

Health Search

From Consumers to Clinicians

Slides available at

<https://github.com/ielab/health-search-tutorial>

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Auxiliary Details on UMLS

(not covered in tutorial)

Digging into the UMLS

- There UMLS Metathesaurus:
- The heart of UMLS
- Consists of over 1,000,000 concepts

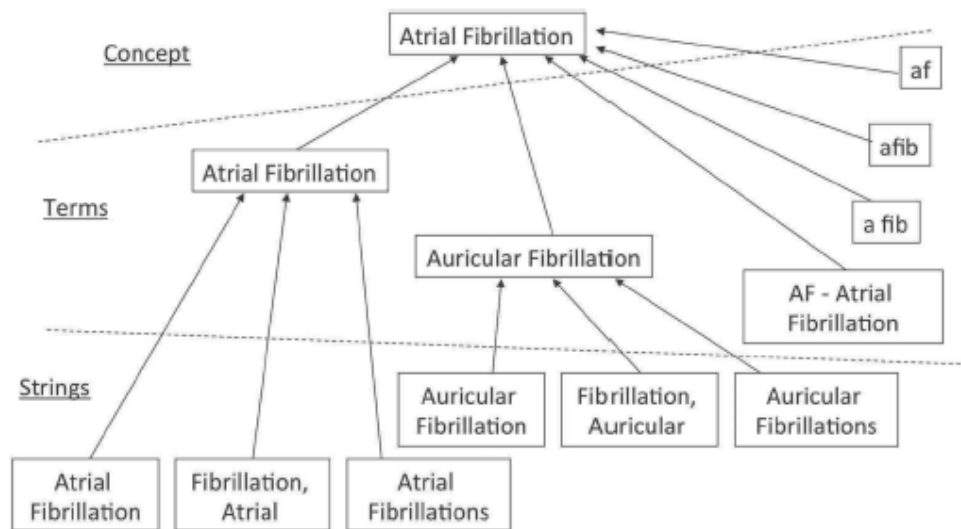


FIGURE 14.3: Unified Medical Language System Metathesaurus concept of atrial fibrillation. (Courtesy of NLM)

Table 1. Concept, Term, Atom, and String Identifiers.

| Concept (CUI) | Terms (LUIs) | Strings (SUIs) | Atoms (AUIs) * RRF Only |
|---|---|--|--|
| C0004238 Atrial Fibrillation (preferred) Atrial Fibrillations Auricular Fibrillation Auricular Fibrillations | L0004238 Atrial Fibrillation (preferred) Atrial Fibrillations | S0016668 Atrial Fibrillation (preferred) | A0027665 Atrial Fibrillation (from MSH) A0027667 Atrial Fibrillation (from PSY) |
| | | S0016669 (plural variant) Atrial Fibrillations | A0027668 Atrial Fibrillations (from MSH) |
| | L0004327 (synonym) Auricular Fibrillation Auricular Fibrillations | S0016899 Auricular Fibrillation (preferred) | A0027930 Auricular Fibrillation (from PSY) |
| | | S0016900 (plural variant) Auricular Fibrillations | A0027932 Auricular Fibrillations (from MSH) |
| | | | |
| | | | |

The UMLS Metathesaurus

- UMLS tables - Many of them, but 2 key tables:

1. MRConso.rtf: Links each concept (CUI) to each unique concept in a source vocabulary (AUI)

Sample Records

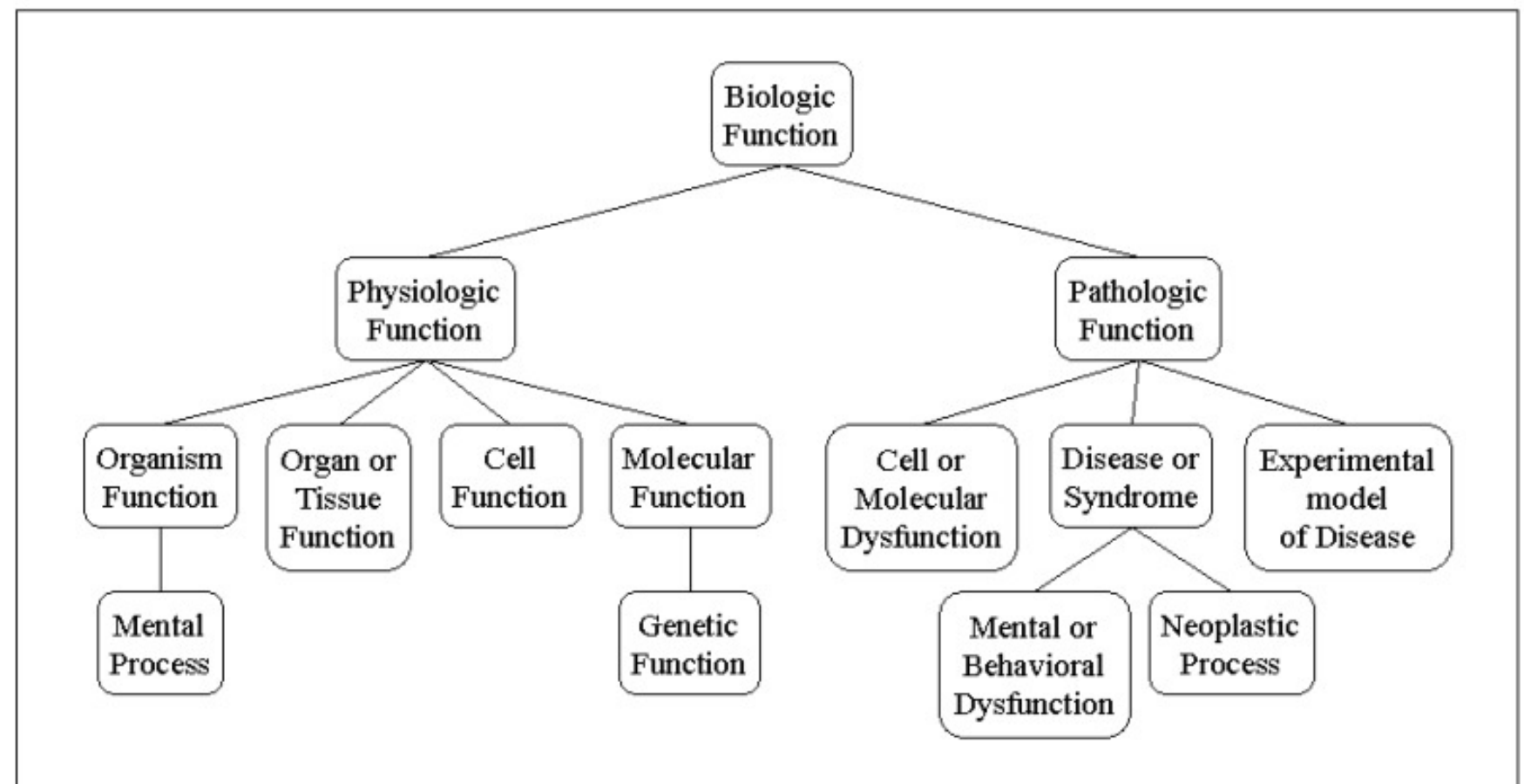
C0001175|ENG|P|L0001175|VO|S0010340|Y|A0019182||M0000245|D000163|MSH|PM|D000163|Acquired Immunodeficiency Syndromes|0|N|1792|
 C0001175|ENG|S|L0001842|PF|S0011877|N|A2878223|103840012|62479008||SNOMEDCT|PT|62479008|AIDS|9|N|3840|
 C0001175|ENG|P|L0001175|VC|S0354232|Y|A2922342|103845019|62479008||SNOMEDCT|SY|62479008|Acquired immunodeficiency syndrome|9|N|3584|
 C0001175|FRE|S|L0162173|PF|S0226654|Y|A7438879||M0000245|D000163|MSHFRE|EN|D000163|SIDA|3|N||
 C0001175|RUS|S|L0904943|PF|S1108760|Y|A13488500||M0000245|D000163|MSHRUS|SY|D000163|SPID|3|N||

2. MRREL.rtf: links 2 concepts with a relationship. Note the relationship is AUI to AUI

| REL | Description |
|------|---|
| AQ | Allowed qualifier |
| CHD | has child relationship in a Metathesaurus source |
| DEL | Deleted concept |
| PAR | has parent relationship in a Metathesaurus source |
| QB | can be qualified by. |
| RB | has a broader relationship |
| RL | the relationship is similar or "alike". the two |
| RN | has a narrower relationship |
| RO | has relationship other than synonymous, |
| RQ | related and possibly synonymous. |
| RU | Related, unspecified |
| SIB | has sibling relationship in a Metathesaurus |
| SUBX | Concept removed from current subset |
| SY | source asserted synonymy. |
| XR | Not related, no mapping |
| | Empty relationship |

UMLS Semantic Network

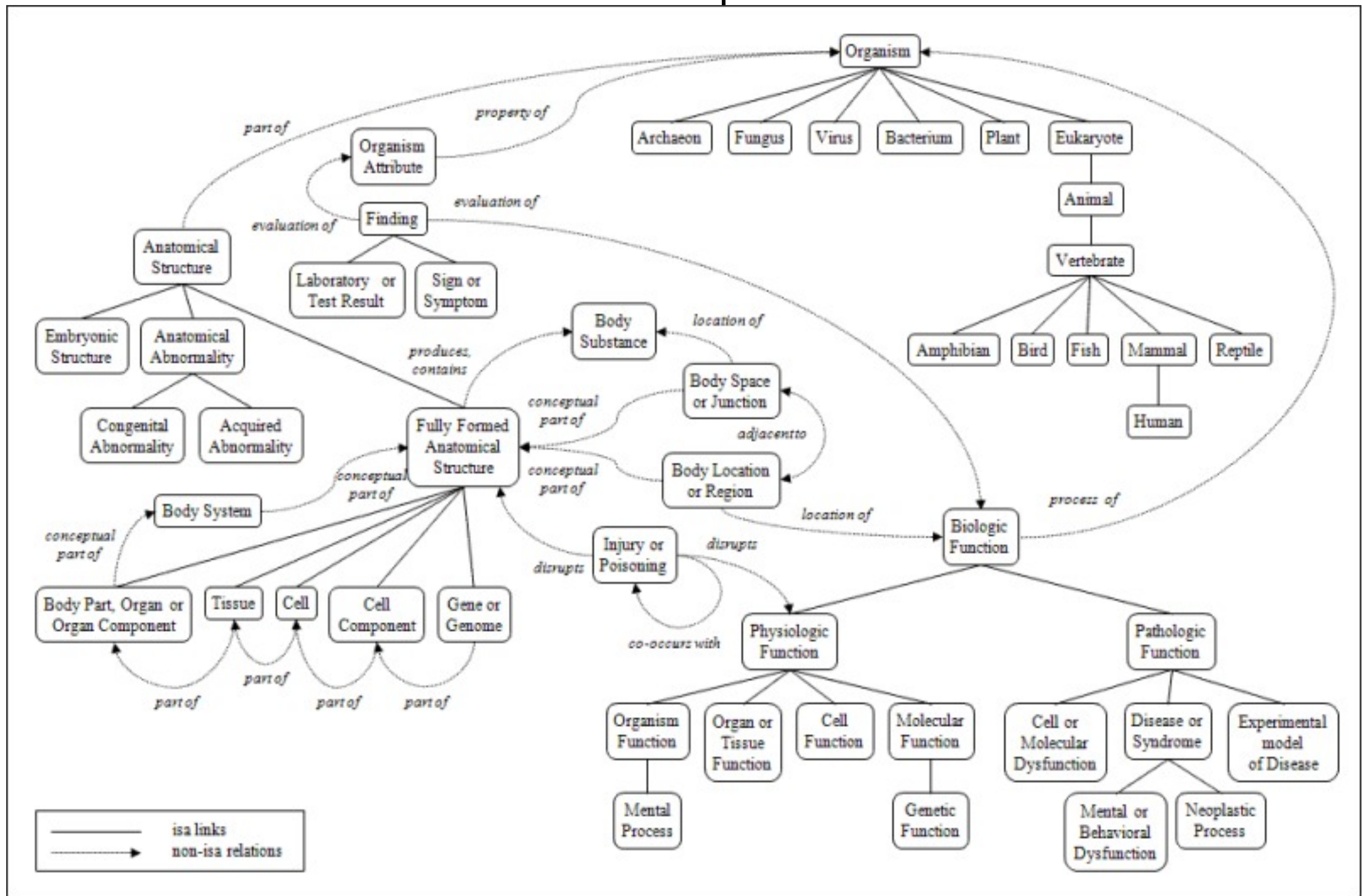
- Categorise concepts and relationships among concepts.
- Each UMLS concept is assigned to at least one semantic type (i.e., defines the concept category).
- Semantic types are related by links: "is a", "part of", "process of", etc.



Part of "is a" semantic hierarchy.

Full hierarchy: https://www.nlm.nih.gov/research/umls/META3_current_semantic_types.html

UMLS Semantic Network Links: Example



UMLS Semantic Type Groups

- To reduce the complexity, semantic types are grouped based on its: semantic validity, parsimony, completeness, exclusivity, naturalness, and utility.

| | | | |
|------|-----------|------|----------------------------------|
| DISO | Disorders | T020 | Acquired Abnormality |
| DISO | Disorders | T190 | Anatomical Abnormality |
| DISO | Disorders | T049 | Cell or Molecular Dysfunction |
| DISO | Disorders | T019 | Congenital Abnormality |
| DISO | Disorders | T047 | Disease or Syndrome |
| DISO | Disorders | T050 | Experimental Model of Disease |
| DISO | Disorders | T033 | Finding |
| DISO | Disorders | T037 | Injury or Poisoning |
| DISO | Disorders | T048 | Mental or Behavioral Dysfunction |
| DISO | Disorders | T191 | Neoplastic Process |
| DISO | Disorders | T046 | Pathologic Function |
| DISO | Disorders | T184 | Sign or Symptom |

Semantic types within Semantic type group: "Disorders"

UMLS semantic network Detail: SRDEF

- Basic information about the semantic types and relations

| Field | Description |
|--------------|---|
| RT: | Record Type (STY = Semantic Type or RL = Relation). |
| UI: | Unique Identifier of the Semantic Type or Relation. |
| STY/RL: | Name of the Semantic Type or Relation. |
| STN/ DEF: | Tree Number of the Semantic Type or Relation. |
| DEF: | Definition of the Semantic Type or Relation. |
| EX: | Examples of Metathesaurus concepts with this Semantic Type (STY |
| UN: | Usage note for Semantic Type assignment (STY records only). |
| NH: | The Semantic Type and its descendants allow the non-human flag |
| ABR: | Abbreviation of the Relation Name or Semantic Type. |
| RIN: | Inverse of the Relation (RL records only). |

SRDEF Sample entries

STY|T047|Disease or Syndrome|B2.2.1.2.1|A condition which alters or interferes with a normal process, state, or activity of an organism. It is usually characterized by the abnormal functioning of one or more of the host's systems, parts, or organs. Included here is a complex of symptoms descriptive of a disorder.||Any specific disease or syndrome that is modified by such modifiers as "acute", "prolonged", etc. will also be assigned to this type. If an anatomic abnormality has a pathologic manifestation, then it will be given this type as well as a type from the 'Anatomical Abnormality' hierarchy, e.g., "Diabetic Cataract" will be double-typed for this reason.||dsyn||S

RL|T186|isa|H|The basic hierarchical link in the Network. If one item "isa" another item then the first item is more specific in meaning than the second item.||||S|inverse_isa|

UMLS semantic network Detail: SRSTR

- Structure of the network

| STY/RL: | Argument 1 (Name of a Semantic Type or Relation). |
|----------------|--|
| RL: | Relation ("isa" or the name of a non-hierarchical Relation). |
| STY/RL: | Argument 2 (Name of a Semantic Type or Relation); if this field is blank this means that the Semantic Type or Relation is one of the top |
| LS: | Link Status (D = Defined for the Arguments and its children; B = Blocked; DNI = Defined but Not Inherited by the children of the |

Disease or Syndrome|conceptually_related_to|Experimental Model of Disease|DNI|
Disease or Syndrome|isa|Pathologic Function|D|
Disease or Syndrome|produces|Tissue|D|

Semantic Network Detail: SRSTRE1 OR SRSTRE2

- Fully inherited set of Relations (UI's or Names).

| | |
|---------|---|
| UI/STY: | Argument 1 (UI or name of a Semantic Type). |
| UI/RL: | Relation (UI or name of a nonhierarchical Relation). |
| UI/STY: | Argument 2 (UI or name of a Semantic Type). N.B.: The relations expressed in this table are binary relations and the arguments are ordered pairs. All relations have been fully inherited in this table. |

SRSTRE1

SRSTRE2

| | |
|--------------------|---|
| T047 T186 T038 | Disease or Syndrome isa Biologic Function |
| T047 T186 T046 | Disease or Syndrome isa Pathologic Function |
| T047 T186 T051 | Disease or Syndrome isa Event |
| T047 T186 T067 | Disease or Syndrome isa Phenomenon or Process |
| T047 T186 T070 | Disease or Syndrome isa Natural Phenomenon or Process |