RESEARCH, CONDITION, AND DISEASE CATEGORIZATION

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NLM Resources for Mining Biomedical Text

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Overview

- An example
- Three types of resources for mining biomedical text
  - Lexical resources
    - SPECIALIST Lexicon
    - Lexical Tools
  - Terminological resources
    - UMLS Metathesaurus
    - MetaMap, MTI
  - Ontological resources
    - UMLS Semantic Network
    - SemRep
- Application: Semantic Medline

An example

Neurofibromatosis 2

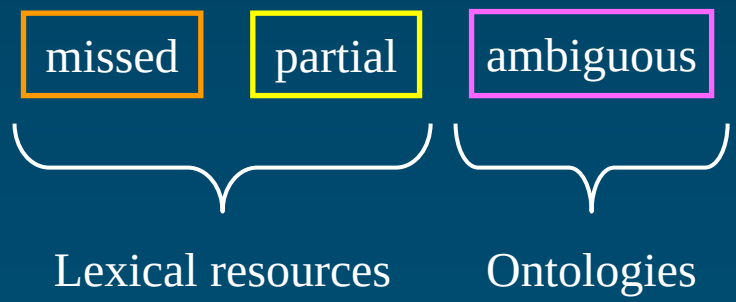
Neurofibromatosis 2

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

NLM resources for mining biomedical text

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

Unified Medical Language System

- SPECIALIST Lexicon
  - 200,000 lexical items
  - Part of speech and variant information
- Metathesaurus
  - 5M names from over 100 terminologies
  - 1.5M concepts
  - 16M relations
- Semantic Network
  - 135 high-level categories
  - 7000 relations among them

Lexical resources

Terminological resources

Ontological resources

Lexical resources

SPECIALIST Lexicon

- *Lexical tools*

Lexical Systems Group

http://umlslex.nlm.nih.gov/

SPECIALIST Lexicon

- Content
  - English lexicon
  - Many words from the biomedical domain
- 200,000+ lexical items
- Word properties
  - morphology
  - orthography
  - syntax
- Used by the lexical tools

```
{
  base=hemoglobin          (base form)
  spelling_variant=haemoglobin
  entry=E0031208           (identifier)
  cat=noun                 (part of speech)
  variants=uncount         (no plural)
  variants=reg             (plural: hemoglobins , haemoglobins)
}
```

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

Terminological resources

UMLS Metathesaurus

- *MetaMap*
- *Medical Text Indexer (MTI)*

http://www.nlm.nih.gov/research/umls/

INDEXING INITIATIVE

http://ii.nlm.nih.gov/

Source Vocabularies (2007AB)

- 143 source vocabularies
  - 17 languages
- Broad coverage of biomedicine
  - 5.9M names
  - 1.4M concepts
  - 16M relations
- Common presentation

Organize terms

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

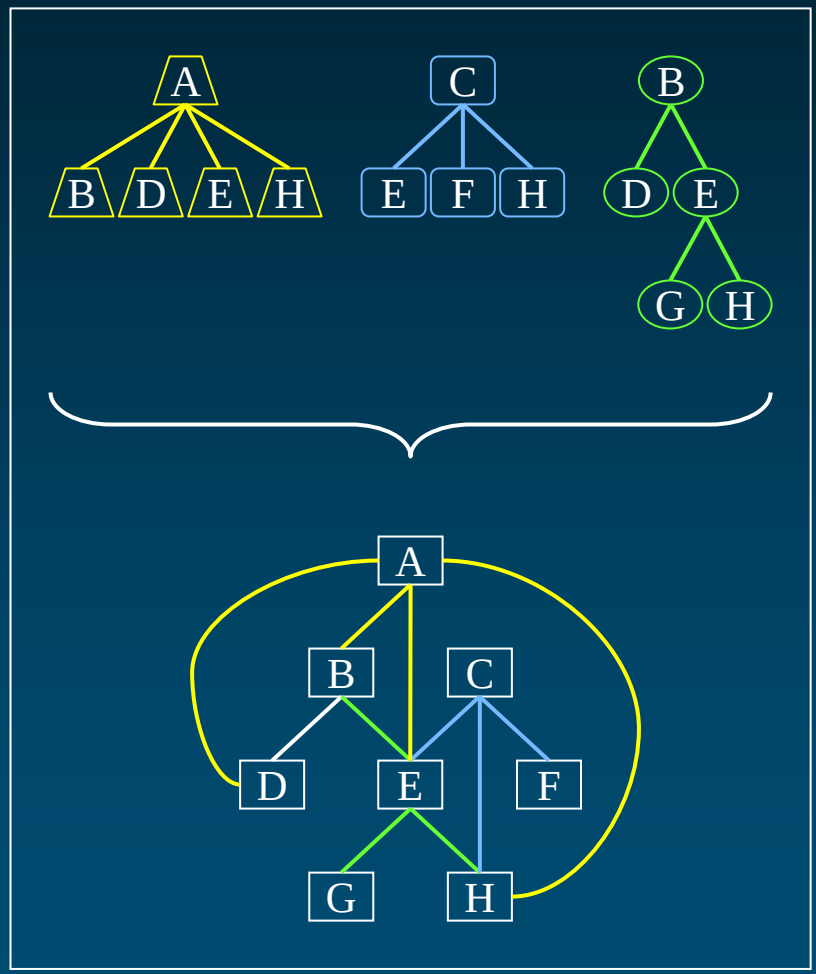
Addison Disease	MeSH	D000224
Primary hypoadrenalism	MedDRA	10036696
Primary adrenocortical insufficiency	ICD-10	E27.1
Addison's disease (disorder)	SNOMED CT	363732003

C0001403

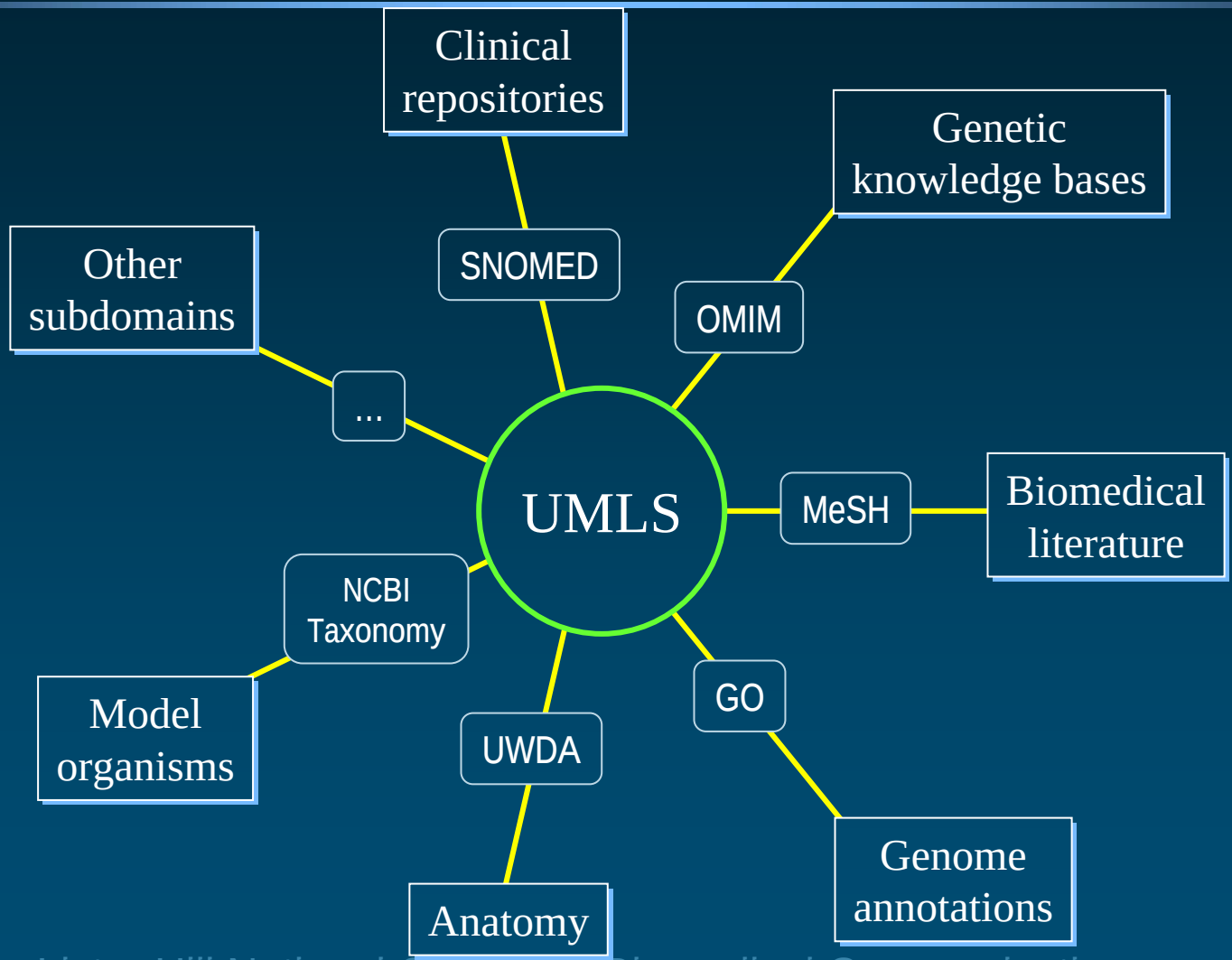
Addison's disease

Organize concepts

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



Integrating subdomains



MetaMap

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.

C0027832 C0027832 C0027831 C0027832 C0027859 C0027832 C0026882 C0254123 C0008665

C0254123

Neurofibromin 2	MeSH
Merlin	SNOMED CT
Schwannomin	MeSH
Schwannomerlin	NCI Thesaurus

INDEXING INITIATIVE

http://ii.nlm.nih.gov/

Medical Text Indexer

- Semi-automatic indexing of MEDLINE citations
  - Suggest MeSH main headings
  - Complement to manual indexing
  - Integrated into the DCMS indexing environment
- Automatic indexing of collections not indexed previously

INDEXING INITIATIVE

http://ii.nlm.nih.gov/

Ontological resources

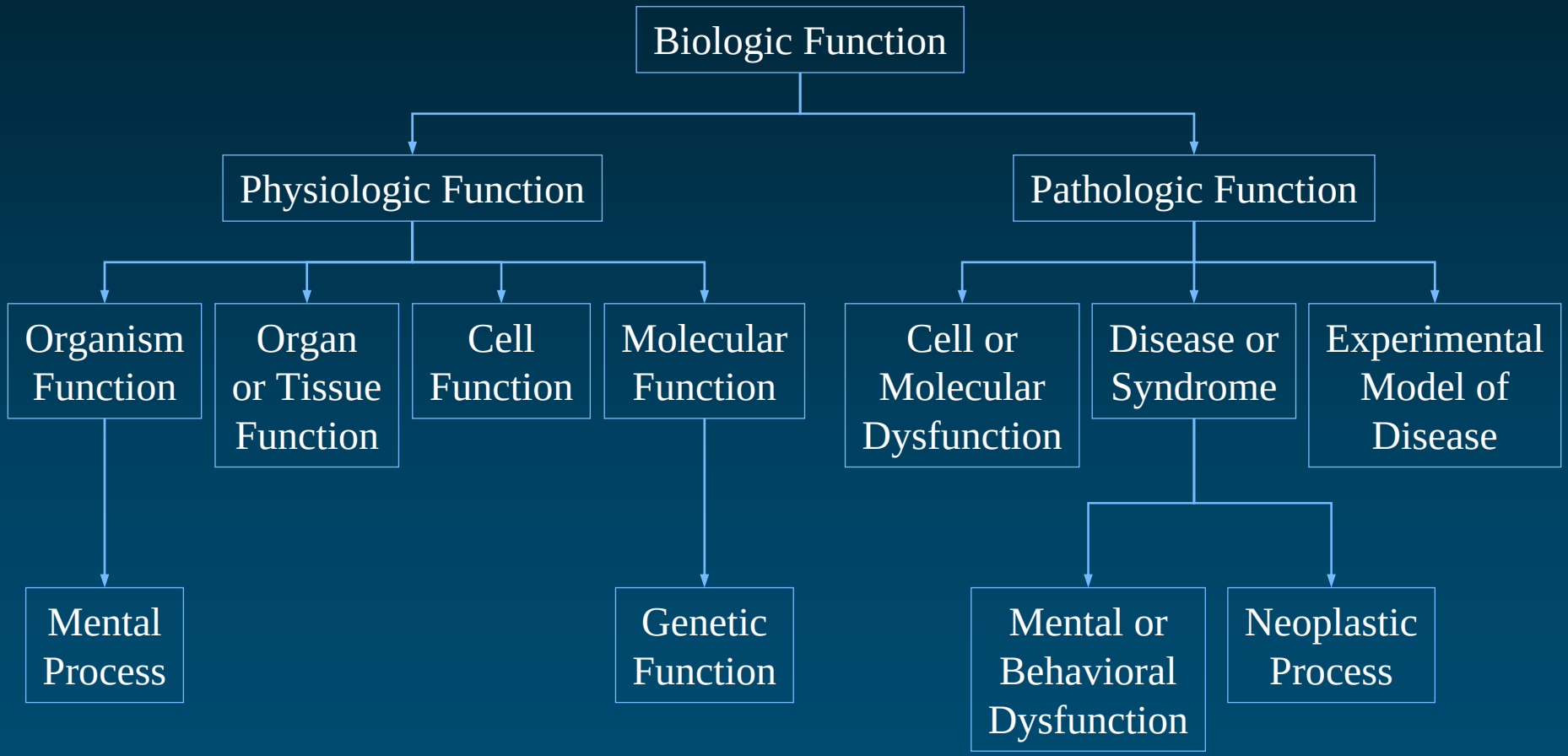
UMLS Semantic Network

http://semanticnetwork.nlm.nih.gov/

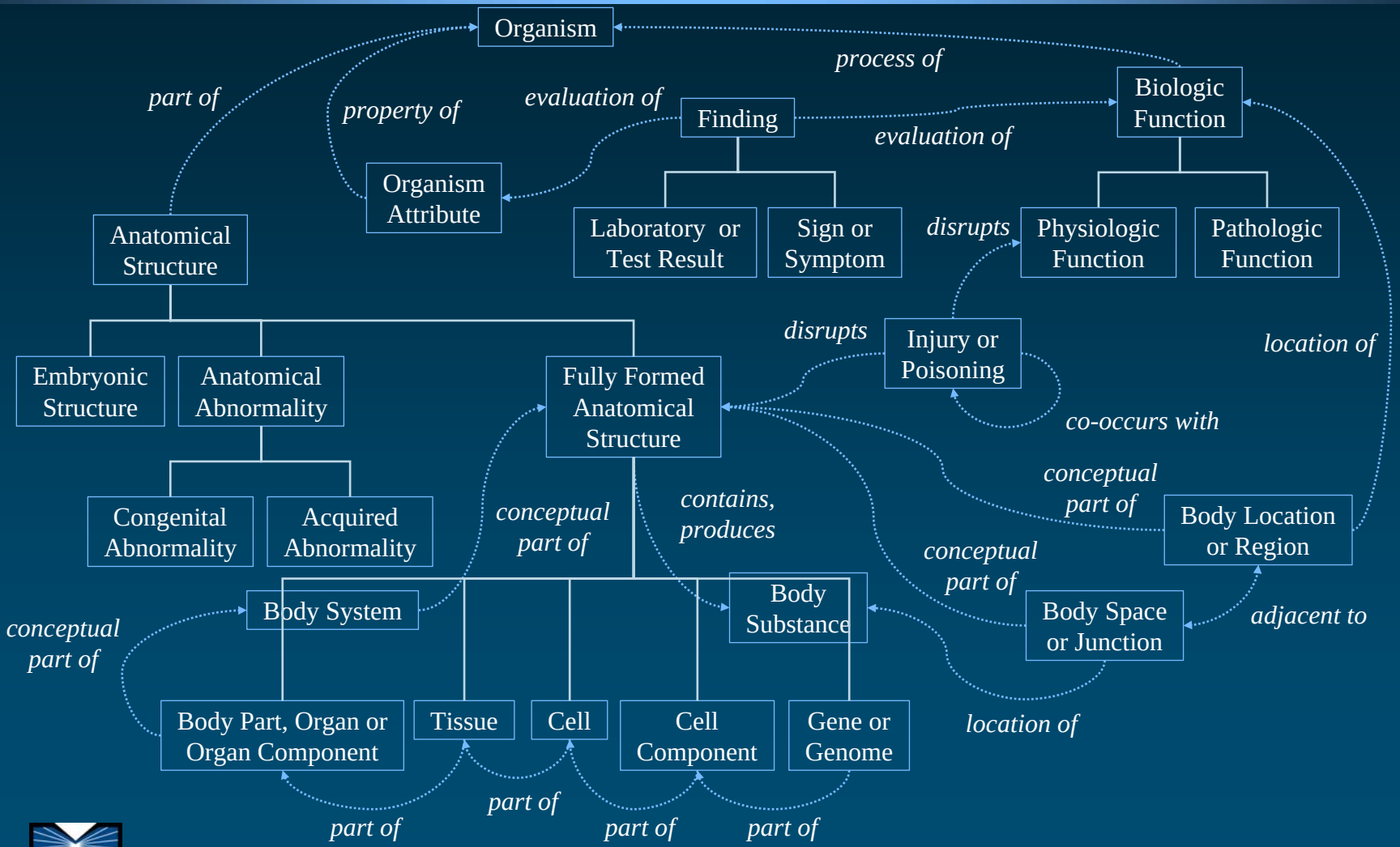
- *SemRep*

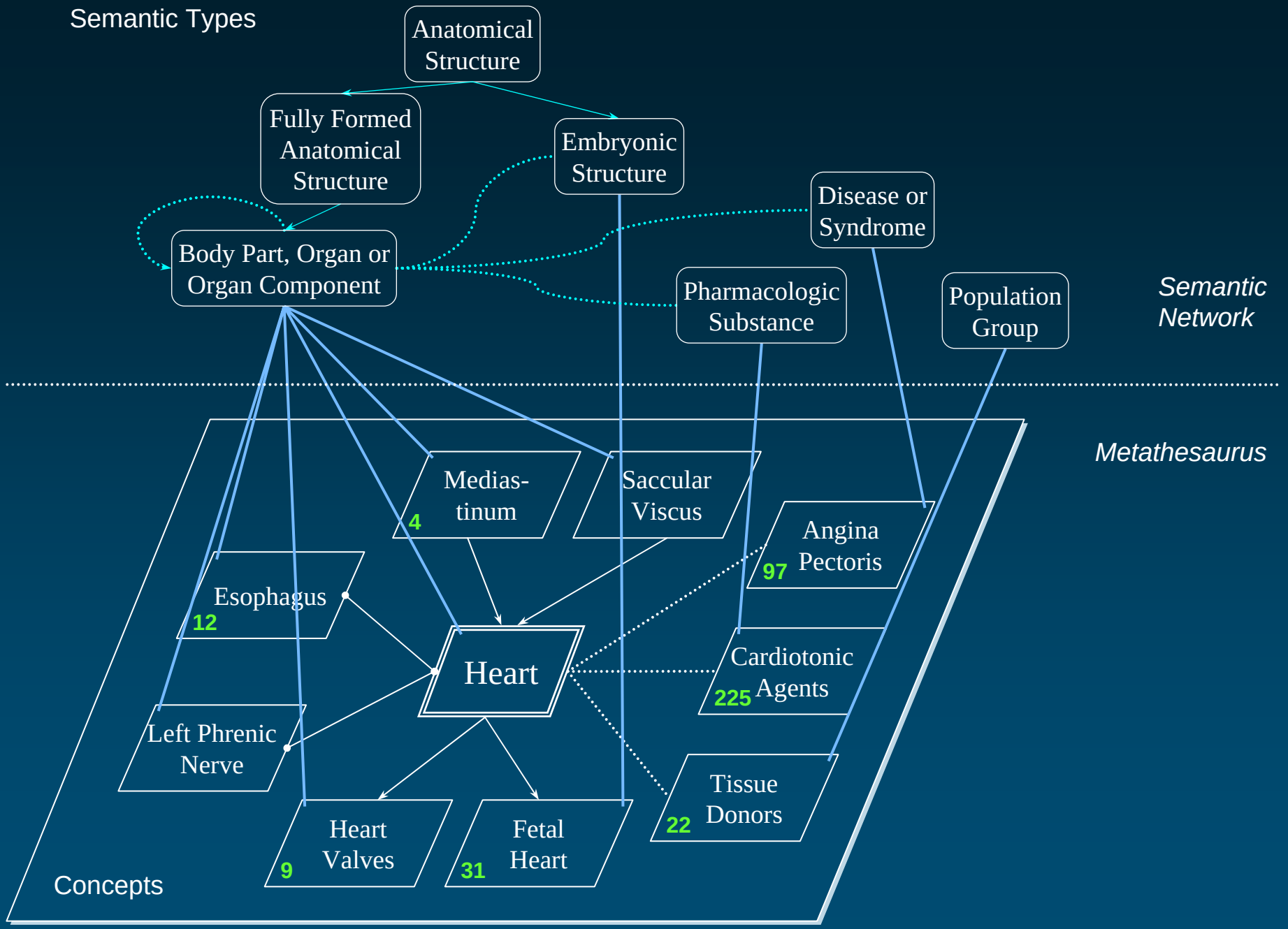
http://skr.nlm.nih.gov/

“Biologic Function” hierarchy (isa)

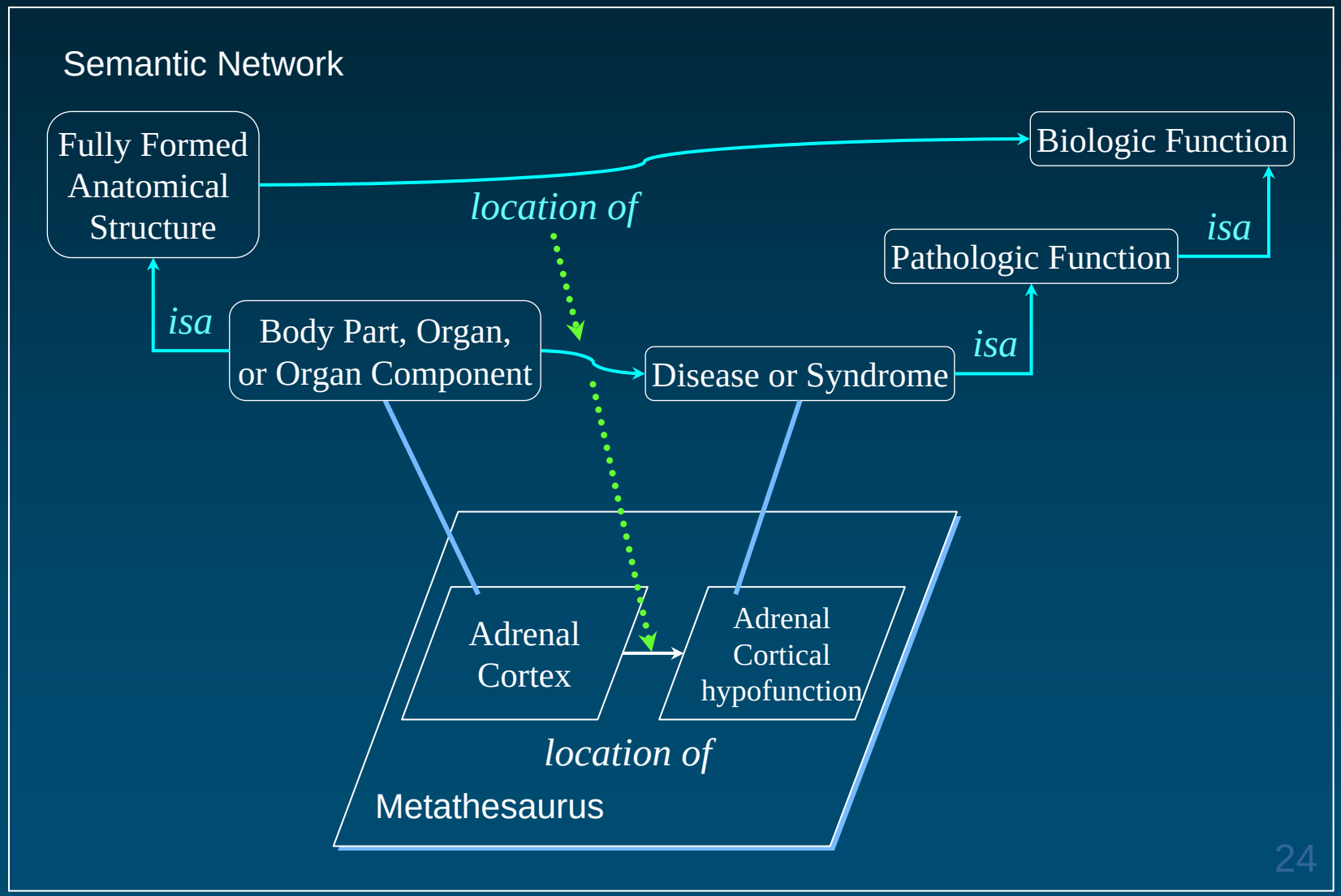


Associative (non-isa) relationships




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
4
Saccular Viscus
Angina Pectoris
97
Esophagus
12
Heart
Cardiotonic Agents
225
Left Phrenic Nerve
Tissue Donors
22
Heart Valves
9
Fetal Heart
31
Concepts

Relationships can inherit semantics



SemRep Relation extraction

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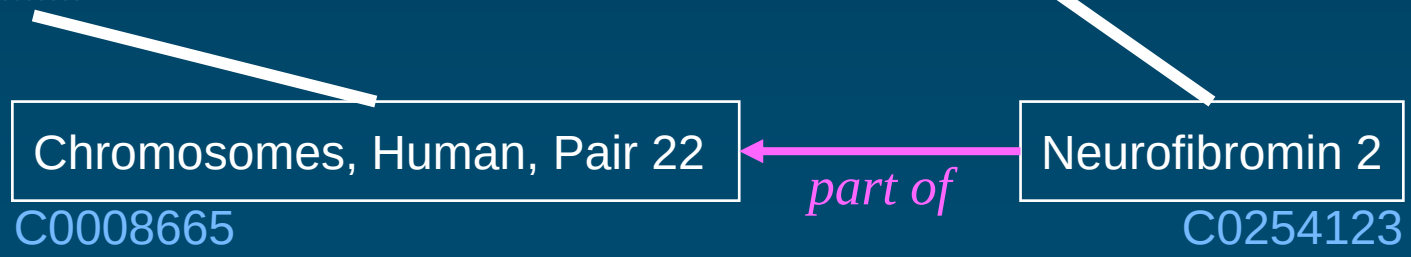
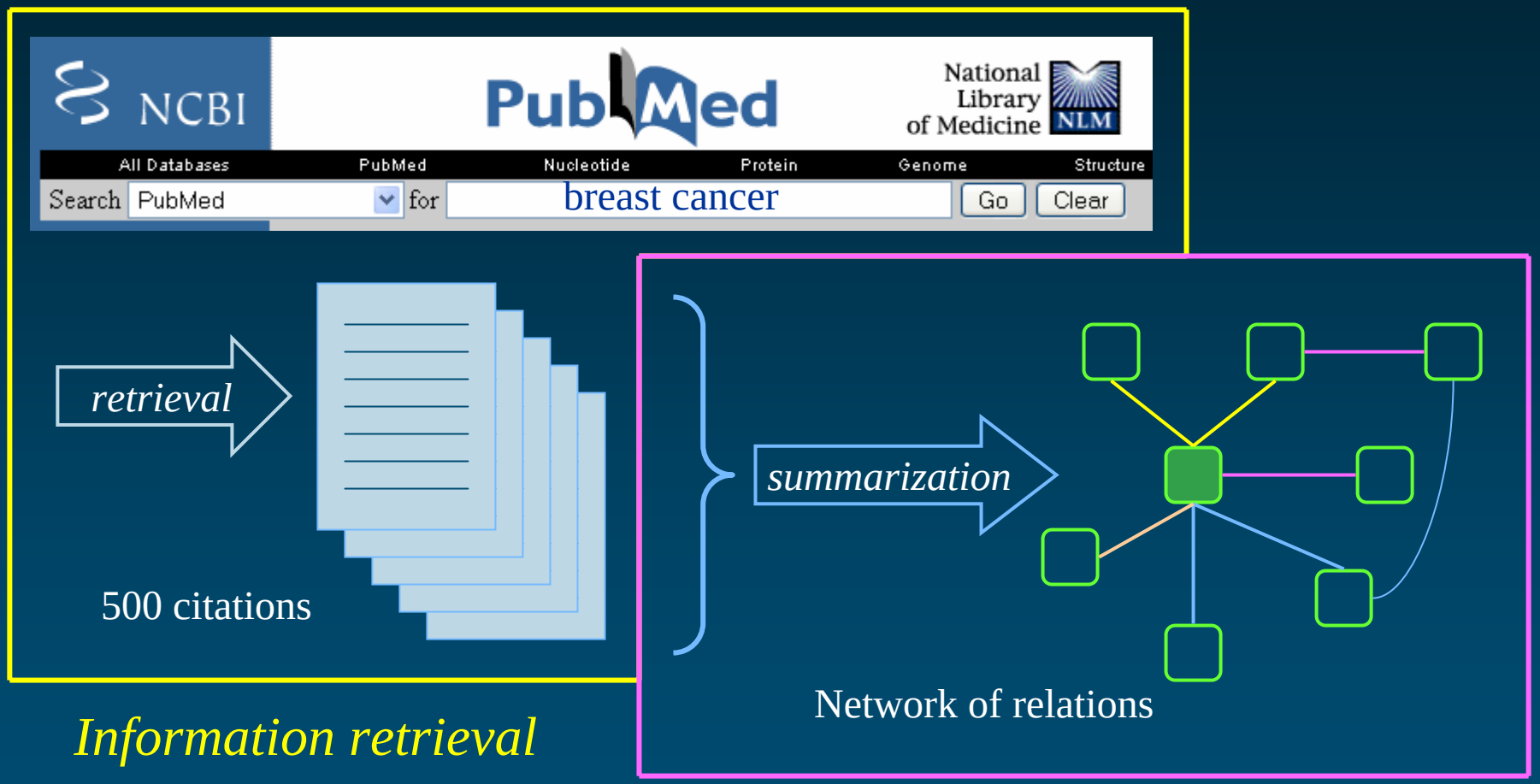


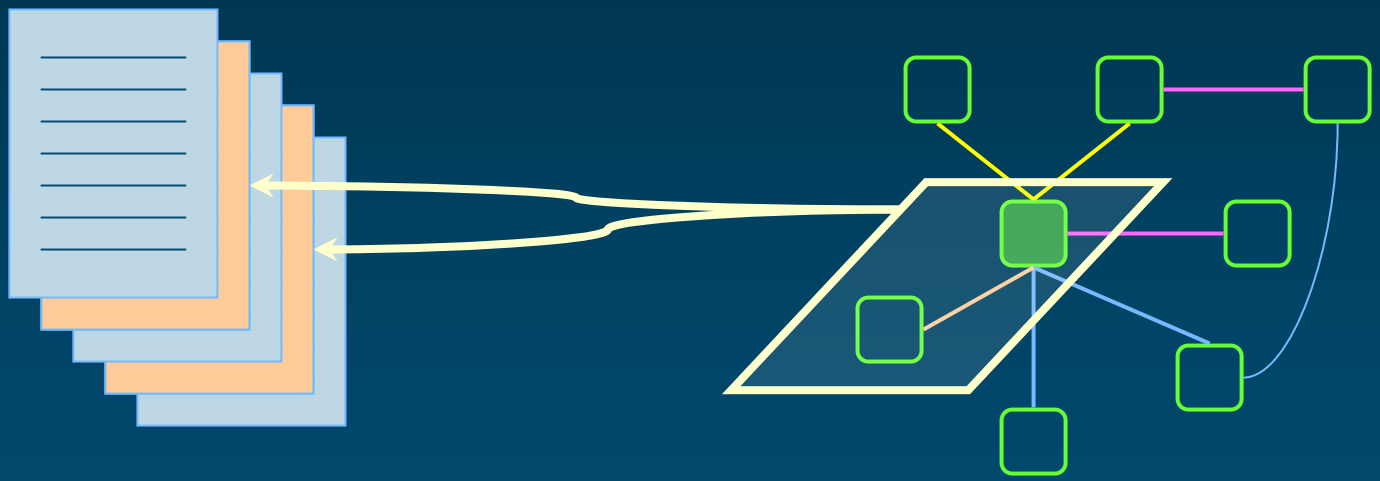
NLM resources for mining biomedical text in action

Semantic Medline

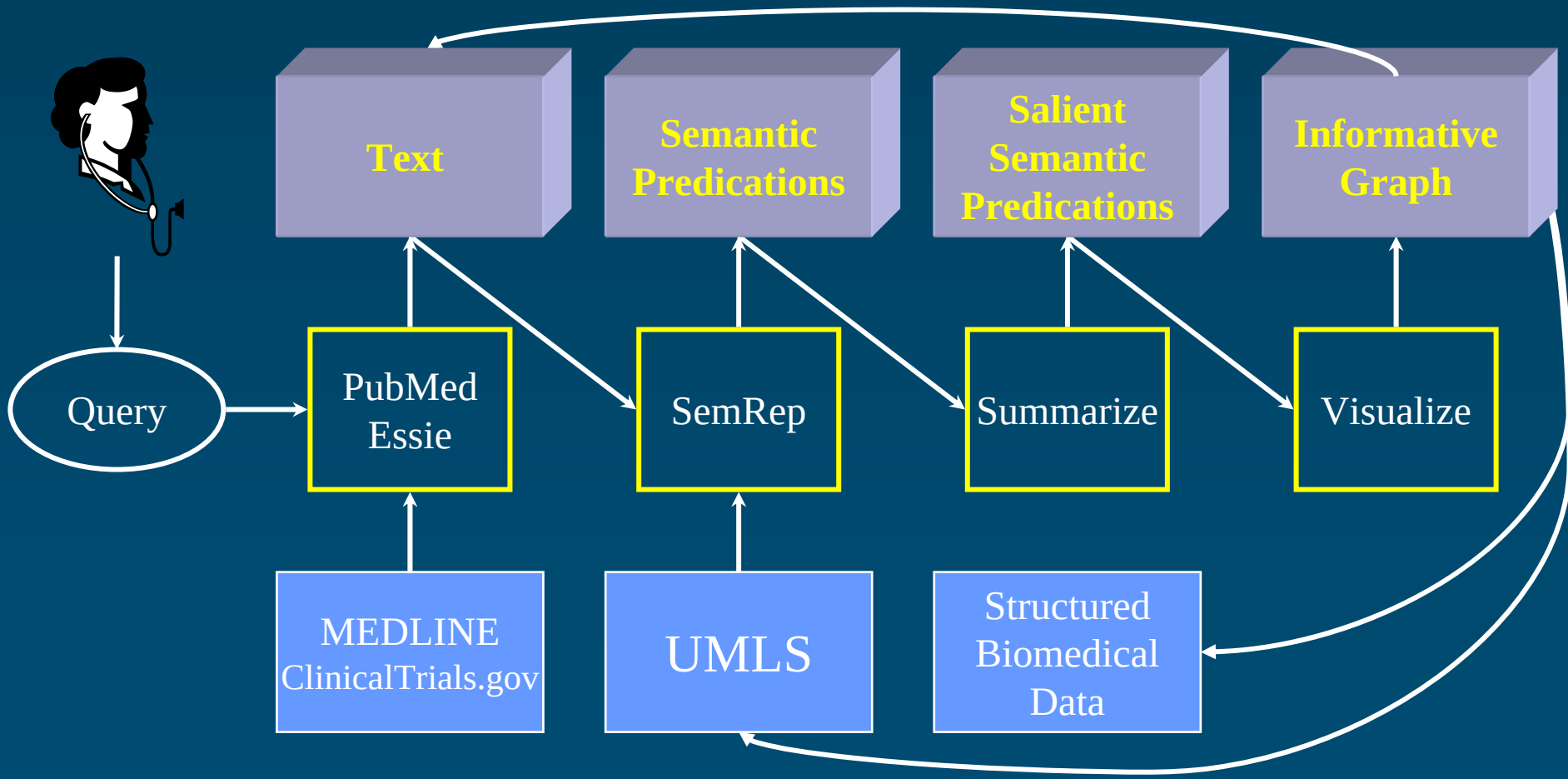
Managing retrieval results



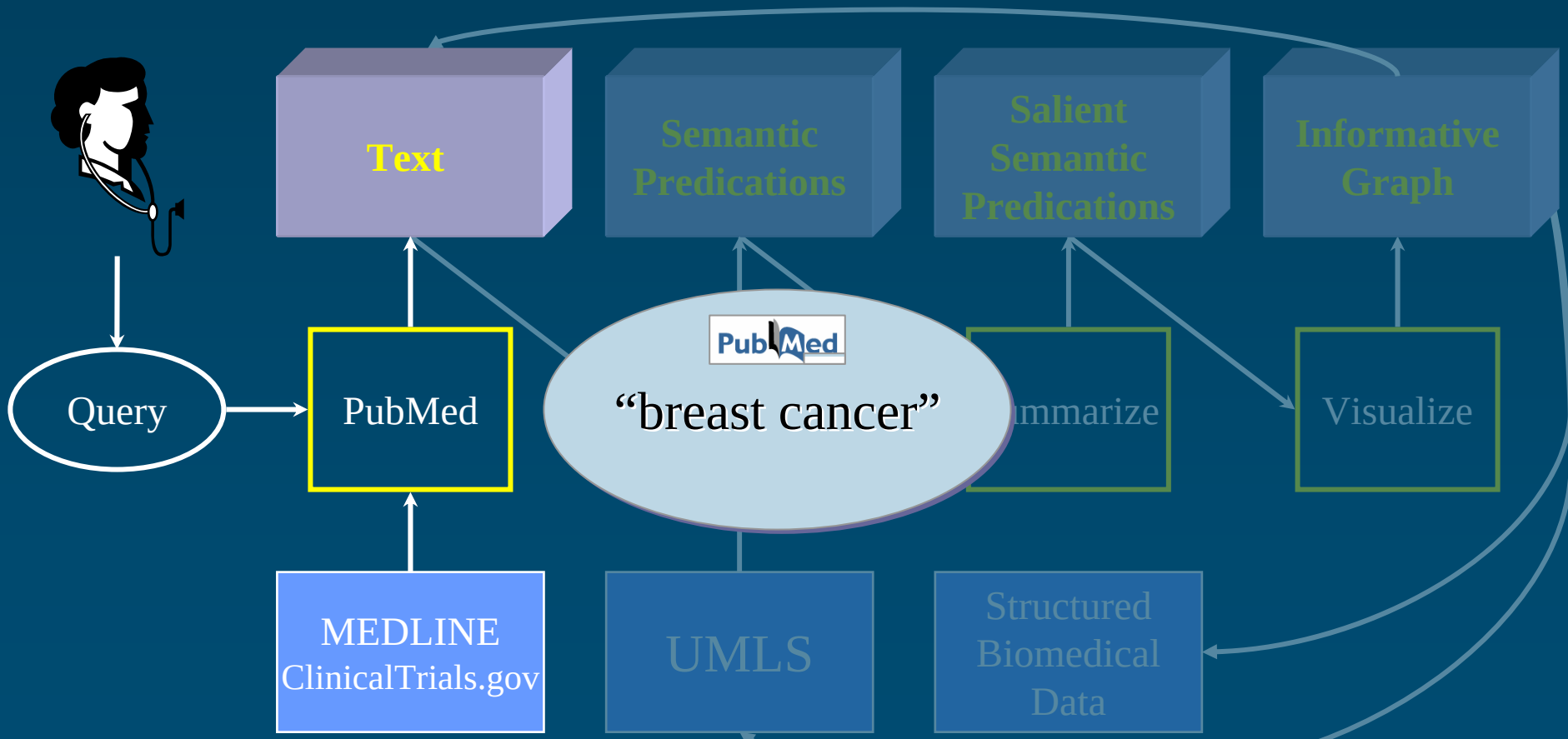
Managing retrieval results



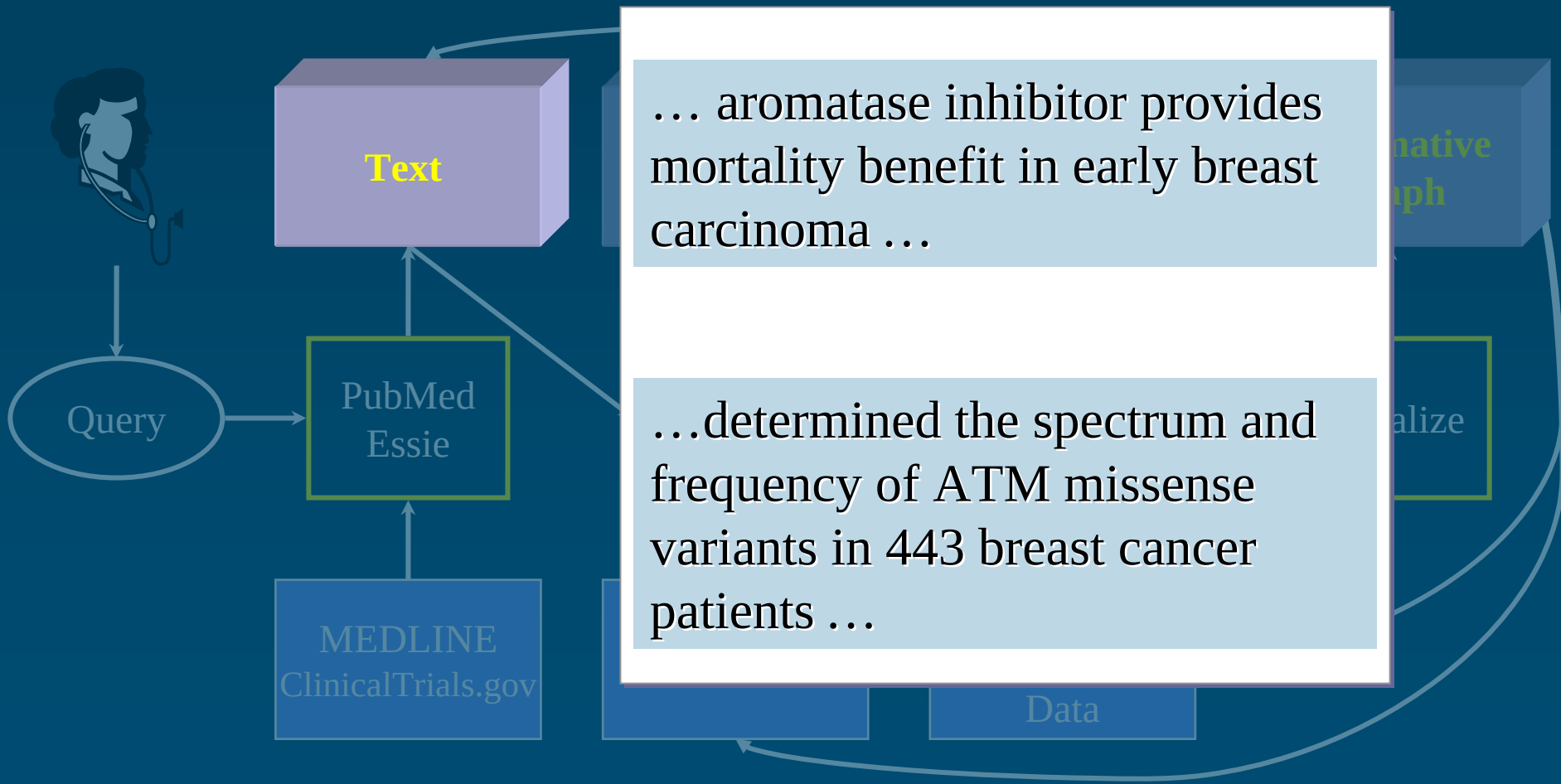
Semantic Medline Overview



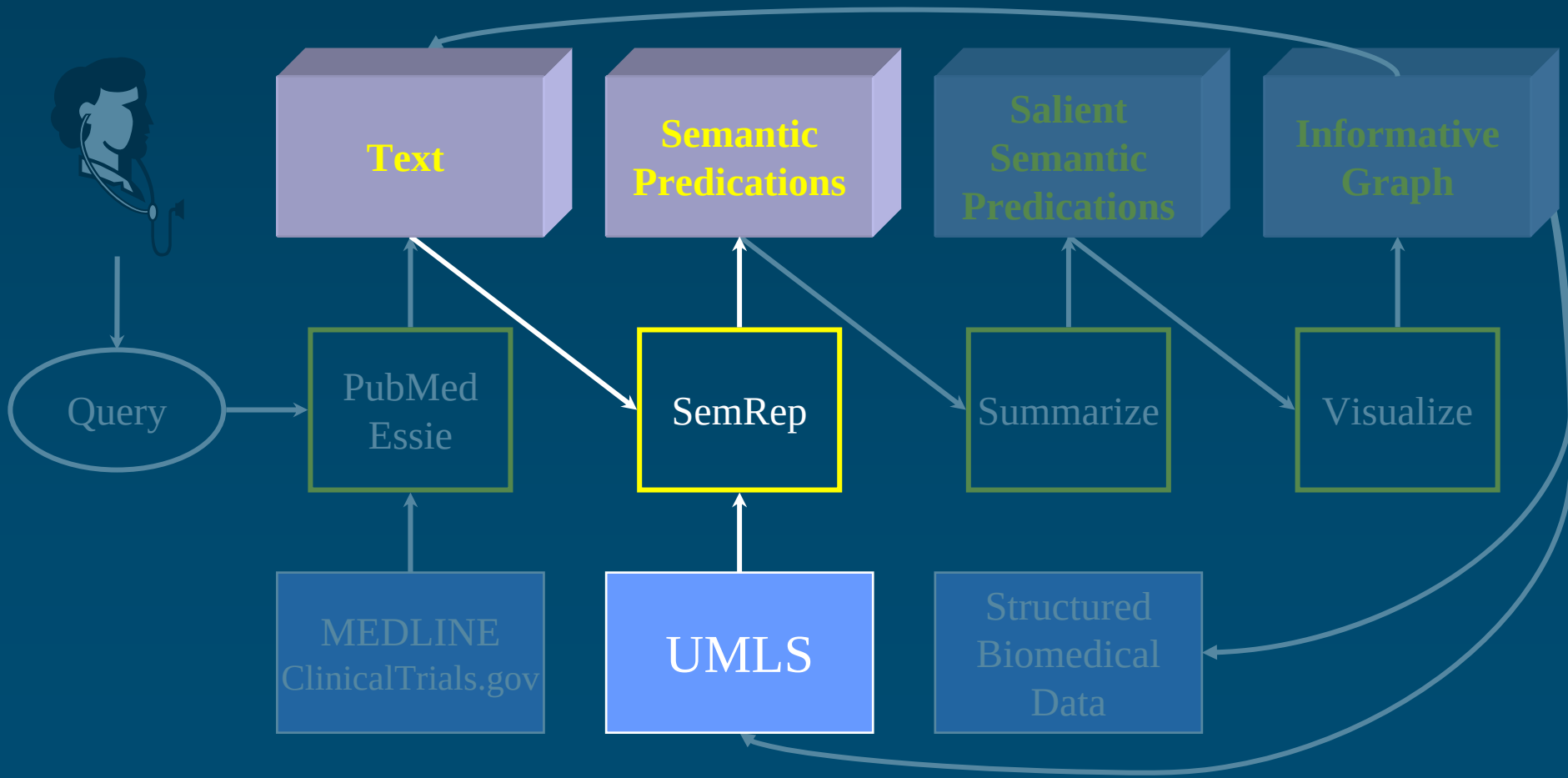
Document selection



MEDLINE citations

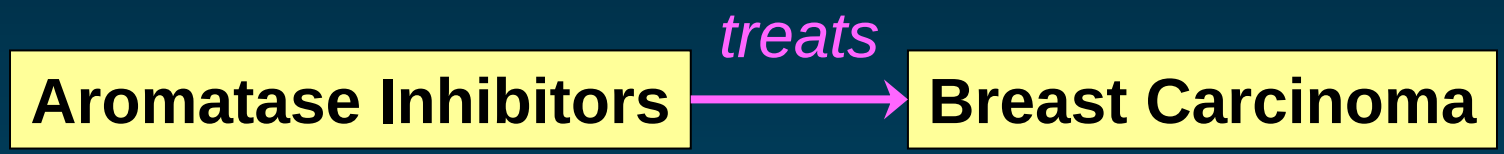


Semantic intepretation



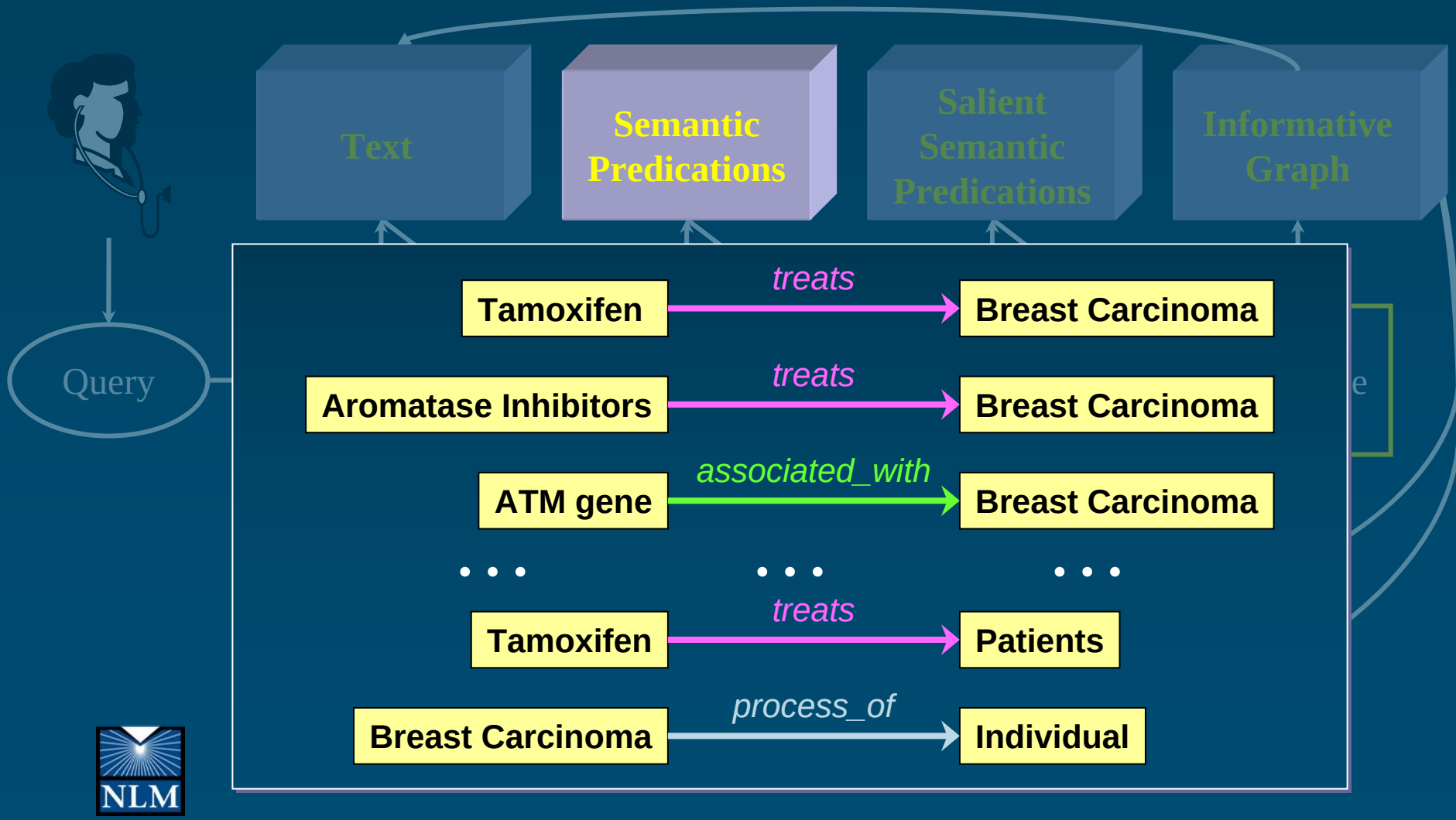
Semantic interpretation

... aromatase inhibitor provides mortality benefit in early breast carcinoma …

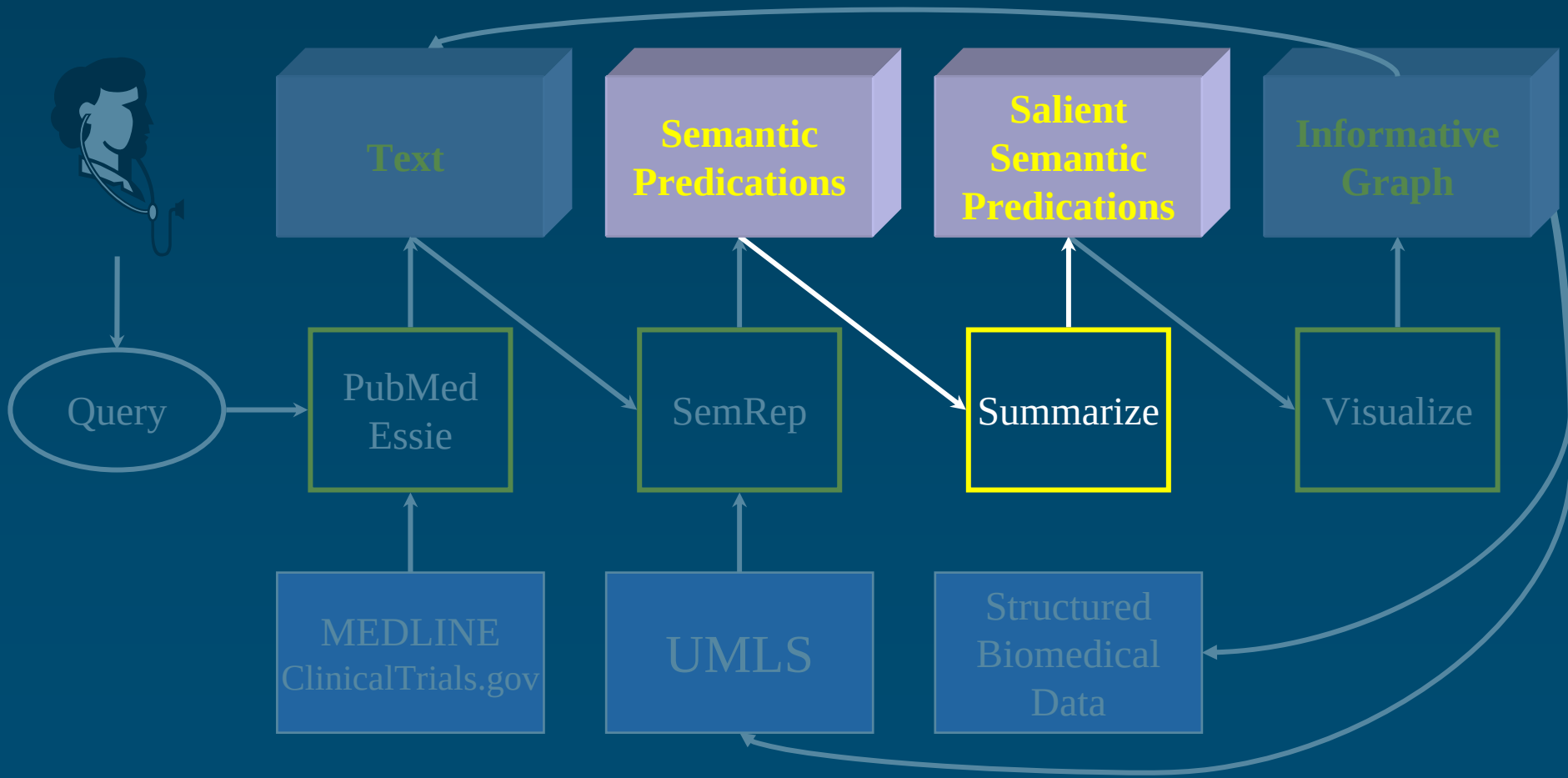


… determined the spectrum and frequency of ATM missense variants in 443 breast cancer patients …

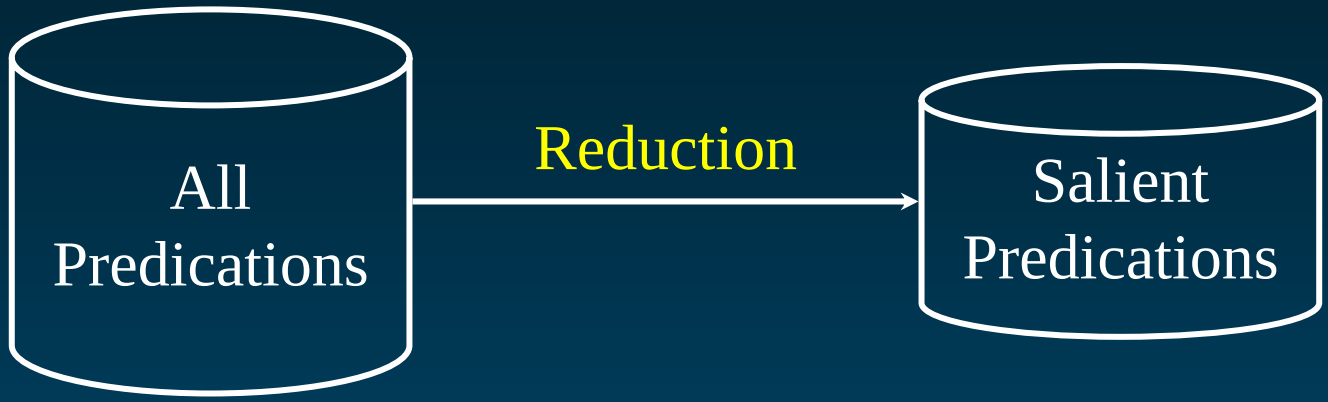
Semantic predications



Summarization

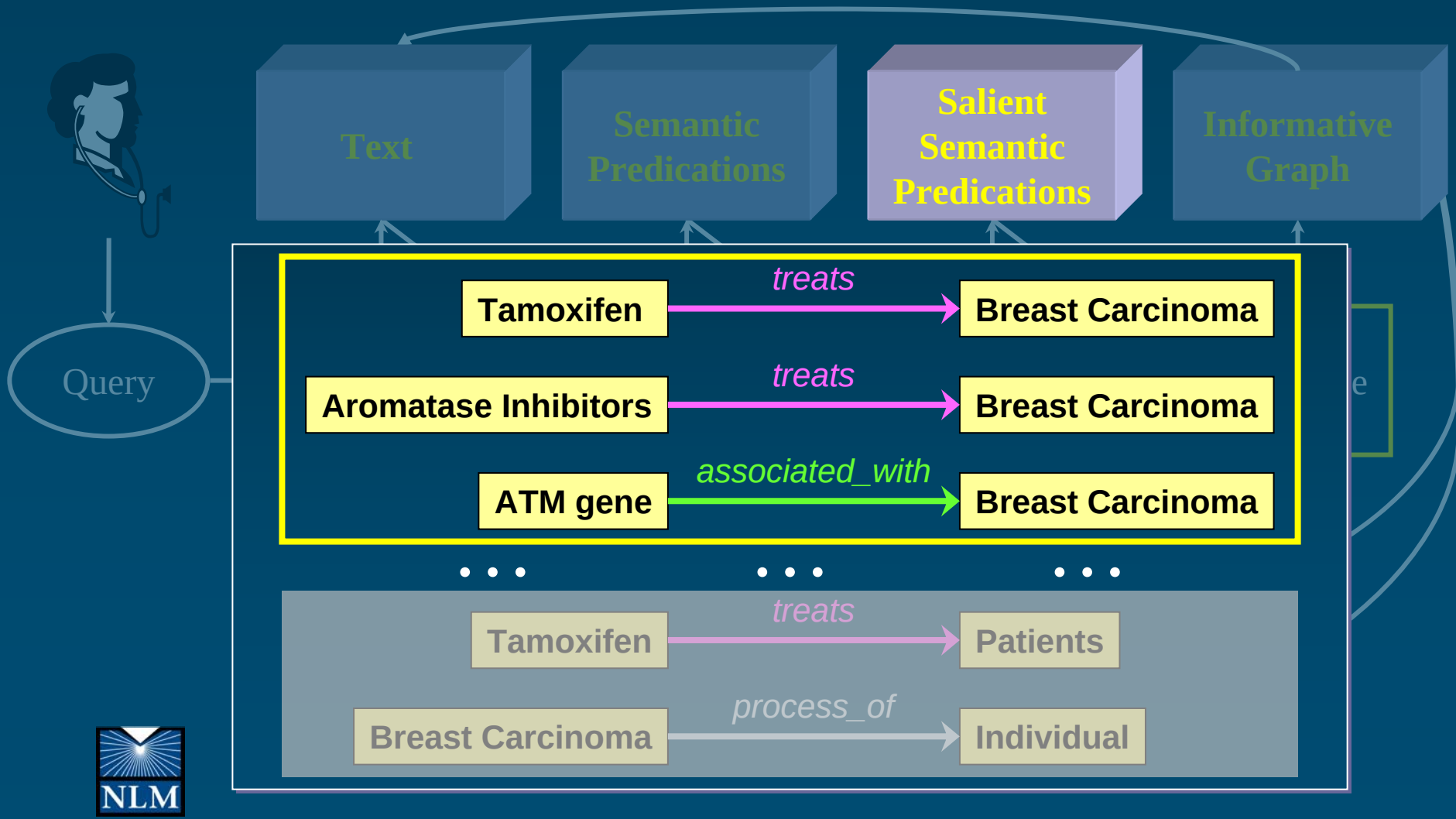


Abstraction summarization

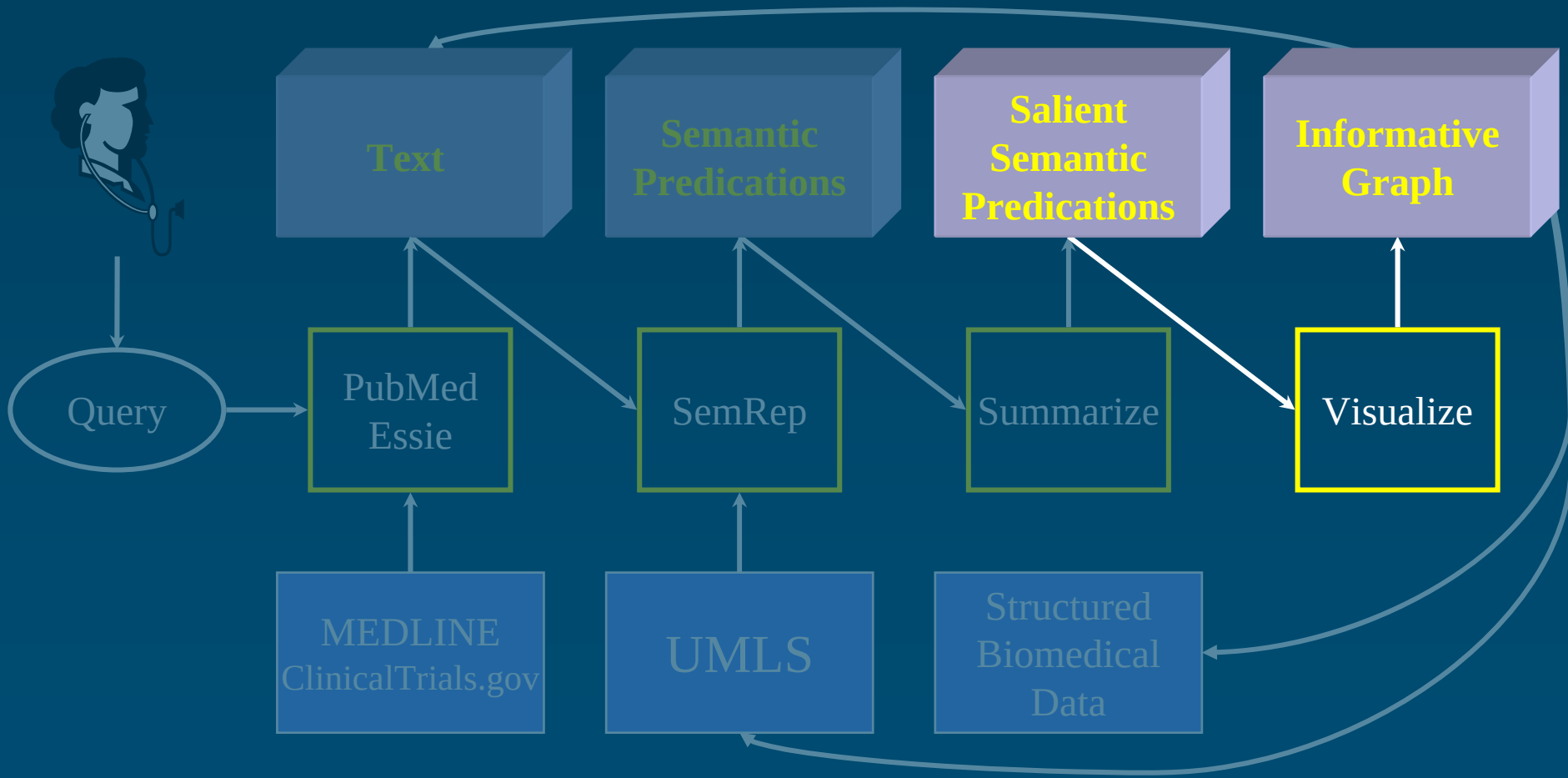


- Specify a topic
- Retain predications on the topic
- Eliminate uninformative predications
- Retain most frequent predications

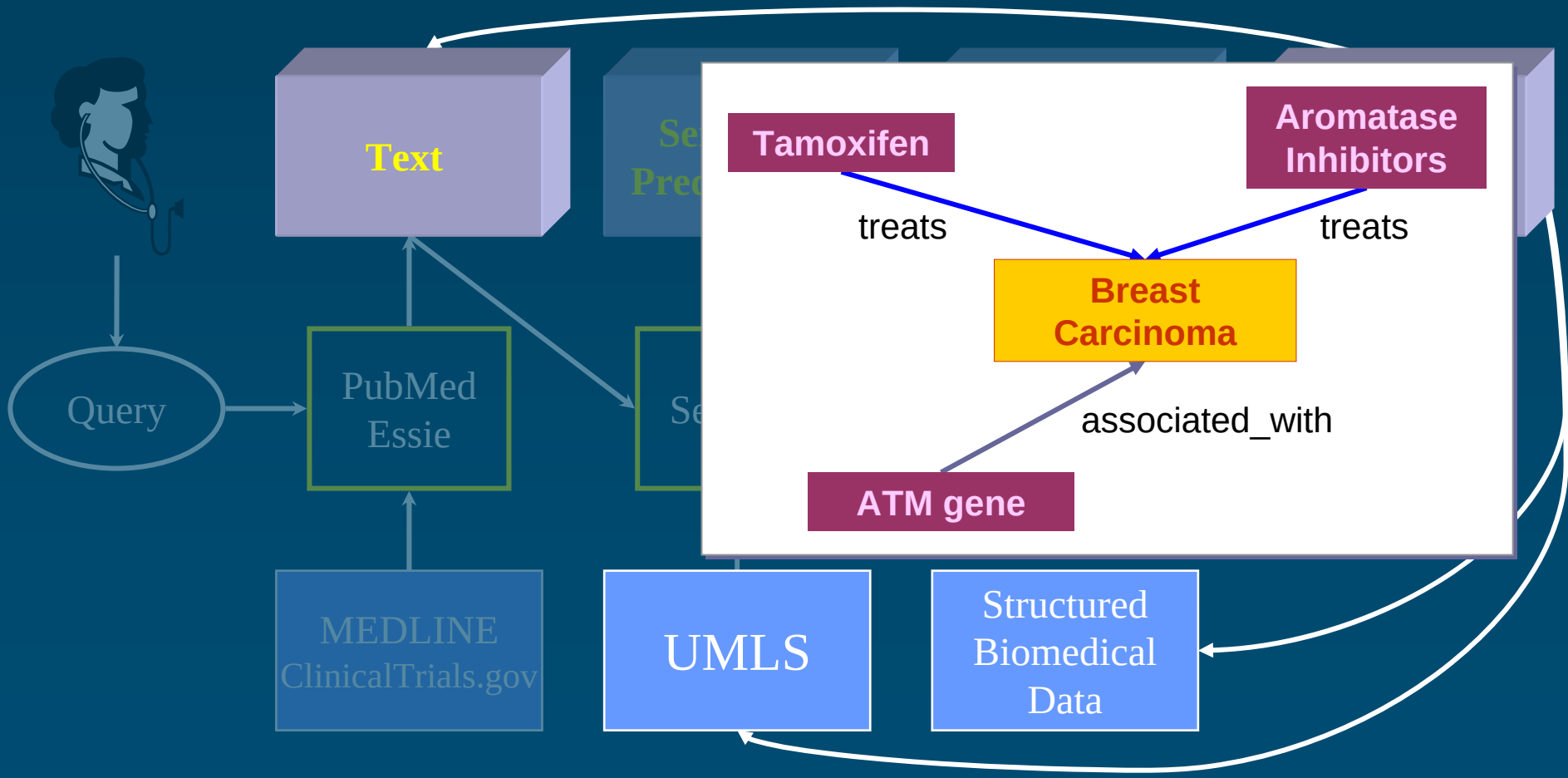
Salient semantic predications



Visualization



Informative graph



Semantic Medline Live