

Scalable Ontology-Based Information Systems

Ian Horrocks

<ian.horrocks@comlab.ox.ac.uk>

Information Systems Group

Oxford University Computing Laboratory





What is an Ontology?





What is an Ontology?

A model of (some aspect of) the world

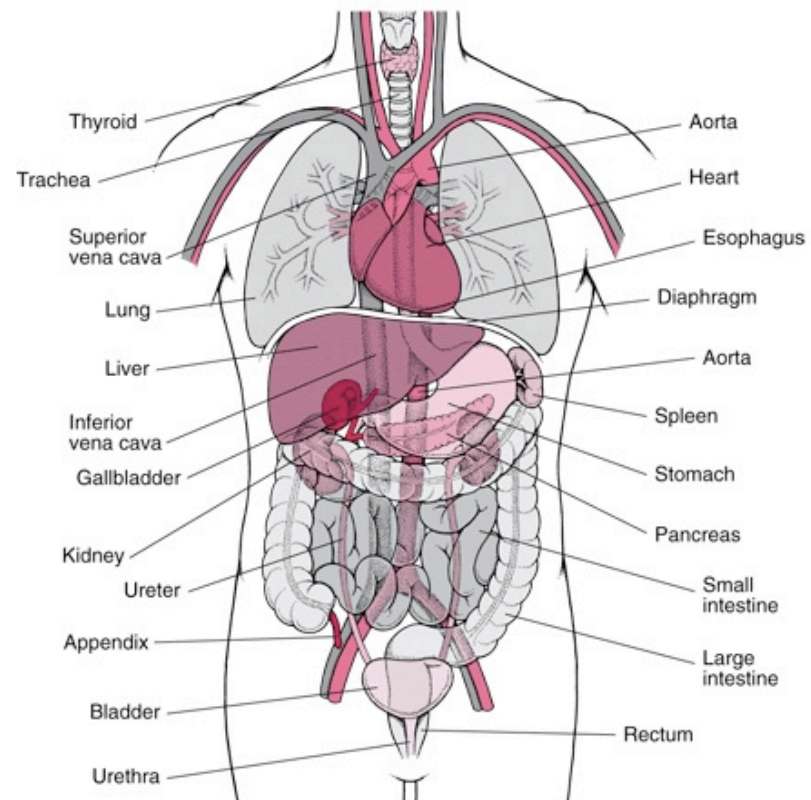




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- Introduces **vocabulary** relevant to domain, e.g.:
 - Anatomy

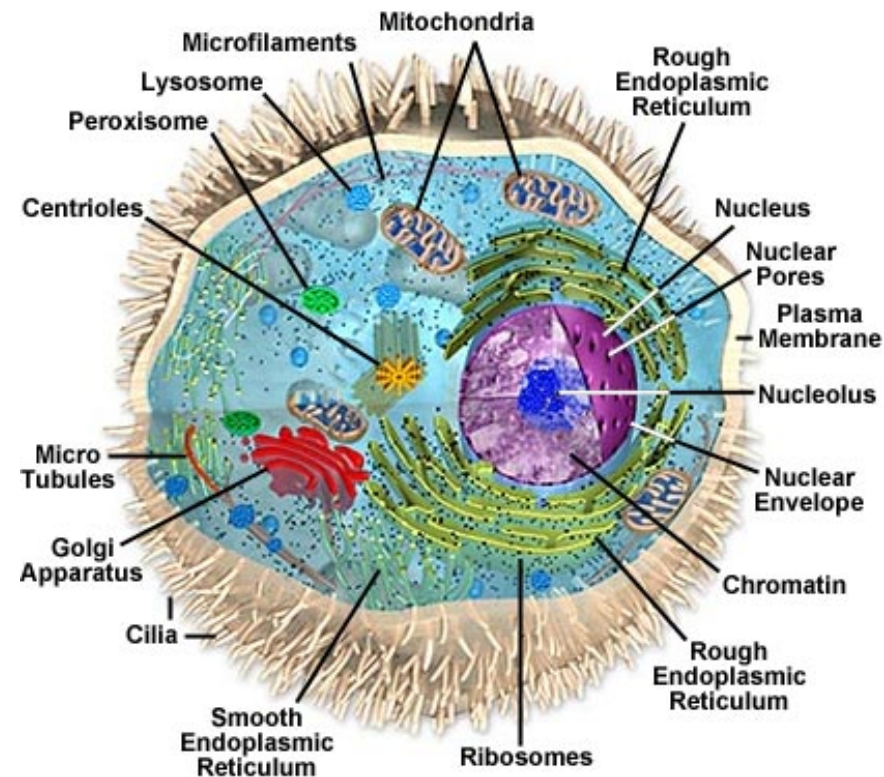




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 - Cellular biology

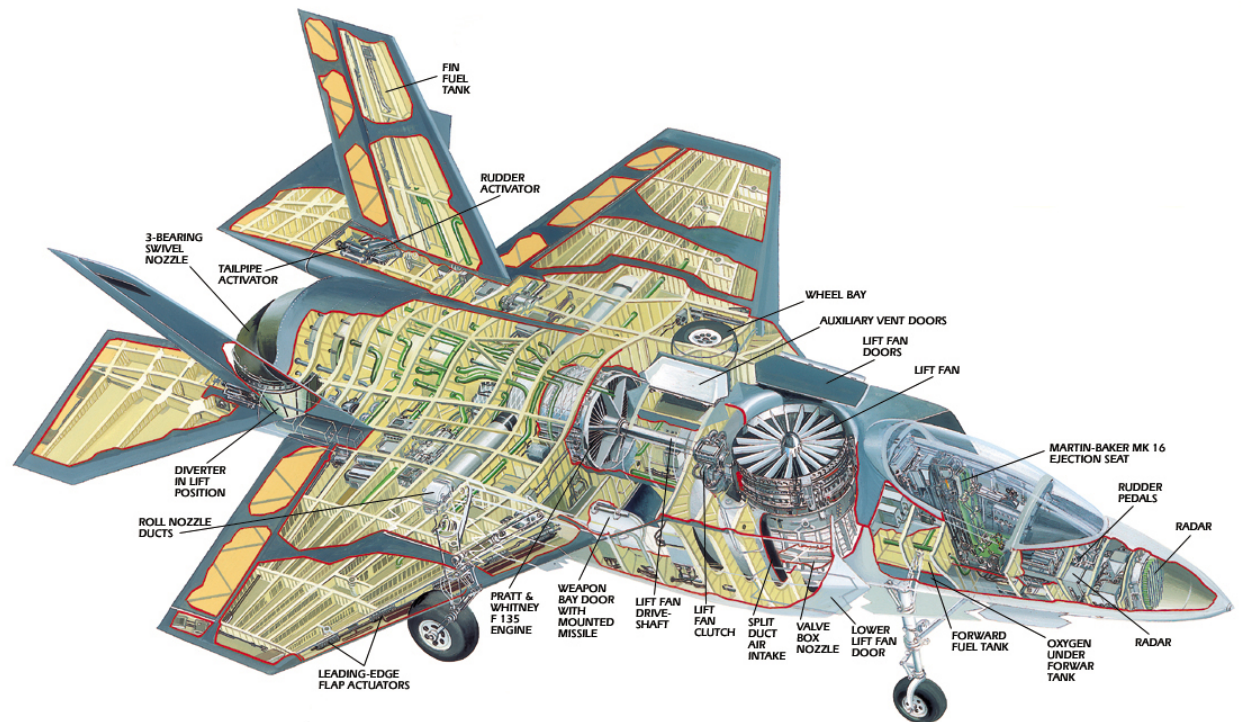




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 - Cellular biology
 - Aerospace

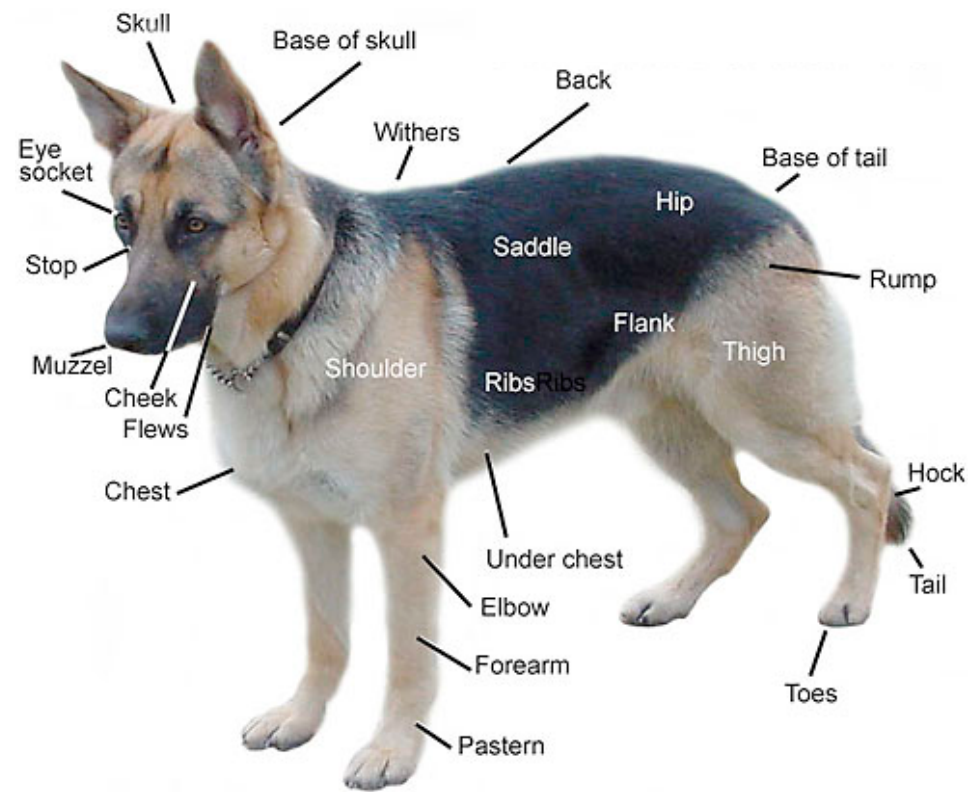




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- Introduces **vocabulary** relevant to domain, e.g.:

- Anatomy
- Cellular biology
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- Hotdogs
- ...



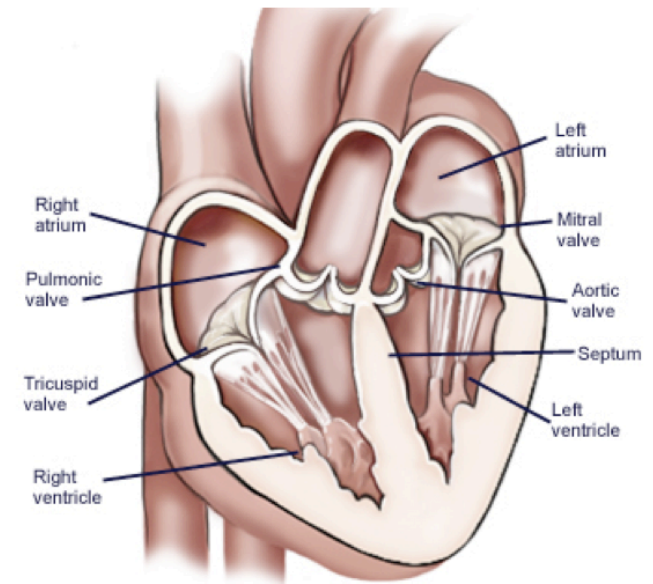


What is an Ontology?

A model of (some aspect of) the world

- Introduces **vocabulary** relevant to domain
- Specifies **meaning** (semantics) of terms

Heart **is** a muscular organ that **is part of** the circulatory system





What is an Ontology?

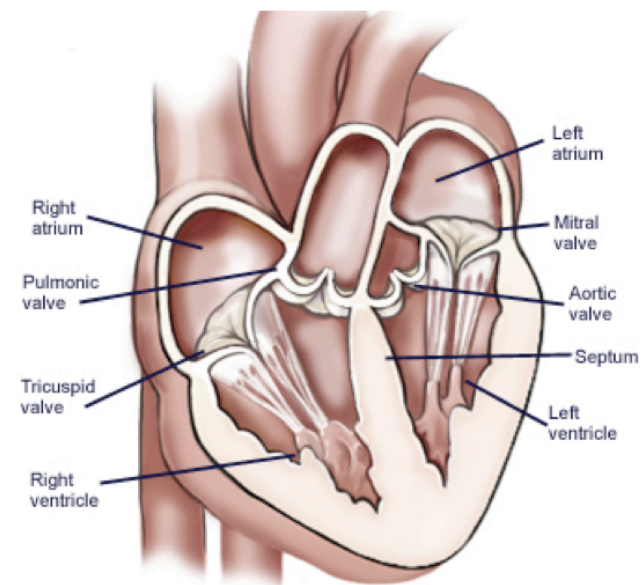
A model of (some aspect of) the world

- Introduces **vocabulary** relevant to domain
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Heart **is** a muscular organ that **is part of** the circulatory system

- **Formalised** using suitable logic

$$\forall x. [\text{Heart}(x) \rightarrow \text{MuscularOrgan}(x) \wedge \exists y. [\text{isPartOf}(x, y) \wedge \text{CirculatorySystem}(y)]]$$





Web Ontology Language OWL (2)

- **W3C** recommendation(s)
- Motivated by **Semantic Web** activity
 - Add meaning to web content by annotating it with terms defined in ontologies
- Supported by **tools and infrastructure**
 - APIs (e.g., OWL API, Thea, OWLink)
 - Development environments (e.g., Protégé, Swoop, TopBraid Composer, Neon)
 - Reasoners & Information Systems (e.g., Pellet, Racer, HermiT, Quonto, ...)
- Based on **Description Logics** (*SHOIN / SROIQ*)





Description Logics (DLs)

- Fragments of **first order logic** designed for KR
- Desirable computational properties
 - **Decidable** (essential)
 - Low complexity (desirable)
- Succinct and **variable free syntax**

$$\forall x. [\text{Heart}(x) \rightarrow \text{MuscularOrgan}(x) \wedge \\ \exists y. [\text{isPartOf}(x, y) \wedge \\ \text{CirculatorySystem}(y)]]$$

$$\text{Heart} \sqsubseteq \text{MuscularOrgan} \sqcap \\ \exists \text{isPartOf}. \text{CirculatorySystem}$$



Description Logics (DLs)

DL **Knowledge Base** (KB) consists of two parts:

- Ontology (aka **TBox**) axioms define terminology (schema)

$\text{Heart} \sqsubseteq \text{MuscularOrgan} \sqcap$
 $\exists \text{isPartOf}.\text{CirculatorySystem}$

$\text{HeartDisease} \equiv \text{Disease} \sqcap$
 $\exists \text{affects}.\text{Heart}$

$\text{VascularDisease} \equiv \text{Disease} \sqcap$
 $\exists \text{affects} . (\exists \text{isPartOf} . \text{CirculatorySystem})$

- Ground facts (aka **ABox**) use the terminology (data)

$\text{John} : \text{Patient} \sqcap$
 $\exists \text{suffersFrom} . \text{HeartDisease}$

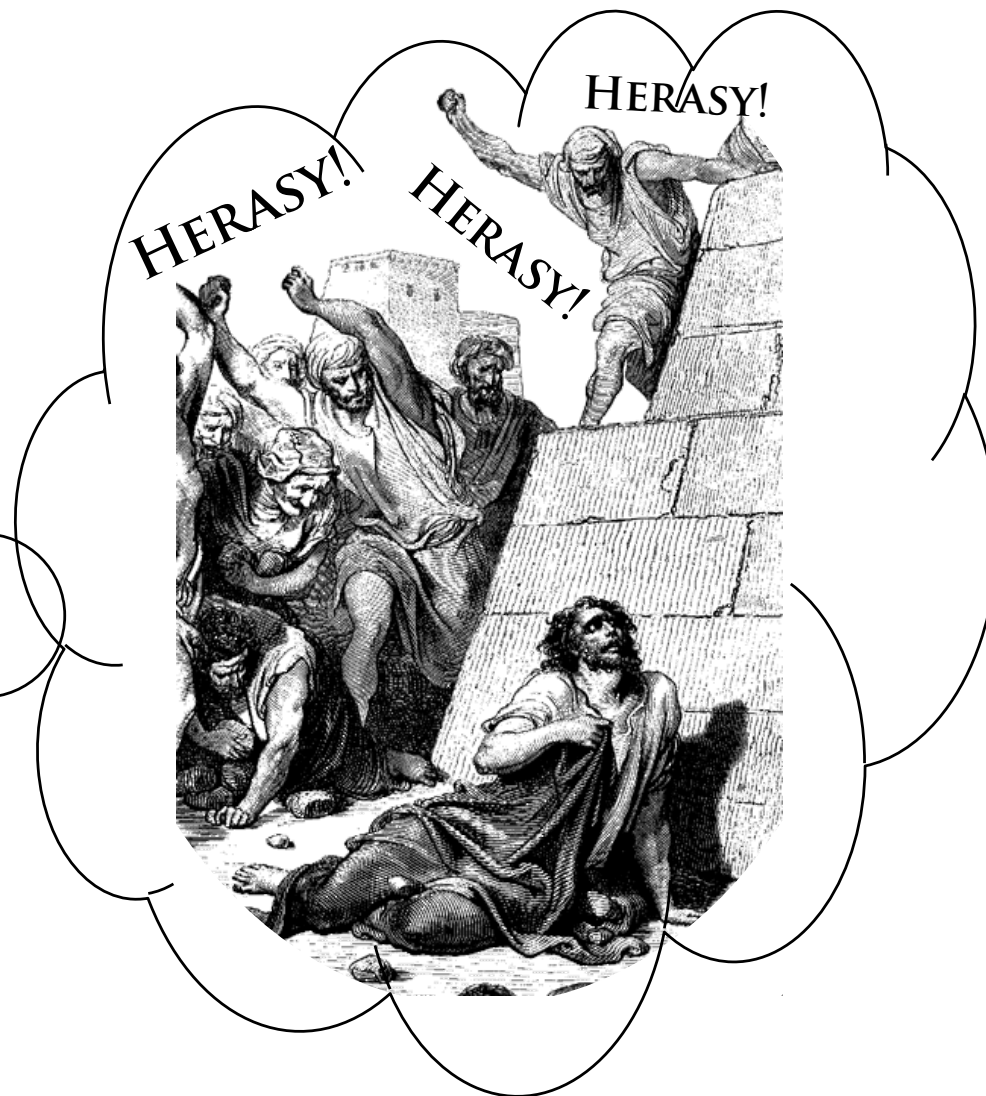


Why Care About Semantics?





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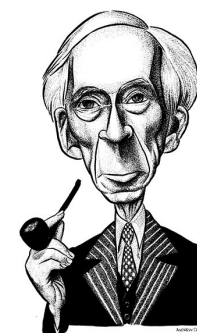
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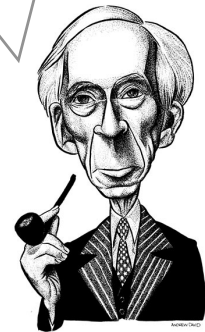




Why Care About Semantics?

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Well, from a philosophical POV, we need to specify the relationship between statements in the logic and the existential phenomena they describe.



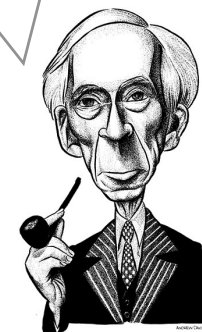


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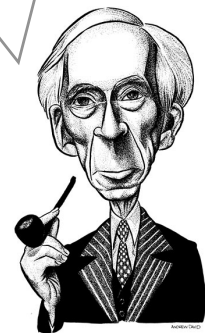
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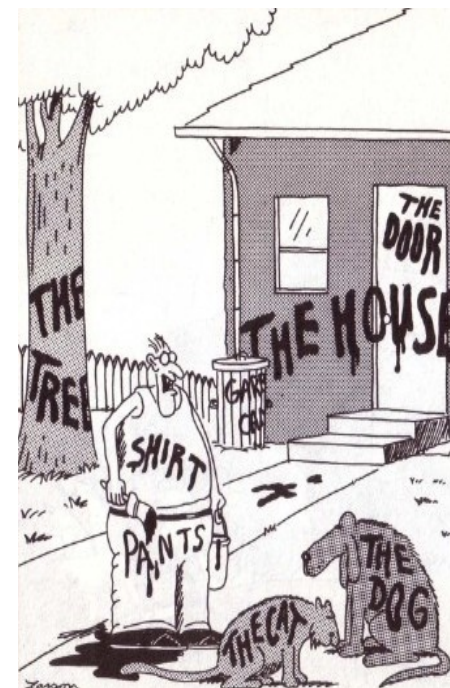
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From a practical POV, in order to specify and test (ontology-based) information systems we need to precisely define their intended behaviour



What are Ontologies Good For?

- Coherent **user-centric view** of domain
 - Help identify and resolve disagreements
- Ontology-based **Information Systems**
 - View of data that is independent of logical/physical schema
 - Answers reflect schema & data, e.g.:
“Patients suffering from Vascular Disease”



Now... *that* should clear up a few things around here





What are Ontologies Good For?

Heart \sqsubseteq MuscularOrgan \sqcap

\exists isPartOf.CirculatorySystem

HeartDisease \equiv Disease \sqcap

\exists affects.Heart

VascularDisease \equiv Disease \sqcap

\exists affects.(\exists isPartOf.CirculatorySystem)

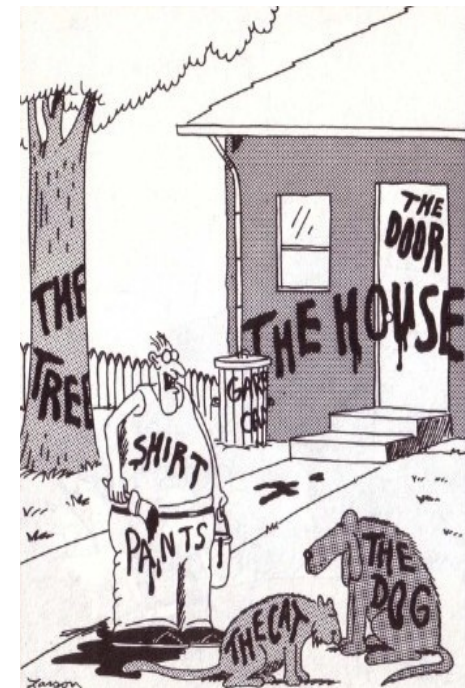
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 - View of data that is independent of logical/physical schema
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 - Query expansion/navigation/refinement
 - Incomplete and semi-structured data
 - Integration of heterogeneous sources



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Information-Based Decisions

Increasingly critical in many areas:

- In Healthcare industry
 - Too much screening harms patients and wastes money
 - Too little screening costs lives





Information-Based Decisions

Increasingly critical in many areas:

- In Oil and Gas industry
 - Better quality information could add €1B/year net value to Statoil production
 - Poorer quality information and analysis costs €6M/weekend!





Information-Based Decisions

Increasingly critical in many areas:

- In IT industry
 - SAP deals with 80,000 queries/month at a cost of approx. €16M
 - SAP estimate 50% of support staff time spent searching for relevant information





Information-Based Decisions

Increasingly critical in many areas:

- In Transport Security
 - Failures can cost hundreds of lives



“We had sufficient information, but failed to integrate and understand it”





Healthcare

- UK NHS **£10 billion** “Connecting for Health” IT programme
- Key component is **Care Records Service** (CRS)
 - “Live, interactive patient record service accessible 24/7”
 - Patient **data distributed** across local centres in 5 regional clusters, and a national DB
 - **SNOMED-CT** ontology provides common **vocabulary** for data
 - Clinical data uses terms drawn from ontology





SNOMED-CT

- It's **BIG** – over **400,000** concepts
- Language used is **EL** profile of **OWL 2**
- **Multiple hierarchies** and **rich definitions**





CliniClue 2006: SNOMED CT(International 0801int[Release]) [Registered user: phendler@hotmail.com]

File Edit Subsets Restrict Language Layout Tools Help

Concept Id 154283005 **TB - Pulmonary tuberculosis**

Description Id 1784750013 clinical finding

Words - any order

Find pulmonary tuber

- P pulmonary tuberculosis
- S TB - Pulmonary tuberculosis**
- P pulmonary tuberosc sclerosis
- S PTB - Pulmonary tuberculosis
- S inactive pulmonary tuberculosis

Hierarchy Subtype hierarchy

- C 205237003 pneumonitis
- C 56717001 tuberculosis
- C 84353005 pulmonary disease due to Mycobacteria
 - 154283005 **pulmonary tuberculosis**
 - C 428897002 inactive tuberculosis of lung
 - C 186175002 infiltrative lung tuberculosis
 - C 186188004 isolated tracheal or bronchial tuberculosis
 - C 77688003 isolated tracheal tuberculosis
 - C 80602006 nodular tuberculosis of lung
 - C 186192006 respiratory tuberculosis, bacteriologically and histologically confirmed
 - C 186202007 respiratory tuberculosis, not confirmed bacteriologically
 - C 186177005 tuberculosis of lung with cavitation
 - C 81554001 tuberculosis of lung with involvement of bronchus
 - C 186204008 tuberculosis of lung, bacteriologically and histologically confirmed
 - C 186194007 tuberculosis of lung, confirmed by culture only
 - C 186193001 tuberculosis of lung, confirmed by sputum microscopy
 - C 186195008 tuberculosis of lung, confirmed histologically
 - C 23022004 tuberculous bronchiectasis
 - C 90117007 tuberculous fibrosis of lung

pulmonary tuberculosis - Definition
Concept Status: **Current**

Descriptions

- F pulmonary tuberculosis (disorder)
- P pulmonary tuberculosis
- S PTB - Pulmonary tuberculosis
- S TB - Pulmonary tuberculosis

Definition: Fully defined by ...

is a

- D pneumonitis
- D inflammatory disorder of lower respiratory tract
- D disorder of lung
- D inflammation of specific body organs
- D tuberculosis
- D pulmonary disease due to Mycobacteria
- D infectious disease of lung
- D bacterial lower respiratory infection
- D mycobacteriosis

causative agent

- D Mycobacterium tuberculosis complex

Group

- associated morphology
- finding site
 - D lung structure

Qualifiers

- severity
 - p severities
- episodicity
 - p episodicities
- clinical course
 - p courses

Codes

Original SnomedId : R-F46B3

Pulmonary Tuberculosis

kind of pneumonitis

kind of tuberculosis

kind of Pulmonary disease due to Mycobacteria

found in lung structure



What About Scalability?

- Only **useful in practice** if we can deal with large ontologies and/or large data sets
- Unfortunately, many ontology languages are highly intractable
 - OWL 2 satisfiability is **2NEXPTIME-complete** w.r.t. schema
 - and **NP-Hard** w.r.t. data (upper bound open)
- Problem addressed in practice by
 - Algorithms that work well in **typical cases**
 - Highly **optimised implementations**
 - Use of tractable fragments (aka **profiles**)



Tableau Reasoning Algorithms





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Standard technique based on (hyper-) **tableau**

- Reasoning tasks reducible to (un)**satisfiability**
 - E.g., $KB \models \text{HeartDisease} \sqsubseteq \text{VascularDisease}$ iff
 $KB \cup \{x:(\text{HeartDisease} \sqcap \neg \text{VascularDisease})\}$ is *not* satisfiable



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 - E.g., $\text{KB} \models \text{HeartDisease} \sqsubseteq \text{VascularDisease}$ iff $\text{KB} \cup \{x:(\text{HeartDisease} \sqcap \neg\text{VascularDisease})\}$ is *not* satisfiable
- Algorithm tries to construct (an abstraction of) a model

$x : \text{HeartDisease} \sqcap \neg\text{VascularDisease}$	$x : \neg\text{VascularDisease}$
$x : \text{HeartDisease}$	$x : \neg\text{Disease} \sqcup$
$x : \text{Disease}$	$\neg\exists\text{affects} . (\exists\text{isPartOf} . \text{CirculatorySystem})$
$x : \exists\text{affects} . \text{Heart}$	$x : \neg\exists\text{affects} . (\exists\text{isPartOf} . \text{CirculatorySystem})$
$(x, y) : \text{affects}$	$x : \forall\text{affects} . (\forall\text{isPartOf} . \neg\text{CirculatorySystem})$
$y : \text{Heart}$	$y : \forall\text{isPartOf} . \neg\text{CirculatorySystem}$
$y : \text{MuscularOrgan}$	$z : \neg\text{CirculatorySystem}$
$y : \exists\text{isPartOf} . \text{CirculatorySystem}$	
$(y, z) : \text{isPartOf}$	
$z : \text{CirculatorySystem}$	



Tableau Reasoning Algorithms

Standard technique based on (hyper-) **tableau**

- Reasoning tasks reducible to (un)**satisfiability**
 - E.g., $\text{KB} \models \text{HeartDisease} \sqsubseteq \text{VascularDisease}$ iff $\text{KB} \cup \{x:(\text{HeartDisease} \sqcap \neg\text{VascularDisease})\}$ is *not* satisfiable
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$x : \text{HeartDisease} \sqcap \neg\text{VascularDisease}$	$x : \neg\text{VascularDisease}$
$x : \text{HeartDisease}$	$x : \neg\text{Disease} \sqcup$
$x : \text{Disease}$	$\neg\exists\text{affects} . (\exists\text{isPartOf} . \text{CirculatorySystem})$
$x : \exists\text{affects} . \text{Heart}$	$x : \neg\exists\text{affects} . (\exists\text{isPartOf} . \text{CirculatorySystem})$
$(x, y) : \text{affects}$	$x : \forall\text{affects} . (\forall\text{isPartOf} . \neg\text{CirculatorySystem})$
$y : \text{Heart}$	$y : \forall\text{isPartOf} . \neg\text{CirculatorySystem}$
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$z : \text{CirculatorySystem}$	



Highly Optimised Implementations

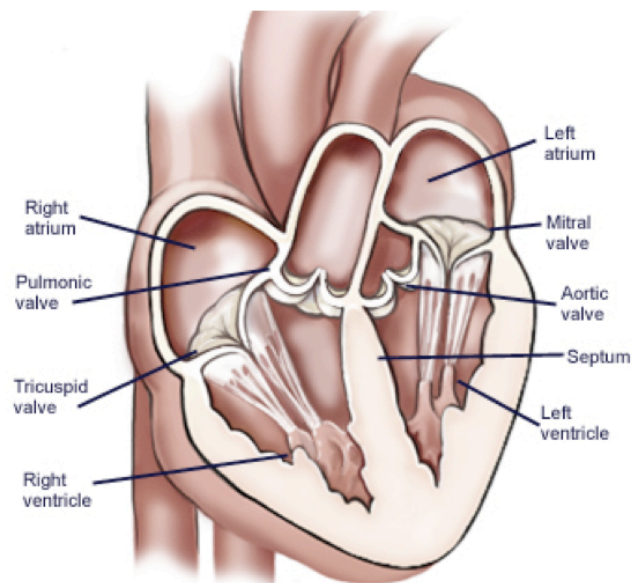
- Lazy unfolding
- Simplification and rewriting,
e.g., $A \sqcap B \sqsubseteq C \longrightarrow A \sqsubseteq C \sqcup \neg B$
- HyperTableau (reduces non-determinism)
- Fast semi-decision procedures
- Search optimisations
- Reuse of previous computations
- Heuristics

**Not computationally optimal,
but effective with many realistic ontologies**



Scalability Issues

- Problems with very **large and/or cyclical ontologies**



LeftSide $\sqsubseteq \exists$ hasComponent.AorticValve
 LeftSide $\sqsubseteq \exists$ hasComponent.MitralValve
 AorticValve $\sqsubseteq \exists$ hasConnection.LeftVentricle
 MitralValve $\sqsubseteq \exists$ hasConnection.LeftVentricle
 LeftVentricle $\sqsubseteq \exists$ isDivisionOf.LeftSide

- Ontologies may define 10s/100s of thousands of terms
- Can lead to construction of *very* large models



Scalability Issues

- Problems with **large data sets** (ABoxes)
 - Main reasoning problem is (conjunctive) query answering, e.g., retrieve all patients suffering from vascular disease:
 $Q(x) \leftarrow \text{Patient}(x) \wedge \text{suffersFrom}(x, y) \wedge \text{VascularDisease}(y)$
 - Decidability still open for OWL, although minor restrictions (on cycles in non-distinguished variables) restore decidability
 - Query answering reduced to standard decision problem, e.g., by checking for each individual x if $\text{KB} \models Q(x)$
 - Model construction starts with *all* ground facts (data)
- Typical applications may use data sets with **10s/100s of millions** of individuals (or more)



OWL 2 Profiles

- OWL recommendation now updated to **OWL 2**
- OWL 2 defines several **profiles** – fragments with desirable computational properties
 - **OWL 2 EL** targeted at very large ontologies
 - **OWL 2 QL** targeted at very large data sets



OWL 2 EL

- A (near maximal) fragment of OWL 2 such that
 - Satisfiability checking is in PTime (**PTime-Complete**)
 - Data complexity of query answering also PTime-Complete
- Based on \mathcal{EL} family of description logics
- Can exploit **saturation** based reasoning techniques
 - Computes classification in “one pass”
 - Computationally optimal
 - Can be extended to Horn fragment of OWL DL



Saturation-based Technique (basics)

- Normalise ontology axioms to standard form:

$$A \sqsubseteq B \quad A \sqcap B \sqsubseteq C \quad A \sqsubseteq \exists R.B \quad \exists R.B \sqsubseteq C$$

- Saturate using inference rules:

$$\frac{A \sqsubseteq B \quad B \sqsubseteq C}{A \sqsubseteq C} \quad \frac{A \sqsubseteq B \quad A \sqsubseteq C \quad B \sqcap C \sqsubseteq D}{A \sqsubseteq D}$$

$$\frac{A \sqsubseteq \exists R.B \quad B \sqsubseteq C \quad \exists R.C \sqsubseteq D}{A \sqsubseteq D}$$

- Extension to Horn fragment requires (many) more rules



Saturation-based Technique (basics)

Example:

OrganTransplant \equiv Transplant $\sqcap \exists$ site.Organ

HeartTransplant \equiv Transplant $\sqcap \exists$ site.Heart

Heart \sqsubseteq Organ





Saturation-based Technique (basics)

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Saturation-based Technique (basics)

Example:

OrganTransplant \equiv Transplant \sqcap \exists site.Organ

HeartTransplant \equiv Transplant \sqcap \exists site.Heart

Heart \sqsubseteq Organ

OrganTransplant \sqsubseteq Transplant

OrganTransplant \sqsubseteq \exists site.Organ





Saturation-based Technique (basics)

Example:

$\text{OrganTransplant} \equiv \text{Transplant} \sqcap \exists \text{site}.\text{Organ}$

$\text{HeartTransplant} \equiv \text{Transplant} \sqcap \exists \text{site}.\text{Heart}$

$\text{Heart} \sqsubseteq \text{Organ}$

$\text{OrganTransplant} \sqsubseteq \text{Transplant}$

$\text{OrganTransplant} \sqsubseteq \exists \text{site}.\text{Organ}$

$\exists \text{site}.\text{Organ} \sqsubseteq \text{SO}$

$\text{Transplant} \sqcap \text{SO} \sqsubseteq \text{OrganTransplant}$





Saturation-based Technique (basics)

Example:

OrganTransplant \equiv Transplant \sqcap \exists site.Organ

HeartTransplant \equiv Transplant \sqcap \exists site.Heart

Heart \sqsubseteq Organ

OrganTransplant \sqsubseteq Transplant

OrganTransplant \sqsubseteq \exists site.Organ

\exists site.Organ \sqsubseteq SO

Transplant \sqcap SO \sqsubseteq OrganTransplant





Saturation-based Technique (basics)

Example:

OrganTransplant \equiv Transplant \sqcap \exists site.Organ

HeartTransplant \equiv Transplant \sqcap \exists site.Heart

Heart \sqsubseteq Organ

OrganTransplant \sqsubseteq Transplant

OrganTransplant \sqsubseteq \exists site.Organ

\exists site.Organ \sqsubseteq SO

Transplant \sqcap SO \sqsubseteq OrganTransplant

HeartTransplant \sqsubseteq Transplant

HeartTransplant \sqsubseteq \exists site.Heart

\exists site.Heart \sqsubseteq SH

Transplant \sqcap SH \sqsubseteq HeartTransplant



Saturation-based Technique (basics)

Example:

OrganTransplant \equiv Transplant \sqcap \exists site.Organ

HeartTransplant \equiv Transplant \sqcap \exists site.Heart

Heart \sqsubseteq Organ

OrganTransplant \sqsubseteq Transplant

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Transplant \sqcap SO \sqsubseteq OrganTransplant

HeartTransplant \sqsubseteq Transplant

HeartTransplant \sqsubseteq \exists site.Heart

\exists site.Heart \sqsubseteq SH

Transplant \sqcap SH \sqsubseteq HeartTransplant

Heart \sqsubseteq Organ



Saturation-based Technique (basics)

Example:

OrganTransplant \equiv Transplant \sqcap \exists site.Organ
HeartTransplant \equiv Transplant \sqcap \exists site.Heart
Heart \sqsubseteq Organ

$$\frac{A \sqsubseteq \exists R.B \quad B \sqsubseteq C \quad \exists R.C \sqsubseteq D}{A \sqsubseteq D}$$

OrganTransplant \sqsubseteq Transplant
OrganTransplant \sqsubseteq \exists site.Organ
 \exists site.Organ \sqsubseteq SO
Transplant \sqcap SO \sqsubseteq OrganTransplant
HeartTransplant \sqsubseteq Transplant
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$\text{HeartTransplant} \sqsubseteq \text{SO}$
 $\text{HeartTransplant} \sqsubseteq \text{OrganTransplant}$



Saturation-based Technique

Performance with large bio-medical ontologies:

	GO	NCI	Galen v.0	Galen v.7	SNOMED
Concepts:	20465	27652	2748	23136	389472
FACT++	15.24	6.05	465.35	—	650.37
HERMIT	199.52	169.47	45.72	—	—
PELLET	72.02	26.47	—	—	—
CEL	1.84	5.76	—	—	1185.70
CB	1.17	3.57	0.32	9.58	49.44
Speed-Up:	1.57X	1.61X	143X	∞	13.15X



OWL 2 QL

- A (near maximal) fragment of OWL 2 such that
 - Data complexity of conjunctive query answering in **AC⁰**
- Based on **DL-Lite** family of description logics
- Can exploit **query rewriting** based reasoning technique
 - Computationally optimal
 - Data storage and query evaluation can be delegated to standard RDBMS
 - Can be extended to more expressive languages (beyond AC⁰) by delegating query answering to a Datalog engine



Query Rewriting Technique (basics)

- Given ontology \mathcal{O} and query Q , use \mathcal{O} to rewrite Q as Q' s.t., for any set of ground facts \mathcal{A} :
 - $\text{ans}(Q, \mathcal{O}, \mathcal{A}) = \text{ans}(Q', \emptyset, \mathcal{A})$



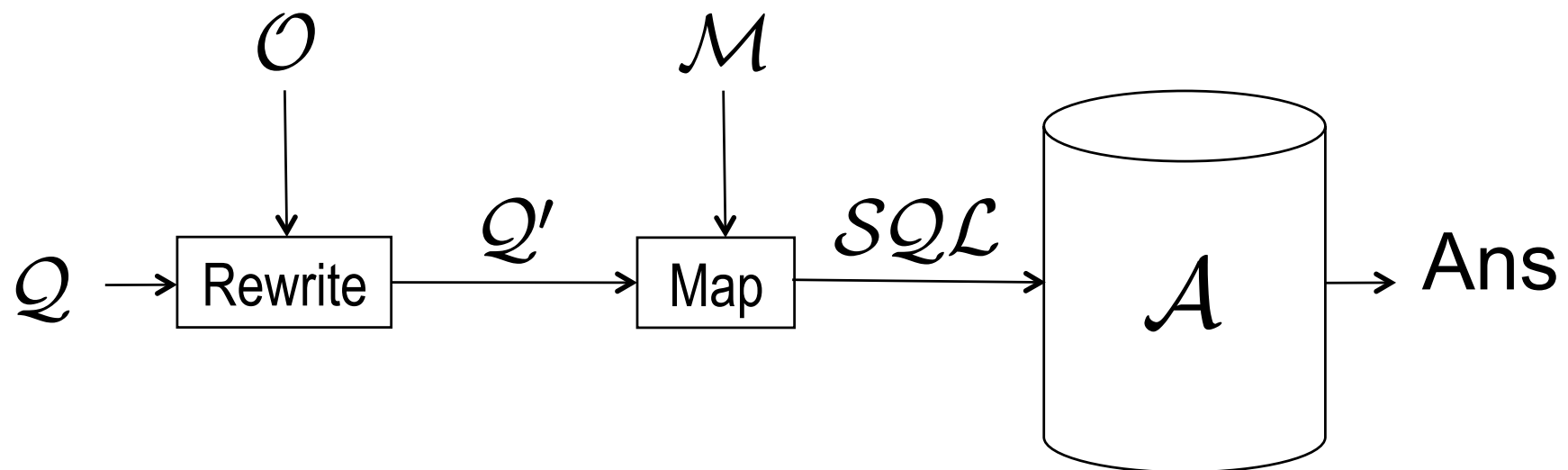
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 - $\text{ans}(Q, \mathcal{O}, \mathcal{A}) = \text{ans}(Q', \emptyset, \mathcal{A})$
- Use (GAV) mapping \mathcal{M} to map Q' to SQL query
- Resolution based query rewriting
 - **Clausify** ontology axioms
 - **Saturate** (clausified) ontology and query using resolution
 - **Prune** redundant query clauses



Query Rewriting Technique (basics)

- Example:

Doctor \sqsubseteq \exists treats. Patient

Consultant \sqsubseteq Doctor

$$Q(x) \leftarrow \text{treats}(x, y) \wedge \text{Patient}(y)$$



Query Rewriting Technique (basics)

- Example:

Doctor $\sqsubseteq \exists \text{treats.Patient}$

Consultant \sqsubseteq Doctor

$\text{treats}(x, f(x)) \leftarrow \text{Doctor}(x)$

$\text{Patient}(f(x)) \leftarrow \text{Doctor}(x)$

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$Q(x) \leftarrow \text{Doctor}(x)$

$Q(x) \leftarrow \text{Consultant}(x)$

- For DL-Lite, result is a union of conjunctive queries

$Q(x) \leftarrow (\text{treats}(x, y) \wedge \text{Patient}(y)) \vee \text{Doctor}(x) \vee \text{Consultant}(x)$





Query Rewriting Technique (basics)

- Data can be stored/left in **RDBMS**
- Relationship between ontology and DB defined by **mappings**, e.g.:

Doctor \mapsto SELECT Name FROM Doctor

Patient \mapsto SELECT Name FROM Patient

treats \mapsto SELECT DName, PName FROM Treats



Query Rewriting Technique (basics)

- Data can be stored/left in **RDBMS**
- Relationship between ontology and DB defined by **mappings**, e.g.:

Doctor \mapsto SELECT Name FROM Doctor

Patient \mapsto SELECT Name FROM Patient

treats \mapsto SELECT DName, PName FROM Treats

- UCQ translated into **SQL query**:

$$Q(x) \leftarrow (\text{treats}(x, y) \wedge \text{Patient}(y)) \vee \text{Doctor}(x) \vee \text{Consultant}(x)$$

\Downarrow

SELECT Name FROM Doctor UNION

SELECT DName FROM Treats, Patient WHERE PName=Name



Problems & Research Challenges

- Combining best features of DLs & DBs
 - In particular, integrating OWA and CWA
- Hard to find a coherent semantic framework
 - Problems mainly due to existential quantifiers: should existentially implied objects be considered different?
 - Does a person owning a phone and an ipod own 2 things?
 - Does a person owning a phone and an iphone own 2 things?
 - Does a person owning a phone and a phone own 2 things?
- Interesting ideas emerging in DL & DB communities, e.g.:
 - *Calì et al. Datalog \pm : a unified approach to ontologies and integrity constraints. ICDT 2009.*
 - *Motik et al. Bridging the gap between OWL and relational databases. WWW 2007.*



Problems & Research Challenges

- Open questions w.r.t. query rewriting





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 - Currently only for very weak ontology languages





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 - Even for these languages, queries can get very large (order $(|\mathcal{O}| \cdot |Q|)^{|\mathcal{Q}|}$), and existing RDBMSs may behave poorly
 - Not clear if this will be a problem in practice, see, e.g., *Savo et al. MASTRO at Work: Experiences on Ontology-based Data Access. DL 2010.*



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 - Not clear if this will be a problem in practice, see, e.g., *Savo et al. MASTRO at Work: Experiences on Ontology-based Data Access. DL 2010.*
 - Larger fragments require (at least) Datalog engines and/or extension to technique (e.g., partial materialisation)
 - Promising new work in this area, see, e.g., *Lutz et al. Conjunctive Query Answering in the Description Logic EL Using a Relational Database System. IJCAI 2009.*



Problems & Research Challenges

- Infrastructure





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 - Standardised query language
 - SPARQL standard for RDF
 - Currently being extended for OWL, see http://www.w3.org/2009/sparql/wiki/Main_Page





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 - Currently being extended for OWL, see http://www.w3.org/2009/sparql/wiki/Main_Page
 - Privacy and information hiding
 - May want to keep parts of data/schema private
 - Difficulties compounded when information can be inferred, see, e.g., *Cuenca Grau et al. Privacy-preserving query answering in logic-based information systems. ECAI 2008.*





Problems & Research Challenges

- Infrastructure
 - Standardised query language
 - SPARQL standard for RDF
 - Currently being extended for OWL, see http://www.w3.org/2009/sparql/wiki/Main_Page
 - Privacy and information hiding
 - May want to keep parts of data/schema private
 - Difficulties compounded when information can be inferred, see, e.g., *Cuenca Grau et al. Privacy-preserving query answering in logic-based information systems. ECAI 2008.*
 - ...



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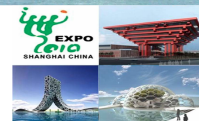
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Thanks To

- Boris Motik
- Yevgeny Kazakov
- Héctor Pérez-Urbina
- Rob Shearer
- Bernardo Cuenca Grau
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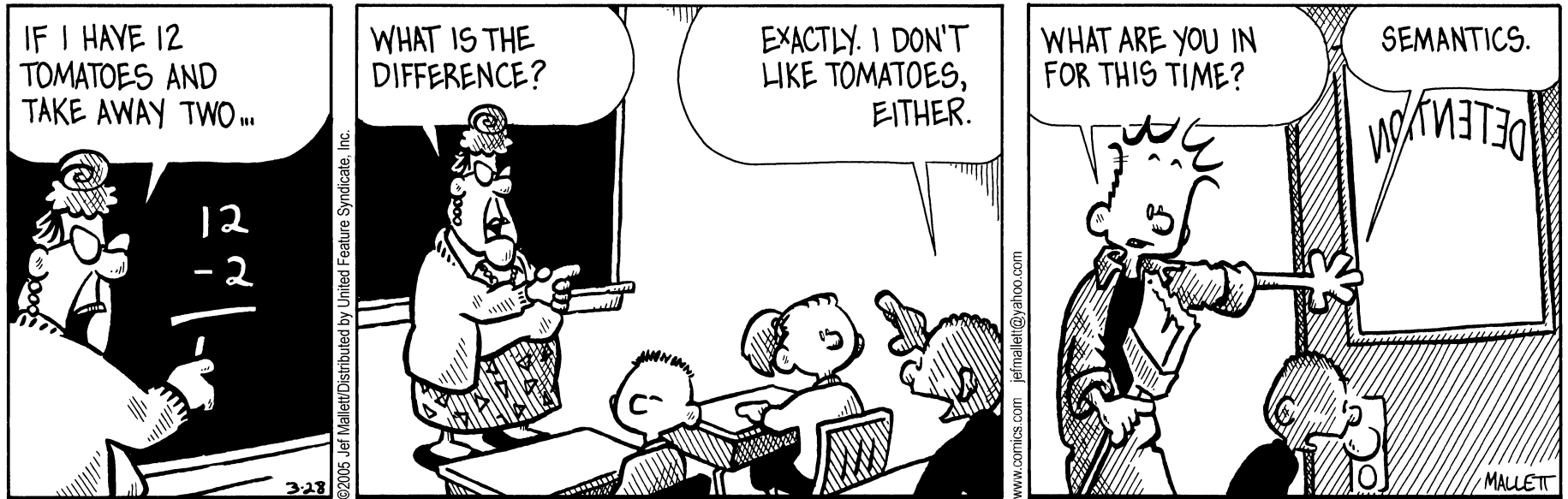


Thank you for listening





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Any questions?





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