Session 33
November 1, 2005

Ontological research
and its applications to the biomedical domain

Biomedical resources for text mining

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Overview

- An example
- Three types of resources
  - Lexical resources
  - Terminological resources
  - Ontological resources
- Some issues

An example

Neurofibromatosis 2

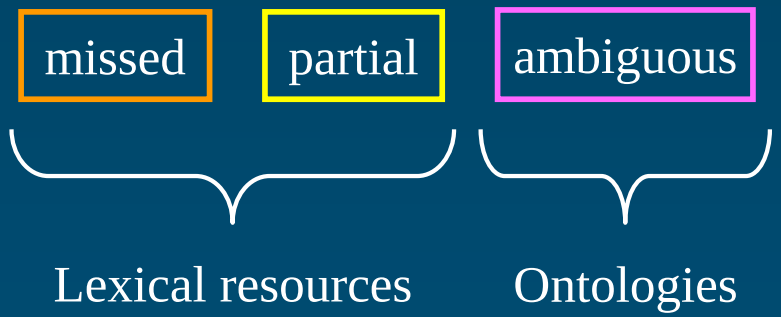
Neurofibromatosis 2

Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from peripheral neurofibromatosis. NF2 is a predominantly intracranial condition whose hallmark is bilateral vestibular schwannomas. NF2 results from a mutation in the gene named merlin, located on chromosome 22.

[Uppal, S., and A. P. Coatesworth. “Neurofibromatosis Type 2.” *Int J Clin Pract*, 57, no. 8, 2003, pp. 698-703.]

Entity recognition

Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from peripheral neurofibromatosis. NF2 is a predominantly intracranial condition whose hallmark is bilateral vestibular schwannomas. NF2 results from a mutation in the gene named merlin, located on chromosome 22.



Relation extraction

Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from peripheral neurofibromatosis. NF2 is a predominantly intracranial condition whose hallmark is bilateral vestibular schwannomas. NF2 results from a mutation in the gene named merlin, located on chromosome 22.

- vestibular schwannomas *manifestation of* neurofibromatosis 2
- neurofibromatosis 2 *associated with* mutation of NF2 gene
- NF2 gene *located on* chromosome 22

Ontologies

Resources for text mining

Types of resources

- Lexical resources
  - Collections of lexical items
  - Additional information
    - Part of speech
    - Spelling variants
  - Useful for entity recognition
  - UMLS SPECIALIST Lexicon, WordNet
- Ontological resources
  - Collections of
    - kinds of entities (substances, qualities, processes)
    - relations among them
  - Useful for relation extraction
  - UMLS Semantic Network, SNOMED CT

Types of resources (revisited)

- Lexical and terminological resources
  - Mostly collections of names for biomedical entities
  - Often have some kind or hierarchical organization (e.g., relations)
- Ontological resources
  - Mostly collections of relations among biomedical entities
  - Sometimes also collect names

Unified Medical Language System

- SPECIALIST Lexicon
  - 200,000 lexical items
  - Part of speech and variant information
- Metathesaurus
  - 5M names from over 100 terminologies
  - 1M concepts
  - 16M relations
- Semantic Network
  - 135 high-level categories
  - 7000 relations among them

Lexical resources

Terminological resources

Ontological resources

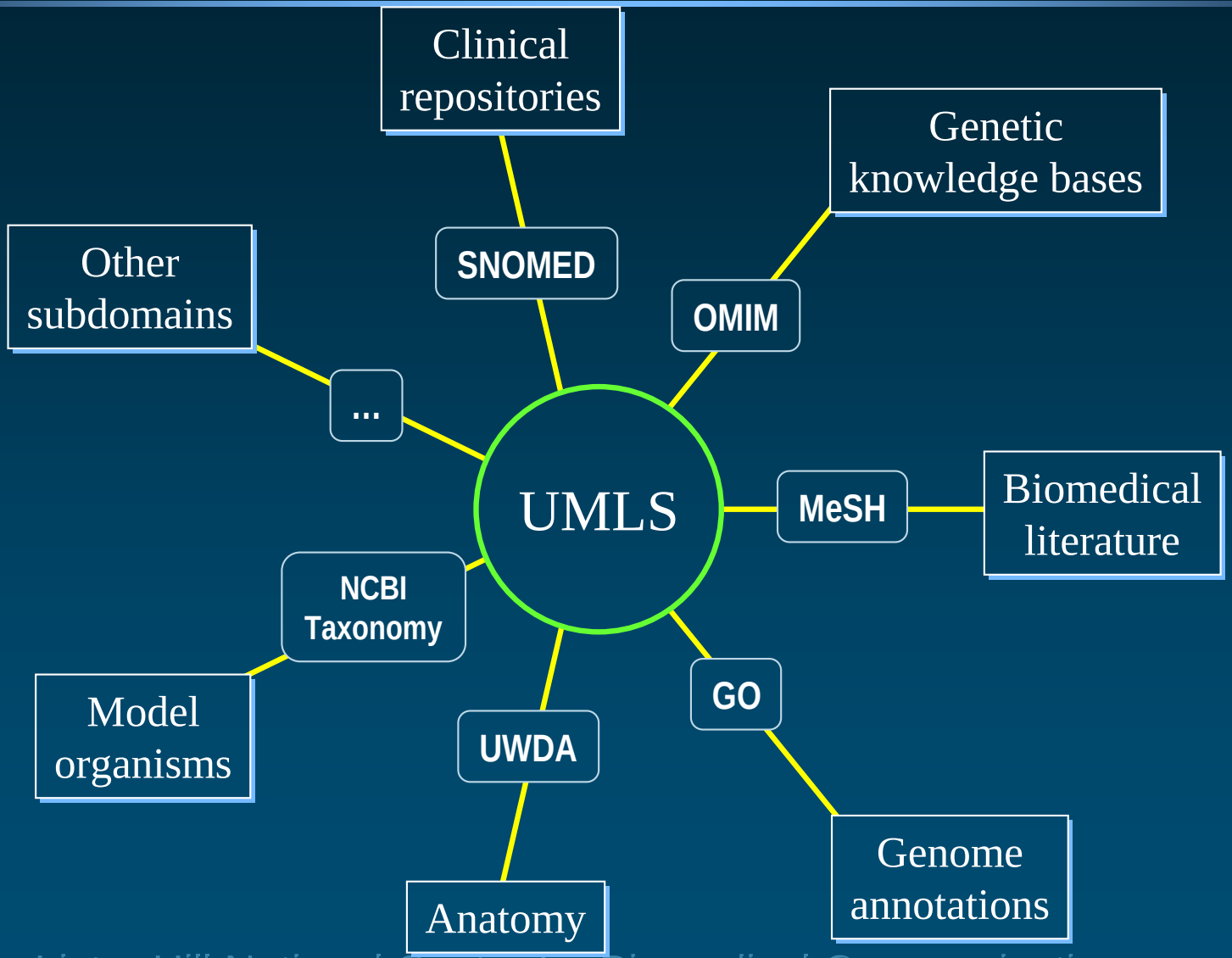
Terminological resources

UMLS Metathesaurus

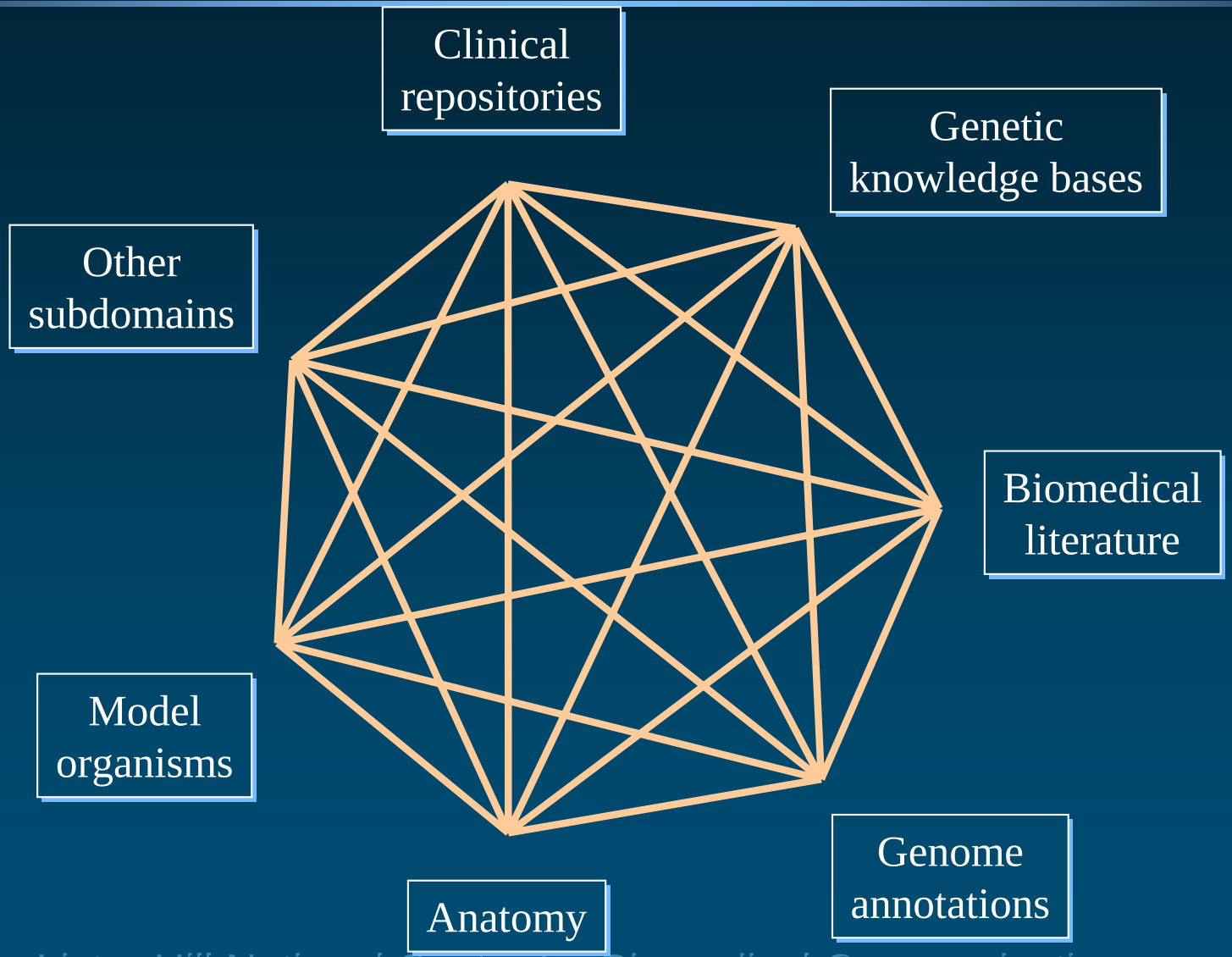
Source Vocabularies (2005AB)

- 133 source vocabularies contributing concept names
- ~80 families of vocabularies
  - multiple translations (e.g., MeSH, ICPC, ICD-10)
  - variants (American-English equivalents, Australian extension/adaptation)
  - subsequent editions usually considered distinct families (ICD: 9-10; DSM: IIIR-IV)
- Broad coverage of biomedicine
- Common presentation

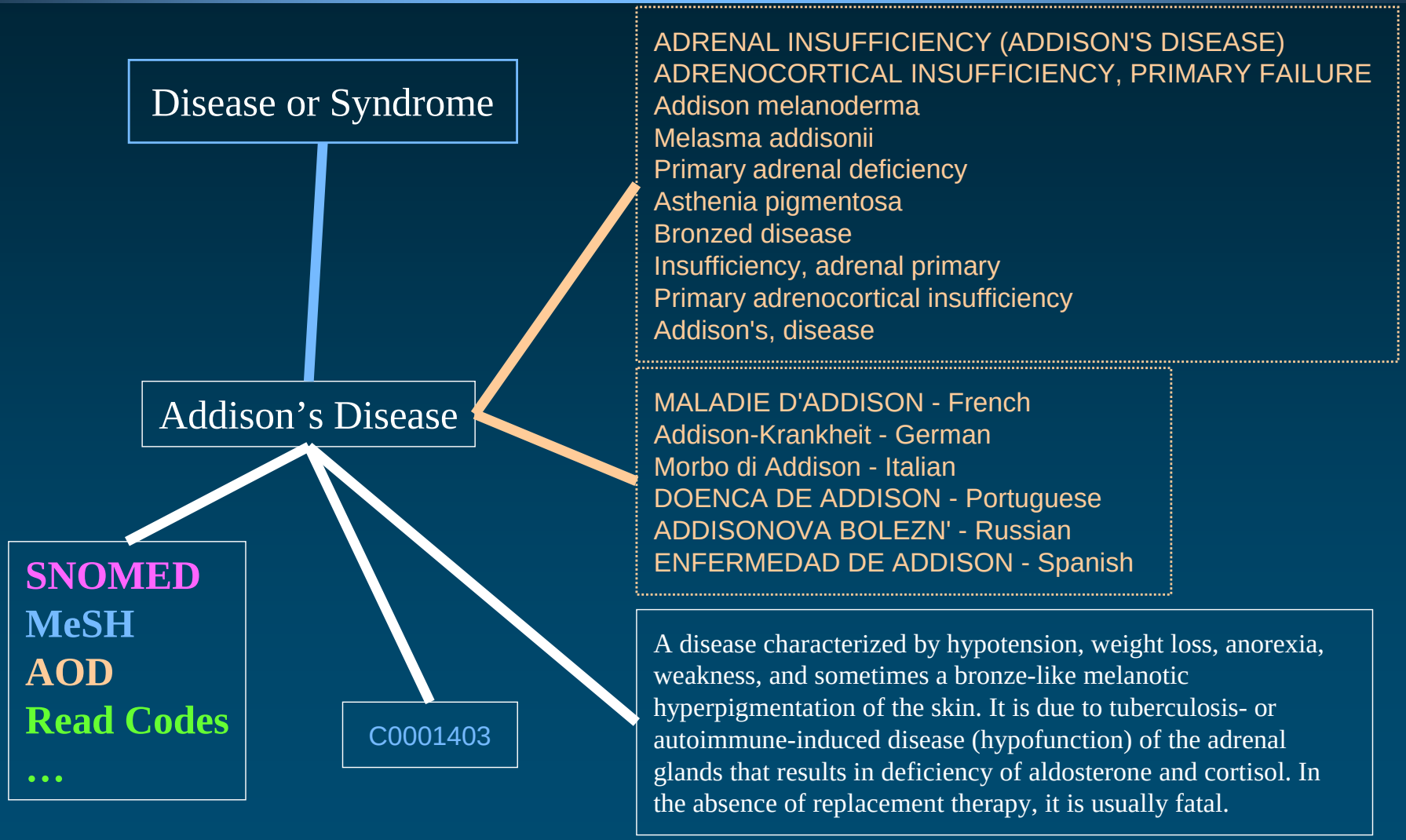
Integrating subdomains



Integrating subdomains

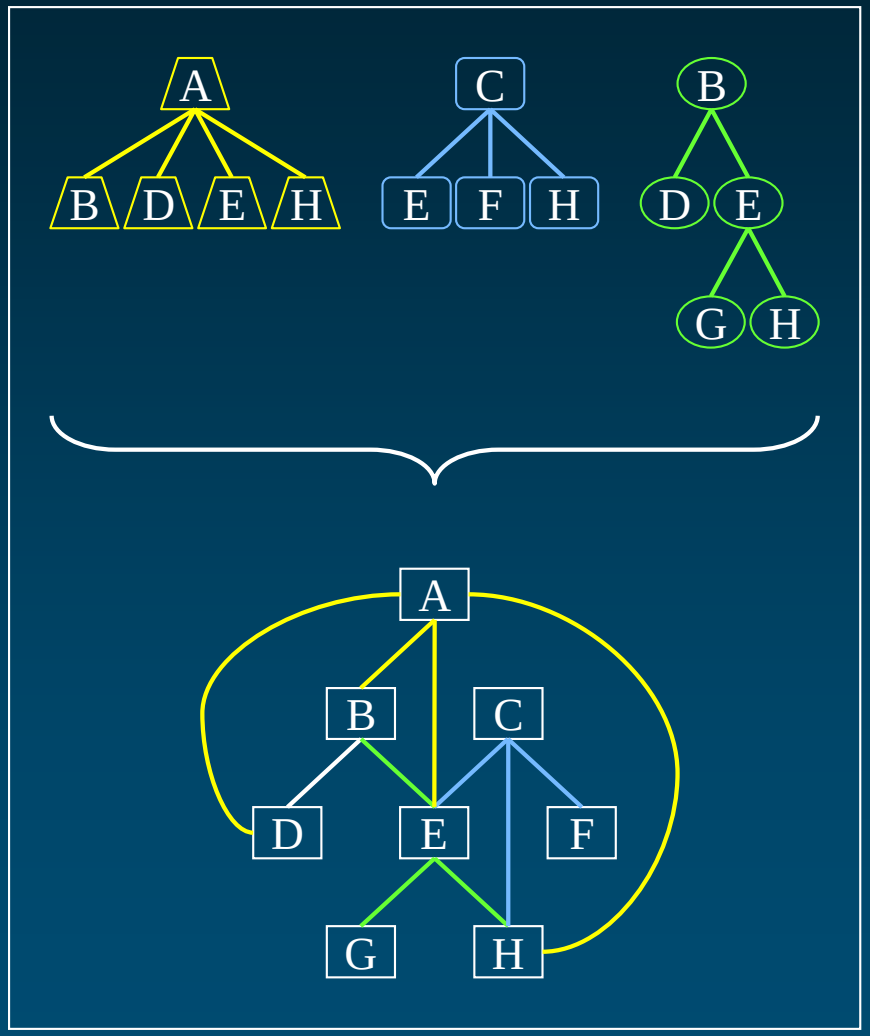


Addison's Disease: Concept



Organize concepts

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



Metahesaurus relations Examples

- Neurofibromin 2
  - Multiple parent concepts
    - Membrane proteins [MeSH]
    - Tumor suppressor proteins [MeSH]
    - Signaling protein [NCI Thesaurus]
  - 1 child concept
    - Merlin, Drosophila [MeSH]
  - Co-occurring concepts in MEDLINE
    - Neurofibromatosis 2 [13]
    - Membrane proteins [8]
    - …

Finding Metathesaurus concepts in text

- MetaMap (MMTx)
  - Developed at NLM
  - Named entity recognition
  - Approximate matches
  - Used in many projects
  - Not distributed with the UMLS

Neurofibromatosis type 2 (**NF2**) is often not recognised as a distinct entity from **peripheral neurofibromatosis**. **NF2** is a predominantly intracranial condition whose hallmark is bilateral **vestibular schwannomas**. **NF2** results from a **mutation** in the **gene** named **merlin**, located on **chromosome 22**.

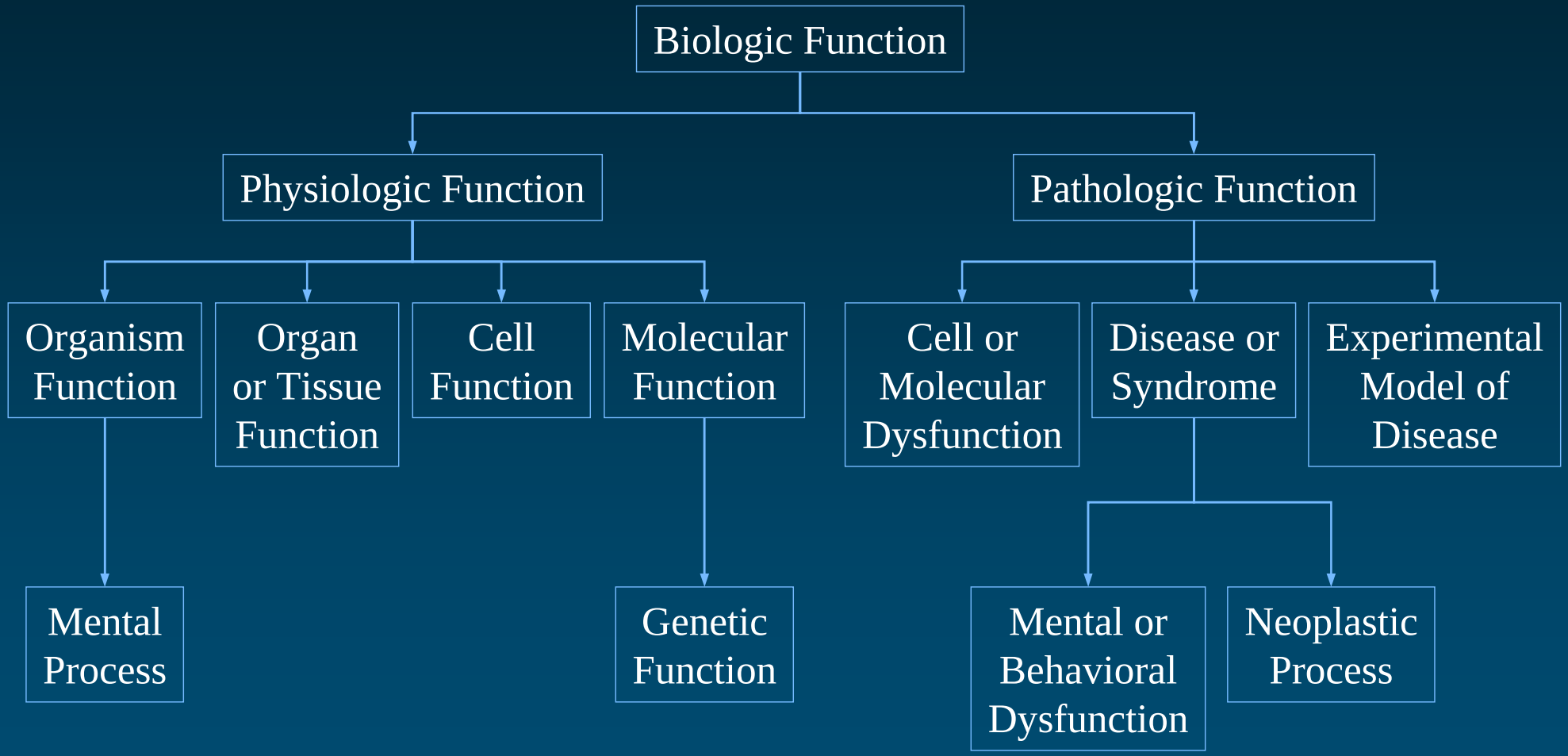
Ontological resources

UMLS Semantic Network

Semantic Network

- Semantic types (135)
  - tree structure
  - 2 major hierarchies
    - Entity
      - Physical Object
      - Conceptual Entity
    - Event
      - Activity
      - Phenomenon or Process

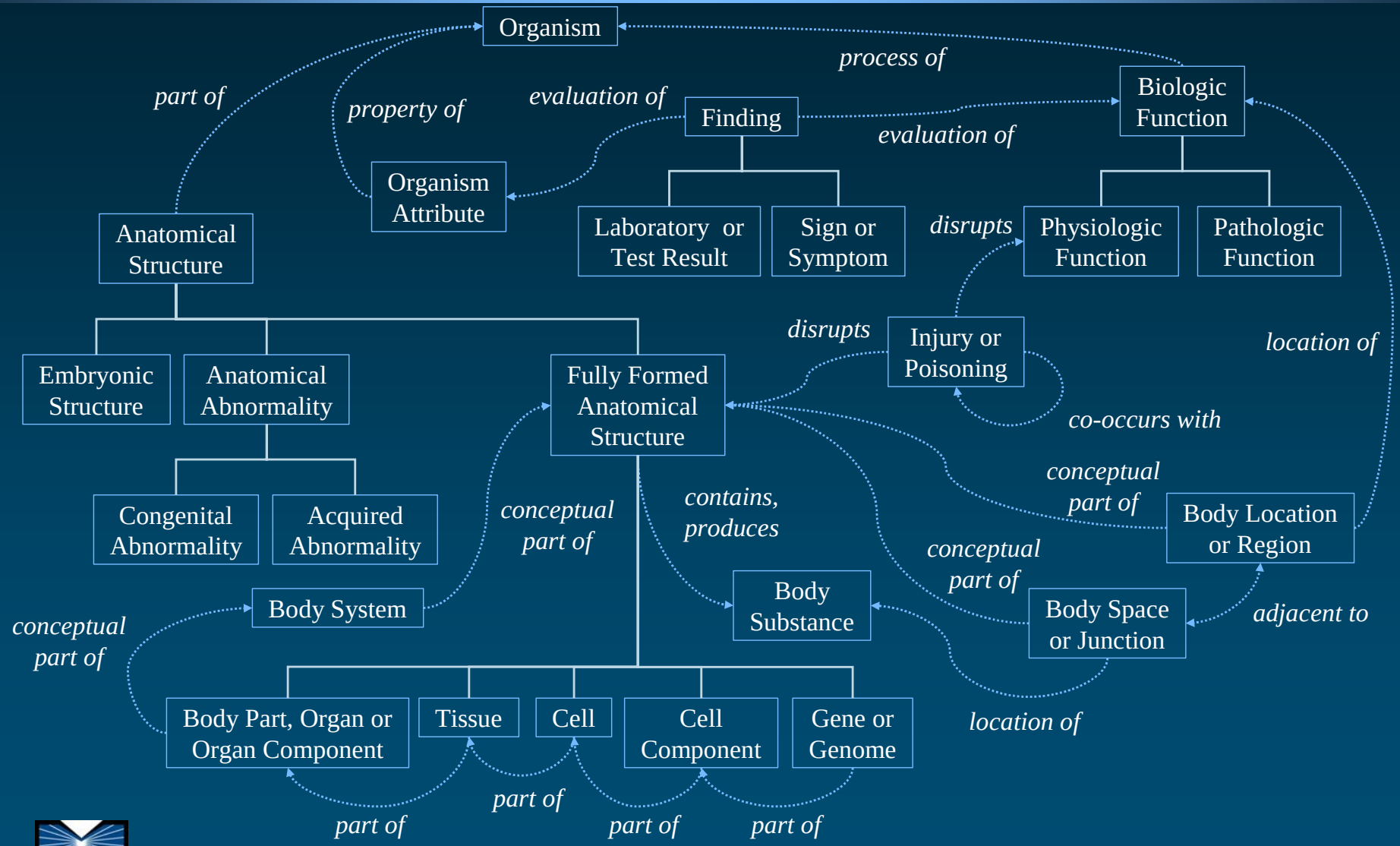
“Biologic Function” hierarchy (isa)



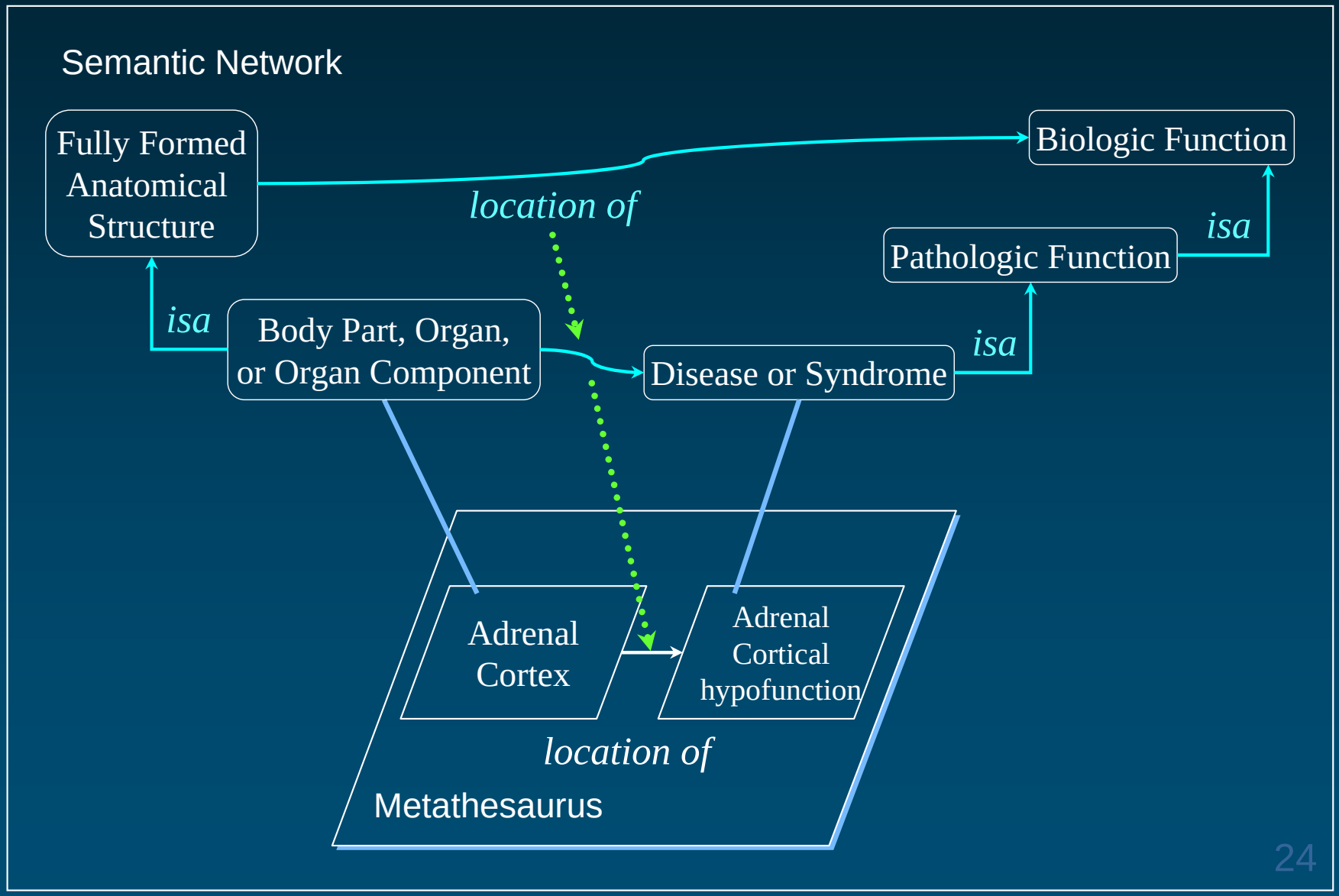
Semantic Network

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

Associative (non-isa) relationships

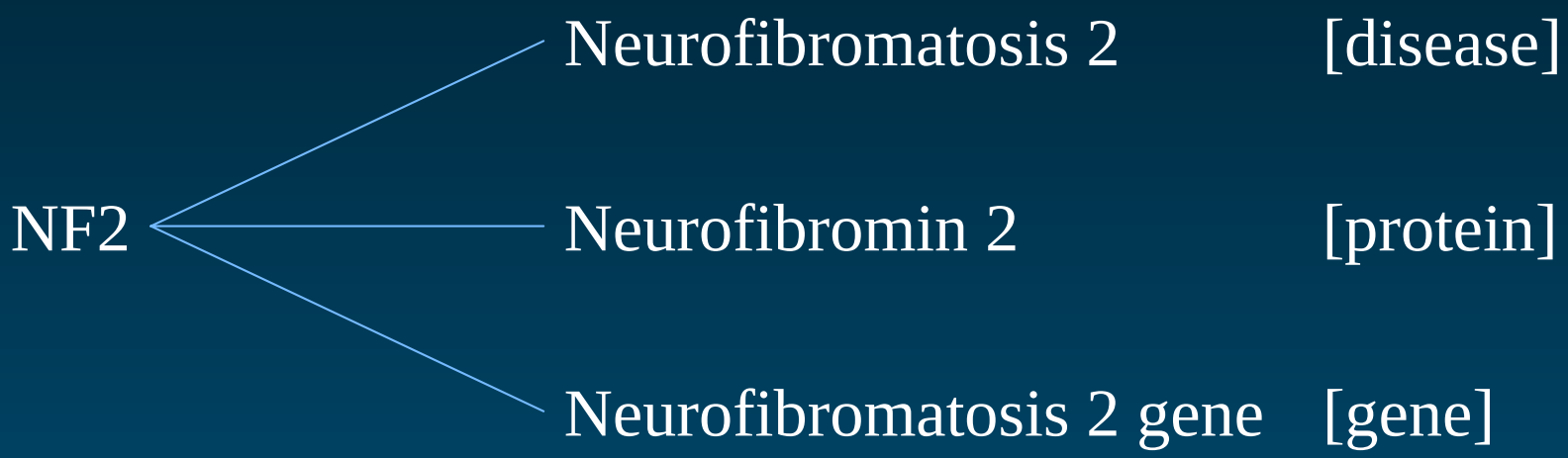


Relationships can inherit semantics



Some issues related to these resources

Ambiguity


NF2
Neurofibromatosis 2 [disease]
Neurofibromin 2 [protein]
Neurofibromatosis 2 gene [gene]

Limited coverage

- e.g., Gene and protein names
  - Additional sources
  - Additional identification methods

Genew	http://www.gene.ucl.ac.uk/nomenclature/
Entrez Gene	http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=gene
UniProt	http://www.ebi.uniprot.org/index.shtml

Conclusions

Conclusions

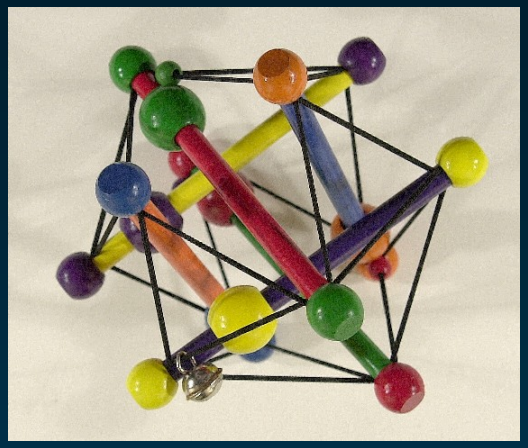
- Lexical and terminological resources enable entity recognition
- Terminological and ontological resources enable relation extraction

But…

- Text mining techniques can also benefit
  - Terminologies: term extraction
  - Ontologies: ontology population

UMLS documentation and support

- UMLS homepage http://umlsinfo.nlm.nih.gov/
  - with links to all other UMLS information
- UMLSKS homepage http://umlsks.nlm.nih.gov/
  - with links to the User's and Developer's guides
- Email address for support custserv@nlm.nih.gov



Medical Ontology Research

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