

Department of Biomedical Informatics, University at Buffalo
Grand Rounds September 9th, 2016

Knowledge acquisition and management for clinical decision-making

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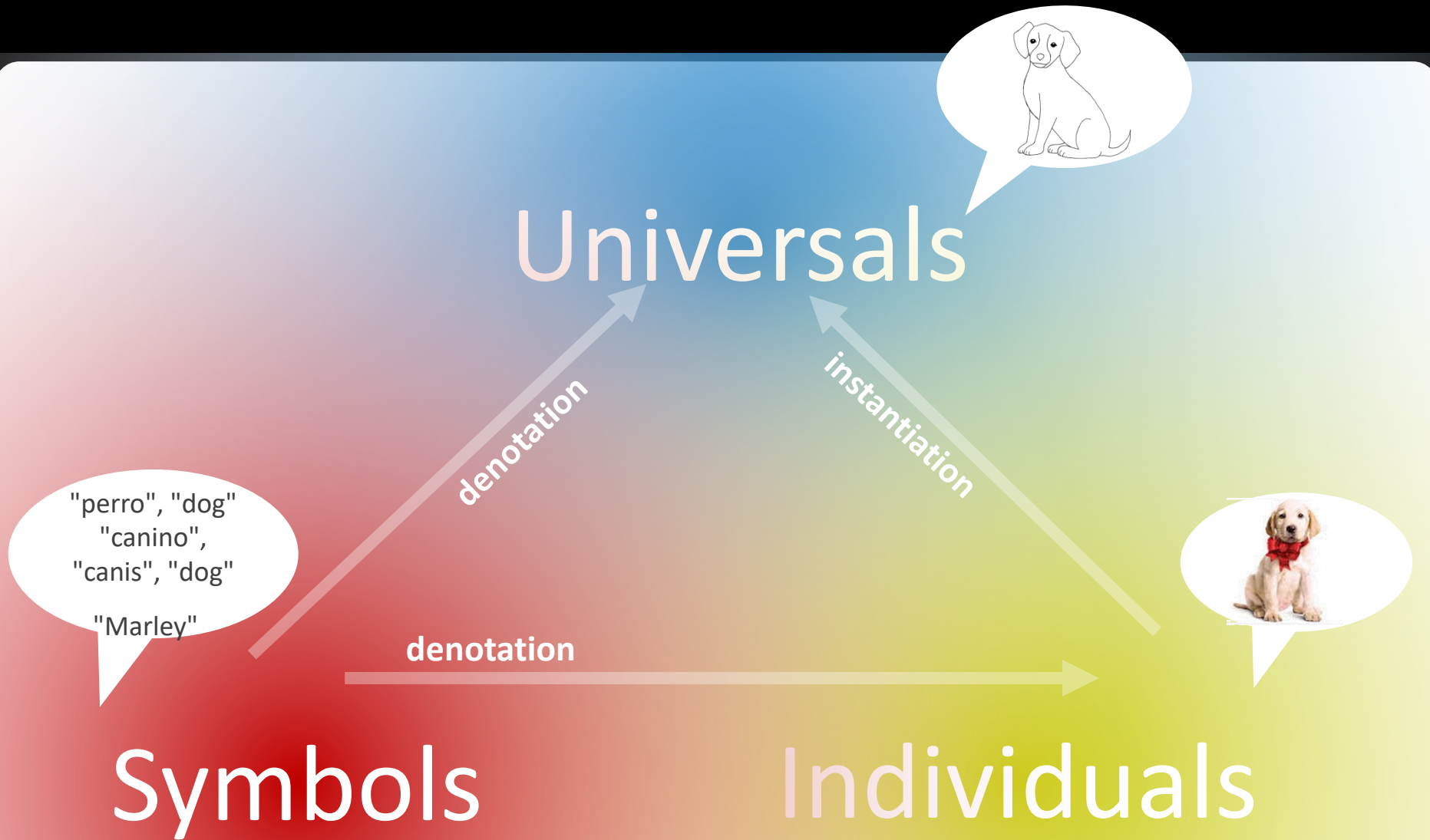


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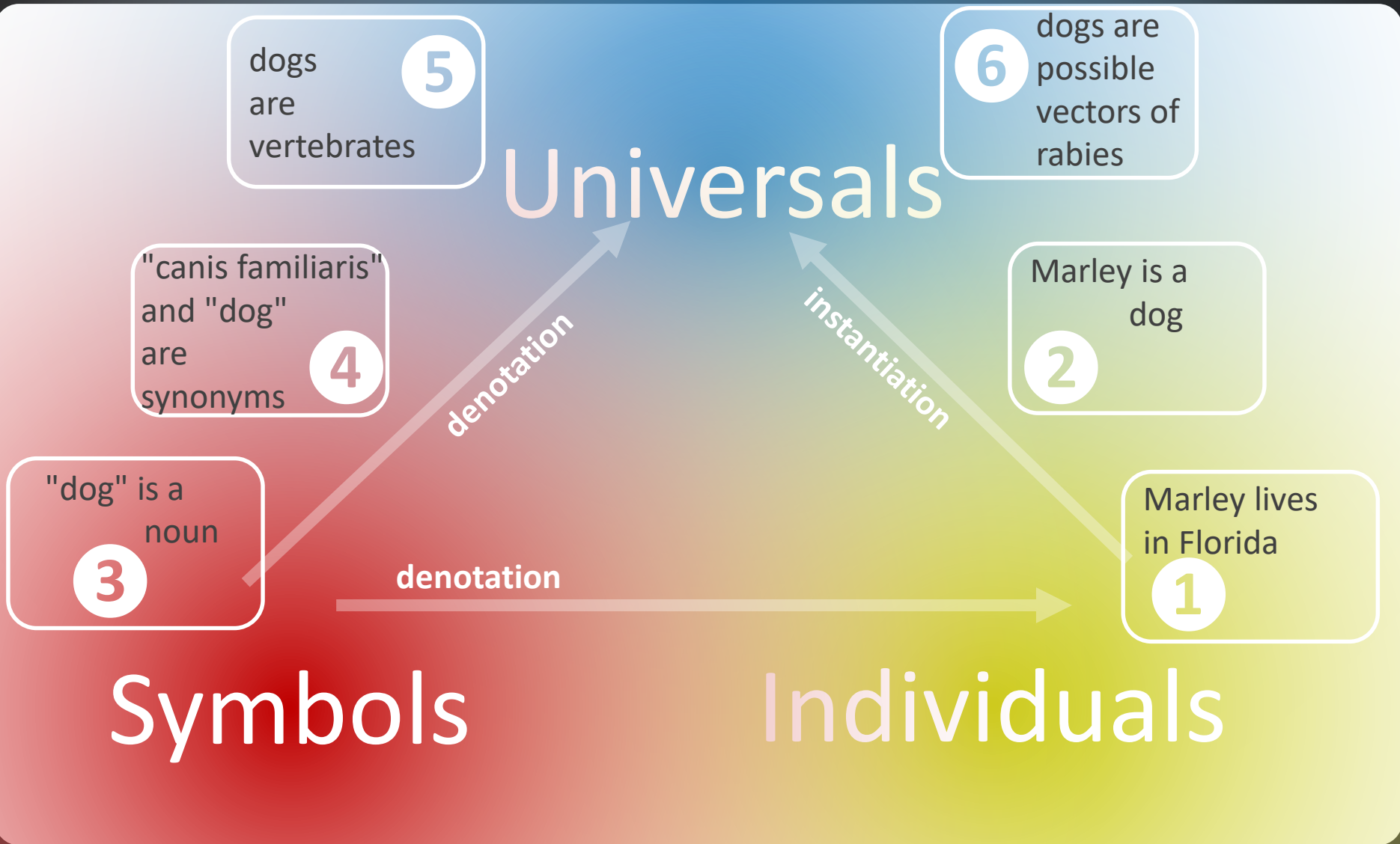
Topics of the talk

- Context: representation of knowledge in computer science
- What are the types of knowledge to be distinguished
- How are they connected
- How can data help acquire and maintain knowledge?

Knowledge map



Knowledge map

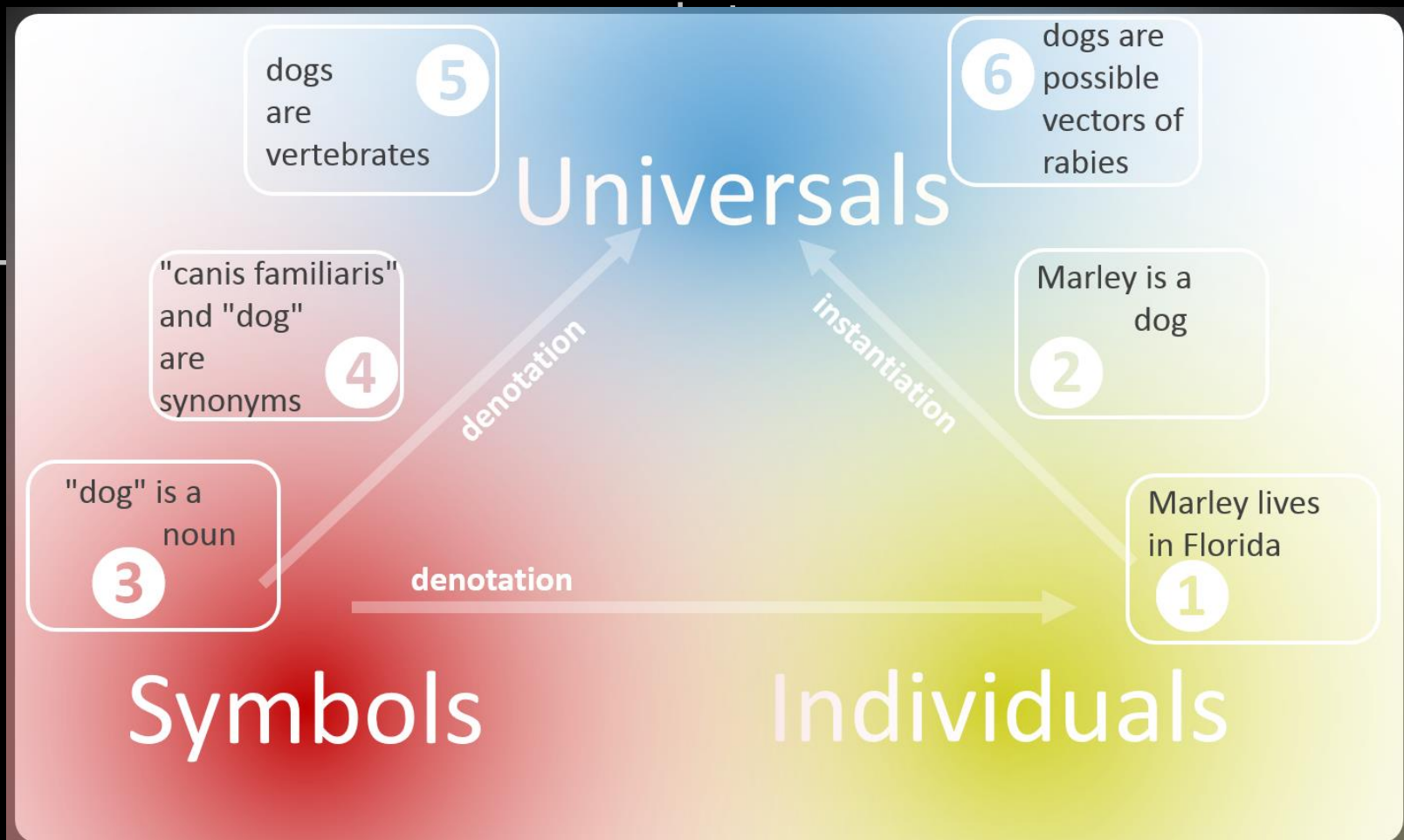


Ontological knowledge:

Axioms that are universally true

Contingent knowledge:

typical, likely, possible

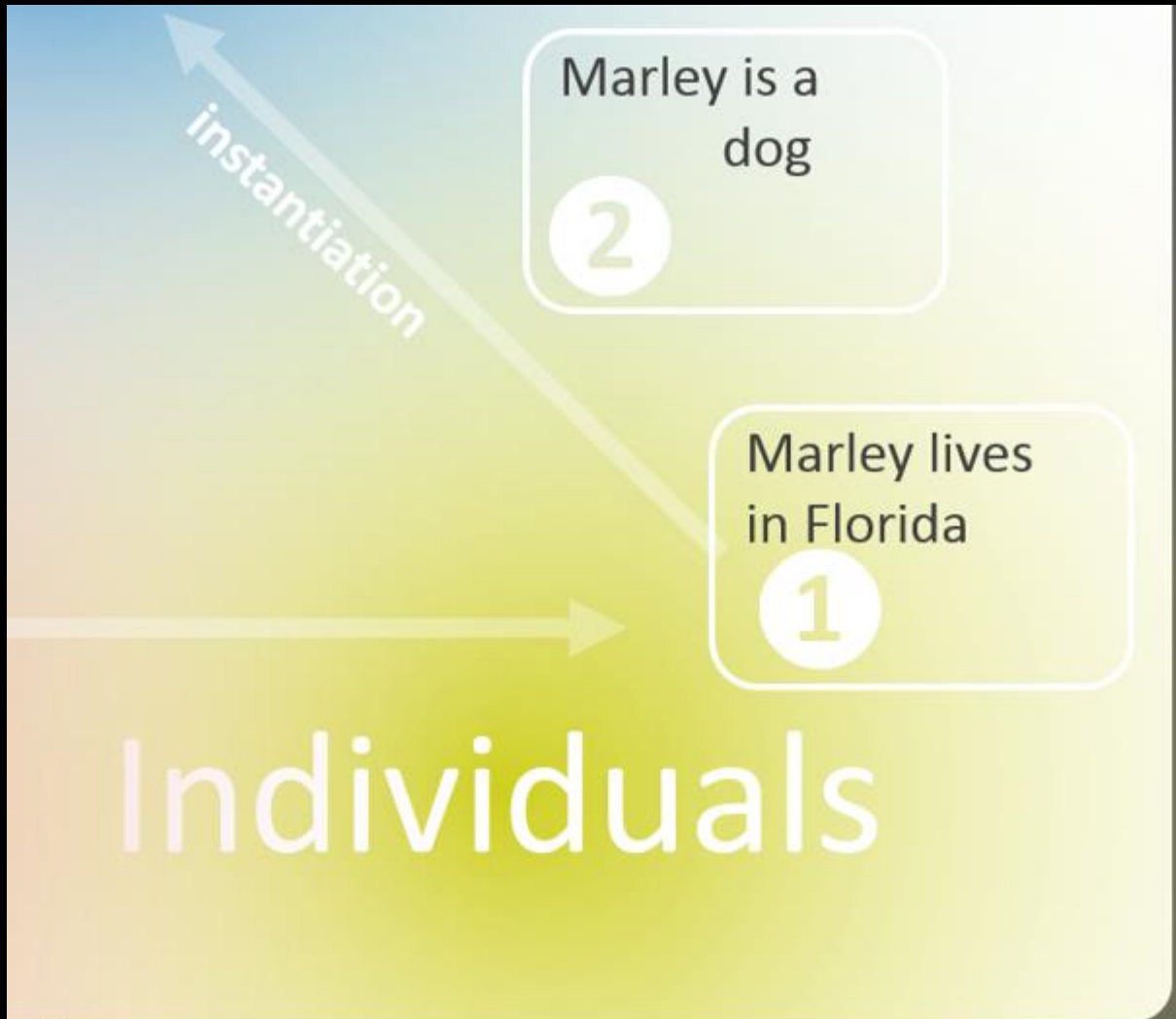


Linguistic knowledge:

Statements about properties and meaning of signs of language

Factual knowledge:

Statements about concrete entities and their relationships



Factual knowledge:

Statements about concrete entities
and their relationships

Statements about individuals

<Subject> <Predicate> <Object>

:Florida rdf:type :state

:Marley rdf:type :dog

:Marley :lives :Florida

Syntax TURTLE : <https://www.w3.org/TR/turtle/>

Information extraction

Physical examination on admission revealed purpura of the upper and lower extremities, swelling of the gums and tonsils, but no symptoms showing the complication of myasthenia gravis. Hematological tests revealed leucocytosis: WBC count 68 700/ μ l (blasts 11.5%, myelocytes 0.5%, bands 2.0%, segments 16.0%, monocytes 65.5%, lymphocytes 4.0%, atypical lymphocytes 0.5%), Hb 7.1 g/dl (reticulocytes 12%) and a platelet count of 9.1×10^4 / μ l. A bone marrow aspiration revealed hypercellular bone marrow with a decreased number of erythroblasts and megakaryocytes and an increased number of monoblasts



<http://www.beverlyhillsneurology.com/myastheniagravis-symptoms.html>

Instantiation

rdf:type

```
ADM{419620001
110714004
65124004 113279002
NEG{116223007 91637004 252275004
111583006} 767002[68700] 271040006[11.5]
313696224[0.5] 313696667[2.0]
313696009[16.0] 271037006[65.5]
271036002[4.0] 271036013[0.5]
365809007[7.1] 45995003[12]
365632008[91000] }
40401002 76107007 14016002
```

● Myasthenia gravis (disorder) ☆ 📄

SCTID: 91637004

91637004 | Myasthenia gravis (disorder) |

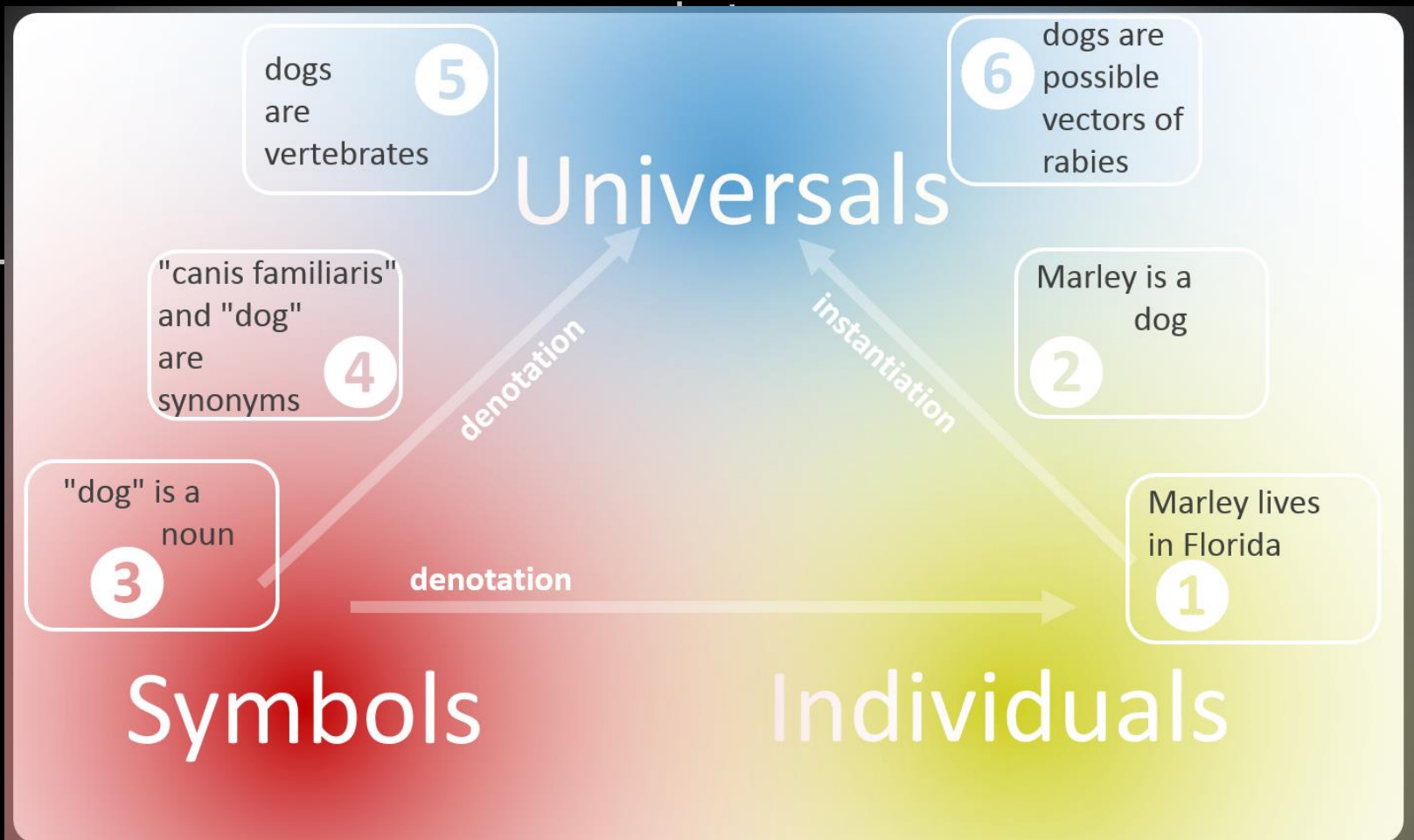
- Myasthenia gravis
- Erb-Goldflam disease
- Myasthenia gravis (disorder)
- MG - Myasthenia gravis

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"canis familiaris"
and "dog"
are
synonyms **4**

"dog" is a
noun **3**

denotation

denotation

Symbols

Linguistic knowledge:

Statements about properties and
meaning of signs of language

Representations SKOS / Linked Data

```
:ex:Dog rdf:type skos:Concept  
:ex:Dog skos:prefLabel "dog"@en;  
:ex:Dog skos:prefLabel "perro"@es;
```

```
:ex:Animal rdf:type skos:Concept  
:ex:Animal skos:broader ex:Dog
```

```
wr:dog      lemon:sense      wr:dog-English-Noun-1  
wr:dog      lemon:sense      wr:dog-English-Verb-1  
wr:dog-English-Noun-1      wt:hasPoS wt:Noun
```

Syntax TURTLE : <https://www.w3.org/TR/turtle/>

Wiktionary: <http://wiki.dbpedia.org/wiktionary-rdf-extraction>

3 Linguistic knowledge: POS tagging

"Myasthenia" is a noun

JJ Physical NN examination IN on NN admission VB of DT the JJ upper CC and JJR lower NNS extremities VBG swelling IN of DT the NN gums CC and NNS tonsils , , CC but DT no NNS symptoms VBG showing DT the NN complication IN of NN myasthenia NN gravis . . JJ Hemato logical NNS tests VBD revealed NN leucocytosis : : NNP WBC VBP count CD 68 CD 700 NN / NN μ NN | -LRB- (NNS blasts CD 11.5 NN % , , VBZ myelocytes CD 0.5 NN % , , NNS bands CD 2.0 NN % , , NNS segments CD 16.0 NN % , , VBZ monocytes CD 65.5 NN % , , VBZ lymphocytes CD 4.0 NN % , , NN atypical VBZ lymphocytes CD 0.5 NN % -RRB-) , , NNP Hb CD 7.1 NN g NN / NN dl -LRB- (VBZ reticulocytes CD 12 NN % -RRB-) CC and DT a NN platelet NN count IN of CD 9.1 NN \times CD 104 NN / NN μ NN | . . DT A NN bone NN marrow NN aspiration VBD revealed JJ hypercellular NN bone NN marrow IN with DT a VBN decreased NN number IN of NNS erythroblasts CC and NNS megakaryocytes CC and DT an VBN increased NN number IN of NNS monoblasts

4 Linguistic knowledge: Expansion of short forms

- pattern to extract acronym definitions from a corpus

Pattern	Example
Acronym (Definition)	CVA (Cerebrovascular accident)
Definition (Acronym)	Cerebrovascular accident (CVA)
Acronym – Definition	CVA – Cerebrovascular accident–
Definition – Acronym	Cerebrovascular accident–ACV–

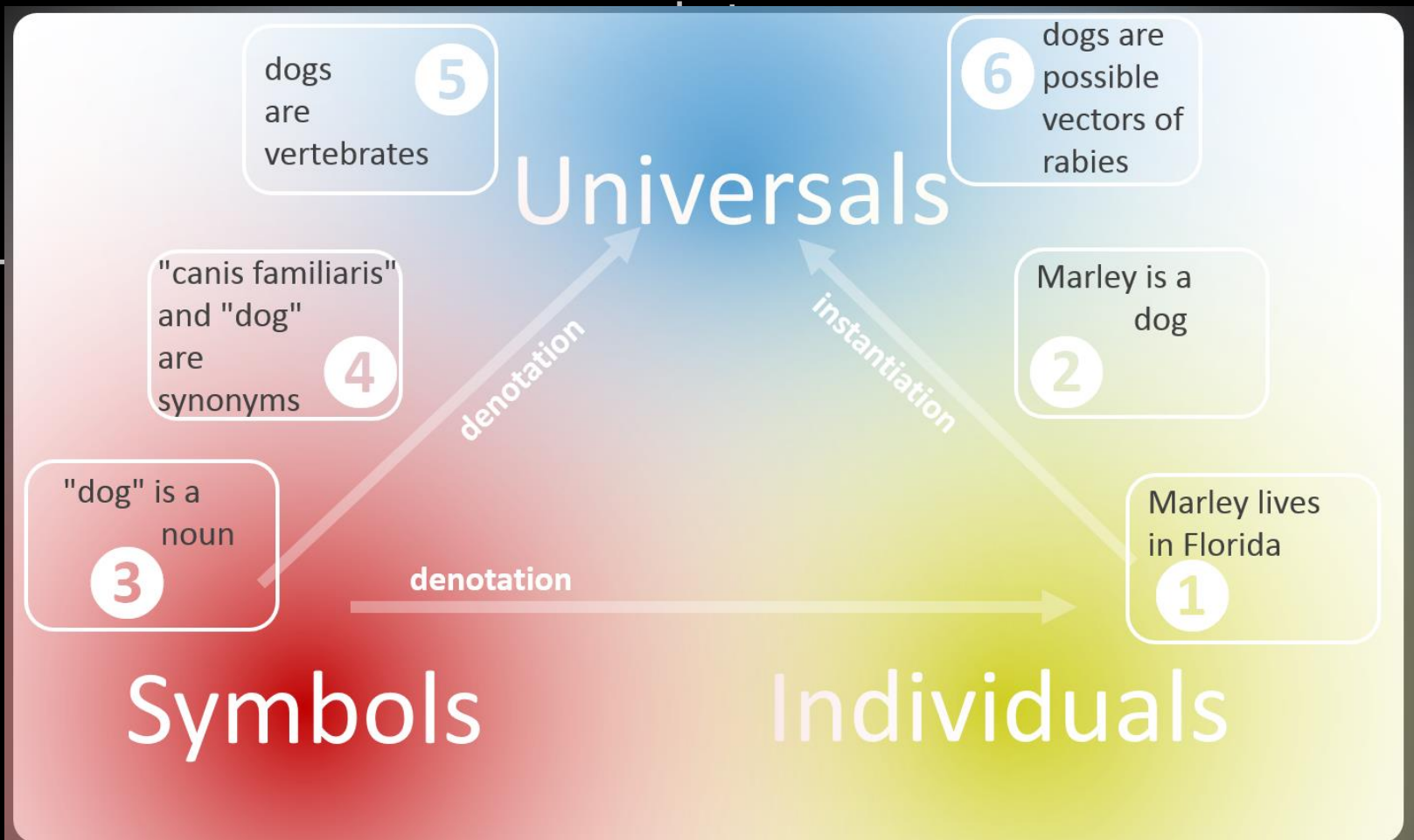
C < V < A

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6

Ontological knowledge:

Axioms that are universally true

dogs
are
vertebrates

5

Universals

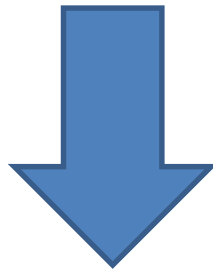
Representation OWL

Dog subclassOf Vertebrate

Vertebrate subclassOf Animal

Vertebra subclassOf Bone

Vertebrate equivalentTo Animal and
has-part some Bone



computable inference
(e.g. Hermit or Fact++
OWL reasoner)

There is no dog that has no bones

OWL Manchester Syntax: <https://www.w3.org/TR/owl2-manchester-syntax/>

Hermit reasoner: <http://www.hermit-reasoner.com/>

Fact++ reasoner: <http://owl.man.ac.uk/factplusplus/>

Active Ontology x Entities x Individuals by class x DL Query x

Class hierarchy Class hierarchy (inferred)

Class hierarchy: vertebrado

Annotations: vertebrado

Description: vertebrado

Equivalent To +

- **hasPart some vertebra**

SubClass Of +

- **animal**

General class axioms +

SubClass Of (Anonymous Ancestor)

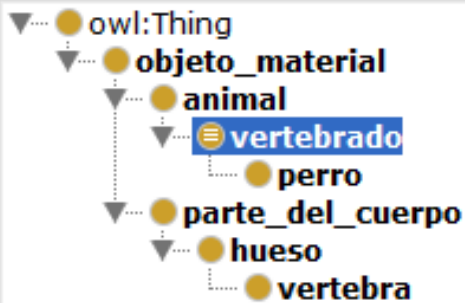
owl:Thing

- objeto_material
 - animal
 - **vertebrado**
 - perro
 - parte_del_cuerpo
 - hueso
 - vertebra

Active Ontology x Entities x Individuals by class x DL Query x

Class hierarchy Class hierarchy (inferred)

Class hierarchy: vertebrado



Class Annotations Class Usage

Annotations: vertebrado

Query (class expression)

perro and not (hasPart some hueso)

Execute

Add to ontology

Query results

Equivalent classes (1)

owl:Nothing

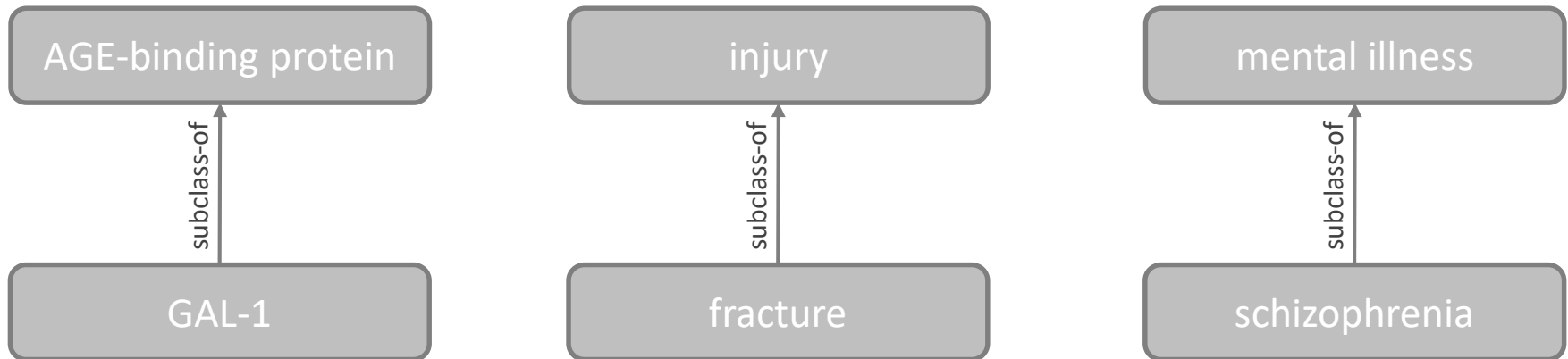
Direct subclasses (0)

Subclasses (0)

Extraction of taxonomic relations

- Lexical-semantic patterns Hearst:

Patrón	Ejemplo
NP such as {NP}* (and or) NP	"AGE-binding proteins, such as GAL-3"
NP {NP}* (and or) other NP	"fractures or other Injuries"
NP including {NP}* (or and) NP	"mental illnesses including schizophrenia"



5 Extracción de otras relaciones

- Validation of SNOMED CT axioms by Web mining
- Query:
"Gastritis is an inflammation of"

Current Concept:

Fully Specified Name: Gastritis (disorder)

ConceptId: 4556007

Defining Relationships:

Is a Disorder of stomach (disorder)

Is a Inflammation of specific body organs (disorder)

Is a Inflammatory disorder of digestive tract (disorder)

Group 1

Associated morphology [Inflammation \(morphologic abnormality\)](#)

Finding site [Stomach structure \(body structure\)](#)

This concept is fully defined.

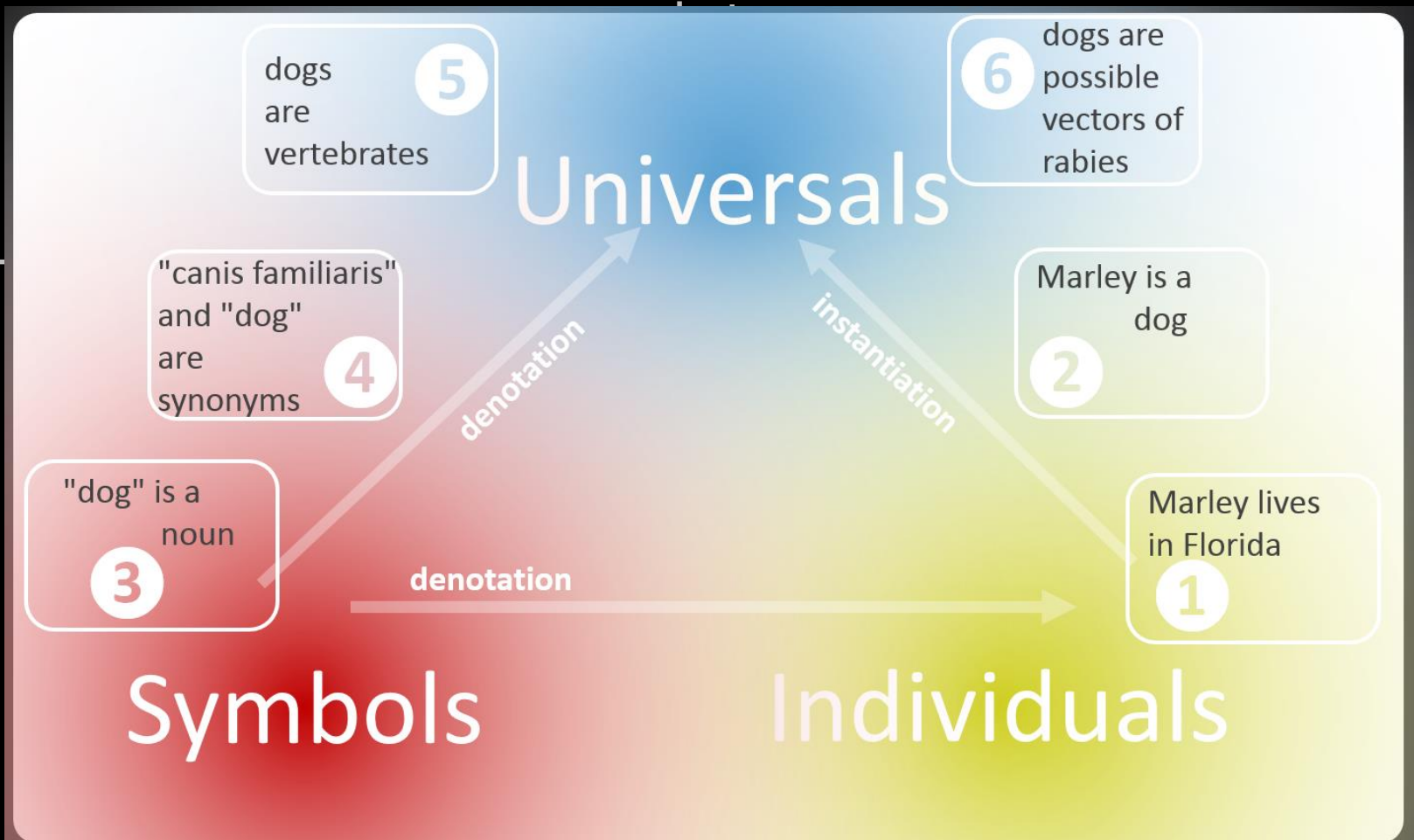
Result	Frequency
"stomach lining"	44
"lining of the stomach"	22
"lining of your stomach"	3
"lining of stomach"	1

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e

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typical, likely, possible

Universals

6

dogs are
possible
vectors of
rabies

Contingent knowledge

Alan Rector (2008):

" very few interesting items of knowledge that are truly ontological...". Much current work on informatics ontologies is aimed at integrating probabilistic and typical reasoning with universal "ontological" reasoning effectively. Hence, the background information for a clinical system often goes well beyond the ontology, in this strict sense. Brachman introduced the notion of the ontology as a "conceptual coat rack" on which other information is held."

Don't do this !

~~Dog subclassOf
vector-of some Rabies~~

~~Rabies subclassOf
has-vector some Dog~~

~~Tobacco subclassOf
causes some Cancer~~

~~Cancer subclassOf
caused-by some Tobacco~~

~~Aspirin subclassOf
treats some Pain~~

~~Pain subclassOf
treated-by some Aspirin~~

Triple representation

- No formal semantics!
- Different, mostly complex interpretations

<Subject>	<Predicate>	<Object>
:Dog	:vector-of	:Rabies
:Tobacco	:causes	:Cancer
:Aspirin	:treats	:Pain
:Fever	:suggests	:Malaria
:Bird	:capable-of	:Flying

6 Example: extraction of contingent (non ontological knowledge) from MEDLINE

- Co-occurrence analysis

Source concept	<i>Name</i>	Bipolar disorder
	<i>Type</i>	Disorder
Target concept	<i>Name</i>	Tricyclic antidepressant
	<i>Type</i>	Substance
MeSH subheadings		DT=9,CI=7,DI=5,PX=4,CO=2,EP=2,GE=2,BL=1,ET=1,PA=1,PC=1,PP=1,TH=1
<i>Absolute co-occurrence</i>		17
<i>Log-likelihood</i>		54.57

Qualifies source concept, e.g:

DT = "drug therapy"

PC = "prevention and control"

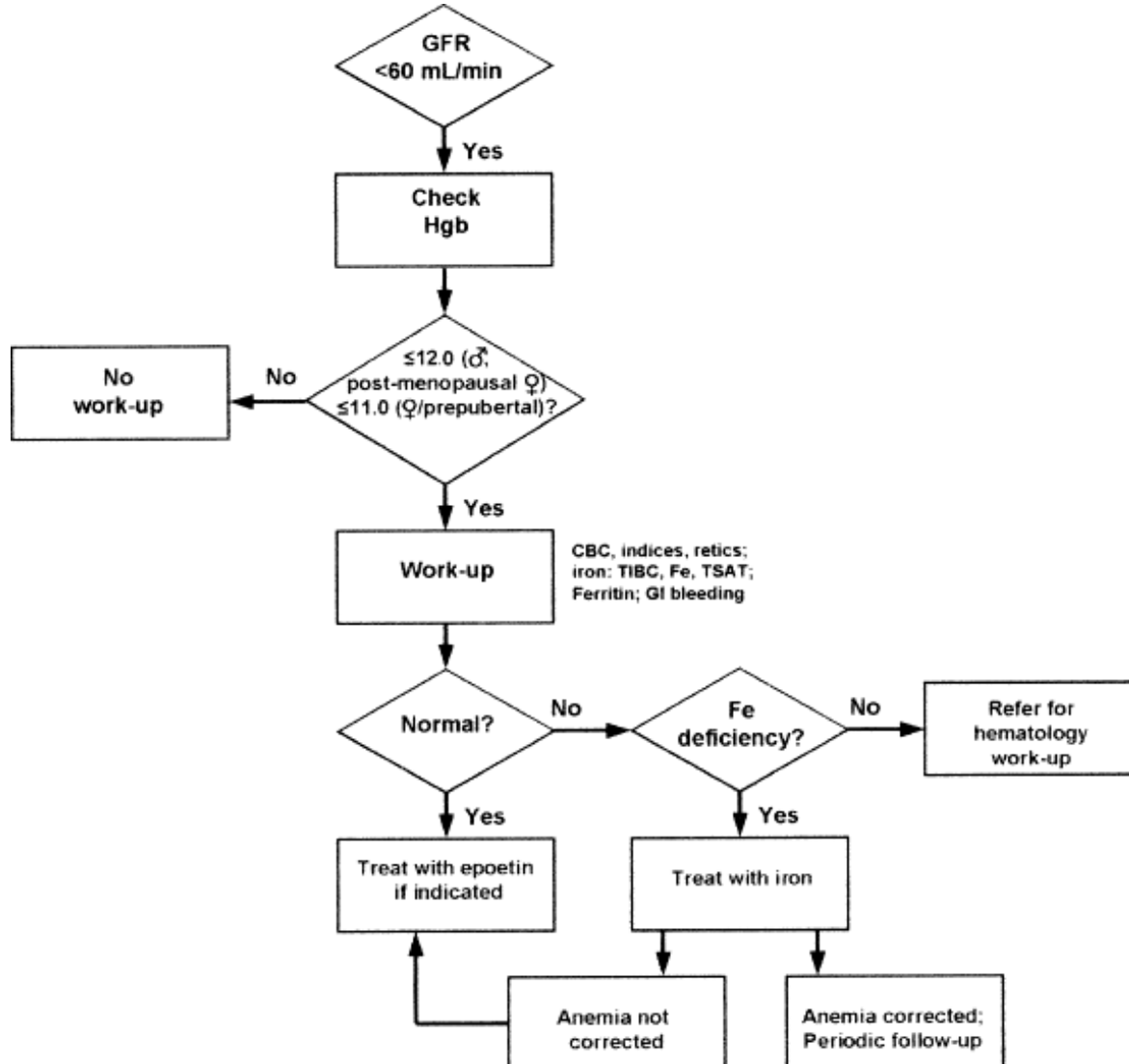
CO = "complication"

Predicate extractions according to MeSH subheadings analysis

		Object			
		Disease	Finding	Substance	Organism
Subject	Finding	<i>sign of symptom of</i>	<i>accompanied by</i>	<i>treated by</i>	<i>affects caused by</i>
	Substance	<i>causes treats prevents metabolite of</i>	<i>causes treats prevents</i>	<i>Interacts with</i>	<i>affects produced by</i>
	Organism	<i>causes affected by</i>	<i>causes</i>	<i>sensitive to</i>	<i>interacts with</i>
	Body part	<i>possible location of</i>	<i>possible location of</i>	<i>targeted by</i>	<i>targeted by</i>

Simplified: high frequency of "TU" ("therapeutic use") suggests the predicate "treats", high frequency of "PC" (prevention & control) suggests "prevents"

Clinical practice guidelines

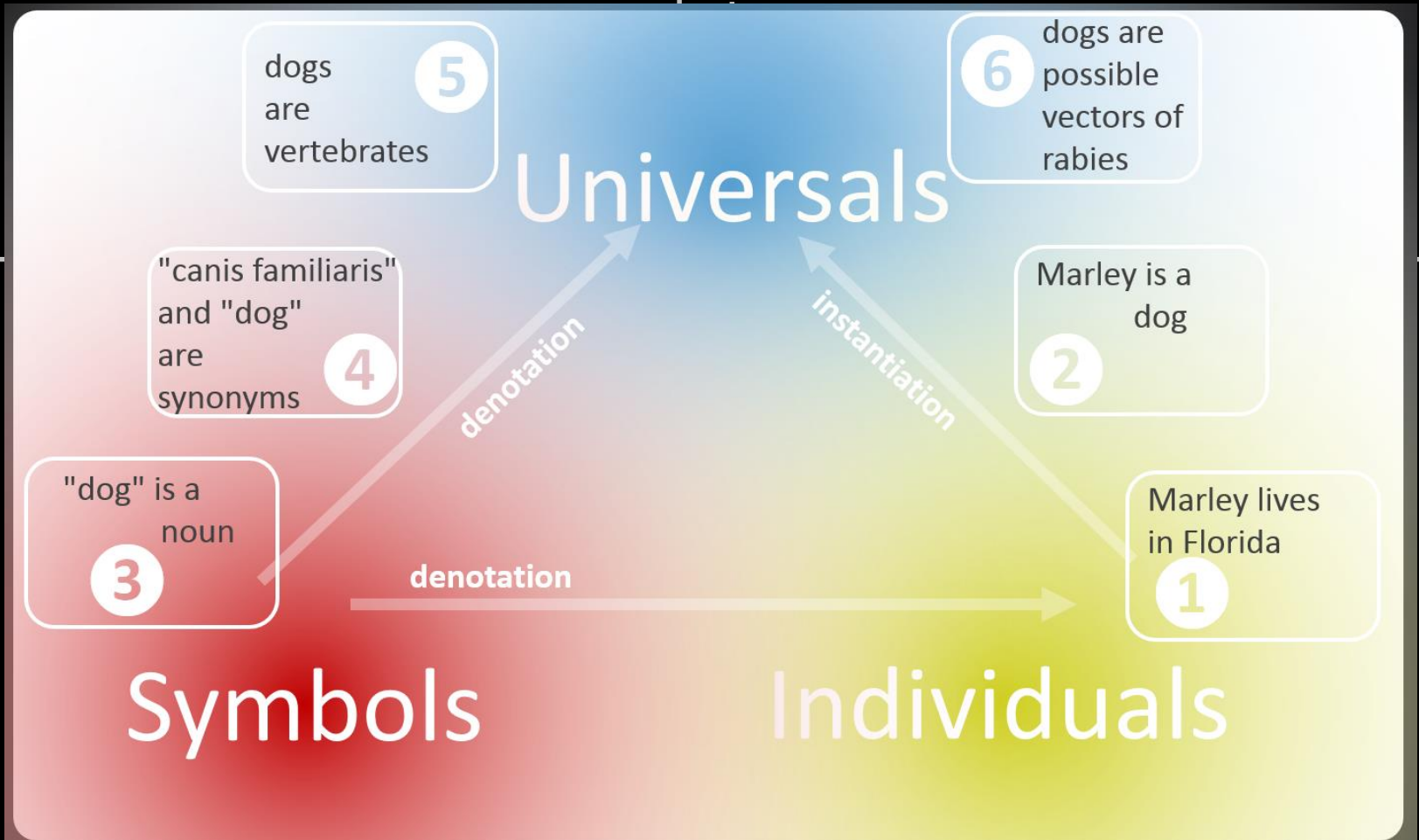


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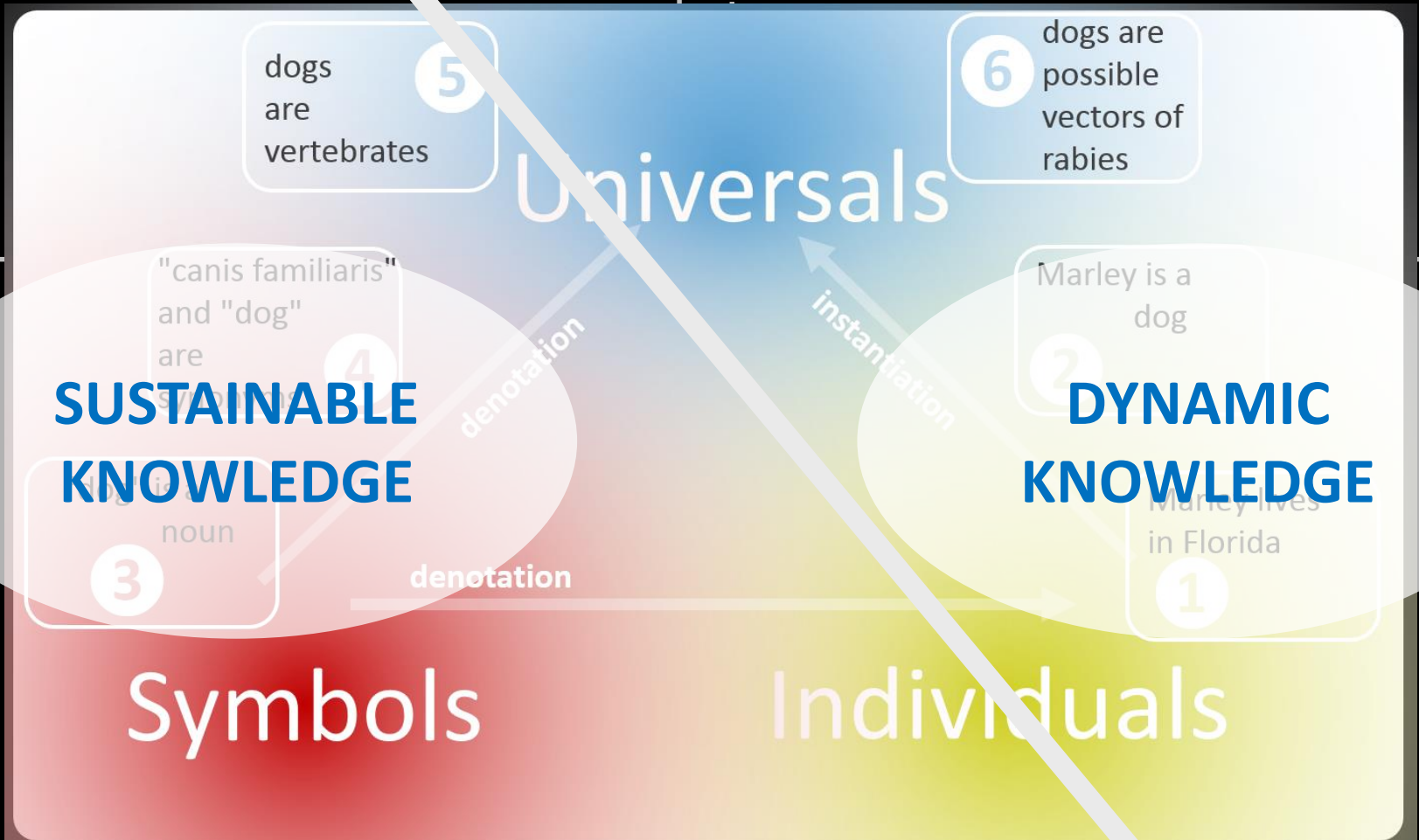
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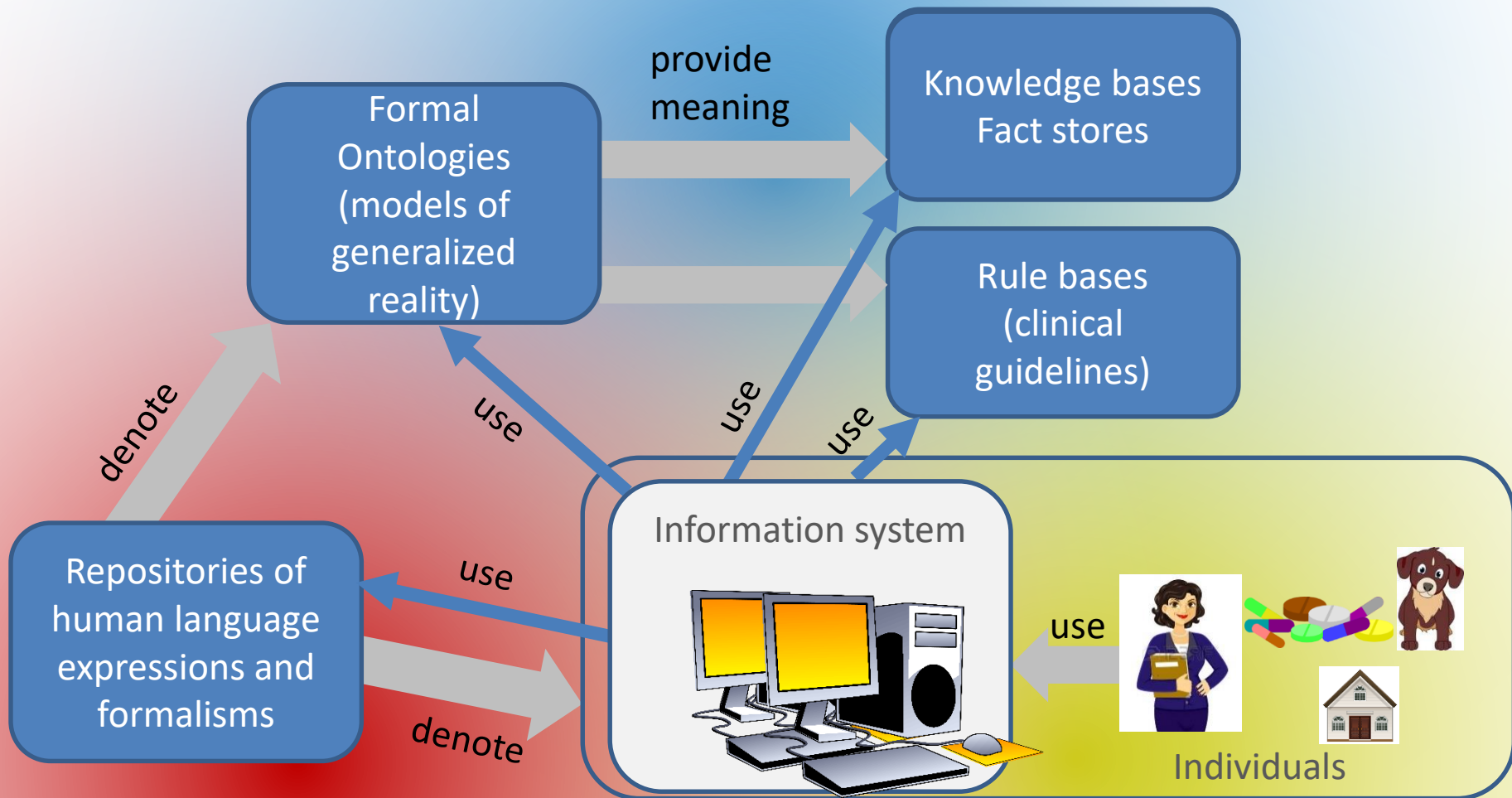
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Knowledge assets, users and information systems



Perspectives

- Use big data to help construct and maintain large knowledge resources
- Importance of unstructured data in clinical data management
 - Used for knowledge construction
 - Used as input for decision-making
- Sensitize knowledge engineers to distinguish between qualitatively different knowledge assets
- Distinction between sustainable and permanent knowledge assets

Questions?

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