

Introduction aux ontologies biomédicales

École d'été interdisciplinaire en numérique de la santé (EINS 2024)

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Plan

- **Partie I :** Les ontologies comme solution au problème de Babel de l'informatique médicale
- **Partie II :** Les différents niveaux d'ontologies
- **Partie III :** Définitions textuelles et formelles

Application à la classification : l'exemple de la maladie



Partie 1 : Les ontologies comme solution au problème de Babel de l'informatique médicale



Les données médicales au Québec aujourd'hui

- Cabinets de médecin
- Hôpitaux
- RAMQ
- Centre Local de Service Communautaire (CLSC)
- Ministère de la Santé et des Services Sociaux (MSSS)
- Cohortes
- Essais cliniques
- Biobanques
- ...



Tour de Babel des systèmes
d'information

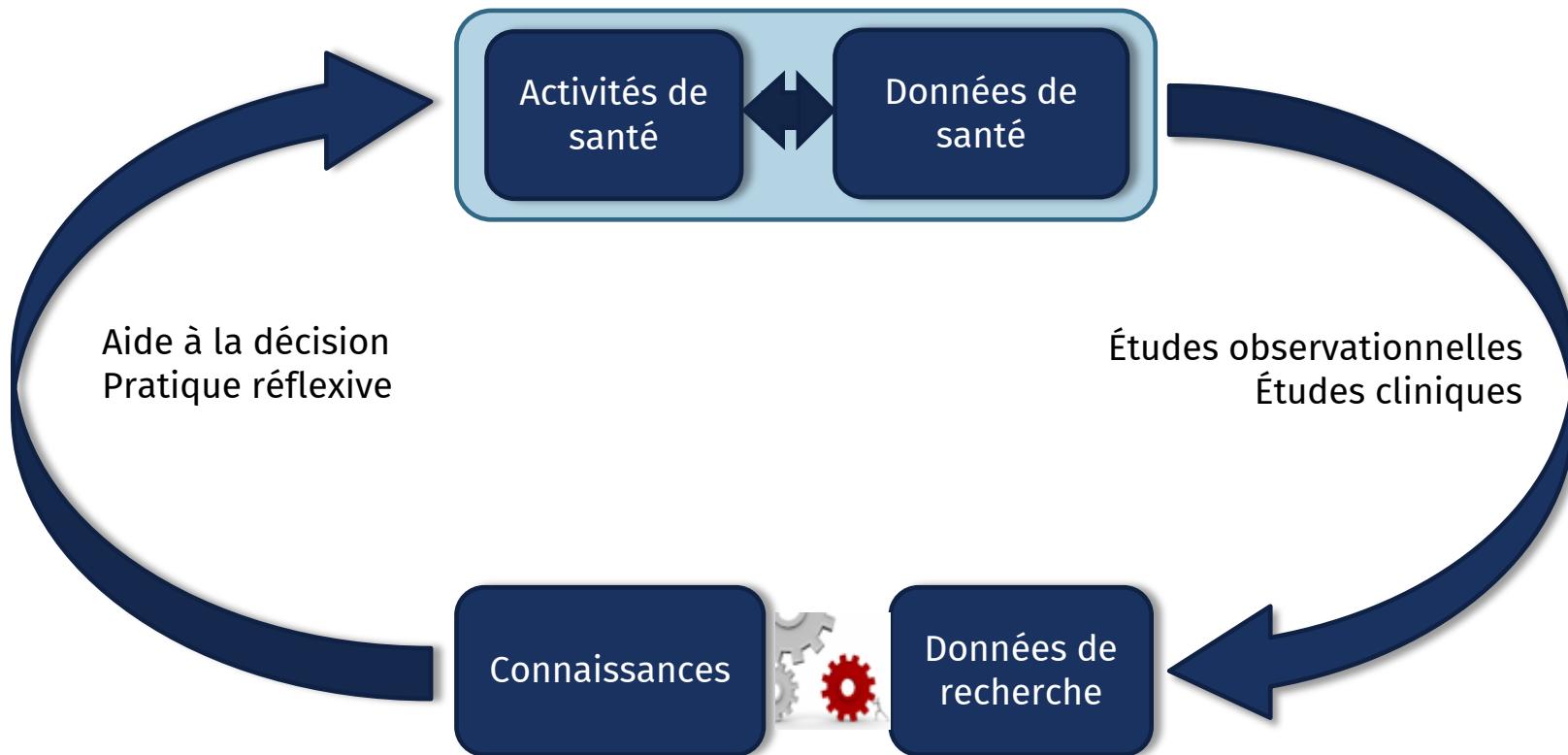
Les données médicales au Québec aujourd’hui

- Défi : rendre l’information scientifique interopérable, partageable, réutilisable
- Obstacle principal : diversité des données, plutôt que leur quantité
- “Problème de Babel” :
 - Idiosyncrasie technologique
 - Idiosyncrasie humaine



Tour de Babel des systèmes d’information

Un défi pour la mise au point de systèmes de santé apprenants



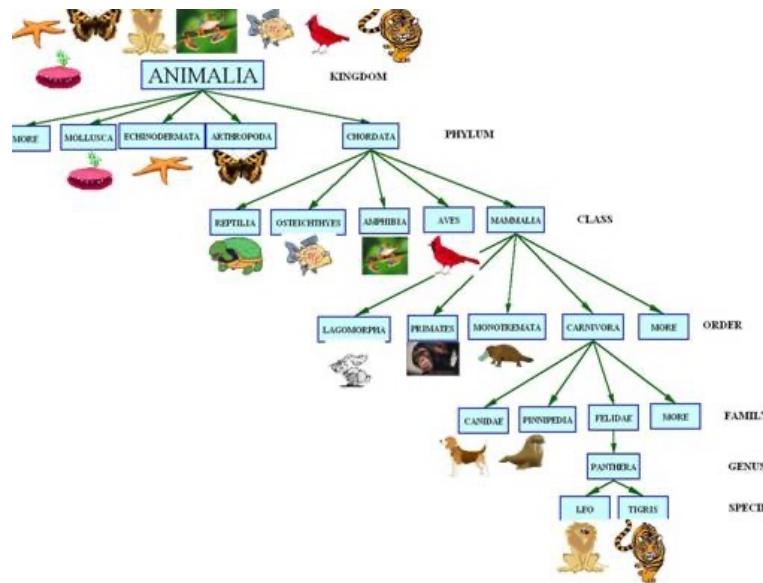
Approche du GRIIS : Utiliser les ontologies



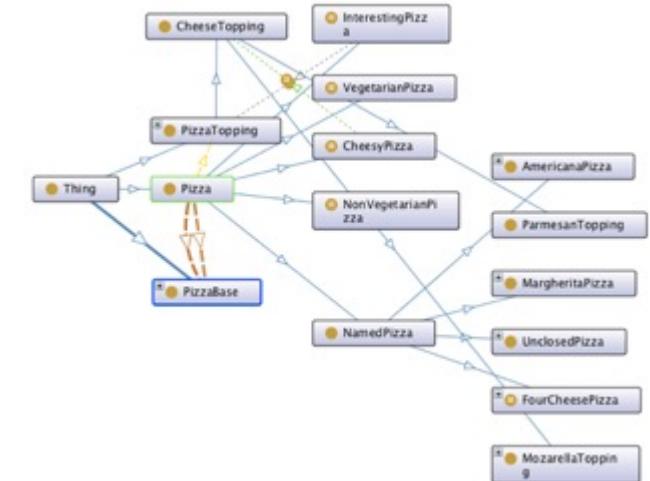
Terminologies, taxonomies et ontologies

Medicines and Drugs	
Term	Definition
abortifacients	used to stimulate uterine contraction and promote evacuation of the uterus to cause abortion
ACE inhibitor	used to block the enzyme responsible for converting Angiotensin 1 to angiotensin 2 in the lungs; this blocking prevents vasoconstriction
antiarrhythmics	this affects the action potential of cardiac cells and are used to treat arrhythmias and return normal rate and rhythm of the heart muscles
anticoagulant	drugs that inhibit any step of the coagulation process, preventing or slowing clot formation

Terminologie



Taxonomie

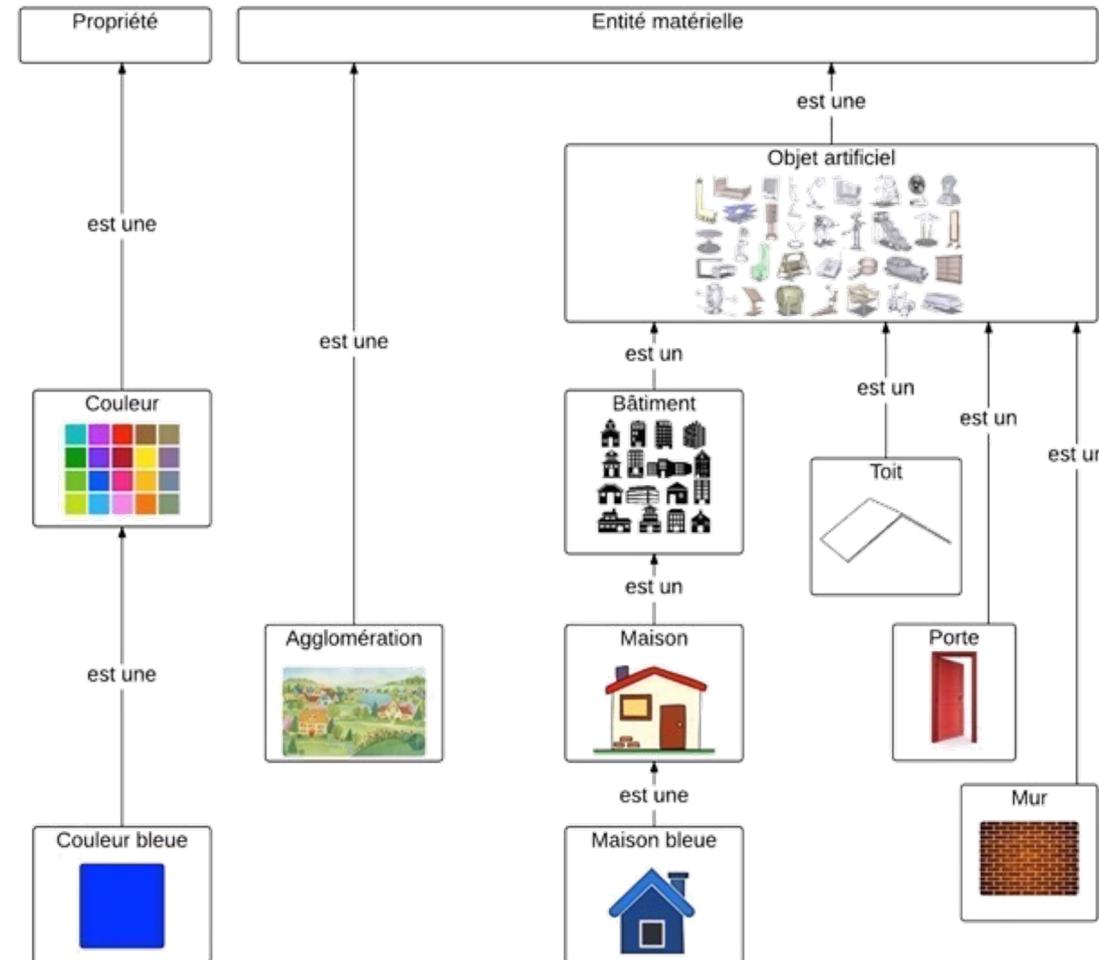


Ontologie

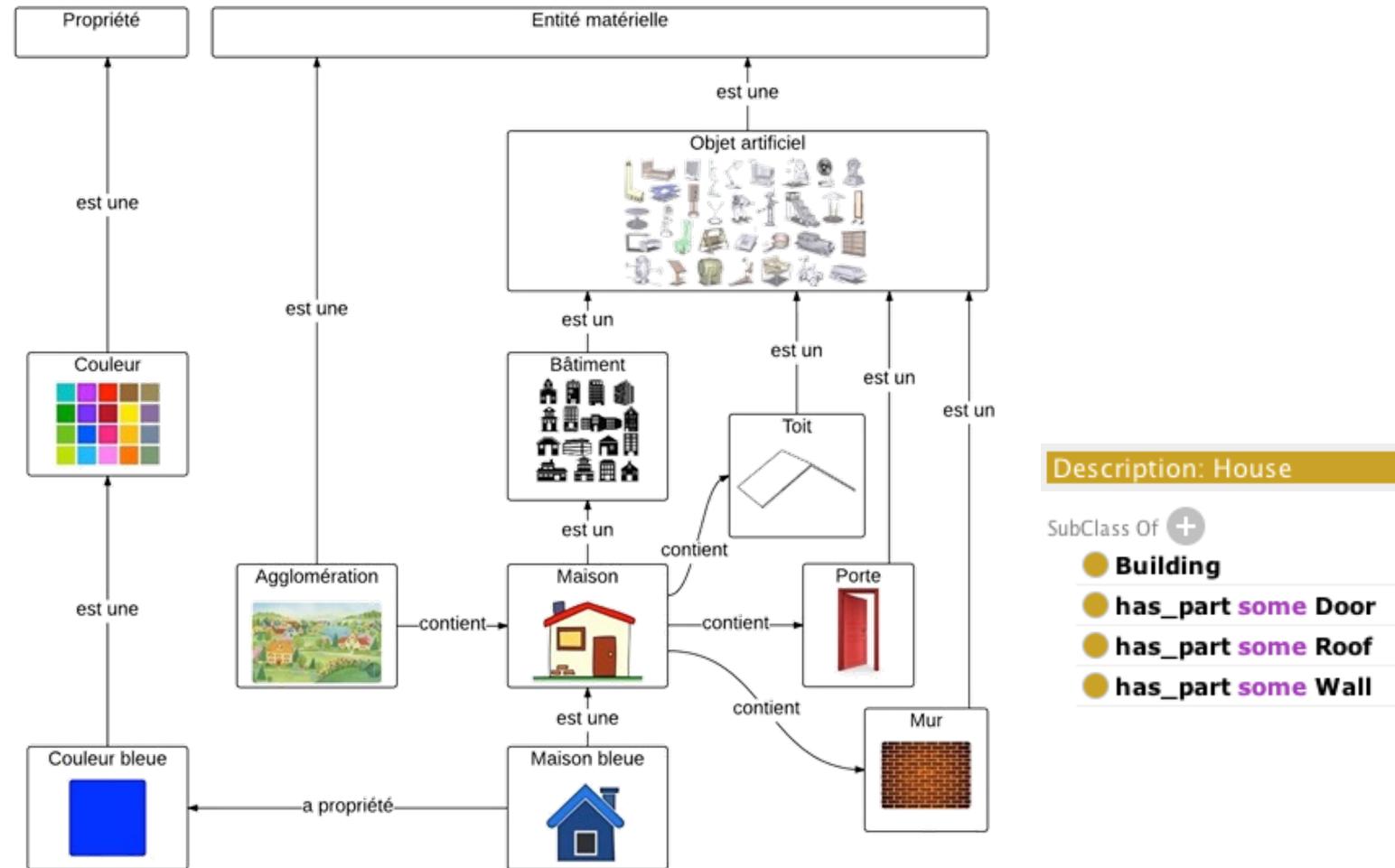
Terminologie

Terme	Définition
Maison	Bâtiment destiné à servir d'habitation à des humains.
Toit	Surface supérieure d'un bâtiment.
Porte	Ouverture limitant un espace clos, permettant la communication entre cet espace et ce qui est extérieur à cet espace.
Mur	Ouvrage de maçonnerie vertical (parfois oblique), élevé sur une certaine longueur pour constituer le côté d'un bâtiment.

Taxonomie



Ontologie



Ontologie de domaine

Représentation canonique des entités et relations d'un domaine donné, visant à l'exhaustivité.

Exemple : Cardiovascular Disease Ontology (CVDO)

Définition formelle de la sténose aortique valvulaire (OWL)

The screenshot displays the Protégé ontology editor interface. At the top, a yellow bar shows the description: 'aortic valve stenosis'. Below it, the 'Equivalent To' section contains the OWL definition:

```
disease
and ('realized in' some
('disease course'
and ('has occurred part' some 'aortic valve improper opening during left ventricular systole'))
and ('has material basis at all times' some 'aortic valve disorder'))
```

The middle section, titled 'Hiérarchie de classes', shows the class hierarchy starting from 'Thing' and leading to 'aortic valve stenosis' and its subtypes.

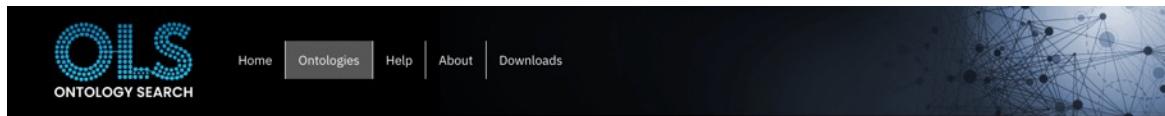
```
Thing
|--- Nothing
|--- entity
|   |--- continuant
|   |   |--- 'generically dependent continuant'
|   |   |--- 'independent continuant'
|   |   |--- 'specifically dependent continuant'
|   |   |--- quality
|   |   |--- 'realizable entity'
|   |   |--- disposition
|   |   |--- disease
|   |   |   |--- 'aneurysm (disease)'
|   |   |   |--- 'aortic disease'
|   |   |   |--- 'aortic valve disease'
|   |   |   |--- 'aortic valve insufficiency'
|   |   |   |--- 'aortic valve prolapse'
|   |   |--- 'aortic valve stenosis'
|   |   |   |--- 'rheumatic aortic valve stenosis'
|   |   |   |--- aortitis
|   |   |   |--- 'arrhythmogenic right ventricular dysplasia'
|   |   |   |--- 'arterial occlusive disease'
|   |   |   |--- 'Barlow's syndrome'
|   |   |   |--- 'cardiac tamponade'
```

The right side, titled 'Quelques relations', lists various relations:

- 'has location at some time'
- 'has material basis at all times'
- 'has occurred part'
- 'has participant at some time'
- 'has profile'
- 'has spatial occupant at some time'
- 'has spatiotemporal occupant'
- 'has specific dependent at some time'
- 'located in at some time'
- 'material basis of at some time'
- 'occupies spatial region at some time'
- 'occupies spatiotemporal region'
- 'occurs in'
- 'part of continuant at some time'
- 'part of occurrent'
- 'participates in at some time'
- 'process profile of'
- 'projects onto spatial region at some time'
- 'realized in'
- 'realizes'
- 'spatial projection of spatiotemporal at some time'
- 'specifically depends on at some time'

Vues d'écran du logiciel Protégé

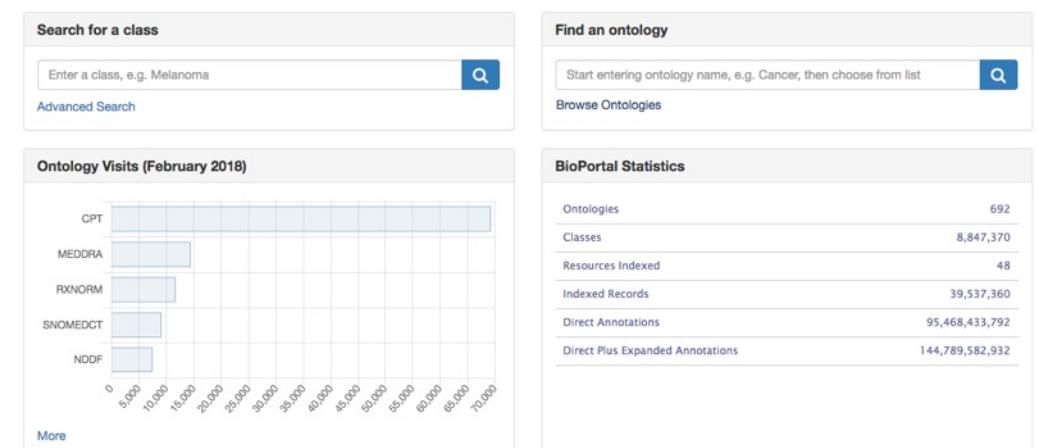
Moteurs de recherche pour ontologies



Ontology	ID	Description
Alzheimer's Disease Ontology (ADO)	ADO	Alzheimer's Disease Ontology is a knowledge-based ontology that encompasses varieties of concepts related to Alzheimer's Disease, fundamentally structured by upper level Basic Formal Ontology (BFO). This Ontology is enriched by the interrelational entities that demonstrate the network of the understanding on Alzheimer's disease and can be readily applied for text mining.
African Population Ontology	AFPO	AFPO is an ontology that enables annotating African individuals and combines knowledge accumulated about existing populations with their genetic fingerprints in a standardized format. The AFPO can be employed to classify African study participants comprehensively in prospective research studies. It can also be used to classify past study participants by mapping them using a language or ethnicity identifier or synonyms.
Agronomy Ontology	AGRO	Agro is an ontology for representing agronomic practices, techniques, variables and related entities

The screenshot shows the Ontobee interface. At the top is a dark header with the Ontobee logo and a network graph icon. Below it is a navigation bar with links for Home, Intro, Statistics, SPARQL, Ontobee, Annotator, Tutorial, FAQs, References, Links, Contact, Acknowledge, and News. A search bar with the placeholder "Welcome to Ontobee!" is positioned below the navigation. Below the search bar is a paragraph of text about Ontobee. Further down are input fields for "Please select an ontology (optional)" and "Keywords:", a "Search terms" button, and a "Batch Search" button. There is also a "Jump to http://purl.obolibrary.org/obo/" input field and a "Go" button. A table titled "Currently Ontobee has been applied for the following ontologies:" lists three ontologies: ADO, AEO, and AFO, each with its full name, OBO status (L or N), and a small icon.

Welcome to BioPortal, the world's most comprehensive repository of biomedical ontologies



Les ontologies comme légendes pour les bases de données

GlyProt

GOL	Glycoside-amino acid ligase
SMO2	Smoothened (cycling) 1
Mnk1-A	Nucleophosmin Mnk1-A
Erythrocyte membrane S100 glycoprotein	
C24	Carnitine acyltransferase II
NF1N	Nuclear factor 1/X
WORMPROTEIN_RN-45	
Biosat1	Protein biosat1
ELAMON16	ELAMON16 gene, partial cds
ELM1	Elastatin 1, neutrophil
M60	M60 glycoprotein
CNTS	Cystatin C
AHHS	Ahhs homolog, member 6 (rho 6)
ANX1	Anxinin 1 (lipocortin 1)
CNSR	
SP11	SP11 integration exonease
D	D component of complement
NP-115-sets	
BLB-Related Proteins	
MM02	Alpha-amidomodidase 2, incyte
BB	
BB-1	Binding suppressor-of-white-apricot
CG1	Antigen (p51)
Myo1A	Myo1A (M1B)
SPRR1B	Small proline-rich protein 1B
ACTN1	Actinin alpha 2
ELAM0006	ELAM0006 gene, partial cds
DYKE	Dicarboxylyl dipeptide kinase
Carrilactoprotin	Carrilactoprotin antigen precursor
LTS	Lysophosphatidic-acid-beta
GNAT3	GNAT-binding protein 3
GB_SF7	Escherichia coli unknown
GUAYLATE_CICLOSE	ECM-1
Spinal Muscular Atrophy 4	
ELAM0026	ELAM0026 gene, partial cds
Batrachosiphon (BTF4)	
MEC-exoceted proteases LAMP7-EL	
NP11	Replication protein 11
Clone 22	NP11
CNS	CNS synthetase
UNP0007-LIKE PROTEIN_N01	
[NP11]	NP11
S100	S100 calcium-binding protein A12
SLK	Protein-tyrosine kinase slk
DB	(B5)
CPV-1	
TBL	
CDS	CDS antigen
Skeletal muscle abundant protein	

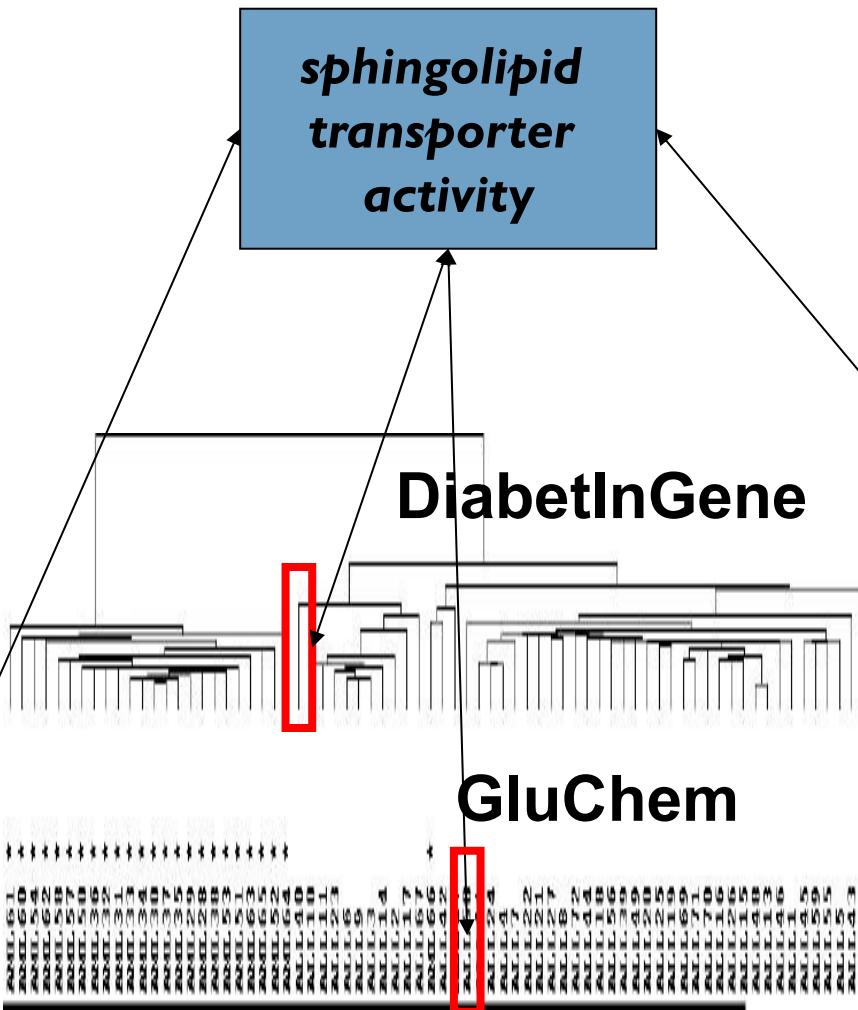
MouseEcotope

Tool	Statistical model	Correction for multiple experiments	GO Visualization	Microarray supported	Time to process 200 genes (s)
Osteo-Express	χ^2 , binomial, Hypergeometric, Fisher's exact test	Sabatti, Hoban, Boufahoum, FDR	Flat, Tree	172 commercial arrays (Affymetrix, SuperArray, Sigma-Genosys, ClonTech, PerSeptive Biosystems, Operon, Takara, NSI); can also upload a user-defined list	7, 8, 16, 28
Goldman	Fisher's exact test	Relative enrichment	Tree, DAG	Uploads from user	77, 128, 228, 340
DAVID	None			Not applicable	13, 17, 27, 54
EASEonline	Fisher's exact test	Boufahoum		Not available	27 arrays (Affymetrix only); can also upload a user-defined list
Geneledge	Hypergeometric	Boufahoum	Flat, no hierarchical structure	Uploads from user	6, 6, 6, 8
PathAssociate	Fisher's exact test			Uploads from user	22, 27, 29, 50
GOTM	Hypergeometric	None		Not available	58, 60, 157,
TwoGO	Percentage	Step-down used? FDR (Boguski and Rockwell, 1997), FDR (Boguski and Yeastock, 2001)	Flat, Tree	Uploads from user	13, 48, 69, 101
CLEUCH	Hypergeometric, χ^2	Binomial	DAG	Uploads from user	N/A
GOstat	χ^2 , Fisher's exact test	FDR, Hoban	Not available	Uploads from user	12, 26, 46, 57
GOstat-Bin	Hypergeometric, binomial, Fisher's exact test	Boufahoum, Hoban, Hochberg, Henneman, FDR	Not available	Uploads from user	22, 81, 145,
GoGol	χ^2	Binomial	DAG	** arrays (Affymetrix only)	9, 9, 9, 9
Ontology Taximeter	Hypergeometric	FDR	Not available	2 arrays (Affymetrix); uploads from user	N/A
eGOrilla	Binomial	None	Tree	Uploads from user	28, 42, 89, 95

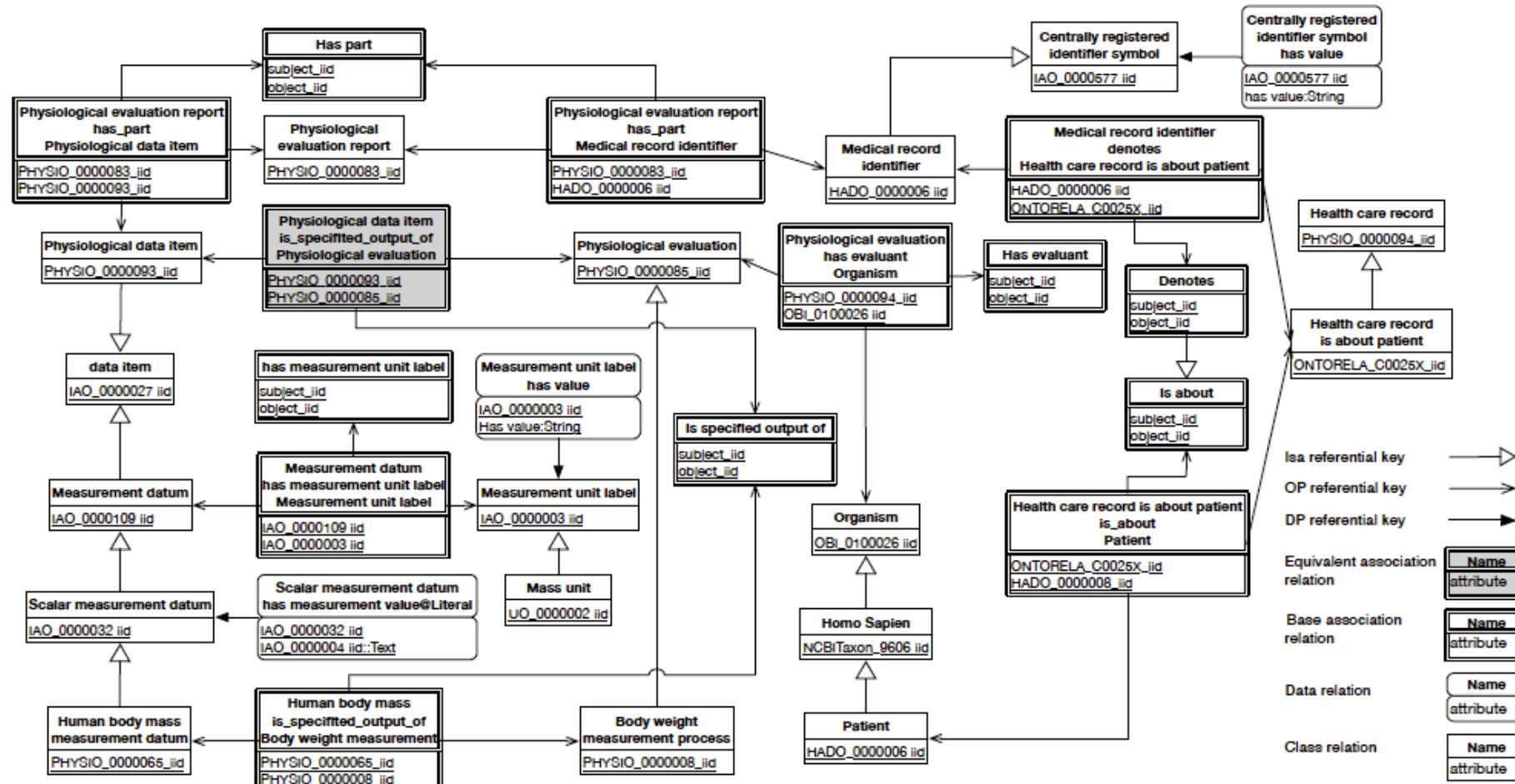
sphingolipid transporter activity

DiabetInGene

GluChem



Utilisation d'une ontologie pour créer un modèle de base de données relationnelle

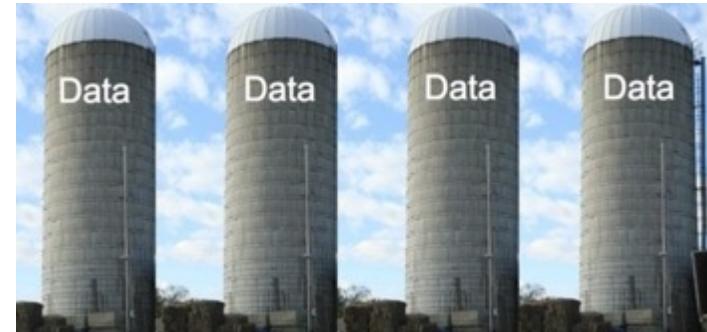


Partie 2 : Les différents niveaux d'ontologie



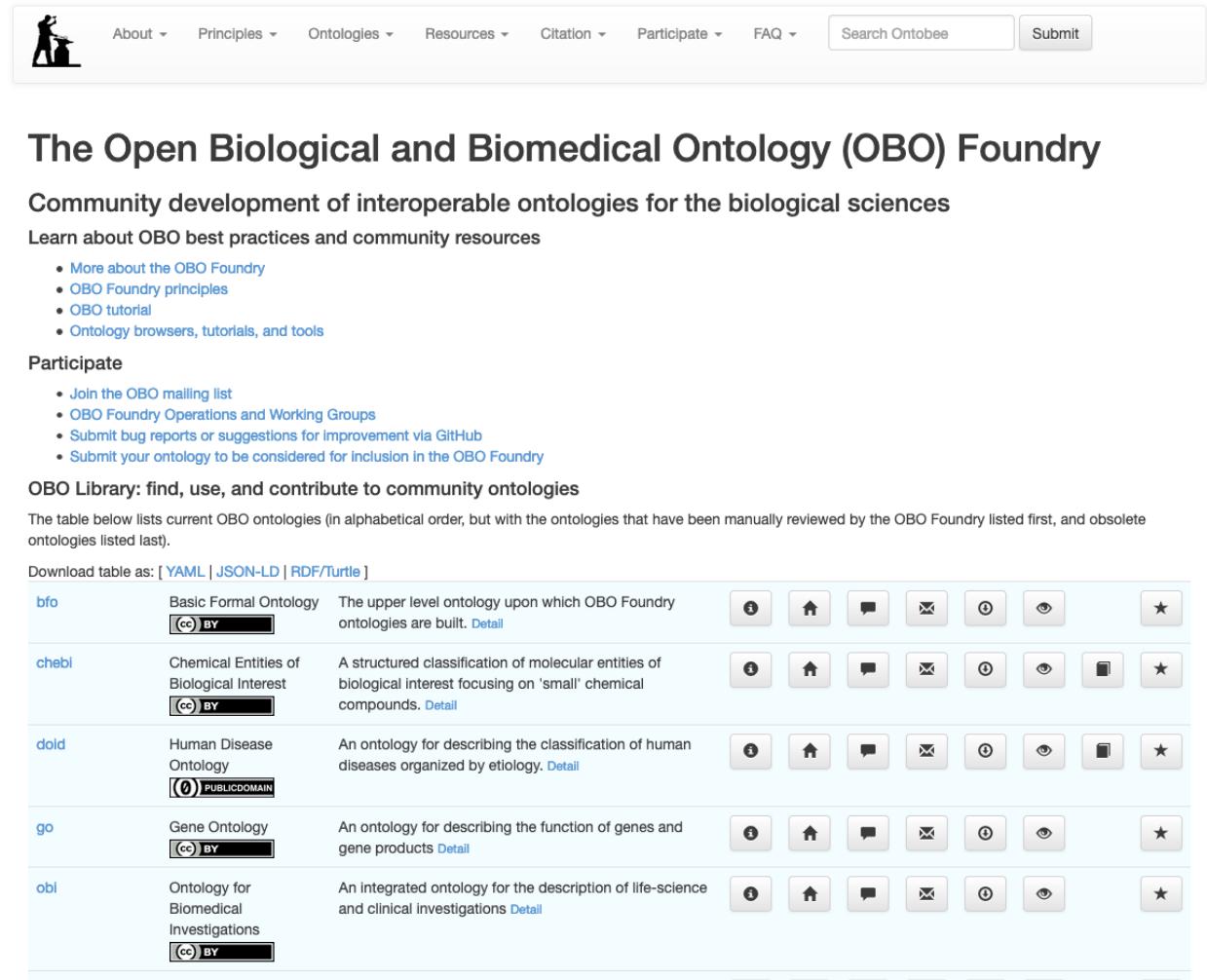
Difficultés

- Formation de silos de données d'un même domaine : la multiplication de terminologies et ontologies recrée le problème de l'interopérabilité à un plus haut niveau.
- La multiplicité des domaines représentés par des ontologies : médecine, biologie, industrie, géographie, éducation...



Solution

- Créer des ensembles d'ontologies de domaine :
 - avec une seule ontologie par domaine
 - unifiées par une ontologie de haut niveau.



The Open Biological and Biomedical Ontology (OBO) Foundry

Community development of interoperable ontologies for the biological sciences

Learn about OBO best practices and community resources

- More about the OBO Foundry
- OBO Foundry principles
- OBO tutorial
- Ontology browsers, tutorials, and tools

Participate

- Join the OBO mailing list
- OBO Foundry Operations and Working Groups
- Submit bug reports or suggestions for improvement via GitHub
- Submit your ontology to be considered for inclusion in the OBO Foundry

OBO Library: find, use, and contribute to community ontologies

The table below lists current OBO ontologies (in alphabetical order, but with the ontologies that have been manually reviewed by the OBO Foundry listed first, and obsolete ontologies listed last).

Download table as: [YAML | JSON-LD | RDF/Turtle]

ontology	description	status	actions
bfo	Basic Formal Ontology [CC BY]	The upper level ontology upon which OBO Foundry ontologies are built. Detail	
chebi	Chemical Entities of Biological Interest [CC BY]	A structured classification of molecular entities of biological interest focusing on 'small' chemical compounds. Detail	
doid	Human Disease Ontology [CC0 PUBLICDOMAIN]	An ontology for describing the classification of human diseases organized by etiology. Detail	
go	Gene Ontology [CC BY]	An ontology for describing the function of genes and gene products Detail	
obi	Ontology for Biomedical Investigations [CC BY]	An integrated ontology for the description of life-science and clinical investigations Detail	

Quatre types d'ontologies

- **Ontologie de haut-niveau** (ou formelle) : indépendante du domaine

Exemple: Basic Formal Ontology (BFO)

- **Ontologie de niveau intermédiaire** : utilisable par plusieurs ontologies de domaine (mais pas toutes).

Exemple: Ontology for General Medical Science (OGMS)

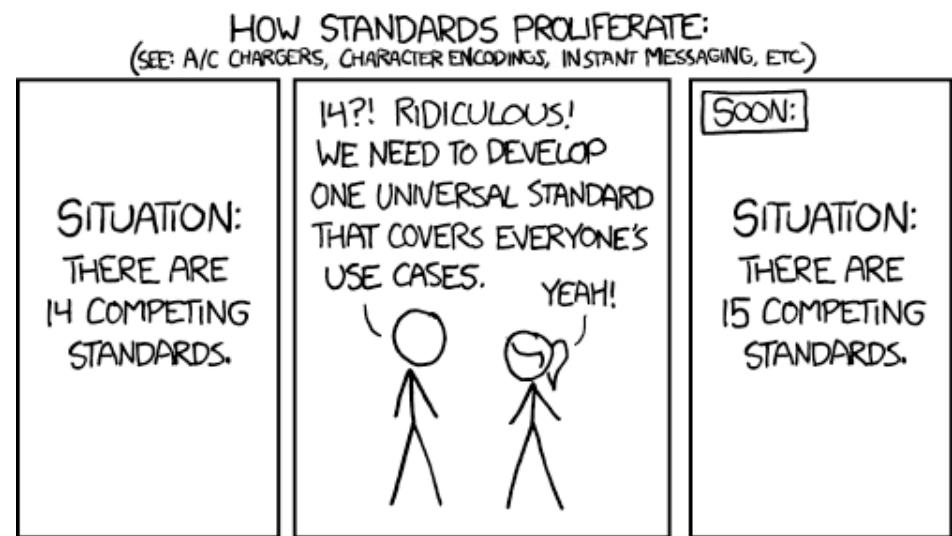
- **Ontologie de domaine** (ou de référence) : représentation canonique des entités et relations d'un domaine donné.

Exemple: Cardiovascular Disease Ontology (CVDO)

- **Ontologie d'application** : créée pour accomplir certains buts spécifiques.

Ontologies de haut niveau

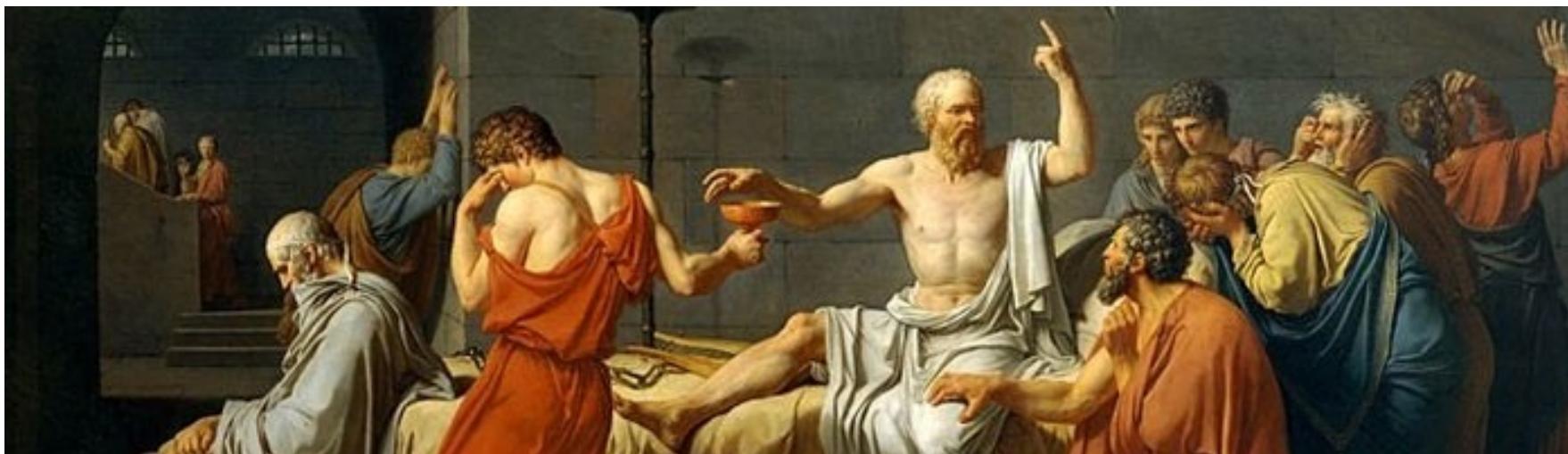
- Plusieurs ontologies de haut niveau proposées :
 - BFO (Basic Formal Ontology)
 - DOLCE (Descriptive Ontology for Linguistic and Cognitive Engineering)
 - UFO (Unified Foundational Ontology)
 - GFO (General Formal Ontology)
 - ...



<https://xkcd.com/927/>

Contexte interdisciplinaire

- La recherche de cohérence en développement d'ontologie a mené à des collaborations entre :
 - informaticiens et spécialistes des sciences de l'information
 - biologistes et cliniciens
 - mais aussi philosophes, logiciens, et linguistes.

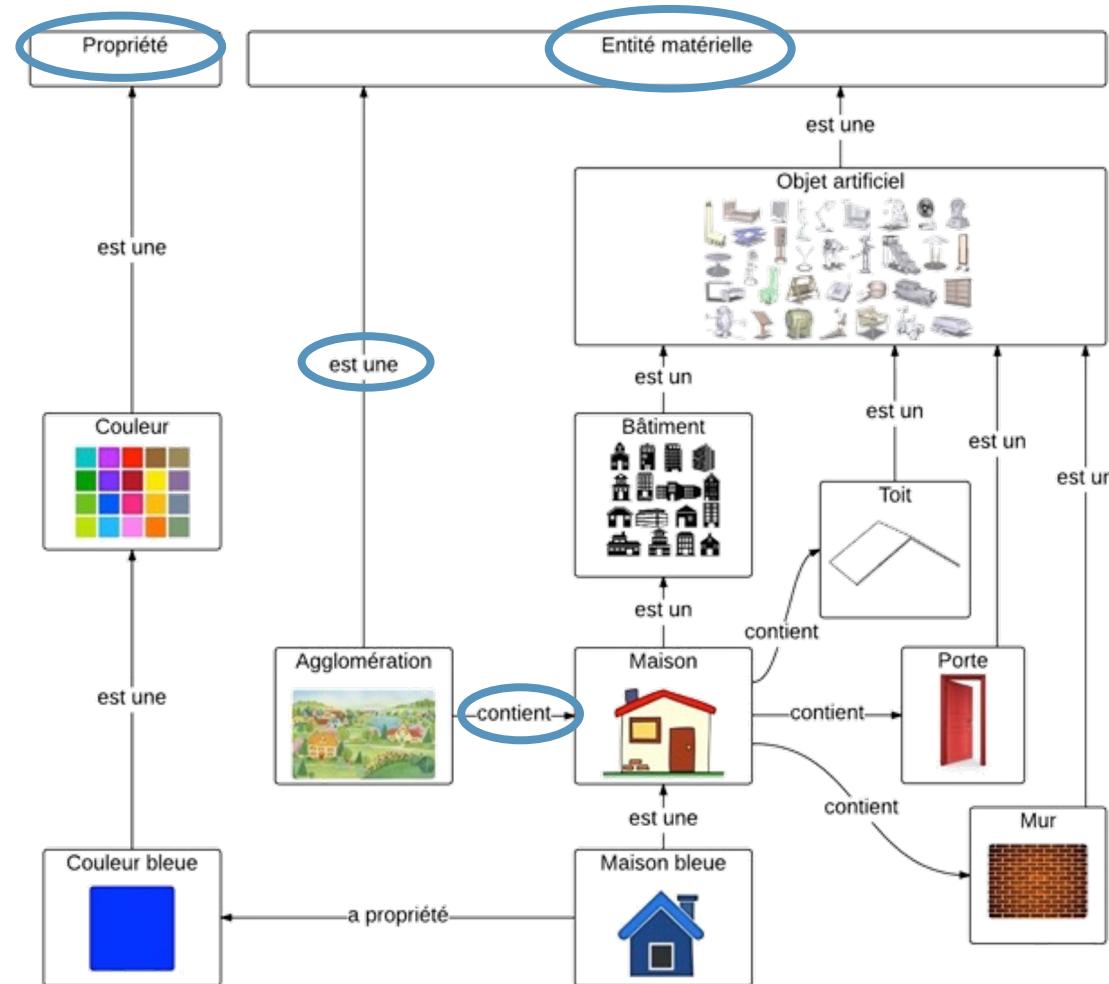


Pourquoi des philosophes ???

L'apport de la philosophie

Apports de la métaphysique :

- similarité
- identité et changement
- causalité
- méréologie (relation de tout à partie)
- ...



Apports de la philosophie des sciences :

- maladie
- risque
- probabilités
- ...

BFO (Basic Formal Ontology)

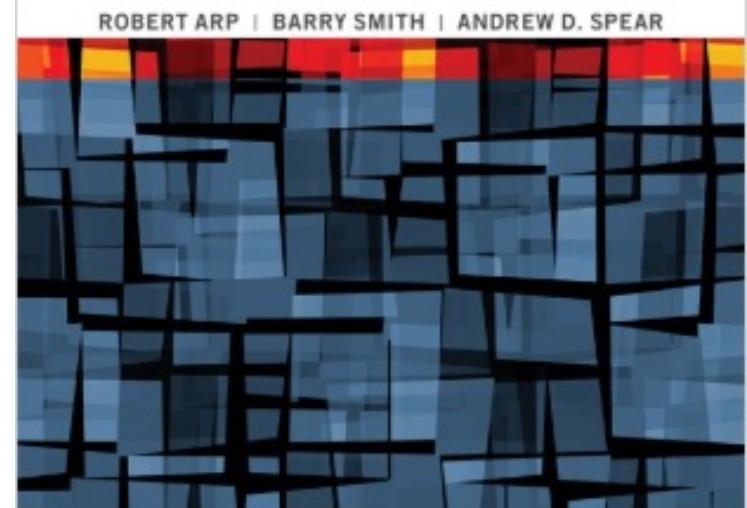
- Ontologie à méthodologie dite « réaliste » : vise à décrire le réel en s'alignant sur nos meilleures théories scientifiques.
- Développée par différentes institutions, notamment à l'université de Buffalo
- Ontologie très utilisée dans le domaine biomédical, de plus en plus dans le domaine industriel.
- Largement utilisée par les ontologies de la OBO Foundry



The Open Biological and Biomedical Ontology (OBO) Foundry

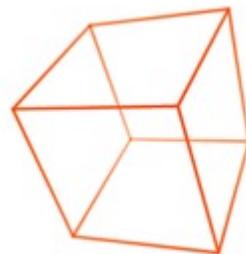
Community development of interoperable ontologies for the biological sciences

BUILDING ONTOLOGIES WITH
BASIC FORMAL ONTOLOGY



Continuants et occurrents

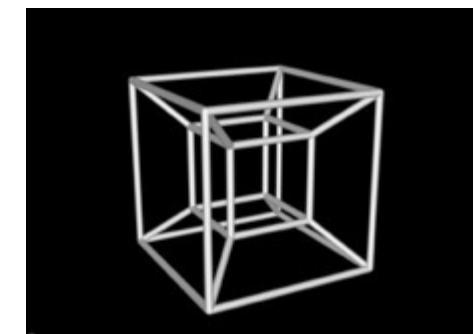
Continuants (perspective 3D)



Mitochondrie
Cellule
Organe
Organisme
Espèce
...

existent pleinement à tout instant de leur existence

Occurrents (perspective 4D)



Circulation sanguine
Division de cellule
Enfance
Croissance
...

n'existent pas pleinement à un instant : ils ont des parties temporelles

Continuants indépendants et dépendants Universaux et particuliers

	Continuant indépendant	Continuant dépendant	Occurrent
	<i>Humain</i> <i>Chat</i> <i>Table</i>	<i>Rougeur</i> <i>Forme rectangulaire</i> <i>Fragilité</i>	<i>Conférence</i> <i>Souper</i> <i>Circulation sanguine</i>

Continuants indépendants et dépendants

Universaux et particuliers

	Continuant indépendant	Continuant dépendant	Occurrent
Universaux / Classes	<i>Humain</i> <i>Chat</i> <i>Table</i>	<i>Rougeur</i> <i>Forme rectangulaire</i> <i>Fragilité</i>	<i>Conférence</i> <i>Souper</i> <i>Circulation sanguine</i>
Particuliers	cet humain ce chat cette table	cette rougeur cette forme rectangulaire cette fragilité	cette conférence ce souper cette circulation sanguine

Partie 3 : Définitions textuelles et formelles

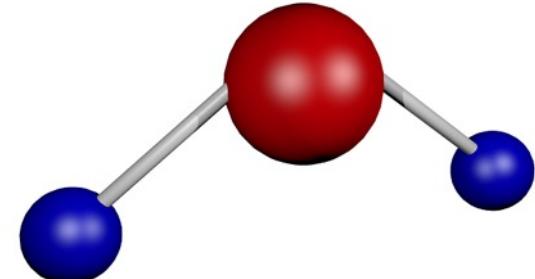


Le besoin de définitions textuelles : Eviter les imprécisions de pensée



Ambiguïté

- La “International Classification of Nursing Practice” (ICNP) définit "water" comme :
"A type of Nursing Phenomenon of Physical Environment with the specific characteristics: clear liquid compound of hydrogen and oxygen that is essential for most plant and animal life influencing life and development of human beings."



Circularité

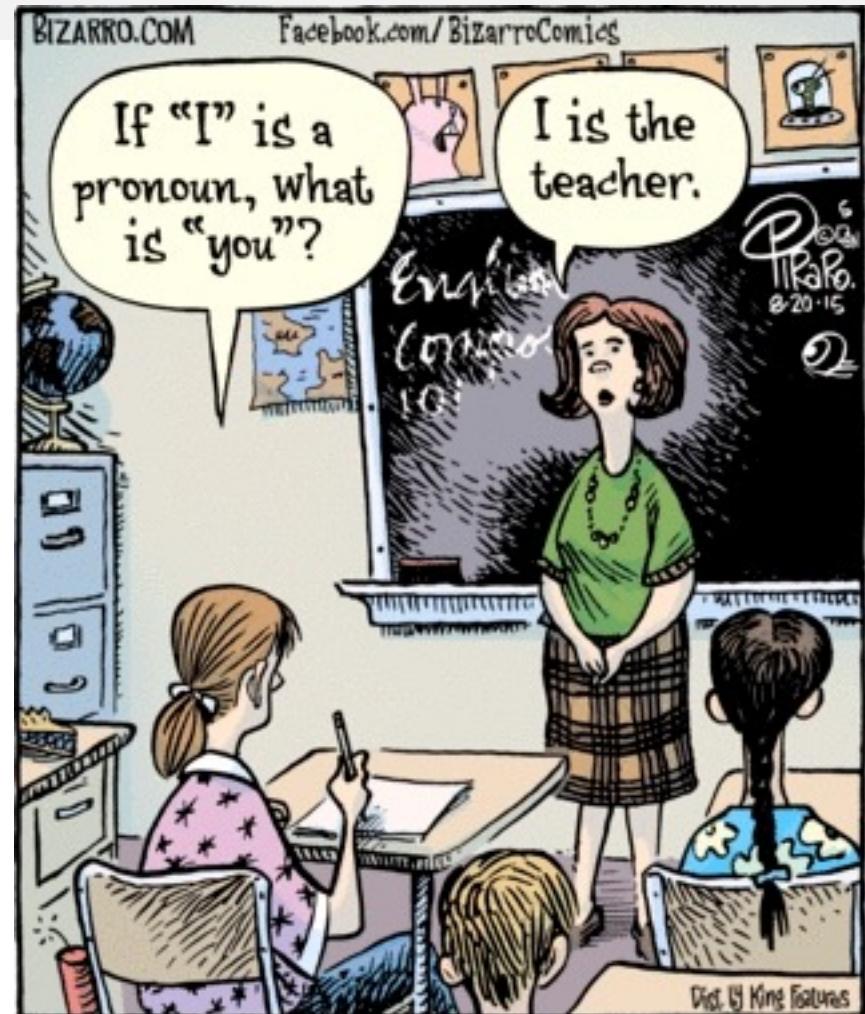
First Healthcare Interoperability Resources
Specification (FHIR) :

- Nourriture (“food”) défini comme “naturally occurring, processed, or manufactured entities that are primarily used as food for humans and animals.”
- Contenant (“container”) défini comme “a container of other entities”



Confusion “utilisation-mention” (“use-mention”)

- HL7 définit un sujet vivant (“living subject”) comme : “a subtype of Entity representing an organism or complex animal, alive or not.”
- Confond un objet (un sujet vivant) avec sa représentation (“Entity representing [...]”).
- E.g. : “Le sommeil est bon pour la santé et contient trois voyelles.”



Un modèle utile : la définition aristotélicienne

- Exemple historique chez Aristote :

Humain =_{def} Un animal qui est rationnel.

- Définition aristotélicienne d'une classe fille (“espèce” E) par sa classe parente (“genre” G), en la particularisant avec un “*differentia*” (D) :

E =_{def} Un G qui D.

Exemple (FMA) : Heart =_{def} An organ with cavitated organ parts, which is continuous with the systemic and pulmonary arterial and venous trees.



Ethology

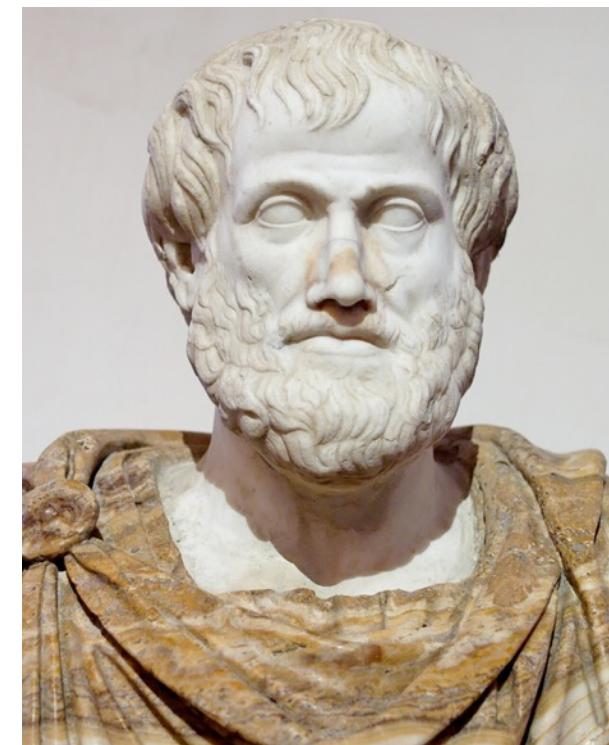
CURRENT ISSUES - PERSPECTIVES AND REVIEWS

Transitive or Not: A Critical Appraisal of Transitive Inference in Animals

David Guez* & Charles Audley†

* School of Psychology, The University of Newcastle, Newcastle, NSW, Australia

† Centre for Applied Psychology, Faculty of Health, University of Canberra, Canberra, ACT, Australia

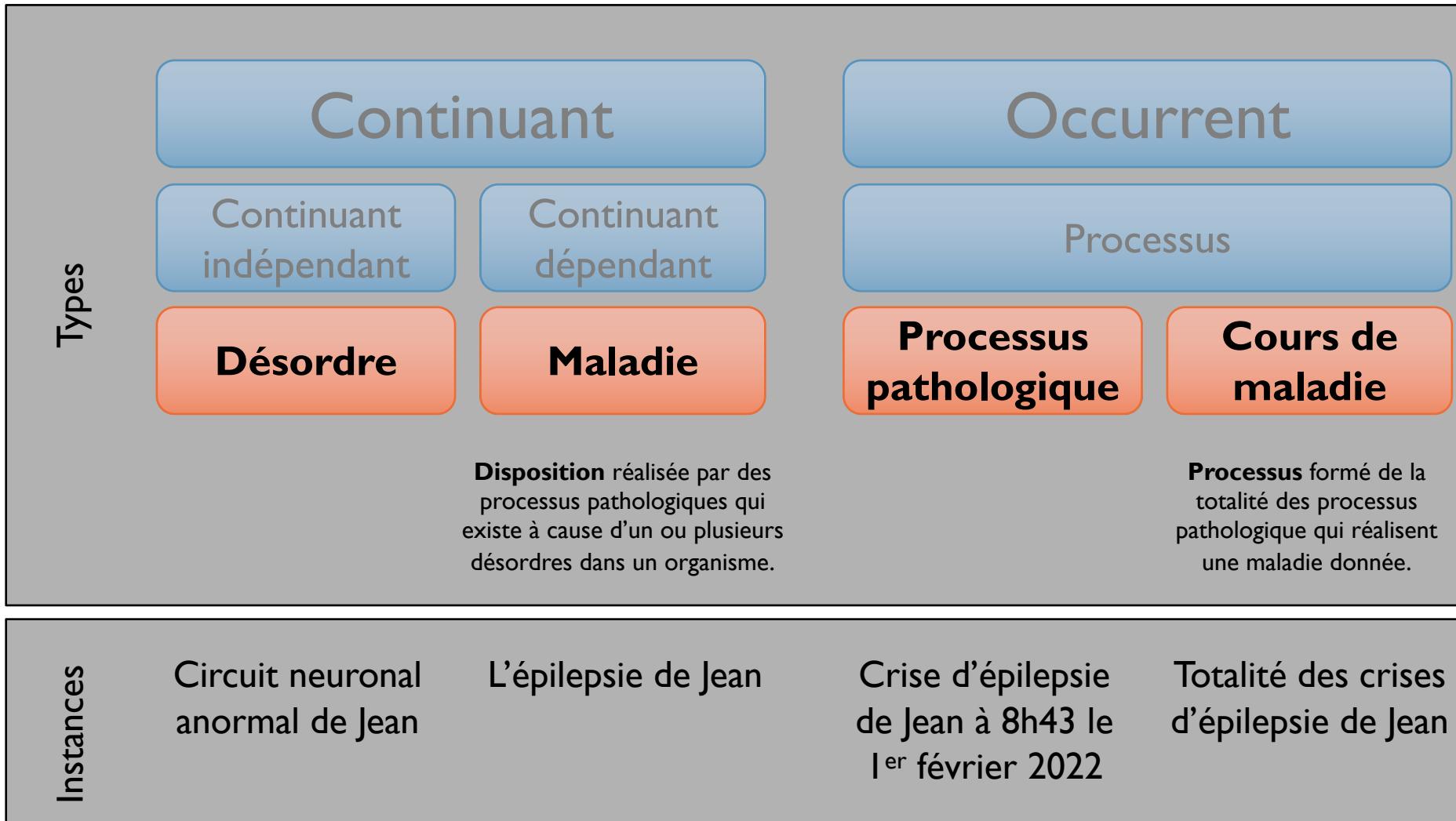


Application à la classification : l'exemple de la maladie

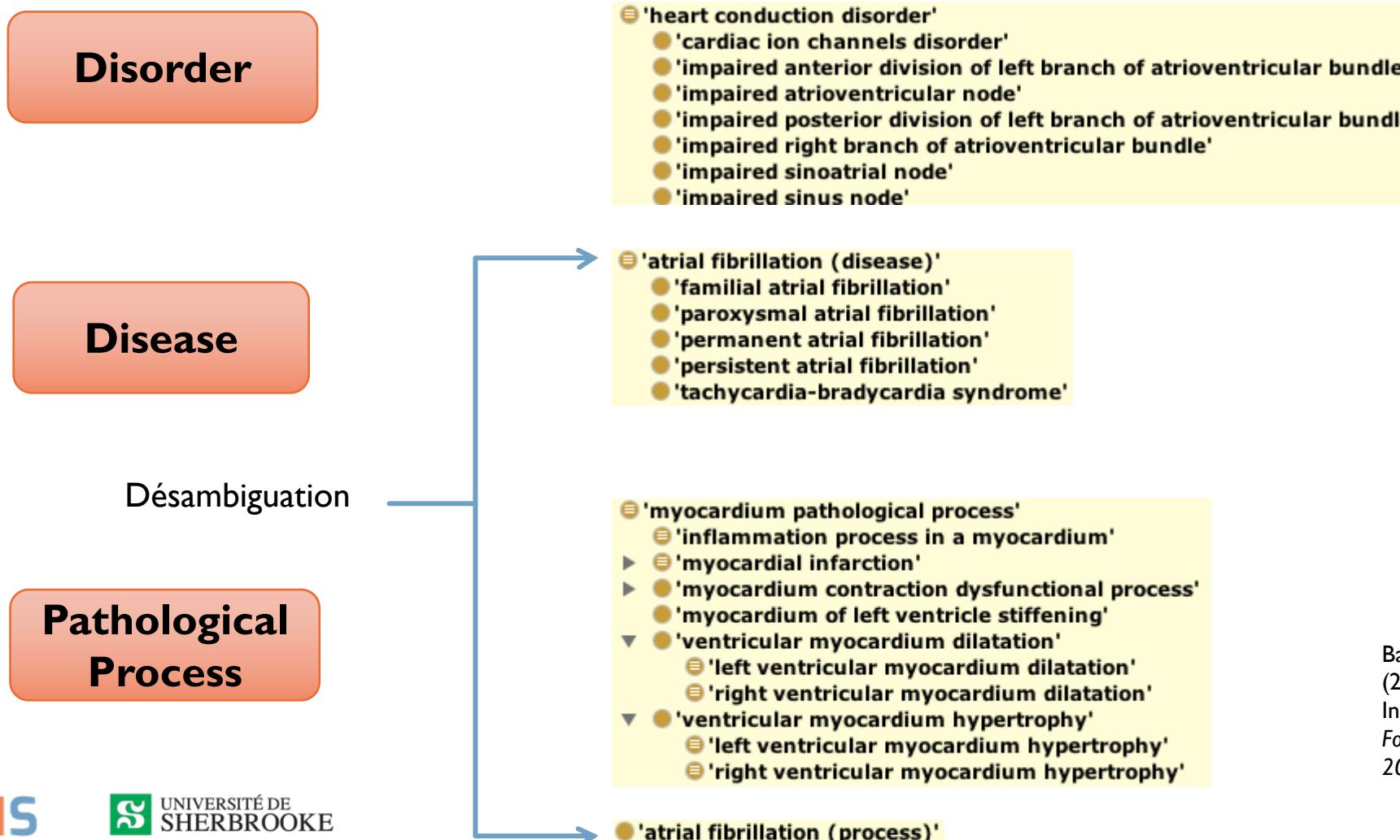


OGMS : Une ontologie générale de la maladie

Scheuermann, R. H., Ceusters, W., & Smith, B. (2009). Toward an ontological treatment of disease and diagnosis. *Summit on translational bioinformatics, 2009*, 116.



Cardiovascular Disease Ontology: Quelques exemples de classes



Barton, A., Rosier, A. Burgun, A & Ethier, J.-F. (2014) The Cardiovascular Disease Ontology. In: *Proceedings of the 8th International Conference on Formal Ontology in Information Systems (FOIS-2014)*, Amsterdam: IOS Press, 409-414.

Les définitions en ontologie

Définitions en langage courant (aristotéliciennes)

Définitions logiques

Annotations: 'cardiomyopathy (1995 definition)'

'CVDO definition'

Disease of the myocardium associated with cardiac dysfunction.
(Report of the 1995 World Health Organization/International Society and Federation of Cardiology Task Force on the Definition and Classification of Cardiomyopathies)



Description: 'cardiomyopathy (1995 definition)'

Equivalent To +

● disease
and ('realized in' some
'disease course'
and ('has occurred part' some 'ventricular myocardium contraction dysfunctional process'))

Classé comme consistant par le raisonneur Pellet

Annotations: 'cor pulmonale'

'CVDO definition' [language: en]

A disease characterized by a dilatation or hypertrophy of the myocardium of right ventricle of the heart as a response to increased resistance or high blood pressure in the lungs, involving a failure of the right side of the heart.

Assertions et inférences

Description: 'cor pulmonale'

Equivalent To +

SubClass Of +

● 'realized in' some
'disease course'
and (('has occurred part' some 'right ventricular myocardium dilatation')
or ('has occurred part' some 'right ventricular myocardium hypertrophy'))
and ('has occurred part' some 'right ventricular myocardium contraction dysfunctional process'))

● disease

→ 'cardiomyopathy (1995 definition)'

→ 'right ventricle disease'

Rend compte de l'évolution des définitions

● 'myocardium disease'

→ ● 'cardiomyopathy (1995 definition)'

→ ● 'cardiomyopathy (2008 definition)'
► ● 'dilated cardiomyopathy'

► ● 'genetic cardiomyopathy'
► ● 'hypertrophic cardiomyopathy'

► ● 'left ventricular noncompaction'
► ● 'restrictive cardiomyopathy'

► ● 'Tako Tsubo cardiomyopathy'

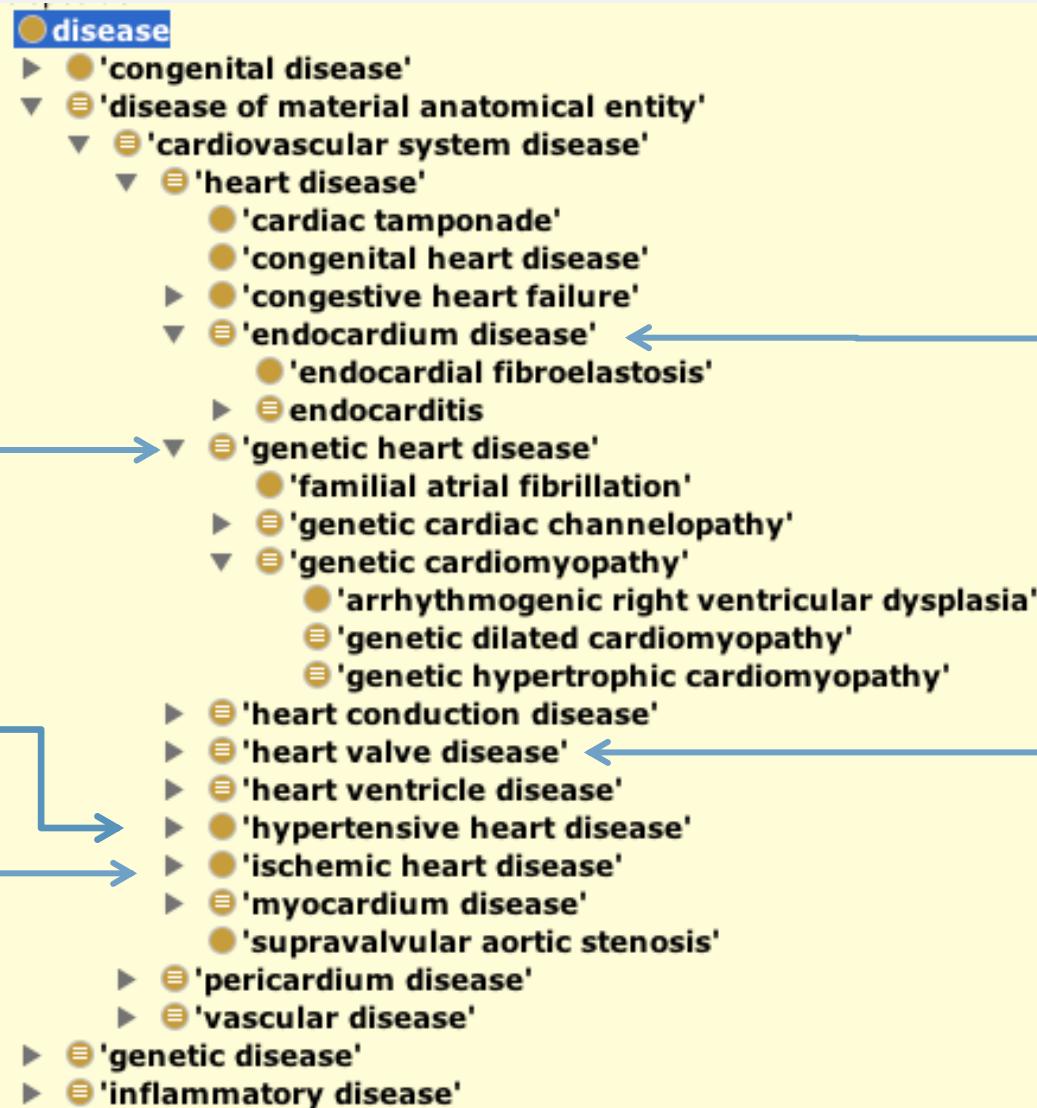
► ● 'cor pulmonale'
► ● 'hypertensive cardiomyopathy'

► ● 'ischemic cardiomyopathy'
► ● 'valvular cardiomyopathy'

Révèlent des défauts logiques dans des définitions courantes

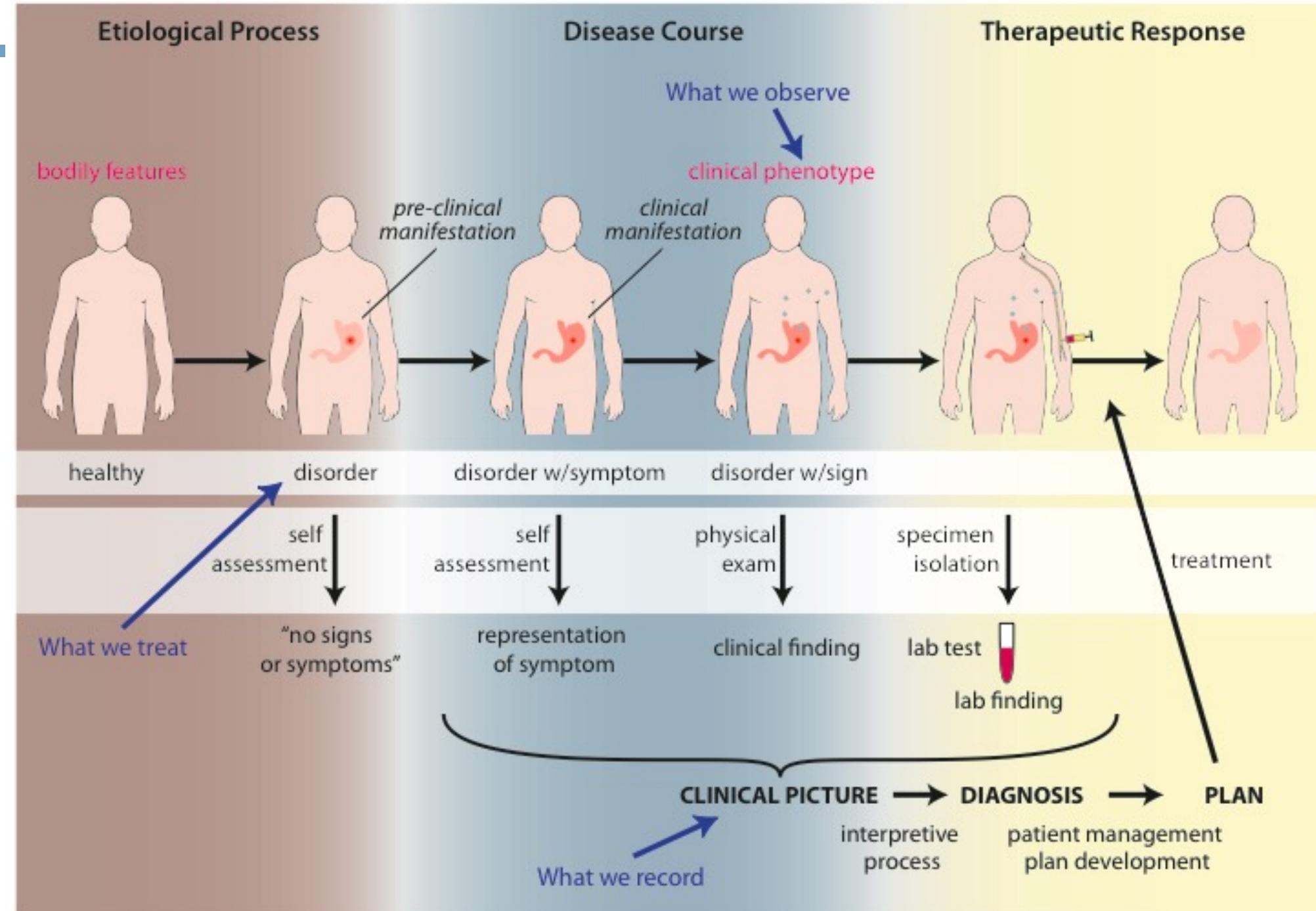
Classification automatique des maladies

par
étiologie

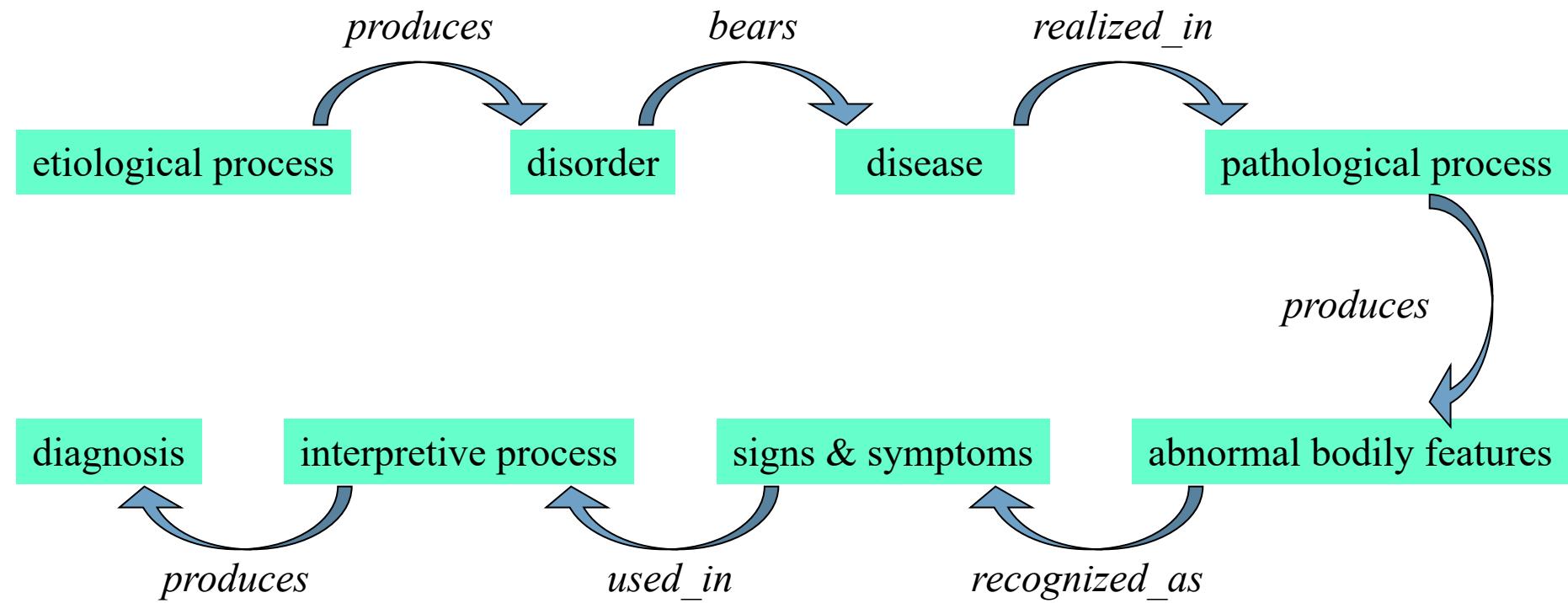


par organe
affecté

Scheuermann, R. H., Ceusters, W., & Smith, B. (2009). Toward an ontological treatment of disease and diagnosis. *Summit on translational bioinformatics, 2009*, 116.



Aperçu de possible relations pertinentes

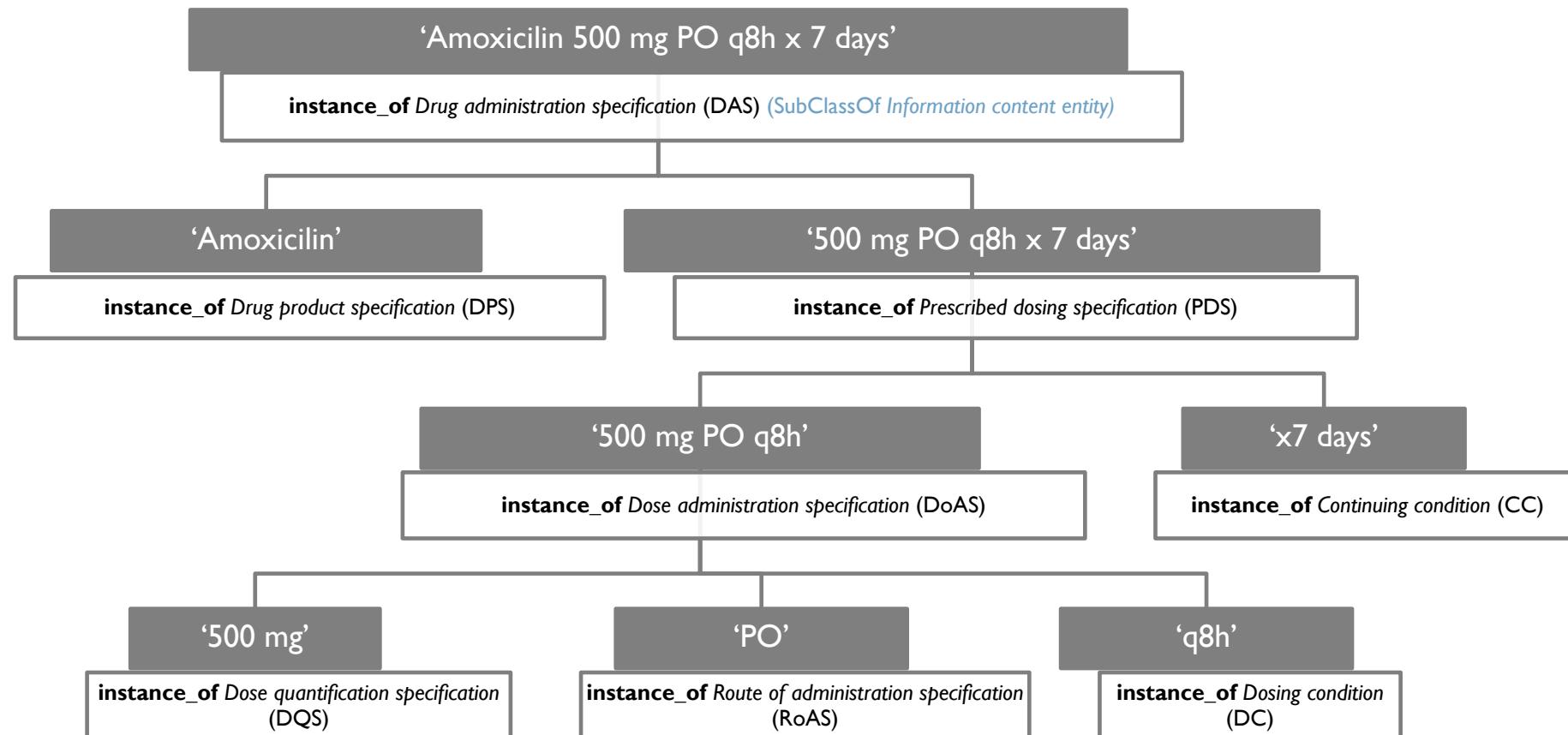


Influenza (infectious disease)

- Etiological process - infection of airway epithelial cells with influenza virus
 - *produces*
- Disorder - viable cells with influenza virus
 - *bears*
- Disposition (disease) - flu
 - *realized_in*
- Pathological process - acute inflammation
 - *produces*
- Abnormal bodily features
 - *recognized_as*
- Symptoms - weakness, dizziness
- Signs - fever
- Symptoms & Signs
 - *used_in*
- Interpretive process
 - *produces*
- Hypothesis - rule out influenza
 - *suggests*
- Laboratory tests
 - *produces*
- Test results - elevated serum antibody titers
 - *used_in*
- Interpretive process
 - *produces*
- Result - diagnosis that patient X has a disorder that bears the disease flu

PDRO: Prescription of Drugs Ontology (extrait)

Ethier, J. F., Goyer, F., Fabry, P., & Barton, A. (2021). The prescription of drug ontology 2.0 (PDRO): more than the sum of its parts. *International Journal of Environmental Research and Public Health*, 18(22), 12025.



Comment représenter formellement une ontologie ?

- Langages logiques :

- logique du premier ordre
- logique des descriptions
- ...

$\forall x(\text{Univ}(x) \rightarrow \text{EducInst}(x)), \forall x(\text{Student}(x) \rightarrow \text{Person}(x)) \dots$

`Univ ⊑ EducInst, Student ⊑ Person, Teacher ⊑ Person`

- Langages informatiques :

- OWL
- CLIF
- ...

`Univ SubClassOf EducInst, Student SubClassOf Person,
Teacher SubClassOf Person`



14:00 – 15:30 :
Ontologie des pizzas

18:00
Souper pizza

Questions ?

adrien.barton@gmail.com

École d'été interdisciplinaire en numérique de la santé (EINS 2024)

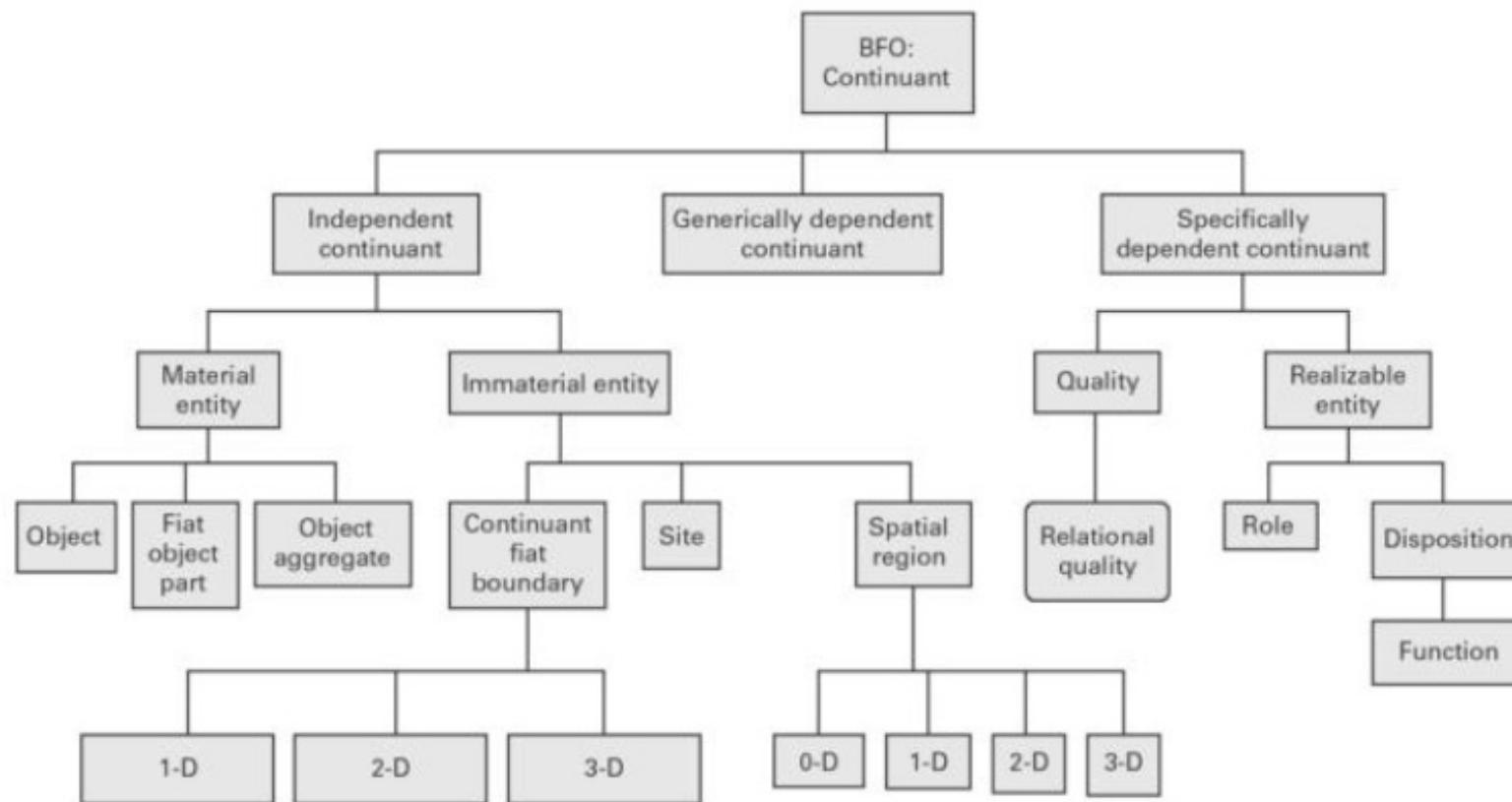
Collaborateurs (ontologies) du GRIIS :

- Anita Burgun
- Jean-François Ethier
- Paul Fabry
- François Goyer
- Olivier Grenier
- Christina Khnaissar
- Luc Lavoie
- Ryeyan Taseen
- ...

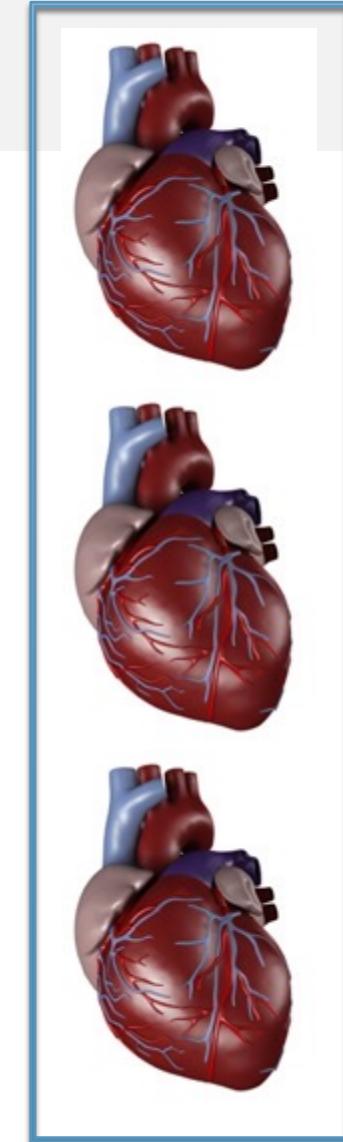
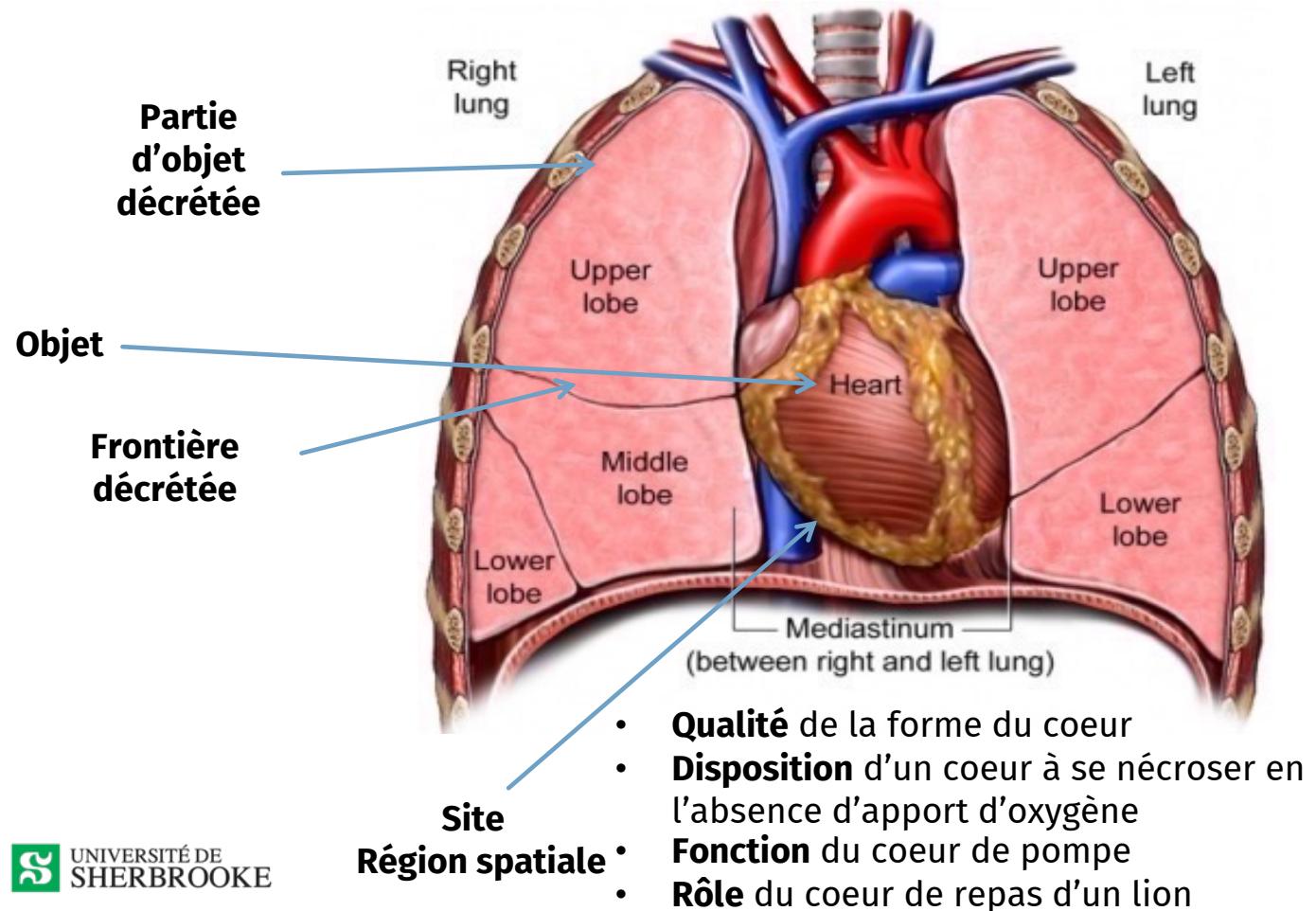
Collaborateurs (ontologies) extérieurs au GRIIS :

- William Duncan (Lawerence Berkeley National Laboratory, USA)
- Ludger Jansen (Institut de Philosophie, Université de Rostock, Allemagne)
- Arnaud Rosier (Implicity, Paris, France)
- Cédric Tarbouriech (IRIT, Toulouse, France)
- Fumiaki Toyoshima (IRIT, Toulouse, France)
- Laure Vieu (IRIT, Toulouse, France)
- ...

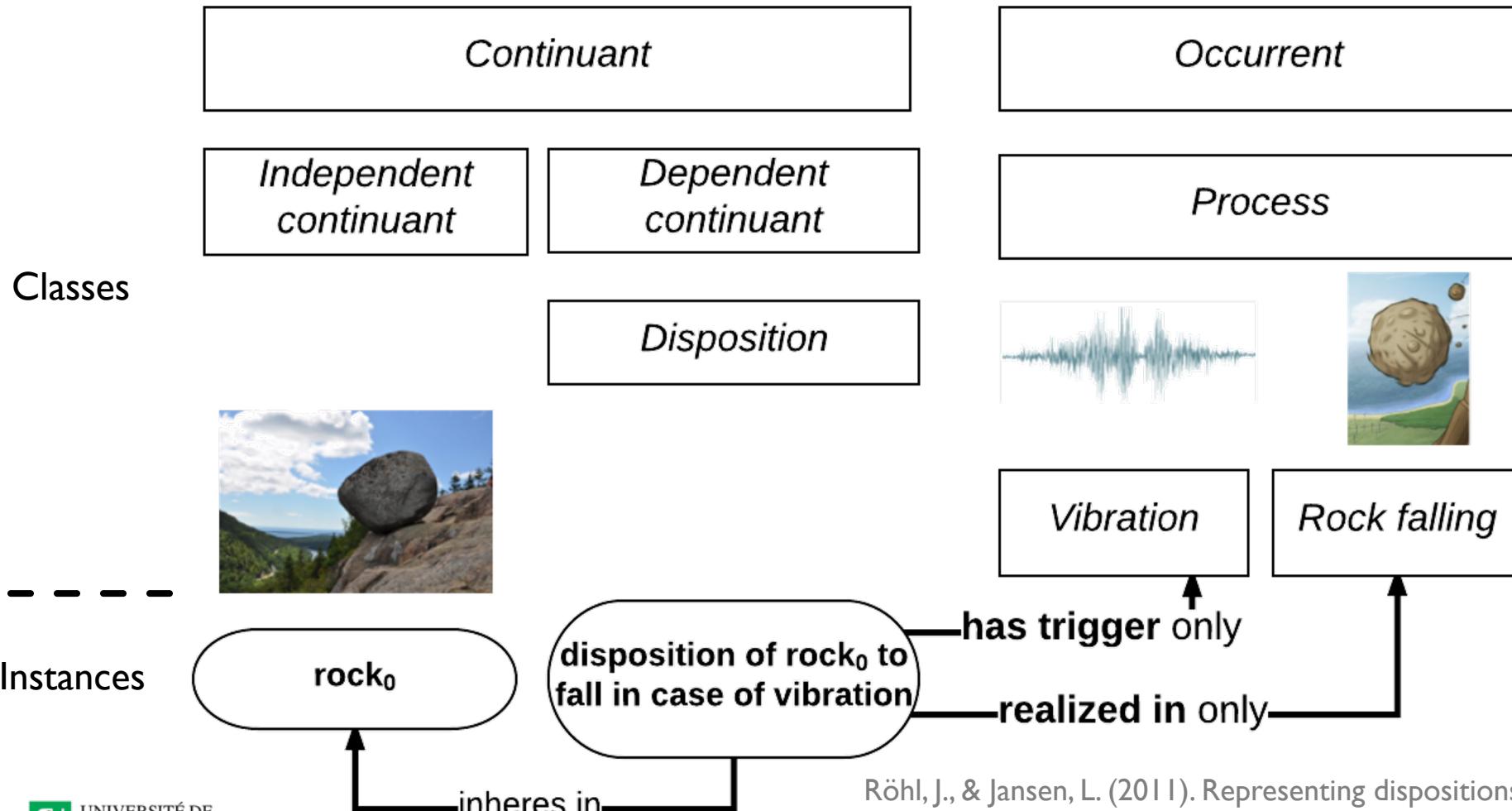
BFO, continuant



Continuants : exemples

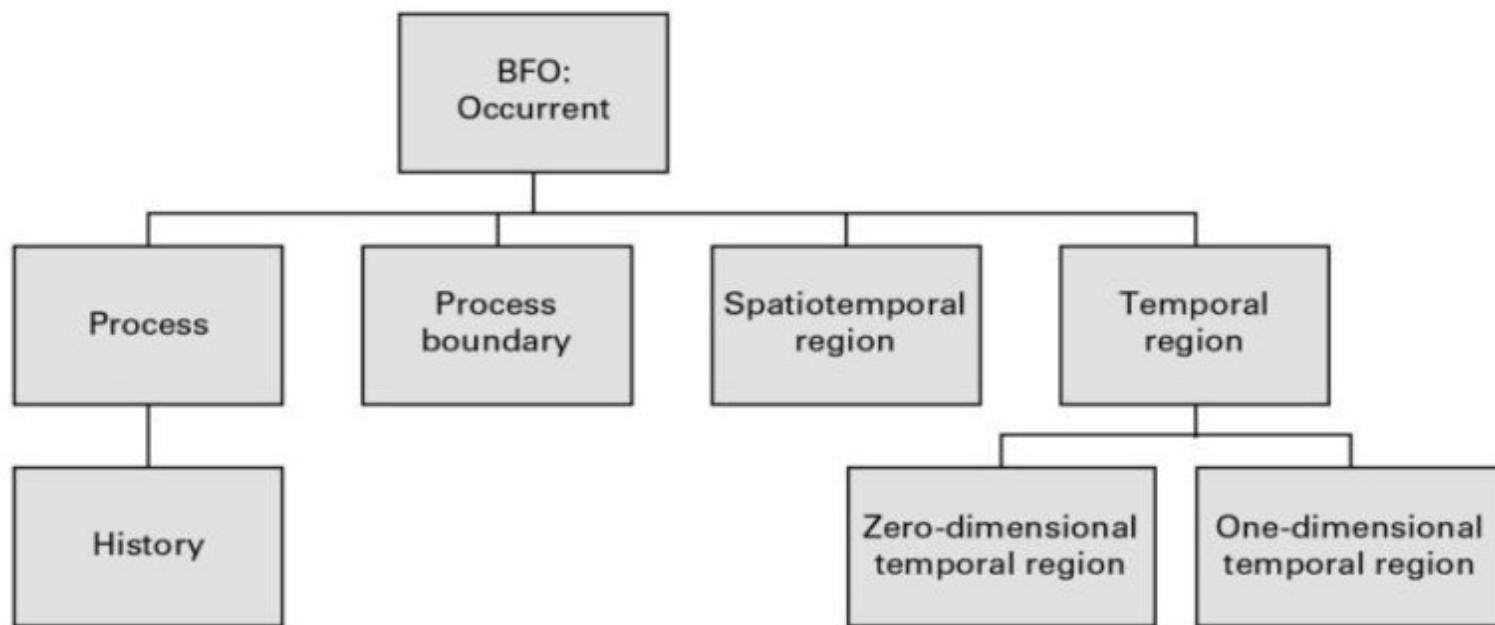


Un type important de propriété : les dispositions

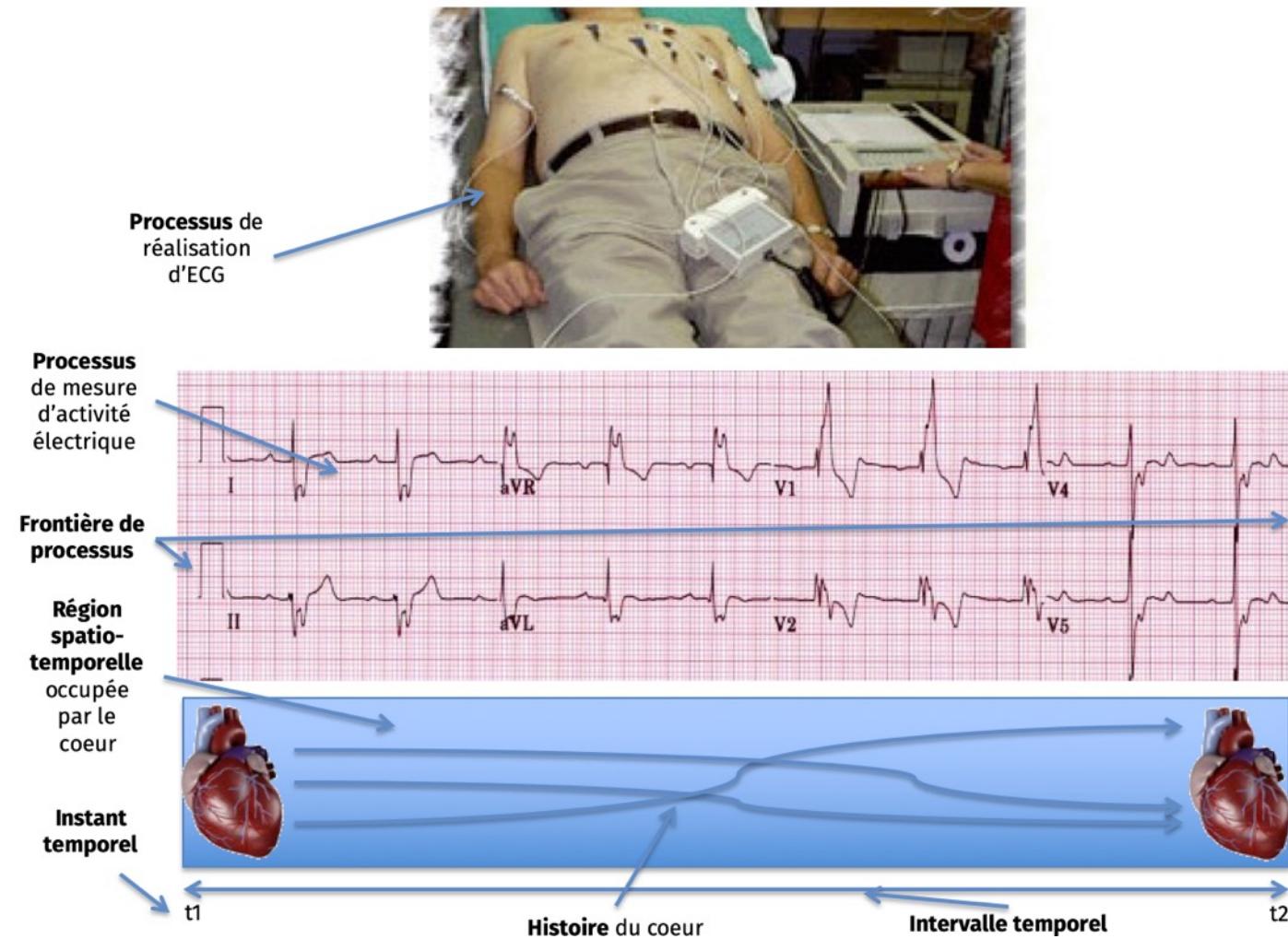


Röhl, J., & Jansen, L. (2011). Representing dispositions. In *Journal of Biomedical Semantics* (Vol. 2, No. 4, pp. 1-13)

BFO, occurrents



Occurrents : exemples



Cirrhosis - environmental exposure

- Etiological process - phenobarbitol-induced hepatic cell death
 - *produces*
- Disorder - necrotic liver
 - *bears*
- Disposition (disease) - cirrhosis
 - *realized_in*
- Pathological process - abnormal tissue repair with cell proliferation and fibrosis that exceed a certain threshold; hypoxia-induced cell death
 - *produces*
- Abnormal bodily features
 - *recognized_as*
- Symptoms - fatigue, anorexia
- Signs - jaundice, splenomegaly
- Symptoms & Signs
 - *used_in*
- Interpretive process
 - *produces*
- Hypothesis - rule out cirrhosis
 - *suggests*
- Laboratory tests
 - *produces*
- Test results - elevated liver enzymes in serum
 - *used_in*
- Interpretive process
 - *produces*
- Result - diagnosis that patient X has a disorder that bears the disease cirrhosis

Huntington's Disease - genetic

- Etiological process - inheritance of >39 CAG repeats in the HTT gene
 - *produces*
- Disorder - chromosome 4 with abnormal mHTT
 - *bears*
- Disposition (disease) - Huntington's disease
 - *realized_in*
- Pathological process - accumulation of mHTT protein fragments, abnormal transcription regulation, neuronal cell death in striatum
 - *produces*
- Abnormal bodily features
 - *recognized_as*
- Symptoms - anxiety, depression
- Signs - difficulties in speaking and swallowing
- Symptoms & Signs
 - *used_in*
- Interpretive process
 - *produces*
- Hypothesis - rule out Huntington's
 - *suggests*
- Laboratory tests
 - *produces*
- Test results - molecular detection of the HTT gene with >39CAG repeats
 - *used_in*
- Interpretive process
 - *produces*
- Result - diagnosis that patient X has a disorder that bears the disease Huntington's disease

Les conditions existentielles d'une maladie

Description: 'hypertrophic cardiomyopathy'

Equivalent To +

SubClass Of +

● 'realized in' **some**
('disease course'
and ('has occurred part' **some** 'left ventricular myocardium hypertrophy')
and ('has occurred part' **some** 'left ventricular myocardium contraction dysfunctional process'))

● **disease**

● 'cardiomyopathy (2008 definition)'
● 'left ventricle disease'

Maladie qui a
besoin d'être
réalisée pour
exister

Description: 'Brugada syndrome'

Equivalent To +

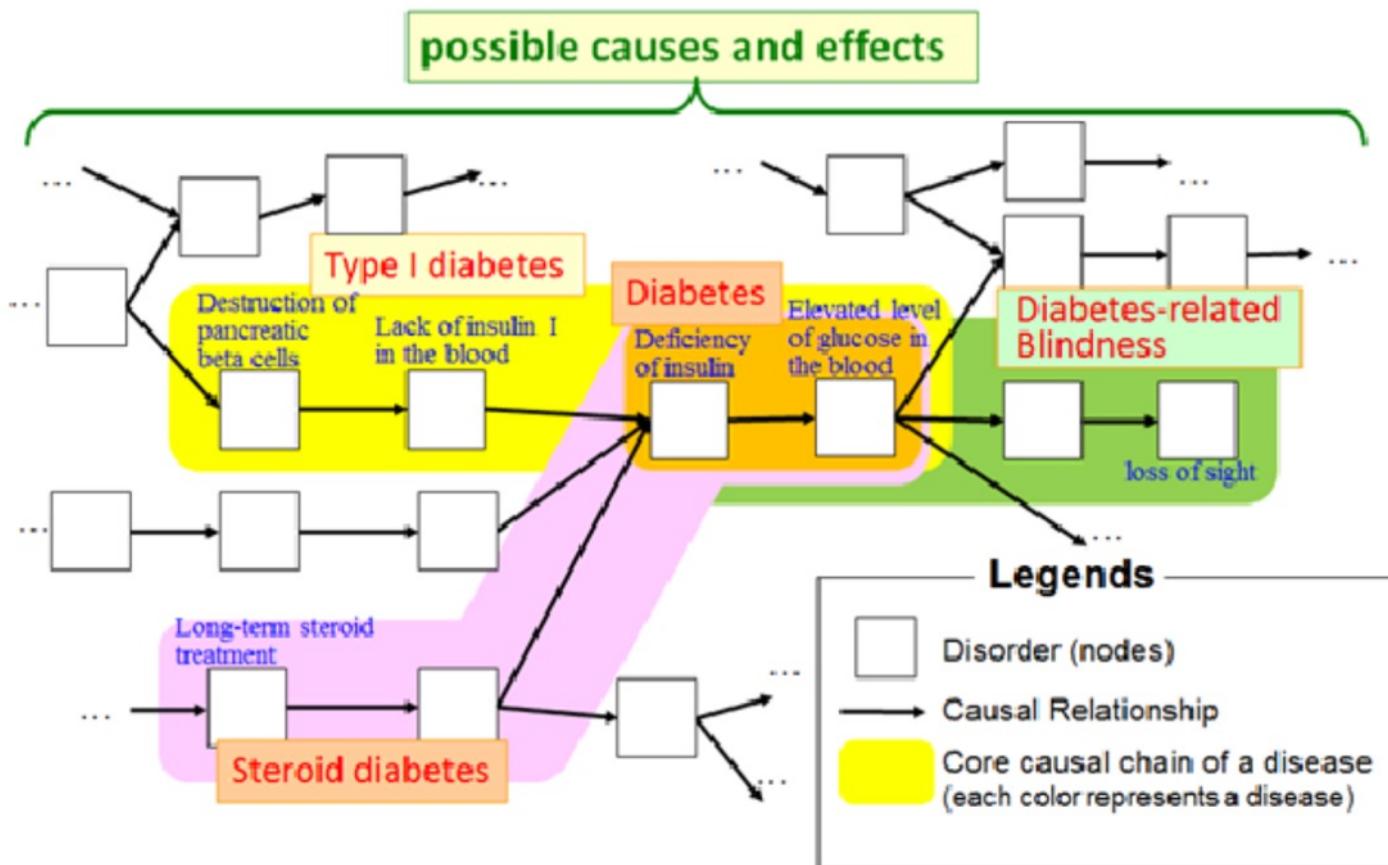
SubClass Of +

● 'cardiac channelopathy'
● 'has material basis at all times' **some** 'genetic disorder'
● 'realized in' **only**
('disease course'
and ('has occurred part' **some** 'ventricular tachyarrhythmia (process)'))
● 'genetic cardiac channelopathy'

Maladie qui n'a
pas besoin d'être
réalisée pour
exister

River-flow model of disease

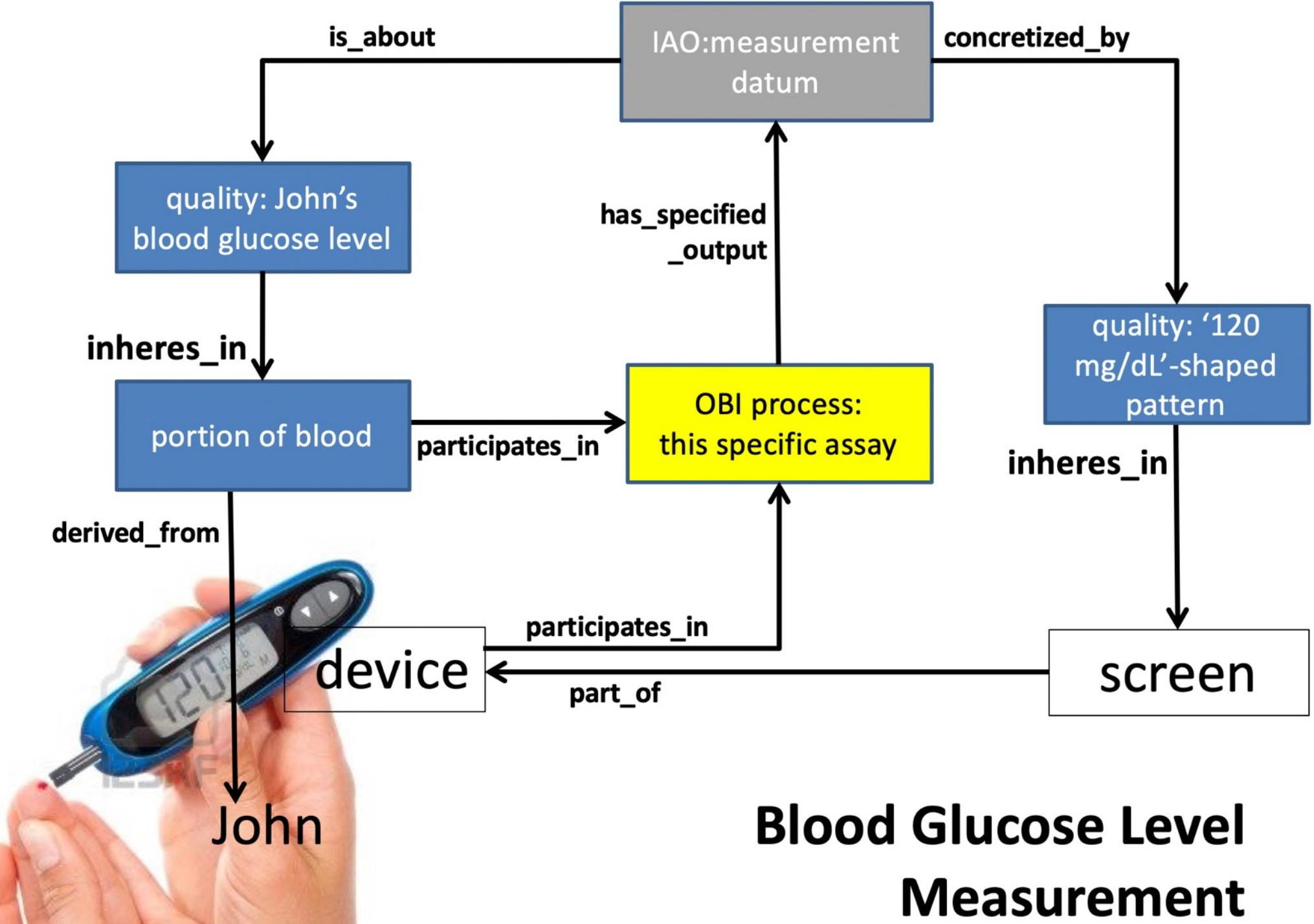
(R. Mizoguchi, K. Kozaki, H. Kou, Y. Yamagata, T. Imai, K. Waki, and K. Ohe. (2011). River Flow Model of Diseases. In Proceedings of the 2nd International Conference on Biomedical Ontology (ICBO2011) (pp. 63-70))



La maladie est un continuant dépendant : elle apparaît, évolue et disparaît.

Compatibilité avec OGMS : la même disposition se retrouve à différentes étapes :

- manifestée par différents processus
- portée par des désordres de nature éventuellement différente.



La variété des documents cliniques

Les ontologies permettent d'analyser et désambiguer les documents cliniques pour permettre un meilleur partage des données :

- **Prescriptions de médicaments**

J.-F. Ethier, A. Barton, R. Taseen (2018). An ontological analysis of drug prescriptions, *Applied Ontology* 13, 273–294.

- **Rapports de distribution de médicaments écrits par les pharmaciens**

A. Barton, P. Fabry, J.-F. Ethier (2020). A classification of instructions in drug prescriptions and pharmacist documents, in: *Proceedings of the 10th International Conference on Biomedical Ontology (ICBO 2019)*, Buffalo, New York, USA, pp. 1–7.

- **Prescriptions de tests de laboratoire et rapports de tests de laboratoire**

A. Barton, P. Fabry, L. Lavoie, J.-F. Ethier (2019) LABO: An ontology for laboratory test prescription and reporting, in: *Proceedings of the Joint Ontology Workshops 2019 (JOWO 2019)*, Graz, Autriche, 2019, pp. 1–9.

- **Questionnaires et sondages**

Fabry, P., Barton, A., Ethier, J.-F. (2021) QUESTO – An Ontology for Questionnaires. In J. Hastings & F. Loebe (Eds.), ICBO|ODLS 2020 International Conference on Biomedical Ontologies 2020, CEUR-WS.org, B.1-12.

- **Documentation de procédures médicales**

Fabry, P., Goyer, F., Barton, A., Ethier, J.-F. (2022) An Ontological Analysis of Health Procedure Information. In *ICBO 2021 - International Conference on Biomedical Ontologies 2021*, CEUR Workshop proceedings, Vol-3073, 36-47.

Mais aussi :

- **Formulaires de consentement**

Lin, Y., Zheng, J., & He, Y. (2016). VICO: Ontology-based representation and integrative analysis of Vaccination Informed Consent forms. *Journal of biomedical semantics*, 7(1), 1-14.