

# Introduction aux ontologies biomédicales

École d'été interdisciplinaire en numérique de la santé (EINS 2025)

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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



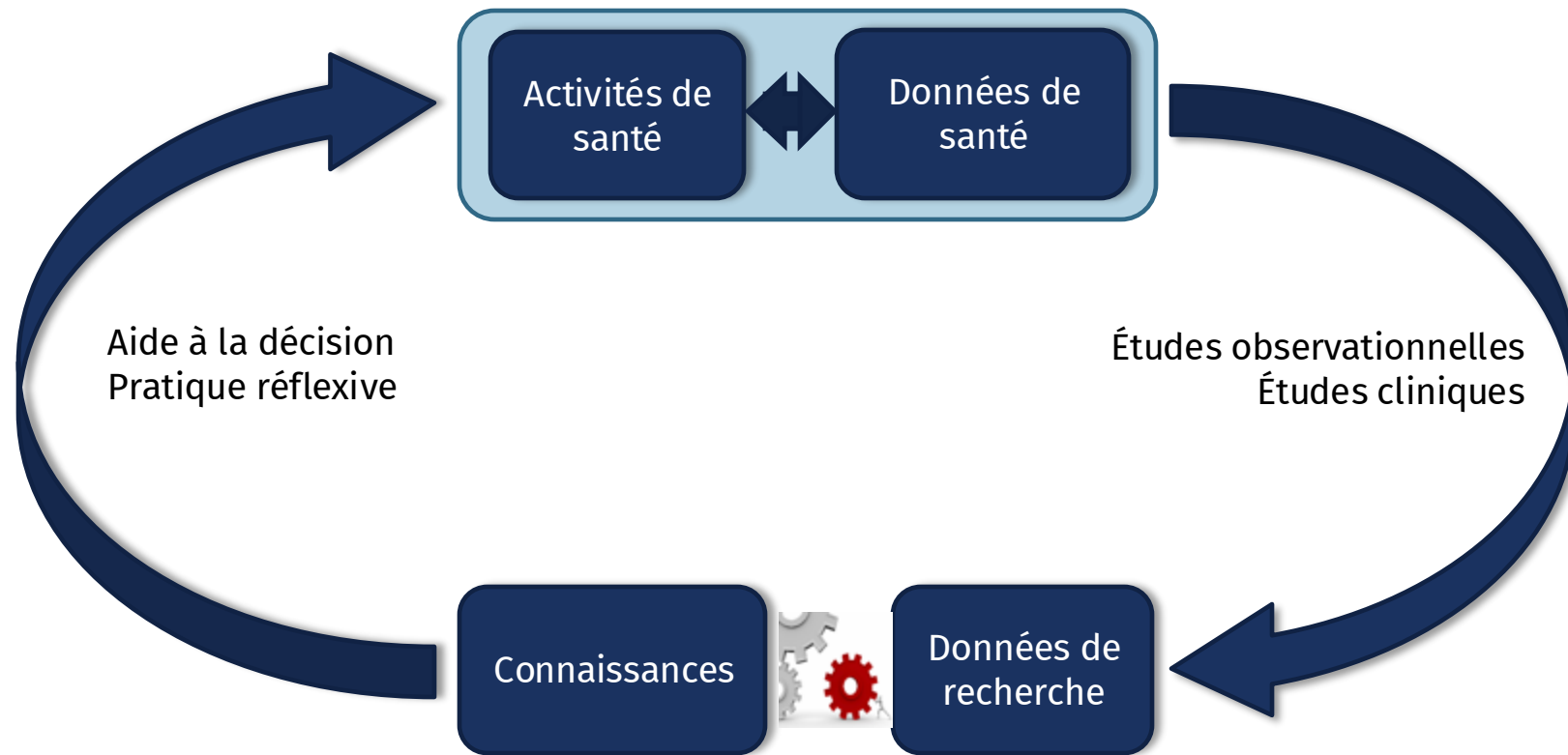
# Les données médicales au Québec aujourd'hui

- Défi : rendre l'information scientifique interoperable, 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



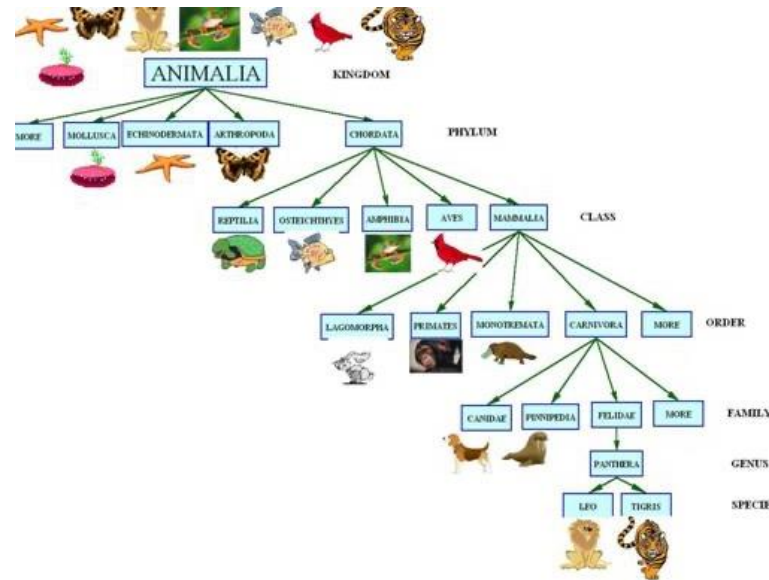
# Approche du GRIIS : Utiliser les ontologies



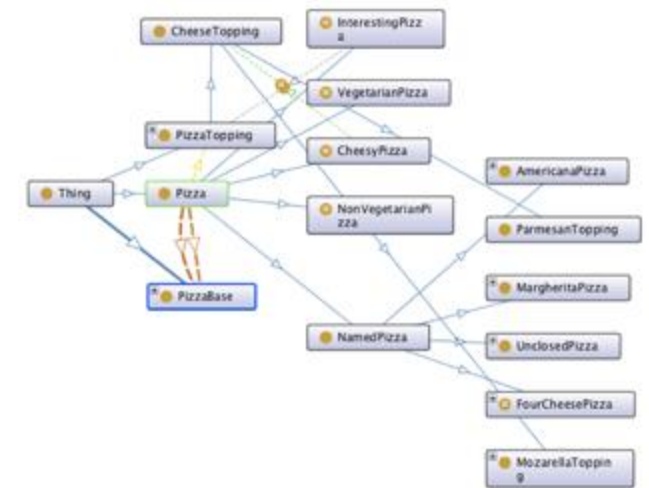
# Terminologies, taxonomies et ontologies

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 down blood clot formation

**Terminologie**



**Taxonomie**



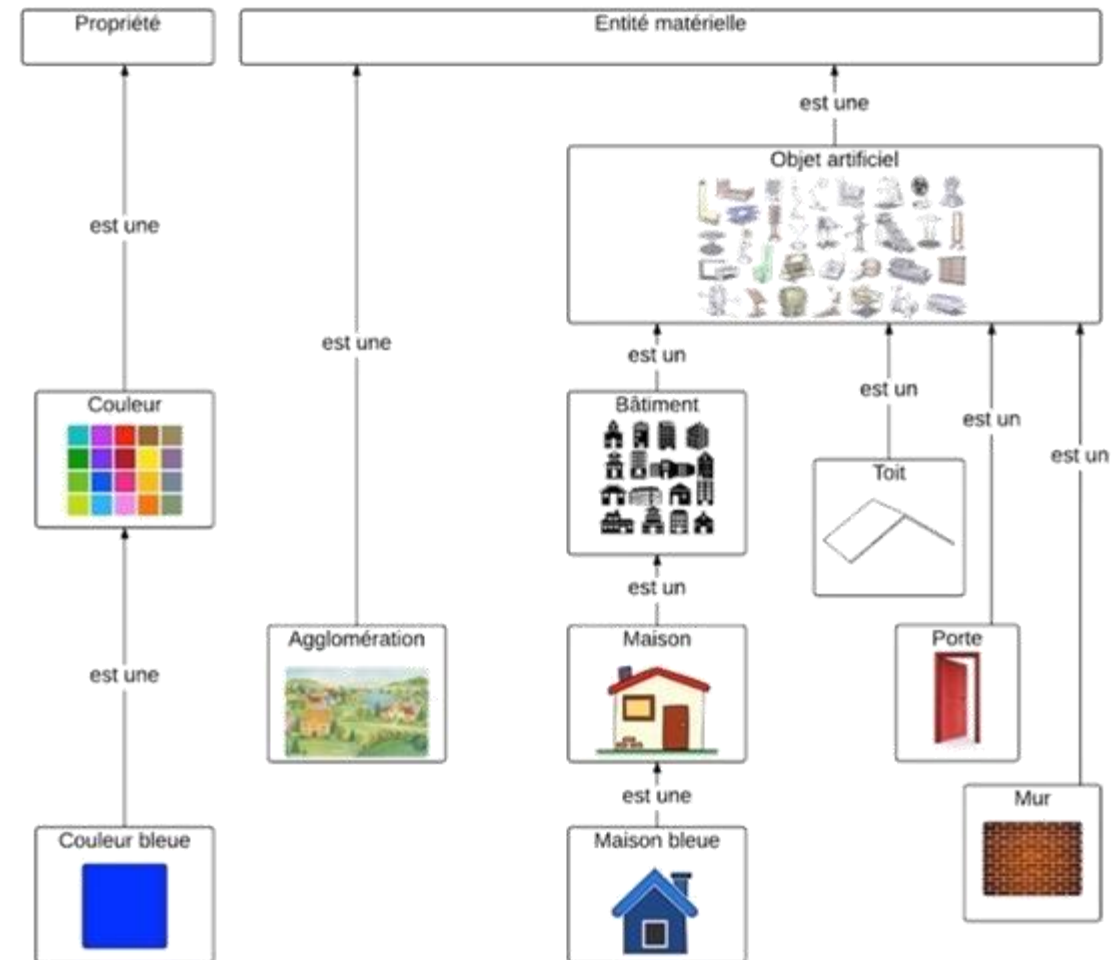
**Ontologie**



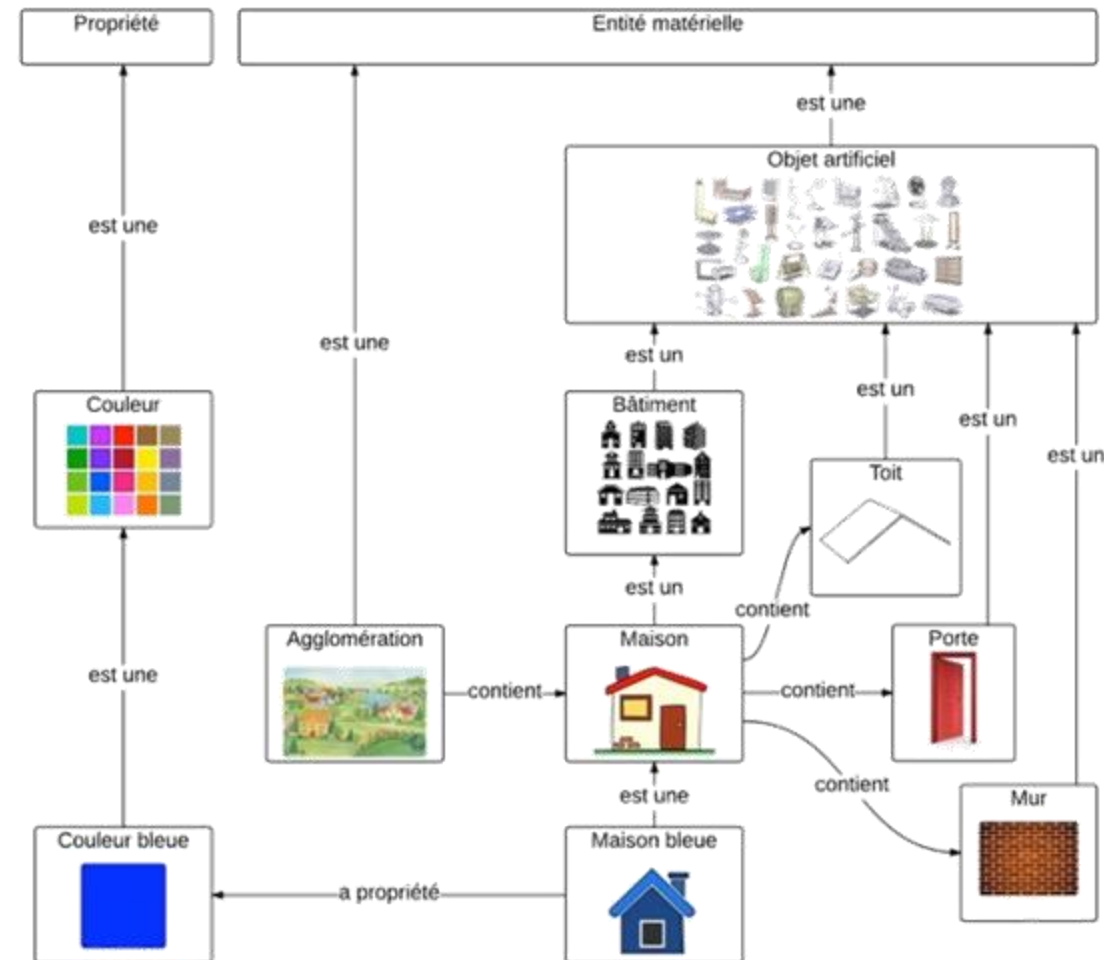
# Terminologie

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



OWL (Web Ontology Language),  
fondé sur la logique  
de description (DL)

Description: House

SubClass Of +

- **Building**
- **has\_part some Door**
- **has\_part some Roof**
- **has\_part some Wall**

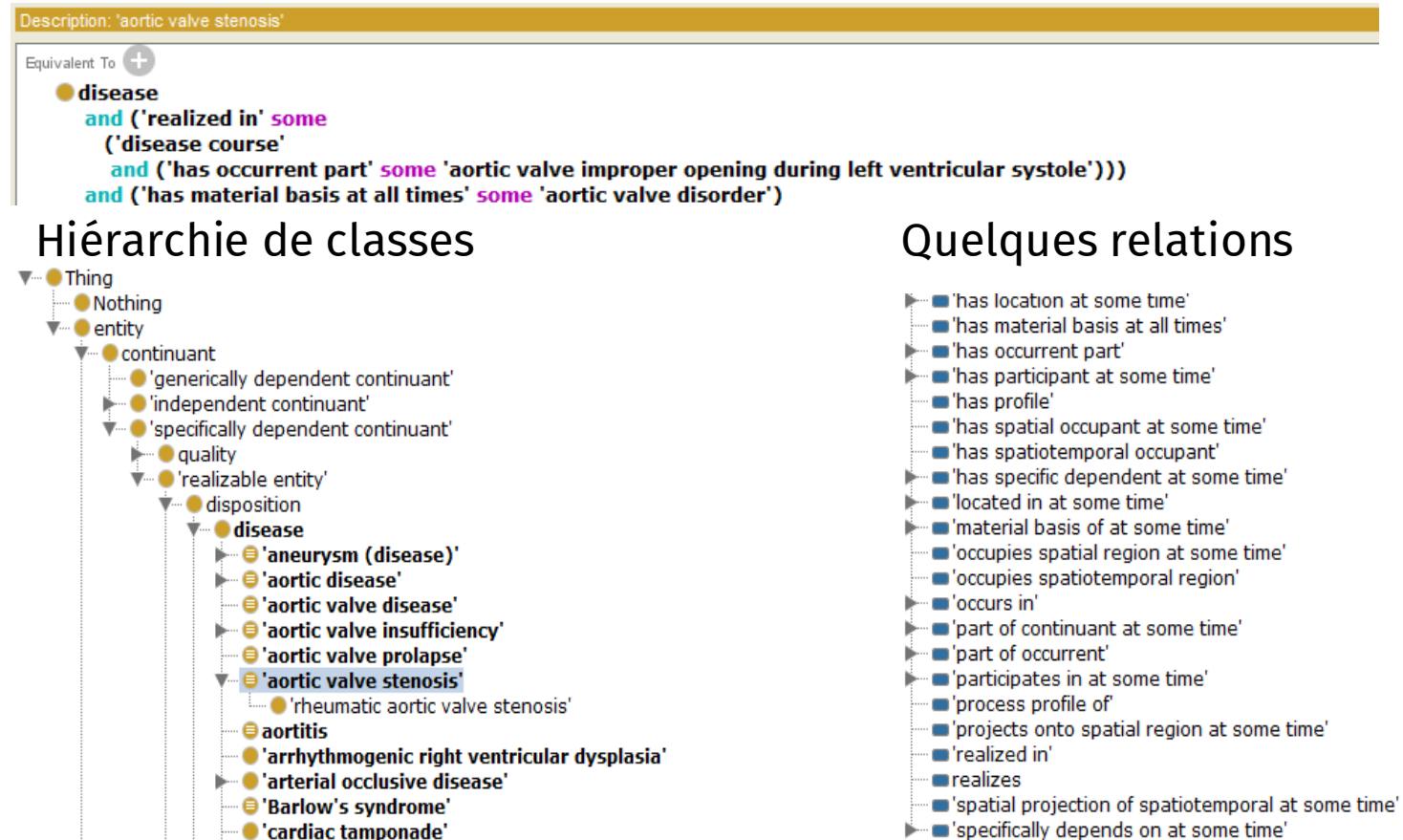
Capture d'écran de Protégé

# Ontologie de domaine

## Définition formelle de la sténose aortique valvulaire (OWL)

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

Exemple : Cardiovascular Disease Ontology (CVDO)



Vues d'écran du logiciel Protégé



# Moteurs de recherche pour ontologies

**OLS ONTOLOGY SEARCH**

Home | **Ontologies** | Help | About | Downloads

Show 10

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 nextwork 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

**Ontobee**

Home | Intro | Statistics | SPARQL | Ontobee | Annotator | Tutorial | FAQs | References | Links | Contact | Acknowledge | News

**Welcome to Ontobee!**

Ontobee: A [linked data](#) server designed for ontologies. Ontobee is aimed to facilitate ontology data sharing, visualization, query, integration, and analysis. Ontobee dynamically [dereferences](#) and presents individual ontology term URIs to (i) [HTML](#) web pages for user-friendly web browsing and navigation, and to (ii) [RDF](#) source code for [Semantic Web](#) applications. Ontobee is the default linked data server for most [OBO Foundry library ontologies](#). Ontobee has also been used for many non-OBO ontologies.

Please select an ontology (optional)

Keywords:  Search terms Batch Search

Jump to <http://purl.obolibrary.org/obo/>  Go

Currently Ontobee has been applied for the following ontologies:

No.	Ontology Prefix	Ontology Full Name	OBO	List of Terms
1	ADO	Alzheimer's Disease Ontology	L	
2	AEO	Anatomical Entity Ontology	L	
3	AFO	Allotrope Foundation Ontology	N	

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

**Search for a class**

Enter a class, e.g. Melanoma

[Advanced Search](#)

**Find an ontology**

Start entering ontology name, e.g. Cancer, then choose from list

[Browse Ontologies](#)

**Ontology Visits (February 2018)**

[More](#)

**BioPortal Statistics**

Ontologies	692
Classes	8,847,370
Resources Indexed	48
Indexed Records	39,537,360
Direct Annotations	95,468,433,792
Direct Plus Expanded Annotations	144,789,582,932

# Les ontologies comme légendes pour les bases de données

MouseEcotope

Tool	Statistical model	Correction for multiple experiments	GO Visualization	Microarrays supported	Time to process 200 genes (s)
Omni-Express	$\chi^2$ , binomial, hypergeometric, Fisher's exact test	Sidak, Holm, Bonferroni, FDR	Flat, Tree	172 commercial arrays (Affymetrix, SuperArray, Agilent, PerkinElmer, Qiagen, Takeda, NDA); can also upload a user-defined list	7, 8, 16, 28
Golden	Fisher's exact test	Relative enrichment	Tree, DAG	Uploads from user	77, 128, 223, 340
DAVID	Fisher's exact test	None	Not available	15, 17, 27, 54	15, 17, 27, 54
EASE online	Fisher's exact test	Bonferroni	Not available	27 arrays (Affymetrix only); can also upload a user-defined list	15, 18, 34, 74
GeneRage	Hypergeometric	Bonferroni	Flat, no hierarchical structure	Uploads from user	6, 6, 6, 8
TransAssociate	Fisher's exact test	None	Not available	Uploads from user	22, 27, 29, 50
GOTM	Hypergeometric	None	Tree	37 arrays (Affymetrix only); uploads from user	39, 60, 137
FastGO	Percentage	Step-down and FDR (Benjamini and Hochberg, 1995), FDR (Benjamini and Yekutieli, 2001)	Flat, Tree	Uploads from user	15, 48, 68, 93
CLEVER	Hypergeometric, $\chi^2$	None	DAG	Uploads from user	NA
GOSort	$\chi^2$ , Fisher's exact test	FDR, Holm, Bonferroni, Holm, Hochberg	Not available	Uploads from user	12, 28, 48, 80
GOSortBox	Hypergeometric, binomial, Fisher's exact test	Benjamini, FDR	Not available	Uploads from user	22, 81, 143
GOshifter	$\chi^2$	None	DAG	90 arrays (Affymetrix only)	5, 7, 7, 7
Ontology Taster	Hypergeometric	FDR	Not available	Uploads from user	NA
eGOn	Binomial	None	Tree	Uploads from user	28, 43, 80, 95

sphingolipid transporter activity

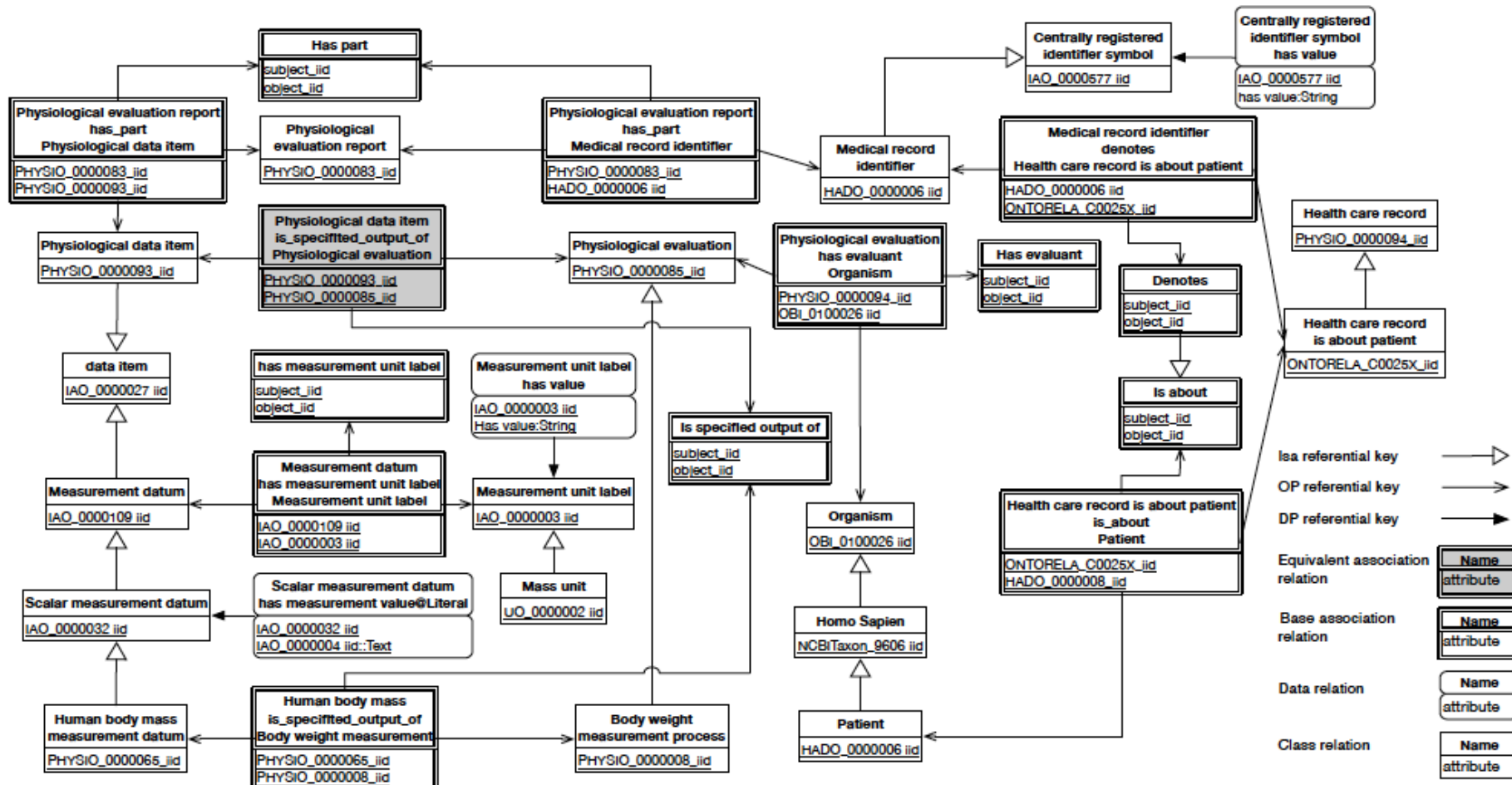
DiabetInGene

GluChem

GlyProt

GALT Glutamine-oxaloacetate transaminase  
 GALT2 Glutamine-oxaloacetate transaminase 2  
 GALT3 Glutamine-oxaloacetate transaminase 3  
 GALT4 Glutamine-oxaloacetate transaminase 4  
 GALT5 Glutamine-oxaloacetate transaminase 5  
 GALT6 Glutamine-oxaloacetate transaminase 6  
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 GALT99 Glutamine-oxaloacetate transaminase 99  
 GALT100 Glutamine-oxaloacetate transaminase 100

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



Khnaisser, Christina, et al. Using an ontology to derive a sharable and interoperable relational data model for heterogeneous healthcare data and various applications. *Methods of Information in Medicine* 61.S 02 (2022): e73-e88.

## Partie 2 : Les différents niveaux d'ontologie





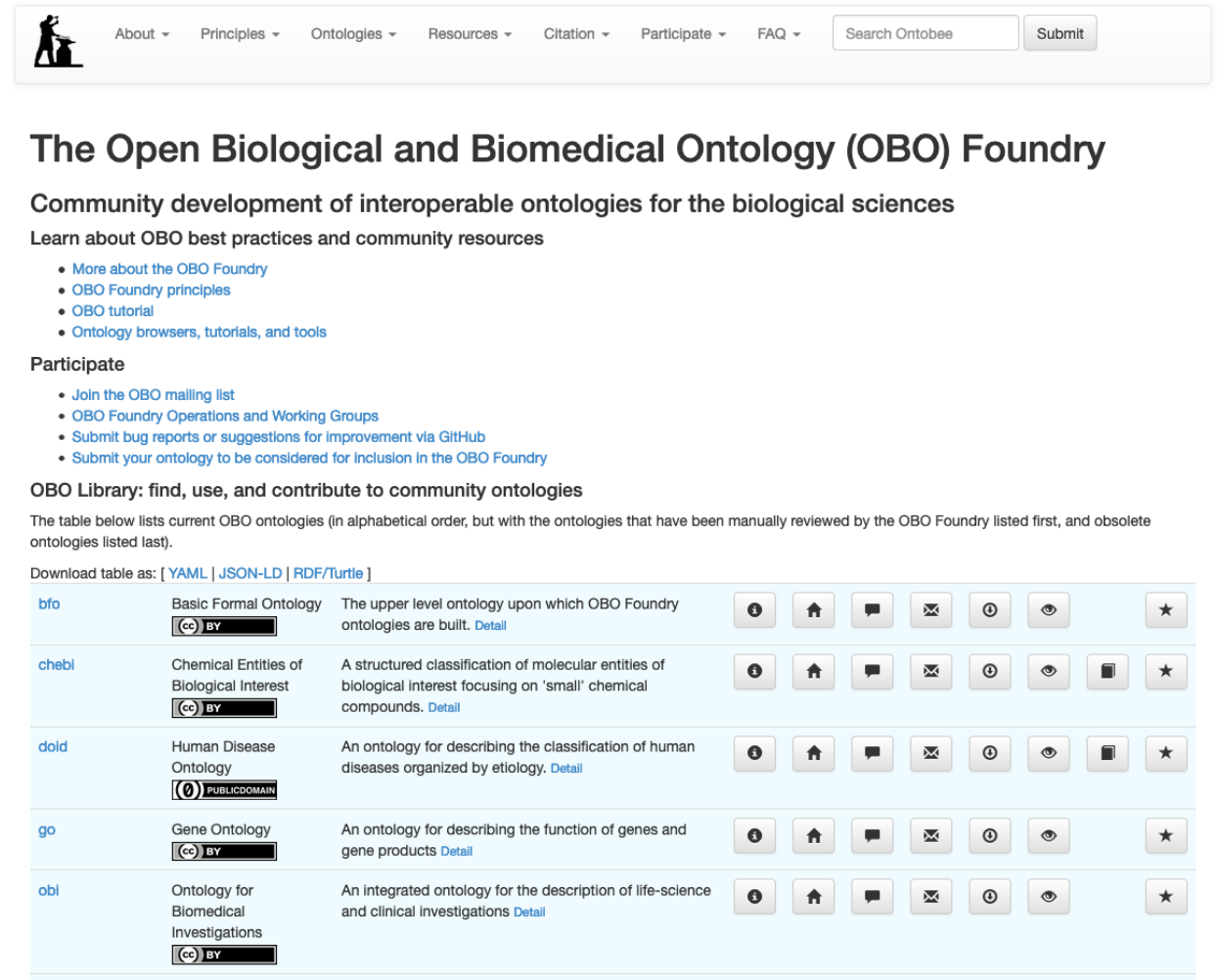
# Difficultés

- 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 screenshot shows the OBO Foundry website. At the top is a navigation bar with links: About, Principles, Ontologies, Resources, Citation, Participate, and FAQ. There is also a search bar labeled 'Search Ontobee' and a 'Submit' button. Below the navigation bar is the main heading 'The Open Biological and Biomedical Ontology (OBO) Foundry' followed by the subtitle 'Community development of interoperable ontologies for the biological sciences'. A section titled 'Learn about OBO best practices and community resources' lists links for 'More about the OBO Foundry', 'OBO Foundry principles', 'OBO tutorial', and 'Ontology browsers, tutorials, and tools'. A 'Participate' section lists links for 'Join the OBO mailing list', 'OBO Foundry Operations and Working Groups', 'Submit bug reports or suggestions for improvement via GitHub', and 'Submit your ontology to be considered for inclusion in the OBO Foundry'. Below this is the 'OBO Library: find, use, and contribute to community ontologies' section, which includes a table of current OBO ontologies. The table lists ontologies like bfo, chebi, doid, go, and obi, each with a brief description and a set of icons for various actions.

**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	License	Description	Icons
<a href="#">bfo</a>	Basic Formal Ontology (cc) BY	The upper level ontology upon which OBO Foundry ontologies are built. <a href="#">Detail</a>	Info, Home, Chat, Email, Print, View, Star
<a href="#">chebi</a>	Chemical Entities of Biological Interest (cc) BY	A structured classification of molecular entities of biological interest focusing on 'small' chemical compounds. <a href="#">Detail</a>	Info, Home, Chat, Email, Print, View, Star
<a href="#">doid</a>	Human Disease Ontology (0) PUBLICDOMAIN	An ontology for describing the classification of human diseases organized by etiology. <a href="#">Detail</a>	Info, Home, Chat, Email, Print, View, Star
<a href="#">go</a>	Gene Ontology (cc) BY	An ontology for describing the function of genes and gene products <a href="#">Detail</a>	Info, Home, Chat, Email, Print, View, Star
<a href="#">obi</a>	Ontology for Biomedical Investigations (cc) BY	An integrated ontology for the description of life-science and clinical investigations <a href="#">Detail</a>	Info, Home, Chat, Email, Print, View, Star

# Quatre types d'ontologies

- **Ontologie de haut-niveau** : indépendante du domaine

*Exemple:* Basic Formal Ontology (BFO): Material entity, Process...

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

*Exemple:* Ontology for General Medical Science (OGMS): Disease, Pathological process...

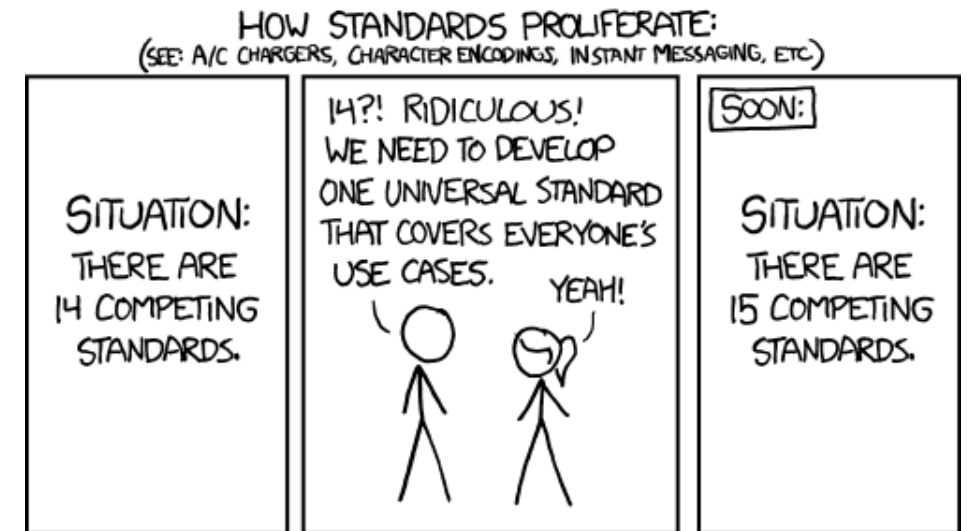
- **Ontologie de domaine** (ou de référence): représentation canonique des entités et relations d'un domaine donné.

*Exemple:* Cardiovascular Disease Ontology (CVDO): Aortic disease, Atrial fibrillation...

- **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.

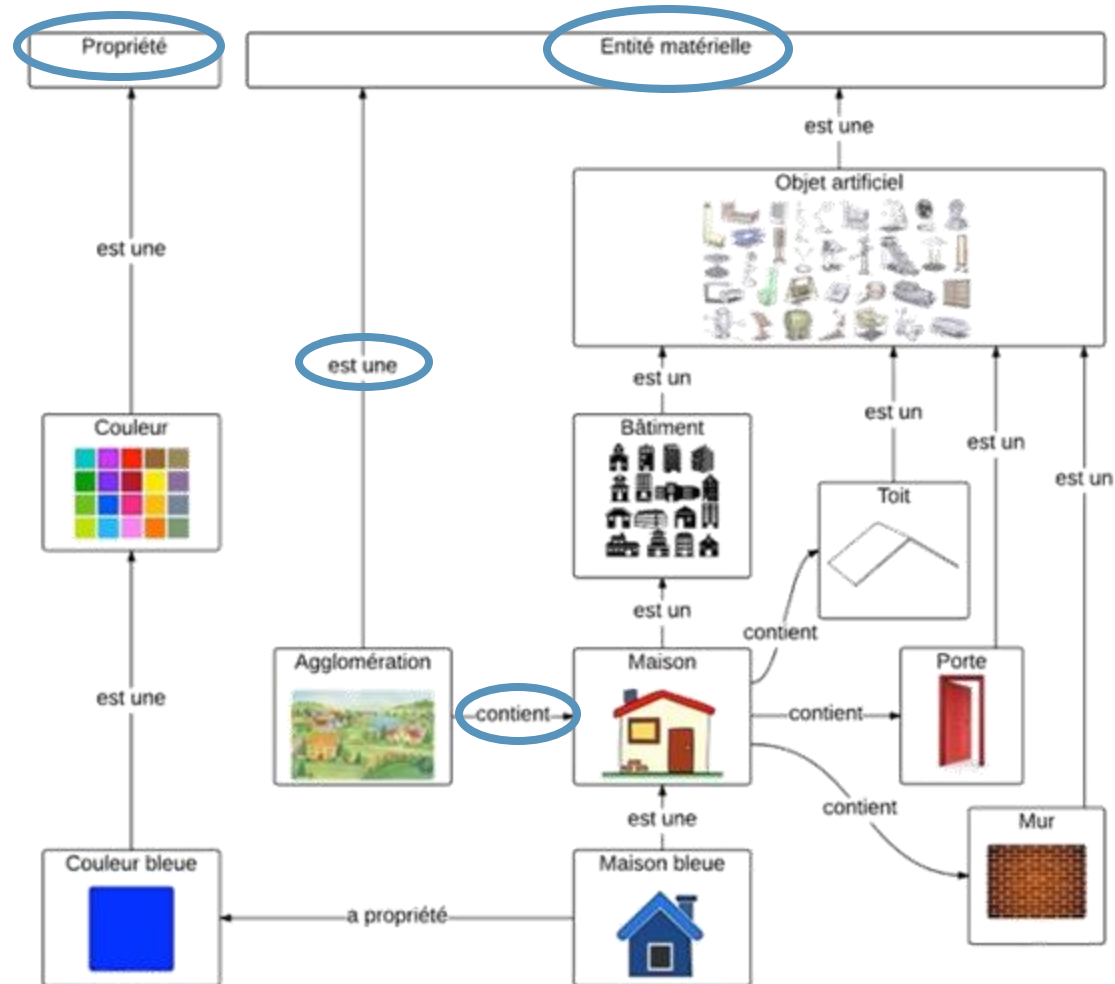


Pourquoi des philosophes ???

# 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 :

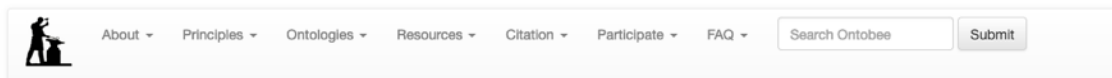
- maladie
- risque
- probabilités
- ...

# 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
- Standard ISO IEC 21838-2:2021

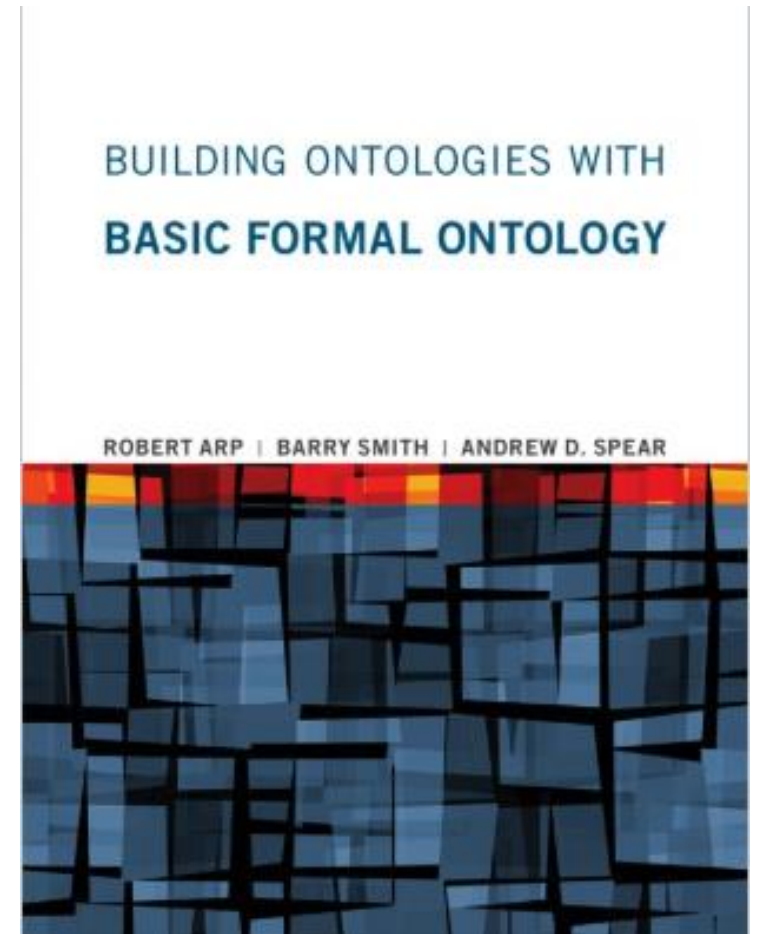
[https://standards.iso.org/ittf/PubliclyAvailableStandards/c074572\\_ISO\\_IEC\\_21838-2\\_2021\(E\).zip](https://standards.iso.org/ittf/PubliclyAvailableStandards/c074572_ISO_IEC_21838-2_2021(E).zip)

- 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



# Continuants et occurrents

## Continuants (perspective 3D)



Mitochondrie

Cellule

Organe

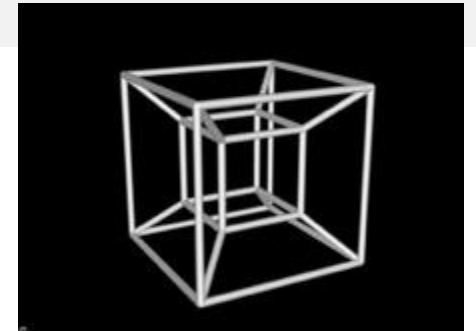
Organisme

Espèce

...

existent  
pleinement à  
tout instant de  
leur existence

## Occurrents (perspective 4D)



Production d'ATP

Division cellulaire

Circulation sanguine

Enfance

Spéciation

...

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
<b>Universaux / Classes</b>	<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>
<b>Particuliers</b>	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

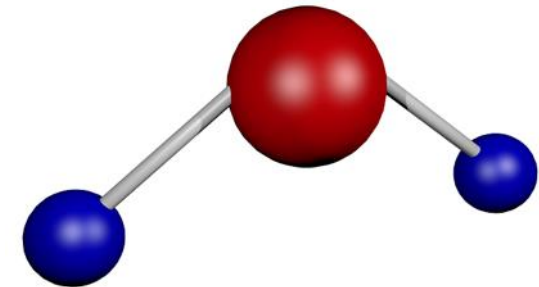


# Le besoin de définitions textuelles : Eviter les imprécisions de pensée



# 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) :

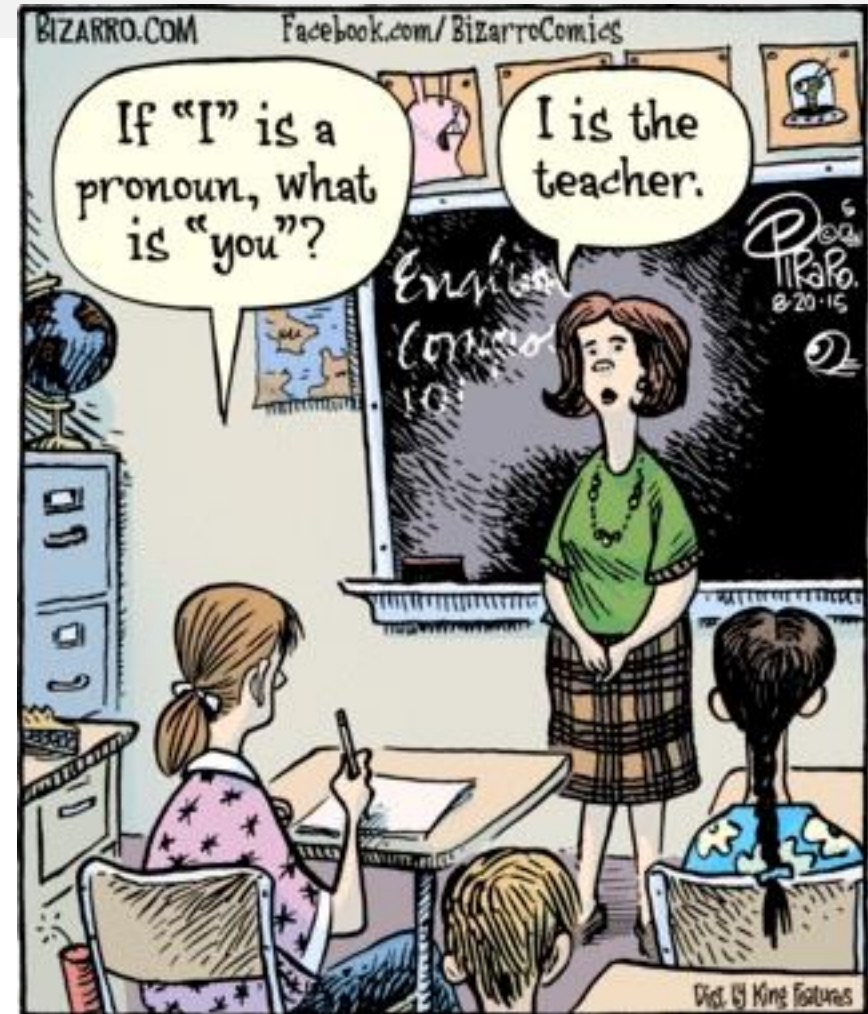
- Nourriture (“food”) défini comme “naturally occurring, processed, or manufactured entities that are primarily used as food for humans and animals.”
- Contenant (“container”) défini comme “a container of other entities”





# 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 =<sub>def</sub> 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 =<sub>def</sub> Un G qui D.

Exemple (FMA) : Heart =<sub>def</sub> An organ with cavitated organ parts, which is continuous with the systemic and pulmonary arterial and venous trees.



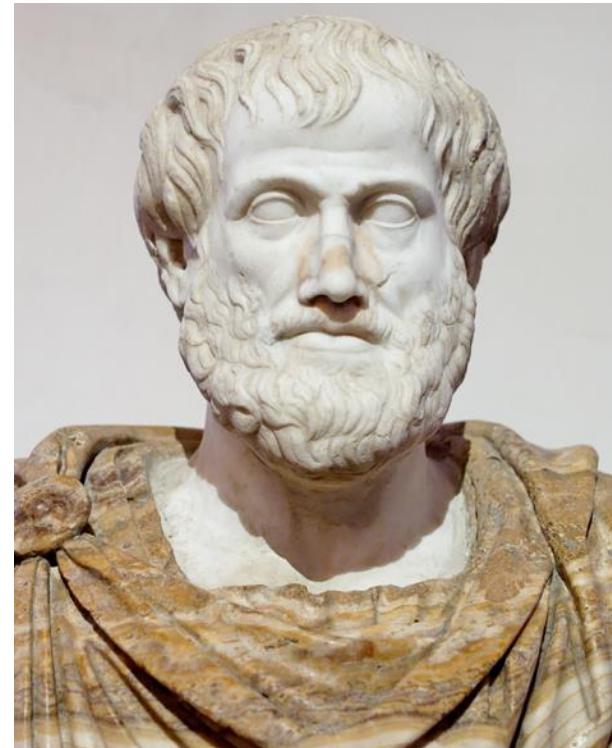
CURRENT ISSUES - PERSPECTIVES AND REVIEWS

## 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



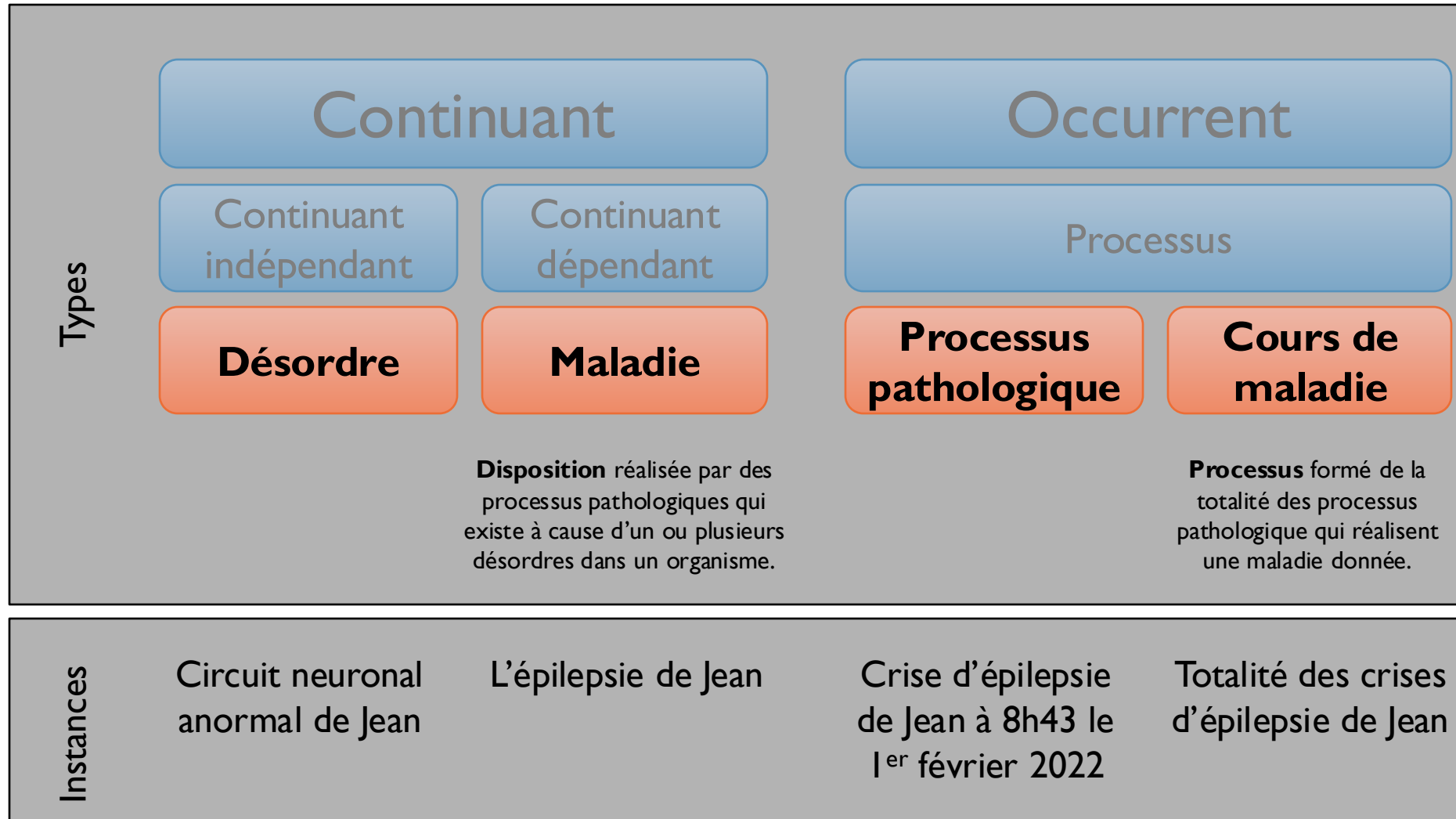
# Application à la classification : l'exemple de la maladie





# 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

**Disorder**

- ⊖ 'heart conduction disorder'
  - 'cardiac ion channels disorder'
  - 'impaired anterior division of left branch of atrioventricular bundle'
  - 'impaired atrioventricular node'
  - 'impaired posterior division of left branch of atrioventricular bundle'
  - 'impaired right branch of atrioventricular bundle'
  - 'impaired sinoatrial node'
  - 'impaired sinus node'

**Disease**

- ⊖ 'atrial fibrillation (disease)'
  - 'familial atrial fibrillation'
  - 'paroxysmal atrial fibrillation'
  - 'permanent atrial fibrillation'
  - 'persistent atrial fibrillation'
  - 'tachycardia-bradycardia syndrome'

Désambiguation

**Pathological  
Process**

- ⊖ 'myocardium pathological process'
  - ⊖ 'inflammation process in a myocardium'
  - ▶ ⊖ 'myocardial infarction'
  - ▶ ● 'myocardium contraction dysfunctional process'
  - 'myocardium of left ventricle stiffening'
  - ▼ ● 'ventricular myocardium dilatation'
    - ⊖ 'left ventricular myocardium dilatation'
    - ⊖ 'right ventricular myocardium dilatation'
  - ▼ ● 'ventricular myocardium hypertrophy'
    - ⊖ 'left ventricular myocardium hypertrophy'
    - ⊖ 'right ventricular myocardium hypertrophy'

- 'atrial fibrillation (process)'

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  
(aristotéliennes)

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 occurrent 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 occurrent part' some 'right ventricular myocardium dilatation')  
or ('has occurrent part' some 'right ventricular myocardium hypertrophy'))  
and ('has occurrent part' some 'right ventricular myocardium contraction dysfunctional process'))

● **disease**

→ 'cardiomyopathy (1995 definition)'

→ 'right ventricle disease'

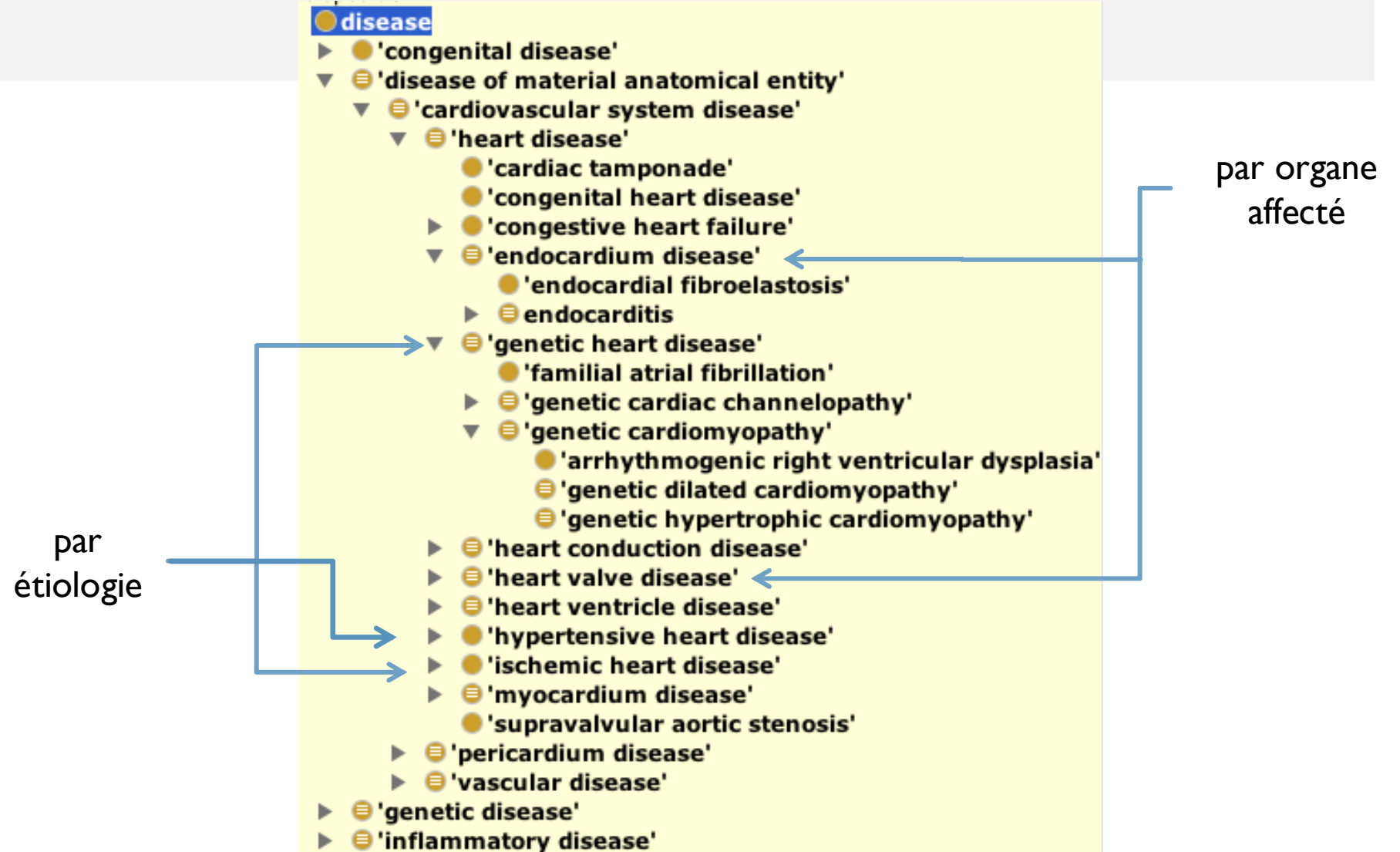
Rend compte de l'évolution des définitions

- **'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'**

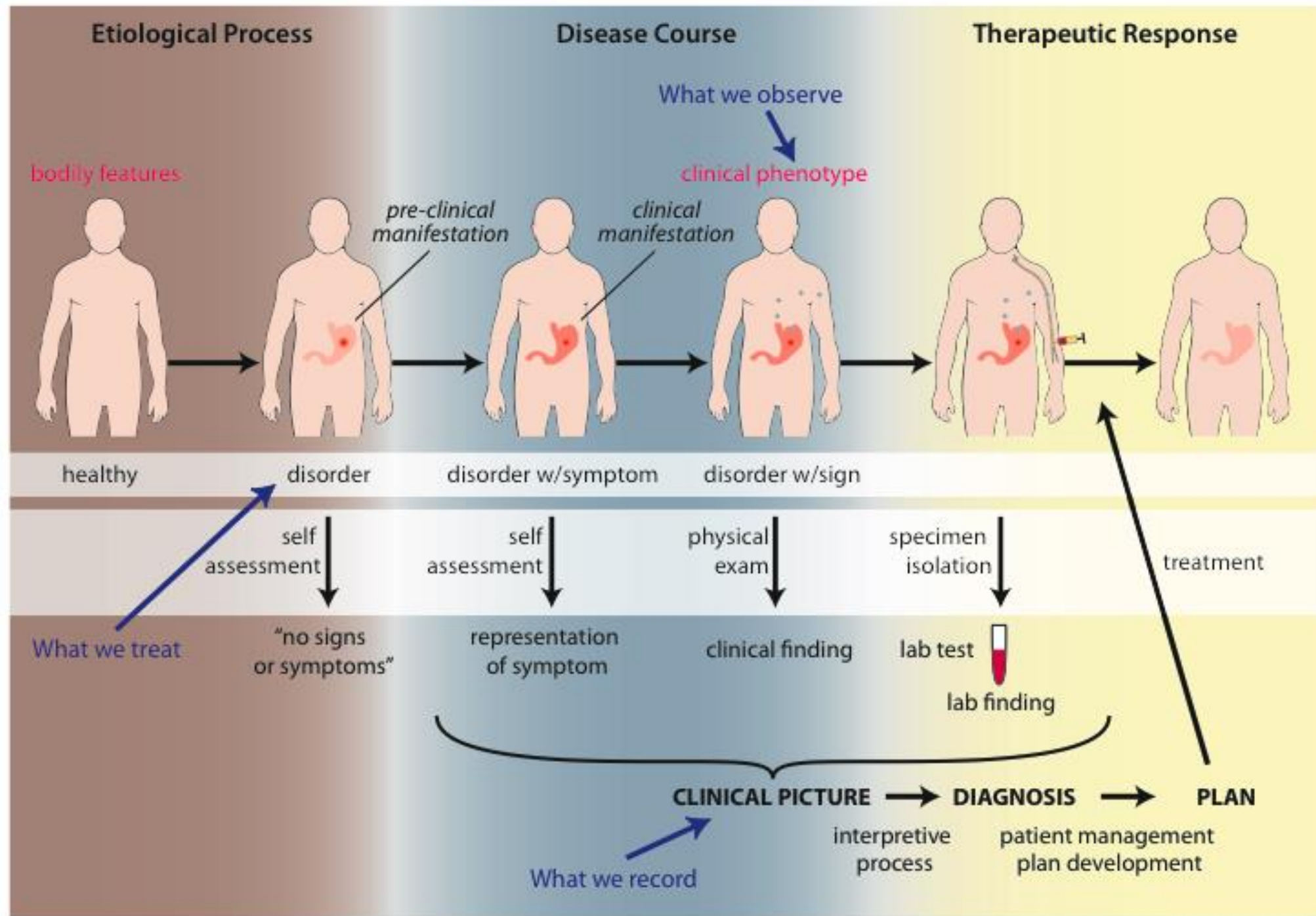
Révèlent des défauts logiques dans des  
définitions courantes



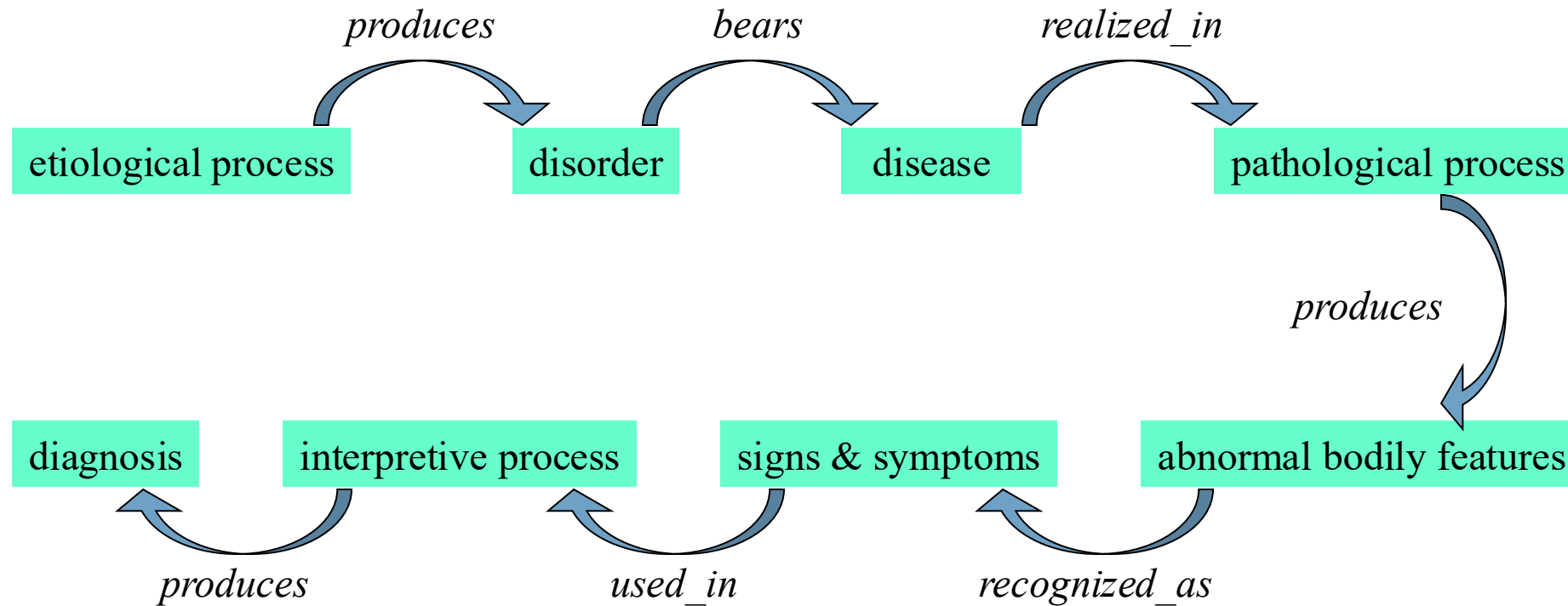
# Classification automatique des maladies



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

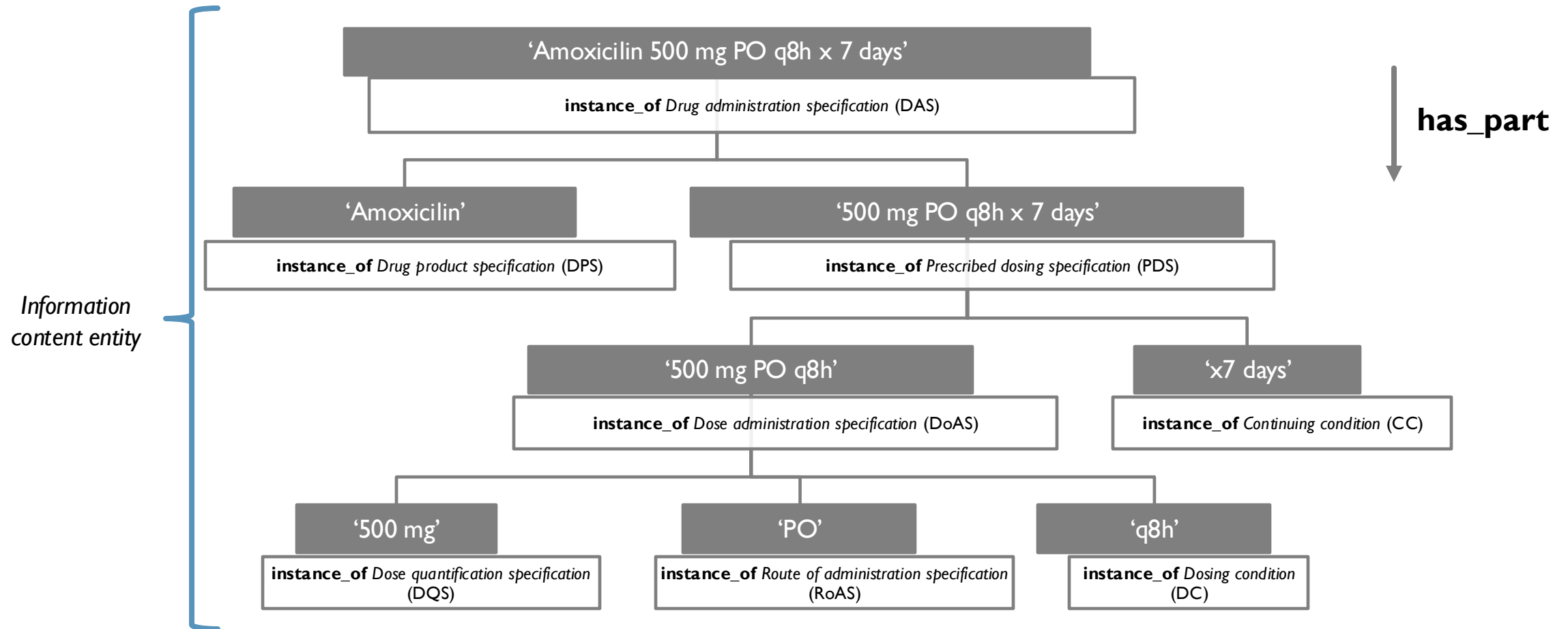


# Influenza (infectious disease)

- 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

# PDR0: Prescription of Drugs Ontology (extrait)

Ethier, J. F., Goyer, F., Fabry, P., & Barton, A. (2021). The prescription of drug ontology 2.0 (PDR0): 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 description
- ...

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

$\text{Univ} \sqsubseteq \text{EducInst}, \text{Student} \sqsubseteq \text{Person}, \text{Teacher} \sqsubseteq \text{Person}$

- Langages informatiques :

- RDF, RDFS
- OWL
- CLIF
- ...

Pizza SubClassOf  
(has\_topping min 1 Topping)



13:30 – 16:30  
Ontologie des pizzas

Snape teaches Harry  
Student SubClassOf Person



10:45 – 12:30  
Introduction RDF, RDFS et SPARQL



## Questions ?

adrien.barton@gmail.com

### École d'été interdisciplinaire en numérique de la santé (EINS 2025)

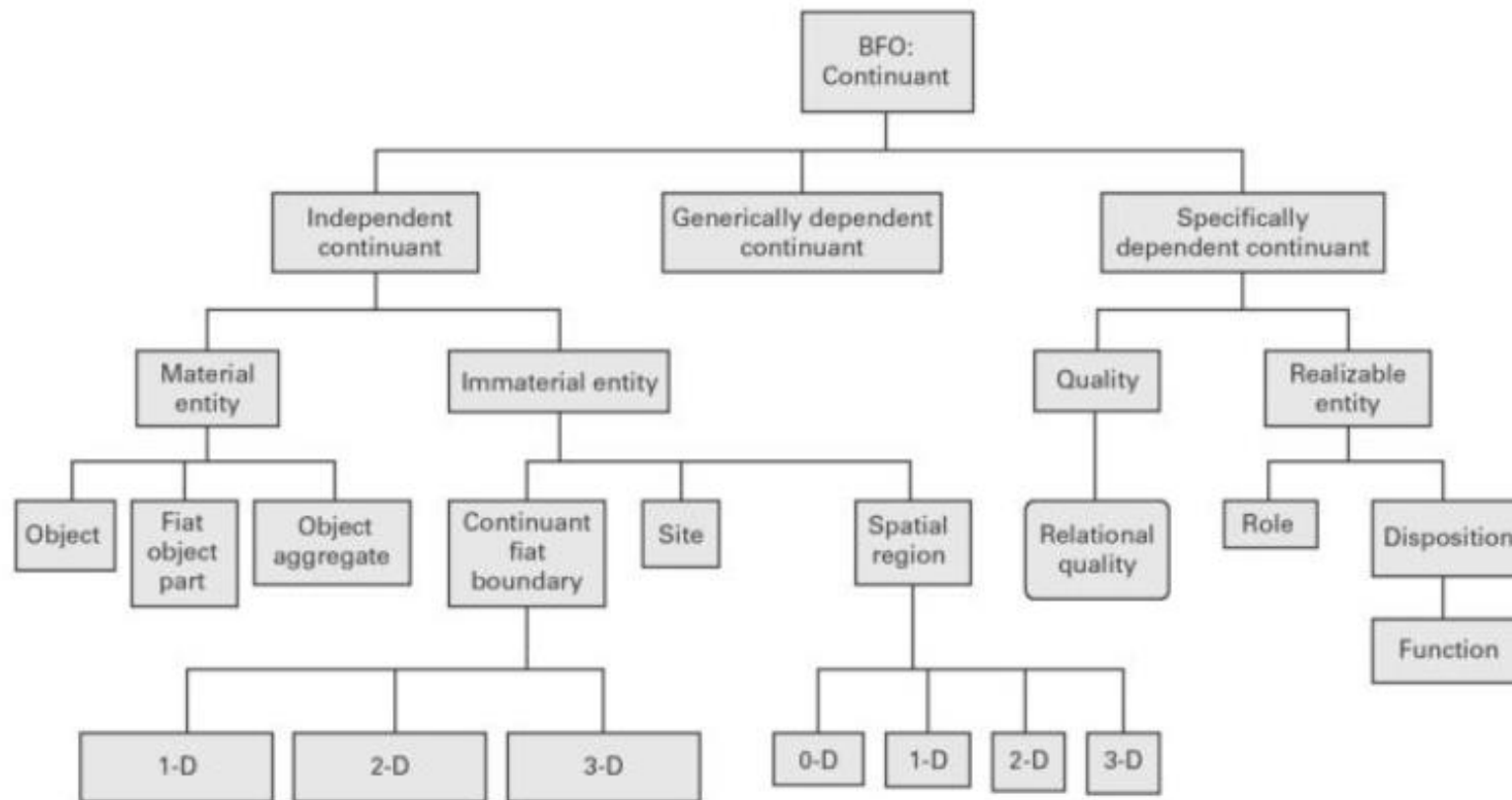
#### Collaborateurs (ontologies) du GRIIS :

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

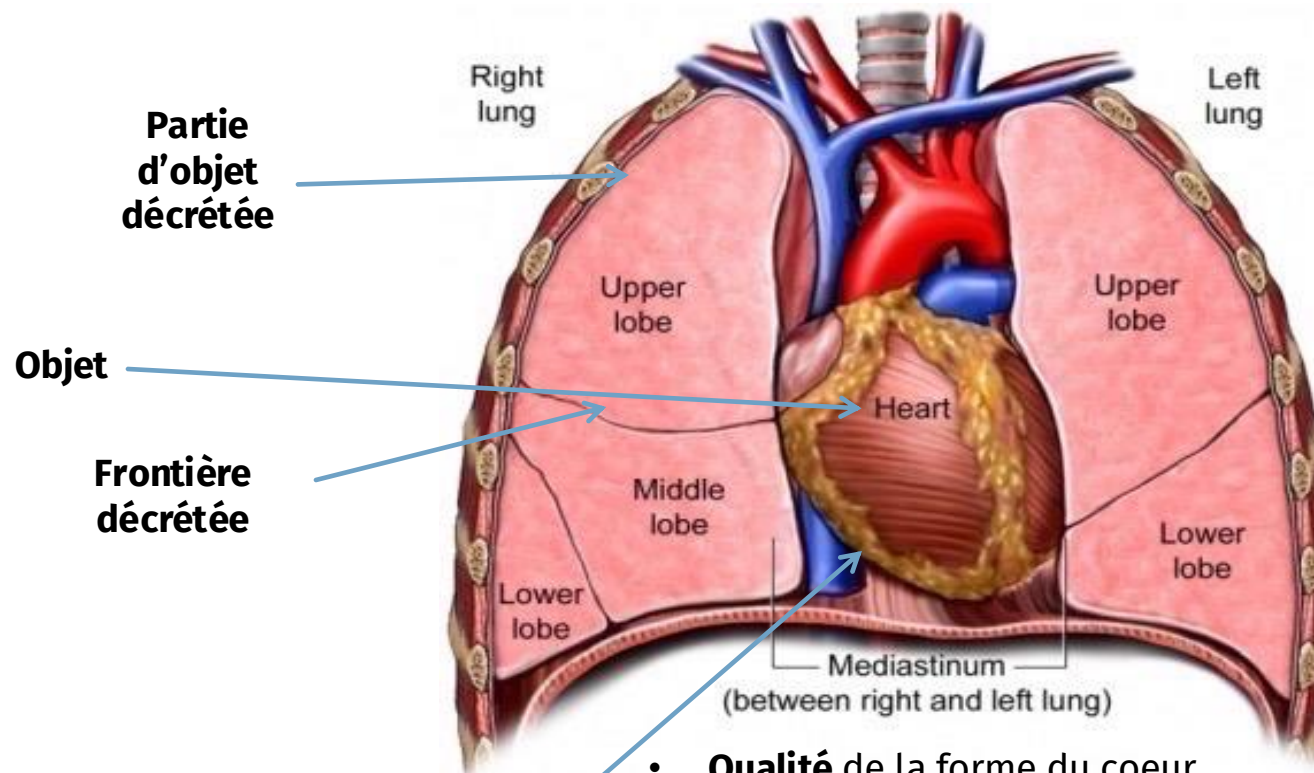
#### Collaborateurs (ontologies) extérieurs au GRIIS :

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

# BFO, continuant



# Continuants : exemples



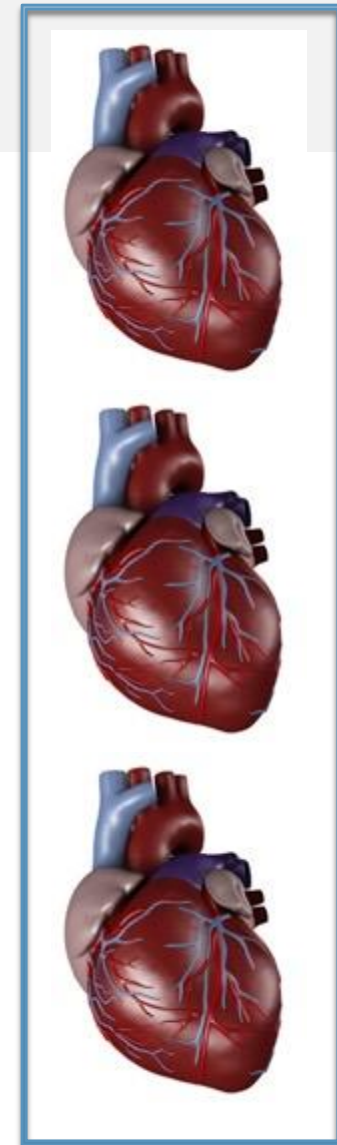
Partie  
d'objet  
décrétée

Objet

Frontière  
décrétée

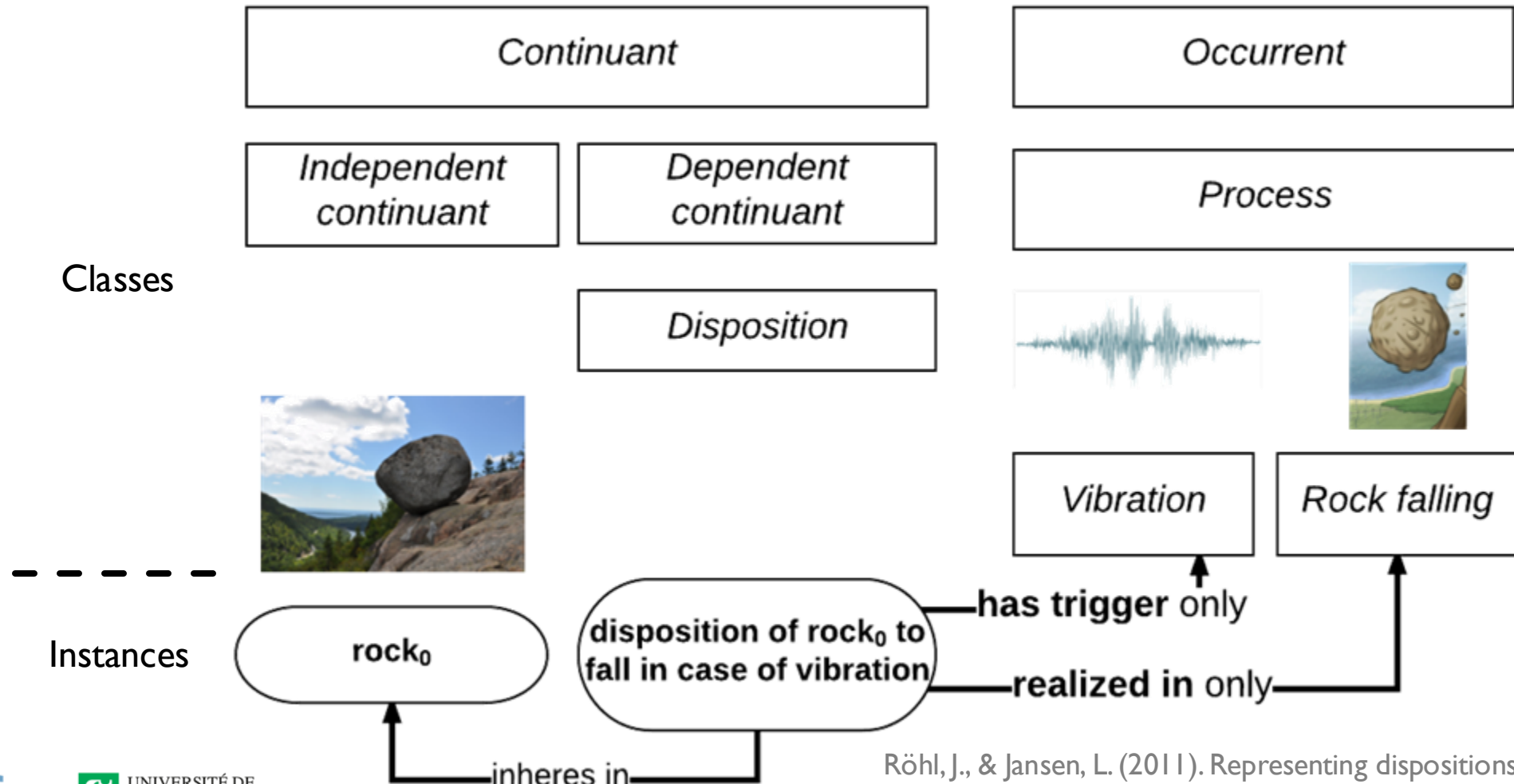
Site  
Région spatiale

- **Qualité** de la forme du coeur
- **Disposition** d'un coeur à se nécroser en l'absence d'apport d'oxygène
- **Fonction** du coeur de pompe
- **Rôle** du coeur de repas d'un lion

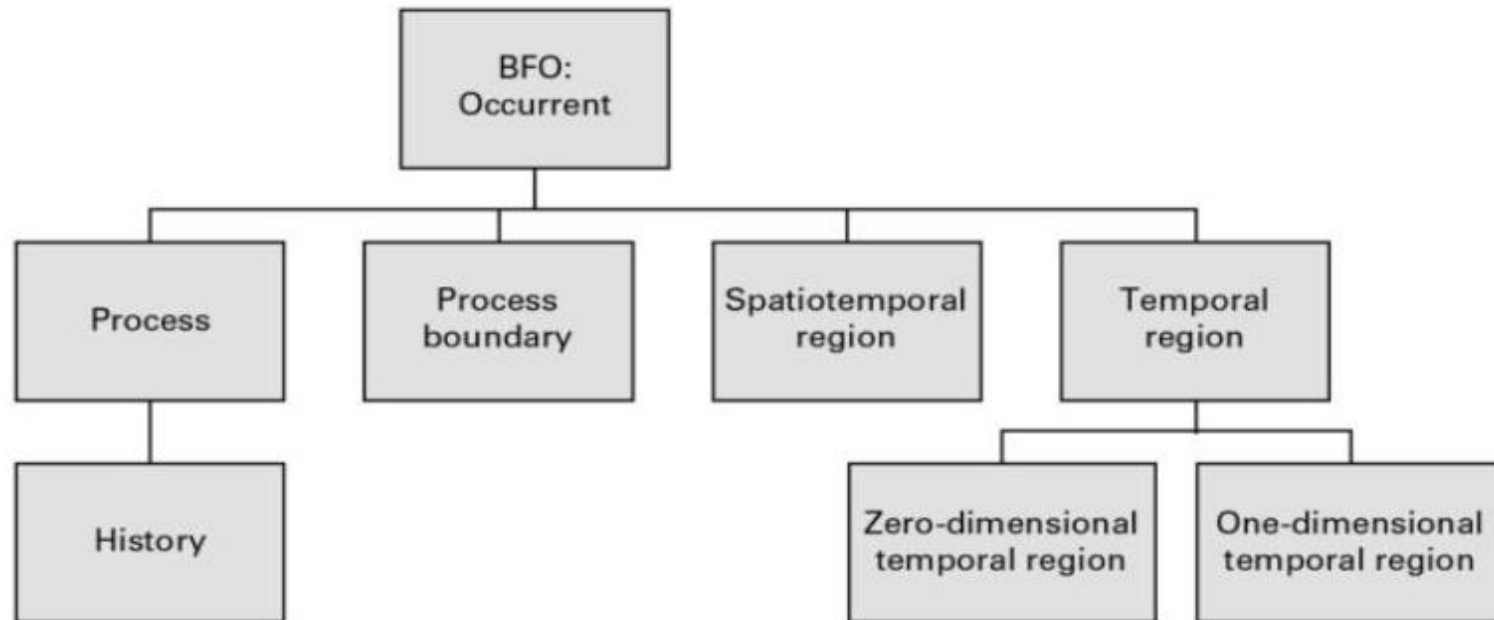


Aggrégat  
d'objets

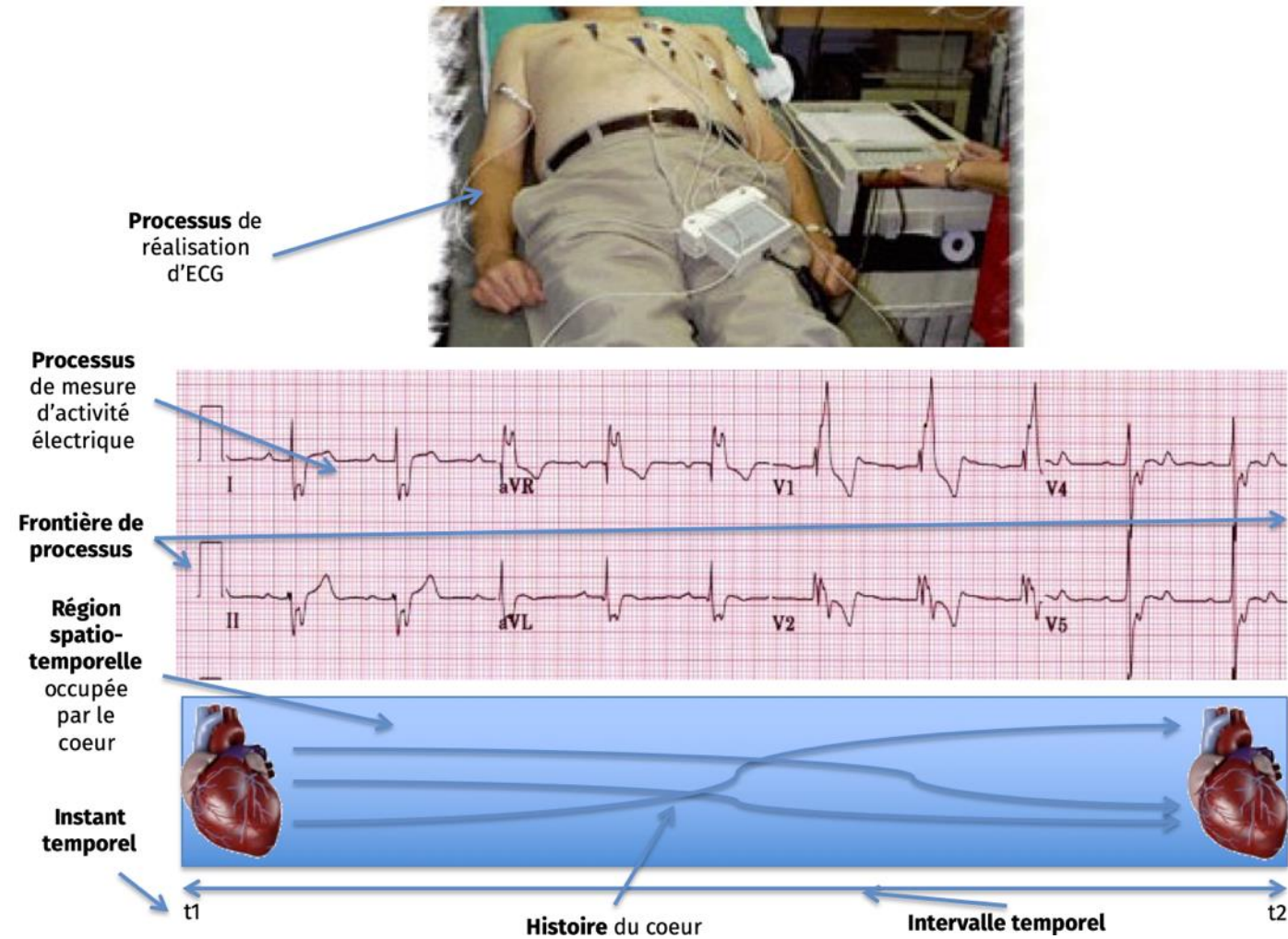
# Un type important de propriété : les dispositions



# BFO, occurrents



# Occurrents : exemples





# Cirrhosis - environmental exposure

- Etiological process - phenobarbital-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 occurrent part' **some** 'left ventricular myocardium hypertrophy')  
and ( 'has occurrent 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 +

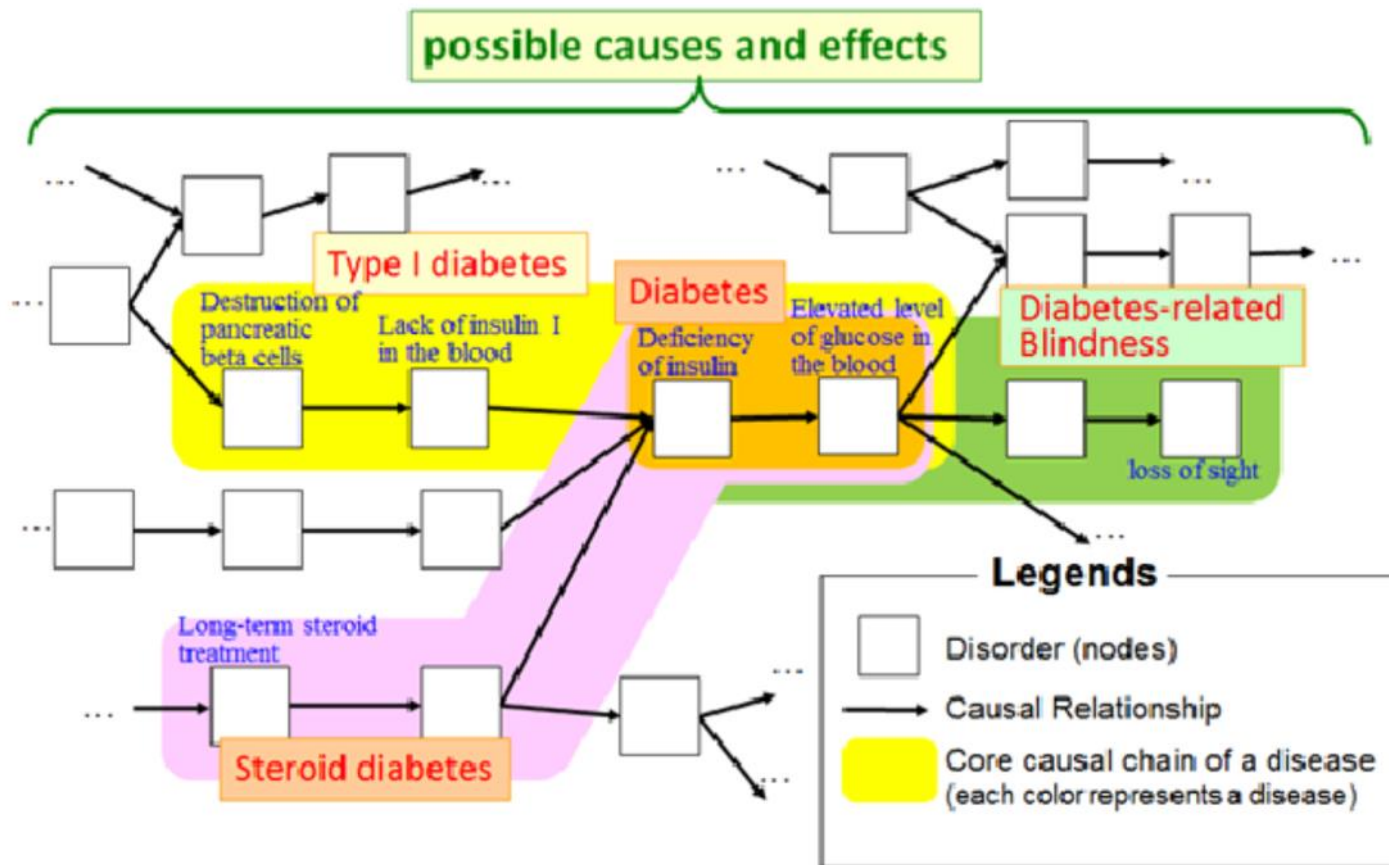
- 'cardiac channelopathy'
- 'has material basis at all times' **some** 'genetic disorder'
- 'realized in' **only**  
( 'disease course'  
and ( 'has occurrent part' **some** 'ventricular tachyarrhythmia (process)'))

- 'genetic cardiac channelopathy'

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

# River-flow model of disease

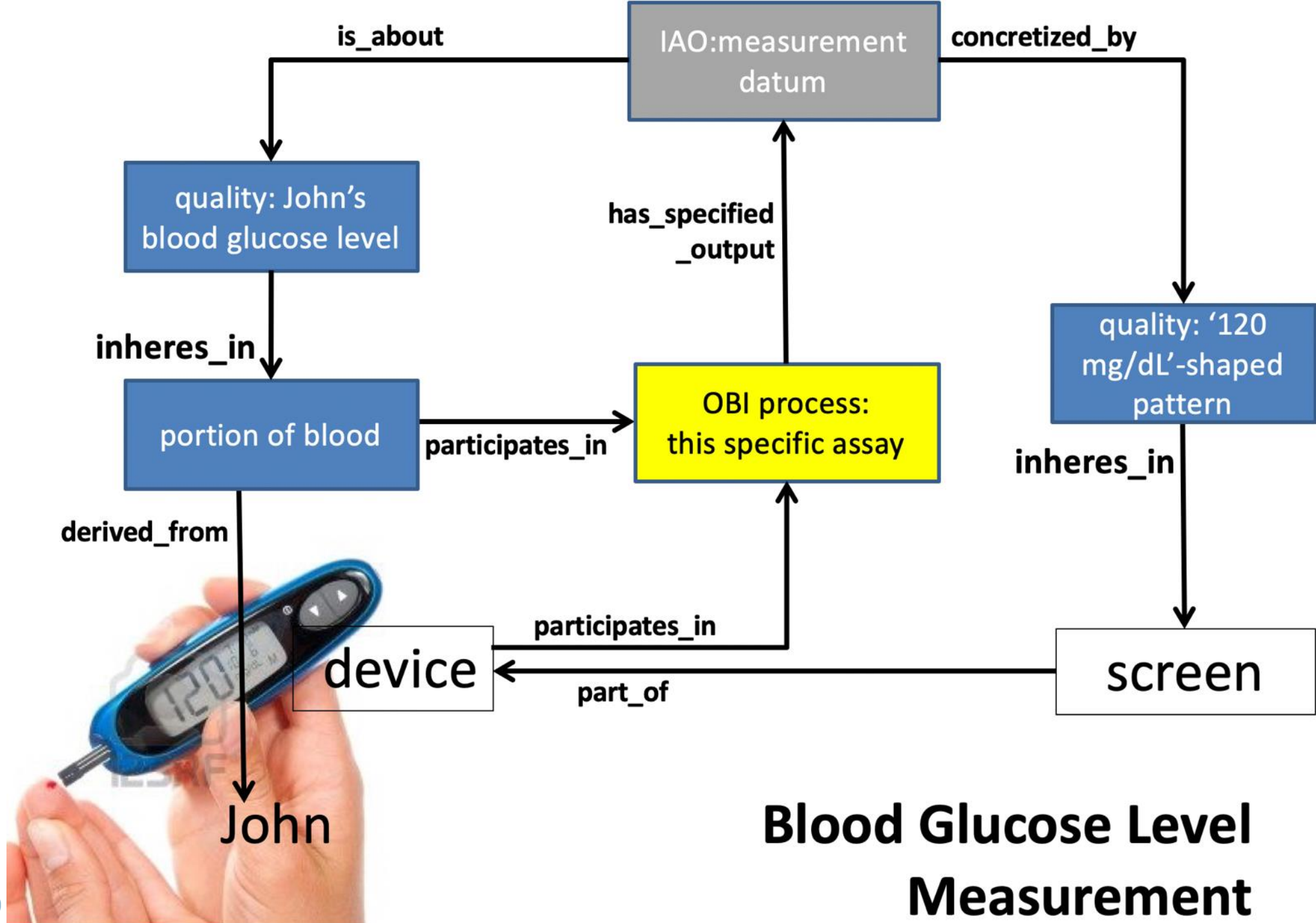
(R. Mizoguchi, K. Kozaki, H. Kou, Y. Yamagata, T. Imai, K. Waki, and K. Ohe. (2011). River Flow Model of Diseases. In Proceedings of the 2nd International Conference on Biomedical Ontology (ICBO2011) (pp. 63-70))



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 document 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 :

- **Formulaire 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.