

Developing a UMLS-based Ontology of Cardiology Procedures for Cognitive Support in Medical Decision Making

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2002

Goal of the research:

- **ontology** of medical procedures in the cardiology domain
- based on knowledge and information found in **UMLS**
- increase **automation** of ontology development
- theoretical background: *KR, ontology*, **cognition**

Why UMLS?

- **content:** knowledge corpus, knowledge sources
- **comprehensiveness:** more than 60 vocabularies and classifications
- **integration:** of sources and vocabularies
- **repository:** of concepts and relations
- **computability:** data, tables, links, applications (*browsers, NLP, etc*)

Cognition

- Principles
- paradigm shift
- present paradigm
- levels of cognitive processes

Cognition principles:

- Modularity of mental processes
 - structural
 - functional
 - evolutionary
- Parallel distributed multi-level processing
 - autonomy, parallelisms
 - distribution, multi-functionalism
 - multi-level, shifts (*level, direction*), integrations

Paradigm shift:

- setting:
 - laboratory => naturalistic medical
- object:
 - individual => social and technological settings

Expertise-novice paradigm:

- **levels** of training, expertise, competency
- **3** basic levels of competency
- **consistent** results in different professional fields

Parallel and interconnected levels of cognition

- **Factual:** *identification, recognition, discrimination, etc.*
- **Semantic:** *meanings, causes, consequences, etc.*
- **Schematic:** *classification, variables, composition, etc.*
- **Strategic:** *plan, action order, values, options, etc.*

Knowledge-base

“Knowledge-based systems emphasize **meaning**. Instead of processing data as a string of bits, they **represent** the meaning of data in terms of the **real world**. They carry on **conversations** with people in ordinary language, they find important **facts** before they are requested, and they solve complex problems at expert **levels** of performance. ...

Two fields devoted to knowledge-based systems are cognitive science and artificial intelligence. **Cognitive science** is a merger of philosophy, linguistics, and psychology with a strong influence from computer science. Artificial intelligence (AI) is the engineering counterpart.”

John Sowa

Knowledge Representation

- **Logic** provides the formal structure and rules of inference.
- **Ontology** defines the kinds of things that exist in the application domain.
- **Computation** supports the applications that distinguish knowledge representation from pure philosophy.

Ontology

- What:
 - formal conceptualization of a particular universe of knowledge about the world
- How:
 - explicit representation of concepts, relations between them, and inference rules about them and the things they represent

Domain ontologies

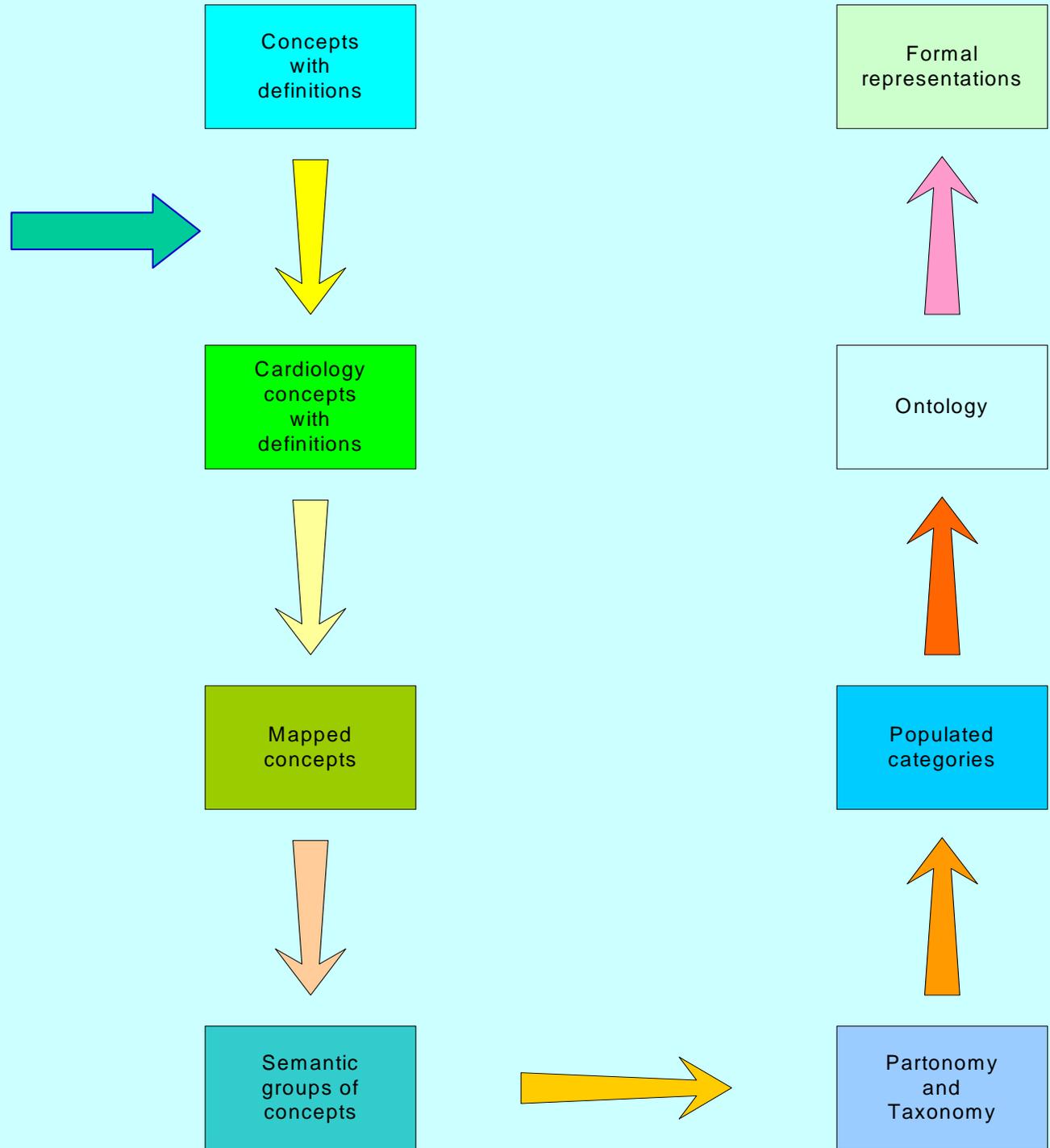
can be used to provide:

- **knowledge support** in aspects of underlying cognitive processes and their inter-relations
- methodology for **connecting** different **databases** through the use of common knowledge structures
- efficient means for facilitating **professional communication** about specific domain subjects and tasks

Medical domain ontology field

- Previous researches:
 - Knowledge:
 - elicitation from experts
 - Development:
 - from scratch
 - Foundations:
 - domain, KR
 - Purposes:
 - data collection, database connection, knowledge support, professional communication
- Present research:
 - Knowledge:
 - acquisition from corpus
 - Development:
 - to increase its automation
 - Foundations:
 - domain, KR, and cognition
 - Purposes:
 - same, with emphasis in cognitive properties of medical decision making and medical education processes

*Diagram
of the
methodology
phases:*

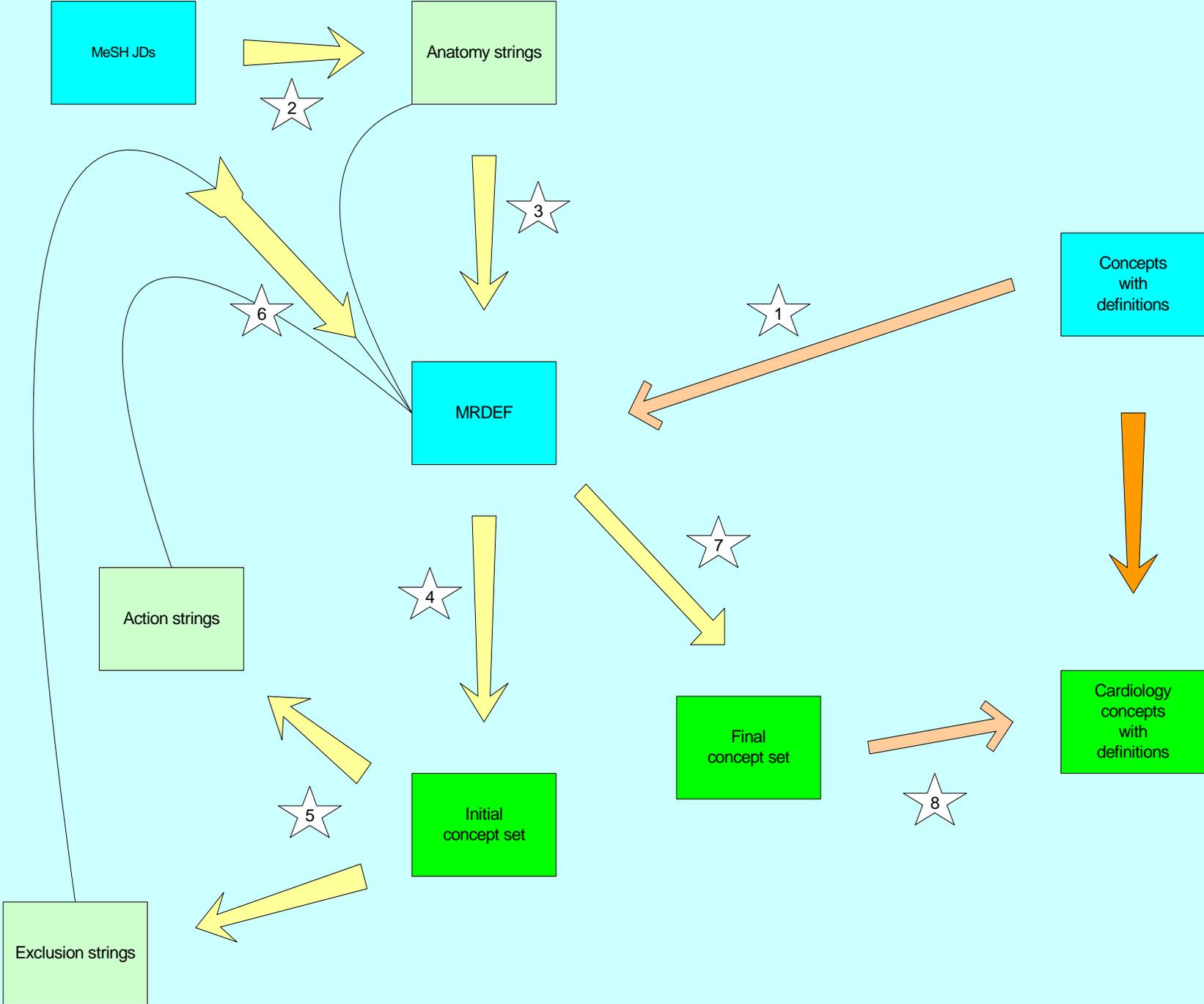


Selecting concepts with definitions:

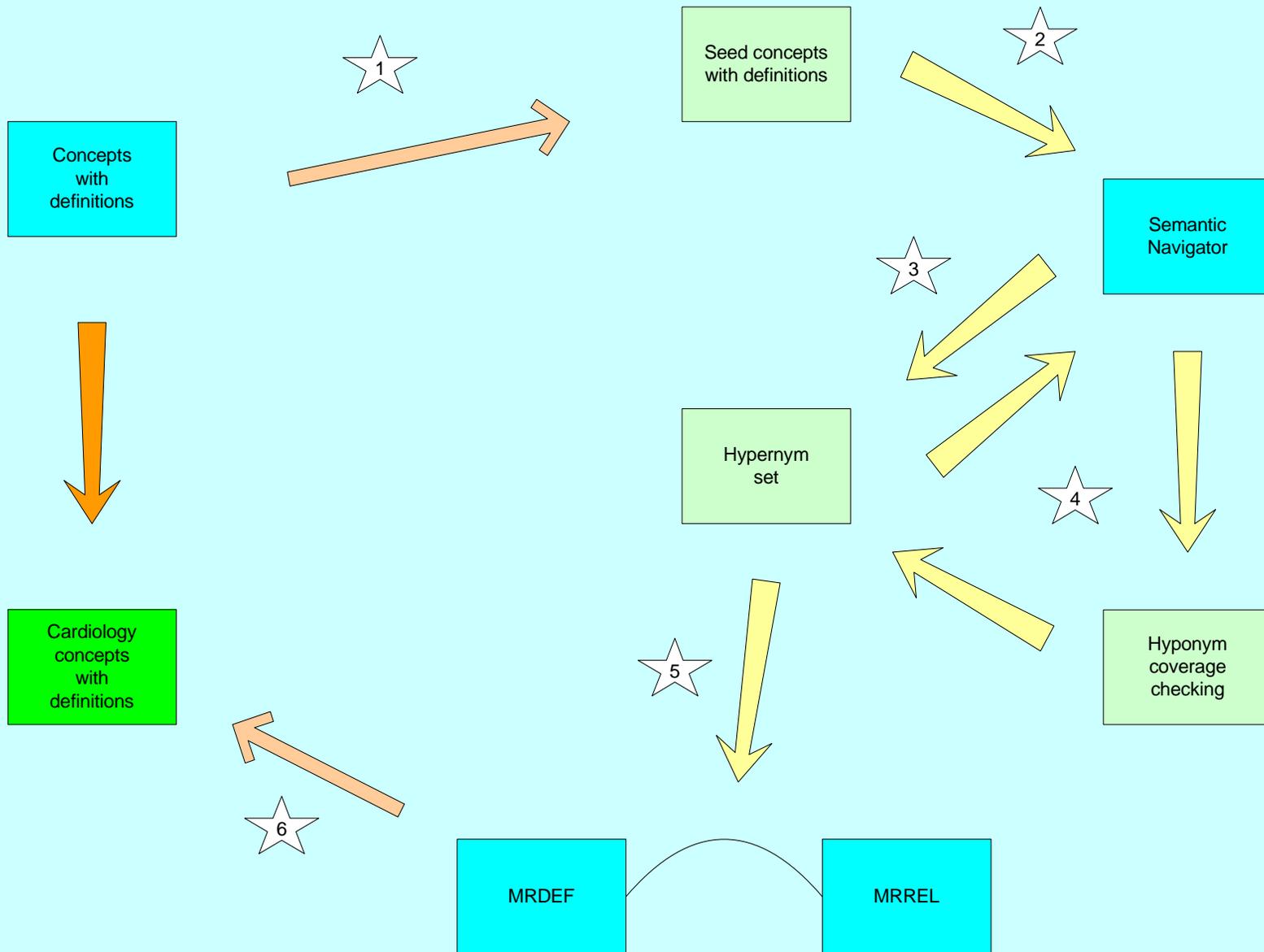
- From domain words to domain concepts:
 - lexical-based method

- From seed concepts to domain concepts:
 - hierarchical-based method

Lexical-based method:



Hierarchical-based method:



Metathesaurus: - 2001 edition -

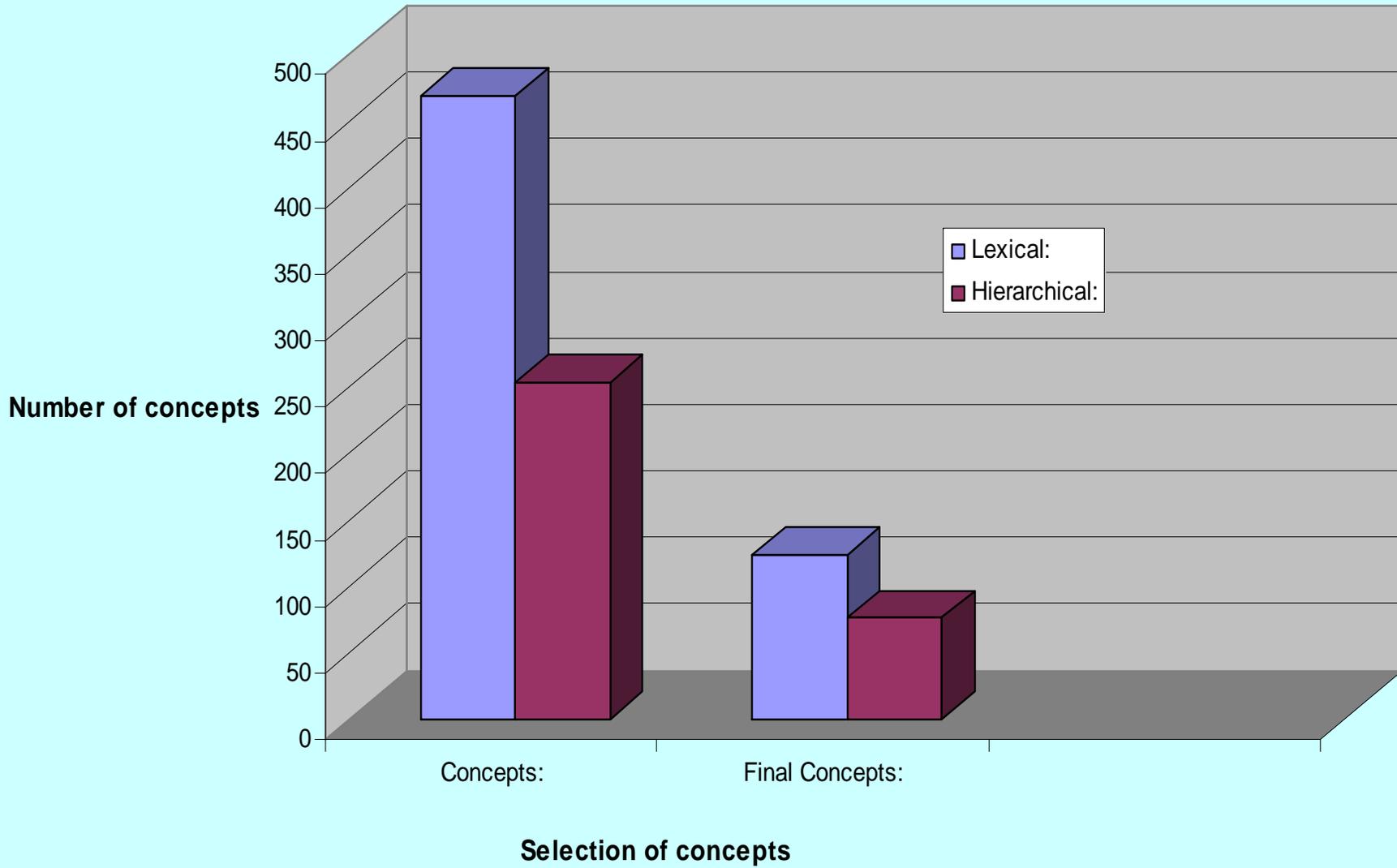
- about 800,000 concepts
- about 1.9 million concept names
- concepts with definitions = 34,095
- = 4.26 % of Metathesaurus concepts

Comparison of sets:

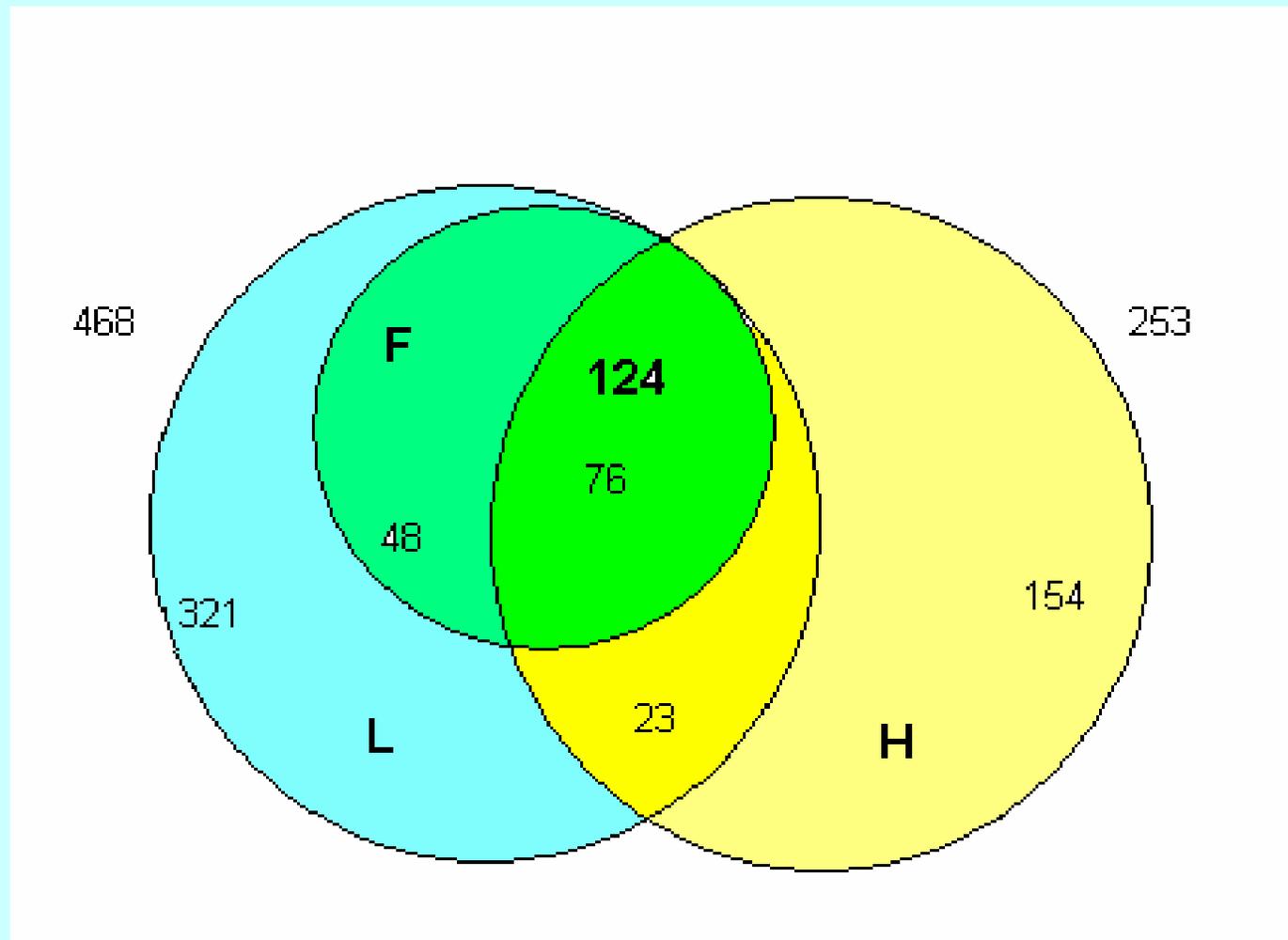
(lexical-based and hierarchical-based)

	Concepts:	Concepts/MRDEF:	Final Concepts:	Final Concepts / MRDEF (Final Concepts / UMLS):
Lexical:	468	1.37 %	124	0.36 % (0.000155 %)
Hierarchical:	253	0.74 %	76	

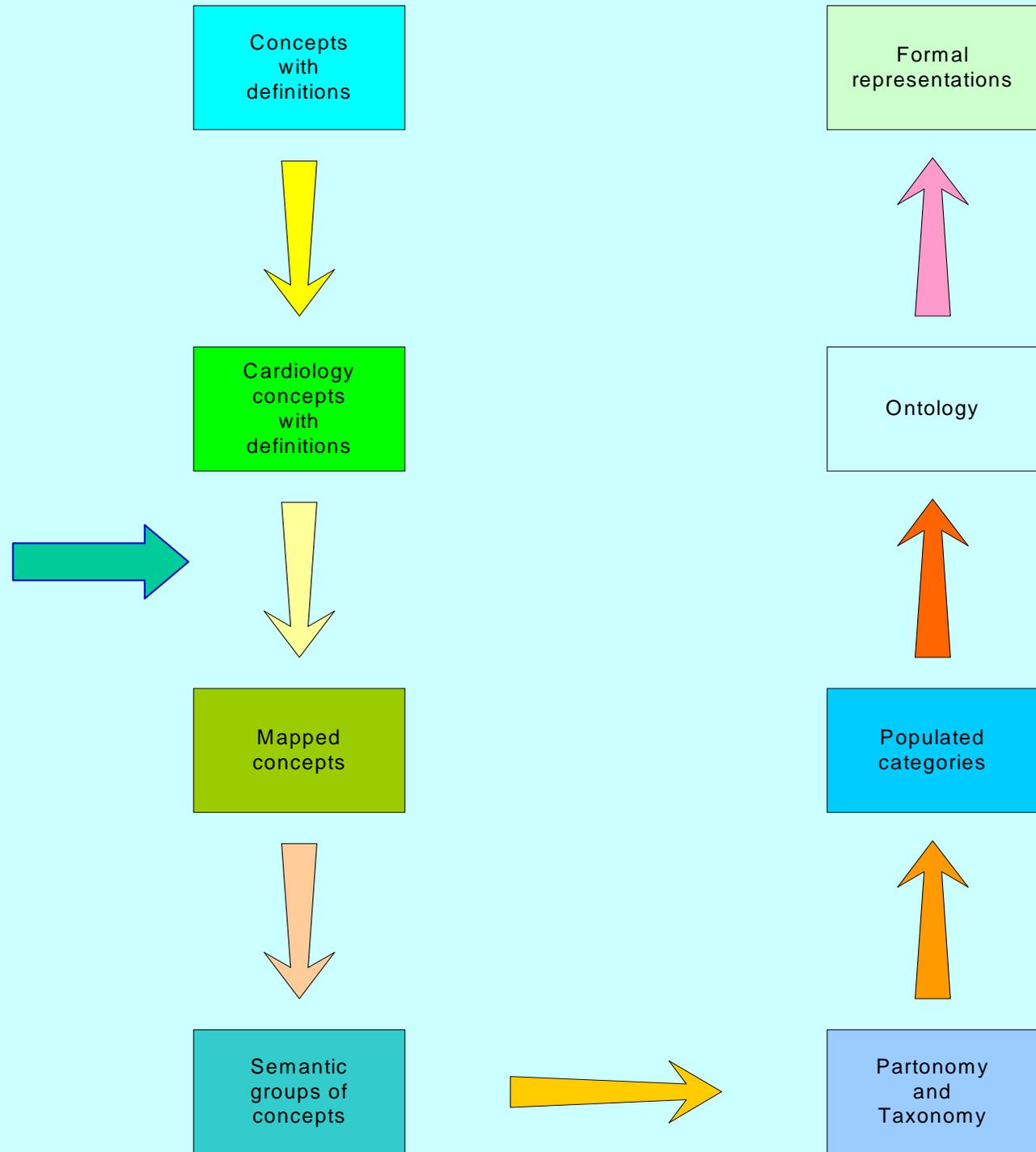
Comparison of sets



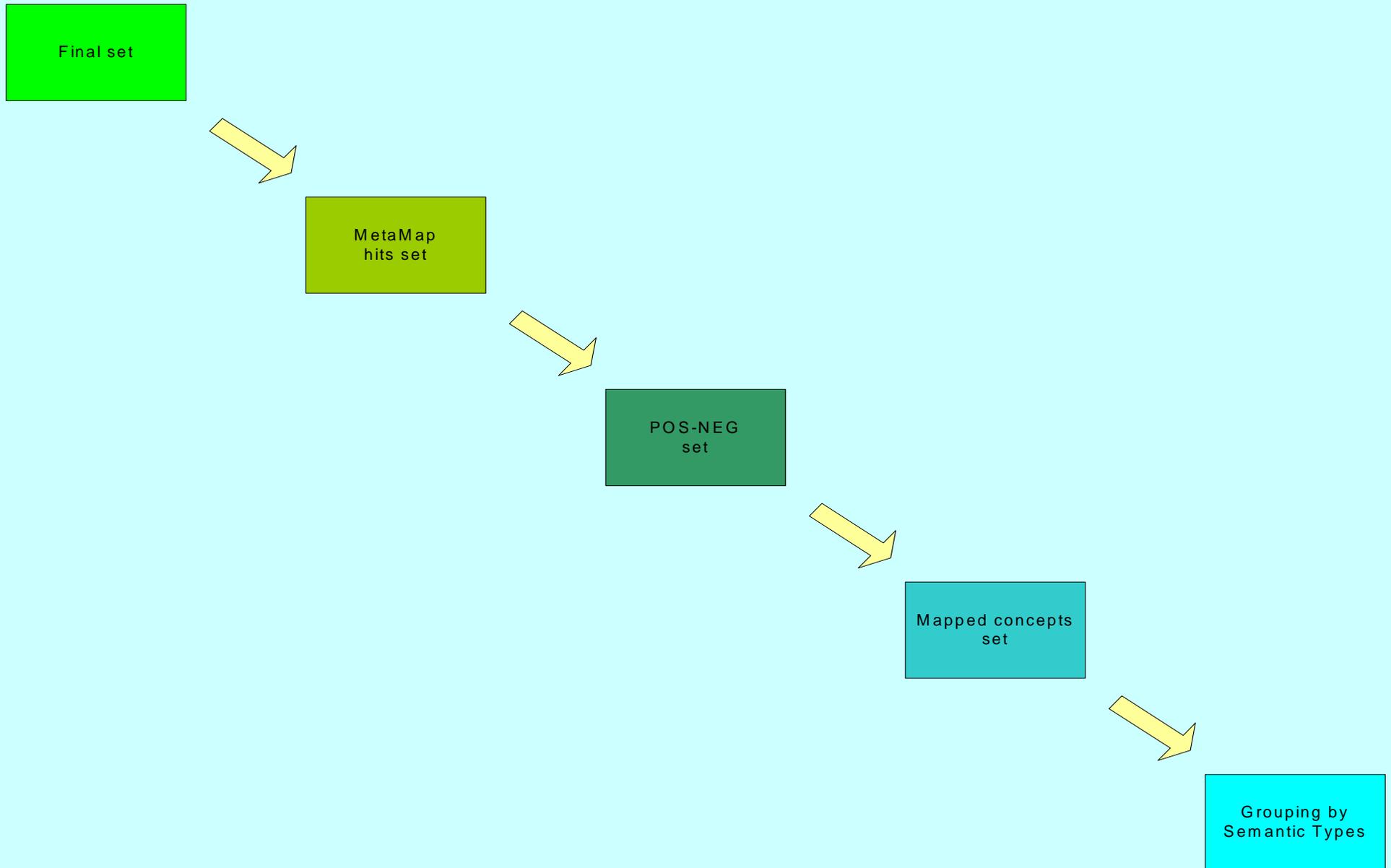
Comparison of sub-sets of lexical-based and hierarchical-based methods:



*Diagram
of the
methodology
phases:*



From domain concepts to domain semantics:



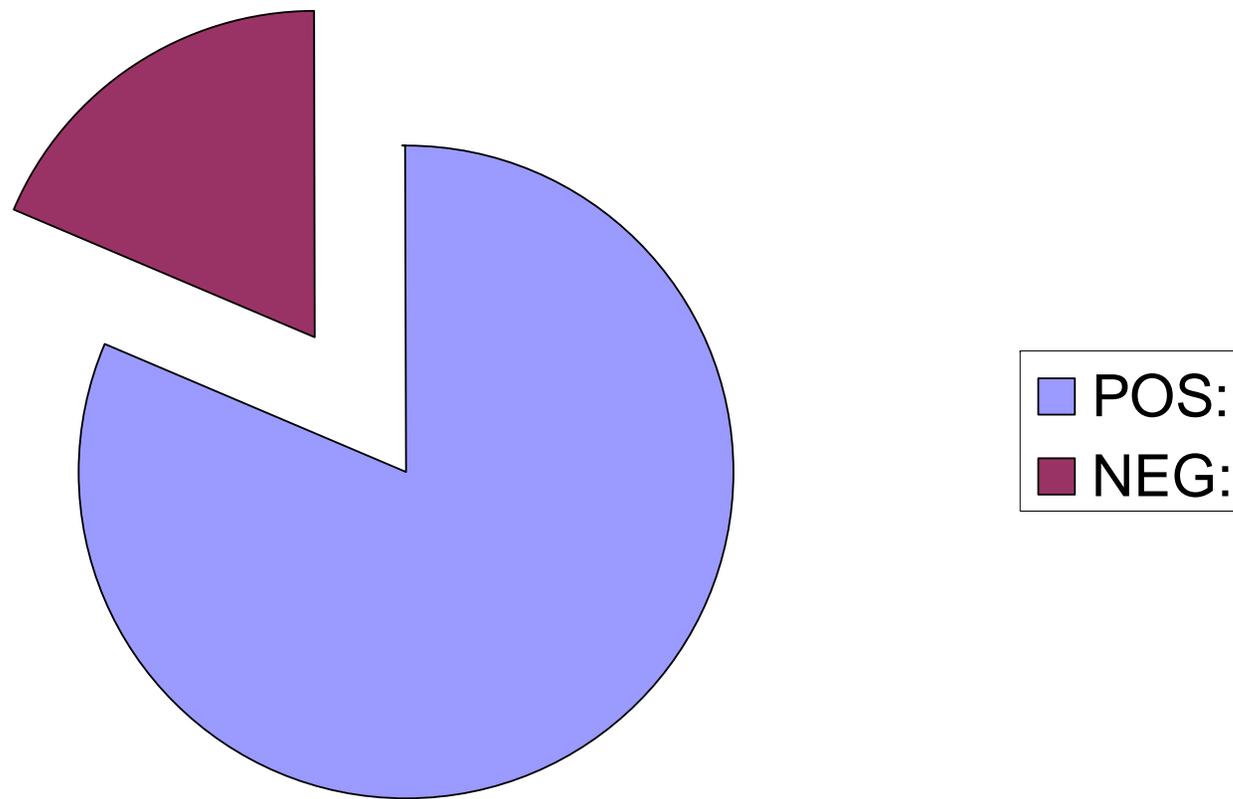
METAMAP HITS:

Total:	POS:	NEG:
2152	1835	317
	85.27 %	14.73 %

MAPPED CONCEPTS:

Total:	POS:	NEG:
987	804	183
	81.46 %	18.54 %

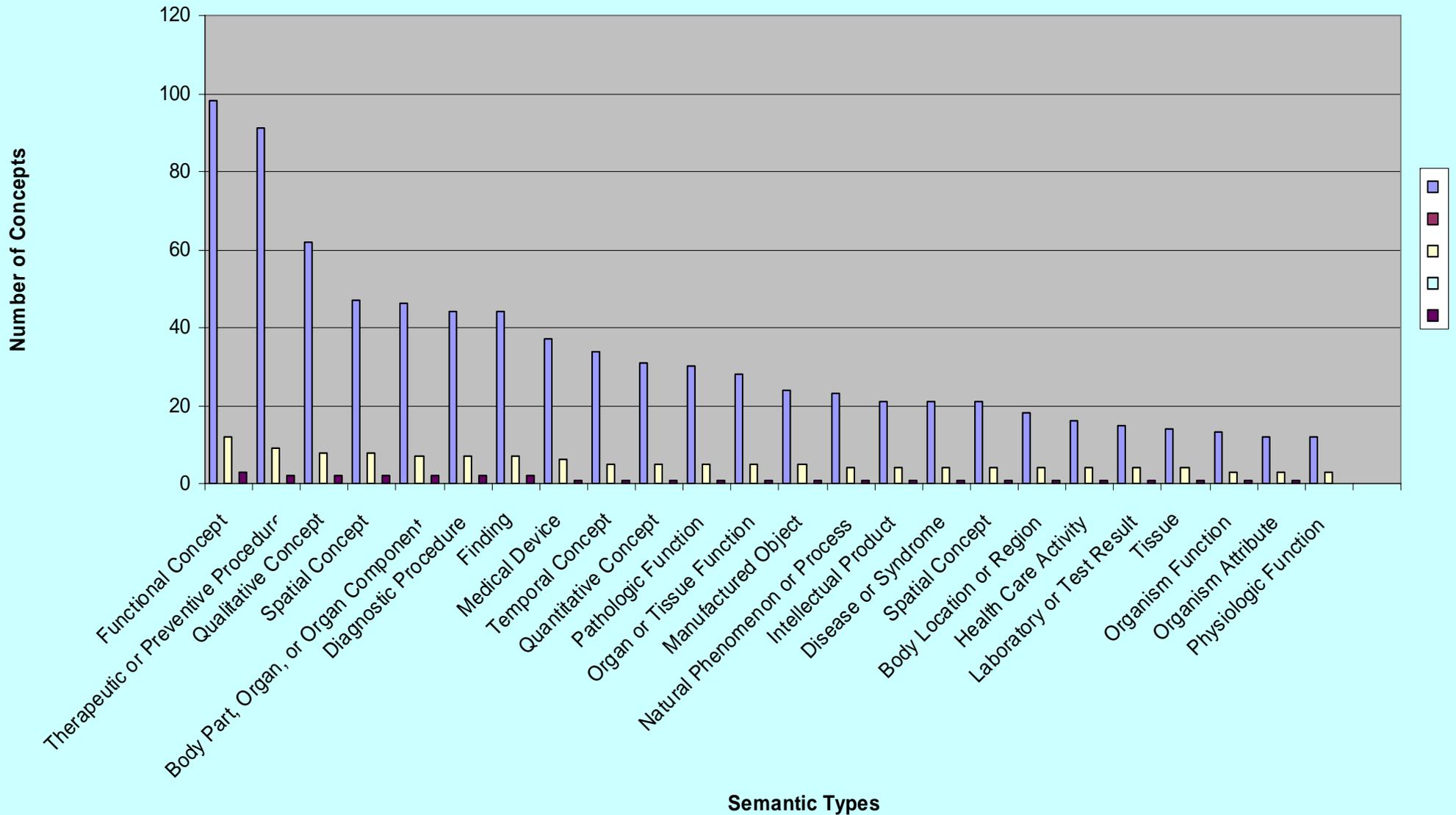
MAPPED CONCEPTS:



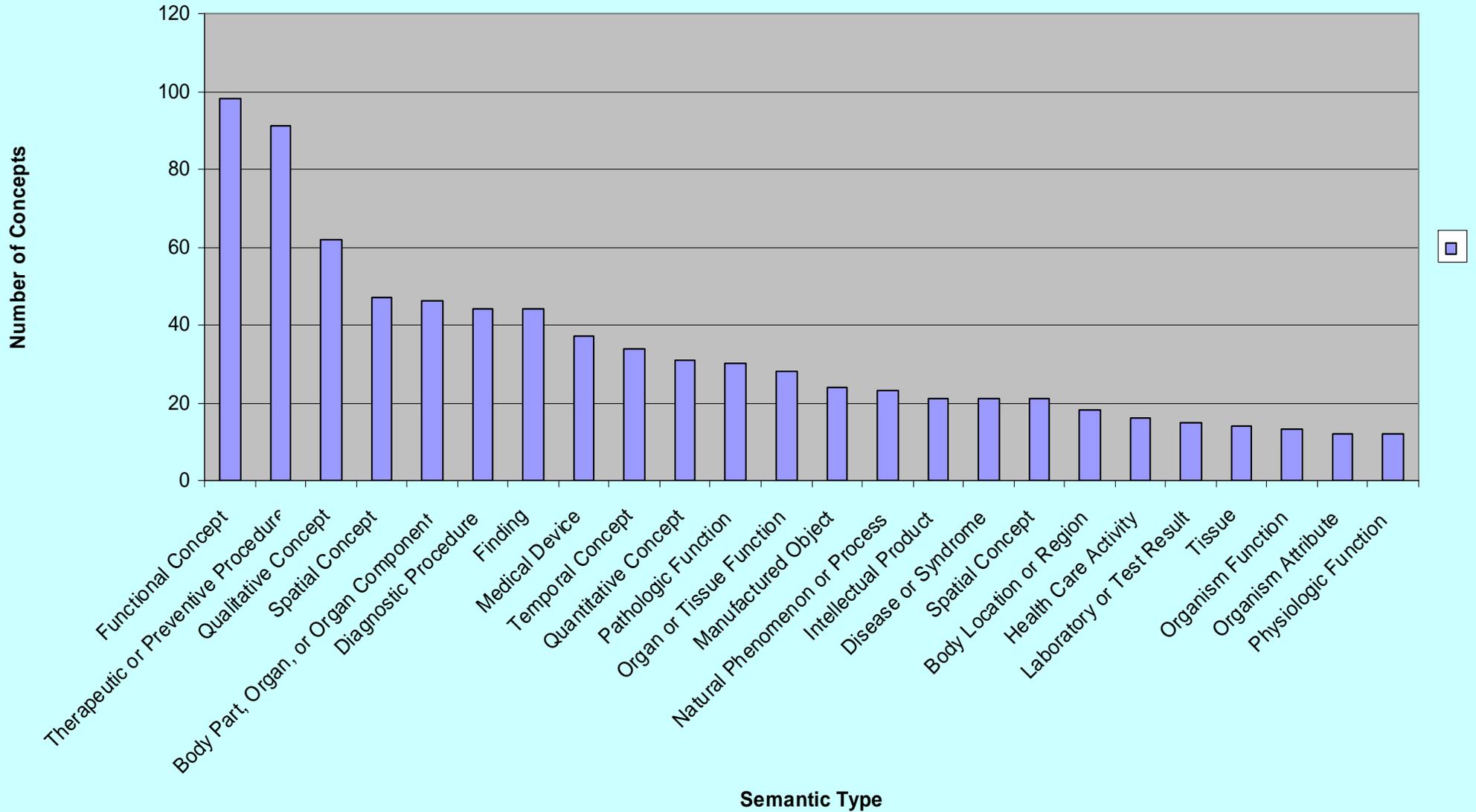
Number of Concepts by Semantic Type:

Functional Concept	98	Injury or Poisoning	1 2	Educational Activity	3
Therapeutic or Preventive Procedure	91	Sign or Symptom	9	Body Space or Junction	2
Qualitative Concept	62	Research Activity	8	Cell	2
Spatial Concept	47	Substance	8	Cell Function	2
Body Part, Organ, or Organ Component	46	Body Substance	7	Human	2
Diagnostic Procedure	44	Pharmacologic Substance	7	Population Group	2
Finding	44	Mental Process	7	Neoplastic Process	2
Medical Device	37	Laboratory Procedure	6	Classification	1
Temporal Concept	34	Congenital Abnormality	5	Group	1
Quantitative Concept	31	Acquired Abnormality	5	Environmental Effect of Humans	1
Pathologic Function	30	Occupational Activity	5	Group Attribute	1
Organ or Tissue Function	28	Phenomenon or Process	5	Conceptual Entity	1
Manufactured Object	24	Biomedical or Dental Material	5	Idea or Concept	1
Natural Phenomenon or Process	23	Inorganic Chemical	4	Biologically Active Substance	1
Intellectual Product	21	Chemical Viewed Structurally	4	Lipid	1
Disease or Syndrome	21	Idea or Concept	4	Clinical Attribute	1
Spatial Concept	21	Anatomical Structure	4	Patient or Disabled Group	1
Body Location or Region	18	Body System	4	Group	1
Health Care Activity	16	Indicator, Reagent, or Diagnostic Aid	4	Biologic Function	1
Laboratory or Test Result	15	Daily or Recreational Activity	4	Chemical	1
Tissue	14	Organic Chemical	4	Embryonic Structure	1
Organism Function	13	Anatomical Abnormality	3	Individual Behavior	1
Organism Attribute	12	Animal	3	Age Group	1
Physiologic Function	12	Element, Ion, or Isotope	3		

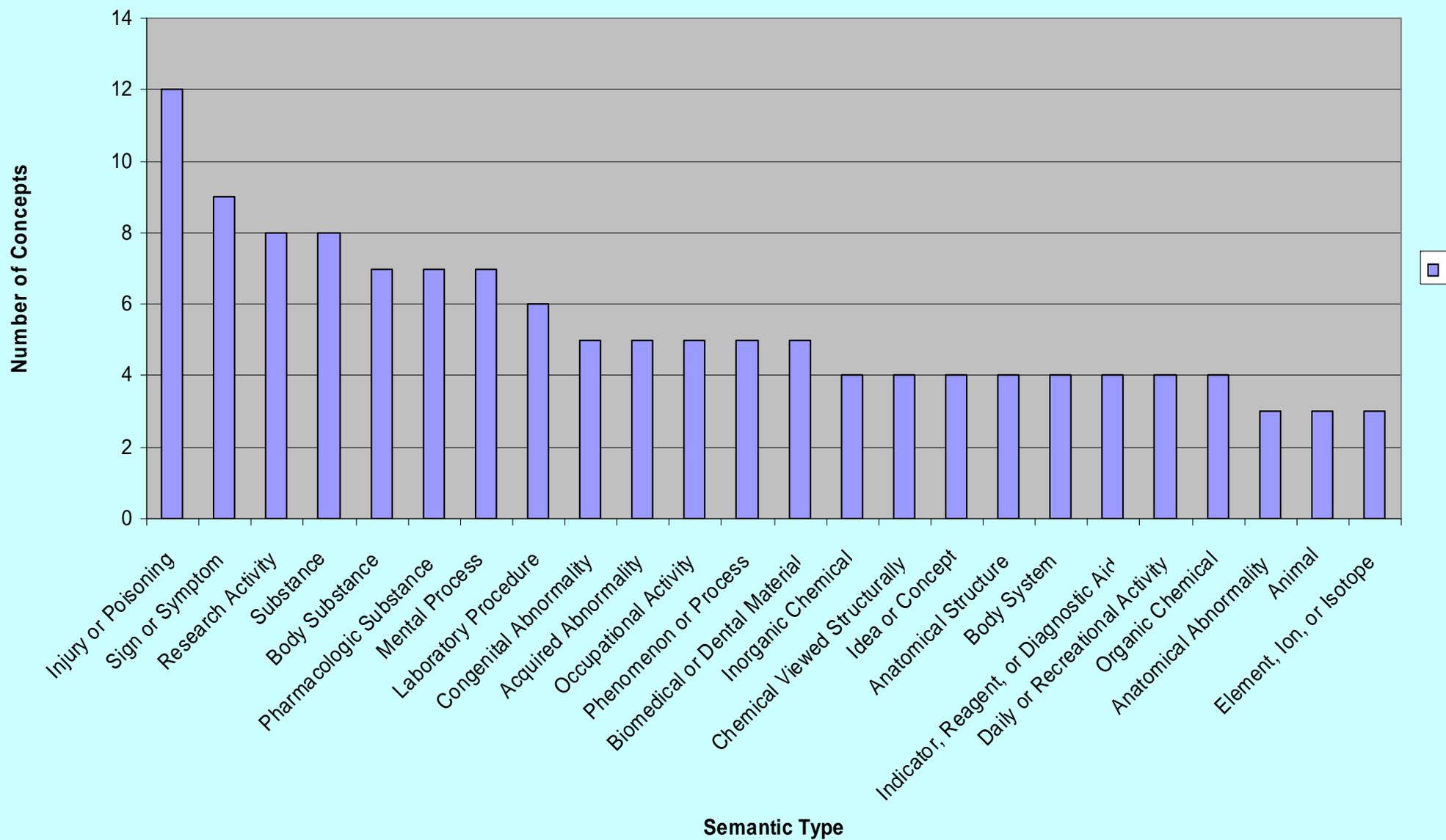
Number of concepts by Semantic Type:



First third:



Second third:



*Diagram
of the
methodology
phases:*

Concepts
with
definitions



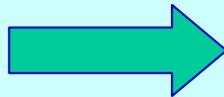
Cardiology
concepts
with
definitions



Mapped
concepts



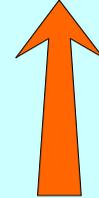
Semantic
groups of
concepts



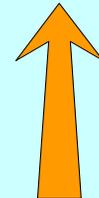
Formal
representations



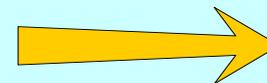
Ontology



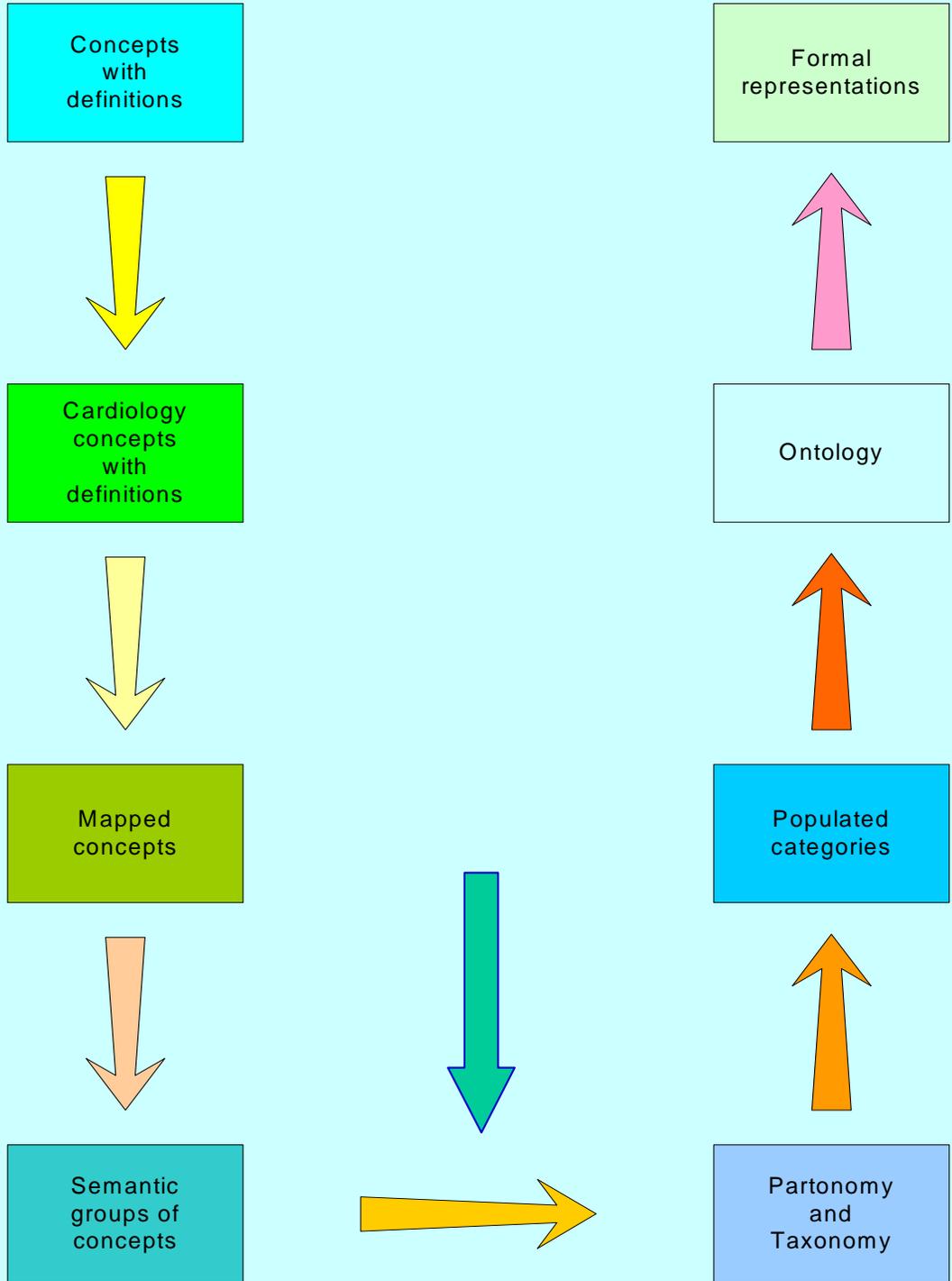
Populated
categories



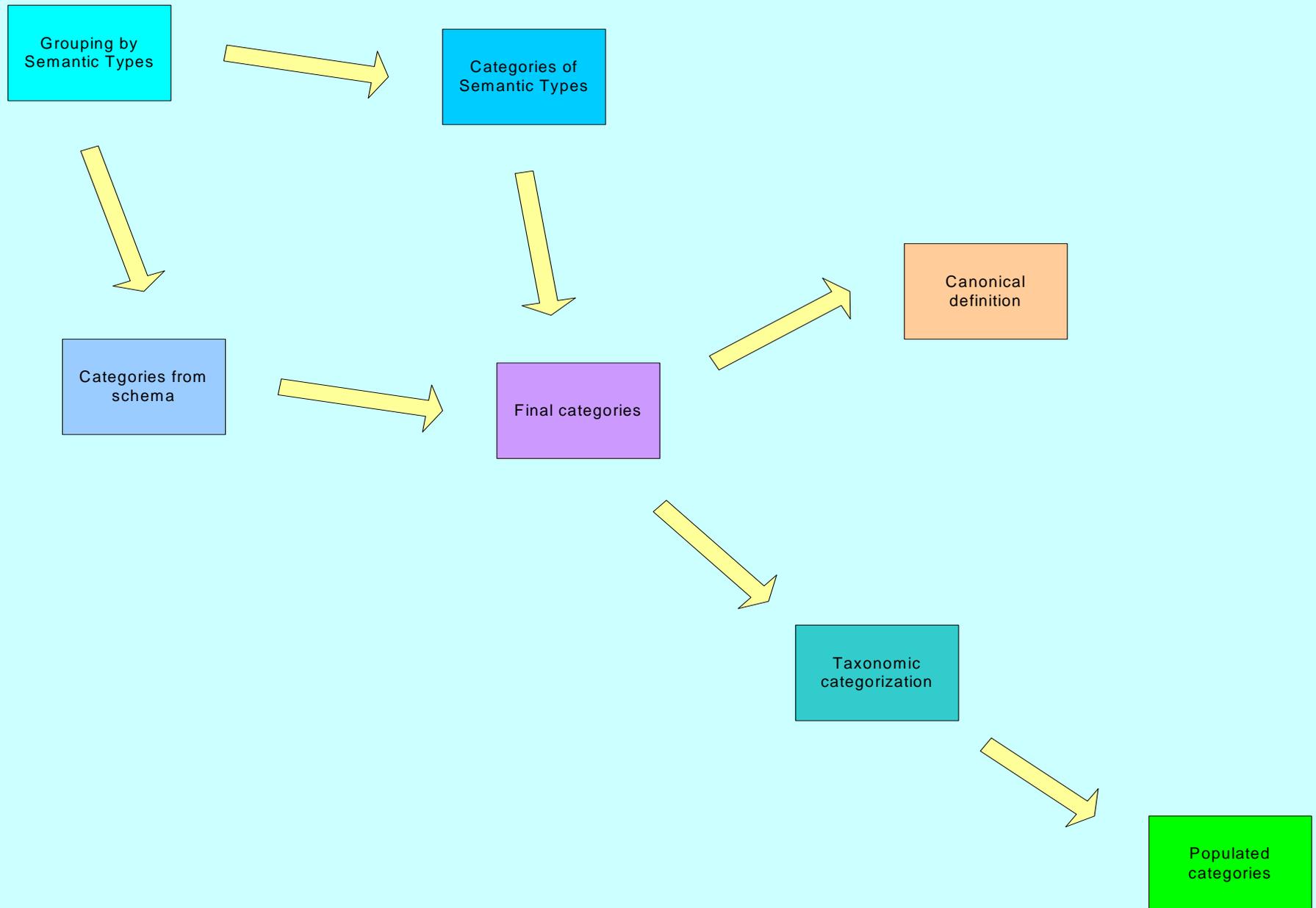
Partonomy
and
Taxonomy



*Diagram
of the
methodology
phases:*



From domain semantics to domain ontology:



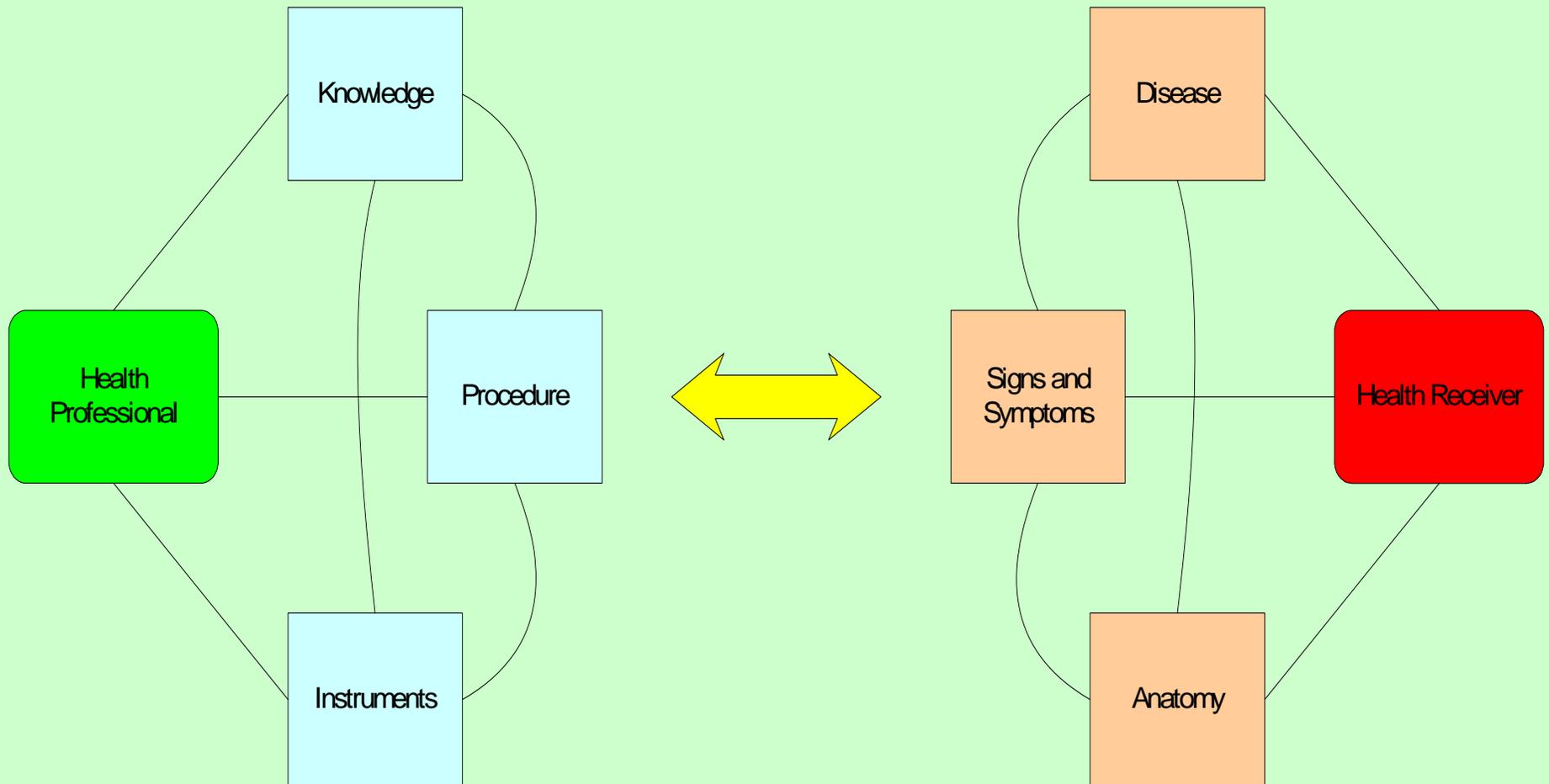
From domain semantics to domain ontology: (*approaches*)

- Bottom-up:
 - comparison of Semantic Types
 - semantic comparison of concepts
 - grouping of Semantic Types in categories

From domain semantics to domain ontology: (*approaches*)

- Top-down:
 - basic schema of agents and processes of medical act
 - comparison of schema with Semantic Types and concepts
 - other comparisons

Agents and Processes of a Medical Act



Comparisons of categorization:

- Original information model
- Semantic Groups of Semantic Types
- MAOUSSC

Categories of the Ontology:

<u>Nuclear:</u>	<u>Auxiliary:</u>
Action	Conditional Action
Anatomical Entity	Method
Pathology	Phenomenon
Instrument	Measure
Material or Energy	Result or Product
Physiology	Time or Circumstance
Purpose	Space
-	Receiver

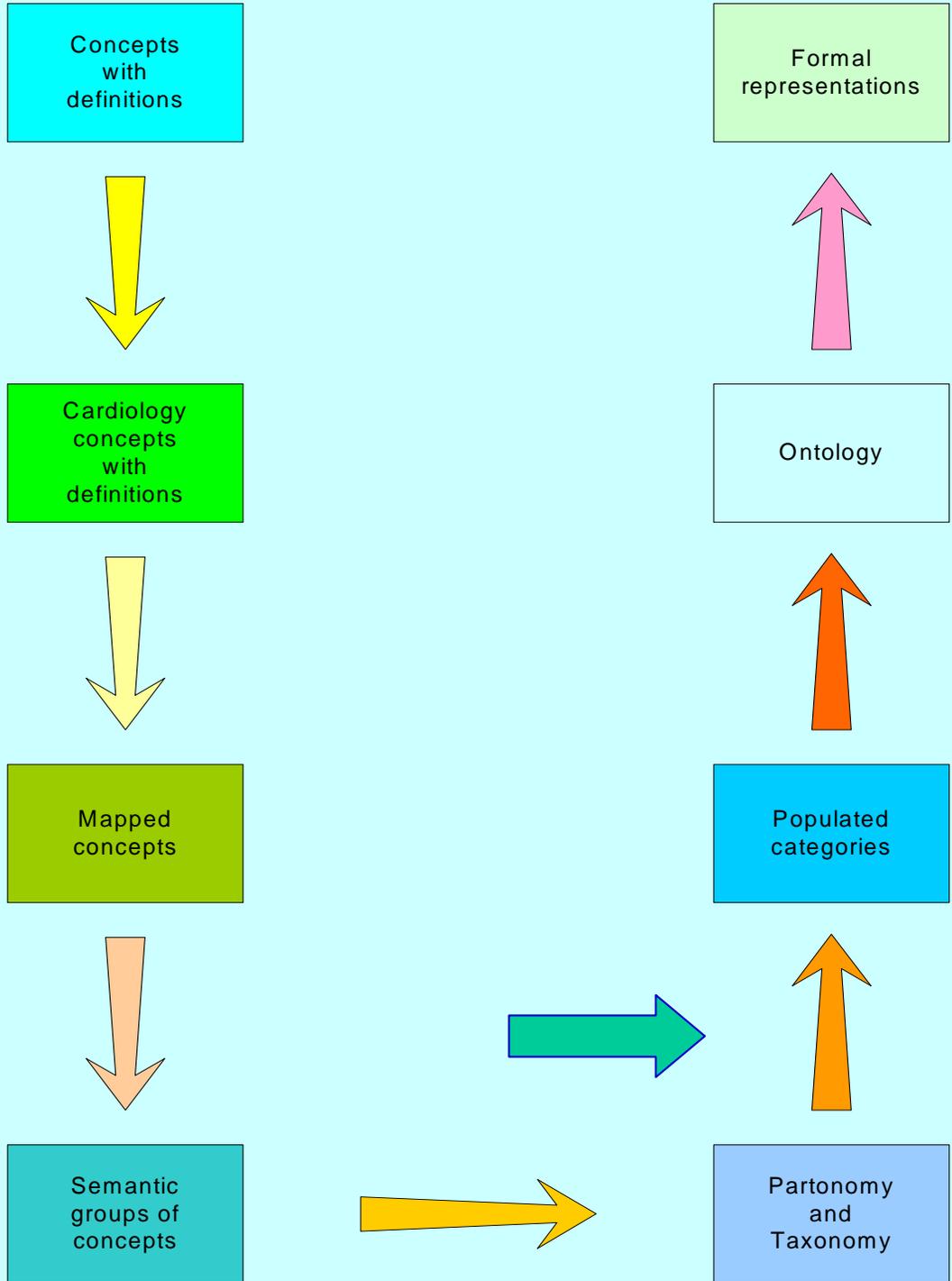
Canonical formal definition of a cardiology procedure concept definition:

“A cardiology procedure concept definition is a conceptual entity, which is composed by an **action**, which: has location in an **anatomical entity**, and affects a **pathology**, and affects the **physiology**, and has a **purpose**, and uses an **instrument**, and uses some **material or energy**, and can be preceded by a **conditional action**, and has a **method**, and uses or affects a **phenomenon**, and uses a **measure**, and has a **result or product**, and occurs in a certain circumstance in reference to **time**, and has some **space** references, and affects a **receiver**.”

From paronomy to taxonomy:

- Bottom-up approach
 - levels of specification-generalization
 - represent a compositional paronomy
 - parallelisms in structure for some sub-categories:
 - external and internal consistency
 - restrictions in relationships between sub-classes

*Diagram
of the
methodology
phases:*



Populating the categories:

- Human X Automatic processes
- comparison between both methods
- potential generalizability

Human and Automatic populating methods:

<div style="text-align: right;">Author</div> <div style="text-align: left;">Automatic</div>	POS	NEG	
POS	797	172	969
	80.75 %	17.43 %	98.18 %
NEG	7	11	18
	0.71 %	1.11 %	1.82 %
	804	183	987
	81.46 %	18.54 %	

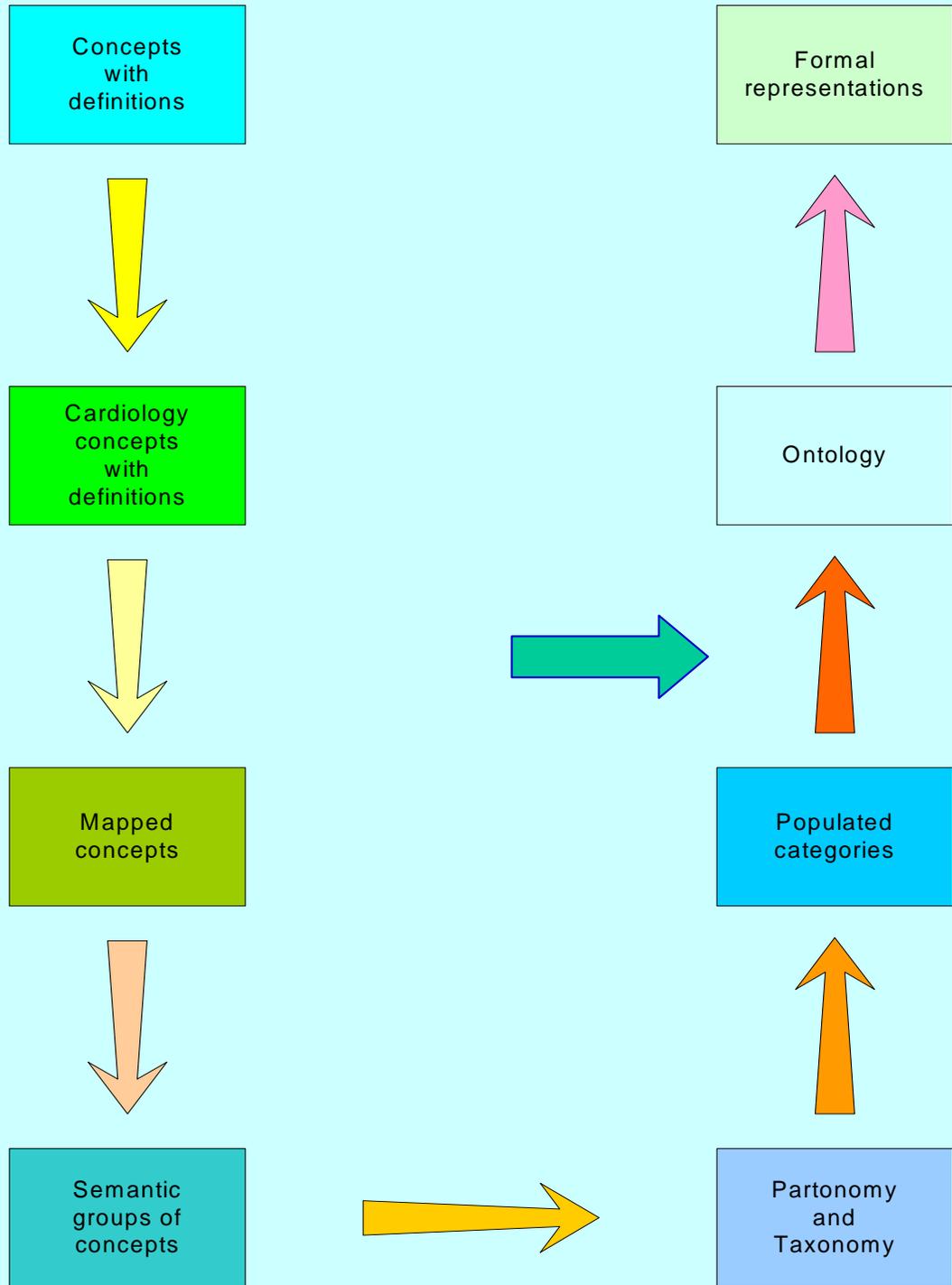
Number of Semantic Types for
kinds of result of populating methods:

<u>Type of Result:</u>	H(+)-A(+)	H(+)-A(-)	H(-)-A(+)	H(-)-A(-)
<u># Semantic Types:</u>	64	44	4	4

Number of equivalent and different
Semantic Types for both methods:

\times	=	H(+)-A(+)	H(+)-A(-)	H(-)-A(+)	H(-)-A(-)
H(+)-A(+)			0	43	0
H(+)-A(-)		4		0	1
H(-)-A(+)		1	4		0
H(-)-A(-)		4	3	4	

*Diagram
of the
methodology
phases:*





Superclasses

Name	Type	Cardinality	Other Facets
------	------	-------------	--------------

:THING

Name	Documentation	Constraints
:THING		

Role

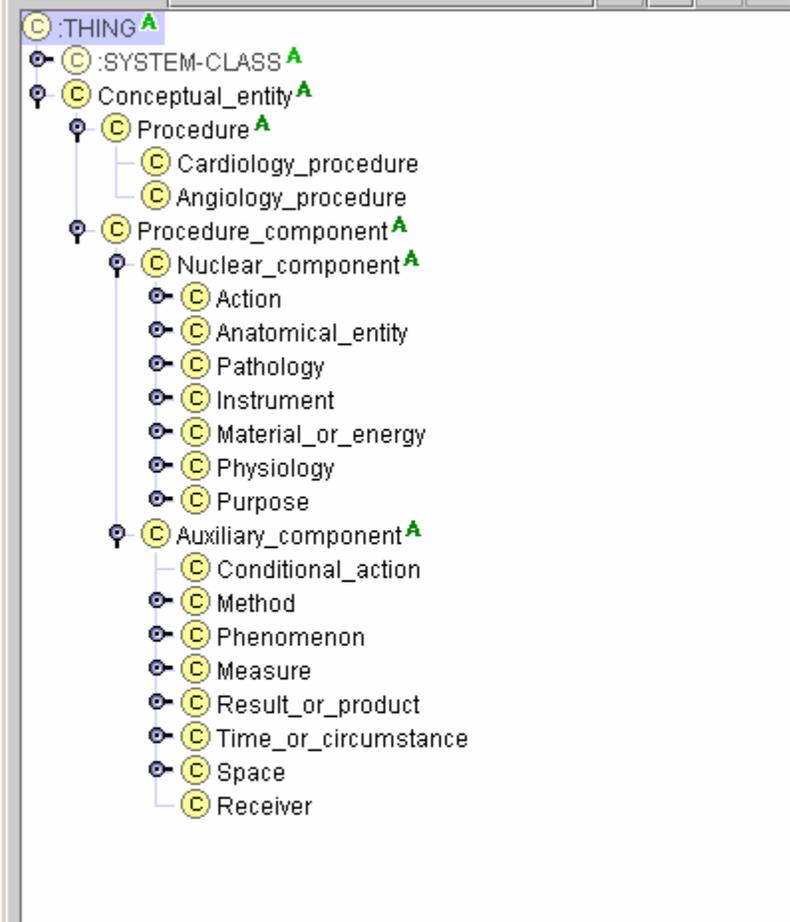
Abstract A

Template Slots

Name	Type	Cardinality	Other Facets
------	------	-------------	--------------



Relationship Superclass



Superclasses

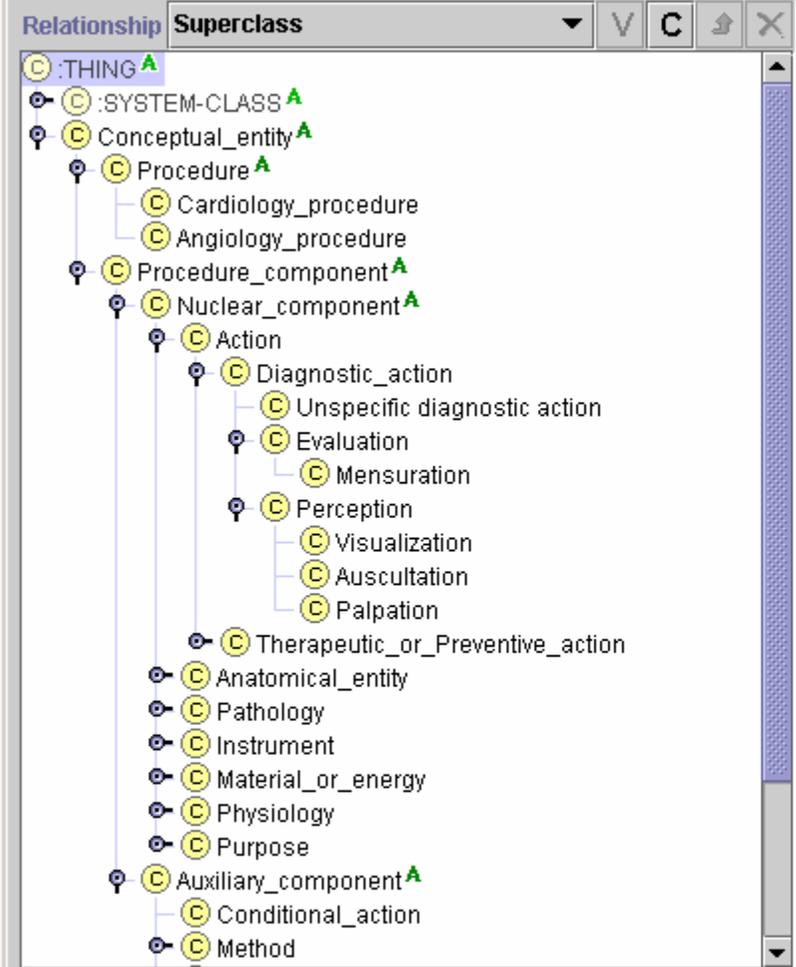
	+	-
--	---	---

:THING

Name	Documentation	Constraints
:THING		
Role		
Abstract		

Template Slots

Name	Type	Cardinality	Other Facets



Superclasses

Superclass

:THING

Name	Documentation	Constraints
:THING		

Role

Abstract A

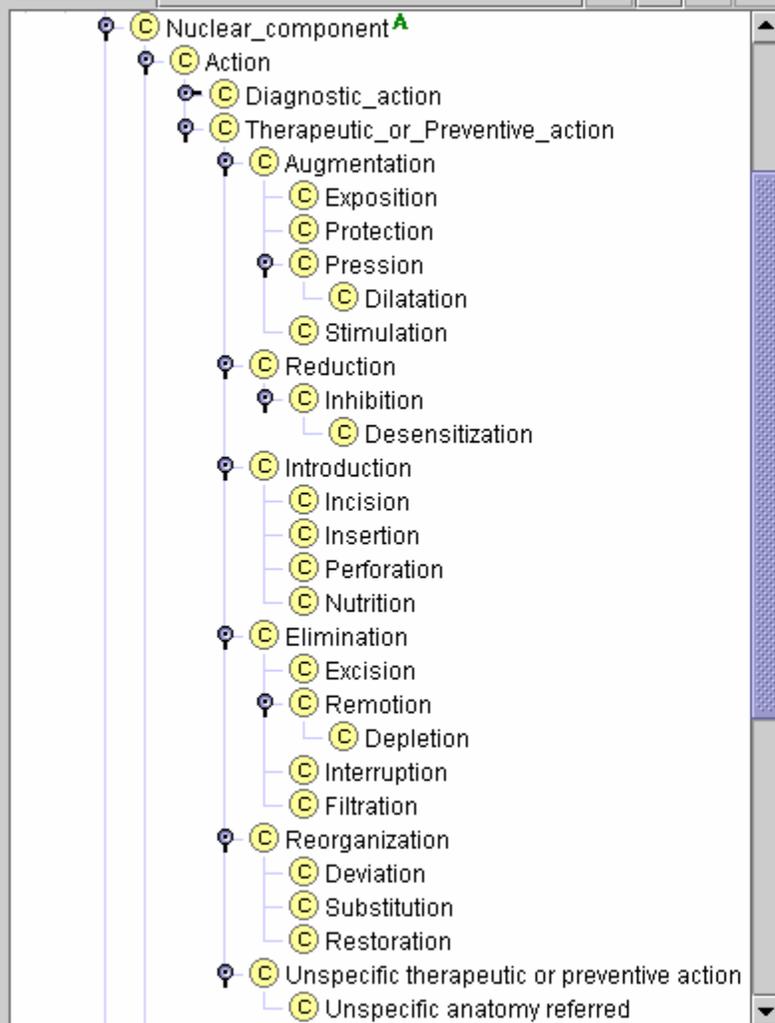
Template Slots

Name	Type	Cardinality	Other Facets



Classes Slots Forms Instances Queries

Relationship Superclass



Superclasses

:THING

Name	Documentation	Constraints
:THING		
Role		
Abstract		

Template Slots

Name	Type	Cardinality	Other Facets
------	------	-------------	--------------



Relationship Superclass

- Nuclear_component^A
 - Action
 - Diagnostic_action
 - Therapeutic_or_Preventive_action
 - Anatomical_entity
 - Pathology
 - Instrument
 - Accoustic_instrument
 - Aerodynamic_instrument
 - Mechanical_instrument
 - Thermal_instrument
 - Chemical_instrument
 - Electrical_instrument
 - Light_or_laser_instrument
 - Magnetic_instrument
 - Radiological_instrument
 - Unspecific_instrument
 - Hydrodynamic_instrument
 - Material_or_energy
 - Physiology
 - Aerodynamic_process
 - Hydrodynamic_process
 - Mechanical_process
 - Chemical_process
 - Electrical_process
 - Unspecific_physiology
 - Thermal_process

Superclasses

+ -

○ :THING

Name	Documentation	Constraints
:THING		

Role

Abstract^A

Template Slots

Name	Type	Cardinality	Other Facets



Relationship Superclass

- Nuclear_component^A
 - Action
 - Anatomical_entity
 - Pathology
 - Instrument
 - Accoustic_instrument
 - Aerodynamic_instrument
 - Mechanical_instrument
 - Thermal_instrument
 - Chemical_instrument
 - Electrical_instrument
 - Light_or_laser_instrument
 - Magnetic_instrument
 - Radiological_instrument
 - Unspecific_instrument
 - Hydrodynamic_instrument
 - Material_or_energy
 - Physiology
 - Purpose
- Auxiliary_component^A
 - Conditional_action
 - Method
 - Phenomenon
 - Mechanical_phenomenon
 - Accoustic_phenomenon
 - Thermal_phenomenon
 - Chemical_phenomenon
 - Electrical_phenomenon
 - Light_phenomenon
 - Unspecific_phenomenon
 - Radiological_phenomenon



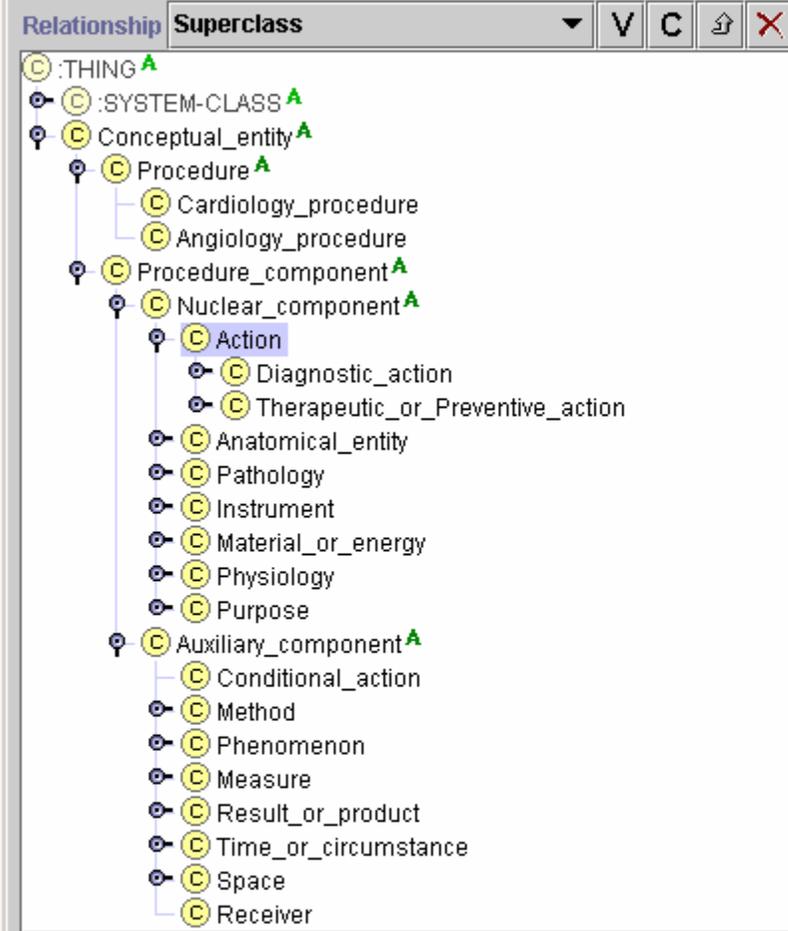
Superclasses

:THING

Name	Documentation	Constraints
:THING		
Role		
Abstract ^A		

Template Slots

Name	Type	Cardinality	Other Facets
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Superclasses + -

- Nuclear_component A

Action C X

Name: Action

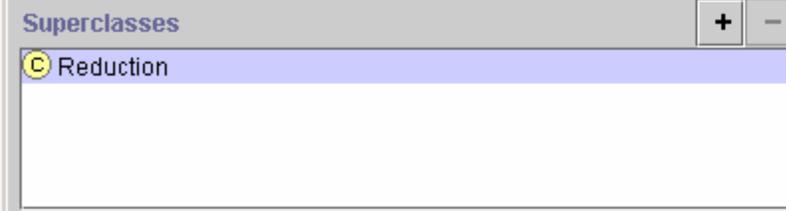
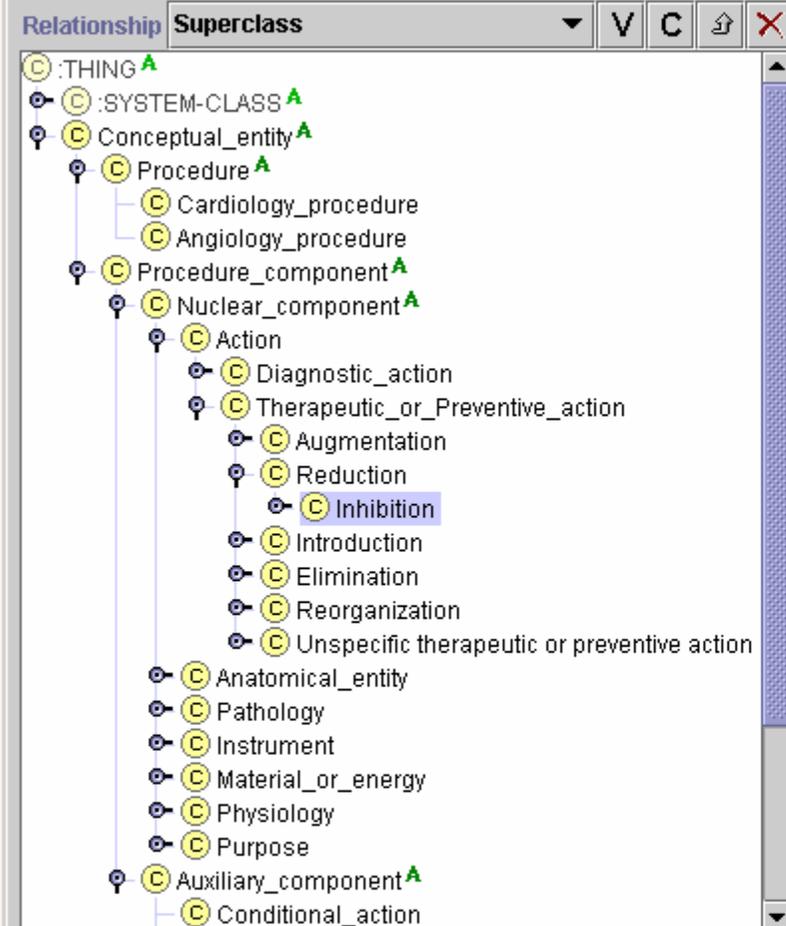
Documentation: Action realized in medical procedures, either 'Diagnostic' or 'Therapeutic or Preventive'.

Constraints: [Empty]

Role: Concrete

Template Slots

Name	Type	Cardinality	Other Facets
indicates	Instance	single	classes={Instrument}
has_goal_action	Instance	single	classes={Purpose}
uses_action	Instance	single	classes={Specific_method,Material_...}
has_location_action	Instance	single	classes={Anatomical_entity}
treats	Instance	single	classes={Pathology}
has_approach_action	Instance	single	classes={Anatomical approach}
diagnoses	Instance	single	classes={Pathology}
has_duration_action	Instance	single	classes={Time_duration}
evaluates	Instance	single	classes={Phenomenon}
concept_of_action	Symbol	single	allowed-values={Action,Application, ...}
affects	Instance	single	classes={Receiver,Pathology,Physic...}
produces	Instance	single	classes={Result_or_product}
prevents	Instance	single	classes={Pathology}
has_circumstance_action	Instance	single	classes={Time_phase}
has_act_action	Instance	single	classes={Conditional_action}



Inhibition

Name: Inhibition

Documentation: Action of reducing the intensity of a body function or rhythm by inducing an inhibiting response to a stimulus.

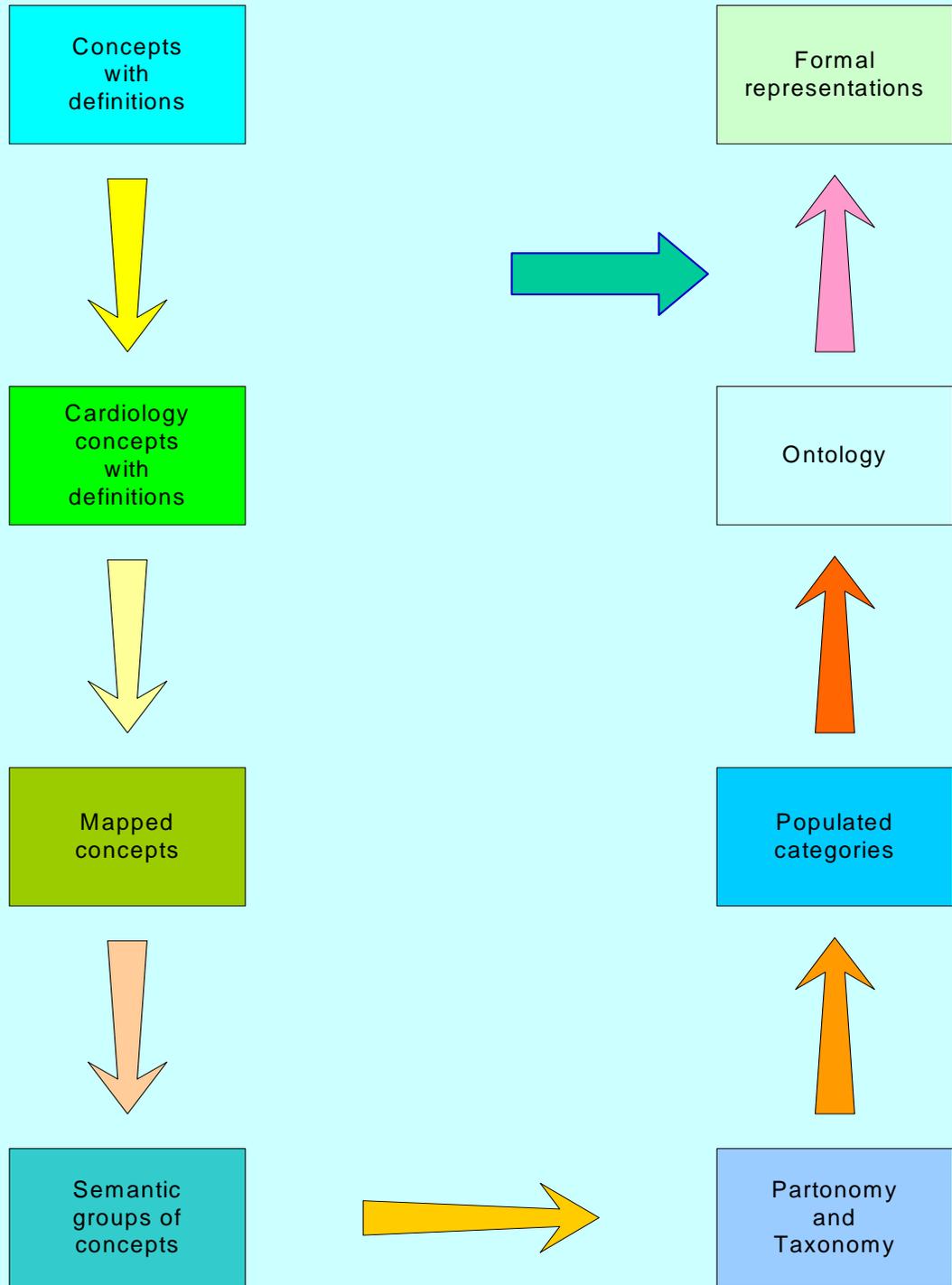
Constraints: [Empty]

Role: Concrete

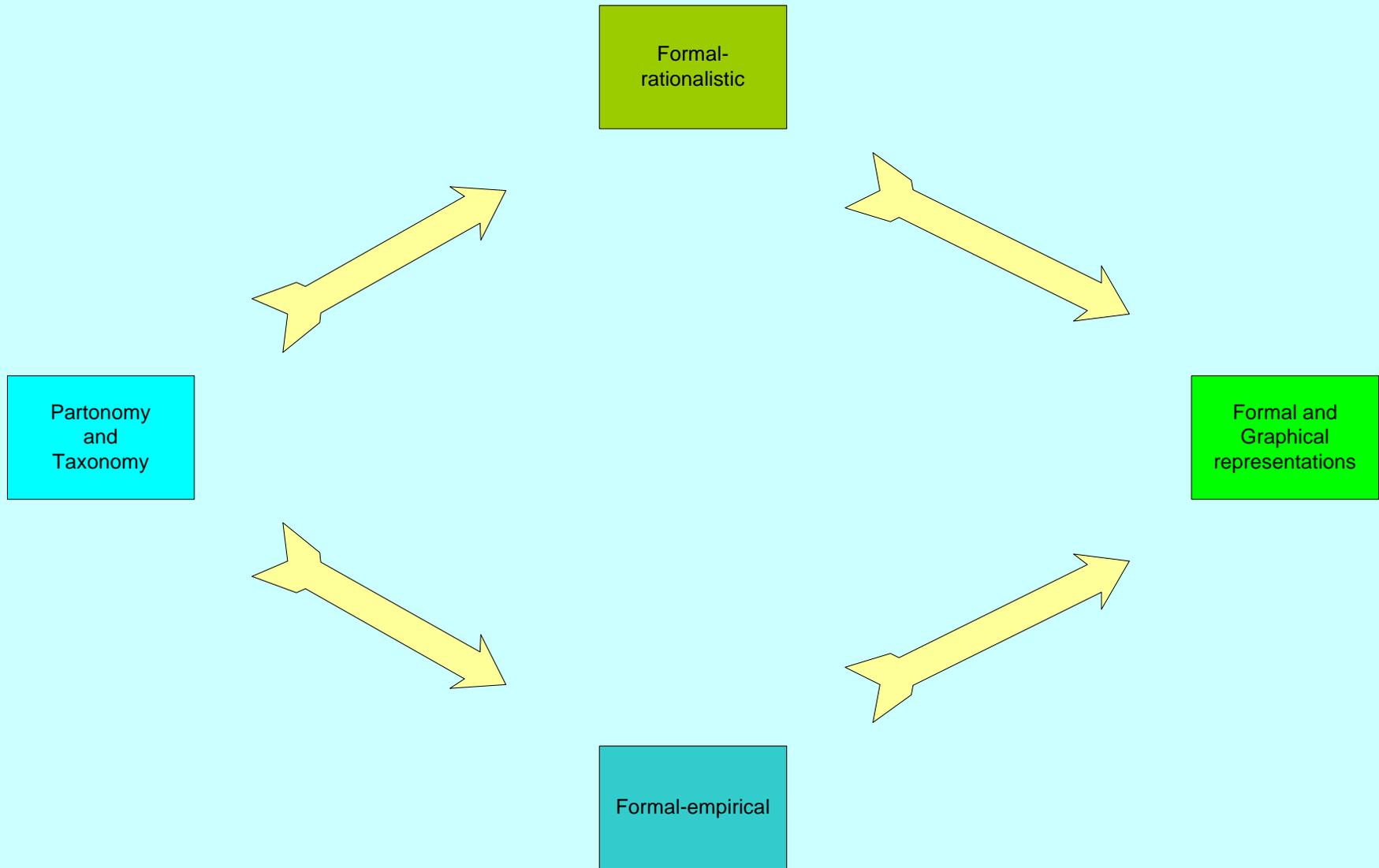
Template Slots

Name	Type	Cardinality	Other Facets
concept_of_inhibition	Symbol	single	allowed-values={hypothermia, induc...
concept_of_reduction	Symbol	single	allowed-values={Reduction - action, ...
concept_of_therapeutic_or_preve...	Symbol	single	allowed-values={pharmacotherape...
indicates	Instance	single	classes={Instrument}
has_goal_action	Instance	single	classes={Purpose}
uses_action	Instance	single	classes={Specific_method, Material_...
has_location_action	Instance	single	classes={Anatomical_entity}
treats	Instance	single	classes={Pathology}
has_approach_action	Instance	single	classes={Anatomical approach}
diagnoses	Instance	single	classes={Pathology}
has_duration_action	Instance	single	classes={Time_duration}
evaluates	Instance	single	classes={Phenomenon}
concept_of_action	Symbol	single	allowed-values={Action, Application, ...
affects	Instance	single	classes={Receiver, Pathology, Physic...
produces	Instance	single	classes={Result_or_product}
prevents	Instance	single	classes={Pathology}
has_circumstance_action	Instance	single	classes={Time_phase}
has_act_action	Instance	single	classes={Conditional_action}

*Diagram
of the
methodology
phases:*



From categories to relations:

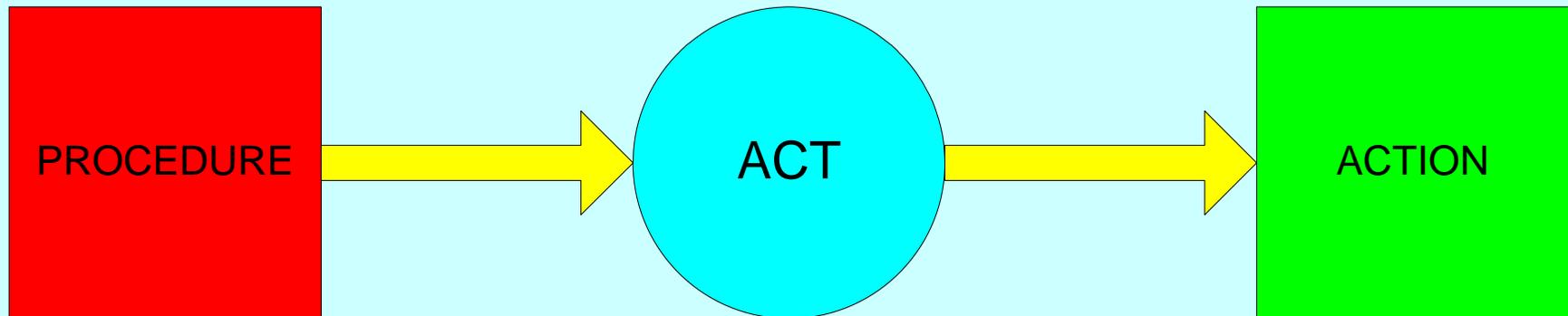


From categories to relations: *(approaches)*

- formal-rationalistic:
 - from basic categories
 - associative relationships for each pair
 - Sowa's model of conceptual structures
 - check for its concrete specific plausibility
- formal-empirical:
 - from original textual definitions
 - semantic relations of UMLS Semantic Network as syntax
 - translate plausible textual relations into formal ones
 - check abstract level with the Semantic Types

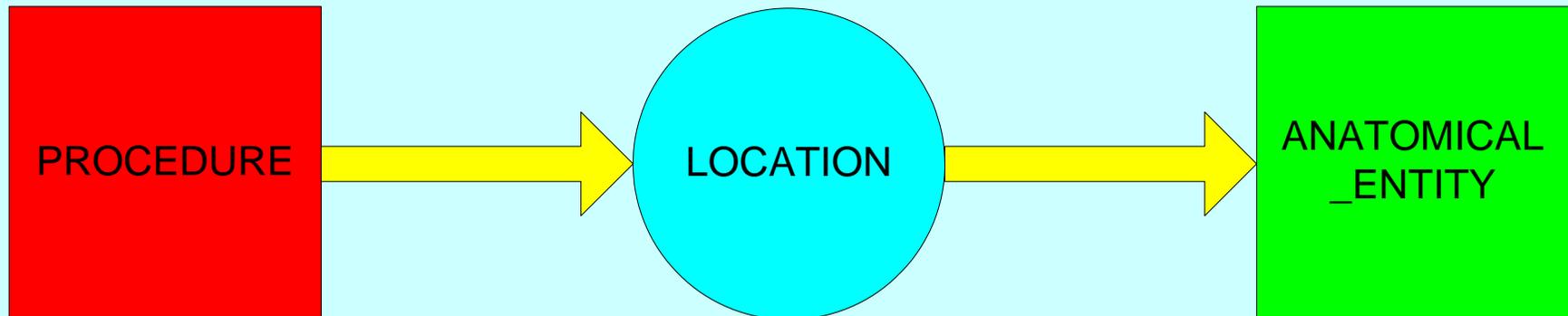
Procedure has_act Action

[PROCEDURE] > (ACT) > [ACTION]



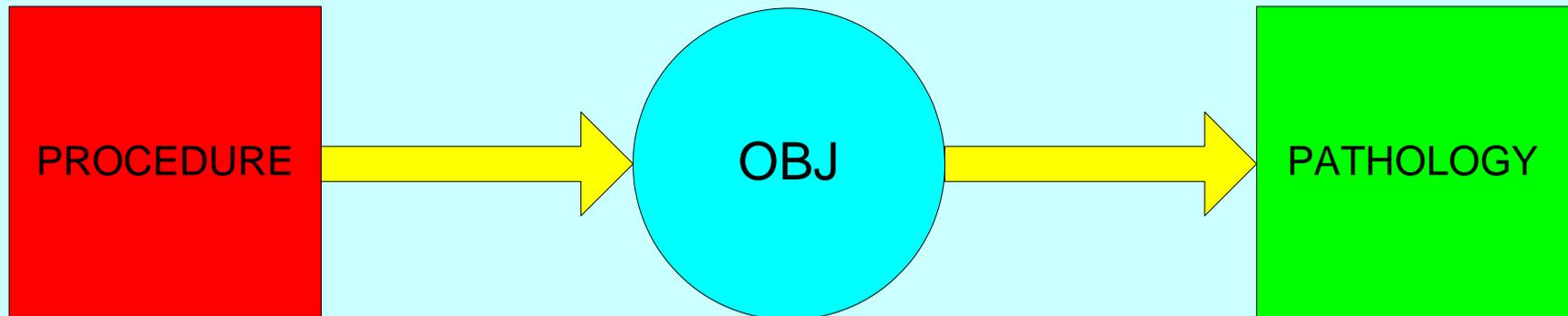
Procedure has_location Anatomical_entity

[PROCEDURE] > (LOC) > [ANATOMICAL-ENTITY]



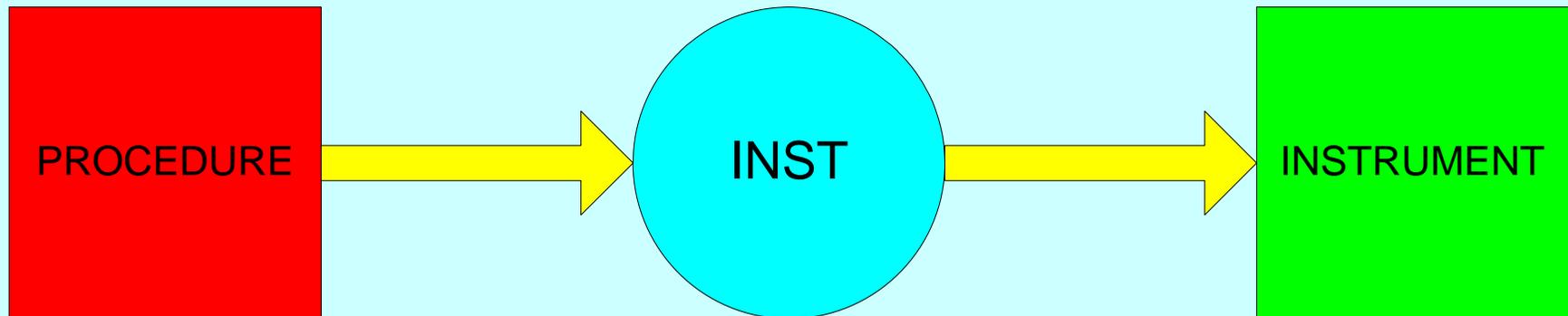
Procedure has_object Pathology

[PROCEDURE] > (OBJ) > [PATHOLOGY]



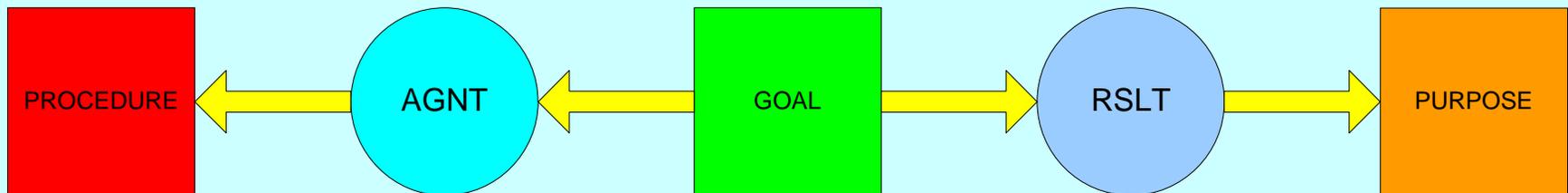
Procedure has_instrument Instrument

[PROCEDURE] > (INST) > [INSTRUMENT]



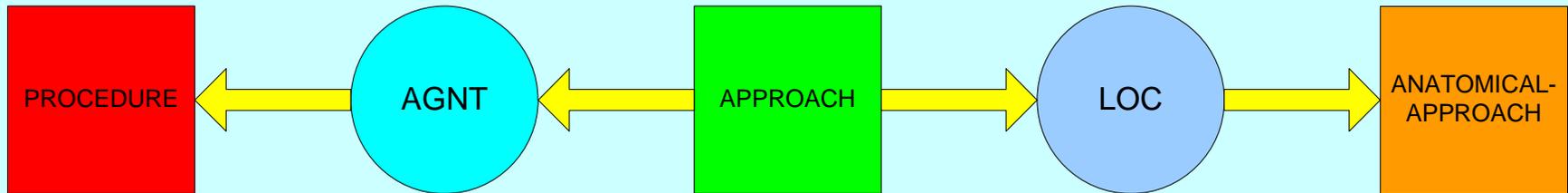
Procedure has_goal Purpose

[PROCEDURE] < (AGNT) < [GOAL] > (RSLT) > [PURPOSE]



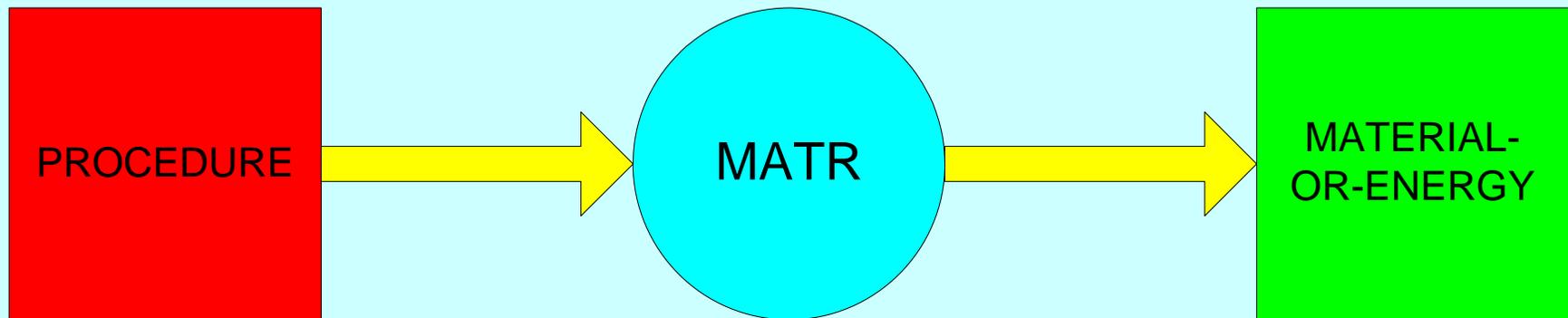
Procedure has_approach Anatomical_approach

[PROCEDURE] < (AGNT) < [APPROACH] > (LOC) > [ANATOMICAL-APPROACH]



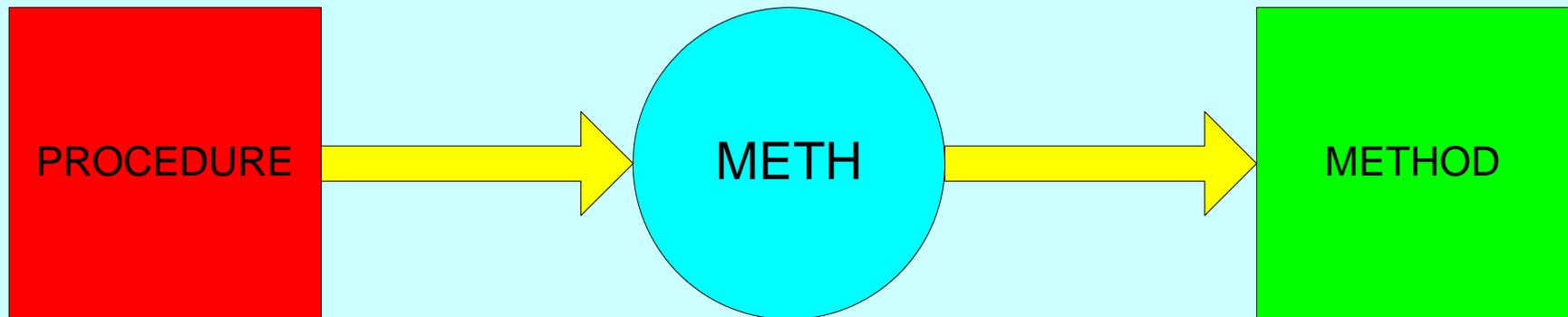
Procedure has_material Material_or_energy

[PROCEDURE] > (MATR) > [MATERIAL-OR-ENERGY]



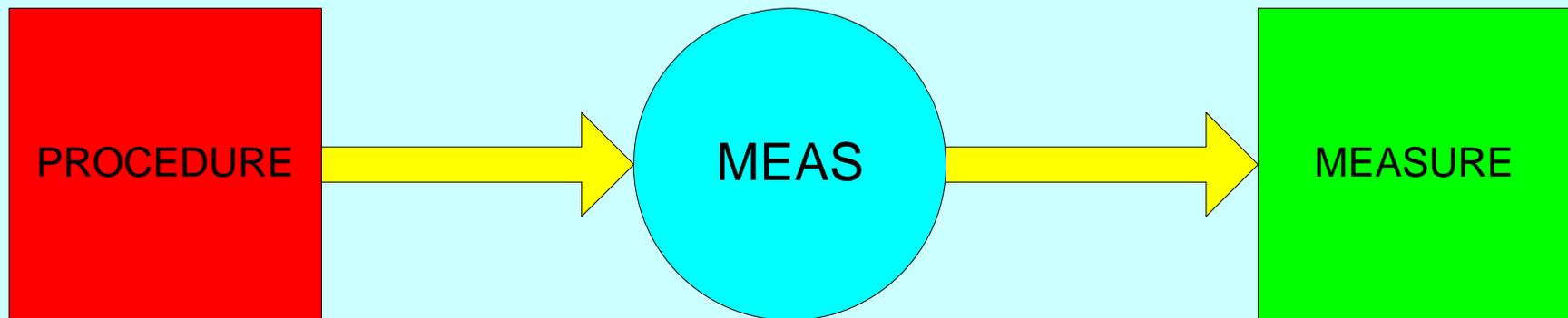
Procedure has_method Method

[PROCEDURE] > (METH) > [METHOD]



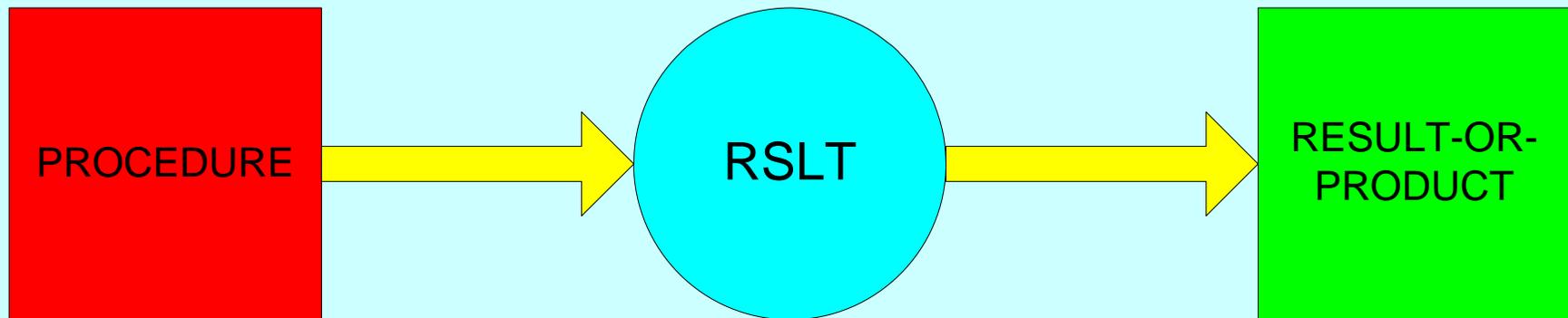
Procedure has_measure Measure

[PROCEDURE] > (MEAS) > [MEASURE]



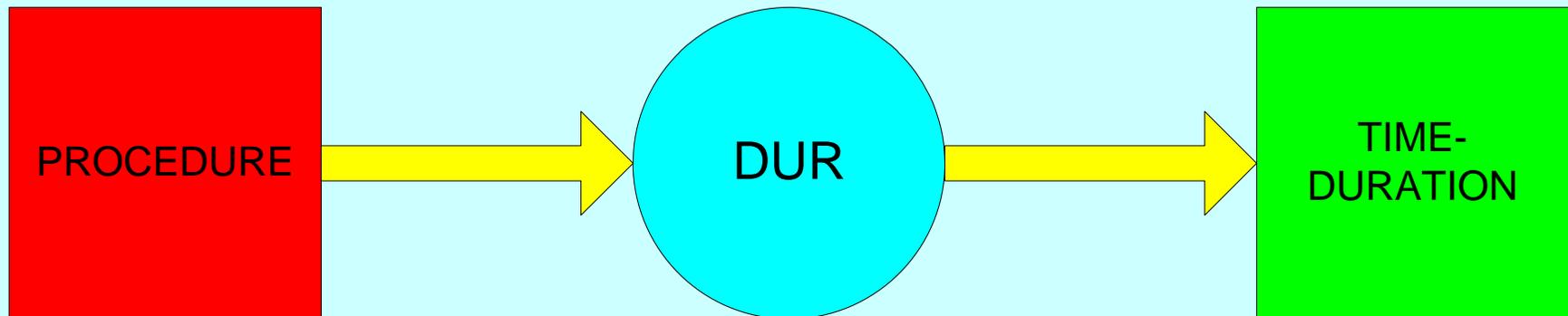
Procedure has_result Result_or_product

[PROCEDURE] > (RSLT) > [RESULT-OR-PRODUCT]



Procedure has_duration Time_duration

[PROCEDURE] > (DUR) > [TIME-DURATION]



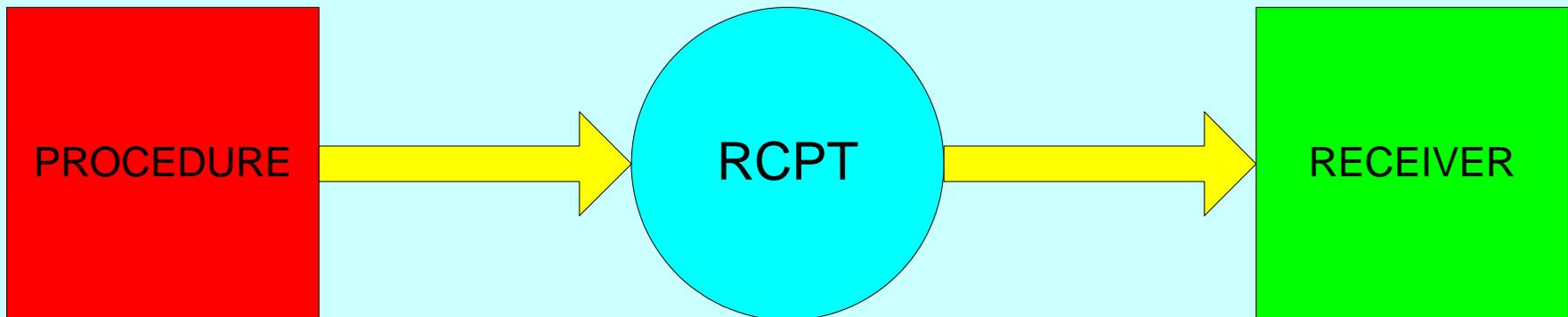
Procedure has_circumstance Time_phase

[PROCEDURE] < (STRT) < [CIRCUMSTANCE] > (PTIM) > [EVENT]



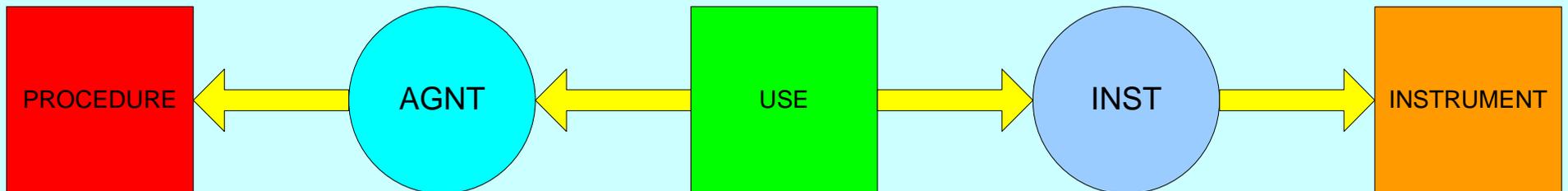
Procedure has_recipient Receiver

[PROCEDURE] > (RCPT) > [RECEIVER]



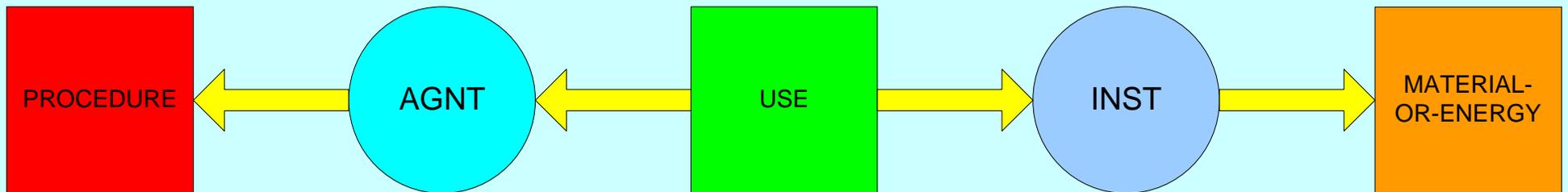
Procedure uses Instrument

[PROCEDURE] < (AGNT) < [USE] > (INST) > [INSTRUMENT]



Procedure uses Material_or_energy

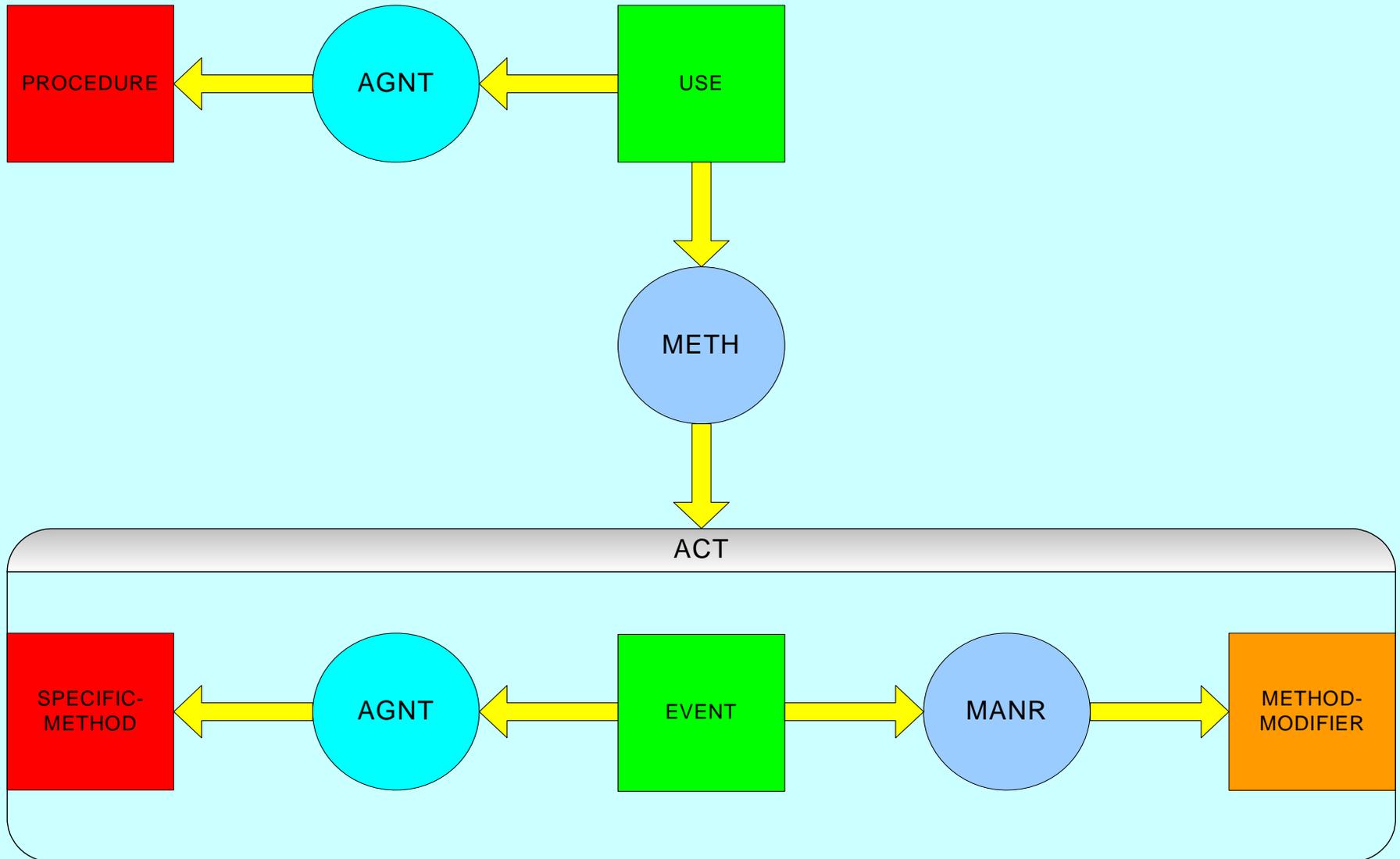
[PROCEDURE] < (AGNT) < [USE] > (INST) > [MATERIAL-OR-ENERGY]



Procedure uses Method

[PROCEDURE] < (AGNT) < [USE] > (METH) >

[ACT: [SPECIFIC-METHOD] < (AGNT) < [EVENT] > (MANR) > [METHOD-MODIFIER]]



Procedure uses Measure

[PROCEDURE] < (AGNT) < [USE] > (INST) >
[MEASURE: [DIMENSION] > (QTY) > [NUMBER]]

