

# ONTOLOGIES & DESCRIPTION LOGICS

Parcours IA - Représentation des connaissances

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# INTRODUCTION TO ONTOLOGIES

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For example, in the university domain:

- terms: **student, PhD student, professor, course, teaches, takes, supervises**
- relationships between terms:
  - **every PhD student is a student**
  - **cannot be both student and professor**
  - **every course is taught by some professor**

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To support **automated reasoning**

- **uncover errors in modelling (debugging)**
- **exploit knowledge in the ontology during query answering**, to get back a **more complete set of answers** to queries

# APPLICATIONS OF ONTOLOGIES: MEDICINE

General medical ontologies: **SNOMED CT**, GALEN

Specialized ontologies: FMA (anatomy), NCI (cancer), ...

The image displays an anatomical diagram of the interior view of the heart and a screenshot of the SNOMED CT ontology interface for the 'Aortic valve'.

**Interior View of the Heart:** The diagram shows the internal structures of the heart, including the Aorta, Pulmonary Artery, Left Atrium, Mitral Valve, Aortic Valve, Left Ventricle, Right Ventricle, Papillary Muscles, Orifices of Coronary Arteries, Inferior Vena Cava, Right Atrium, Tricuspid Valve, Pulmonary Valve, and Superior Vena Cava.

**SNOMED CT Interface:** The screenshot shows the 'Class hierarchy: Aortic valve' and 'Annotations: Aortic valve' panels. The class hierarchy lists various anatomical structures, including 'Aortic valve' and its subclasses. The annotations panel shows the 'Aortic valve' class with its label, definition, and various relationships, such as 'attaches\_to' and 'constititional\_part\_of'.

Querying & exchanging medical records (find patients for medical trials)

Supports analysis of health data for medical research

### Large-scale comprehensive medical ontology

- more than **350,000 terms** on all aspects of clinical healthcare:
  - symptoms, diagnosis, medical procedures, body structures, organisms, substances, pharmaceutical products, etc.

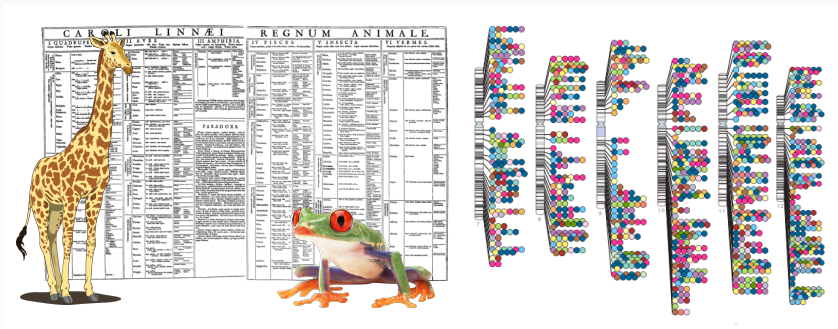


### Widely adopted standard for healthcare terminology

- in use in **> 80 countries** (including U.S., Canada, UK)
- utilized in **health IT** (e.g. IBM Watson Health, Babylon Health)



Hundreds of ontologies at BioPortal (<http://bioportal.bioontology.org/>):  
Gene Ontology (GO), Cell Ontology, Pathway Ontology, Plant Anatomy, ...

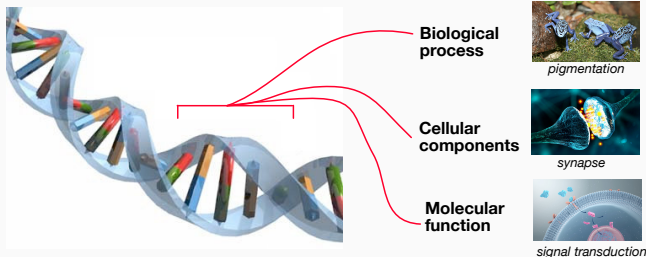


Help scientists share, query, & visualize experimental data

## ZOOM ON: GENE ONTOLOGY

Aim: **rigorous shared vocabulary** to describe the **roles of genes across different organisms**

Annotations: **evidence-based statements relating specific gene product to specific ontology terms**



Very successful endeavour:

- **> 100K published scientific articles** with keyword “Gene Ontology”
- **> 700K experimentally-supported annotations**

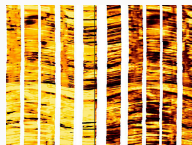
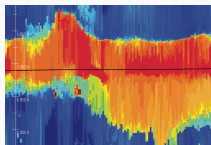
# APPLICATIONS OF OMQA: ENTREPRISE INFORMATION SYSTEMS

Companies and organizations have **lots of data**

- **need easy and flexible access** to **support decision-making**



Goal: aid geologists in gathering information for analysis



Difficulties:

- relevant data stored across **> 3000 database tables**
  - geologist question = **complex query** (thousands of terms, 50-200 joins)
- must ask IT staff, may take **several days to get answer**

Solution:

- **ontology provides familiar vocabulary** for query formulation
- **mappings link database tables to ontology terms**
- use reasoning to **automatically transform ontology query into executable database query**

Which languages can we use to **specify an ontology**?

## Natural language?

- **imprecise**, ambiguous, **not machine-interpretable**

## Formal logic?

(long studied in math, CS, philosophy)

- **first-order logic (FOL)**
  - **expressive**, well understood, BUT reasoning **undecidable**
- **'nice' fragments of FOL** (**description logics**, existential rules)
  - expressivity vs computational complexity tradeoff

## Standardized languages for the Web (OWL, RDFS)

- closely related and **based upon logic**
- geared to **broader public**, offer various formats

## DESCRIPTION LOGICS

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Computational properties well understood (decidability, complexity)

Many implemented reasoners and tools available for use

## Concept names

- correspond to sets of entities
- unary predicates in logic

Parent

Scientist

FrenchPastry

CapitalCity

Whale

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## Individual names

- denote **particular entities** (**constants** in logic)

kim

csc415

univBordeaux

tramStopPeixotto

panda35

**Combine** concepts/classes and roles/properties **using constructors** to **build complex descriptions**

**Example:** courses taught by Meghyn which are attended by at least two students not from computer science and only by Master's or Phd students

$$\text{Course} \sqcap \exists \text{taughtBy}.\{\text{meghyn}\} \sqcap \geq 2 \text{attendedBy} . (\neg \text{CompSciStudent})$$
$$\forall \text{attendedBy} . (\text{MasterStudent} \sqcup \text{PhDStudent})$$

Next slides: introduce some of the **most common constructors**

**Notation:**  $C \sqcap D$  (C, D potentially complex concepts)

**Meaning:** class of entities that belong to **both C and D**

Female scientist

Female  $\sqcap$  Scientist

Spicy vegetarian dish

Spicy  $\sqcap$  VegetarianDish

Notation:  $C \sqcup D$

Meaning: class of entities that belong to **either  $C$  or  $D$  (possibly both)**

Cat or dog

$\text{Cat} \sqcup \text{Dog}$

Cake or cookie or muffin

$\text{Cake} \sqcup (\text{Cookie} \sqcup \text{Muffin})$



Notation:  $\neg C$

Meaning: class of entities that are **not in C**

Anything/anyone who is **not a parent**

$\neg \text{Parent}$

**Person who is not a parent**

$\text{Person} \sqcap (\neg \text{Parent})$

**Notation:**  $\exists r.C$  ( $r$  possibly complex role,  $C$  possibly complex concept)

**Meaning:** class of entities that **are related by  $r$  to an entity in  $C$**

Those who teach a doctoral course offered by a French university

$\exists \text{teaches} . (\text{DoctoralCourse} \sqcap \exists \text{ offeredBy} . \text{FrenchUniv})$

Those who teach (something)

$\exists \text{teaches} . \top$

Here  $\top$  designates the set of all things

**Notation:**  $\forall r.C$  ( $r$  possibly complex role,  $C$  possibly complex concept)

**Meaning:** class of entities that are **only related by  $r$  to entities from  $C$**

Those who teach only doctoral courses

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If we only want to include those who teach, need to specify this:

$(\forall \text{teaches}.\text{DoctoralCourse}) \sqcap (\exists \text{teaches}.\top)$

Notation:  $\geq kr.C, \leq kr.C$  (r role, C concept, k non-negative integer)

Meaning: class of entities that **are connected via r to at least / at most k elements of C**

Those having at least 3 adult children

$\geq 3\text{hasChild.Adult}$

Things that have at most 5 ingredients

$\leq 5\text{hasIngredient.T}$

Notation:  $\{a\}$

( $a$  an individual name)

Meaning: class consisting **solely of**  $a$

Canada, USA, or Mexico

$$\{\text{canada}\} \sqcup \{\text{usa}\} \sqcup \{\text{mexico}\}$$

Courses taught by Meghyn

$$\text{Course} \sqcap \exists \text{taughtBy}.\{\text{meghyn}\}$$



So far we've seen how to describe classes and binary relationships

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We can now use them to express different kinds of knowledge

Common to **distinguish between two kinds of knowledge**:

- **general domain knowledge** **TBox (ontology)**
  - finite set of **axioms** (details on next slides)
- **factual knowledge about particular individuals** **ABox (data)**
  - finite set of **assertions**  $C(a), r(a, b)$  ( $C$  concept,  $r$  role,  $a, b$  inds)

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**DL knowledge base (KB) = TBox (ontology) + ABox (data)**

Note: usage varies, word "ontology" is sometimes used for whole KB

Suppose  $C, D$  are (possibly complex) concepts

**(General) concept inclusion:**  $C \sqsubseteq D$

- every member of  $C$  is also a member of  $D$  (all  $C$ s are  $D$ s)

**Concept equivalence:**  $C \equiv D$

- $C$  and  $D$  describe precisely the same sets
- abbreviation for  $C \sqsubseteq D$  and  $D \sqsubseteq C$

Professors and lecturers are disjoint classes of faculty

$$\text{Prof} \sqsubseteq \text{Faculty} \quad \text{Lect} \sqsubseteq \text{Faculty} \quad \text{Prof} \sqsubseteq \neg \text{Lect}$$

Every course is either an undergrad or grad course

$$\text{Course} \equiv \text{UCourse} \sqcup \text{GCourse}$$

The relation takesCourse connects students to courses

$$\exists \text{takesCourse}.T \sqsubseteq \text{Student} \quad \exists \text{takesCourse}^-.T \sqsubseteq \text{Course}$$

Note: speak of **domain** (1st position) / **range** (2nd position) of roles

Every student takes **at least 2** and **at most 5 courses**

$$\text{Student} \sqsubseteq \geq 2 \text{takesCourse.T} \sqcap \leq 5 \text{takesCourse.T}$$

Every **grad student** is **supervised by some faculty member**

$$\text{GStudent} \sqsubseteq \exists \text{supervisedBy.Faculty}$$

**Students** who **only take grad-level courses** are **grad students**

$$\text{Student} \sqcap \forall \text{takesCourse.GCourse} \sqsubseteq \text{GStudent}$$

Axioms giving **relationship between  $r$  and  $s$**

- $r$  is **contained** in  $s$  (every pair in  $r$  also belongs to  $s$ )

$$r \sqsubseteq s \quad (\text{role inclusion})$$

- $r$  and  $s$  are **disjoint** (no pairs in common)

$$r \sqsubseteq \neg s$$

- $r$  corresponds to the **inverse** of  $s$

$$r \equiv s^{-}$$

**ParentOf** and **ChildOf** are the **inverses of one another**

$$\text{parentOf} \equiv \text{childOf}^{-}$$

The relation **parentOf** is included in **ancestorOf**

$$\text{parentOf} \sqsubseteq \text{ancestorOf}$$

The **friendOf** and **enemyOf** relations are **disjoint**

$$\text{friendOf} \sqsubseteq \neg \text{enemyOf}$$



Can also state that  $r$  is:

- **transitive**: if  $r(x, y)$  and  $r(y, z)$ , then  $r(x, z)$
- **functional**: if  $r(x, y)$  and  $r(x, z)$ , then  $y = z$
- **symmetric**: if  $r(x, y)$ , then  $r(y, x)$
- **reflexive**:  $r(x, x)$  (for any  $x$ )
- also: inverse functional, asymmetric, irreflexive

For example:

funct(hasBirthplace)

trans(locatedIn)

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- **concept constructors:**  $\perp, \top, \neg, \sqcup, \sqcap, \exists r.C, \forall r.C$
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The **highly expressive DL  $\mathcal{SHIQ}$**  is defined by:

- all  $\mathcal{ALC}$  concept constructors, **plus:**  $\geq n r.C, \leq n r.C$
- **inverse roles ( $r^-$ )**
- concept inclusions, **role inclusions**, and **transitivity** axioms

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So far we have introduced

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However, we need **semantics** to **give the symbols meaning**:

- do two concept expressions **designate the same set**?
- does a given axiom **logically follow** from a set of axioms?
- does our knowledge base contain **contradictory information**?

**DL semantics**: based upon **interpretations** (like first-order logic, FOL)

Interpretation  $\mathcal{I}$  takes the form of a pair  $(\Delta^{\mathcal{I}}, \cdot^{\mathcal{I}})$

- $\Delta^{\mathcal{I}}$  is a non-empty set
  - (called the interpretation domain or universe)
- $\cdot^{\mathcal{I}}$  is a function which maps
  - individual name  $a \mapsto$  an element of the universe  $a^{\mathcal{I}} \in \Delta^{\mathcal{I}}$
  - concept name  $A \mapsto$  unary relation  $A^{\mathcal{I}} \subseteq \Delta^{\mathcal{I}}$
  - role name  $r \mapsto$  binary relation  $r^{\mathcal{I}} \subseteq \Delta^{\mathcal{I}} \times \Delta^{\mathcal{I}}$

Each interpretation describes a particular state-of-affairs

Can think of interpretations as possible worlds



## EXAMPLE INTERPRETATION

Interpretation  $\mathcal{I} = (\Delta^{\mathcal{I}}, \cdot^{\mathcal{I}})$  where

$$\Delta^{\mathcal{I}} = \{e_1, e_2, \dots, e_{15}\}$$

$$\textit{Student}^{\mathcal{I}} = \{e_1, \dots, e_7\}$$

$$\textit{Musician}^{\mathcal{I}} = \{e_4, e_5, e_6, e_8, e_{10}, e_{13}\}$$

$$\textit{supervises}^{\mathcal{I}} = \{(e_5, e_2), (e_8, e_7), (e_{10}, e_4), (e_{10}, e_6), \\ (e_9, e_3), (e_9, e_4), (e_{12}, e_{15})\}$$

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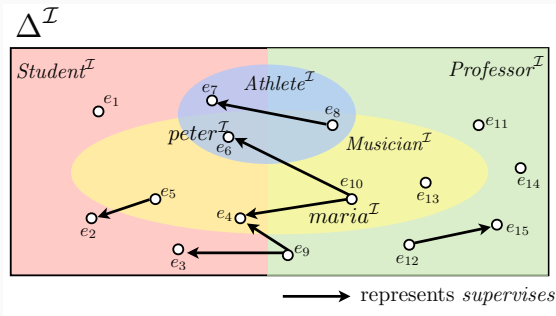
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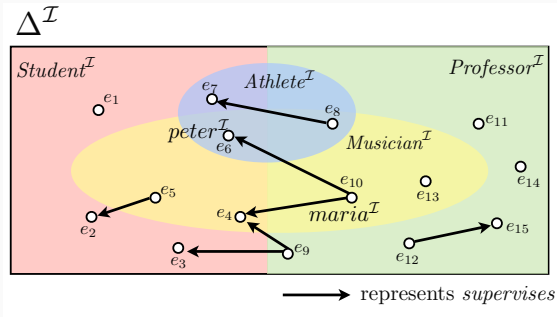
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- $(\exists r.C)^{\mathcal{I}} = \{u \mid \text{exists } v \text{ such that } (u, v) \in r^{\mathcal{I}} \text{ and } v \in C^{\mathcal{I}}\}$
- $(\forall r.C)^{\mathcal{I}} = \{u \mid \text{for every } v \text{ such that } (u, v) \in r^{\mathcal{I}} \text{ we have } v \in C^{\mathcal{I}}\}$
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- $\{a\}^{\mathcal{I}} = \{a^{\mathcal{I}}\}$

We next **extend the function  $\cdot^{\mathcal{I}}$  to complex concepts and roles**, to formalize the meaning of the constructors

- $\top^{\mathcal{I}} = \Delta^{\mathcal{I}}$  and  $\perp^{\mathcal{I}} = \emptyset$
- $(C \sqcap D)^{\mathcal{I}} = C^{\mathcal{I}} \cap D^{\mathcal{I}}$
- $(C \sqcup D)^{\mathcal{I}} = C^{\mathcal{I}} \cup D^{\mathcal{I}}$
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- $\{a\}^{\mathcal{I}} = \{a^{\mathcal{I}}\}$
- $(r^{-})^{\mathcal{I}} = \{(u, v) \mid (v, u) \in r^{\mathcal{I}}\}$

## EXAMPLE: SEMANTICS OF CONSTRUCTORS

Reconsider the interpretation  $\mathcal{I}$ :



For each of the following concepts, give the corresponding set in  $\mathcal{I}$ :

- |                                  |                                    |
|----------------------------------|------------------------------------|
| (1) $Athlete \sqcup Musician$    | (2) $Athlete \sqcap \neg Musician$ |
| (3) $\exists supervises.Student$ | (4) $\exists supervises^-.Student$ |
| (5) $\geq 2 supervises.\top$     | (6) $\forall supervises.Student$   |

## Satisfaction of axioms:

- $\mathcal{I}$  satisfies a concept inclusion  $C \sqsubseteq D$  if  $C^{\mathcal{I}} \subseteq D^{\mathcal{I}}$
- $\mathcal{I}$  satisfies a concept equivalence  $C \equiv D$  if  $C^{\mathcal{I}} = D^{\mathcal{I}}$
- $\mathcal{I}$  satisfies a role inclusion  $r \sqsubseteq s$  if  $r^{\mathcal{I}} \subseteq s^{\mathcal{I}}$
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## Satisfaction of ABox assertions:

- $\mathcal{I}$  satisfies an assertion  $C(a)$  if  $a^{\mathcal{I}} \in C^{\mathcal{I}}$
- $\mathcal{I}$  satisfies an assertion  $r(a, b)$  if  $(a^{\mathcal{I}}, b^{\mathcal{I}}) \in r^{\mathcal{I}}$

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**Important:** ABoxes are interpreted under **open-world assumption**

- provide **incomplete information**,  $\alpha \notin \mathcal{A}$  does not mean  $\alpha$  is false  
(might be able to infer it using axioms)

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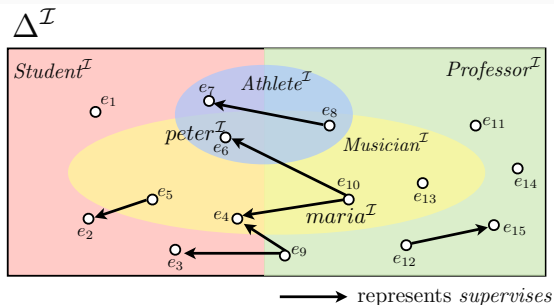
**Important:** ABoxes are interpreted under **open-world assumption**

- provide **incomplete information**,  $\alpha \notin \mathcal{A}$  does not mean  $\alpha$  is false (might be able to infer it using axioms)
- differs from **closed-world assumption for databases** (where **absent means false**)



# EXAMPLE: SATISFACTION OF AXIOMS & ASSERTIONS

Reconsider the interpretation  $\mathcal{I}$ :



Which of the following are satisfied in  $\mathcal{I}$ ?

- |  |  |
|--|--|
| (1) $Athlete \sqsubseteq Musician$                     | (2) $Student \sqsubseteq \neg Professor$             |
| (3) $\exists supervises.Athlete \sqsubseteq Professor$ | (4) $Professor \equiv \exists supervises.\top$       |
| (5) $\exists supervises^-. \top \sqsubseteq Student$   | (6) $Student \sqsubseteq \forall supervises.Student$ |
| (7) $Musician(peter)$                                  | (8) $(\exists supervises.Musician)(maria)$           |

## Models:

- $\mathcal{I}$  is a **model of a TBox**  $\mathcal{T}$  if  $\mathcal{I}$  satisfies every axiom in  $\mathcal{T}$
- $\mathcal{I}$  is a **model of an ABox**  $\mathcal{A}$  if  $\mathcal{I}$  satisfies every assertion in  $\mathcal{A}$
- $\mathcal{I}$  is a **model of a KB**  $(\mathcal{T}, \mathcal{A})$  if  $\mathcal{I}$  is a model of  $\mathcal{T}$  and  $\mathcal{A}$

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## Satisfiability:

- A **concept**  $C$  is **satisfiable w.r.t. TBox**  $\mathcal{T}$  if there exists a model  $\mathcal{I}$  of  $\mathcal{T}$  with  $C^{\mathcal{I}} \neq \emptyset$
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## Entailment:

- A TBox  $\mathcal{T}$  **entails an axiom**  $\alpha$  (written  $\mathcal{T} \models \alpha$ )  
if every model of  $\mathcal{T}$  satisfies  $\alpha$
- A KB  $\mathcal{K} = (\mathcal{T}, \mathcal{A})$  **entails an ABox assertion**  $\alpha$  (written  $\mathcal{K} \models \alpha$ )  
if every model of  $(\mathcal{T}, \mathcal{A})$  satisfies  $\alpha$

The axiom *RattleSnake*  $\sqsubseteq$  *Animal* is entailed from the following TBox:

*RattleSnake*  $\sqsubseteq$  *Reptile*

*Snake*  $\sqsubseteq$  *Reptile*

*Reptile*  $\sqsubseteq$  *Animal*

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The assertion *TeachingStaff*(*rami*) is entailed from the following KB:

*teaches*(*rami*, *phy305*)

$\exists teaches.T \sqsubseteq TeachingStaff$

## EXAMPLES: ENTAILMENT, SATISFIABILITY

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*Reptile*  $\sqsubseteq$  *Animal*

The assertion *TeachingStaff*(*rami*) is entailed from the following KB:

*teaches*(*rami*, *phy305*)

$\exists \text{teaches.T} \sqsubseteq \text{TeachingStaff}$

The following KB is unsatisfiable:

*Childless*(*tom*)

*hasChild*(*tom*, *fred*)

*Childless*  $\equiv \forall \text{hasChild}.\perp$

Suppose that the ontology  $\mathcal{T}$  contains the following axioms:

$$\text{Sheep} \sqsubseteq \text{Animal} \sqcap \forall \text{eats}.\text{Grass} \quad (1)$$

$$\text{Grass} \sqsubseteq \text{Plant} \quad (2)$$

$$\begin{aligned} \text{Vegetarian} \equiv \text{Animal} \sqcap (\forall \text{eats}.\neg \text{Animal}) \quad (3) \\ \sqcap (\forall \text{eats}.\neg (\exists \text{partOf}.\text{Animal})) \end{aligned}$$

$$\text{Animal} \sqcup \exists \text{partOf}.\text{Animal} \sqsubseteq \neg (\text{Plant} \sqcup \exists \text{partOf}.\text{Plant}) \quad (4)$$

Claim:  $\mathcal{T} \models \text{Sheep} \sqsubseteq \text{Vegetarian}$       **Why?**

(animal examples taken from:

<http://owl.man.ac.uk/2003/why/latest/>)



## EXAMPLE: UNSATISFIABLE CONCEPT

Now suppose  $\mathcal{T}$  contains the following axioms:

$$\textit{Sheep} \sqsubseteq \textit{Animal} \sqcap \forall \textit{eats}.\textit{Grass} \quad (1)$$

$$\textit{Cow} \sqsubseteq \textit{Vegetarian} \quad (2)$$

$$\textit{MadCow} \equiv \textit{Cow} \sqcap \exists \textit{eats} . (\textit{Brain} \sqcap \exists \textit{partOf} . \textit{Sheep}) \quad (3)$$

$$\begin{aligned} \textit{Vegetarian} \equiv \textit{Animal} \sqcap (\forall \textit{eats} . \neg \textit{Animal}) \quad (4) \\ \sqcap (\forall \textit{eats} . \neg (\exists \textit{partOf} . \textit{Animal})) \end{aligned}$$

$$\textit{Animal} \sqcup \exists \textit{partOf} . \textit{Animal} \sqsubseteq \neg (\textit{Plant} \sqcup \exists \textit{partOf} . \textit{Plant}) \quad (5)$$

Is *MadCow* is unsatisfiable w.r.t.  $\mathcal{T}$       **Why?**

### Concept satisfiability

- Input: (possibly complex) concept  $C$ , TBox  $\mathcal{T}$
- Task: **determine whether  $C$  is satisfiable w.r.t.  $\mathcal{T}$**
- Use: debugging ontologies

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## Classification

- Input: TBox  $\mathcal{T}$
- Task: **determine, for every pair of concept names  $A, B$  from  $\mathcal{T}$ , whether  $\mathcal{T} \models A \sqsubseteq B$**
- Use: visualizing / understanding ontology (also debugging)

### Knowledge base satisfiability

- Input: KB  $\mathcal{K} = (\mathcal{T}, \mathcal{A})$
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- Use: checking for contradictory information

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### Instance checking

- Input: KB  $\mathcal{K} = (\mathcal{T}, \mathcal{A})$ , concept  $C$
- Task: **find all individuals  $a$  such that  $\mathcal{K} \models C(a)$**
- Use: basic way to query the ABox

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### Ontology-mediated query answering (OMQA)

- Input: KB  $\mathcal{K} = (\mathcal{T}, \mathcal{A})$ , **database query  $q(\vec{x})$** 
  - typically,  $q$  is a conjunctive query (discussed later in course)
- Task: **determine the certain answer to  $q(\vec{x})$  w.r.t.  $\mathcal{K}$** , i.e. answers that **hold in every model of  $\mathcal{K}$**
- Use: database-style querying of the data

## COURSE PLAN

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Rest of today's session: **TD on DL basics**

Following sessions:

- **OWL, Protégé** (TP)
- **Reasoning with expressive DLs** (TD)
- **Reasoning with lightweight DLs** (TD) - two sessions
- **More with Protégé** (TP)

Project on ontologies - presented during Thursday's session