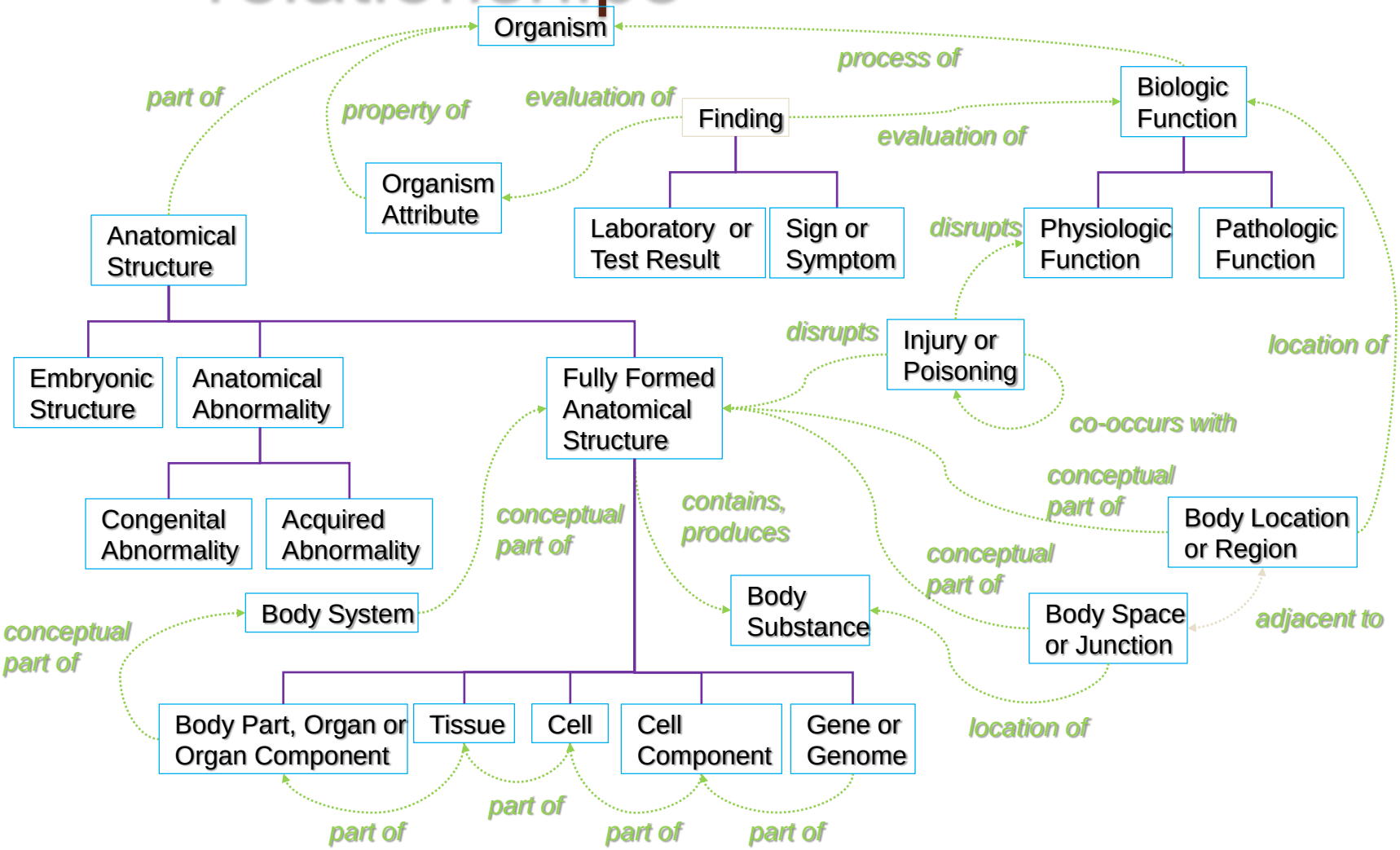
Semantic Network

- 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

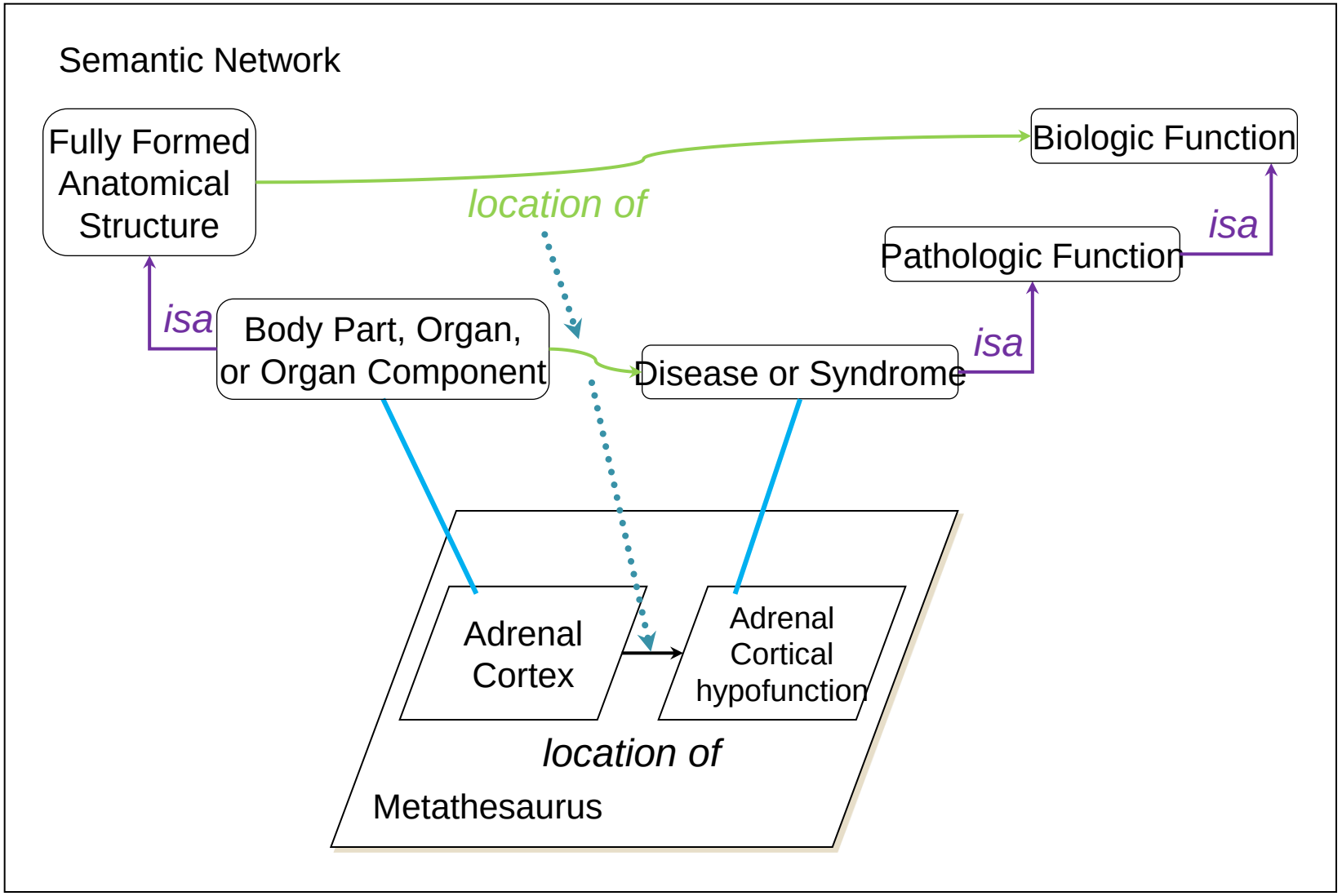


Why a semantic network?

- 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
  - verb
  - adjective
- Derivation
  - verb noun
  - adjective noun

Orthography

- Spelling variants
  - oe/e
  - ae/e
  - ise/ize
  - genitive mark

oe e

ae e

ise ize

's

s

Syntax

- Complementation
  - verbs
    - intransitive
    - transitive
    - ditransitive
  - nouns
    - prepositional phrase

treat

treated the patient

treated the patient with a drug

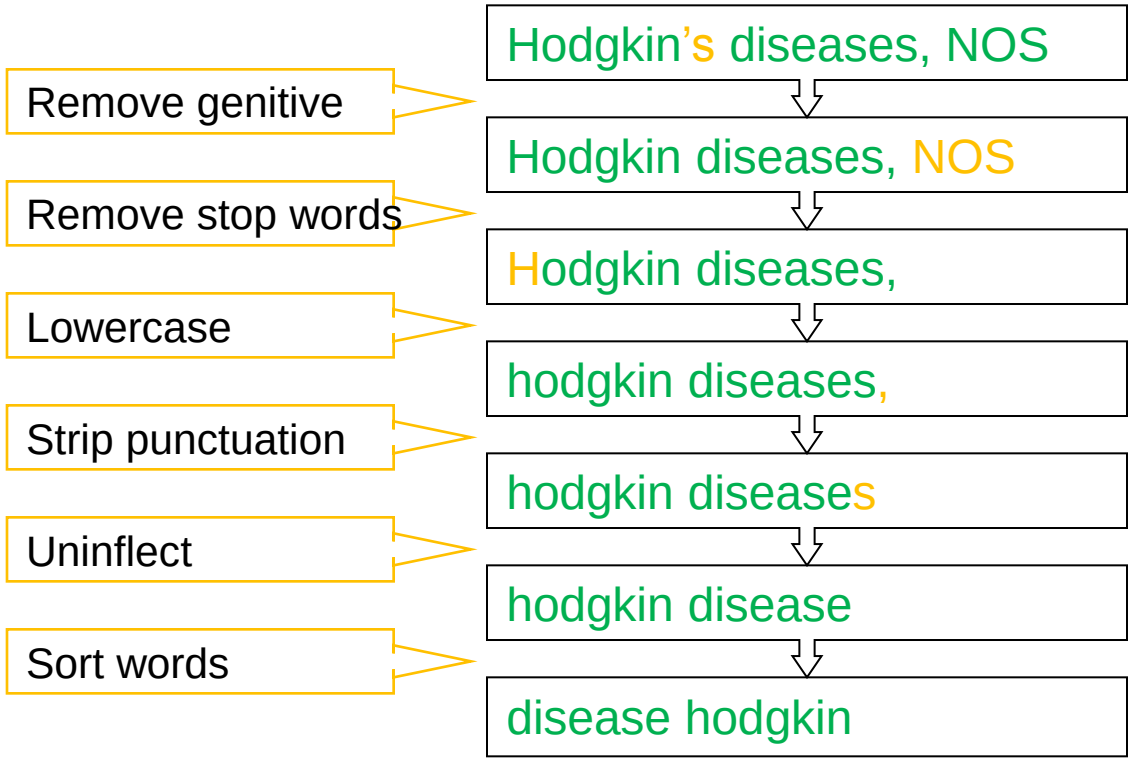
Valve of coronary sinus

- 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

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

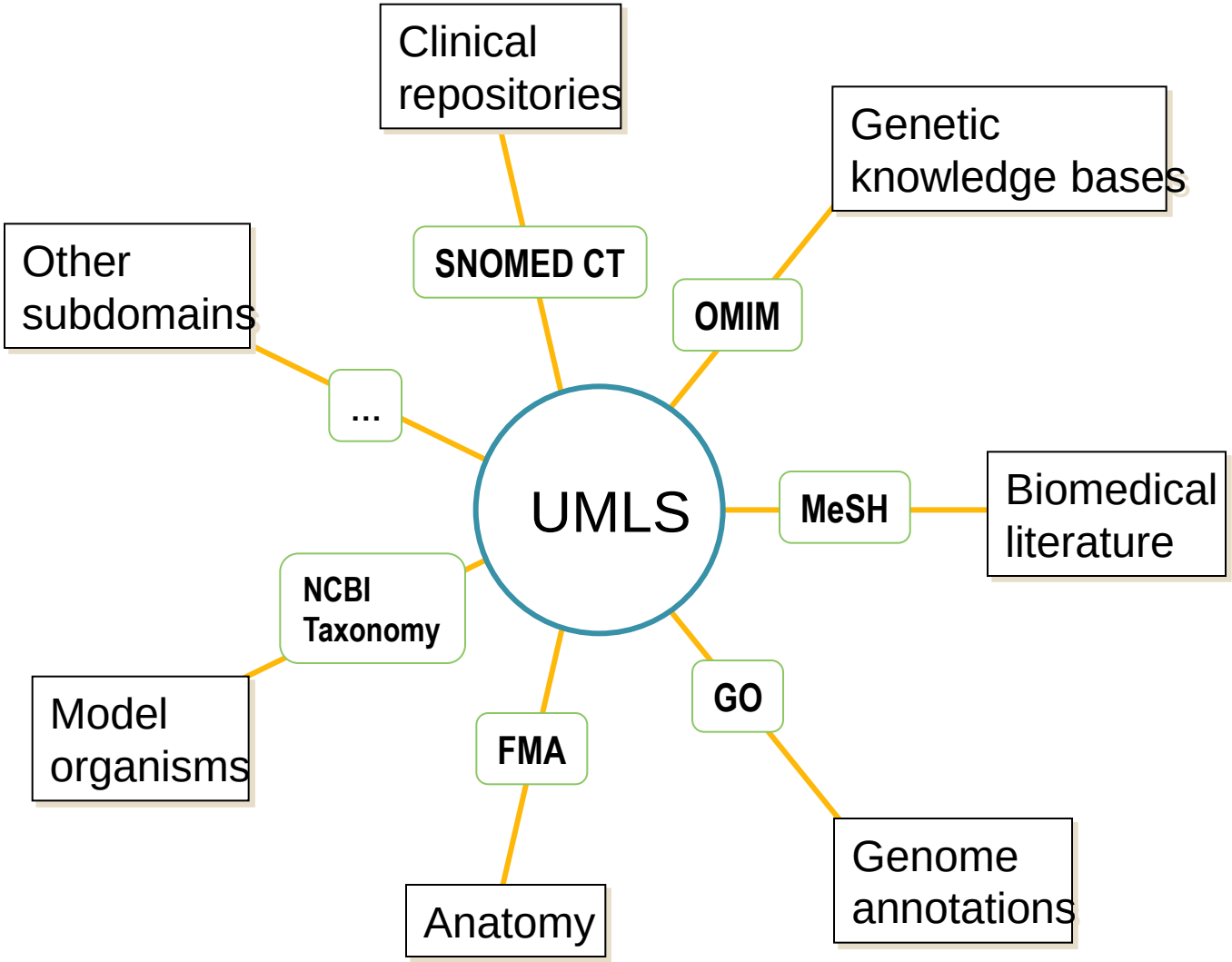
Lexical Variant Generation program

- 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)

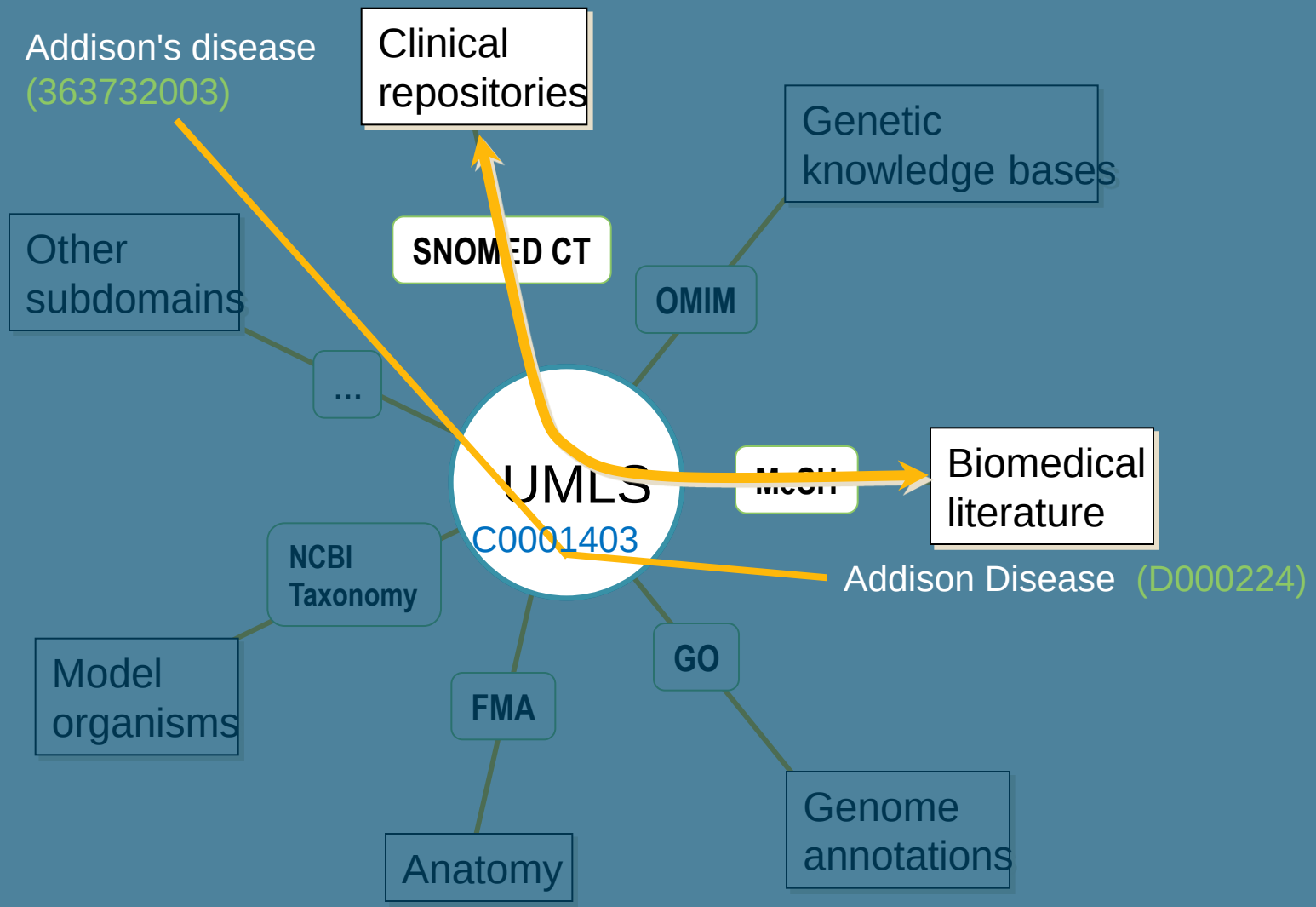
Overview of the UMLS

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) (formerly, UMLS Knowledge Source Server)
  - https://uts.nlm.nih.gov/

Dr. Kin Wah Fung

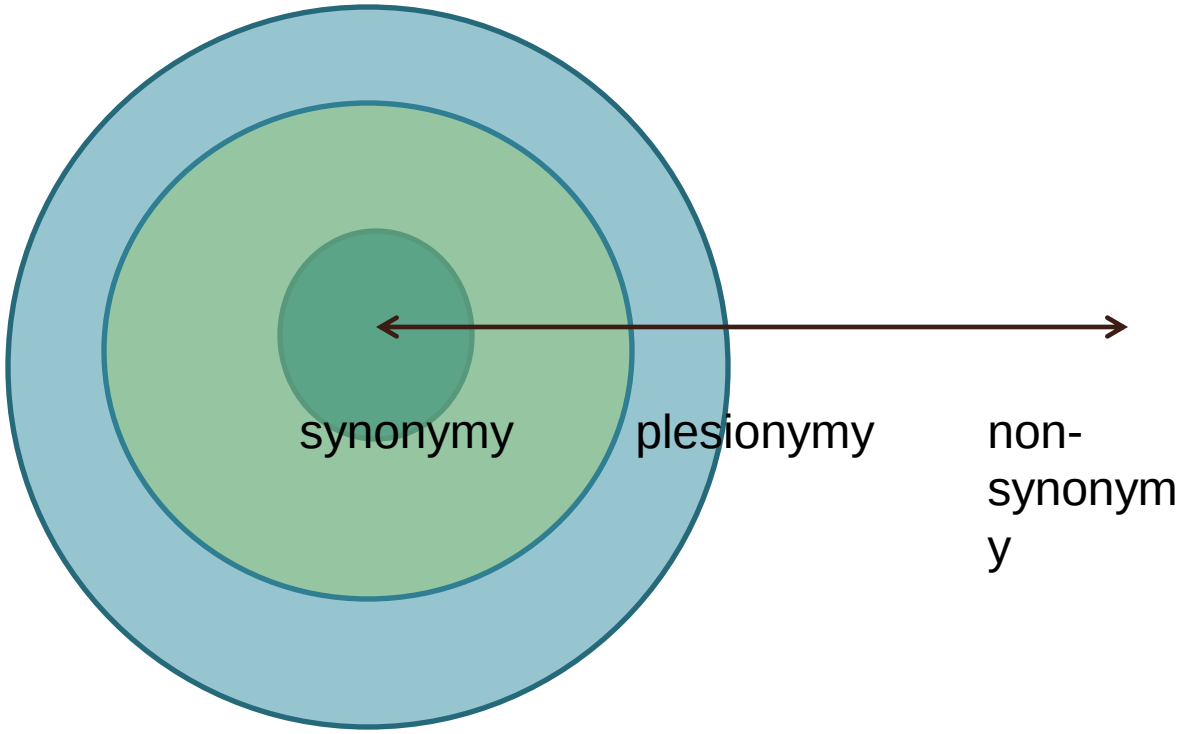
SNOMED CT integration inTO UMLS

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

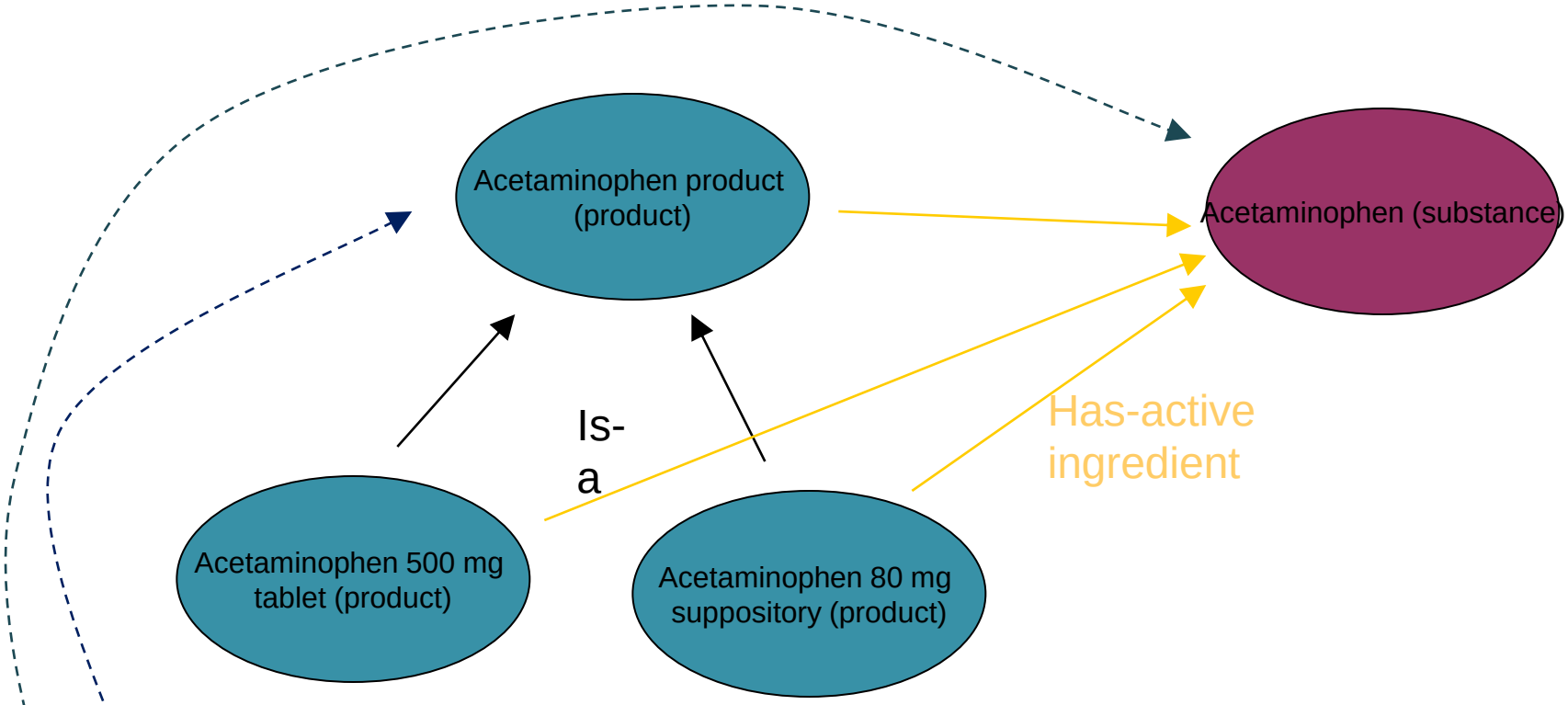
Fully-specified name	Synonym
Adolescence (finding)	Youth
Blinking (finding)	Winking
Candidiasis (disorder)	Thrush
Motor vehicle accident victim (finding)	Motor vehicle accident
Eczema (disorder)	Dermatitis

- 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

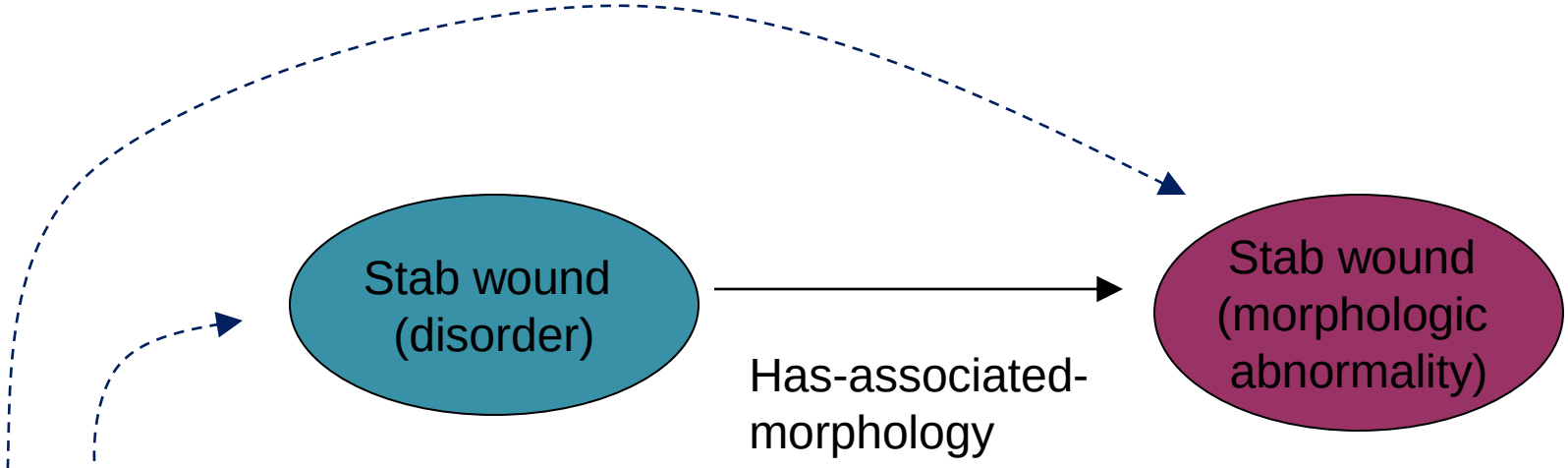
SNOMED CT Concept View



‘This patient is on acetaminophen.’

‘Tylenol contains acetaminophen.’

SNOMED CT Concept View



‘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 B>12< 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|

271737000|0|Anemia (disorder)|XM05A|DC-10009|1|

MRCONSO.RRF:

CUI|…|AUI|…|SCUI|…|SAB|TTY|…|STR|…|

C0002871|A3597593|271737000|SNOMEDCT|FN|Anemia (disorder)|

C0002871|A2878480|271737000|SNOMEDCT|PT|Anemia|

C0002871|A2952250|271737000|SNOMEDCT|SY|Absolute anemia|

C0002871|A3095181|271737000|SNOMEDCT|PTGB|Anaemia|

C0002871|A3089808|271737000|SNOMEDCT|SYGB|Absolute anaemia|

MRSAT.RRF:

CUI|…|STYPE|CODE|…|ATN|SAB|ATV|…|

C0002871|SCUI|271737000|CONCEPTSTATUS|SNOMEDCT|0|

C0002871|SCUI|271737000|CTV3ID|SNOMEDCT|XM05A|

C0002871|SCUI|271737000|SNOMEDID|SNOMEDCT|DC-10009|

C0002871|SCUI|271737000|ISPRIMITIVE|SNOMEDCT|1|

SNOMED CT Descriptions

SNOMED CT Descriptions Table:

DescriptionId|DescriptionStatus|ConceptId|Term|InitialCapitalStatus|DescriptionTyp
e|
LanguageCode|

406636013|0|271737000|Anemia|0|1|en-US|

MRCONSO.RRF:

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

C0002871|A2878480|406636013|271737000|SNOMEDCT|PT|Anemia|

MRSAT.RRF:

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

C0002871|A2878480|SAUI|271737000|DESCRIPTIONSTATUS|SNOMEDCT|0|

C0002871|A2878480|SAUI|271737000|INITIALCAPITALSTATUS|SNOMEDCT|0|

C0002871|A2878480|SAUI|271737000|DESCRIPTIONTYPE|SNOMEDCT|1|

C0002871|A2878480|SAUI|271737000|LANGUAGECODE|SNOMEDCT|en-US|

SNOMED CT Relationships

SNOMED CT Relationships Table

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

85555020|263245004|116680003|15574005|0|0|0|

MRCONSO

CUI|…|AUI|SCUI|…|SAB|TTY|…|STR|…|

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

C1292718|A3524752|116680003|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|CHARACTERISTICTYPE|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

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

SNOMED CT QA
What we learn from maintaining the UMLS 2

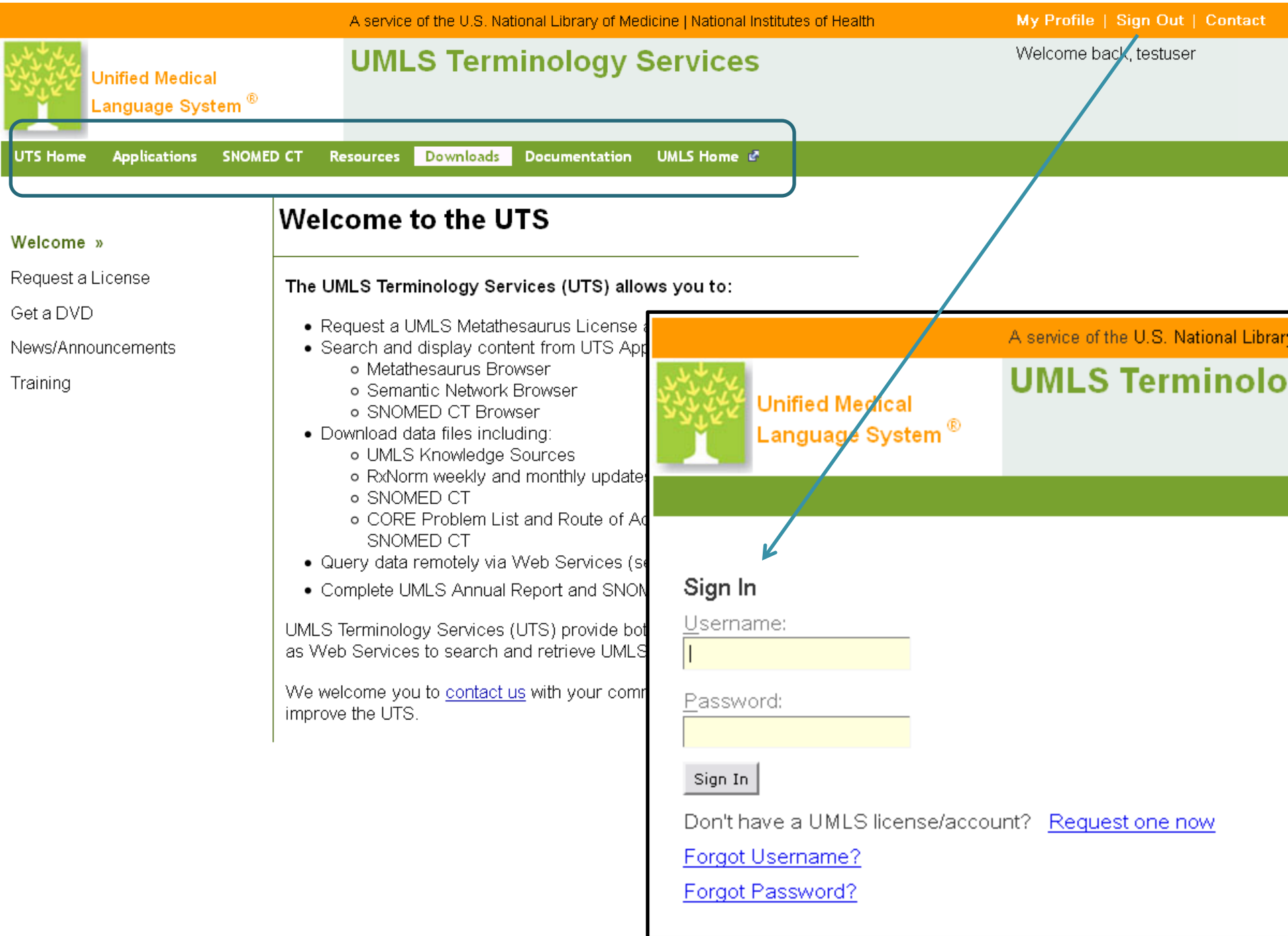
- UMLS Release QA - 2nd round of review, counts, comparisons
  - Individual sources (SNOMED CT) and all sources integrated into Metathesaurus concept structure
- Update Web-based documentation for each UMLS terminology
  - Stats, sample data, conversion specs and details of native and UMLS format
- User 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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RxNorm weekly and monthly update
SNOMED CT
CORE Problem List and Route of Ac
SNOMED CT
Query data remotely via Web Services (s
Complete UMLS Annual Report and SNOM
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as Web Services to search and retrieve UMLS
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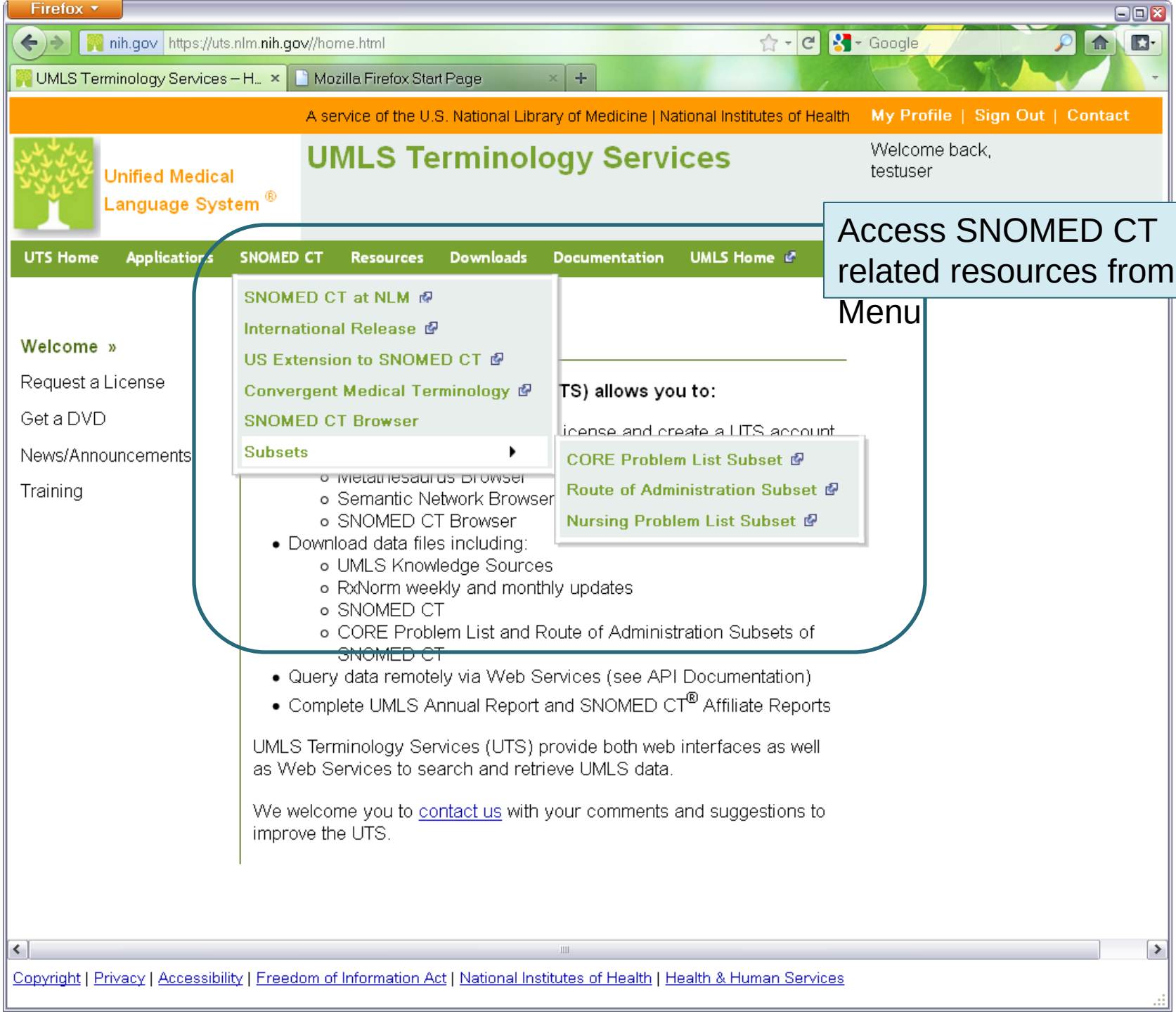
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CORE Problem List Subset
Route of Administration Subset
Nursing Problem List Subset
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Training
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icense and create a UTS account
Semantic Network 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.
We welcome you to contact us with your comments and suggestions to improve the UTS.
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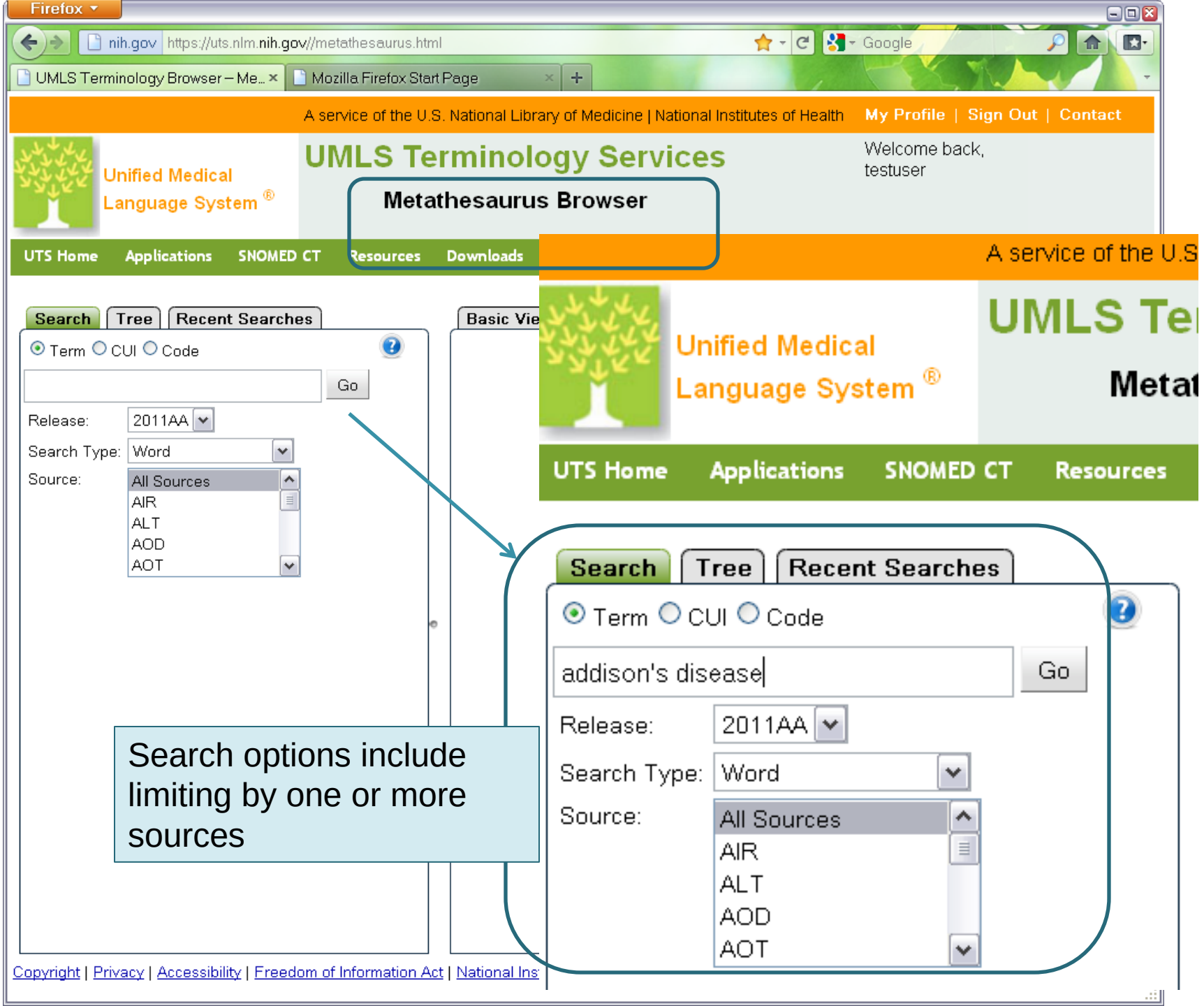
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UMLS Terminology Services
Metathesaurus Browser
Welcome back, testuser
UTS Home
Applications
SNOMED CT
Resources
Downloads
Search
Tree
Recent Searches
Term CUI Code
Go
Release: 2011AA
Search Type: Word
Source:
All Sources
AIR
ALT
AOD
AOT
addison's disease
Search options include limiting by one or more sources
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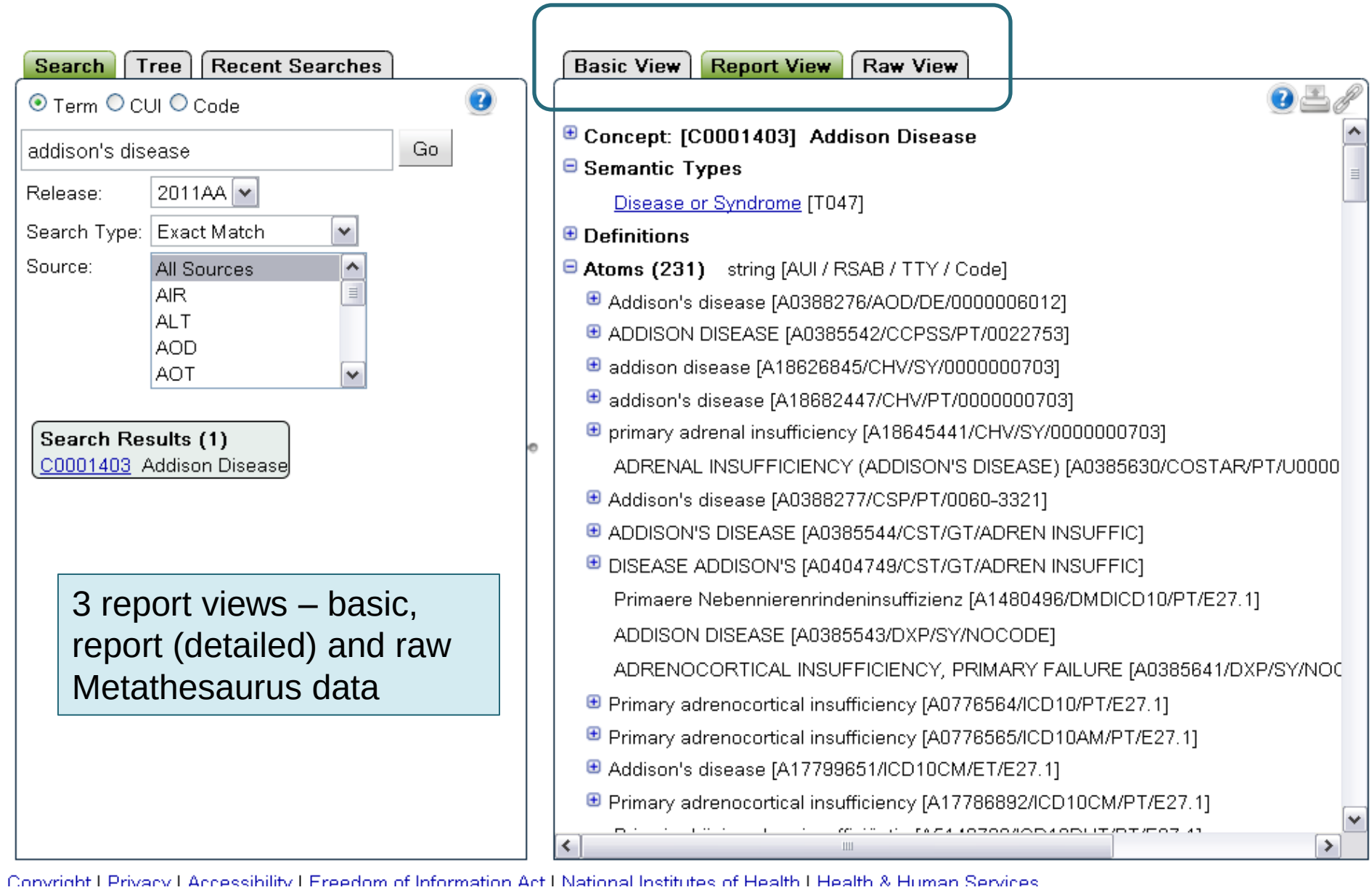
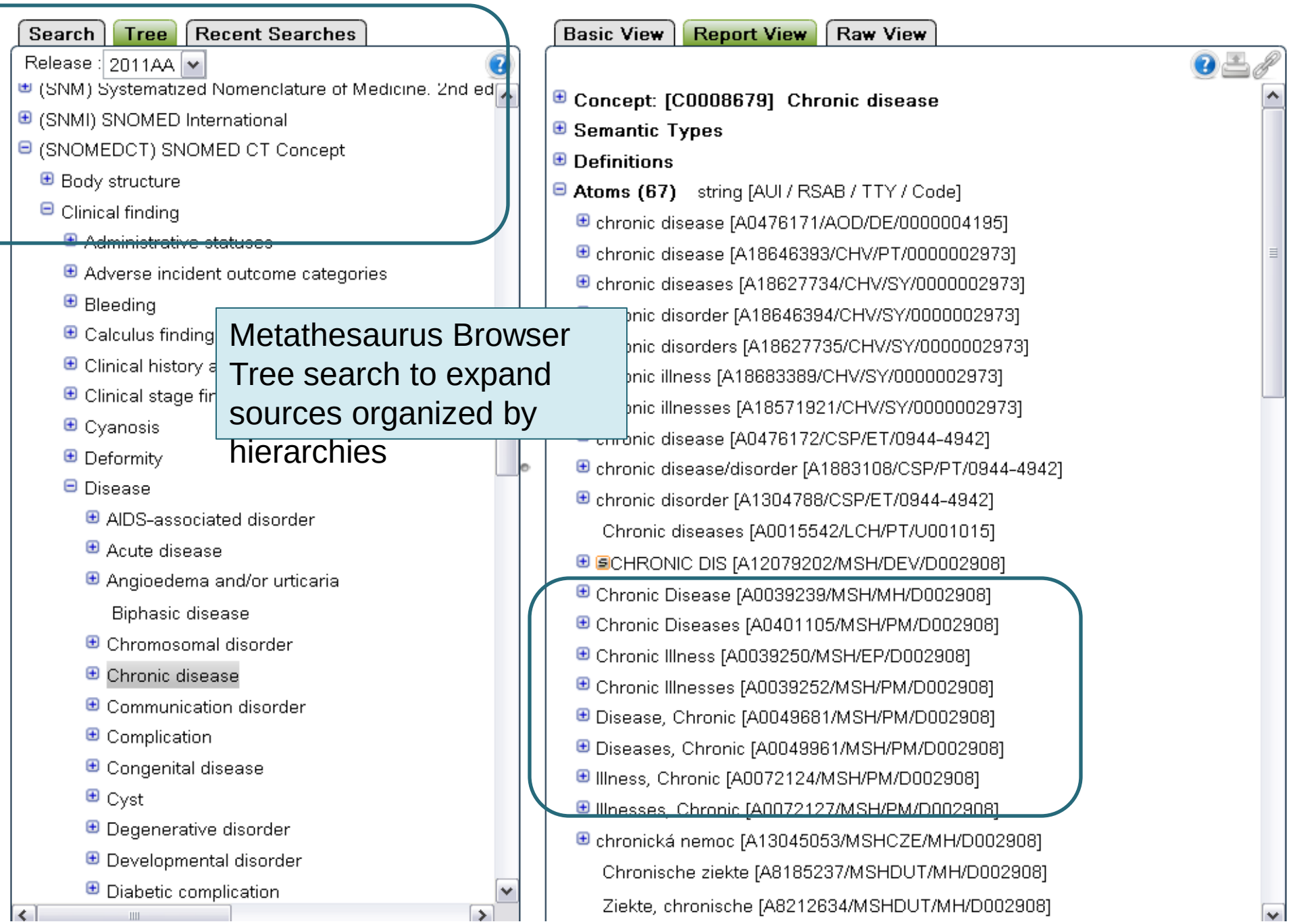
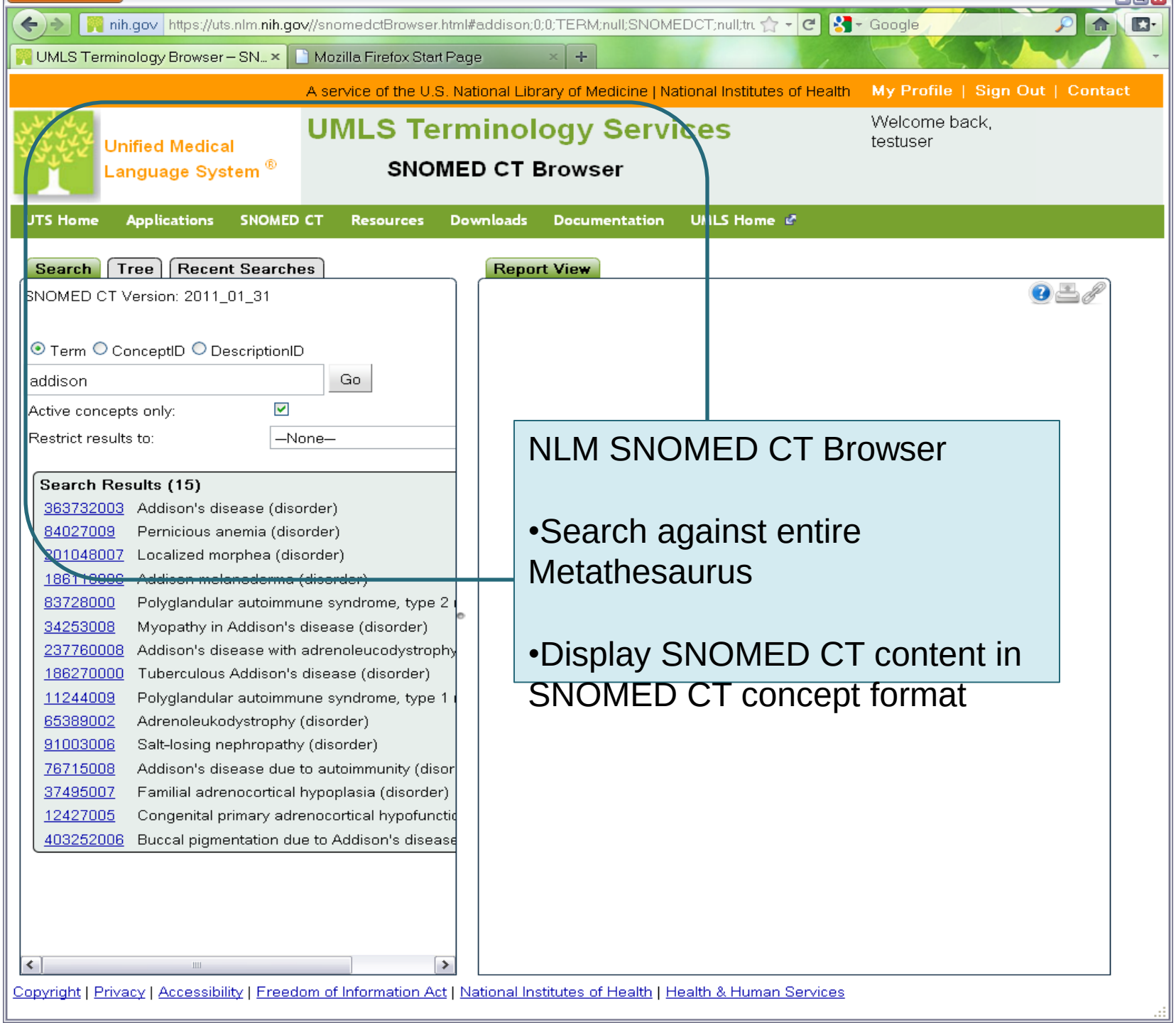


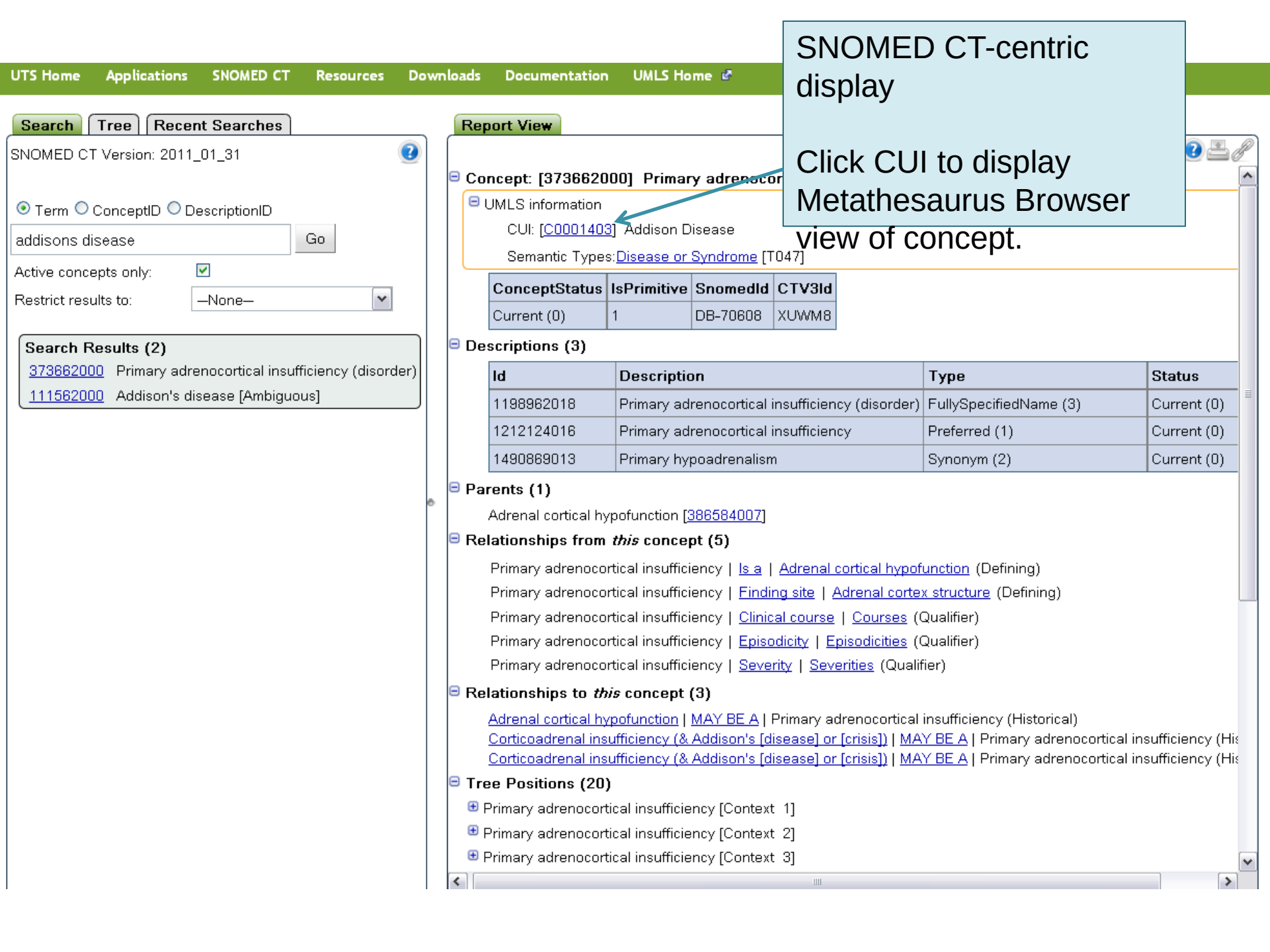
3 report views – basic, report (detailed) and raw Metathesaurus data


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 fi
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]
nic disorder [A18646394/CHV/SY/0000002973]
nic disorders [A18627735/CHV/SY/0000002973]
nic illness [A18683389/CHV/SY/0000002973]
nic illnesses [A18571921/CHV/SY/0000002973]
nic 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]



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.
UTS Home
Applications
SNOMED CT
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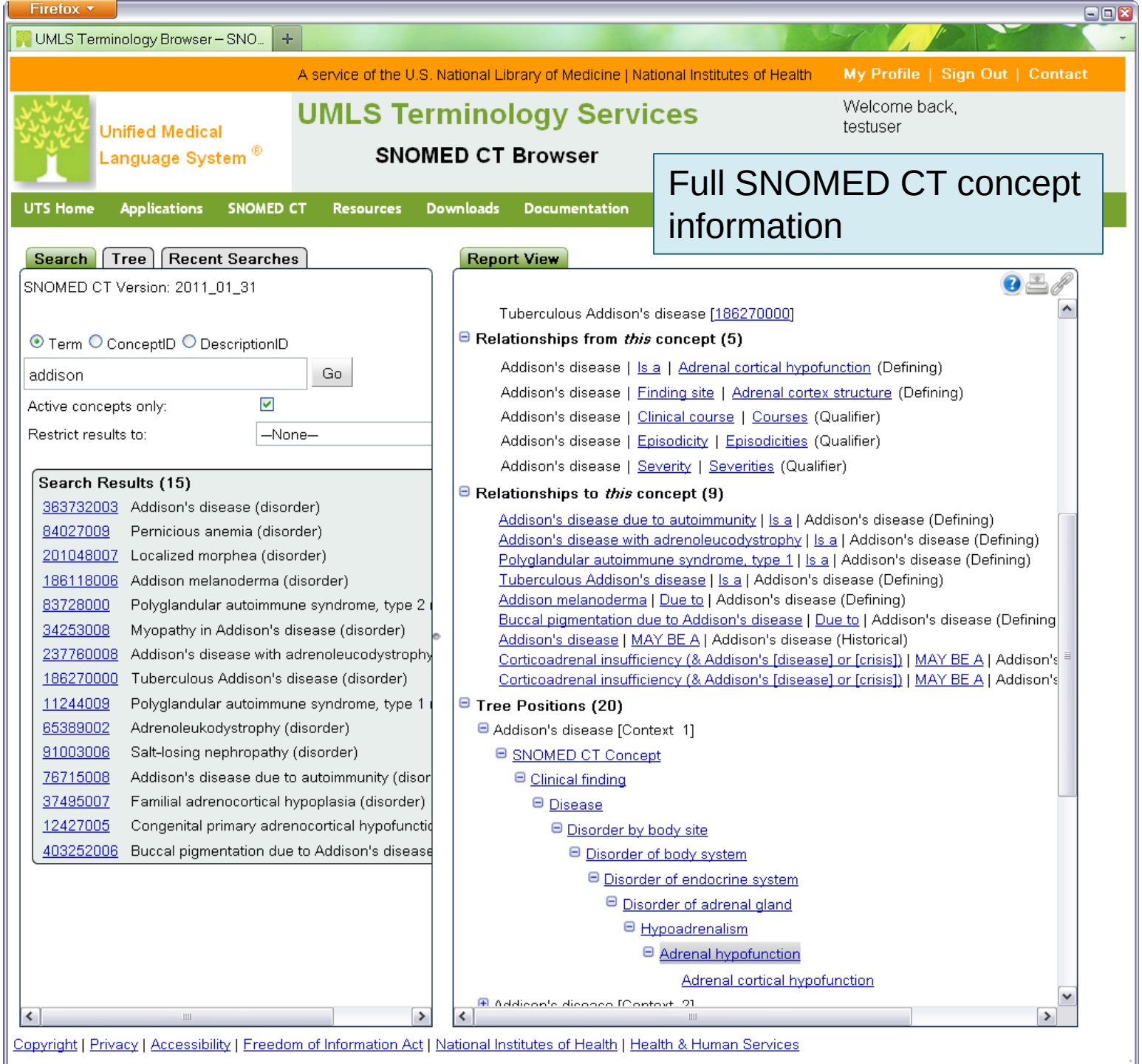
UMLS Home
Search
Tree
Recent Searches
SNOMED CT Version: 2011_01_31
Term
ConceptID
DescriptionID
addisons disease
Go
Active concepts only:
Restrict results to:
—None—
Search Results (2)
373662000 Primary adrenocortical insufficiency (disorder)
111562000 Addison's disease [Ambiguous]
Report View
Concept: [373662000] Primary adrenocor
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 (His
Corticoadrenal insufficiency (& Addison's [disease] or [crisis]) | MAY BE A | Primary adrenocortical insufficiency (His
Tree Positions (20)
Primary adrenocortical insufficiency [Context 1]
Primary adrenocortical insufficiency [Context 2]
Primary adrenocortical insufficiency [Context 3]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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 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
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
Corticoadrenal insufficiency (& Addison's [disease] or [crisis]) | MAY BE A | Addison's
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
Addison's disease [Context 2]
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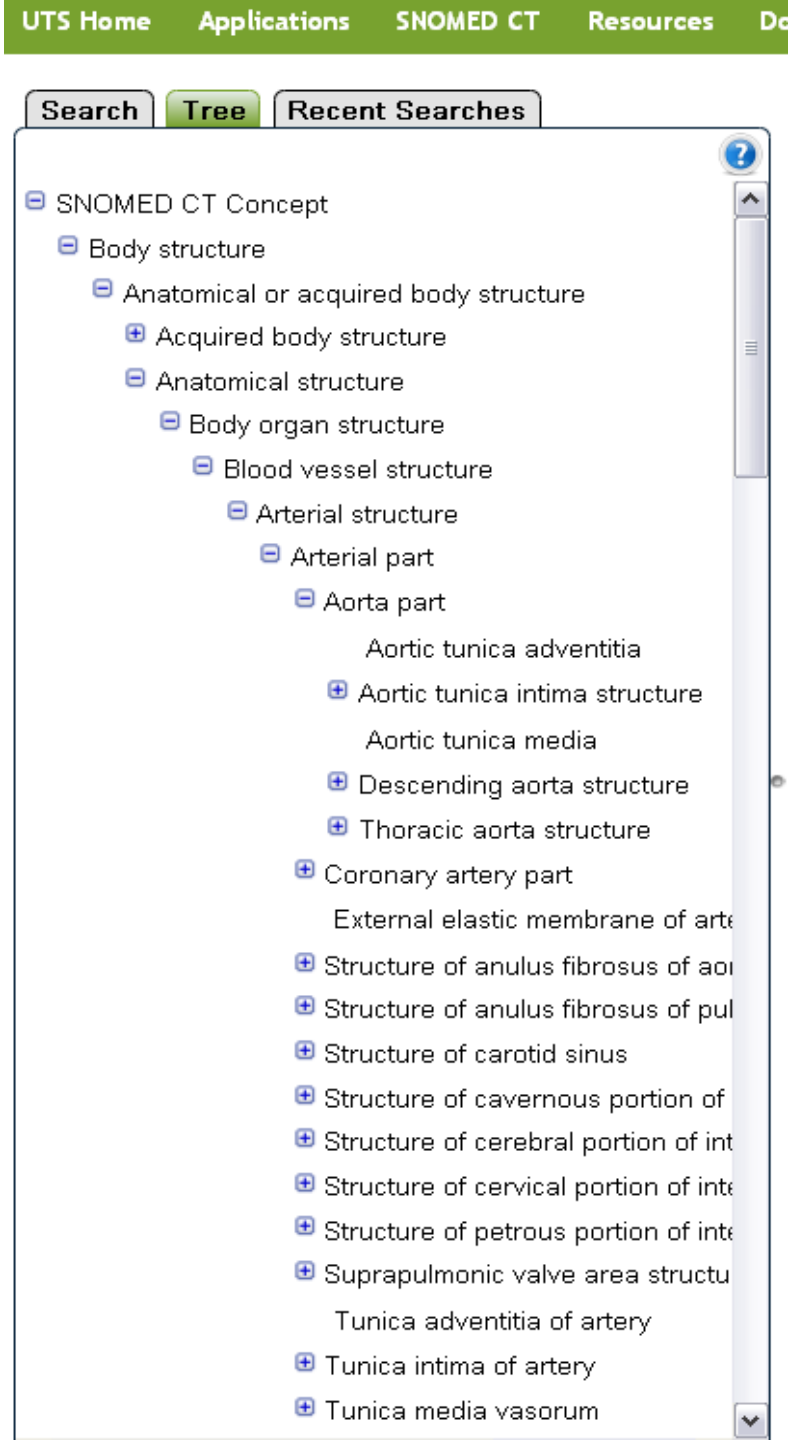

Metathesaurus Browser Tree search display. Click to expand the nodes in the hierarchies.

2011AA SNOMED CT Source Information

Web-based documentation for each source in the UMLS Metathesaurus

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 atribute 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.

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First published: 17 May 2011
Metadata | Permanence level: Permanence Not Guaranteed

2011AA SNOMED CT Source Information

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

see more samples (Click and hold to drag this window) X

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)

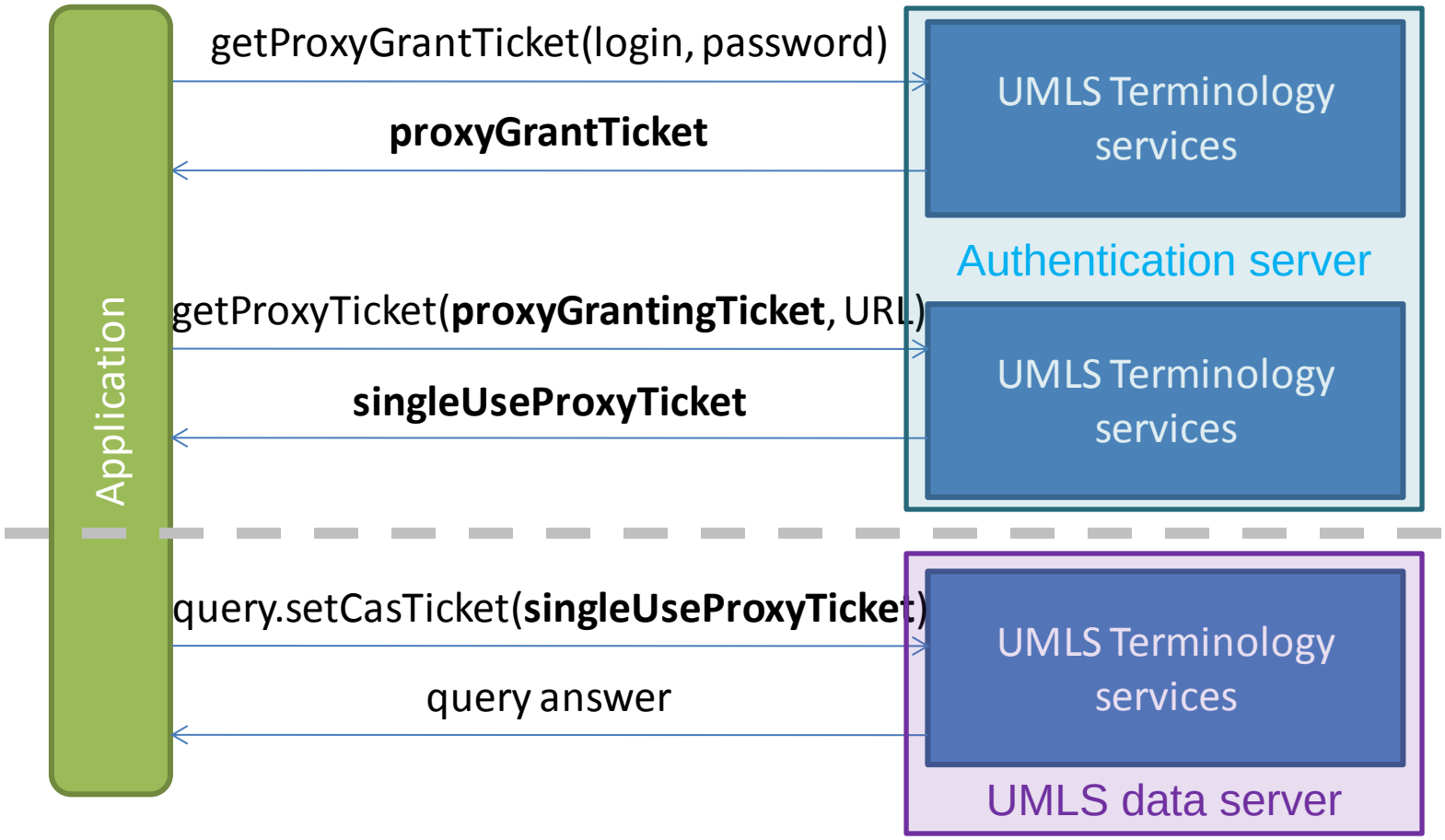
Dr. Olivier Bodenreider
(courtesy of Dr. Bastien Rance, NLM)

Accessing SNOMED CT content with the UTS API

Getting started with the UTS API

- Useful resources
  - UTS Website: signing up for a UMLS license, signing in, browsing the UMLS, access to technical documentations… 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 umlsksService
            = 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 = umlsksService.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

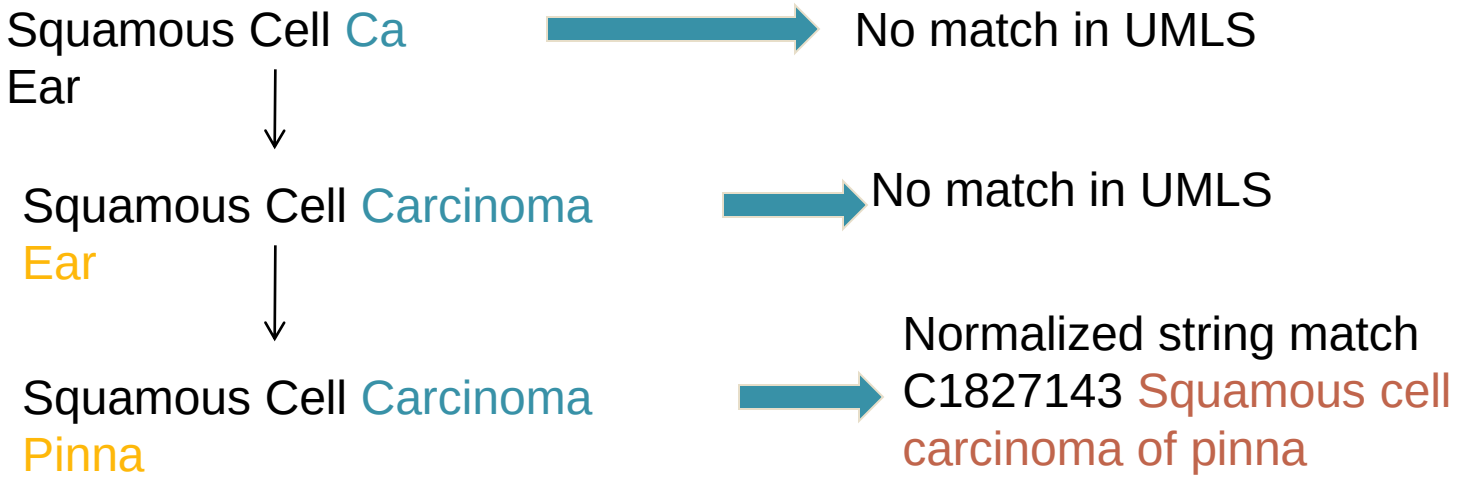
- Over 100 source vocabularies in the UMLS
- content organized according to meaning (concept-based organization) – an interlingua between terminologies
- rich network of relations between concepts – hierarchical relations and associative relations
- various lexical resources and lexical tools

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

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

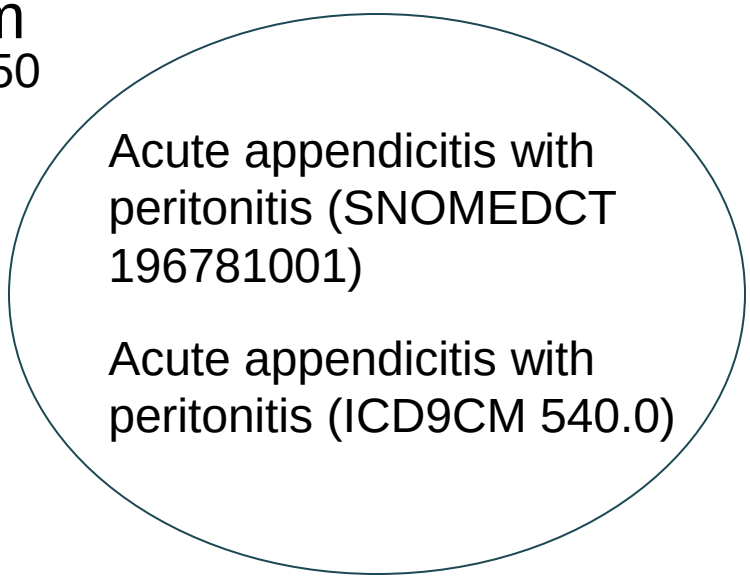
- Make use of the concept structure of the UMLS to identify equivalence between terminologies
- Utilize the relationships between UMLS concepts to improve yield

The IntraMap algorithm (1)

- 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

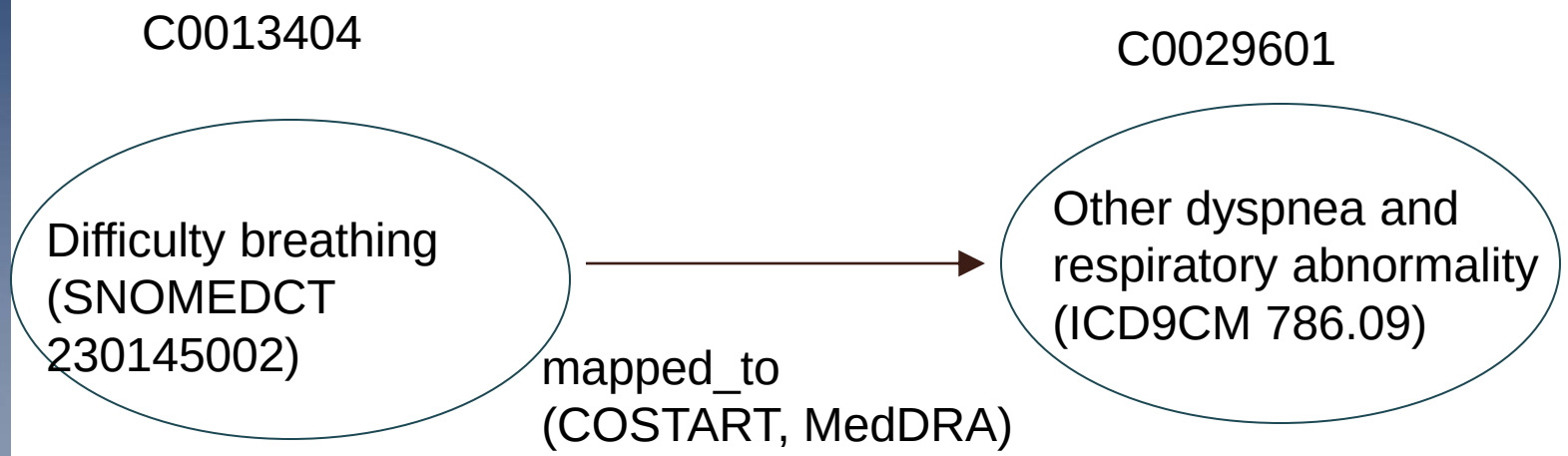
1. A target term is in the same concept as the source term

C0341350



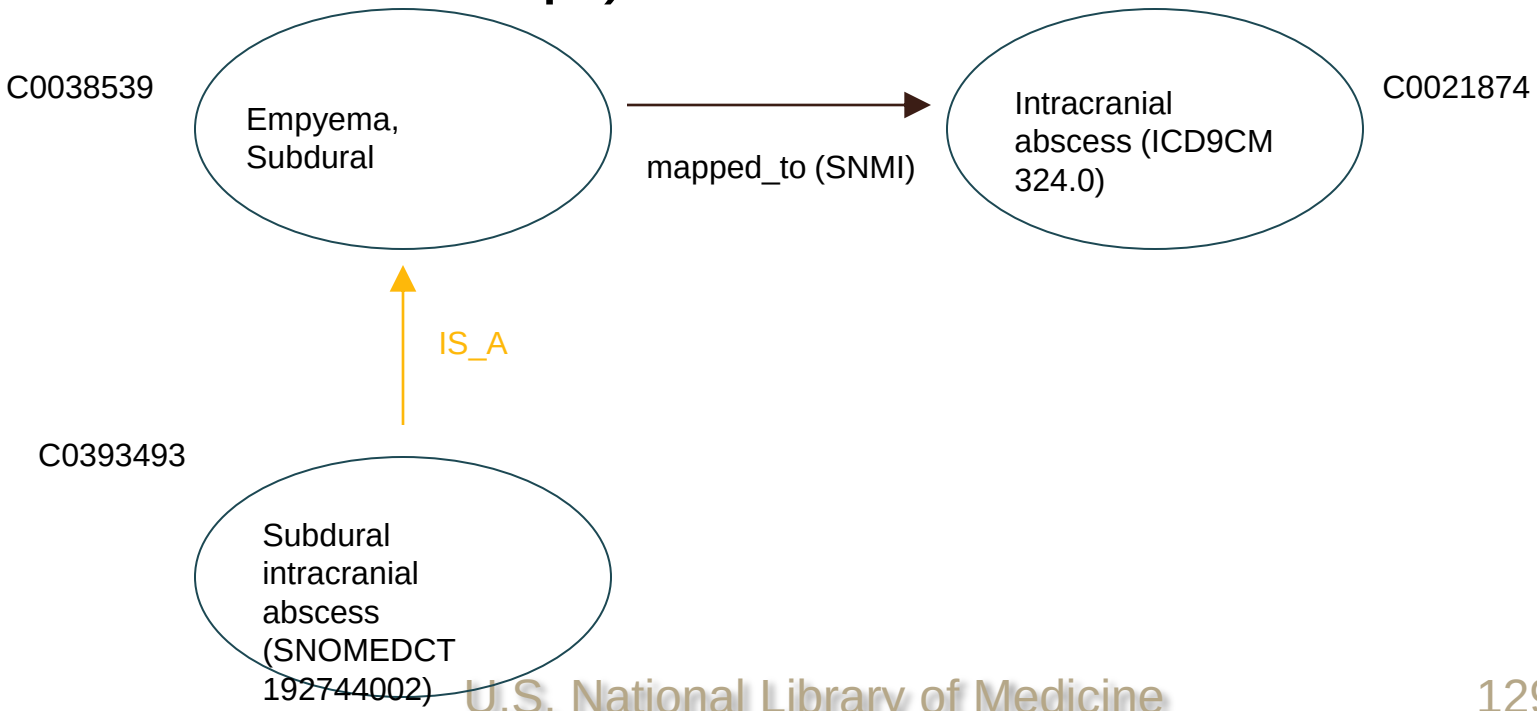
The IntraMap algorithm (2)

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

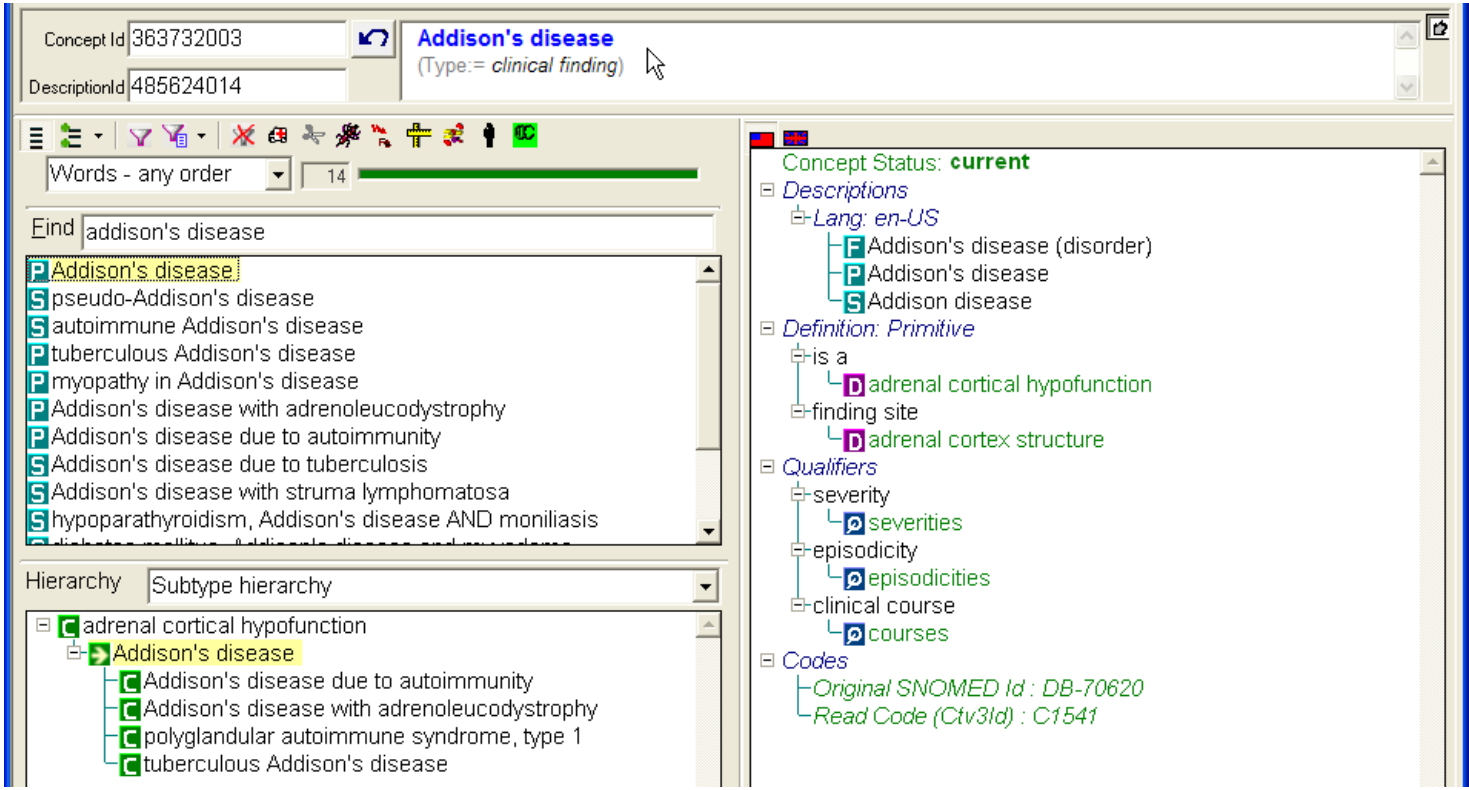
Language	MeSH	ICPC	MedDRA
BAQ		X	
CZE	X		X
DAN		X	
DUT	X	X	X
ENG	X	X	X
FIN	X	X	
FRE	X	X	X
GER	X	X	X
HEB		X	
HUN		X	
ITA	X	X	X
JPN	X		X
KOR			
LAV	X		
NOR		X	
POL	X		
POR	X	X	X
RUS	X		
SCR	X		
SPA	X	X	X
SWE	X	X	

Finding terms in other languages

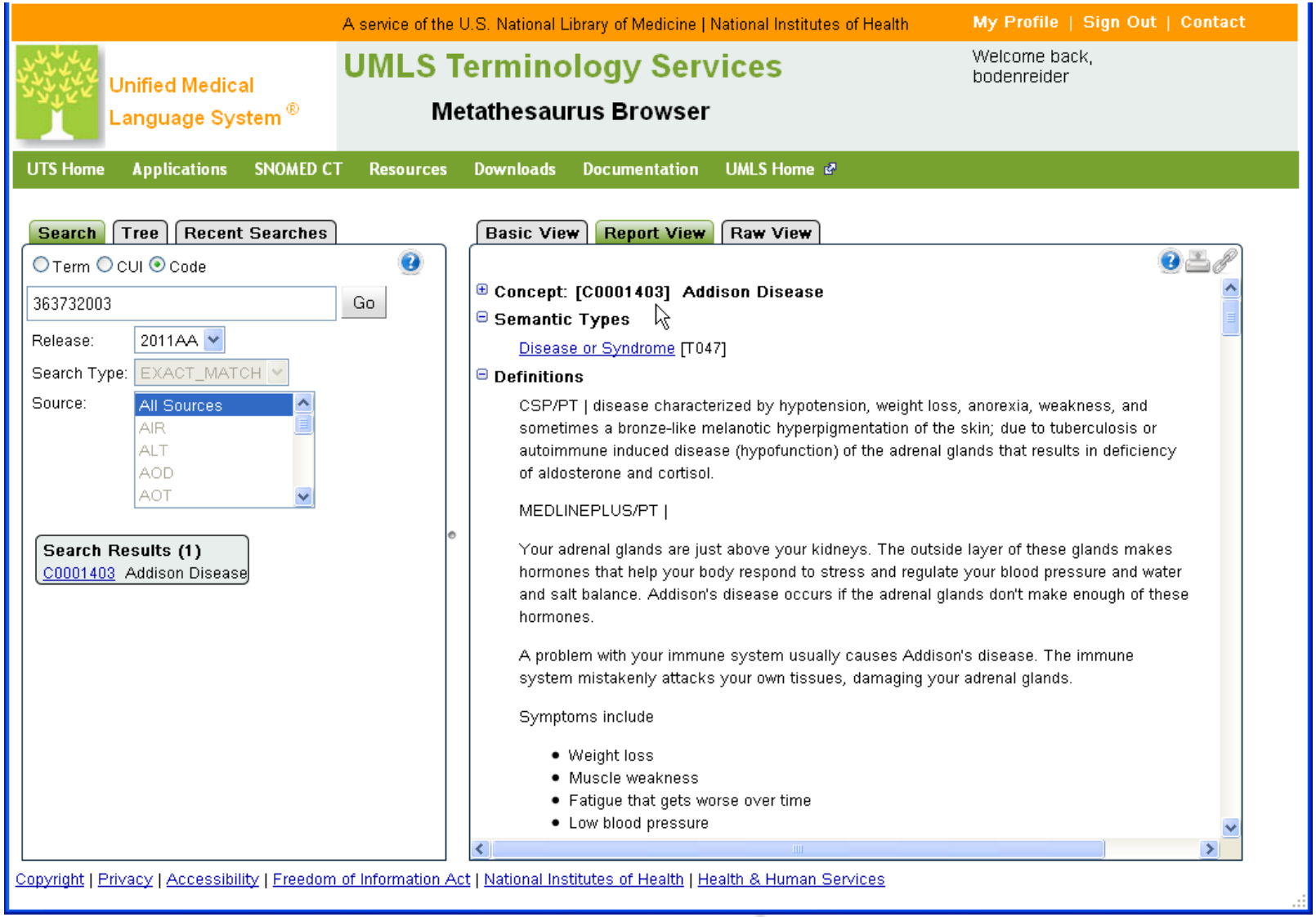
- UTS
  - Search SNOMED CT term in English (or SNOMED CT ID)
  - Find UMLS CUI
  - Explore source vocabularies / languages for this concept

Example

- SNOMED CT concept
  Addison's disease [363732003]



Addison's disease in UMLS



Languages for C0001403

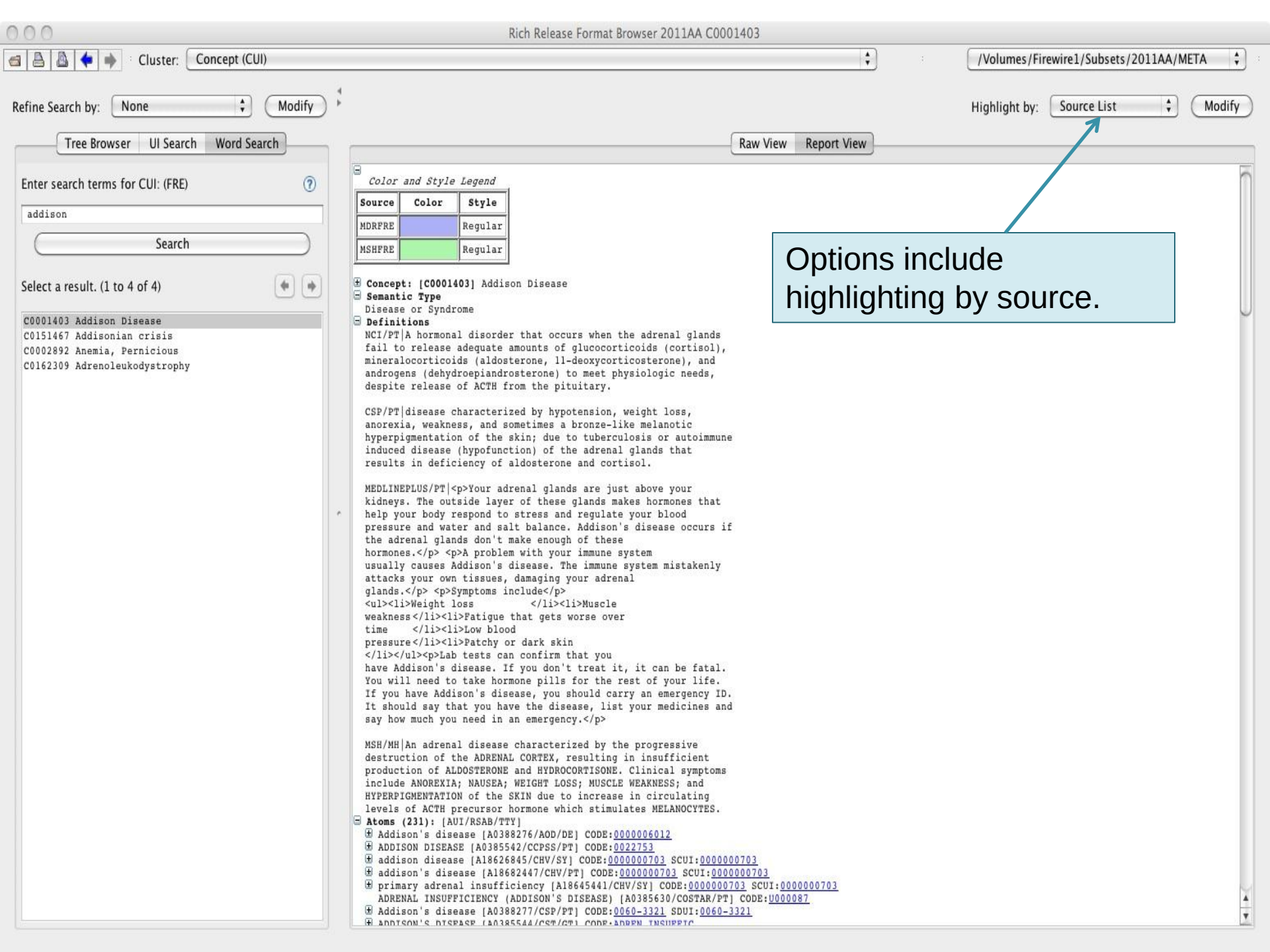
Language	Name
CZE	Addisonova choroba
DUT	Addison, ziekte van
FIN	Addisonin tauti
FRE	Maladie d'Addison
GER	Addisonsche Krankheit
ITA	Malattia di Addison
JPN	アジソン病
KOR	원발성 부신피질 기능부전
POL	Choroba Addisona
POR	Doença de Addison
RUS	АДДИСОНОВА БОЛЕЗНЬ
SCR	ADDISONOVA BOLEST
SPA	Enfermedad de Addison
SWE	Addisons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

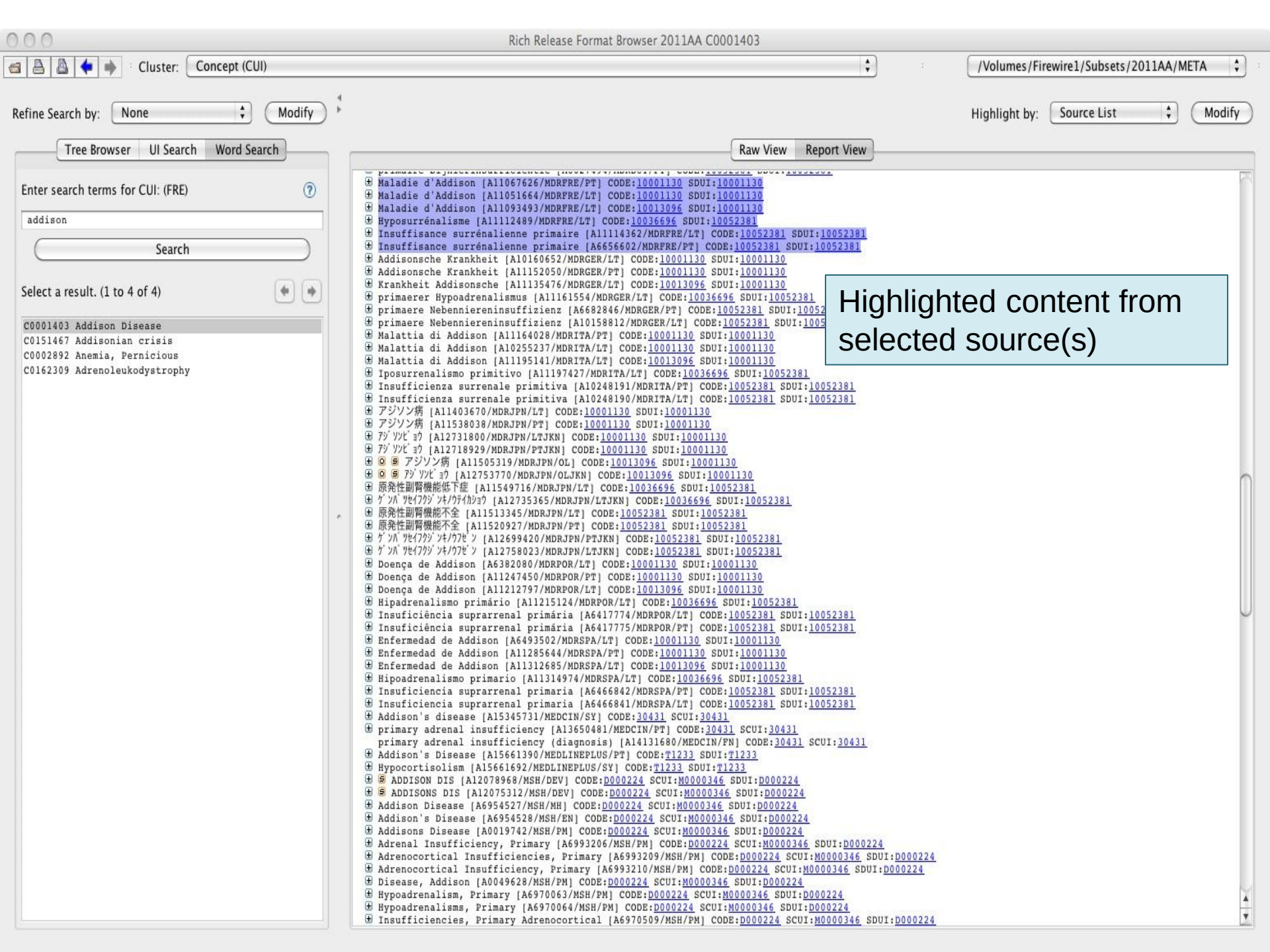
C0001403 in Dutch

Source	Term type	Code	Name
ICD10	PT	E27.1	Primaire bijnierschorsinsufficiëntie
MedDRA	PT	10001130	Addison, ziekte van
MedDRA	PT	10052381	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	10001130	Maladie d'Addison
MedDRA	PT	10052381	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


Rich Release Format Browser 2011AA C0001403
Cluster: Concept (CUI)
/Volumes/Firewire1/Subsets/2011AA/META
Refine Search by: None
Modify
Highlight by: Source List
Modify
Tree Browser
UI Search
Word Search
Raw View
Report View
Enter search terms for CUI: (FRE)
addison
Search
Select a result. (1 to 4 of 4)
C0001403 Addison Disease
C0151467 Addisonian crisis
C0002892 Anemia, Pernicious
C0162309 Adrenoleukodystrophy
Color and Style Legend
Source
Color
Style
MDRFRE
Regular
MSHFRE
Regular
Concept: [C0001403] Addison Disease
Semantic Type
Disease or Syndrome
Definitions
NCI/PT|A hormonal disorder that occurs when the adrenal glands fail to release adequate amounts of glucocorticoids (cortisol), mineralocorticoids (aldosterone, 11-deoxycorticosterone), and androgens (dehydroepiandrosterone) to meet physiologic needs, despite release of ACTH from the pituitary.
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
Patchy or dark skin
 Lab tests can confirm that you have Addison's disease. If you don't treat it, it can be fatal. You will need to take hormone pills for the rest of your life. If you have Addison's disease, you should carry an emergency ID. It should say that you have the disease, list your medicines and say how much you need in an emergency.
MSH/MH|An adrenal disease characterized by the progressive destruction of the ADRENAL CORTEX, resulting in insufficient production of ALDOSTERONE and HYDROCORTISONE. Clinical symptoms include ANOREXIA; NAUSEA; WEIGHT LOSS; MUSCLE WEAKNESS; and HYPERPIGMENTATION of the SKIN due to increase in circulating levels of ACTH precursor hormone which stimulates MELANOCYTES.
Atoms (231): [AUI/RSAB/TTY]
Addison's disease [A0388276/AOD/DE] CODE:0000006012
ADDISON DISEASE [A0385542/CCPSS/PT] CODE:0022753
addison disease [A18626845/CHV/SY] CODE:0000000703 SCUI:0000000703
addison's disease [A18682447/CHV/PT] CODE:0000000703 SCUI:0000000703
primary adrenal insufficiency [A18645441/CHV/SY] CODE:0000000703 SCUI:0000000703
ADRENAL INSUFFICIENCY (ADDISON'S DISEASE) [A0385630/COSTAR/PT] CODE:U000087
Addison's disease [A0388277/CSP/PT] CODE:0060-3321 SDUI:0060-3321
Options include highlighting by source.


Rich Release Format Browser 2011AA C0001403
Cluster:
Concept (CUI)
/Volumes/Firewire1/Subsets/2011AA/META
Refine Search by:
None
Modify
Highlight by:
Source List
Modify
Tree Browser
UI Search
Word Search
Raw View
Report View
Enter search terms for CUI: (FRE)
addison
Search
Select a result. (1 to 4 of 4)
C0001403 Addison Disease
C0151467 Addisonian crisis
C0002892 Anemia, Pernicious
C0162309 Adrenoleukodystrophy
Maladie d'Addison [A11067626/MDRFRE/PT] CODE:10001130 SDUI:10001130
Maladie d'Addison [A11051664/MDRFRE/LT] CODE:10001130 SDUI:10001130
Maladie d'Addison [A11093493/MDRFRE/LT] CODE:10013096 SDUI:10001130
Hyposurrénalisme [A11112489/MDRFRE/LT] CODE:10036696 SDUI:10052381
Insuffisance surrénalienne primaire [A11114362/MDRFRE/LT] CODE:10052381 SDUI:10052381
Insuffisance surrénalienne primaire [A6656602/MDRFRE/PT] CODE:10052381 SDUI:10052381
Addisonsche Krankheit [A10160652/MDRGER/LT] CODE:10001130 SDUI:10001130
Addisonsche Krankheit [A11152050/MDRGER/PT] CODE:10001130 SDUI:10001130
Krankheit Addisonsche [A11135476/MDRGER/LT] CODE:10013096 SDUI:10001130
primaerer Hypoadrenalismus [A11161554/MDRGER/LT] CODE:10036696 SDUI:10052381
primaere Nebenniereninsuffizienz [A6682846/MDRGER/PT] CODE:10052381 SDUI:10052
primaere Nebenniereninsuffizienz [A10158812/MDRGER/LT] CODE:10052381 SDUI:1005
Malattia di Addison [A11164028/MDRITA/PT] CODE:10001130 SDUI:10001130
Malattia di Addison [A10255237/MDRITA/LT] CODE:10001130 SDUI:10001130
Malattia di Addison [A11195141/MDRITA/LT] CODE:10013096 SDUI:10001130
Iposurrenalismo primitivo [A11197427/MDRITA/LT] CODE:10036696 SDUI:10052381
Insufficienza surrenale primitiva [A10248191/MDRITA/PT] CODE:10052381 SDUI:10052381
Insufficienza surrenale primitiva [A10248190/MDRITA/LT] CODE:10052381 SDUI:10052381
アジソン病 [A11403670/MDRJPN/LT] CODE:10001130 SDUI:10001130
アジソン病 [A11538038/MDRJPN/PT] CODE:10001130 SDUI:10001130
ｱｼﾞｿﾝﾋﾞｮｳ [A12731800/MDRJPN/LTJKN] CODE:10001130 SDUI:10001130
ｱｼﾞｿﾝﾋﾞｮｳ [A12718929/MDRJPN/PTJKN] CODE:10001130 SDUI:10001130
O S アジソン病 [A11505319/MDRJPN/OL] CODE:10013096 SDUI:10001130
O S ｱｼﾞｿﾝﾋﾞｮｳ [A12753770/MDRJPN/OLJKN] CODE:10013096 SDUI:10001130
原発性副腎機能低下症 [A11549716/MDRJPN/LT] CODE:10036696 SDUI:10052381
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原発性副腎機能不全 [A11513345/MDRJPN/LT] CODE:10052381 SDUI:10052381
原発性副腎機能不全 [A11520927/MDRJPN/PT] CODE:10052381 SDUI:10052381
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Doença de Addison [A6382080/MDRPOR/LT] CODE:10001130 SDUI:10001130
Doença de Addison [A11247450/MDRPOR/PT] CODE:10001130 SDUI:10001130
Doença de Addison [A11212797/MDRPOR/LT] CODE:10013096 SDUI:10001130
Hipadrenalismo primário [A11215124/MDRPOR/LT] CODE:10036696 SDUI:10052381
Insuficiência suprarrenal primária [A6417774/MDRPOR/LT] CODE:10052381 SDUI:10052381
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Enfermedad de Addison [A11312685/MDRSPA/LT] CODE:10013096 SDUI:10001130
Hipoadrenalismo primario [A11314974/MDRSPA/LT] CODE:10036696 SDUI:10052381
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primary adrenal insufficiency [A13650481/MEDCIN/PT] CODE:30431 SCUI:30431
primary adrenal insufficiency (diagnosis) [A14131680/MEDCIN/FN] CODE:30431 SCUI:30431
Addison's Disease [A15661390/MEDLINEPLUS/PT] CODE:T1233 SDUI:T1233
Hypocortisolism [A15661692/MEDLINEPLUS/SY] CODE:T1233 SDUI:T1233
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S ADDISONS DIS [A12075312/MSH/DEV] CODE:D000224 SCUI:M0000346 SDUI:D000224
Addison Disease [A6954527/MSH/MH] CODE:D000224 SCUI:M0000346 SDUI:D000224
Addison's Disease [A6954528/MSH/EN] CODE:D000224 SCUI:M0000346 SDUI:D000224
Addisons Disease [A0019742/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Adrenal Insufficiency, Primary [A6993206/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Adrenocortical Insufficiencies, Primary [A6993209/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Adrenocortical Insufficiency, Primary [A6993210/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Disease, Addison [A0049628/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Hypoadrenalism, Primary [A6970063/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Hypoadrenalisms, Primary [A6970064/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Insufficiencies, Primary Adrenocortical [A6970509/MSH/PM] CODE:D000224 SCUI:M0000346 SDUI:D000224
Highlighted content from selected source(s)

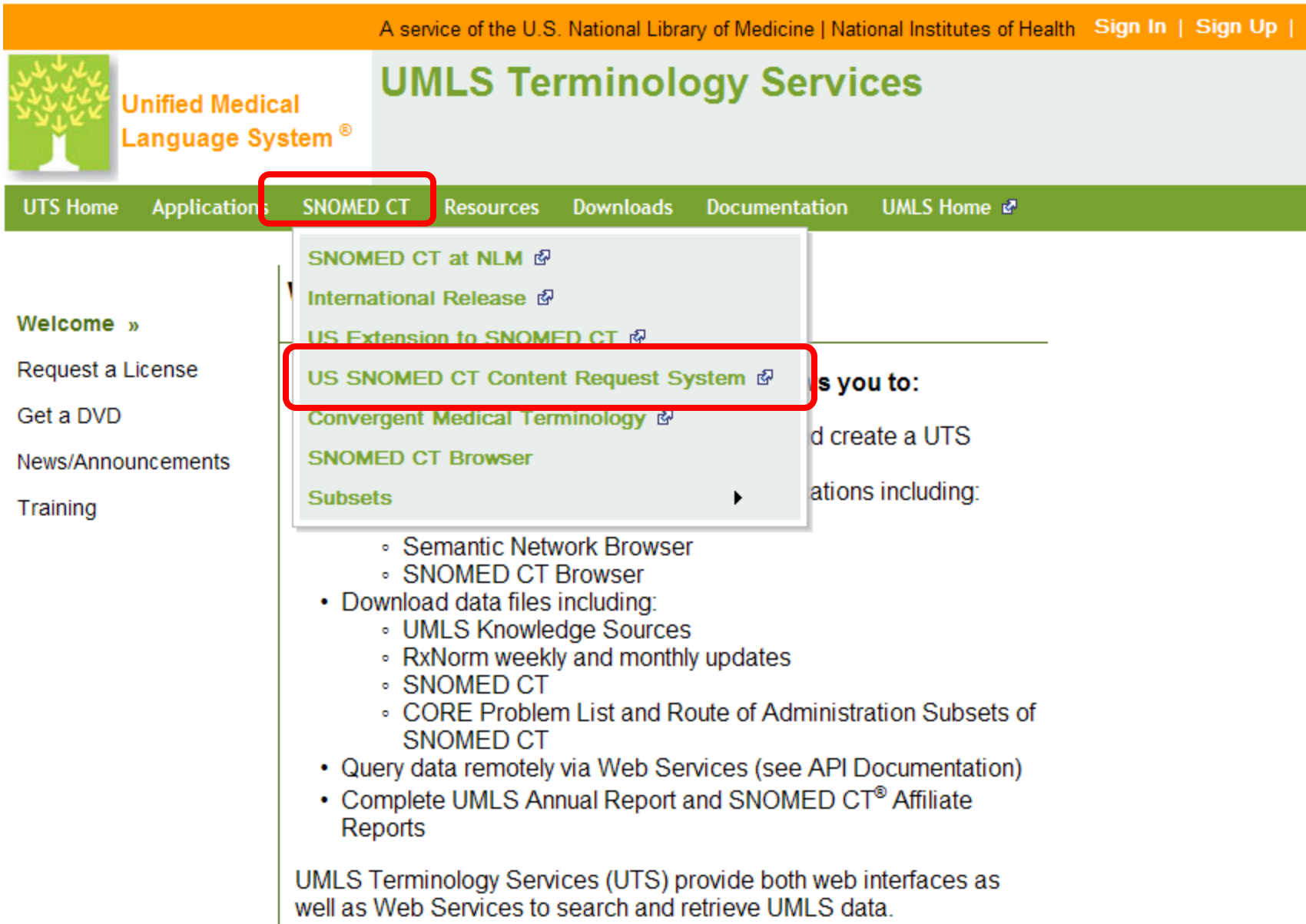
Dr. James T. Case

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



U.S. SNOMED CT Content Request System (USCRS)

U.S. NATIONAL LIBRARY OF MEDICINE

Beta

INTERNATIONAL HEALTH TERMINOLOGY STANDARDS DEVELOPMENT ORGANISATION

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 10 requests per page

Previous Next

Summary

Total Requests: 2416

My Requests:

Submitted: 1795

Accepted: 723

Approved: 21

Draft: 0

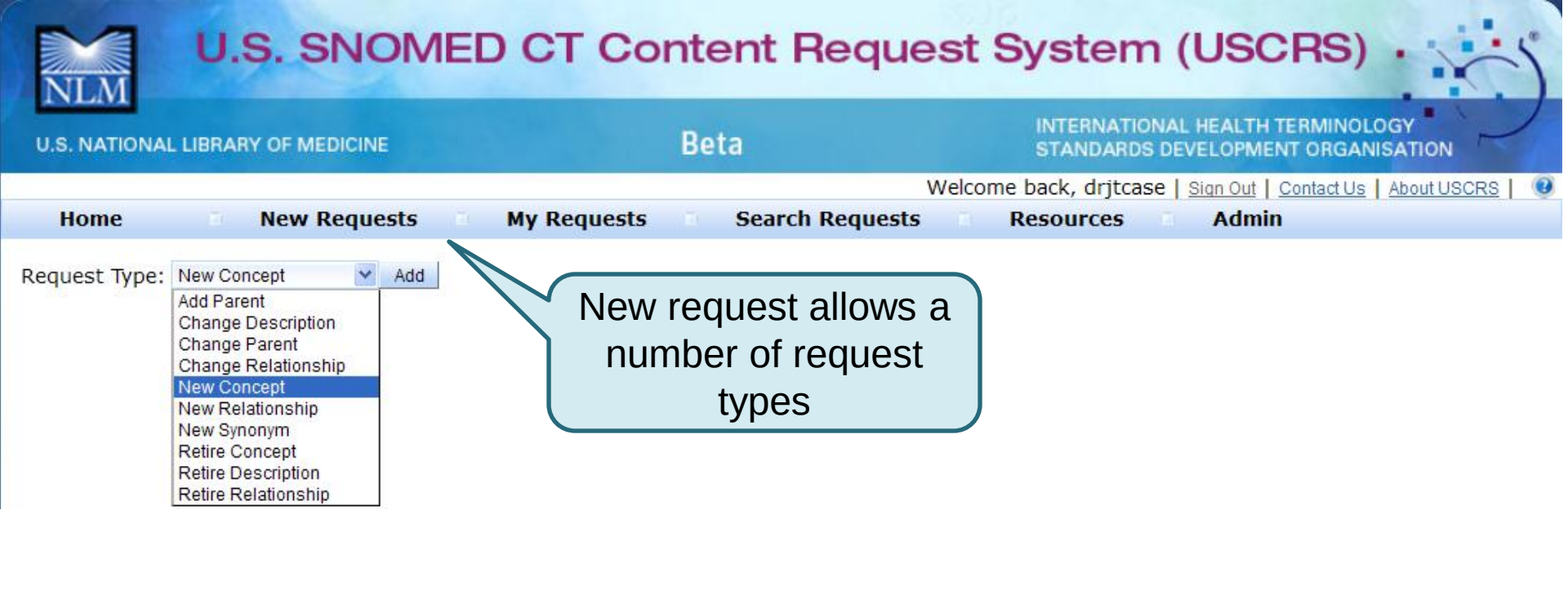
Important Dates

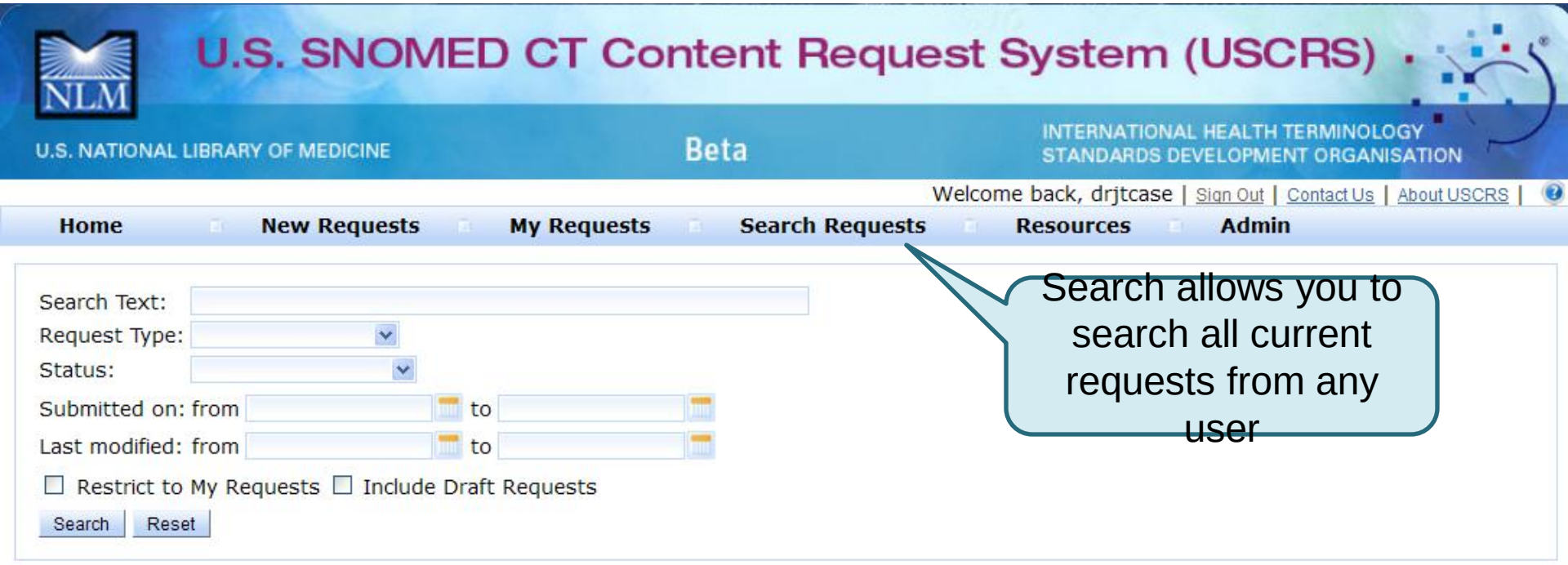
September 15, 2011
Submit requests for next US Extension of SNOMED CT

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.)


U.S. SNOMED CT Content Request System (USCRS)
U.S. NATIONAL LIBRARY OF MEDICINE
Beta
INTERNATIONAL HEALTH TERMINOLOGY STANDARDS DEVELOPMENT ORGANISATION
Welcome back, drjtcase | Sign Out | Contact Us | About USCRS |
Home
New Requests
My Requests
Search Requests
Resources
Admin
Request Type: New Concept Add
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


U.S. SNOMED CT Content Request System (USCRS)
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Beta
INTERNATIONAL HEALTH TERMINOLOGY STANDARDS DEVELOPMENT ORGANISATION
Welcome back, drjtcase | Sign Out | Contact Us | About USCRS |
Home
New Requests
My Requests
Search Requests
Resources
Admin
Search Text:
Request Type:
Status:
Submitted on: from to
Last modified: from to
Restrict to My Requests
Include Draft Requests
Search
Reset
Search allows you to search all current requests from any user

U.S. SNOMED CT Content Request System (USCRS)

U.S. NATIONAL LIBRARY OF MEDICINE — Beta — INTERNATIONAL HEALTH TERMINOLOGY STANDARDS DEVELOPMENT ORGANISATION

Submitter:	James Case (NLM/NIH)
Request Type:	New Concept
Topic:*	Dentistry
Source Terminology:*	SNOMED CT International
Local Code:	123445
Local Term:	
Fully Specified Name:*	Supernumerary second incisor of maxilla
Semantic Tag:*	disorde
Preferred Term:*	Supernumerary second incisor of maxilla
Synonym:	Add
Parent Concept Id:*	Add
UMLS CUI:	95267007
Definition:*	Teeth which appear in addition to the regular number of teeth.
Proposed Use:	
Justification:*	Needed to support dental findings
Note:	

*indicates required information

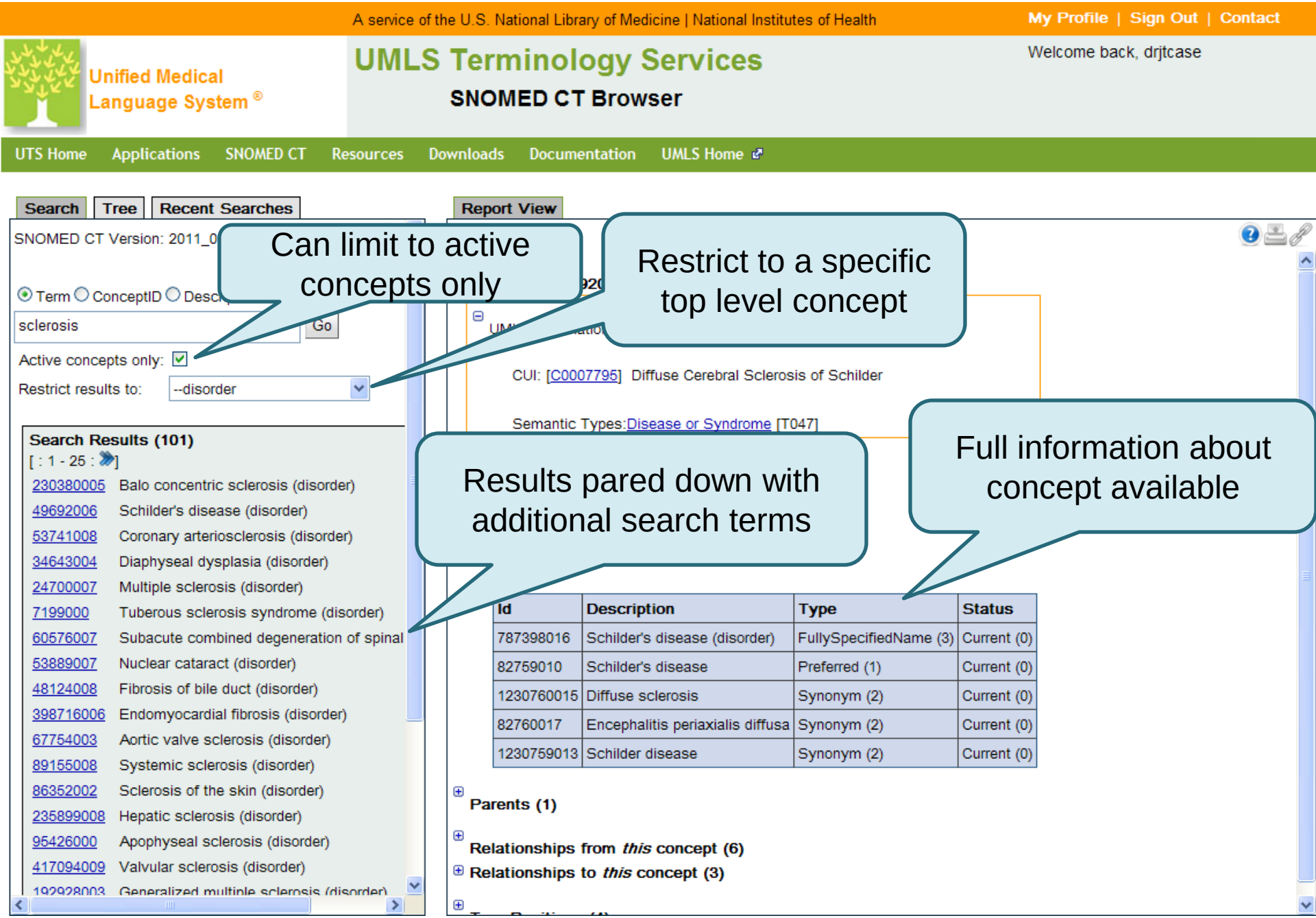
Save Draft | Validate and Save | Discard

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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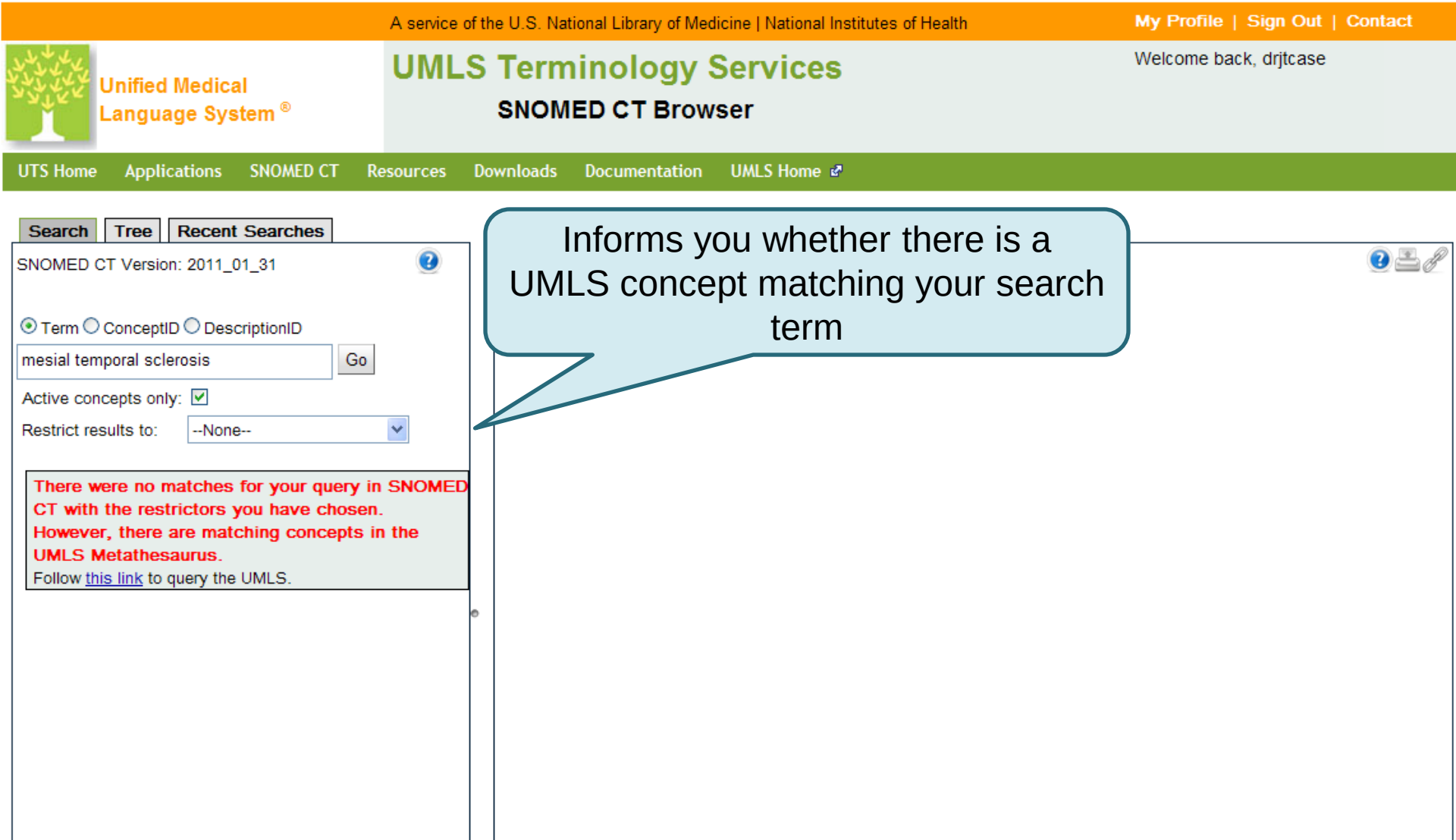
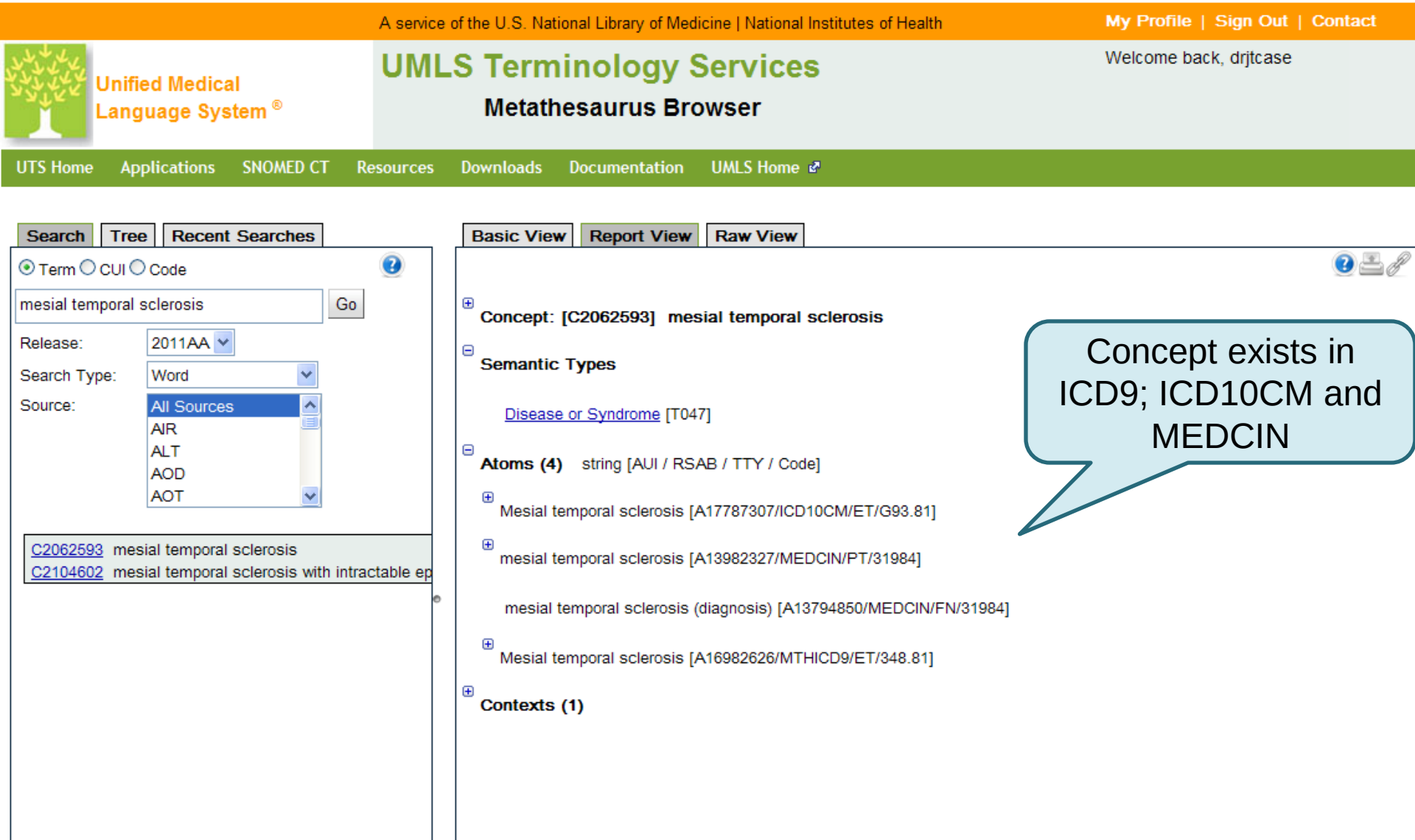
Unified Medical Language System ®
UMLS Terminology Services
SNOMED CT Browser
Welcome back, drjtcase
UTS Home
Applications
SNOMED CT
Resources
Downloads
Documentation
UMLS Home
Search
Tree
Recent Searches
Report View
SNOMED CT Version: 2011_0
Term
ConceptID
sclerosis
Go
Active concepts only:
Restrict 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 Tuberous 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)
192928003 Generalized multiple sclerosis (disorder)
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)
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

No SNOMED CT Description?



UMLS Concept



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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