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Comparing SNOMED CT and the NCI Thesaurus through Semantic Web Technologies

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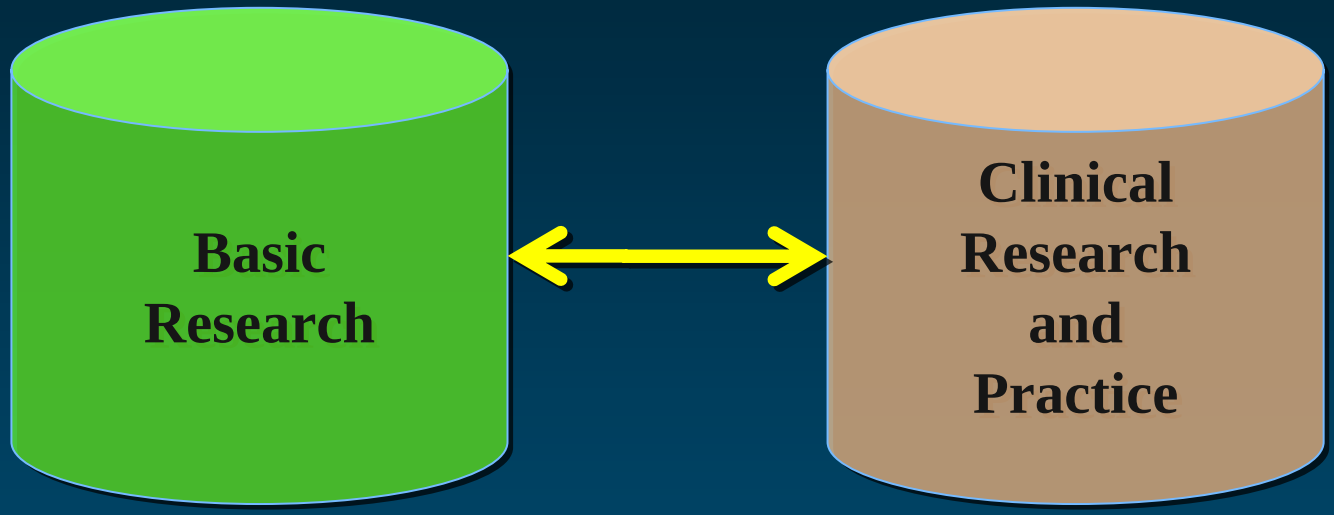
Motivation Translational research

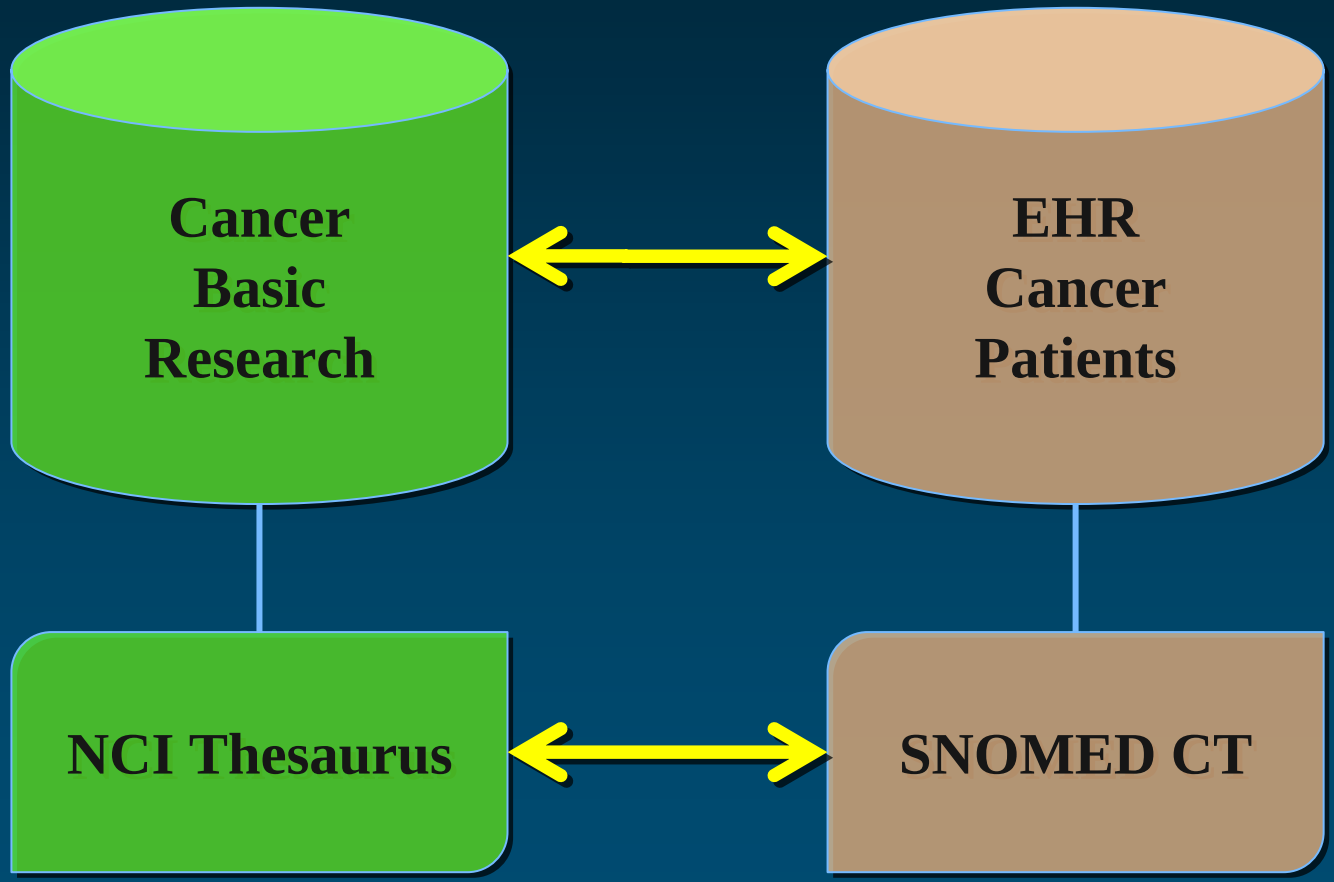
- "Bench to Bedside"
- Integration of clinical and research activities and results
- Supported by research programs
  - NIH Roadmap
  - Clinical and Translational Science Awards (CTSA)
- Requires the effective integration and exchange and of information between
  - Basic research
  - Clinical research

Translational research NIH Roadmap

Motivation Translational research

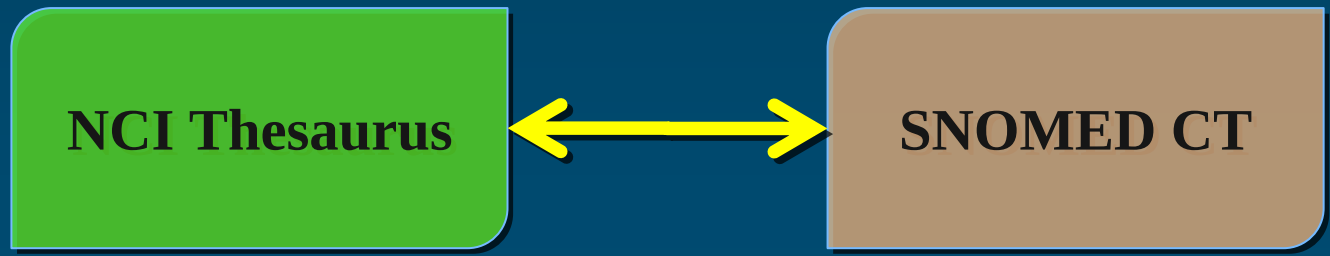


Terminology and translational research



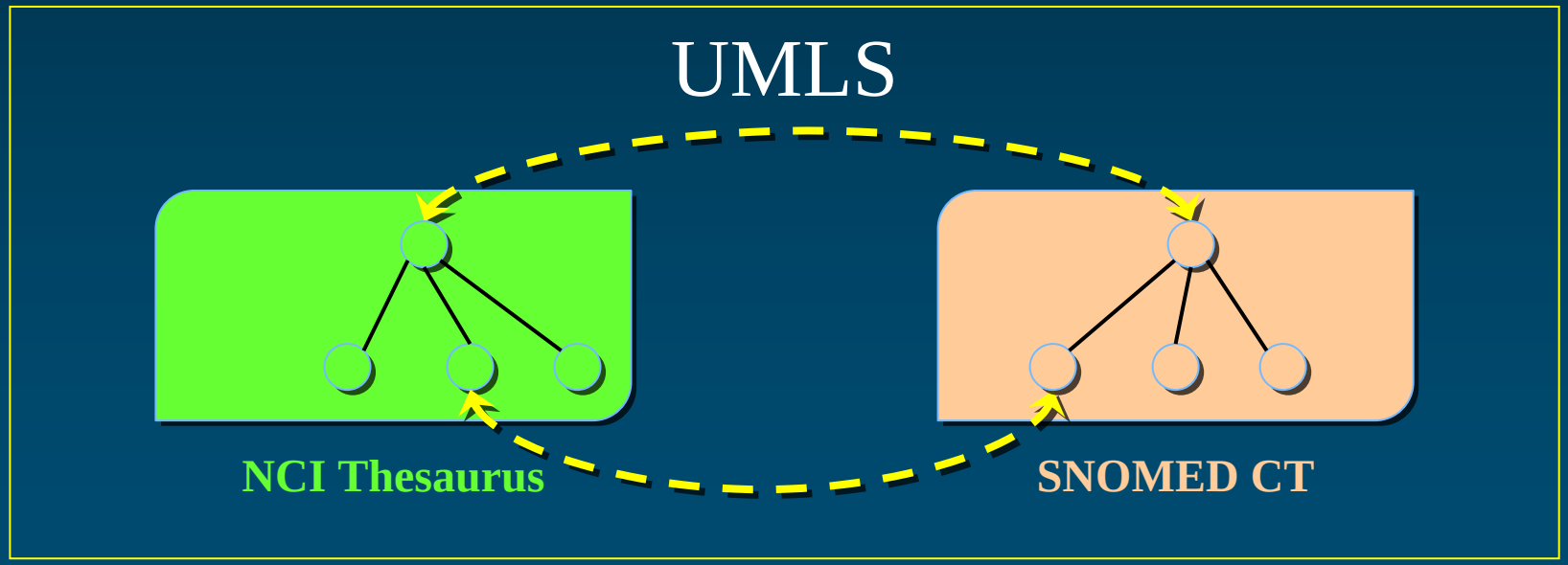
Objectives

- To evaluate interoperability between data repositories for cancer research (basic/clinical) at the terminological level
  - To compare the formal definitions of NCI Thesaurus (NCIt) and SNOMED CT concepts
  - Evaluate Semantic Web technologies in this context



Hypotheses

- Equivalent concepts should have similar formal definitions
  - Concept equivalence is provided by the UMLS



Semantic Web technologies

- Collection of languages, formalisms and tools developed to support the Semantic Web
  - XML
  - Resource Description Framework (RDF)
  - Web Ontology Language (OWL)

- RDF
  - Relations expressed as triples, forming a graph
  - Both NCIt and SNOMED CT can be easily converted to RDF

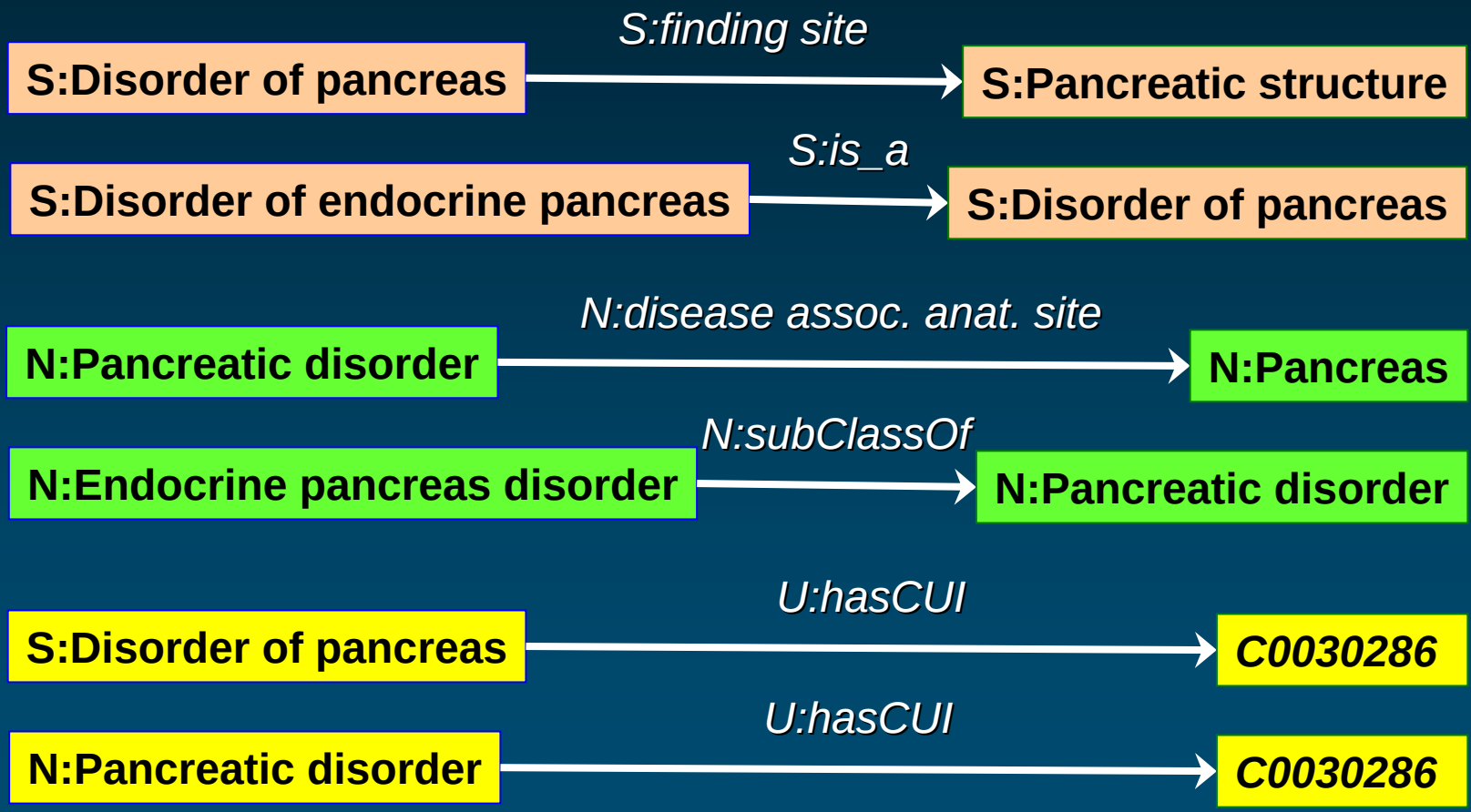
Methods Outline

- Acquiring RDF triples
- Adding entailed statements through inference rules
- Querying the triple store (RDF graph)
- Comparing shared relata of NCIt and SNOMED CT concepts

Methods Acquiring RDF triples

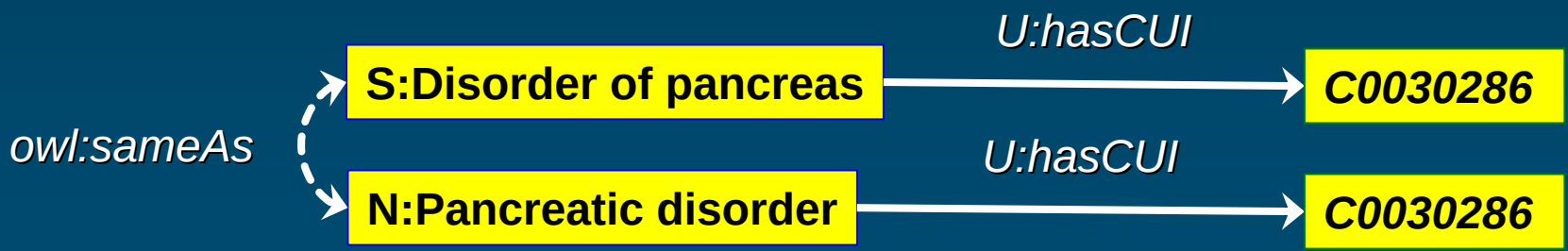
- SNOMED CT
  - CONCEPTID as the identifier
  - Relationships: from the RELATIONSHIP table
- NCIt
  - CODE as the identifier
  - Relationships: from the RELATIONSHIP table
- UMLS
  - Link between SCUI and CUI
  - Relationship: hasCUI

Methods Acquiring RDF triples

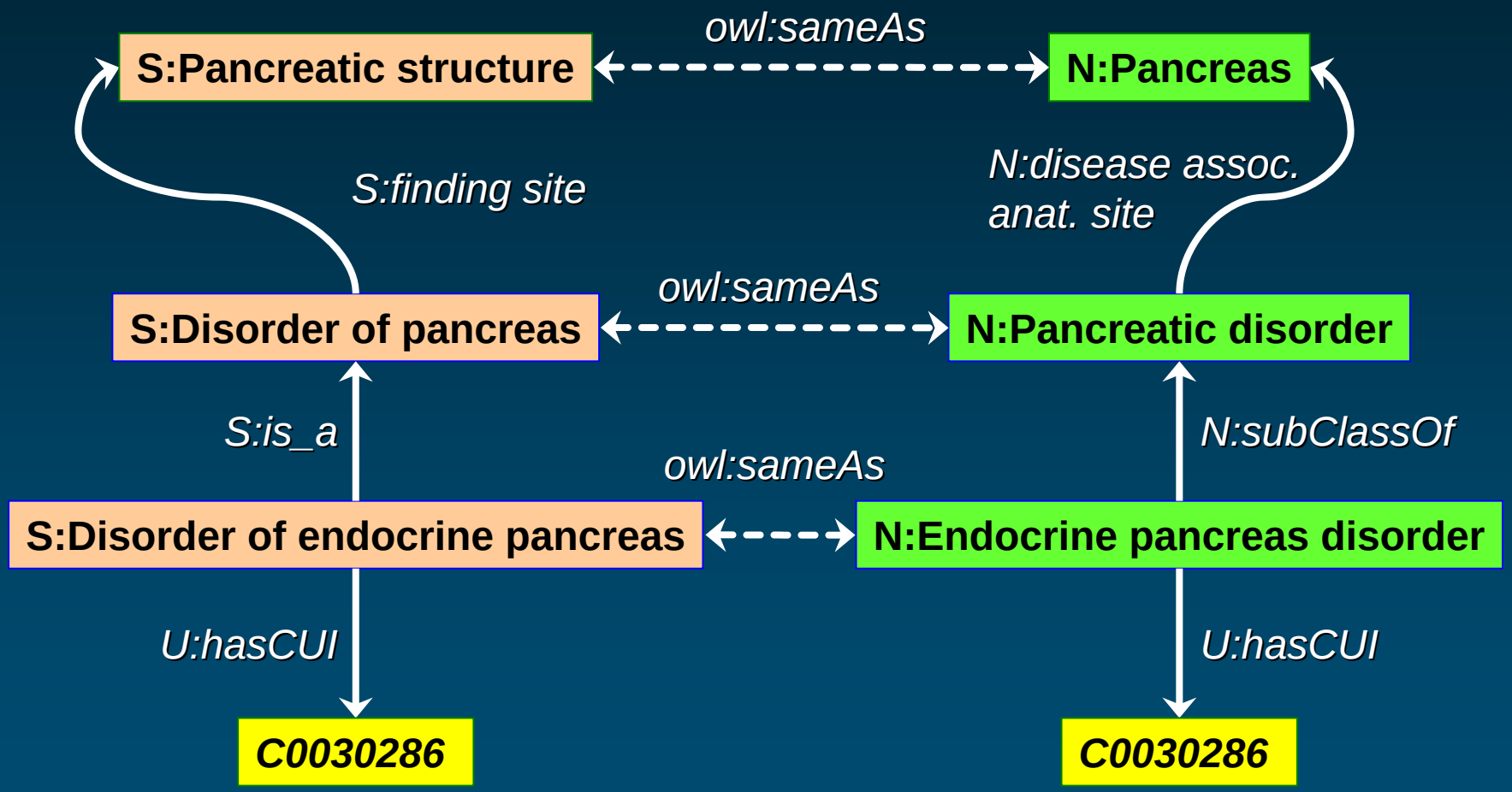


Methods Inference rules

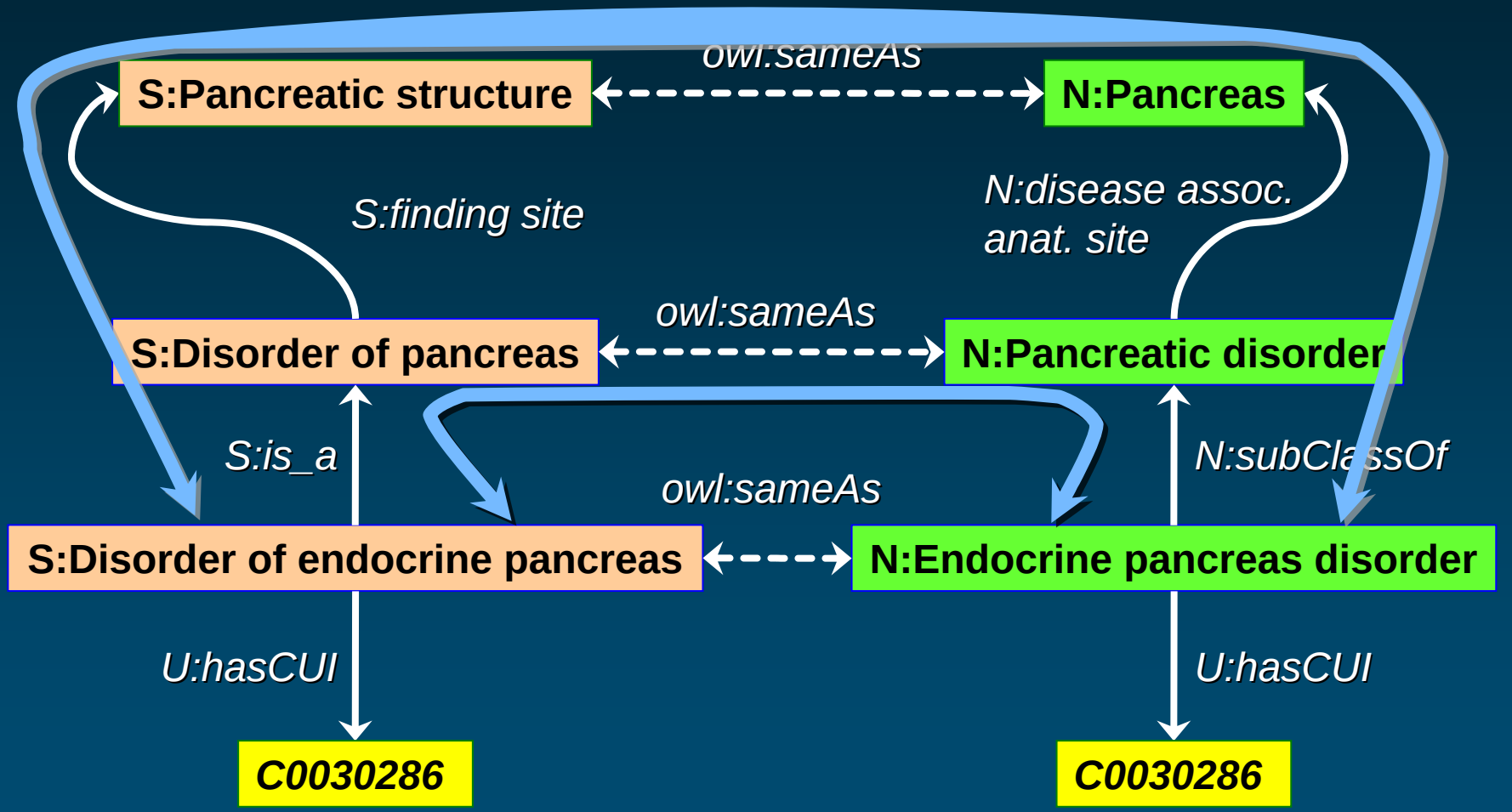
- Triple store: mulgara™ http://www.mulgara.org/
- Could not apply the rules for RDFS semantics on a large, heavily hierarchical graph
- Created one rule for the equivalence of concepts through UMLS
  - 2 concepts are equivalent if they have the same UMLS CUI



Methods RDF graph



Methods Querying the RDF graph

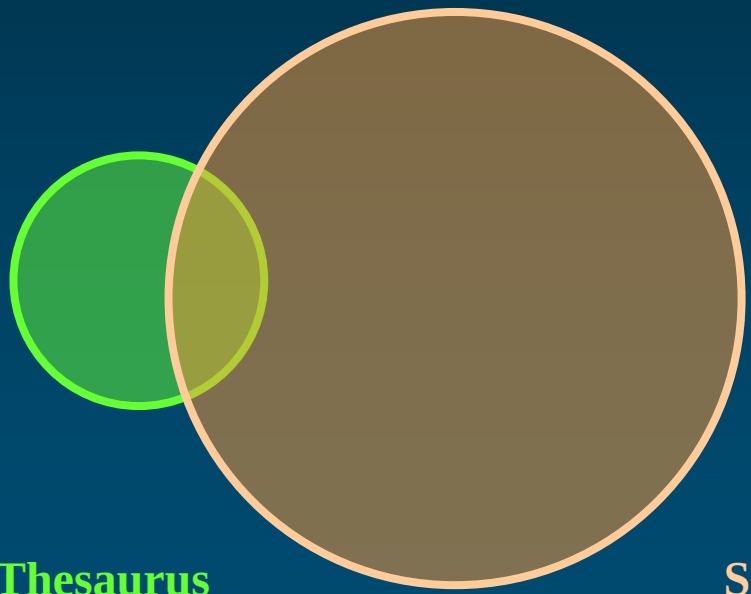


Methods Querying the RDF graph

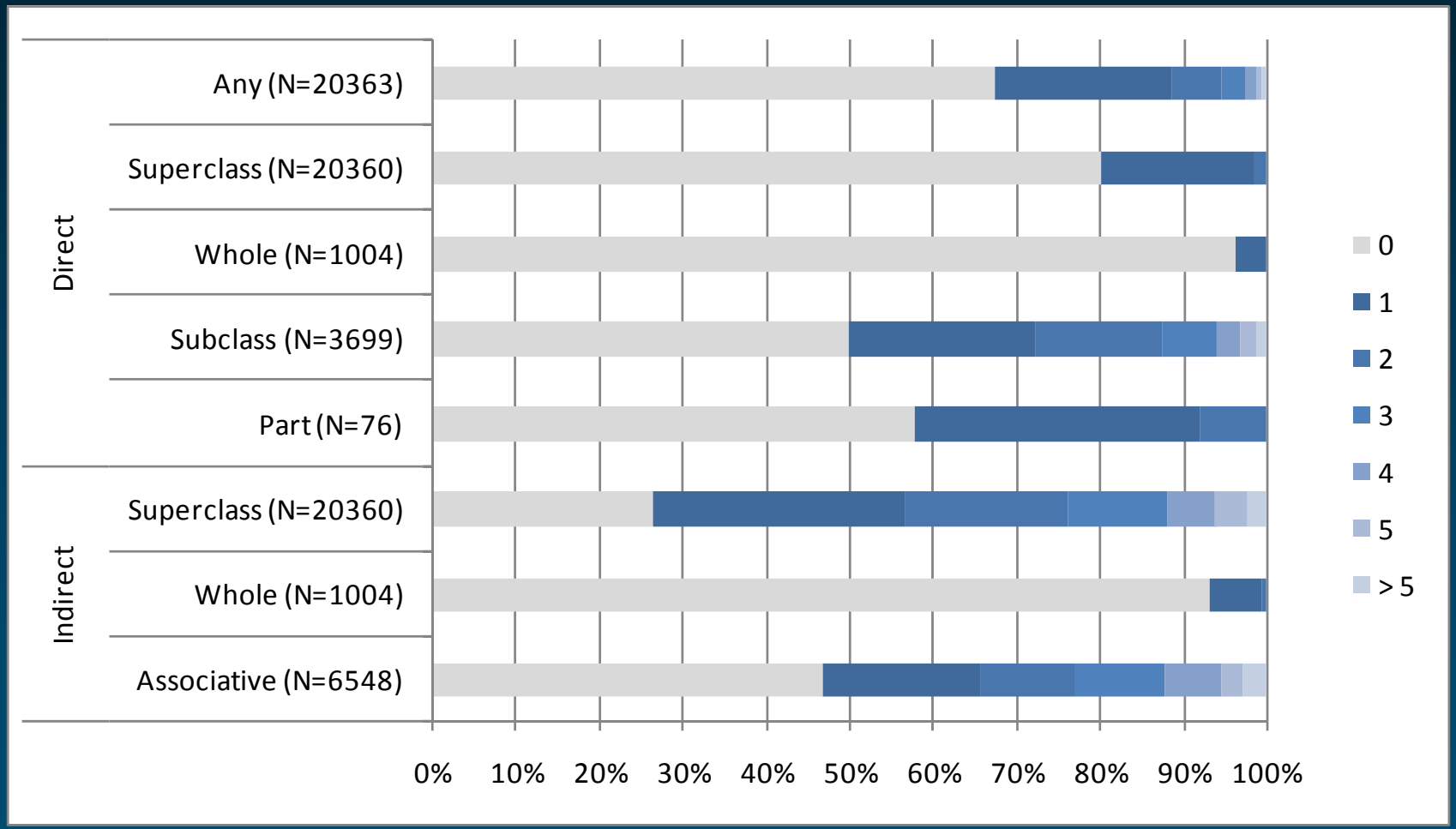
- Comparing the shared relata of concepts
  - For various kinds of relationships (isa, part of, associative relationships)
  - Directly or recursively
- ITQL queries for each pair of equivalent concepts and for each type of relationship queried
  - Functionally equivalent to SPARRL
  - “walk” function compensates for the absence of transitive closure rule

Results Overlap NCIt/SNOMED CT

- 20,369 pairs of equivalent concepts
  - 23.9% of all (active) NCIT concepts
  - 6.3% of all (active) SNOMED CT concepts



Results Shared relata (quantitative)

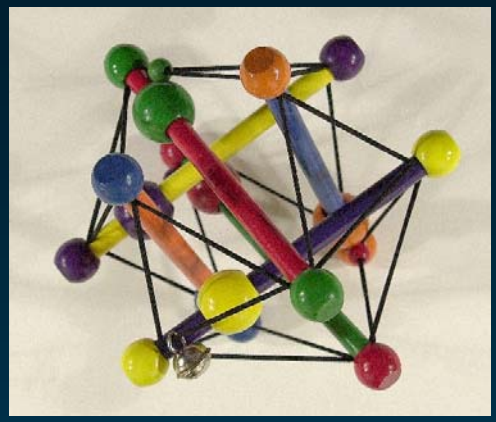


Comparing formal definitions ☹

- Relatively small proportion of relata in common between equivalent concepts from NCIt and SNOMED CT
- Large number of primitive concepts in NCIt and SNOMED CT (70-80%)
- Insufficient for effectively comparing definitions
  - Could not be used for validating the mapping provided by the UMLS

RDF approach to comparing concepts ☺

- RDF is suitable to the comparison of terminologies
  - Large terminologies
  - One is not available in OWL
- Mulgara supported the load
  - Except for applying RDFS rules
- RDF approach
  - No *ad hoc* programming necessary
  - Well adapted to recursive traversal of relationships (notoriously difficult with relational databases)



Medical Ontology Research

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