#### **Ontology Languages and Engineering**

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#### An explicit specification of a conceptualization







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





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  - Cellular biology





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  - Photography





- Introduces **vocabulary** relevant to domain, e.g.:
  - Anatomy
  - Cellular biology
  - Aerospace
  - Photography
  - Pizzas





A model of (some aspect of) the world

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

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



A model of (some aspect of) the world

- Introduces vocabulary
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Heart is a muscular organ that is part of the circulatory system

• Formalised e.g. using suitable logic

 $\begin{array}{l} \mathsf{Heart}\sqsubseteq\mathsf{MuscularOrgan}\sqcap\\ \exists \mathsf{isPartOf}.\mathsf{CirculatorySystem} \end{array}$ 





- Coherent **shared view** of domain
  - Help identify and resolve disagreements
- Ontology-based Information Systems
  - User-centric view of data that is independent of logical/physical schema
  - Answers reflect knowledge & data, e.g.:



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



- $Q(x) \gets \mathsf{Patient}(x) \land \mathsf{suffersFrom}(x,y) \land \mathsf{VascularDisease}(y)$
- i.e., "Patients suffering from Vascular Disease"



 $Q(x) \leftarrow \mathsf{Patient}(x) \land \mathsf{suffersFrom}(x,y) \land \mathsf{VascularDisease}(y)$ 

i.e., "Patients suffering from Vascular Disease"

John : Patient □ ∃suffersFrom.HeartDisease



 $Q(x) \gets \mathsf{Patient}(x) \land \mathsf{suffersFrom}(x,y) \land \mathsf{VascularDisease}(y)$ 

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  - Answers reflect knowledge & data, e.g.:
    "Patients suffering from Vascular Disease"
  - Query expansion/navigation/refinement
  - Incomplete and semi-structured data

#### More "intelligent" applications



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• According to **TBL** circa 1998:

"... a consistent logical web of data ..." in which

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  - RDF provides uniform syntactic structure for data
  - OWL provides machine readable schemas (ontologies)

i.e., a large distributed ontology based information system

- RDF standard first published 1999; revised 2004
- RDF extended to **RDFS**, a primitive ontology language
  - classes and properties; sub/super-classes (and properties); range and domain (of properties)
- But RDFS lacks important features, e.g.:
  - existence/cardinality constraints; transitive/inverse properties;
     localised range and domain constraints, ...
- And RDF(S) has "higher order flavour" with no (later non-standard) formal semantics
  - difficult to understand or to provide reasoning support



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- Efforts soon merged to produce DAML+OIL
  - Further development carried out by "Joint EU/US Committee"





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  - Further development carried out by "Joint EU/US Committee"
- DAML+OIL submitted to WSC as basis for standardisation
  - WebOnt WG developed OWL (2004)
  - OWL WG developed OWL 2 (2009)
- OWL (2) based on SHOIN (SROIQ)
   Description Logics!?

# What are Description Logics (DLs)?

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

 $\begin{array}{l} \mathsf{Heart}\sqsubseteq\mathsf{MuscularOrgan}\sqcap\\ \exists \mathsf{isPartOf}.\mathsf{CirculatorySystem} \end{array}$ 

 $\begin{aligned} \forall x. [\mathsf{Heart}(x) & \to \mathsf{MuscularOrgan}(x) \land \\ & \exists y. [\mathsf{isPartOf}(x, y) \land \\ & \mathsf{CirculatorySystem}(y)]] \end{aligned}$ 

#### Can exploit the results of 20+ years of DL research

- Well defined (model theoretic) semantics

Constructor	DL Syntax	Example	FOL Syntax
intersectionOf	$C_1 \sqcap \ldots \sqcap C_n$	Human ⊓ Male	$C_1(x) \wedge \ldots \wedge C_n(x)$
unionOf	$C_1 \sqcup \ldots \sqcup C_n$	Doctor ⊔ Lawyer	$C_1(x) \lor \ldots \lor C_n(x)$
complementOf	$\neg C$	¬Male	$\neg C(x)$
oneOf	$\{x_1\}\sqcup\ldots\sqcup\{x_n\}$	{john} ⊔ {mary}	$x = x_1 \lor \ldots \lor x = x_n$
allValuesFrom	$\forall P.C$	∀hasChild.Doctor	$\forall y. P(x, y) \to C(y)$
someValuesFrom	$\exists P.C$	∃hasChild.Lawyer	$\exists y. P(x, y) \land C(y)$
maxCardinality	$\leqslant nP$	≤1hasChild	$\exists^{\leqslant n}y.P(x,y)$
minCardinality	$\geqslant nP$	≥2hasChild	$\mid \exists^{\geqslant n} y. P(x, y)$





Can exploit the results of 20+ years of DL research

- Well defined (model theoretic) semantics
- Formal properties well understood (complexity, decidability)



I can't find an efficient algorithm, but neither can all these famous people.

[Garey & Johnson. Computers and Intractability]

Can exploit the results of 20+ years of DL research

- Well defined (model theoretic) semantics
- Formal properties well understood (complexity, decidability)
- Practical reasoning algorithms

□-rule	1. $(C_1 \sqcap C_2) \in \mathcal{L}(v), v \text{ is not inc}$	directly blocked, and
	2. $\{C_1, C_2\} \not\subseteq \mathcal{L}(v)$	
	nen $\mathcal{L}(v) \to \mathcal{L}(v) \cup \{C_1, C_2\}.$	
⊔-rule	E1. $(C_1 \sqcup C_2) \in \mathcal{L}(v), v \text{ is not inc}$	lirectly blocked, and
	2. $\{C_1, C_2\} \cap \mathcal{L}(v) = \emptyset$	-
	then $\mathcal{L}(v) \to \mathcal{L}(v) \cup \{E\}$ for some	$E \in \{C_1, C_2\}$
∃-rule	1. $\exists r. C \in \mathcal{L}(v_1), v_1 \text{ is not block}$	ed, and
	2. $v_1$ has no safe <i>r</i> -neighbour $v_2$	with $C \in \mathcal{L}(v_1)$ ,
	nen create a new node $v_2$ and an e	dge $\langle v_1, v_2 \rangle$
	with $\mathcal{L}(v_2) = \{C\}$ and $\mathcal{L}(\langle v_1, \rangle)$	$v_2\rangle) = \{r\}.$
∀-rule	1. $\forall r.C \in \mathcal{L}(v_1), v_1 \text{ is not indire}$	ctly blocked, and
	2. there is an <i>r</i> -neighbour $v_2$ of $v$	$v_1$ with $C \notin \mathcal{L}(v_2)$
	nen $\mathcal{L}(v_2) \to \mathcal{L}(v_2) \cup \{C\}.$	
∀ <sub>+</sub> -rule	1. $\forall r.C \in \mathcal{L}(v_1), v_1 \text{ is not indire}$	ctly blocked, and
	2. there is some role $r'$ with Tran	$r(r')$ and $r' \equiv r$
	3. there is an $r'$ -neighbour $v_2$ of	$v_1$ with $\forall r'.C \notin \mathcal{L}(v_2)$
	nen $\mathcal{L}(v_2) \to \mathcal{L}(v_2) \cup \{ \forall r'.C \}.$	
choose-rule	1. $\leq n r.C \in \mathcal{L}(v_1), v_1$ is not ind	irectly blocked, and
	2. there is an <i>r</i> -neighbour $v_2$ of $v$	$v_1 \text{ with } \{C, \neg C\} \cap \mathcal{L}(v_2) = \emptyset$
	then $\mathcal{L}(v_2) \to \mathcal{L}(v_2) \cup \{E\}$ for sor	me $E \in \{C, \neg C\}.$
≥-rule	1. $\geq n r.C \in \mathcal{L}(v), v$ is not block	ed, and
-	2. there are not $n$ safe $r$ -neighbor	urs $v_1, \ldots, v_n$ of $v$
	with $C \in \mathcal{L}(v_i)$ and $v_i \neq v_j$ for	or $1 \leq i < j \leq n$
1.000		A LINA A





Can exploit the results of 20+ years of DL research

- Well defined (model theoretic) semantics
- Formal properties well understood (complexity, decidability)
- Practical reasoning algorithms
- Effective implemented systems







#### **Ontologies** before:

Name	Original	de-	primi-	arti-	Σ	de-	primi-
	Language	fined	tive	ficial		fined	tive
		concepts			roles		
CKB	SB-ONE	23	57	58	138	2	46
Companies	BACK	70	45	81	196	1	39
FSS	SB-ONE	34	98	75	207	0	47
Espresso	SB-ONE	0	145	79	224	11	41
Wisber	TURQ	50	81	152	283	6	18
Wines	CLASSIC	50	148	237	435	0	10



#### **Ontologies** after:

Sv Sv	voogle Semantic Web Search Engine	
+ # http://swoogle.um	bc.edu/	C Qr Google
Some semantic v	veb search	Want more results? Login
ontology <u>document</u>	t term more >> Swoogle Search	3
Searchi	ng over 10,000 ontologies	
		11

#### **Ontologies** after:

#### Welcome to the Protege Ontology Library!

#### **OWL** ontologies

- AIM@SHAPE Ontologies A: Ontologies pertaining to digital shapes. Source: AIM@SHAPE NoE Advanced and Innovative Models And Tools for the development of Semantic-based systems for Handling, Acquiring, and Processing knowledge Embedded in multidimensional digital objects.
- amino-acid.owl 

  A small OWL ontology of amino acids and their properties. Source: Amino Acid Ontology Web site 

  Acid Ontology Web site
- Basic Formal Ontology (BFO) 🗗
- bhakti.owl : An OWL ontology for the transcendental states of consciousness experienced by practitioners of bhakti-yoga, a form of Vedic consciousness engineering.
- Biochemical Ontologies &: Over 30 ontologies for knowledge representation and reasoning across scientific domains. Ontologies are normalized into non-disjoint primitive skeletons and



#### "Profile" before:




#### "Profile" after:





**Applications** before:

#### **Applications** after:

- eScience, eCommerce, geography, engineering, defence, ...
- Major impact in healthcare and life sciences
- Mainstream technology supported by, e.g., ORACLE 11g
- Increasing impact in business applications





#### Peter and lan before:





#### Peter and Ian after:





Tools before:

```
> (load-tkb "demo.kb" :verbose T)
                    . . . . . . . . . . . . . . .
> (classify-tkb :mode :stars)
  pppppppppppppppccpcpcccpcppcpcpppcccpcp
  pccccppcpcppcccp
  ጥ
> (direct-supers 'MAN)
  (C[HUMAN] C[MALE])
>
```



#### Tools after:







**Major benefit** of OWL has been huge increase in range and sophistication of tools and infrastructure:

Editors/development environments





- Editors/development environments
- Reasoners





- Editors/development environments
- Reasoners
- Explanation, justification and pinpointing

♠ ● ● SWOOP v2.3 beta 3.1 (Jan 2006)						
File View Bookmarks Resource Holder A	dva	nced About				
Address: http://www.m	ninds	wap.org/ontologies/tambis-full.owl		•		
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tambis-full.owl			Ontology Info Species Validation	1		
		OWL Ontology: tambis-full.owl				
		Annotations:				
		Post/Derived Debugging Informat	ion.			
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Add C Add P Add T root unsat. classes (3)						
Add GCI Remove Rename metal (141)						
Show Imports QNames Pellet	metalloid (140)					
Class Tree Property Tree List						
owl:Thing						
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C process	active-site		macromolecule part, protein, site, protein part,			
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▶ ⓒ substance		alpha-helix	protein-structure, protein-secondary-structure, macromolecular-compound,			
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v e owl:Nothing			organic-molecular-compound			
methylation-site		amino-acid	small-organic-molecular-compound,			
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geranyl-geranyl-attachment-site		astatine	nonmetal, ?, metal, metalloid,			
😑 dna-binding-site		atom	nonmetal, metal, metalloid,			
e alkali-metal			protein-structure, protein-secondary-structure,			
anna-nart L		beta-sheet	macromolecular-compound,			
Lookup All Ontologies?						



Developing and using ontologies is *hard*



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- Reasoning enabled tools allow K-Eng to check if, e.g.:
  - classes are consistent (no "obvious" errors)



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  - expected subsumptions hold (consistent with intuitions)



- Developing and using ontologies is *hard*
- Reasoning enabled tools allow K-Eng to check if, e.g.:
  - classes are consistent (no "obvious" errors)
  - expected subsumptions hold (consistent with intuitions)
  - unexpected equivalences hold (unintended synonyms)





- OBO foundry includes more than 100 biological and biomedical ontologies
- Siemens "actively building OWL based clinical solutions"
- OWL tools used to find and repair critical errors in ontology used at Columbia Presbyterian
- **SNOMED-CT** (Clinical Terms) ontology
  - used in healthcare systems of more than 15 countries, including Australia, Canada, Denmark, Spain, Sweden and the UK
  - also used by major US providers, e.g., Kaiser Permanente
  - ontology provides common vocabulary for recording clinical data



#### SNOMED is **BIG** – over **400,000 concepts**



#### SNOMED is **BIG** – over 400,000 concepts

Image: Second provide the second	Pulmonary Tuberculosis
Words - any order       Image: Constraint of the second seco	pulmonary tuberculosis - Definition Concept Status: Current Descriptions pulmonary tuberculosis (disorder) pulmonary tuberculosis FTB - Pulmonary tuberculosis TB - Pulmonary tuberculosis TB - Pulmonary tuberculosis TB - Pulmonary tuberculosis Definition: Fully defined b inflammatory disorder of lower respiratory trace inflammation of specific body organs Inflammation of specific body or
<ul> <li>84353005 pulmonary disease due to Mycobacteria</li> <li>154283005 pulmonary tuberculosis</li> <li>428697002 inactive tuberculosis of lung</li> <li>186175002 infiltrative lung tuberculosis</li> <li>186188004 isolated tracheal or bronchial tuberculosis</li> <li>77668003 isolated tracheal tuberculosis</li> <li>80602006 nodular tuberculosis of lung</li> <li>186192006 respiratory tuberculosis, bacteriologically and hist</li> <li>186192007 respiratory tuberculosis, not confirmed bacteriolegical and histologic</li> <li>186194007 tuberculosis of lung with cavitation</li> <li>186194007 tuberculosis of lung, bacteriological and histologic</li> <li>186194007 tuberculosis of lung, confirmed by sputum micros tuberculosis of lung, confirmed histologically</li> </ul>	bacterial lower respiratory infection     mycobacteriosis     causative agent     foroup     finding site     foroup     forou

- Kaiser Permanente extending SNOMED to express, e.g.:
  - *non-viral pneumonia* (negation)
  - *infectious pneumonia* is caused by a *virus* or a *bacterium* (disjunction)
  - *double pneumonia* occurs in two *lungs* (cardinalities)
- This is easy in **SNOMED-OWL** 
  - but reasoner failed to find expected subsumptions, e.g., that bacterial pneumonia is a kind of non-viral pneumonia
- Ontology highly under-constrained: need to add disjointness axioms (at least)
  - virus and bacterium must be disjoint

- Adding disjointness led to **surprising results** 
  - many classes become inconsistent, e.g., *percutanious embolization of hepatic artery using fluoroscopy guidance*
- Cause of inconsistencies identified as class groin
  - groin asserted to be subclass of both abdomen and leg
  - abdomen and leg are disjoint
  - modelling of *groin* (and other similar "junction" regions) identified as incorrect

- Correct modelling of groin is quite complex, e.g.:
  - groin has a part that is part of the abdomen, and has a part that is part of the leg (*inverse properties*)

```
Groin \sqsubseteq \exists hasPart.(\exists isPartOf.Abdomen))
```

```
Groin \sqsubseteq \exists hasPart.(\exists isPartOf.Leg)
```

```
hasPart \equiv isPartOf^-
```

 all parts of the groin are part of the abdomen or the leg (disjunction)

 $\mathsf{Groin} \sqsubseteq \forall \mathsf{hasPart.}(\exists \mathsf{isPartOf.}(\mathsf{Abdomen} \sqcup \mathsf{Leg}))$ 

#### What we learned:

- Ontology engineering is error prone
  - errors of omission (e.g., disjointness) and commission (e.g., modelling of groin)
- Expressive features of OWL are sometimes needed
- Sophisticated tool support is essential
  - handling ontologies of this size is challenging
  - domain experts (and logicians!) often need help to understand the (root) cause of both inconsistencies and non-subsumptions
  - surprising and unexplained (non-) inferences are frustrating for users and may cause them to lose faith in the reasoner



#### **Case Study: BBC**





#### **Case Study: BBC**





### **Case Study: BBC**





## Why Ontology Reasoning?



# Why Ontology Reasoning?

 $Q(x) \leftarrow \mathsf{Patient}(x) \land \mathsf{suffersFrom}(x,y) \land \mathsf{VascularDisease}(y)$ 

i.e., "Patients suffering from Vascular Disease"





**Major benefit** of OWL has been huge increase in range and sophistication of tools and infrastructure:

 Integration and modularisation

🛿 JIAO.owl (http://www.semanticweb.org/ontologies/2008/9/JIAO.owl) - [C:\Archivos de programa\Protege_4.0_beta101\contentCVS_local\JI 📃 🔲 🛛							
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JRA 🕥	ojotonno_otot						
RA SubClassOf RA and Systemic Disease	JRA 🕥						
	SJRA subClassOf RA and (suffered_By some Child)						
Poly_JRA 🌖							
Poly_JRA subClassOf JRA and Multi_Joint_Dis	Poly_JRA						
Poly_JRA subClassOf AbnormalRA	Poly_JRA subClassOf JRA and (affects exactly 3 Joint)						
	Poly_JRA subClassOf hasRF only Positive_RF						
	Enviro (P)						
Oly_JRA subClassOf JRA and (not Poly_JRA)							
	Systemic_JRA						
	Systemic_JRA subClassOf JRA and (has_Symptom						
	some Fever)						
VEVIEW IMPACT OF CUSTOM MERGING	PREVIEW MERGING IN A NEW FRAME						



- Integration and modularisation
- Extraction

ctive Ontology Entities Classes Object Properties Data Properties	Individ	uals OWLViz DL Query Pr	roSÉ Manager	
roSÉ Safe Protege Manager:			me	
Select Signature Group Create/Modify External URIs		Extension of the selected Signature (Optional)		
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LimbJoint subClassOf JointsFromGalen	0			
HipJoint subClassOf JointsFromGalen	0	a. Preview Module 🦻	Number of Classes (Module/Ontology): 234 / 2748	
AnkleJoint subClassOf JointsFromGalen	0	b. Import Module		
SynovialJoint subClassOf JointsFromGalen         SO           KneeJoint subClassOf JointsFromGalen         SO			Number of Properties (Module/Ontology): 81 / 413	
		b. Come Back 🛛 🏹		
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		2. Import Whole Ontology		



- Integration and modularisation
- Extraction
- Comparison





- Integration and modularisation
- Extraction
- Comparison
- Protege plugins

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= Help search	Topics	Register your Plugin							
	Browse plugins by tonics of interest	Add a new plugin in 2 steps:							
Go Search	Biomedical Informatics Code Examples Export Import Inference Natural Language Processing Navigation Project Management Reasoning Search	1. Enter the name of your plugin, which will then be the title of a new article in this wiki.							
<ul><li>Related changes</li><li>Special pages</li><li>Printable version</li></ul>	Semantic Web Software Engineering Terminologies Validation Visualization Types	2. Enter as much information as possible in the provided form (don't forget to save your work).							
<ul> <li>Permanent link</li> <li>Browse properties</li> </ul>	Browse plugins by type. API Application Backend Import Export Project Reasoner Slot Widget	New Plugin							
	Tab Widget View	Register your Organization							



**Major benefit** of OWL has been huge increase in range and sophistication of tools and infrastructure:

- Integration and modularisation
- Extraction
- Comparison
- Protege plugins
- APIs, in particular the <u>OWL API</u>

Revision 1403 - (download) (annotate) Fri Dec 18 17:14:37 2009 UTC (4 months, 2 weeks ago) by matthewhorridge File size: 4711 byte(s) package org.coode.owlapi.examples; import org.semanticweb.owlapi.apibinding.OWLManager; import org.semanticweb.owlapi.model.\*; import org.semanticweb.owlapi.util.DefaultPrefixManager; 6 \* Copyright (C) 2009, University of Manchester \* Modifications to the initial code base are copyright of their 10 \* respective authors, or their employers as appropriate. Authorship 11 \* of the modifications may be determined from the ChangeLog placed at 12 \* the end of this file. 13 14 \* This library is free software; you can redistribute it and/or 15 \* modify it under the terms of the GNU Lesser General Public 16 \* License as published by the Free Software Foundation; either 17 \* version 2.1 of the License, or (at your option) any later version. 18 \* This library is distributed in the hope that it will be useful, 19 20 \* but WITHOUT ANY WARRANTY; without even the implied warranty of 21 \* MERCHANTABILITY or FITNESS FOR A PARTICULAR PURPOSE. See the GNU 22 \* Lesser General Public License for more details.





# **Ongoing Research**

- Query answering
  - [Kontchakov et al], [Konev et al], [Baader et al]
- Diagnosis and repair
  - [Horridge et al], [Peñaloza et al]
- Extensions
  - [Motik et al], [Artale et al]
- Optimisation/Profiles
  - [Kazakov], [Glimm et al], [Faddoul et al], [Savo et al]



# **Ongoing Standardisation Efforts**

- Standardised query language
  - SPARQL standard for RDF
  - Currently being extended for OWL, see <a href="http://www.w3.org/TR/sparql11-entailment/">http://www.w3.org/TR/sparql11-entailment/</a>
- RDF
  - Revision currently being considered, see <a href="http://www.w3.org/2009/12/rdf-ws/">http://www.w3.org/2009/12/rdf-ws/</a>




## Thank you for listening





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Any questions?