Public Health – Medical Informatics

Semantic Web, terminology, ontology in health

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Health is the richest domain in terms of existence & development of (followed by law):

- Classifications
- Controlled vocabularies
- Thesaurus
- Terminologies
- Ontologies

Engineering sciences: very poor in terminologies & ontologies (T/O) => also poor in bibliographic databases

In health, around 200 in UMLS,
Over 500 in BioPortal (including biology)
Around 70 in HeTOP, crosslingual but mainly in French
One T/O for each domain: ICD10 for disease, MeSH for documentation, FMA for anatomy…

Documentary language

- Artificial language, constituted of notions and relations between notions
- Goal: in a documentary system, to formalize data contained in the document and data in users' queries
- Two main families:
  - Language with hierarchy structure (classifications), with symbolic indices
  - Language with combination structure (thesaurus), using words of the natural language

Classification vs. thesaurus

- Two main methods to perfom an indexing
  - Synthetic method: from general to specific
  - Analytical method: decomposition of concepts and combination of them
- Two families of documentary languages
  - Synthetic method ⇨ systematic indexing ⇨ language with hierarchy structure = **classification**
  - Analytical method ⇨ analytical indexing or alphabetical indexing ⇨ language with analytical structure or combinatory structure = **thesaurus**

Precoordination & Postcoordination

- Contradictory organization and use of documentary languages
- In precoordinated languages (classifications)
  - Terms of indexing are mainly words or composed indices (e.g. 121.2.1) covering the entire notion
  - Coordination between the concepts are performed when indexing
- In postcoordinated languages (thesaurus)
  - Notions are defined by the most simple constituants
  - Several descriptors are necessary to describe the entire notion
  - Coordination between the concepts are performed when information retrieval using query operators (e.g. Boolean)

Classification

- Knowledge separated in small units
- Domain to cover
  - Separated in subdomains, then subsubdomains… to obtain the smallest granularity
- Language with hierarchy structure
  - ICD10 = monoaxial structure
  - Coded language (e.g. ICD10)
- Possibility to group codes by themes

Dewey Decimal Classification

Def. = system of library classification made up of ten classes, each divided into ten divisions, each having ten sections.

- 110 Metaphysics
- 111 **Ontology**
  - 111.1 - Essence, existence
  - 111.2 - Universaux
  - 111.5 - Néant
  - 111.6 - Fini, infini
  - 111.8 - Propriétés de l'être
    - 111.82 - Unité
    - 111.84 - Bonté
    - 111.85 – Beauté
- 112 *No longer used—formerly Methodology*
- 113 Cosmology (Philosophy of nature)
- 114 Space

International Classification of Diseases

- WHO World Heath Organization
- standard diagnostic tool for epidemiology, health management and clinical purposes
- Translated into 43 languages
- System to report mortality data, a primary indicator of health status
- DRG Diagnosis Related Group (PMSI in France)

⇨ Hospital budget +++

- Version 10 since 1994
- Version 11 in 2018 ???

International Classification of Diseases

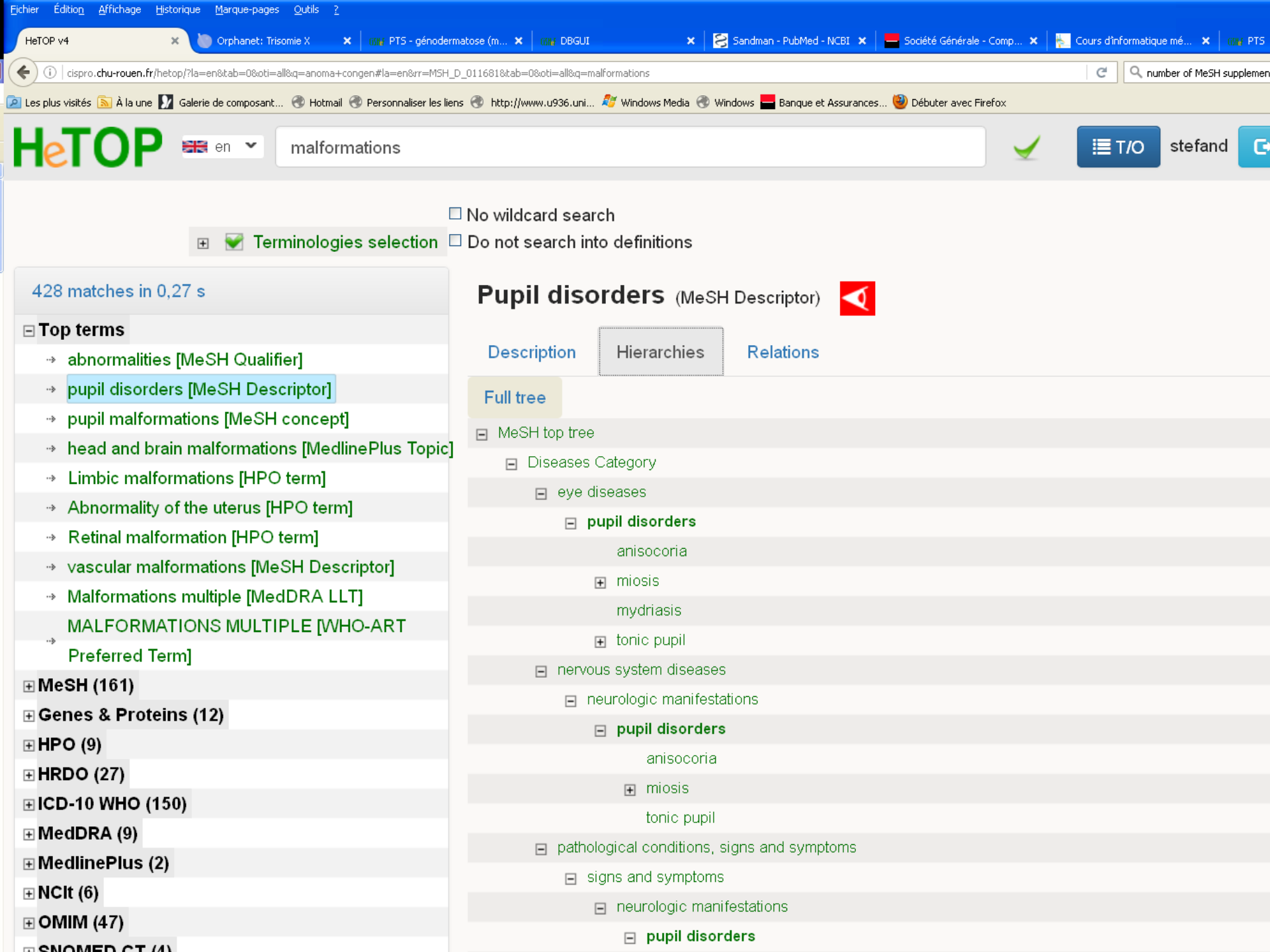
- ICD-10 top tree
  - Diseases of the respiratory system
    - Chronic lower respiratory diseases
      - Asthma
      - Bronchiectasis
      - Bronchitis, not specified as acute or chronic
      - Emphysema
        - Centrilobular emphysema
        - Emphysema, unspecified
        - MacLeods syndrome
        - Other emphysema
        - Panlobular emphysema
      - Other chronic obstructive pulmonary disease
        - Chronic obstructive pulmonary disease with acute exacerbation, unspecified
        - Chronic obstructive pulmonary disease with acute lower respiratory infection
        - Chronic obstructive pulmonary disease, unspecified
        - Other specified chronic obstructive pulmonary disease
      - Simple and mucopurulent chronic bronchitis
        - Mixed simple and mucopurulent chronic bronchitis
        - Mucopurulent chronic bronchitis
        - Simple chronic bronchitis
      - Status asthmaticus
      - Unspecified chronic bronchitis

Thesaurus

- Controlled vocabulary

=> « limited » number of descriptors

- Each descriptor is linked to other via several relations
  - Hierarchy
    - IS A
    - PART OF
    - Merge of these two relations => BTNT & NTBT ; wrong for ontologies with reasoning capabilities
- Most used thesaurus in medicine = MeSH used to index article citations in MEDLINE/PubMed bibliographic database
  - N (MeSH Descriptors) ≃27,000
  - N (MeSH Supplementary Concepts) ≃228,000
  - N (MeSH Concepts) ≃352,000 > N(SNOMED CT)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No wildcard search
Terminologies selection
Do not search into definitions
428 matches in 0,27 s
Top terms
abnormalities [MeSH Qualifier]
pupil disorders [MeSH Descriptor]
pupil malformations [MeSH concept]
head and brain malformations [MedlinePlus Topic]
Limbic malformations [HPO term]
Abnormality of the uterus [HPO term]
Retinal malformation [HPO term]
vascular malformations [MeSH Descriptor]
Malformations multiple [MedDRA LLT]
MALFORMATIONS MULTIPLE [WHO-ART Preferred Term]
MeSH (161)
Genes & Proteins (12)
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ICD-10 WHO (150)
MedDRA (9)
MedlinePlus (2)
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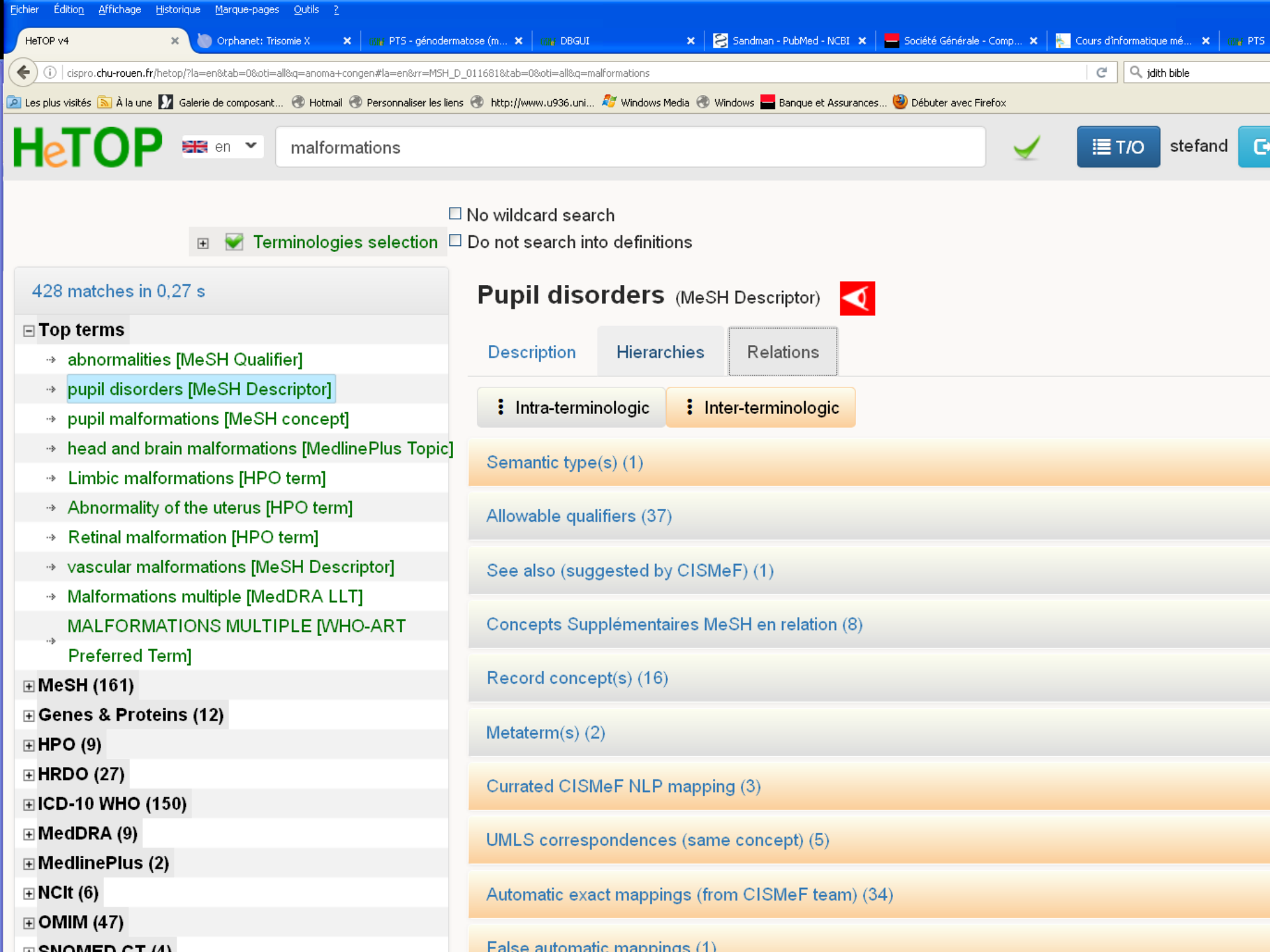
OMIM (47)
Pupil disorders (MeSH Descriptor)
Description
Hierarchies
Relations
Full tree
MeSH top tree
Diseases Category
eye diseases
pupil disorders
anisocoria
miosis
mydriasis
tonic pupil
nervous system diseases
neurologic manifestations
pupil disorders
anisocoria
miosis
tonic pupil
pathological conditions, signs and symptoms
signs and symptoms
neurologic manifestations
pupil disorders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Galerie de composant...
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Personnaliser les liens
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Pupil disorders (MeSH Descriptor)
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Hierarchies
Relations
Intra-terminologic
Inter-terminologic
Semantic type(s) (1)
Allowable qualifiers (37)
See also (suggested by CISMeF) (1)
Concepts Supplémentaires MeSH en relation (8)
Record concept(s) (16)
Metaterm(s) (2)
Currated CISMeF NLP mapping (3)
UMLS correspondences (same concept) (5)
Automatic exact mappings (from CISMeF team) (34)

HeTOP en malformations T/O stefand

☐ No wildcard search
☐ Do not search into definitions
Terminologies selection

428 matches in 0,27 s

- **Top terms**
  - abnormalities [MeSH Qualifier]
  - pupil disorders [MeSH Descriptor]
  - pupil malformations [MeSH concept]
  - head and brain malformations [MedlinePlus Topic]
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- **SNOMED CT (4)**

Pupil disorders (MeSH Descriptor)

Description | Hierarchies | Relations

Intra-terminologic | Inter-terminologic

Semantic type(s) (1)

Allowable qualifiers (37)

See also (suggested by CISMeF) (1)

Concepts Supplémentaires MeSH en relation (8)

congenital corneal opacities, cornea guttata, and corectopia	MeSH Supplementary Concept
pupil disorders/congenital:microcoria, congenital	MeSH Descriptor/MeSH Qualifier: MeSH S
ectopia lentis with ectopia of pupil	MeSH Supplementary Concept
ectopia pupillae	MeSH Supplementary Concept
McPherson robertson Cammarano syndrome	MeSH Supplementary Concept
microphthalmia, isolated, with corectopia	MeSH Supplementary Concept
pierson syndrome	MeSH Supplementary Concept
ptosis, strabismus, and ectopic pupils	MeSH Supplementary Concept

Record concept(s) (16)

Metaterm(s) (2)

Use cases in health of terminologies

- Statistics
  - Since XIXth century, mortality statistics using ICD
- Controlled indexation (information sciences)
  - Bibliographic databases +++ MEDLINE/PubMed
- Use or reuse of clinical (& omics) data
  - Care (aggregated visualization of these data)
  - Epidemiology
  - Clinical trials
  - Indicators
  - …

Towards Ontology

Requête « Pot » sur un moteur de recherche











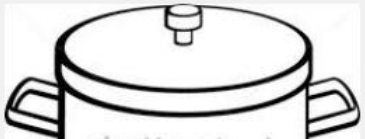












Perhaps a question of “knowledge” ?

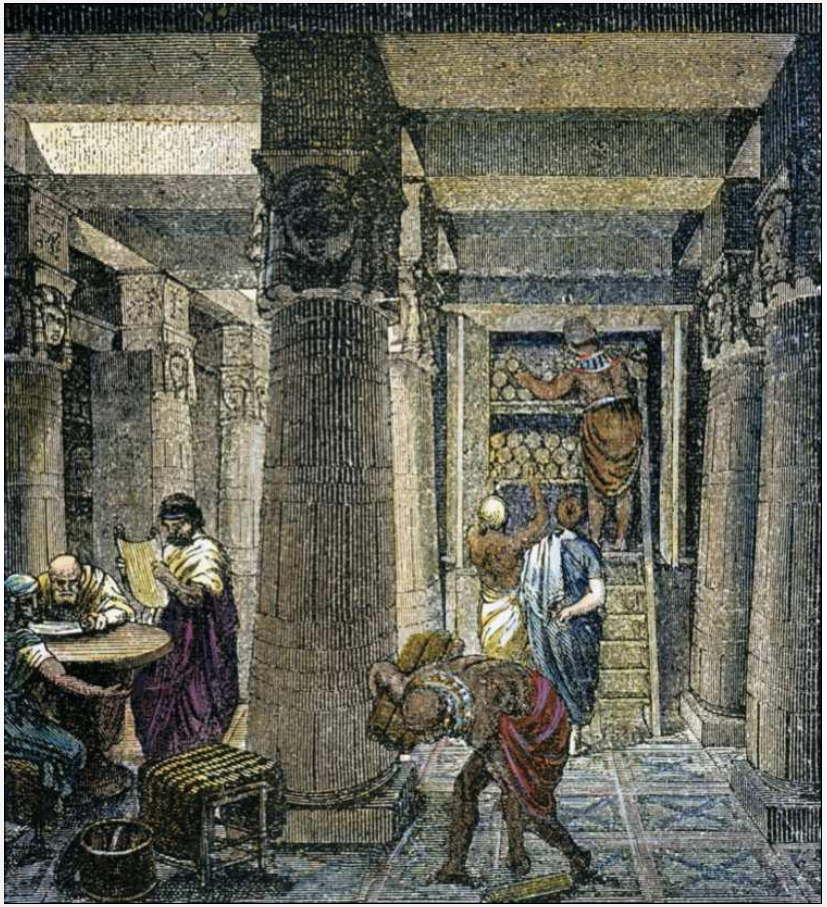
- A question of interpretation of the word “pôt”
- A question of “knowledge”
- A question of shared conceptualization
- If I want that the computer helps me during my work (recognition, research, “reasoning”, etc.)
- I need a computer “ontology”

Definitions of ontology

- Philosophy
  - Part of metaphysics, which applies to the nature of being, becoming, existence, or reality, as well as the basic categories of being and their relations. Traditionally listed as a part of the major branch of philosophy known as metaphysics, ontology often deals with questions concerning what entities exist or may be said to exist, and how such entities may be grouped, related within a hierarchy, and subdivided according to similarities and differences.
- Computer Science & Knowledge Engineering
  - "An ontology is a shared specification of a conceptualization" (by Tom Gruber, 1990)
  - Formal naming and definition of the types, properties, and interrelationships of the entities that really or fundamentally exist for a particular domain of discourse. It is thus a practical application of philosophical ontology, with a taxonomy.

History (1)

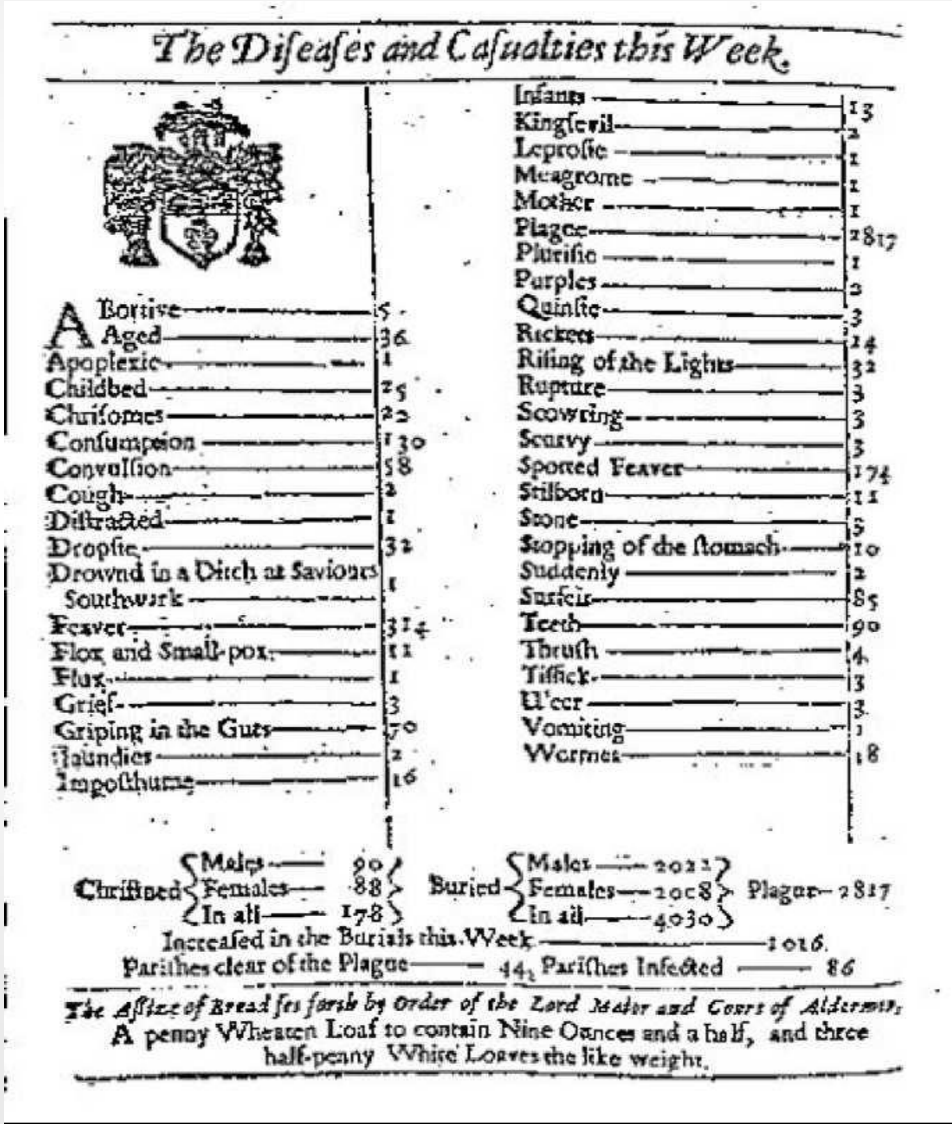
- IIIrd century BC.
- **Library of Alexandria**
- First public Library
- Has an index to manage its collections of 700 000 books



History (2)

- XVIIth century
- **London Bills of mortality**
- Classification of diseases used to make an inventory of deceases
- Published each Thursday from 1603 and during more than 2 centuries !

=> Ancester of ICD



The Diſeaſes and Caſualties this Week.

Abortive	5	Infants	13
Aged	36	Kingſevil	2
Apoplexie	1	Leproſie	1
Childbed	25	Meagrome	1
Chriſomes	20	Mother	1
Conſumpeion	130	Plague	2817
Convulſion	58	Plurifie	1
Cough	2	Purples	2
Diſtracted	1	Quinſie	3
Dropſie	32	Rickets	14
Drowned in a Ditch at Saviours Southwark	1	Riſing of the Lights	32
Feaver	314	Rupture	3
Flox and Small-pox	11	Scowring	3
Flux	1	Scurvy	3
Grief	3	Spotted Feaver	174
Griping in the Guts	70	Stilborn	11
Iaundies	2	Stone	5
Impoſthume	16	Stopping of the ſtomach	10
		Suddenly	2
		Surfeit	85
		Teeth	90
		Thruſh	4
		Tiſſick	3
		Ulcer	3
		Vomiting	1
		Wormes	18

Chriſtned: Males 90, Females 88, In all 178 — Buried: Males 2022, Females 2008, In all 4030 — Plague 2817

Increaſed in the Burials this Week 1016.

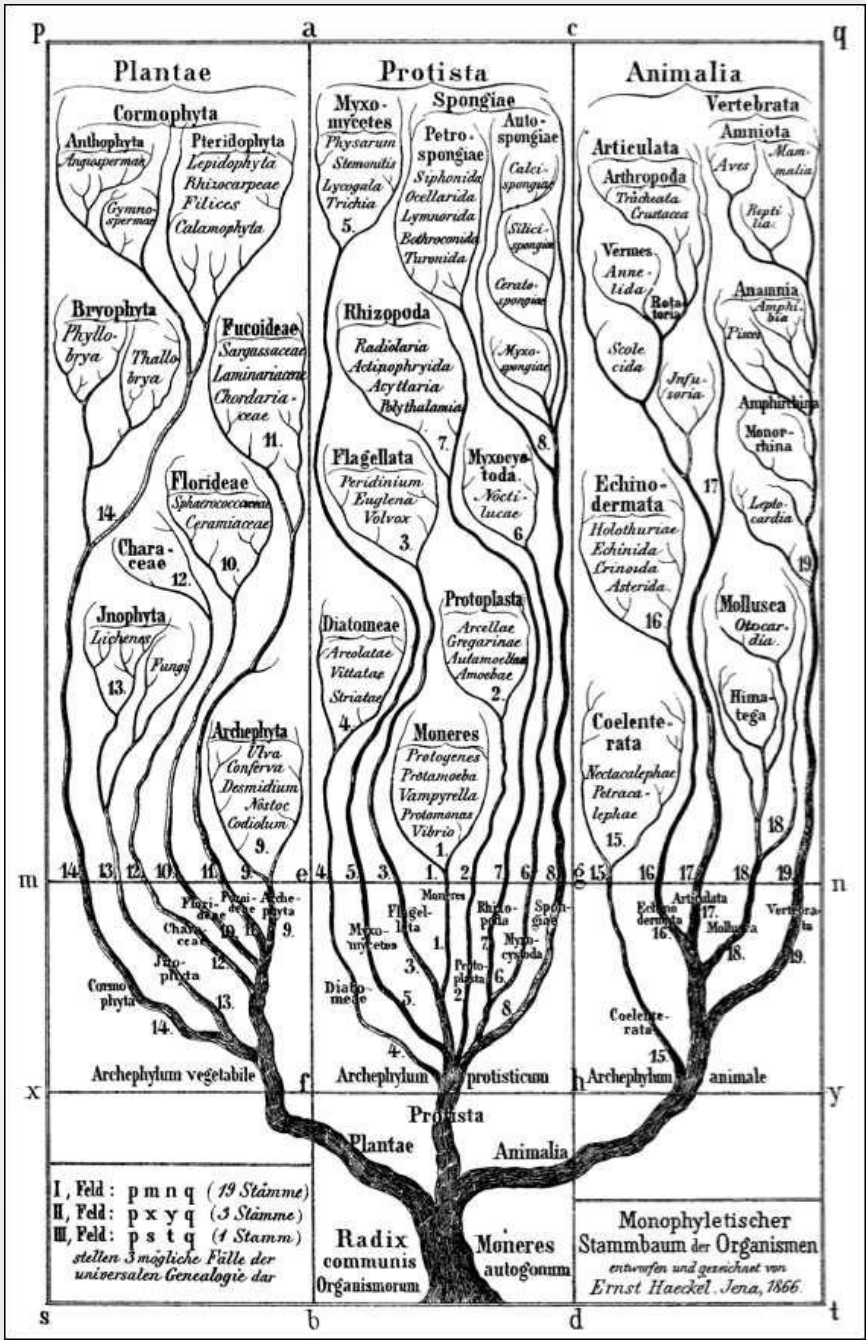
Parithes clear of the Plague 44. Pariſhes Infected 86

The Aſſize of Bread ſet forth by Order of the Lord Maior and Court of Aldermen:

A penny Wheaten Loaf to contain Nine Ounces and a half, and three half-penny White Loaves the like weight.

History (3)

- XVIIIth century
- World exploration and building of real life classifications



Terminology vs. Ontology

Ontology

- Richer than terminology
- Formal definitions
- Inferencing +++
  - Protege tool (Stanford)
- Formal languages
  - RDF
  - OWL, OWL2

Terminology

- More practical approach
- Terminology servers +++
- Rich relations as ontology
- No inferencing
- Solution
  - First, use ontology with inferencing to clean it
  - Then, implement on a terminology server
  - e.g. FMA OWL2 => HeTOP
    Golbreich C et coll. The Foundational Model of Anatomy in OWL 2 and its use. **Artif Intell Med** 2013 ;57(2), 119-132.

Expected Utility

- Pragmatic approaches for KBS and the SW
  - ☞ To create and maintain reusable KB
  - ☞ Interoperability between different KBS
  - ☞ Conceptual vocabulary (referential) of information system
  - ☞ Conceptual vocabulary in order to tag or index documents
  - ☞ Model of RDF triples inside semantic datawarehouses of the Linked Open Data

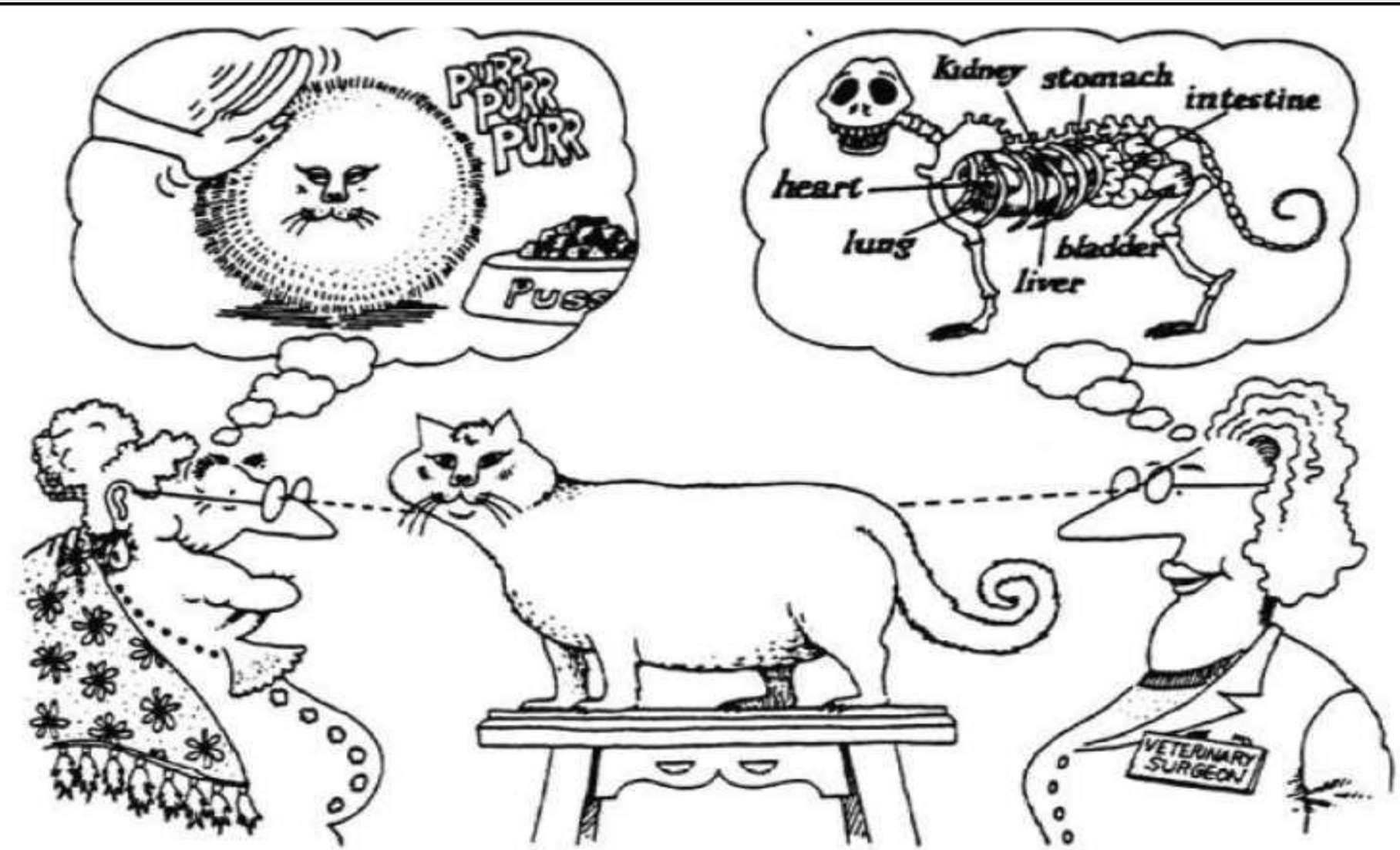
Terminological and Ontological Resources (TOR)

- Which sharable abstractions?
  - Lexicon
  - Thesaurus
  - Ontology (for KBS, as metadata . . .)
  - Domain model
  - Case model
  - Decomposition in recurrent tasks
  - Problems Solving Methods
  - Abstract application tasks

Some opposite points of view?

- First vision
  - An ontology is universal but different from a KB which would be individuated, relative, and finalized
- Second vision
  - A few “ontologies” for a same domain
  - Impossible to deliver an universal ontology including all possible points of view
  - It bears the trace of the particular task for which it have been built and the reasoning for this task

A cat is a cat



Abstraction focuses upon the essential characteristics of some object, relative to the perspective of the viewer.

Remarks

- On the negotiation of meaning
  - Despite different views on the cat, it could imagine that Grandma and the vet can negotiate a shared sense
  - But this meaning is continuously renegotiated, it is not stable and therefore difficult to be represented in an ontology… or terminology

Processus of ontology building (Guarino

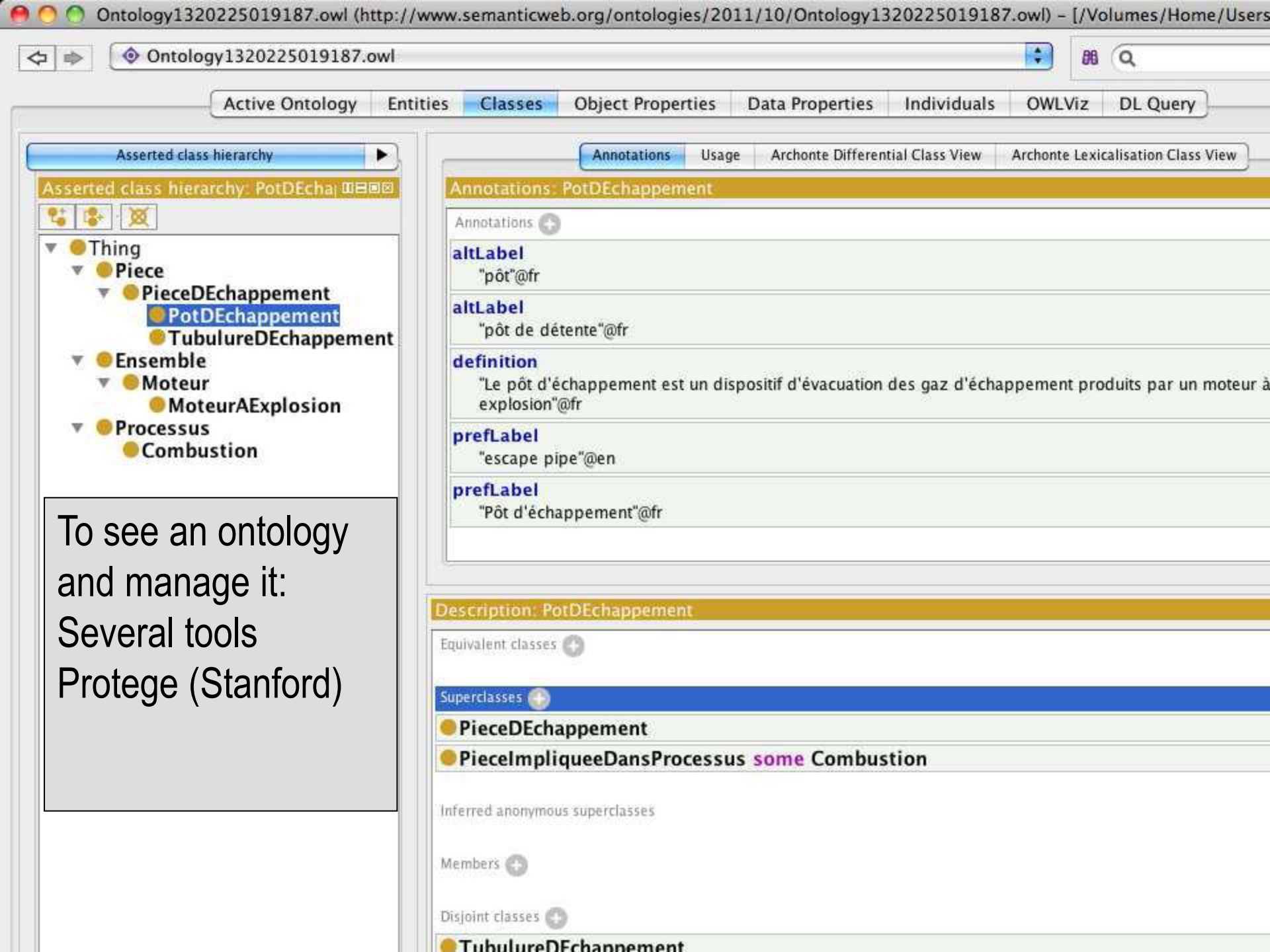
- Guarino 94 :
  - It is necessary to fix precisely and previously
    - The general ontological commitments
    - High level categories : appellations and significations
    - The specialization of theses categories
- Guarino 96 :
  - The determination of a domain ontology must fix expected significations from domain primitives
  - But, this primitives don’t exist in an expertise domain.
  - Explicit ontological modelization process in order to establish a set of primitives as a prerequisite necessary to domain modelization

About Knowledge Representation

- The exposition of a formal language of knowledge representation leaves open the question of functional and relational symbols required and semantics to associate.
- Defining an ontology for the knowledge representation is defining, for a domain and a problem given, the functional and relational signature of a formal representation language and the associated semantics. (Bachimont 2000)
- Defining non-logical primitives of a representation language and associated semantics : identifying basic concepts from which domain knowledge is built.

Two roles of an ontology

- At computer side
  - Define / provide a formal semantics for the information allowing its use by a computer
- At human being side
  - Define / provide an interpretative semantics of real world domain, based on a consensus, and allowing to link the content usable by the computer to its meaning for human being
- A model of knowledge about the world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Active Ontology
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Object Properties
Data Properties
Individuals
OWLViz
DL Query
Asserted class hierarchy
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Moteur
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Processus
Combustion
Annotations
Usage
Archonte Differential Class View
Archonte Lexicalisation Class View
Annotations: PotDEchappement
Annotations
altLabel
"pôt"@fr
altLabel
"pôt de détente"@fr
definition
"Le pôt d'échappement est un dispositif d'évacuation des gaz d'échappement produits par un moteur à
explosion"@fr
prefLabel
"escape pipe"@en
prefLabel
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Equivalent classes
Superclasses
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Members
Disjoint classes
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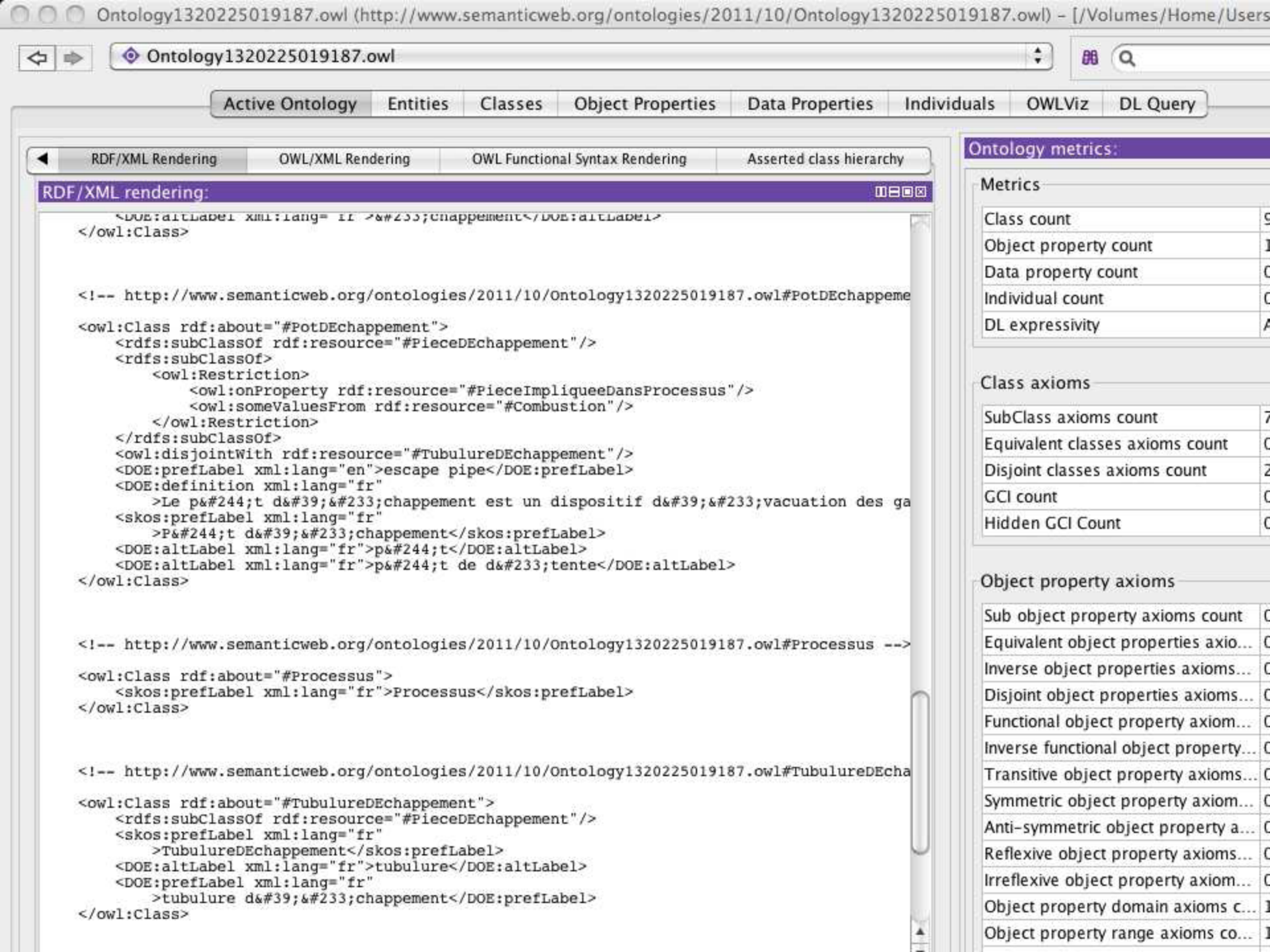
To see an ontology
and manage it:
Several tools
Protege (Stanford)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Asserted class hierarchy
RDF/XML rendering:
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<rdfs:subClassOf>
<owl:Restriction>
<owl:onProperty rdf:resource="#PieceImpliqueeDansProcessus"/>
<owl:someValuesFrom rdf:resource="#Combustion"/>
</owl:Restriction>
</rdfs:subClassOf>
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>Pôt d'échappement</skos:prefLabel>
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</owl:Class>
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<skos:prefLabel xml:lang="fr"
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<DOE:prefLabel xml:lang="fr"
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Ontology metrics:
Metrics
Class count
Object property count
Data property count
Individual count
DL expressivity
Class axioms
SubClass axioms count
Equivalent classes axioms count
Disjoint classes axioms count
GCI count
Hidden GCI Count
Object property axioms
Sub object property axioms count
Equivalent object properties axio...
Inverse object properties axioms...
Disjoint object properties axioms...
Functional object property axiom...
Inverse functional object property...
Transitive object property axioms...
Symmetric object property axiom...
Anti-symmetric object property a...
Reflexive object property axioms...
Irreflexive object property axiom...
Object property domain axioms c...
Object property range axioms co...

Normalize conceptually separating the concepts (about disease)

- shifting the meaning of objects in the speech
  - process or state?
  - « l'évolutivité de la maladie est rapide » versus « la maladie est un état morbide »
  - physiological process or diagnostic measure ?
    - Bowel transit is good vs. Bowel transit is xx.xx
  - Metonymy about localization
    - figure of speech in which a thing or concept is called not by its own name but rather by the name of something associated in meaning with that thing or concept

Query the status of concepts

Shift the meaning of objects in entry forms

- « Hyperglycemia » as reason
  - Result of a declaration by the patient or information from an entry form
- « Hyperglycemia » as finding
  - Result of a biological analysis but requiring verification over time to confirm that we are in front of a…
- « Hyperglycemia » as disease
  - Practitioner diagnostic

Building (medical) ontologies

- By reusing ontologies or parts of ontologies already built
- **By reusing terminological resources (thesaurii, classifications, . . .) <= our approach in Rouen**
  - **Expanding, translating, mapping (EM, CM, BTNT, NTBT)**
- By explaining the underlying conceptualizations in patterns of DBMS
- By analyzing textual data generated during the activity to conceptualize
- By combining theses approaches as appropriate

But

- Which conceptual organization?
- Which granularity?
- Primitive versus defined concepts?
- And about “top-ontologies” reutilization?

Articulation Top/core/domain

- The top ontology
  - The most abstract level structuring knowledge with high-level categories. Its organization depends on philosophical reflections. The question the uniqueness or otherwise of this ontology is discussed.
- The core ontology
  - Provides the structuring concepts of the domain and the relationships between these concepts – in medecine, these concepts are diagnostic, sign, anatomical structure and the relations as diagnostic localisedOn anatomical structure.
- The domain ontology
  - Domain concepts as they are manipulated by Professional. This level can be built with NLP tools because these tools analyse document write during the professional activity => interface terminologies

About differential semantics

- The fact that we follow the differential principles (Aristotle,Rastier), implies that we construct a tree… without cycles
- The sibling concepts of a level represent mutual exclusive notions
- This tree and these principles provide a better maintenance of the ontology ➠ addition of a new concept
- This also allows a better modularity ➠ each branch extracted from the tree at any level is really independent of the rest of the tree.

Ontological Commitments

At formal level, concepts are classes (subclasses) and individuals. . .

- A class
  - A class defines all the properties that characterize a certain set of objects. A class is something abstract, rather than a particular element of the set of the described objects (e.g. employees class)
- An individual
  - An individual is an object that has exactly the properties of its parent class (e.g. Virginie, new employee)

Introduction to metadata

- These technics are still used today in metadata indexation
  - thematic classification
  - controlled vocabulary or not
  - controlled tagging, controlled resource type
  - Type of metadata set chosen (e.g. Dublin Core, LOM)
- Existing resources
  - Dewey & Freinet classification (library & information science)
  - GEMET (environment)
  - MeSH (medical) ←
  - Jurivoc (legal) ←

Dublin Core old metadata set

1	Title	A name given to the resource.
2	Creator	Name of the person, the organisation, primarily responsible for making the resource.
3	Subject	The topic of the resource. Typically, the subject will be represented using keywords, key phrases, or classification codes. Recommended best practice is to use a controlled vocabulary.
4	Description	An account of the resource. Description may include but is not limited to: an abstract, a table of contents, a graphical representation, or a free-text account of the resource
5	Contributor	An entity responsible for making contributions to the resource
6	Publisher	An entity responsible for making the resource available
7	Date	A point or period of time associated with an event in the lifecycle of the resource
8	Resource type	The nature or genre of the resource. Recommended best practice is to use a controlled vocabulary such as the DCMI Type Vocabulary
9	Format	The file format, physical medium, or dimensions of the resource
10	Identifier	An unambiguous reference to the resource within a given context
11	Source	A related resource from which the described resource is derived
12	Language	A language of the resource
13	Relation	A related resource
14	Coverage	The spatial or temporal topic of the resource, the spatial applicability of the resource, or the jurisdiction under which the resource is relevant
15	Rights	Information about rights held in and over the resource

Dublin Core new metadata set

Properties in the /terms/ namespace n=55 (+40)

abstract , accessRights , accrualMethod , accrualPeriodicity , accrualPolicy , alternative , audience , available , bibliographicCitation , conformsTo , contributor , coverage , created , creator , date , dateAccepted , dateCopyrighted , dateSubmitted , description , educationLevel , extent , format , hasFormat , hasPart , hasVersion , identifier , instructionalMethod , isFormatOf , isPartOf , isReferencedBy , isReplacedBy , isRequiredBy , issued , isVersionOf , language , license , mediator , medium , modified , provenance , publisher , references , relation , replaces , requires , rights , rightsHolder , source , spatial , subject , tableOfContents , temporal , title , type , valid

URL: http://dublincore.org/

Semantic Web

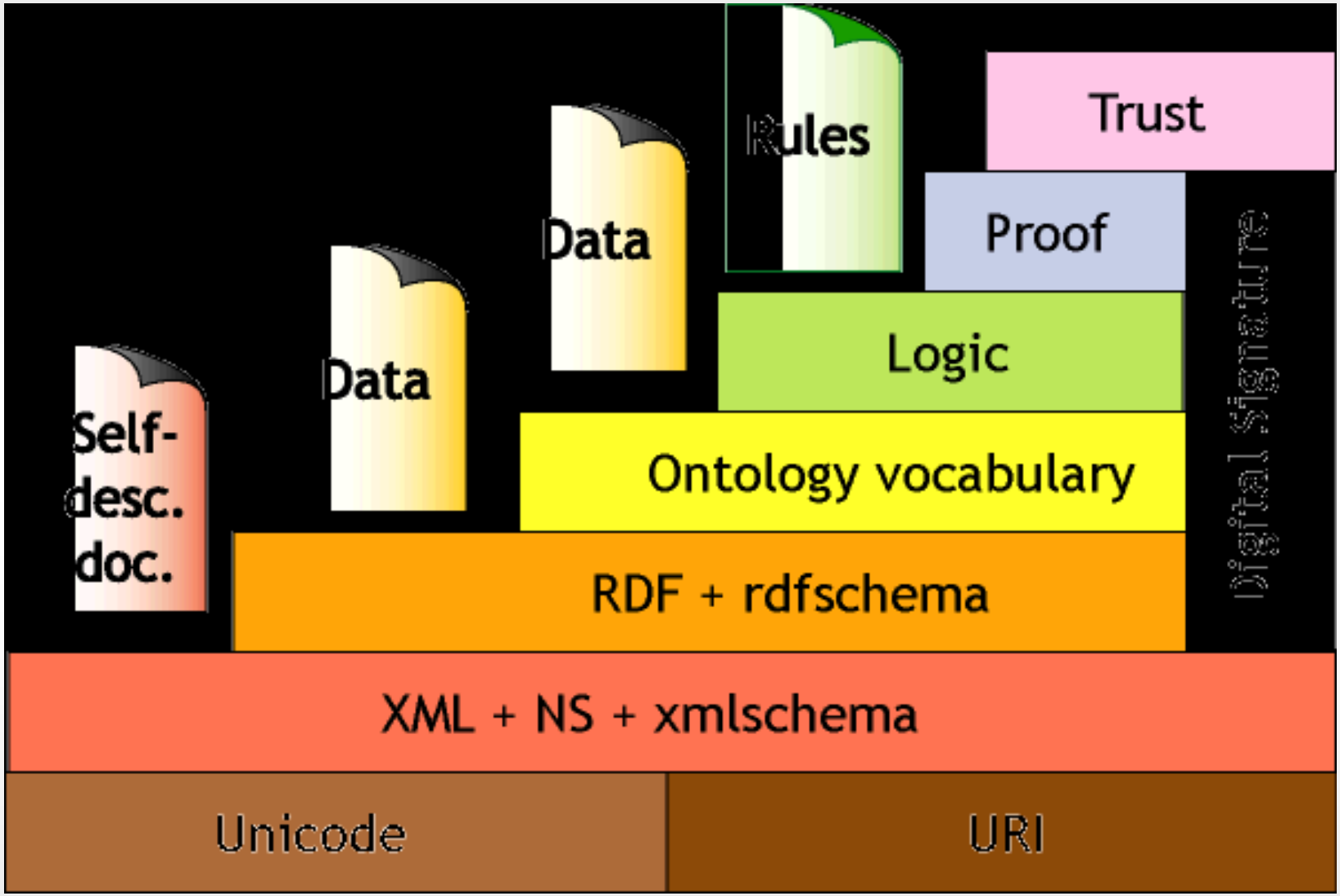
The Web today : properties

- Its “universality”
  - ☞ The homogeneity of the used techniques
    - HTTP, HTML, URI/URL
  - ☞ The power of the hypertexte
    - “each resource” may be link to “each resource”
  - ☞ Web resources are documents primarily elaborated for human use
    - Even if it exists more and more software tools. . .

The first vision of the semantic web

- The Web tomorrow : A huge space of resources exchange between machines enabling users access to large volumes of information and to various services [Tim Berners-Lee (W3C)]
  - ☞ Different languages to describe, exploit and reason about the contents of the resources
  - ☞ Knowledge based on ontologies
  - ☞ Utilization of metadata
  - ☞ Automatic integration of informations from heterogeneous sources
  - ☞ Utilization and automatic combination of Web services
  - ☞ Personnalisation and adaptation
- Towards more relevant answers
- Towards data integration and heterogeneous services integration

The « cake » of the semantic web



Ontologies: different needs

- ☞ Conceptual vocabulary to tag and index documents => terminologies
- ☞ Publish and share database type information => terminologies
- ☞ Semi-automatic integration of information between software agents => ≈ terminologies

Small size ontologies available everywhere versus big size ontologies

Technical point of view

- RDF triple store
  - Using semantic web technologies

Then

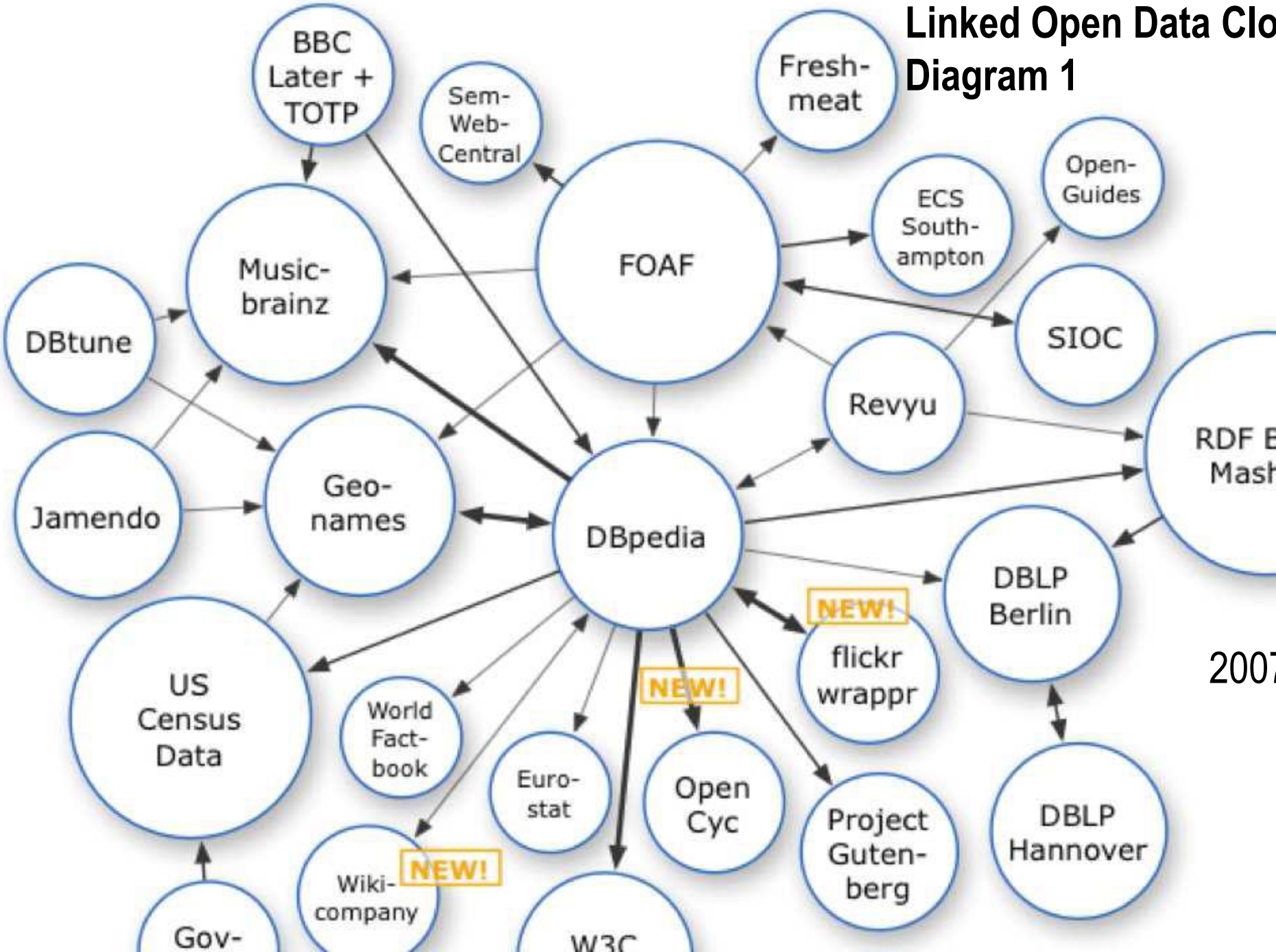
- NoSQL
  - For real implementation
- Benchmark 2014 in our lab to develop HeTOP
  - NoSQL >> RDF triple store
  - Which NoSQL
    - MangoDB, InfiSpan, …

The second vision of the semantic web I

- The Web of data
  - ☞ Create an automatic link to connect the data that is stored in various files and databases of our computers
  - ☞ A huge repository of information buried in all computers of the planet: by linking them, the semantic web will allow to exploit this mine of information in order to to improve our knowledge
  - ☞ RDF to link data to categories defined by OWL ontologies

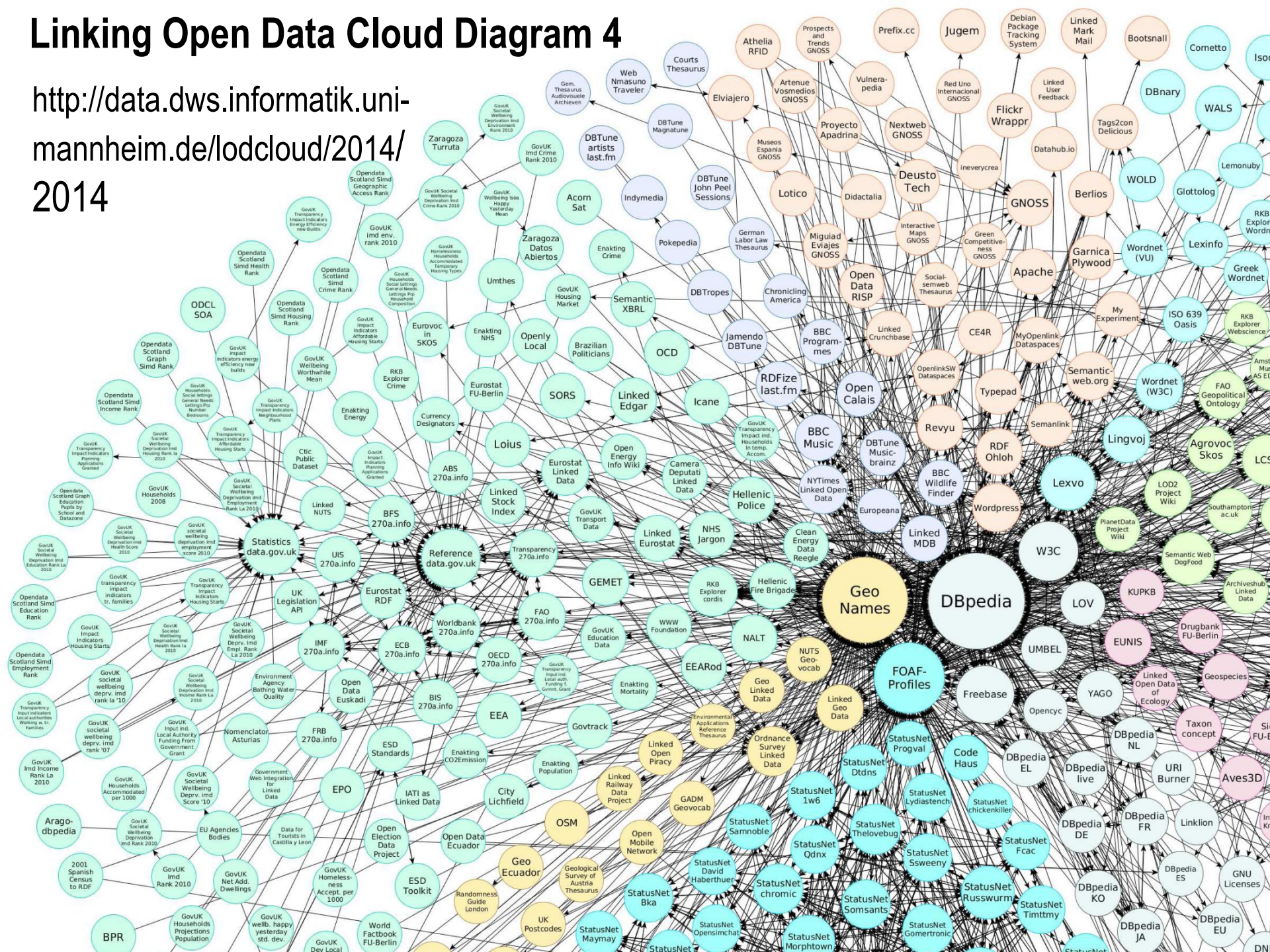
The second vision of the semantic web II

- Each company will have to mark all the data it wants to publish on the semantic web with a description. Tools, such as D2R Server developed by the Free University of Berlin, scans tables of databases and convert them to Semantic Web format according to an ontology
- Access to the huge mass of data, the “deep Web”, through a query language defined by the W3C, SPARQL, using RDF triple
  - **Concept -> Relation -> Concept**
  - Acebutolol -> Contradication -> Asthma
  - Acebutolol -> Indication -> Arterial hypertension


Linked Open Data Clo
Diagram 1
BBC Later + TOTP
Sem-Web-Central
Fresh-meat
Open-Guides
ECS South-ampton
Music-brainz
FOAF
DBtune
SIOC
Revyu
RDF B Mash
Jamendo
Geo-names
DBpedia
DBLP Berlin
NEW!
flickr wrappr
US Census Data
World Fact-book
NEW!
Euro-stat
Open Cyc
Project Guten-berg
DBLP Hannover
NEW!
Wiki-company
Gov-
W3C
2007

Linking Open Data Cloud Diagram 4

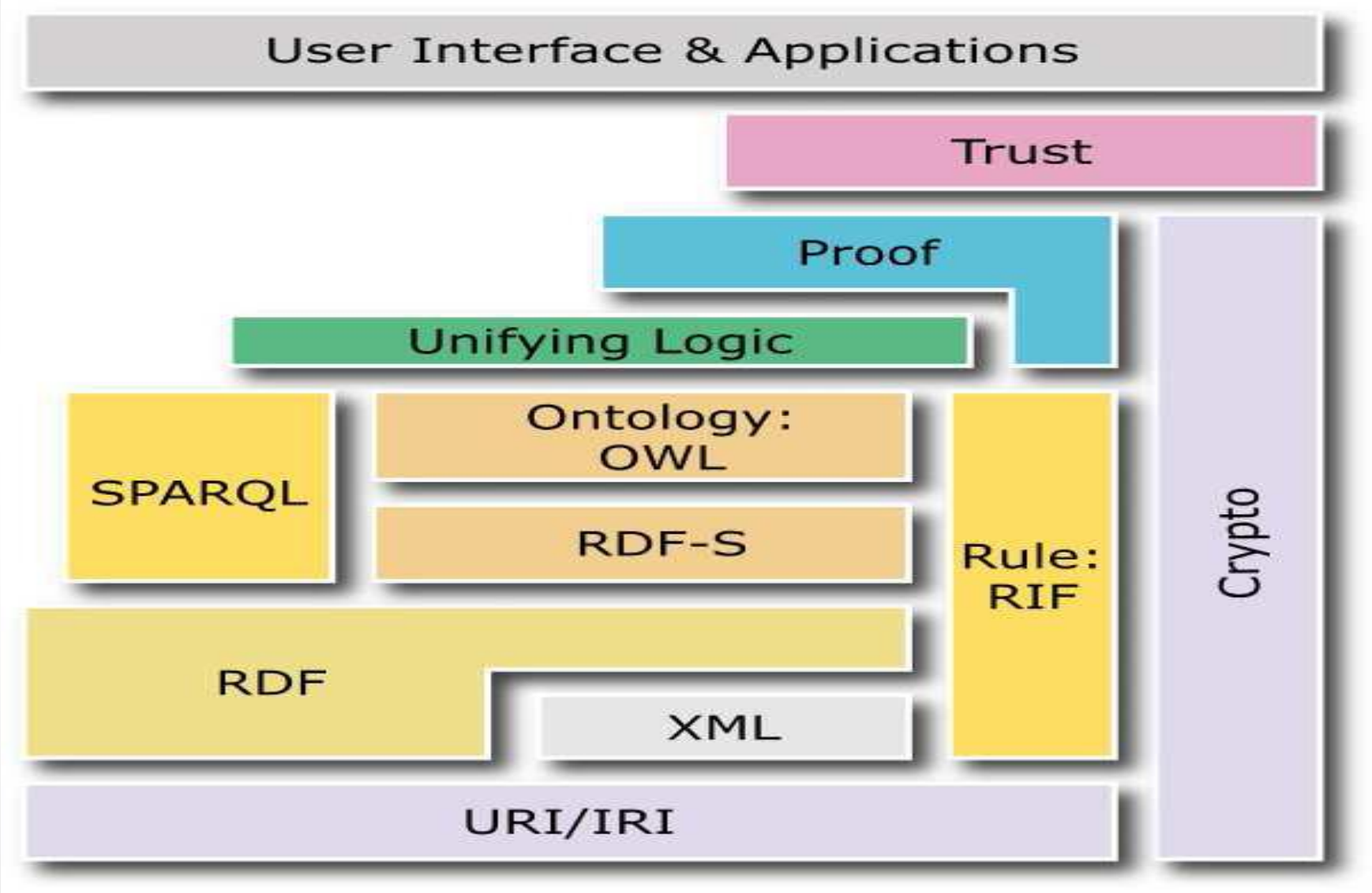
http://data.dws.informatik.uni-mannheim.de/lodcloud/2014/2014



The second vision of the semantic web III

- Great use of small size ontologies – even simplistic like the DC – before specific use of domain ontologies
- Usage of “small” ontologies inversely proportional to their size (Dublin Core, FOAF, . . .)

The « cake » of the Web of data



Three main terminology servers in health

- UMLS URL: https://uts.nlm.nih.gov/home.html
  - NIH, Bethesda (USA)
  - Around 200 T/O
  - Mainly in English
  - The international reference for dissemination, but not for consultation

- BioPortal* URL: bioportal.bioontology.org/
  - NCBO, Stanford (USA)
  - More than 500 T/0 (a lot in biology, with few hundred concepts)
  - Mainly in English (not crosslingual)
  - The reference to post and display an ontology

- HeTOP* URL: www.hetop.eu
  - SIBM, Rouen, Normandy (France)
  - 69 T/0 in 32 languages
  - The crosslingual reference (navigation between languages) and in French

*_Grosjean J et coll._ An Approach to Compare Bio-Ontologies Portals. **Stud Health Technol Inform**, 2014;205:1008-1012.

UMLS

- Unified Medical Language System
- compendium of many controlled vocabularies in the biomedical sciences
- created in 1986 by the US NLM, updated quarterly
- Knowledge Sources
  - Metathesaurus
  - Semantic Network
  - SPECIALIST Lexicon

UMLS Metathesaurus

- The base of the UMLS
- comprises over 1 million biomedical concepts and 5 million concept names
- organized by concept, and each concept has specific attributes defining its meaning and is linked to the corresponding concept names
- Numerous relationships: for instance hierarchical ones such as "isa" for subclasses and "is part of" for subunits
- Around 200 incorporated controlled vocabularies and classification systems
  - ICD-10
  - MeSH
  - SNOMED CT
  - DSM-IV
  - LOINC
  - MedDRA
  - RxNorm
  - Gene Ontology &
  - OMIM

UMLS Semantic Network

- Each concept in the UMLS Metathesaurus is assigned one or more *semantic types,* which are linked with one another through *semantic relationships*
- Semantic network = catalog of these semantic types (semantic groups) and relationships
  - 135 ST and 54 R

UMLS SPECIALIST Lexicon

- Information about:
  - common English vocabulary,
  - biomedical terms found in MEDLINE and in the UMLS Metathesaurus.
- Each entry contains:
  - syntactic (how words are put together to create meaning),
  - morphological (form and structure) and
  - orthographic (spelling) information
- In French, UMLF project (Zweigenbaum et al.)

HeTOP content

- HeTOP is a repository dedicated to (European) health professionals and students.
URL: www.hetop.eu

-HeTOP provides access to 69 health terminologies and ontology (T/O) available mainly in French or in English, but also German, Italian and Dutch (European languages) but also with no Latin alphabet (Greek, Russian) and more recently outside Europe (Japanese, Mandarin, Arabic & Hebrew) (**32 different languages**).

-HeTOP can be used by humans and by computers via Web services.

- The main objective of HeTOP is to provide an access to terminologies and ontology, allowing dynamic browsing and navigation.

  • Free portal for over 20 T/O: e.g. MeSH, CISMeF, ICD10, & CCAM;
  extended access restricted by ID/pwd for academic use only

HeTOP content

- HeTOP provides the usual data for each concept: preferred terms, original code, synonyms, definitions and other attributes, relations and hierarchies.

- Double (matricial) navigation:
  - among T/O
  - among languages

- Time consuming task > 20 man-years (to develop) + 2 man-years per year to maintain (integration & maintenance of T/O + mappings)
- Time consuming task to translate terminologies +++

- Several services on demand
  - access to other resources on the Internet (PubMed, CISMeF, etc.) through a French InfoButton (InfoRoute)
  - access to mappings tools (integrated in a beta version)
  - acces to automatic indexing tool (ECMT)

HeTOP methods

To integrate terminologies and ontology into EHTOP, three steps are necessary:

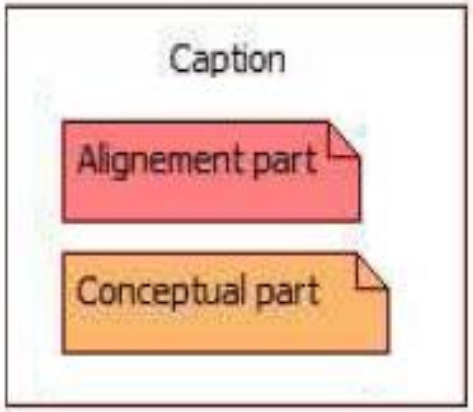
(1) designing a meta-model into which each terminology and ontology can be integrated,

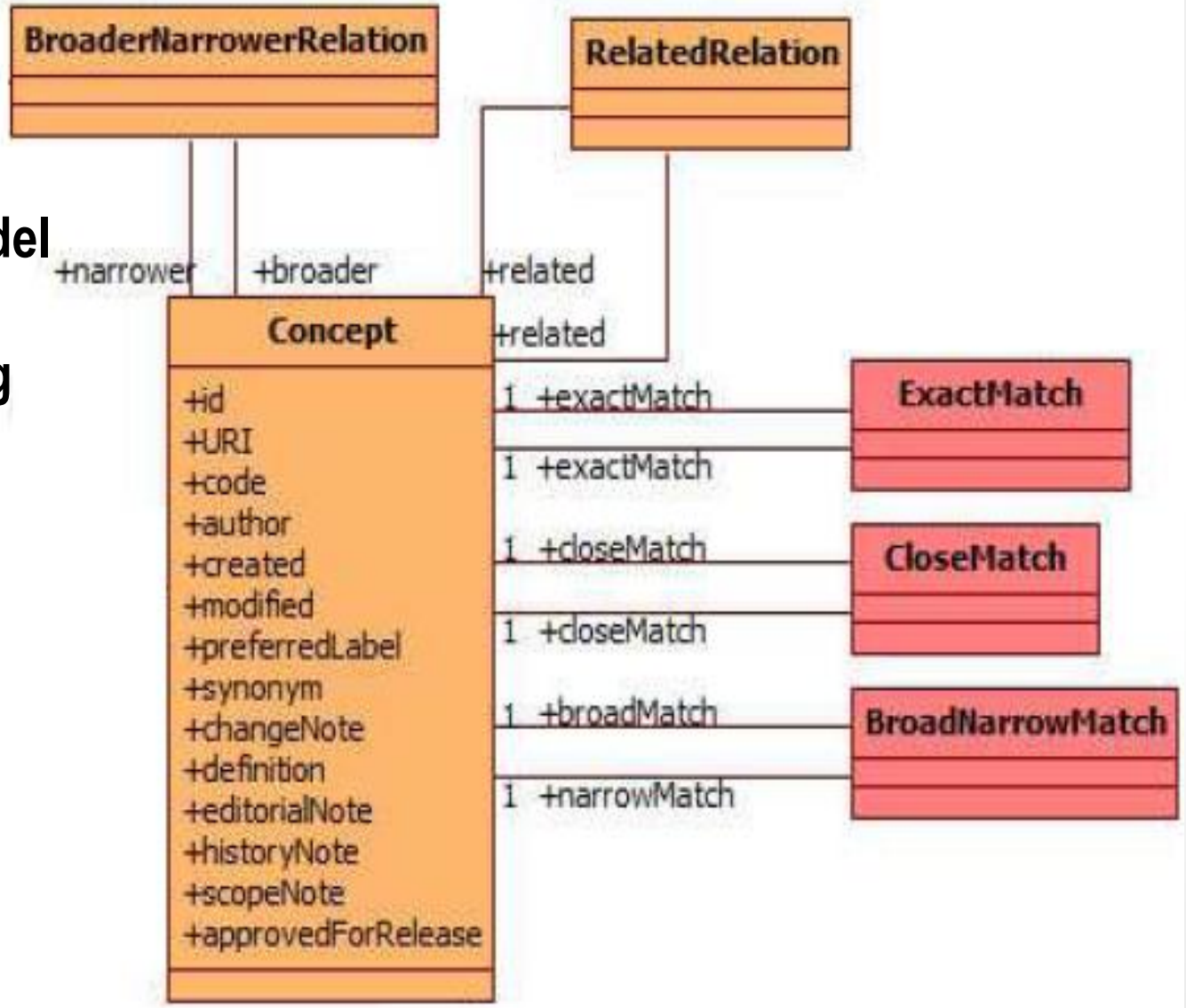
(2) developing a process to include terminologies into EHTOP,

(3) building and integrating existing and new inter & intra-terminology semantic harmonization into EHTOP.

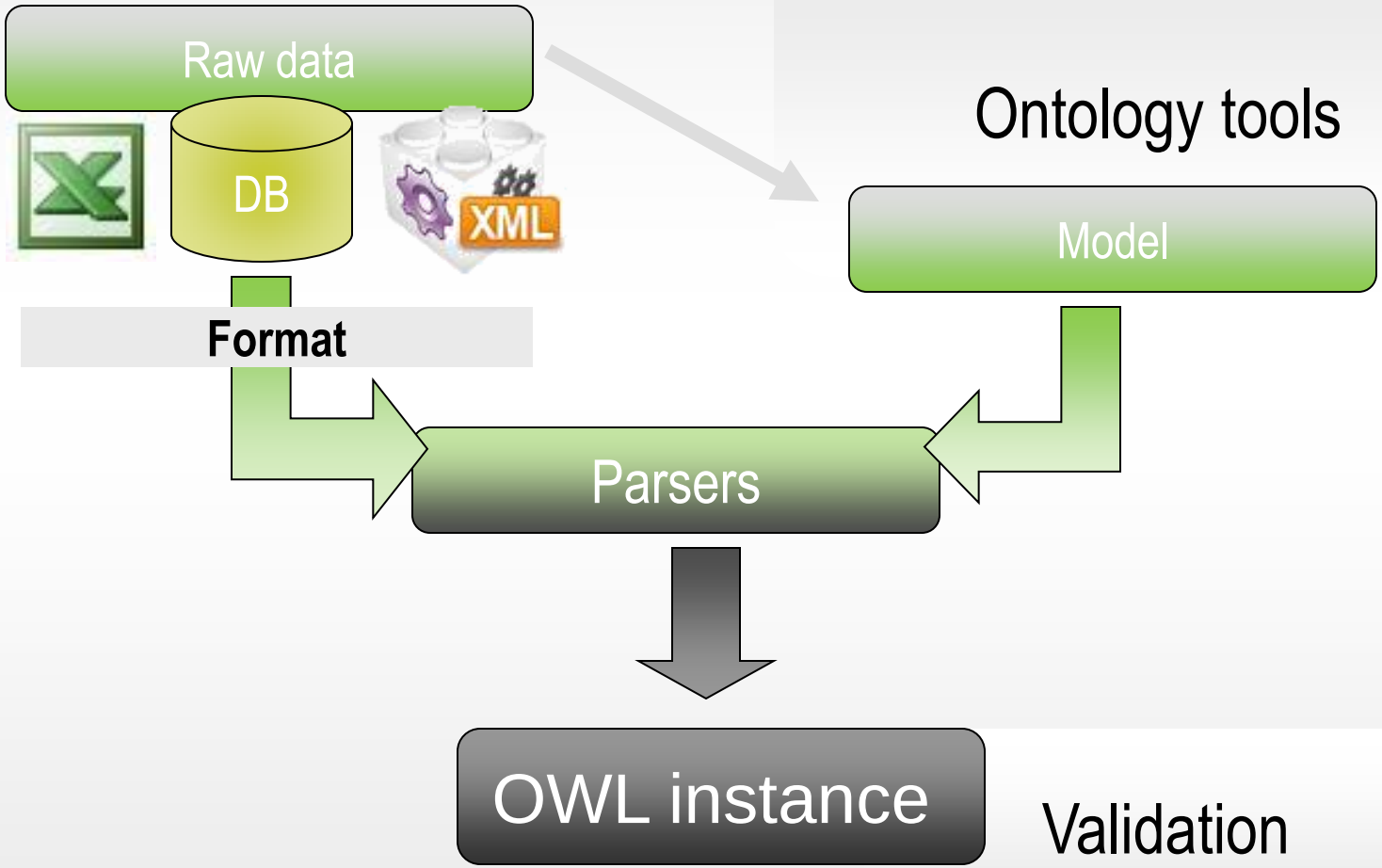
HeTOP generic model

Compliant with
ISO Terminology model
More simple
No versioning

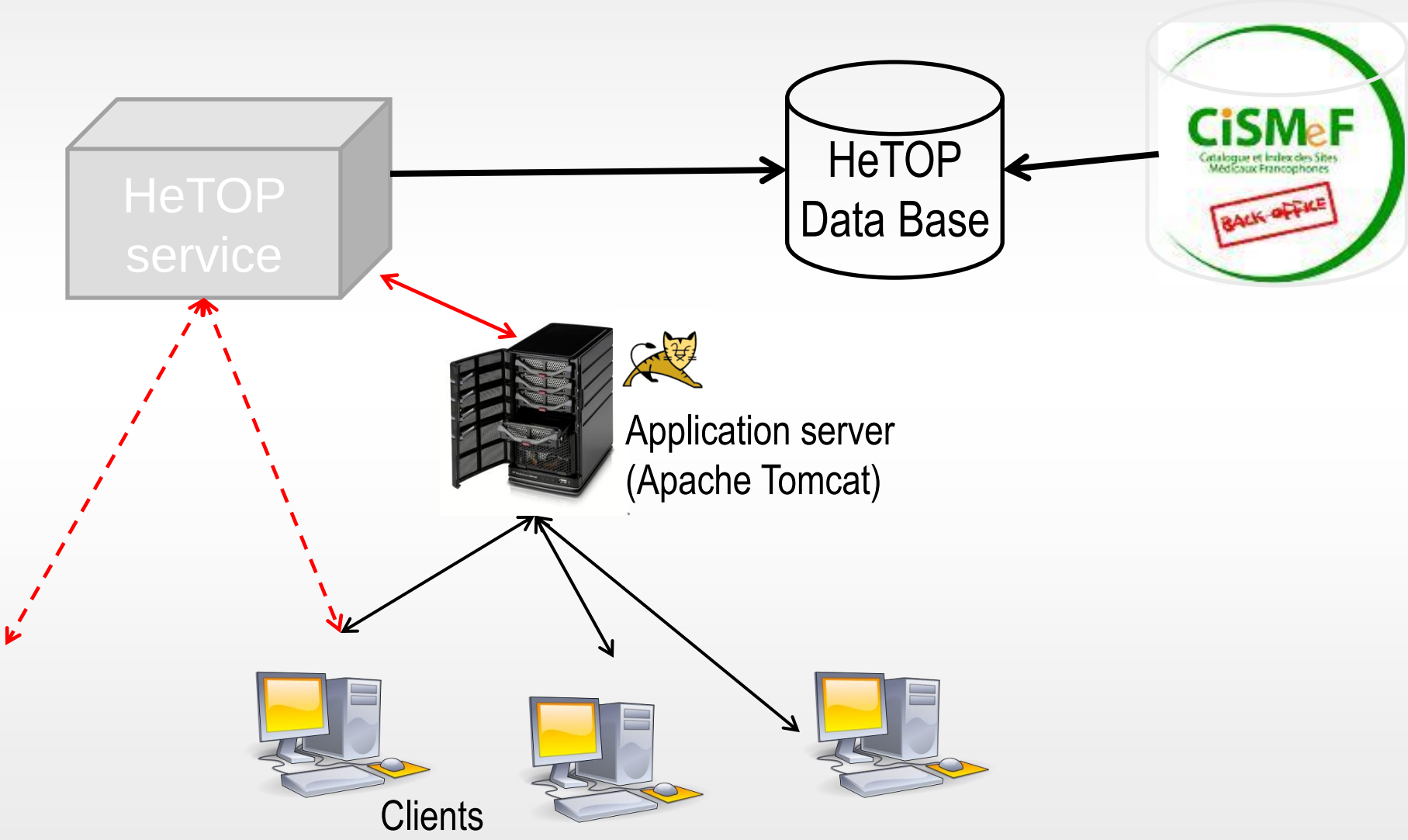




HeTOP integration: OWL instances



HeTOP technologies (1)



HeTOP technologies (2)

- Oracle 11.1g (optimizations & partitionning)
=> NoSQL since 2015

- Java J2EE
- CISMeF APIs

- Apache Tomcat
- Infinispan cache layer

- Cross-browser (Vaadin framework)
=> new framework in 2016 (INSA Rouen Engineering School)

Croslingual Health Multi-Terminology/Ontology Portal

- First version before HeTOP (French & English)
- URL: http://pts.chu-rouen.fr/
- Access for humans and coumputers (Web services)
  - Since September 2010, daily used by CISMeF team to index manually and automatically Web resources
  - Since January 2011, MeSH is freely available (500 unique users per working day)
  - Teaching tool: Rouen Medical School (since Sept. 2010) to teach anatomy and rare diseases
  - Terminology auditing: HPO/Orphanet
  - T/O translations into French: FMA, HPO, SNOMED CT, MEDLINEplus
- Restricted access to the other terminologies (2,250 registred)
- Cooperation with BioPortal: Clement Jonquet & Mark Musen (ANR Jeunes Chercheurs: project SIFR)

HeTOP asthme

exemples : asthme, asthma, D001249.nu, asth,

94 entrées trouvées en 2,39 s

Sélection terminologies

Vos recherches (2)

Historique des consultations (4)

Résultats

- MeSH (21)
  - Descripteur MeSH (8)
    - antiasthmatiques
    - asthme
    - asthme à l'effort
    - asthme induit par l'aspirine
    - asthme professionnel
    - dyspnée paroxystique
    - état de mal asthmatique
    - remodelage des voies aériennes
  - MeSH Concept Supplémentaire (4)
    - asthme et polypes nasaux
    - asthme, petite taille et IgA élevés
    - asthme, polypes nasaux et intolérance à l'aspi
    - dermatite atopique, type 3
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    - Remodelage des voies aériennes dans l'asthm
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- HPO (3)
- MedlinePlus (2)
- NCIt (2)
  - Concept NCIt (2)
    - asthme
    - Asthme chronique obstructif
- OMIM (2)
- RADLEX (1)
- SNOMED int. (30)
- WHO-ART (3)

Description | Hiérarchies | Relations | PubMed / Doc'CISMeF

Asthme (Descripteur MeSH)

Ressource consultée 1675 fois Afficher toutes les langues

BioPortal NLM Inserm

Libellé préféré

asthme
asthma

Identifiant d'origine

D001249

Definition du MeSH

Forme de maladie bronchique présentant une obstruction des voies respiratoires, marquée par des attaques récurrentes de dyspnée paroxysmale avec sifflements dûes à la contraction spasmodique des bronches. [Traduction effectuée avant 2008]
A form of bronchial disorder with three distinct components: airway hyper-responsiveness (RESPIRATORY HYPERSENSITIVITY), airway INFLAMMATION, and intermittent AIRWAY OBSTRUCTION. It is characterized by spasmodic contraction of airway smooth muscle, WHEEZING, and dyspnea (DYSPNEA, PAROXYSMAL).

Synonyme CISMeF

asthmas, bronchial
bronchial asthmas

Synonyme MeSH

Asthme bronchique
asthma, bronchial
asthmas
bronchial asthma

CUI UMLS

C0004096

VIDAL

Maladie caractérisée par une difficulté à respirer, se traduisant souvent par des sifflements. L'asthme, permanent ou survenant par crise, est dû à un spasme et à une inflammation des bronches.

url (CISMeF)

CiSMeF CiSMeF

Annotation MeSH

note specifics; ASTHMA, CARDIAC see DYSPNEA, PAROXYSMAL is also available; in historical literature consider indexing "phthisic" here; "phthisis" probably goes under TUBERCULOSIS, PULMONARY

Icône VCM hiérarchique

HeTOP asthme

exemples : asthme, asthma, D001249.nu, asth,

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Description | Hiérarchies | Relations | PubMed / Doc'CISMeF

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Libellé préféré

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Asma
АСТМА БРОНХИАЛЬНАЯ
Astma
哮喘

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Synonyme CISMeF

asthmas, bronchial
bronchial asthmas
Alternative helseforsikringssystemer

Synonyme MeSH

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asthma, bronchial
asthmas
bronchial asthma

CUI UMLS

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- **WHO-ART (3)**

Description | Hiérarchies | Relations | PubMed / Doc'CISMeF

Asthme (Descripteur MeSH)

Arborescence complète

- arborescence MeSH
  - Maladies
    - maladies de l'appareil respiratoire
      - hypersensibilité respiratoire
        - **asthme**
          - asthme à l'effort
          - asthme induit par l'aspirine
          - asthme professionnel
          - état de mal asthmatique
      - maladies des bronches
        - **asthme**
          - asthme à l'effort
          - asthme induit par l'aspirine
          - asthme professionnel
          - état de mal asthmatique
      - maladies pulmonaires
        - bronchopneumopathies obstructives
          - **asthme**
    - maladies du système immunitaire
      - hypersensibilité
        - hypersensibilité immédiate
          - hypersensibilité respiratoire
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              - asthme à l'effort
              - asthme professionnel
              - état de mal asthmatique

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exemples : asthme, asthma, D001249.nu, asth,

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Description | Hiérarchies | Relations | PubMed / Doc'CISMeF

Asthme (Descripteur MeSH)

Intra-terminologiques | Inter-terminologiques

- Liste des qualificatifs affiliables (37)
- Voir aussi (2)
- Type(s) sémantique(s) (1)
  - maladie ou syndrome — Type sémantique
- Concepts Supplémentaires MeSH en relation (4)
- Concept(s) lié(s) au record (1)
- Métaterme(s) (2)
- Information(s) d'indexation (20)
- Topic(s) MedlinePlus (2)
  - asthme — Topic MedlinePlus
  - asthme chez l'enfant — Topic MedlinePlus
- Alignements manuels CISMeF (1/2)
  - asthme chez l'enfant — Topic MedlinePlus
- Alignements automatiques CISMeF supervisés (9/14)

.0804493 - asthme		Code CIM-9
asthme		Topic MedlinePlus
asthme		Notion SNOMED
asthme		Résultat de consultation DRC
ASTHME		Terme préféré WHO-ART
asthme		RCE DRC
Asthme		Terme HPO
asthme		Concept NCIt
J45 - asthme		Catégorie CIM-10

- Correspondances UMLS (même concept) (7/17)

C0004096	493.9 - asthme, sai	Code CIM-9
C0004096	asthme	Notion SNOMED
C0004096	ASTHME	Terme préféré WHO-ART
C0004096	asthme	Topic MedlinePlus
C0004096	asthme	Concept NCIt
C0004096	J45 - asthme	Catégorie CIM-10
C0004096	J45.9 - asthme, sans précision	Sous Catégorie CIM-10

- Alignements automatiques supervisés en BTNT (1/2)
  - 493.9 - asthme, sai — Code CIM-9
- Alignements automatiques exacts (par équipe CISMeF) (3/9)

493.9 - asthme, sai	Code CIM-9
Asthme	Concept Radlex
prédisposition à l'asmthe	Phénotype OMIM

- Alignements automatiques faux (1/3)

HeTOP asthme

exemples : asthme, asthma, D001249.nu, asth,

94 entrées trouvées en 2,39 s

Sélection terminologies
Vos recherches (2)
Historique des consultations (4)

Résultats

- MeSH (21)
  - Descripteur MeSH (8)
    - antiasthmatiques
    - asthme
    - asthme à l'effort
    - asthme induit par l'aspirine
    - asthme professionnel
    - dyspnée paroxystique
    - état de mal asthmatique
    - remodelage des voies aériennes
  - MeSH Concept Supplémentaire (4)
    - asthme et polypes nasaux
    - asthme, petite taille et IgA élevés
    - asthme, polypes nasaux et intolérance à l'aspi
    - dermatite atopique, type 3
  - MeSH Concept (9)
    - Asthme
    - Asthme à l'effort
    - Asthme cardiaque
    - asthme et polypes nasaux
    - Asthme induit par l'aspirine
    - Asthme professionnel
    - asthme, petite taille et IgA élevés
    - asthme, polypes nasaux et intolérance à l'aspi
    - Remodelage des voies aériennes dans l'asthm
- CIM-9 (8)
- CIM-10 (18)
- DRC (4)
- HPO (3)
- MedlinePlus (2)
- NCIt (2)
  - Concept NCIt (2)
    - asthme
    - Asthme chronique obstructif
- OMIM (2)
- RADLEX (1)
- SNOMED int. (30)
- WHO-ART (3)

Description | Hiérarchies | Relations | PubMed / Doc'CISMeF

Asthme (Descripteur MeSH)

Qualificatif(s) utilisable(s) pour ce mot clé :

Afficher la liste alphabétique des qualificatifs

- analyse
  - liquide céphalorachidien
  - sang
  - urine
- anatomie et histologie
  - cytologie
    - anatomie pathologique
  - embryologie
  - classification
- diagnostic
  - anatomie pathologique
  - échographie
  - radiographie
  - scintigraphie
- étiologie
  - complications
  - congénital
  - embryologie
  - génétique
  - immunologie
  - induit chimiquement
  - microbiologie
    - virologie
  - parasitologie
- histoire
- médecine vétérinaire
- organisation et administration
  - économie
- physiologie
  - génétique
  - immunologie
  - métabolisme
    - enzymologie
    - liquide céphalorachidien
    - sang
    - urine
  - physiopathologie
- psychologie
- statistiques et données numériques
  - épidémiologie
    - ethnologie
    - mortalité
- thérapie
  - chirurgie
  - diététothérapie
  - prévention et contrôle
  - radiothérapie
  - rééducation et réadaptation
  - soins infirmiers
  - traitement médicamenteux

- toutes les ressources / seulement les principales
- sans explosion

- tous les types
- recommandations professionnelles
- documents concernant l'enseignement
- documents concernant les patients
- documents SIGAPS A
- documents SIGAPS A ou B

Three main terminology servers in health

- UMLS URL: https://uts.nlm.nih.gov/home.html
  - NIH, Bethesda (USA)
  - More than 150 T/O
  - Mainly in English
  - The international reference for dissemination, but not for consultation

- BioPortal* URL: bioportal.bioontology.org/
  - NCBO, Stanford (USA)
  - More than 500 T/0 (a lot in biology, with few hundred concepts)
  - Mainly in English (not crosslingual)
  - The reference to post and display an ontology

- HeTOP* URL: www.hetop.eu
  - SIBM, Rouen, Normandy (France)
  - 69 T/0 in 32 languages
  - The crosslingual reference (navigation between languages) and in French

*_Grosjean J et coll._ An Approach to Compare Bio-Ontologies Portals. **Stud Health Technol Inform**, 2014;205:1008-1012.

HeTOP: main figures

May 2010

Terminologies & ontologies	Concepts	Synonymes	Définitions	Relations & hiérarchies
25	> 580 000	> 840 000	> 220 000	> 1 200 000

May 2011

Terminologies	Concepts	Synonymes	Définitions	Relations
32	> 980 000	> 2 300 000	> 220 000	> 4 000 000

April 2013

Terminologies	Concepts	Synonymes	Définitions	Relations
45	≈ 1 620 000	≈ 3 700 000	≈ 220 000	≈ 5 500 000

October 2015

Terminologies	Concepts in English	Concepts in French	Synonyms	Definitions	Relations
69 (17 UMLS)	1,743,772	1,031,230	8,611,170	278,687	9,862,198

Main figures

Registered users	**> 2 200**
traffic	**15 000 hits/day** **(600 users per working day)**

Terminologies in French that are not included in UMLS

- Overall, number of distinct CUI with at least one French translation in HeTOP
  ≈ 333,000 vs. ≈ 88,000 in UMLS
  (x3.68)

- 108 millions of RDF triplets (big data in health) in 2014

HeTOP relationships (examples & numbers)

	Source Term (Terminology)	Target Term (Terminology)	Number of relations in HeTOP
UMLSalignment	*Myocardial Infarction* **(MeSH)**	*Myocardial infarction, NOS* **(SNOMED Int)**	*644,982*
CISMeFmanual	*Riedel thyroiditis* **(HRDO)**	*Riedel's thyroiditis* **(MedDRA)**	*41,673*
CISMeFexact	*appetite stimulants* **(ATC)**	*Appetite stimulated* **(WHOART)**	*653,709*
CISMeFSupervised	*Gonadotropin releasing hormone* **(MeSH)**	*Luteotropin-releasing factor* **(FMA)**	*251,995*

Not an exact match

HeTOP limits

- Formal representation of complex clinical data structures = none
- Formal representation of physiological models = none
- Temporal relations = none
- Data quality = based on T/0 quality and point of view
- Formalism & reasoning capabilities = none
- Collaborative editing/searching/sharing tools = collaboration with BioPortal to share tools (Clement Jonquet)
- T/O versioning = not yet provided by HeTOP
- Semantic resources distribution/dissemination processes = 69 T/0 avalable in OWL format (latest version)/SKOS/RDF in several languages

Other tools integrated to HeTOP

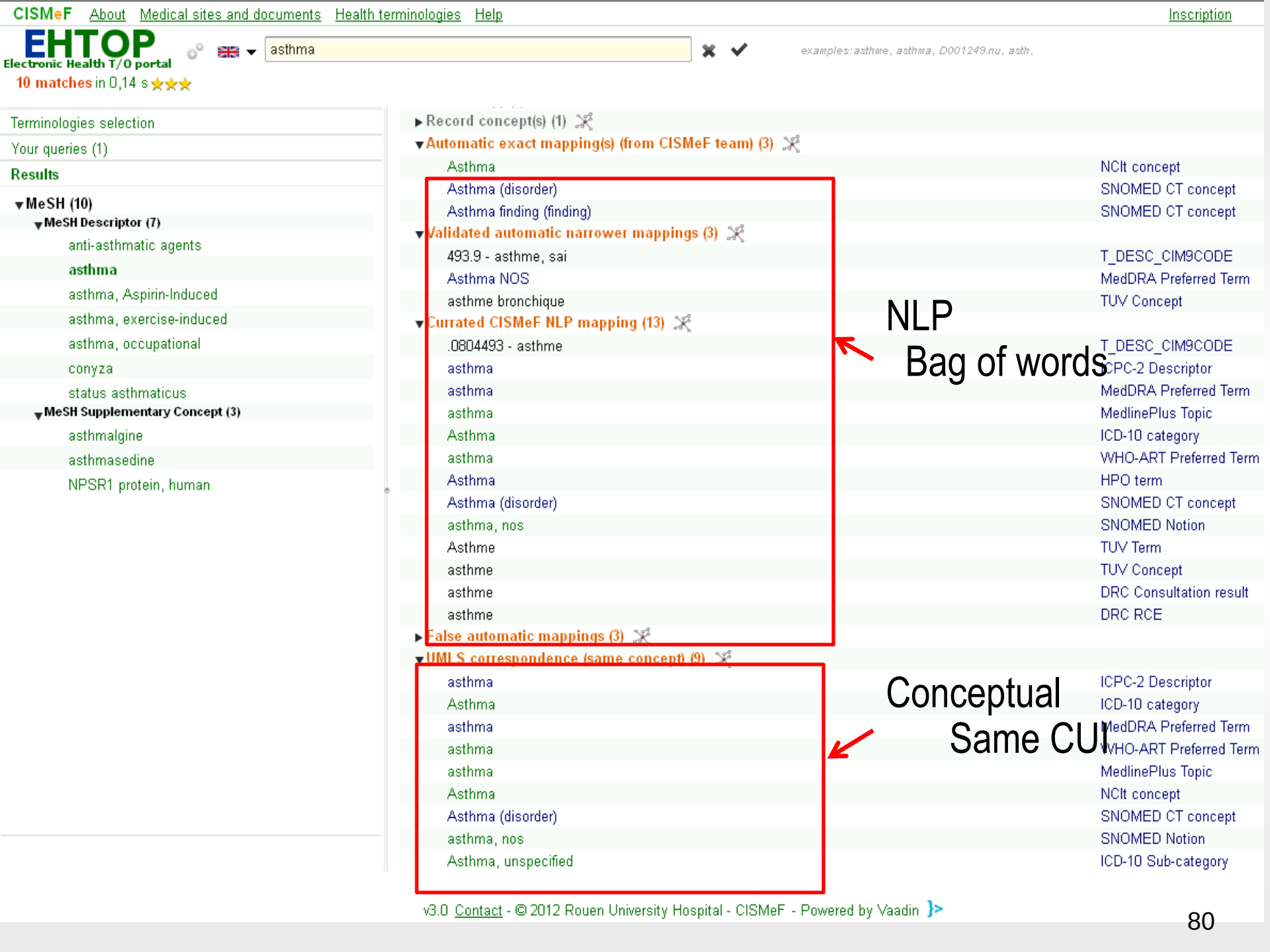
- ECMT Extracteur de Concepts Multi Terminologiques
  - Able to extract health concepts from any text; e.g. discharge summary in ½ second (NoSQL)
  - Valorization with Alicante SME
  - Used in daily practice in the Catholic University Hospital of Lille, France; Dr. Arnaud Hansske; around one million discharge summaries indexed with ECMT
- InfoRoute, a French InfoButton
  - URL: inforoute.chu-rouen.fr
  - Access to a contextualized knowledge based on semantic expansion based on manual & supevised mapping among terminologies
- MT@HeTOP, tool to perform automatic mappings & translations
- Generic semantic search engine
  - Doc'CISMeF (URL: doccismef.chu-rouen.fr) on grey literature about health in French on the Internet (10^5 resources)
  - LISSA (URL : www.lissa.fr), a PubMed in French (0,7 x 10^6 citations d'articles)
  - RIDOPI, search engine in EHR (8 x 10^6 discharge summaries in Rouen; around 10^9 health concepts in these summaries; 10^8 numerical data in Rouen)

Semantic harmonization: mapping, alignment

Three methods employed

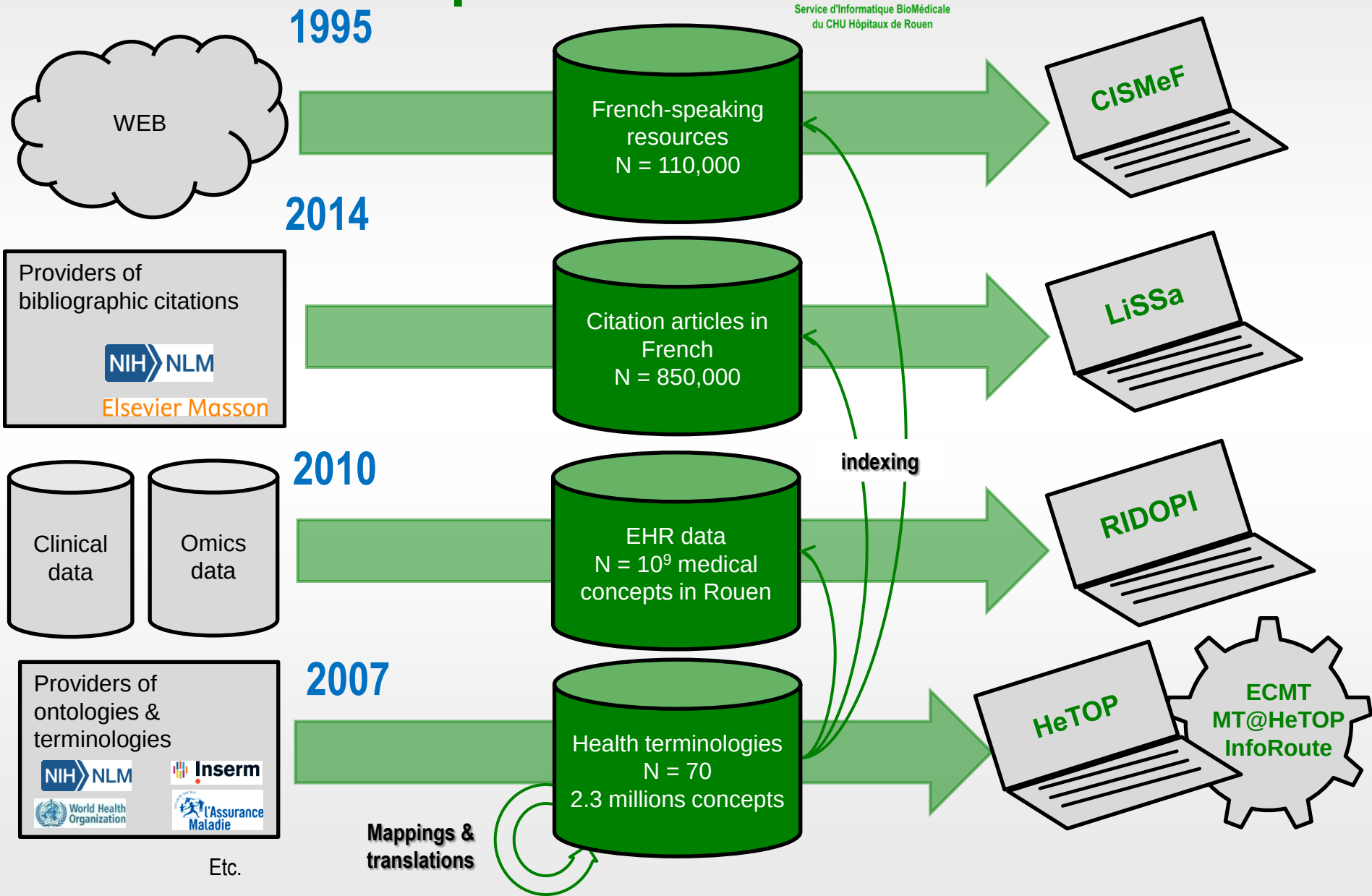
URL: http://cispro.chu-rouen.fr/MT_EHTOP/

- Conceptual
  - Same CUI
  - Other relations: close match, BT-NT, NT-BT (SKOS)
  - On UMLS (n=12 included in HeTOP)
- NLP
  - More or less same algorithm of automatic indexing
  - Bag of words
  - on (N*N-1)/2 T/O (included in the HeTOP)
- Statitistical
  - Co-occurrence matrix
  - CCAM-ICD10; CCAM-LPP


CISMeF About Medical sites and documents Health terminologies Help Inscription
EHTOP
Electronic Health T/O portal
asthma
examples: asthme, asthma, D001249.nu, asth,
10 matches in 0,14 s
Terminologies selection
Your queries (1)
Results
MeSH (10)
MeSH Descriptor (7)
anti-asthmatic agents
asthma
asthma, Aspirin-Induced
asthma, exercise-induced
asthma, occupational
conyza
status asthmaticus
MeSH Supplementary Concept (3)
asthmalgine
asthmasedine
NPSR1 protein, human
Record concept(s) (1)
Automatic exact mapping(s) (from CISMeF team) (3)
Asthma NCIt concept
Asthma (disorder) SNOMED CT concept
Asthma finding (finding) SNOMED CT concept
Validated automatic narrower mappings (3)
493.9 - asthme, sai T_DESC_CIM9CODE
Asthma NOS MedDRA Preferred Term
asthme bronchique TUV Concept
Currated CISMeF NLP mapping (13)
.0804493 - asthme T_DESC_CIM9CODE
asthma ICPC-2 Descriptor
asthma MedDRA Preferred Term
asthma MedlinePlus Topic
Asthma ICD-10 category
asthma WHO-ART Preferred Term
Asthma HPO term
Asthma (disorder) SNOMED CT concept
asthma, nos SNOMED Notion
Asthme TUV Term
asthme TUV Concept
asthme DRC Consultation result
asthme DRC RCE
False automatic mappings (3)
UMLS correspondence (same concept) (9)
asthma ICPC-2 Descriptor
Asthma ICD-10 category
asthma MedDRA Preferred Term
asthma WHO-ART Preferred Term
asthma MedlinePlus Topic
Asthma NCIt concept
Asthma (disorder) SNOMED CT concept
asthma, nos SNOMED Notion
Asthma, unspecified ICD-10 Sub-category
NLP
Bag of words
Conceptual
Same CUI
v3.0 Contact - © 2012 Rouen University Hospital - CISMeF - Powered by Vaadin

Knowledge Engineering platform of SiBM

Service d'Informatique BioMédicale du CHU Hôpitaux de Rouen



Ce document a été réalisé par la Cellule TICE Médecine de la Faculté de Médecine de Rouen (Courriel : Francoise.Charles@univ-rouen.fr).

An ontology in practice

- A differential concepts tree (at level ②)
- A formal concepts lattice (at level ③)
- A relations tree (objectProperty)
- Data (dataProperty)
- Annotations specific to each concept
- And all representations (“necessary”, defined (NSC, ...) which can be built with