

INFECTIOUS DISEASE ONTOLOGY (IDO)

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OVERVIEW

- Goals of IDO project
- IDO architecture
- Glimpse of IDO-Core
- Principle behind building extensions
- Inferring interconnected hierarchies
- New relations

NEED FOR INFECTIOUS DISEASE ONTOLOGIES

- Cover the entire infectious disease domain
 - diseases, hosts, pathogens
 - biological scale (molecule, cell, organism, population)
 - entities in the chain of infection (host, pathogen, vector, reservoir)

CHARACTERIZATION OF AN INFECTIOUS DISEASE

disease	signs and symptoms	diagnostic criteria	disease course stages
host organism type	relevant polymorphisms	host niches	endemic areas
pathogen organism type	pathogen classification	virulence traits	drug susceptibilities
host-pathogen interaction	infection process	host response	epidemiological characteristics
routes of transmission	vector organism types	reservoirs	prevention measures

DEVELOPMENT STRATEGY

- Developed within the OBO Foundry (<http://www.obofoundry.org>)
 - common set of non-overlapping, interoperable ontologies for the biomedical domain
- Core-Extension Approach

OBO FOUNDRY

- Single ontology for each portion of the domain
 - Collaboratively developed
- Common organization
 - Basic Formal Ontology
- Common set of formally defined relations
 - Relation Ontology
- Principled definitions for types
- Availability in a computable format
 - e.g. OWL or OBO

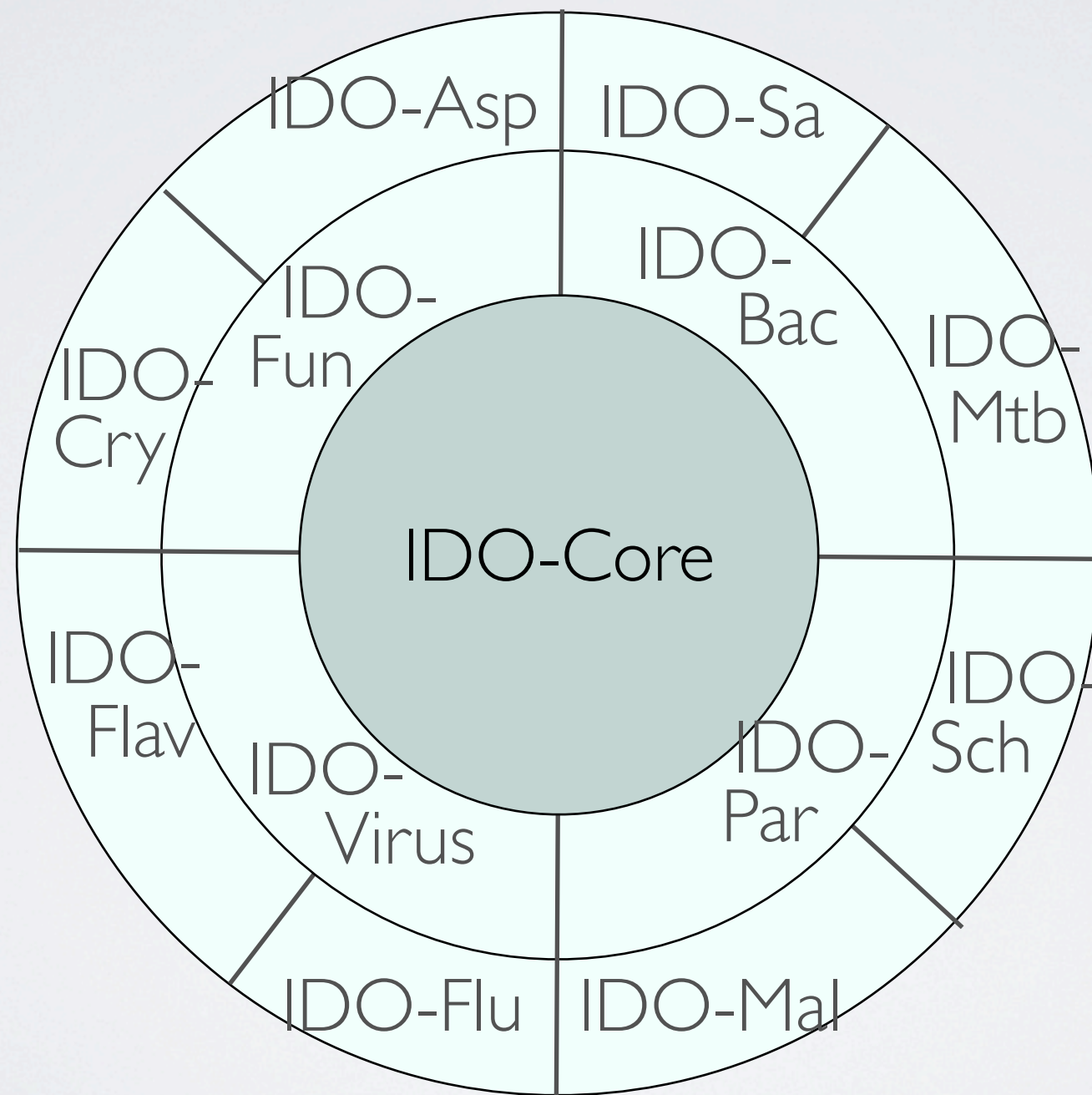
IDO DEVELOPMENT

- importing terms from OBO Foundry ontologies
- defining new terms as needed, adhering to OBO Foundry principles
- enriching the relations asserted between terms

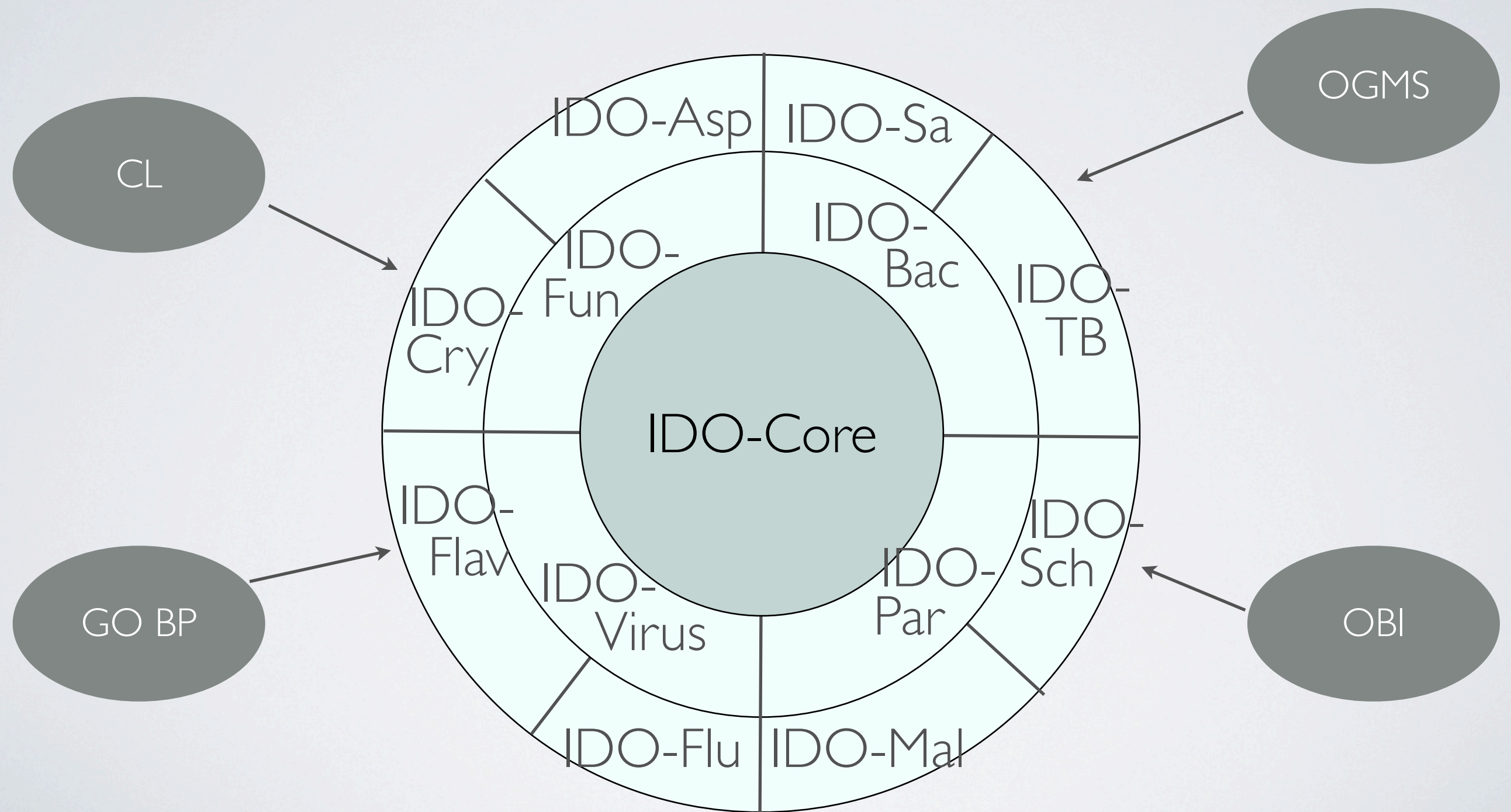
ONTOLOGIES USED BY IDO

Clinical Entities	Biological Entities	Investigational Entities
Ontology of General Medical Science	NCBI Taxonomy	Ontology of Biomedical Investigation
Vital Signs Ontology	Common Anatomy Reference Ontology	
Symptom Ontology	Cell Ontology	
Disease Ontology	Gene Ontology Cellular Component Ontology	
	Protein Ontology	
	Chemical Entities of Biological Interest	
	Phenotype and Trait Ontology	
	Environment Ontology	
	Gene Ontology Biological Process Ontology	

IDO CORE-EXTENSION APPROACH



IDO AND OBO FOUNDRY ONTOLOGIES



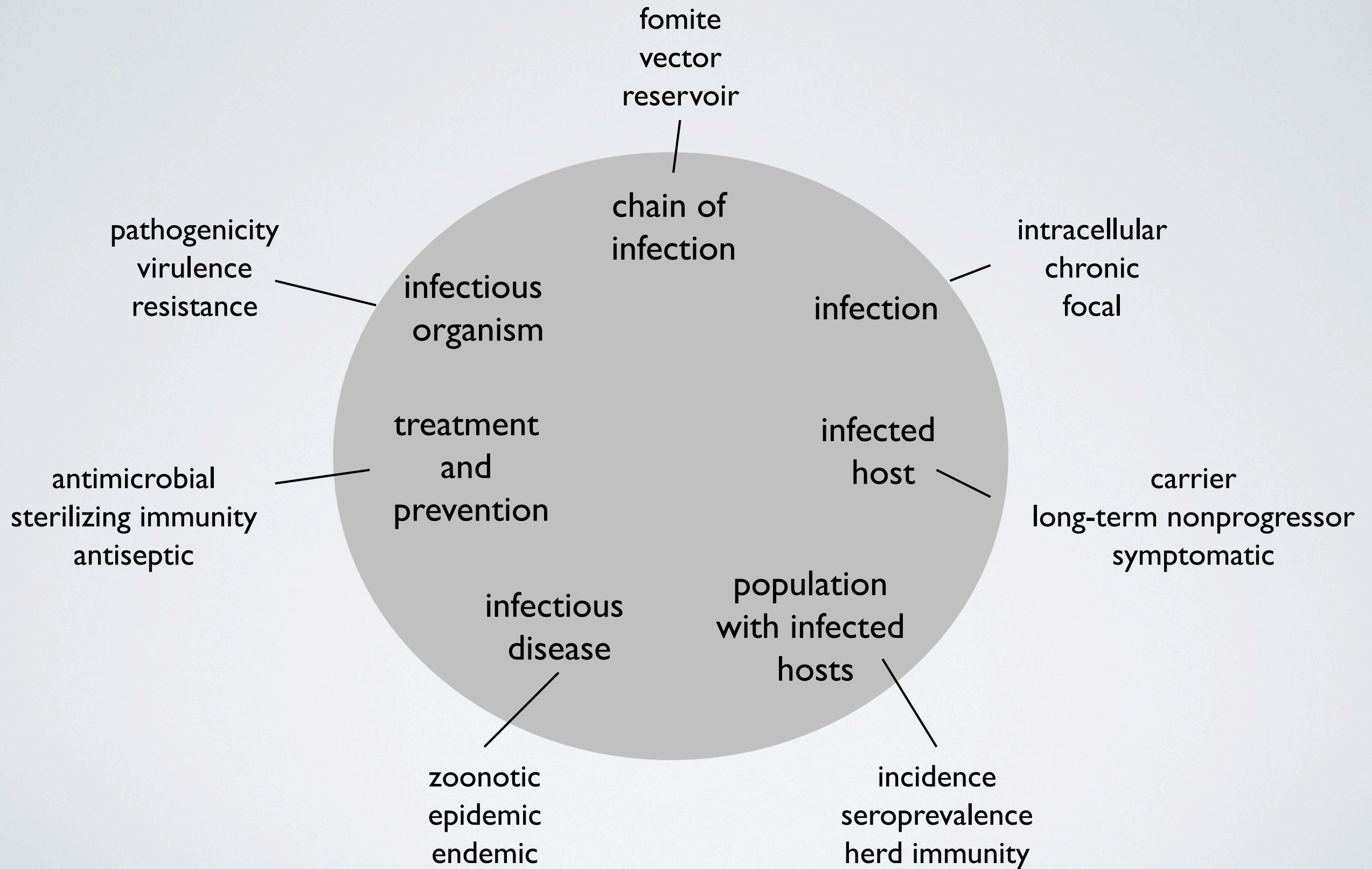
PURPOSE OF IDO-CORE

- Provide terms relevant to infectious diseases generally (e.g. infection, host, pathogen, vector)
 - reducing duplication of effort
- Ensure interoperability between IDO extensions
 - terminological consistency (term names and meanings)
 - definition templates
 - consistent approach to classification
 - consistent approach to asserting relations
- Allows parallel development by domain experts
 - prevent common mistakes
 - ensure utility for computational applications

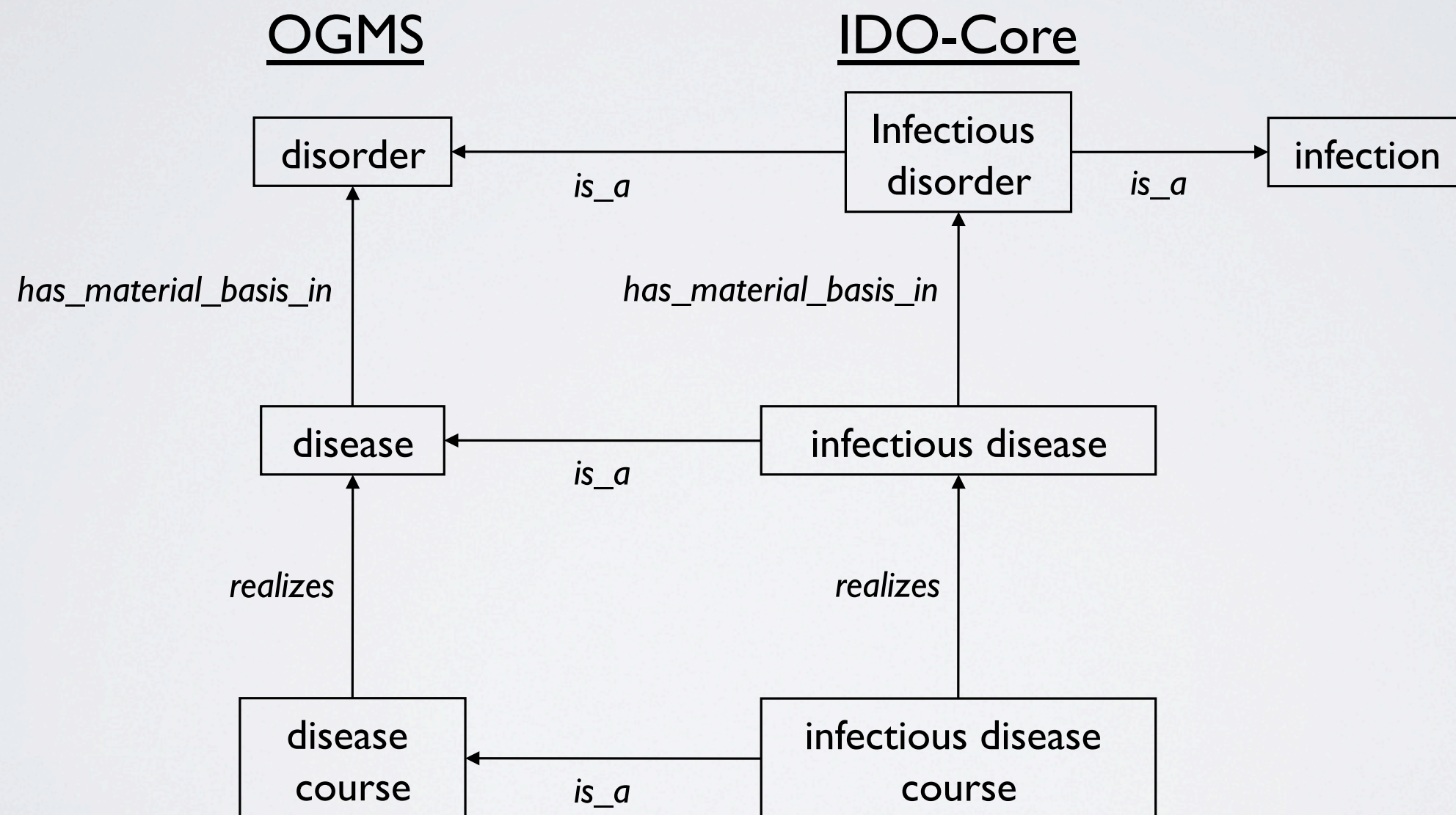
USERS

- Staphylococcus aureus
 - Vance Fowler - Duke University Medical Center
- Brucellosis
 - Oliver He - University of Michigan
- Salmonella
 - Ina Hulsegge - Animal Breeding and Genomics Centre
- Influenza
 - Richard Scheuermann, Burke Squires - UT Southwestern Medical Center
 - Melanie Courtot - BC Cancer Research Center
 - Lynn Schriml - University of Maryland
 - Joanne Luciano - Rensselaer Polytechnic Institute
- HIV
 - Stanley Schwartz, Alex Diehl - University at Buffalo
- Vector-borne diseases (Malaria)
 - Kitsos Louis, Pantelis Topalis - IMBB
- Eukaryotic pathogens
 - Chris Stoeckert - University of Pennsylvania
- Sepsis Use Case in Vital Signs Ontology
 - Albert Goldfain - Blue Highway
- APOLLO SV
 - Bill Hogan - University of Arkansas

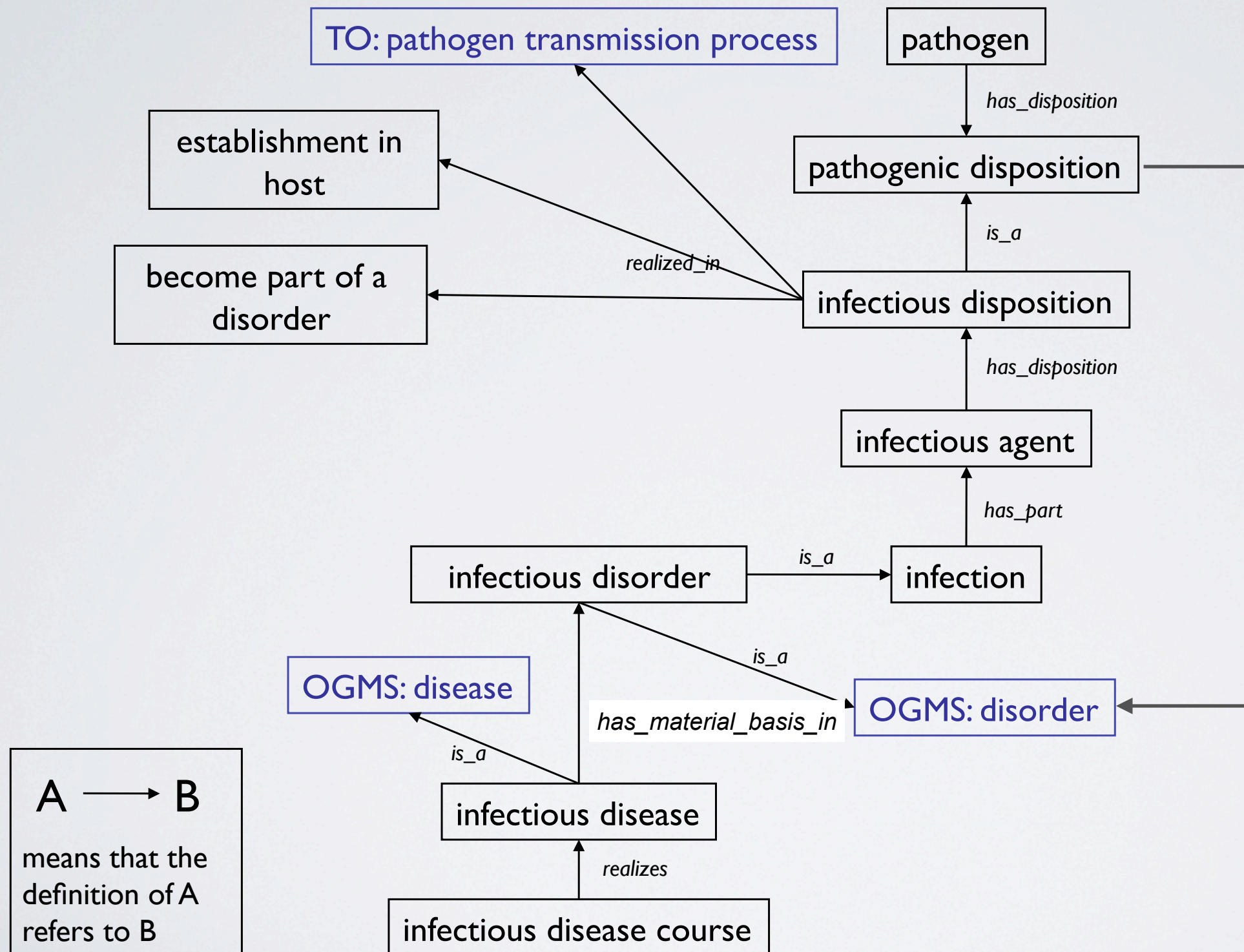
SCOPE OF IDO-CORE



IDO-CORE REPRESENTATION OF INFECTIOUS DISEASE



IDO-CORE CENTRAL TERMS



IDO EXTENSION: TERMS AND DEFINITIONS

- *Staphylococcus aureus* infection $=_{\text{def}}$ An infection that has as part organisms of type *Staphylococcus aureus*.
- *Staphylococcus aureus* bacteremia $=_{\text{def}}$ An infection that has as part organisms of type *Staphylococcus aureus* located in the blood.

IDO EXTENSION: TERMS AND DEFINITIONS

- drug resistance $=_{\text{def}}$ A protective resistance that mitigates the damaging effects of a drug.
- antibiotic resistance $=_{\text{def}}$ A drug resistance that mitigates the damaging effects of an antibiotic.
- resistance to beta-lactam antibiotic $=_{\text{def}}$ An antibiotic resistance that mitigates the damaging effects of a beta-lactam antibiotic.

IDO EXTENSION: TERMS AND DEFINITIONS

- methicillin-resistant *Staphylococcus aureus* =_{def} An organism of type *Staphylococcus aureus* that has resistance to beta-lactam antibiotics.

IDO-CORE AND EXTENSIONS: LOGICAL DEFINITIONS

- useful for error and consistency checking
- connect IDO terms to terms in external ontologies
- utilized for the creation of inferred hierarchies

IDO-CORE AND EXTENSIONS: LOGICAL DEFINITIONS

- infection subClassOf
 - (*part_of* SOME 'extended organism') AND
 - (*has_part* SOME 'infectious agent')
- bacteremia equivalentClass
 - infection AND
 - *has_part* SOME
 - (bacteria AND *located_in* SOME blood)

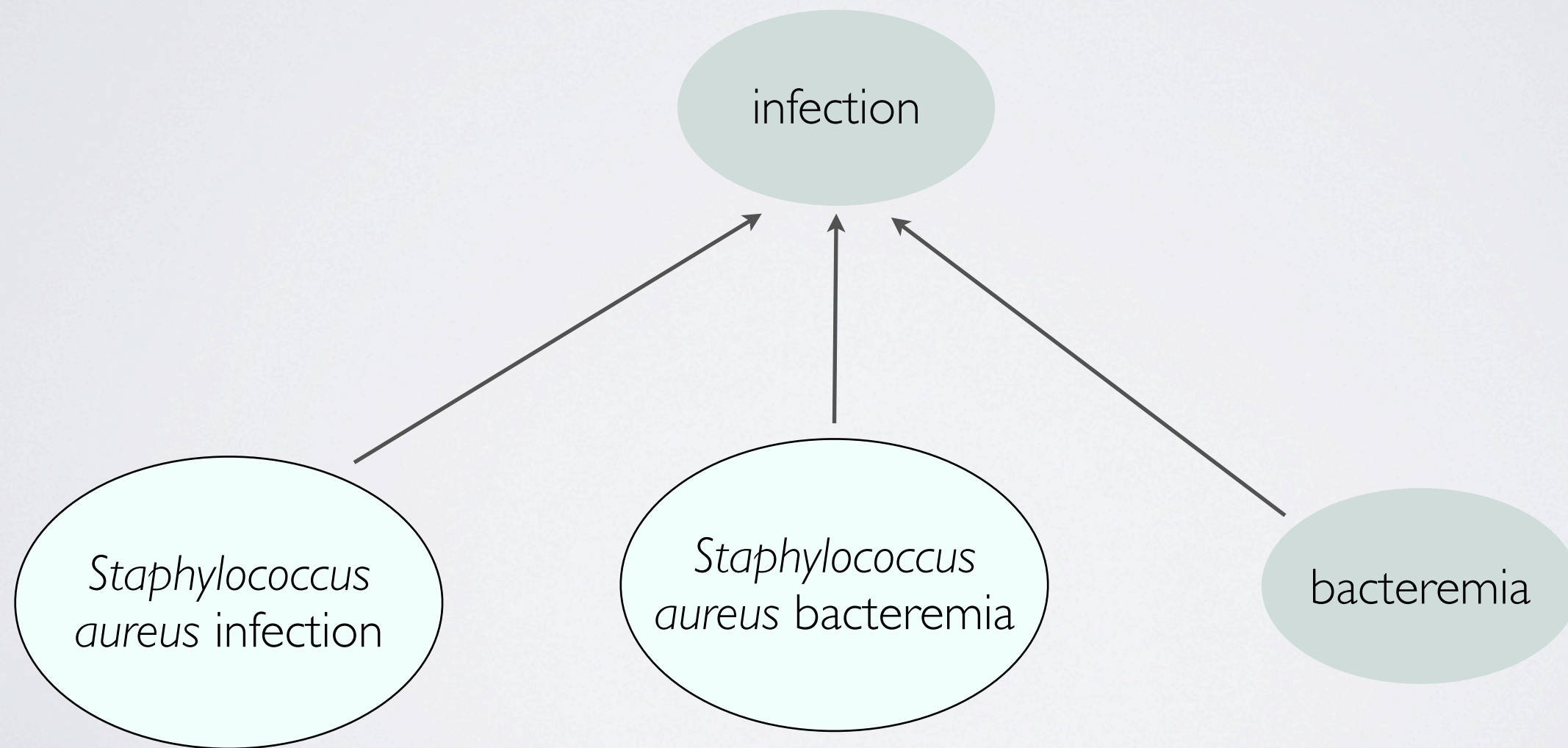
IDO-CORE AND EXTENSIONS: LOGICAL DEFINITIONS

- infection subclassOf
 - (part_of SOME 'extended organism') AND
 - (has_part SOME 'infectious agent')
- *Staphylococcus aureus* infection equivalentClass
 - infection AND
 - has_part SOME '*Staphylococcus aureus*'

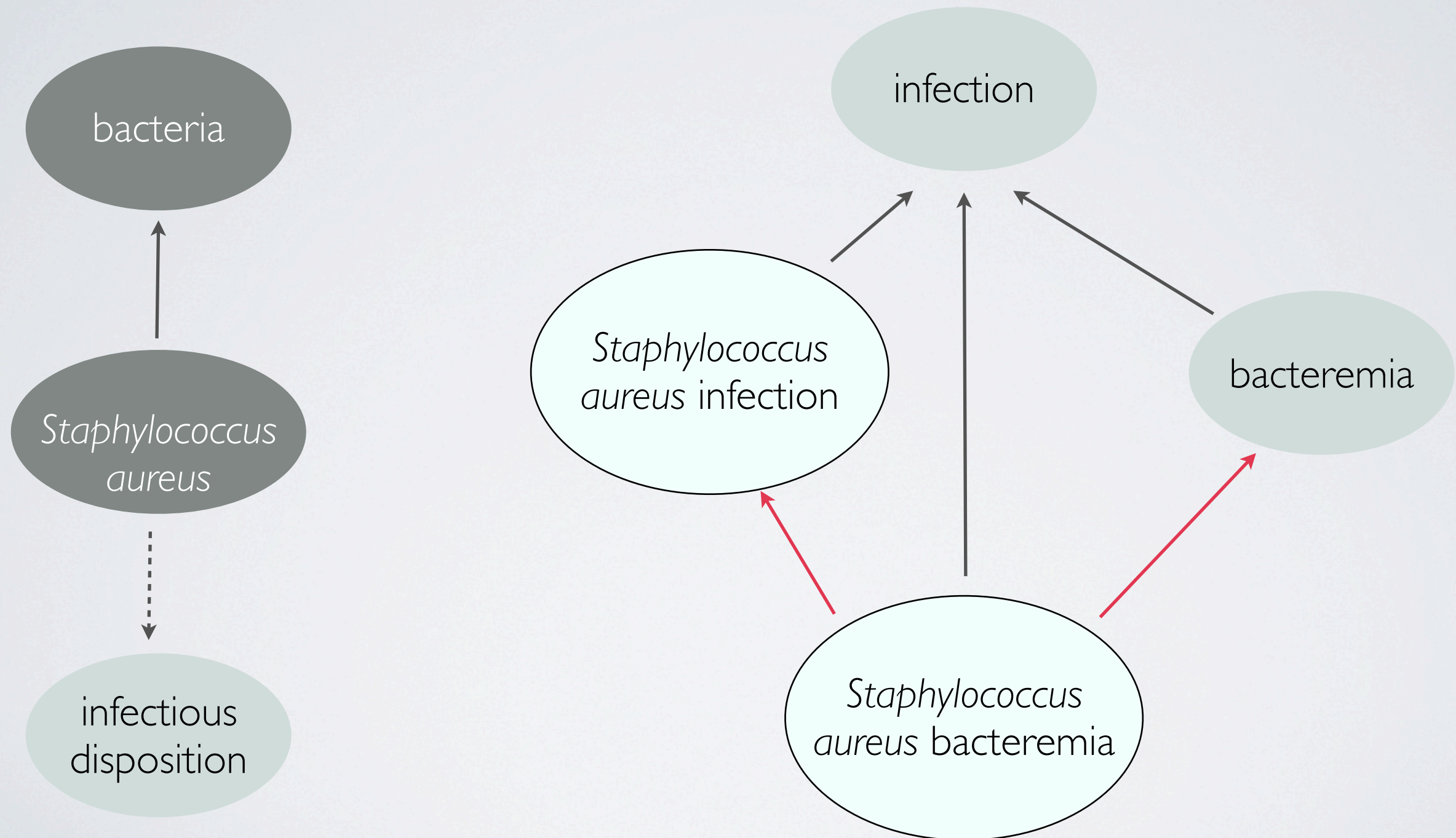
IDO-CORE AND EXTENSIONS: LOGICAL DEFINITIONS

- bacteremia equivalentClass
 - infection AND
 - has_part SOME [
 - (bacteria AND has_disposition SOME 'infectious disposition') AND
 - located_in SOME blood]
- *Staphylococcus aureus* bacteremia equivalentClass
 - infection AND
 - has_part SOME [
 - *Staphylococcus aureus* AND
 - located_in SOME blood]

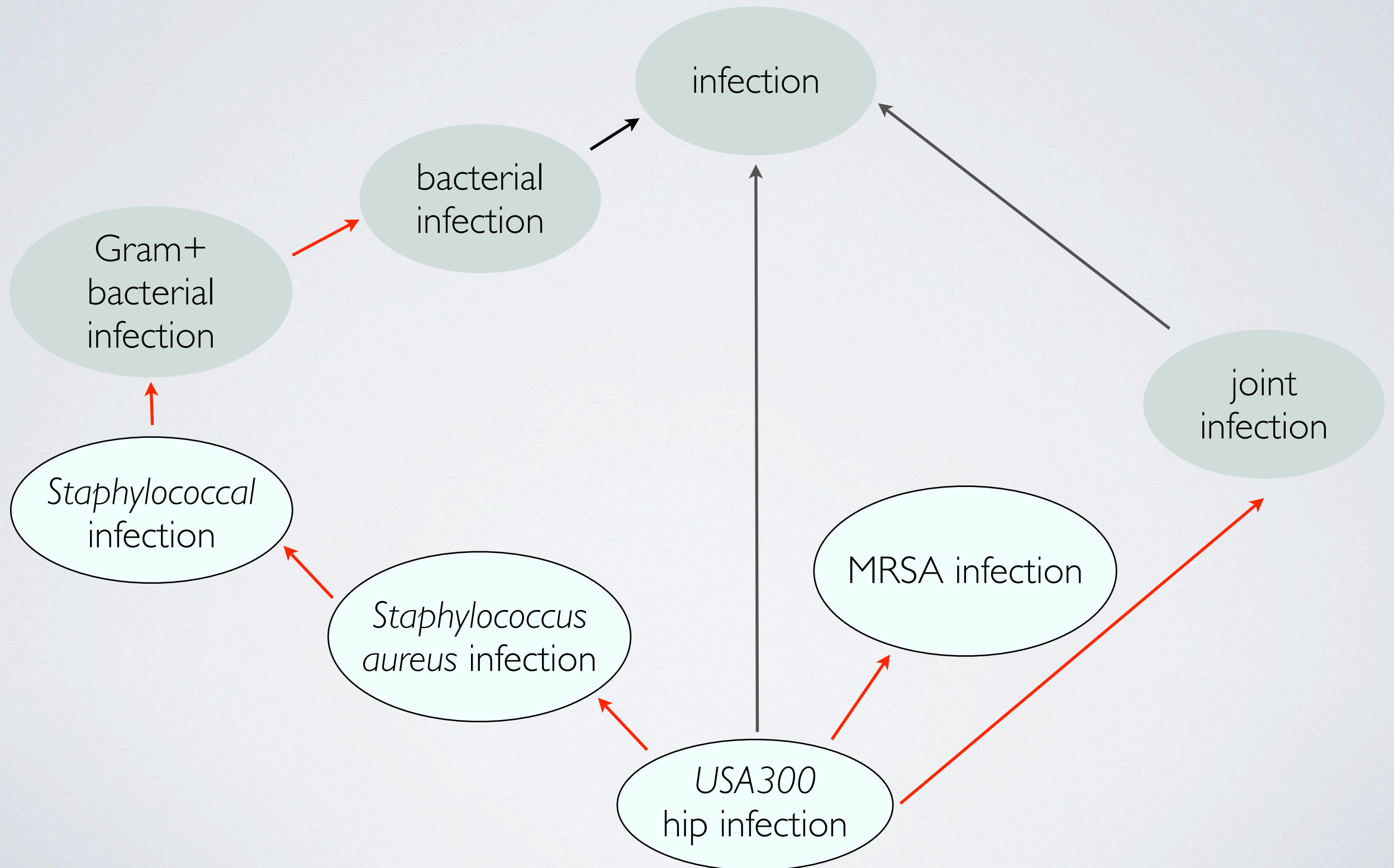
IDO-CORE AND EXTENSIONS: ASSERTED HIERARCHY



IDO-CORE AND EXTENSIONS: INFERRED HIERARCHY



IDO-CORE AND EXTENSIONS: INFERRED HIERARCHY



RELATION BETWEEN DISEASE AND INFECTIOUS AGENT

d* has_infectious_agent *a* at *t =_{def} there exists some *i* and some *o* such that

d* instance_of** IDO:*infectious disease* **at *t

a* instance_of** IDO:*infectious agent* **at *t

i* instance_of** IDO:*infectious disorder* **at *t

o* instance_of** OGMS:*extended organism* **at *t

d* inheres_in *o* at *t

d* has_material_basis *i* at *t implies ***i* has_part *a* at *t***

d* has_material_basis *i* at *t =_{def}

(*d* instance_of BFO:*disposition* **at *t*** and

i* instance_of** BFO:*material entity* **at *t and

o* instance_of** BFO:*material entity* **at *t and

o* has_disposition *d* at *t*)** implies ***o* has_part *i* at *t

RELATION BETWEEN DISEASE AND INFECTIOUS AGENT

$D \text{ has_infectious_agent } A =_{\text{def}} \text{ for all } d \text{ and all times } t,$
if d **instance_of** D **at** t , then there exists some a such that
 a **instance_of** A **at** t and d **has_infectious_agent** a **at** t

RELATION BETWEEN DISEASE AND VECTOR

d **has_vector** v **at** t_2 =_{def} there exists some v such that
 (t_1 **earlier** t_2 and
 d **instance_of** *IDO:infectious disease* **at** t_2 and
 v **instance_of** *OGMS:extended organism* **at** t_1 and
 o **instance_of** *OGMS:extended organism* **at** t_1 and t_2 and
 a_1 **instance_of** *IDO:infectious agent* **at** t_1 and
 a_2 **instance_of** *IDO:infectious agent* **at** t_2 and
 r **instance_of** *IDO:vector role* **at** t_1 and
 t **instance_of** *TRANS:vector-borne transmission process* and
 d **inheres_in** o **at** t_2 and
 d **has_infectious_agent** a_2 **at** t_2) implies
 ($(a_1 = a_2$ or a_2 **derives_from** a_1) and
 t **occurring_at** t_1 and
 t **has_participant** o **at** t_1 and
 t **has_participant** a_1 **at** t_1 and
 t **has_participant** v **at** t_1 and
 r **inheres_in** v **at** t_1 and
 t **realizes** v **at** t_1)

RELATION BETWEEN DISEASE AND VECTOR

$D \text{ has_vector } V =_{\text{def}}$ for all d and all times t ,
if d **instance_of** D **at** t_2 , then there exists some v at t_1 such that
 v **instance_of** V **at** t_1 and d **has_vector** v **at** t_2 and t_1 **earlier** t_2

UPCOMING RELEASE

- Base on BFO 2.0
- Simplified natural language definitions
- Extended IDO-core with MIREOTed terms
- Relations

ACKNOWLEDGEMENTS

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- IDO-Core Contributors

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- Pantelis Topalis



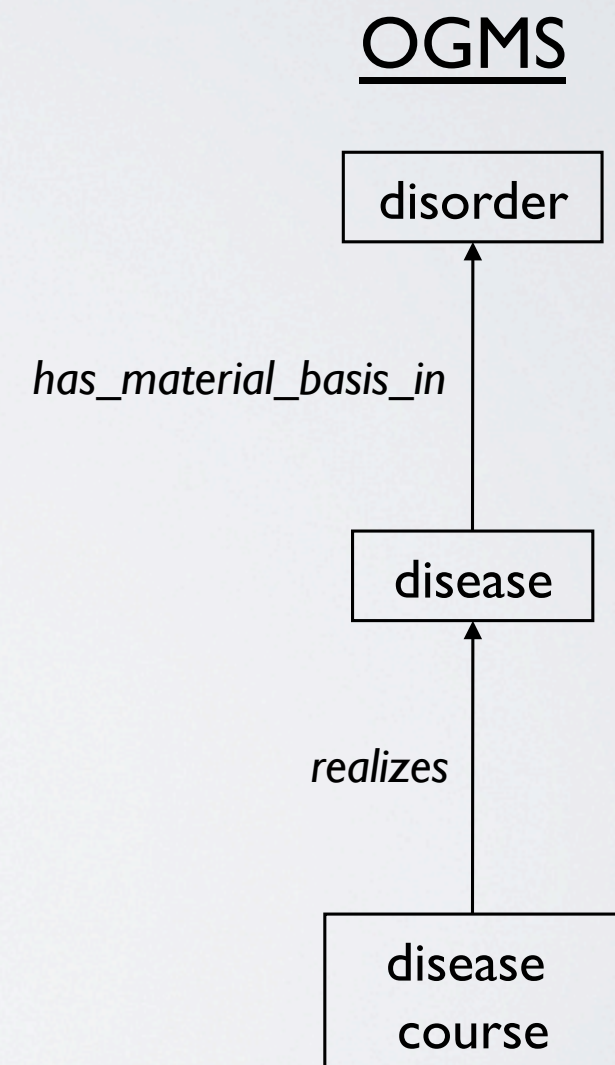
National Institute of Allergy and Infectious Diseases
National Institutes of Health

OGMS REPRESENTATION OF DISEASE

“We use clinically abnormal to characterize those qualities of or in an organism that are causally linked to an elevated risk of pain or other feelings of illness, to dysfunction, or to enhanced morbidity”

OGMS REPRESENTATION OF DISEASE

- disease \equiv_{def} A disposition (i) to undergo pathological processes that (ii) exists in an organism because of one or more disorders in that organism.
- disease course \equiv_{def} The totality of all processes through which a given disease instance is realized.

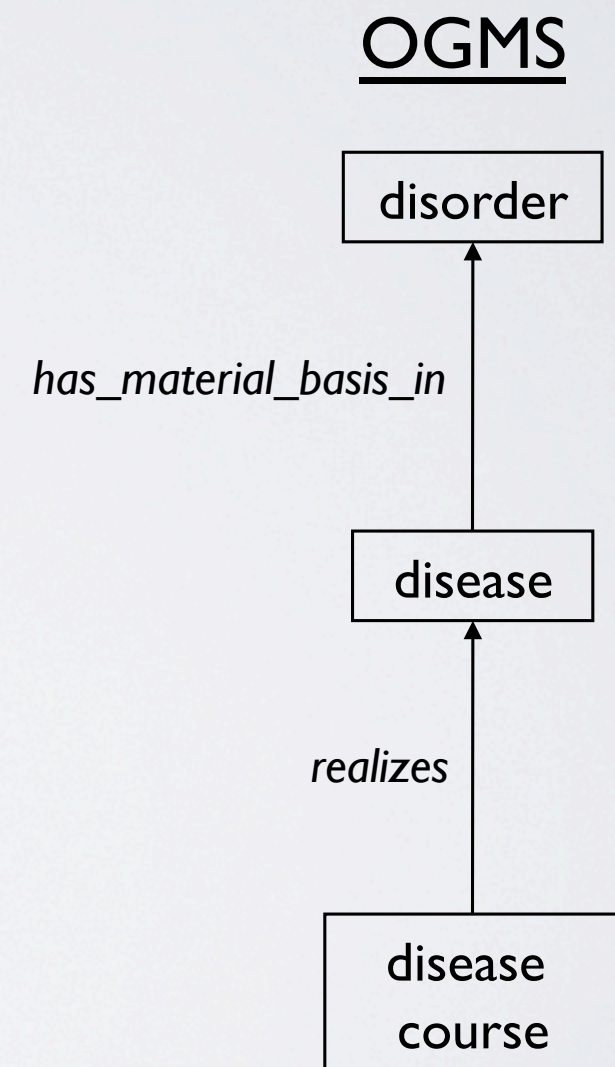


OGMS REPRESENTATION OF DISEASE

“We use clinically abnormal to characterize those qualities of or in an organism that are causally linked to an elevated risk of pain or other feelings of illness, to dysfunction, or to enhanced morbidity, and which (unlike pregnancy or menopause) are not such as to belong to the life plan for an organism of the relevant type. A clinician will judge a quality to be clinically abnormal only where the elevated risk exceeds a certain threshold level of clinical significance.”

OGMS REPRESENTATION OF DISEASE

- disorder =_{def} A material entity which is clinically abnormal and part of an extended organism.
- disease =_{def} A disposition (i) to undergo pathological processes that (ii) exists in an organism because of one or more disorders in that organism.
- disease course =_{def} The totality of all processes through which a given disease instance is realized.



IDO-CORE CENTRAL DEFINITIONS

pathogenic disposition	A disposition to initiate processes that result in a disorder.
pathogen	A material entity with a pathogenic disposition.
infectious disposition	A pathogenic disposition that inheres in an organism and is the disposition for that organism to be transmitted to a host, (2) to establish itself in the host, and (3) to become part of a disorder in the host.
infectious agent	An organism with an infectious disposition.

IDO-CORE CENTRAL DEFINITIONS

- infection \equiv_{def} A part of an extended organism that itself has as part a population of one or more infectious agents and that is
 - (1) clinically abnormal in virtue of the presence of this infectious agent population, or
 - (2) has a disposition to bring clinical abnormality to immunocompetent organisms of the same Species as the host through transmission of a member or offspring of a member of the infectious agent population.

IDO-CORE CENTRAL DEFINITIONS

infectious disorder	An infection that is clinically abnormal.
infectious disease	A disease whose physical basis is an infectious disorder.
infectious disease course	A disease course that is a realization of an infectious disease.

IDO EXTENSION: TERMS AND DEFINITIONS

- bacteremia $=_{\text{def}}$ An infection that has as part bacteria located in the blood.
- Staphylococcal infection $=_{\text{def}}$ An infection that has as part organisms of type *Staphylococcus*.
- *Staphylococcus aureus* infection $=_{\text{def}}$ An infection that has as part organisms of type *Staphylococcus aureus*.
- *Staphylococcus aureus* bacteremia $=_{\text{def}}$ An infection that has as part organisms of type *Staphylococcus aureus* located in the blood.