INFECTIOUS DISEASE ONTOLOGY 2010

December 8 - 9, 2010
Baltimore, Maryland
Sponsored by the National Center for Biomedical Ontology

INTRODUCTION TO THE INFECTIOUS DISEASE ONTOLOGY (IDO)

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TALK OUTLINE

- Need for infectious disease ontologies

- IDO Suite of Ontologies
  - IDO Development Strategy

- Scope and Content of IDO-Core
  - OGMS Representation of Disease
  - IDO-Core Central Terms

- Example IDO Extension Terms

BENEFITS OF BIOMEDICAL ONTOLOGIES

- Generic Model Organism Databases

- GO term analysis for gene expression microarray data

BENEFITS FOR INFECTIOUS DISEASE RESEARCHERS

- data integration for infectious disease research
  - for particular diseases
    - integration of data across studies and institutions
      - genetic susceptibility to *S. aureus* infection
      - Centers for AIDS Research
    - within-site integration of different kinds of data
      - clinical, host genetic, microbiology, and flow cytometry data
  - for co-infection studies

- analysis of high-throughput data types

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)

DEVELOPMENT STRATEGY

- Developed within the OBO Foundry (http://www.obofoundry.org)
  - ensure interoperability with Foundry ontologies (e.g. GO) and the information resources that use them

- 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

ONTOLOGIES USED BY IDO

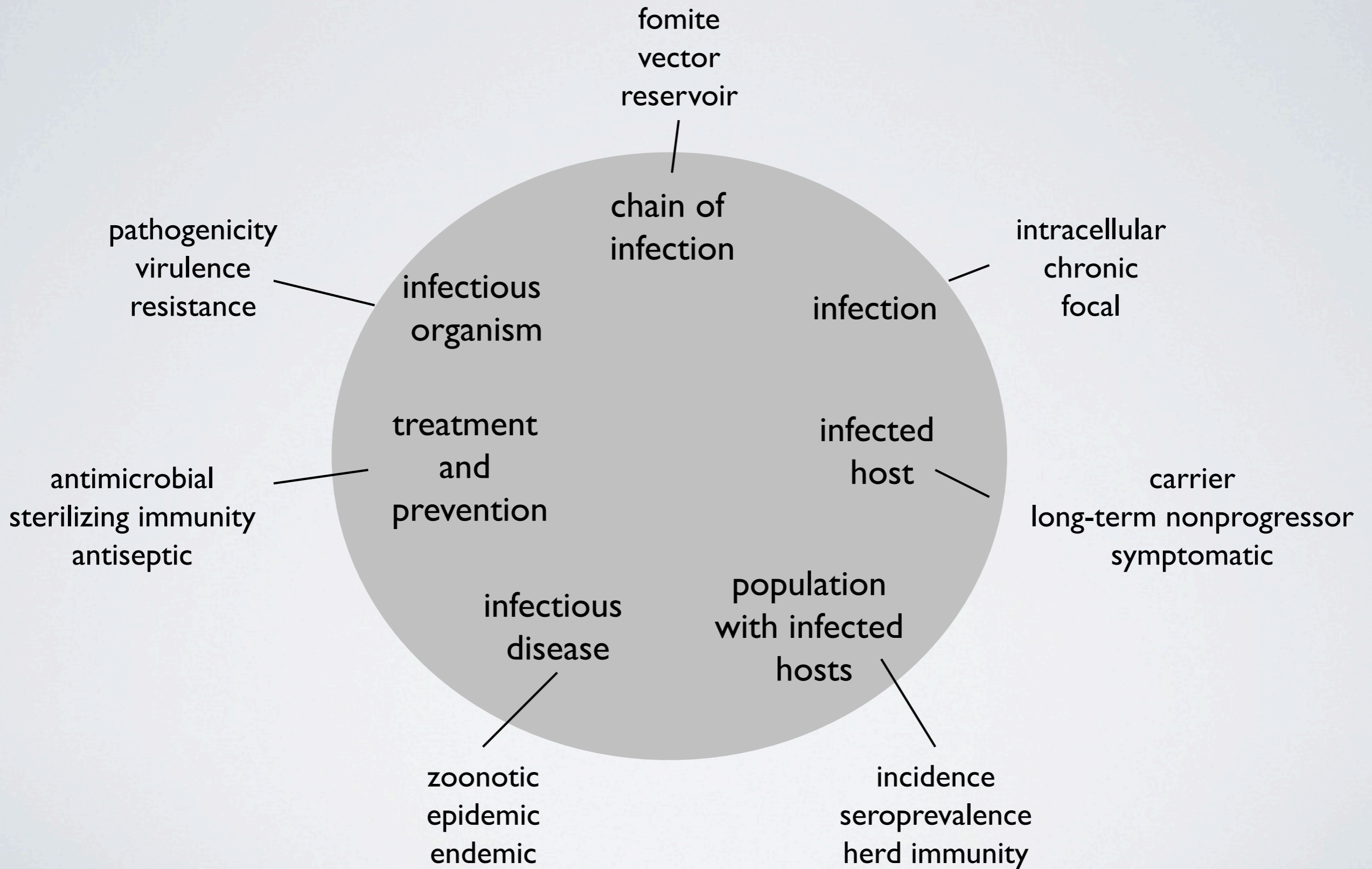| Clinical Entities	Biological Entities	Investigational Entities
Ontology of General Medical Science	NCBI Taxonomy	Ontology of Biomedical Investigation
Vital Signs Ontology	Common Anatomy Reference Ontology	
Symptom Ontology	Cell Ontology	
Disease Ontology	Gene Ontology Cellular Component Ontology	
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

IDO CORE-EXTENSION APPROACH
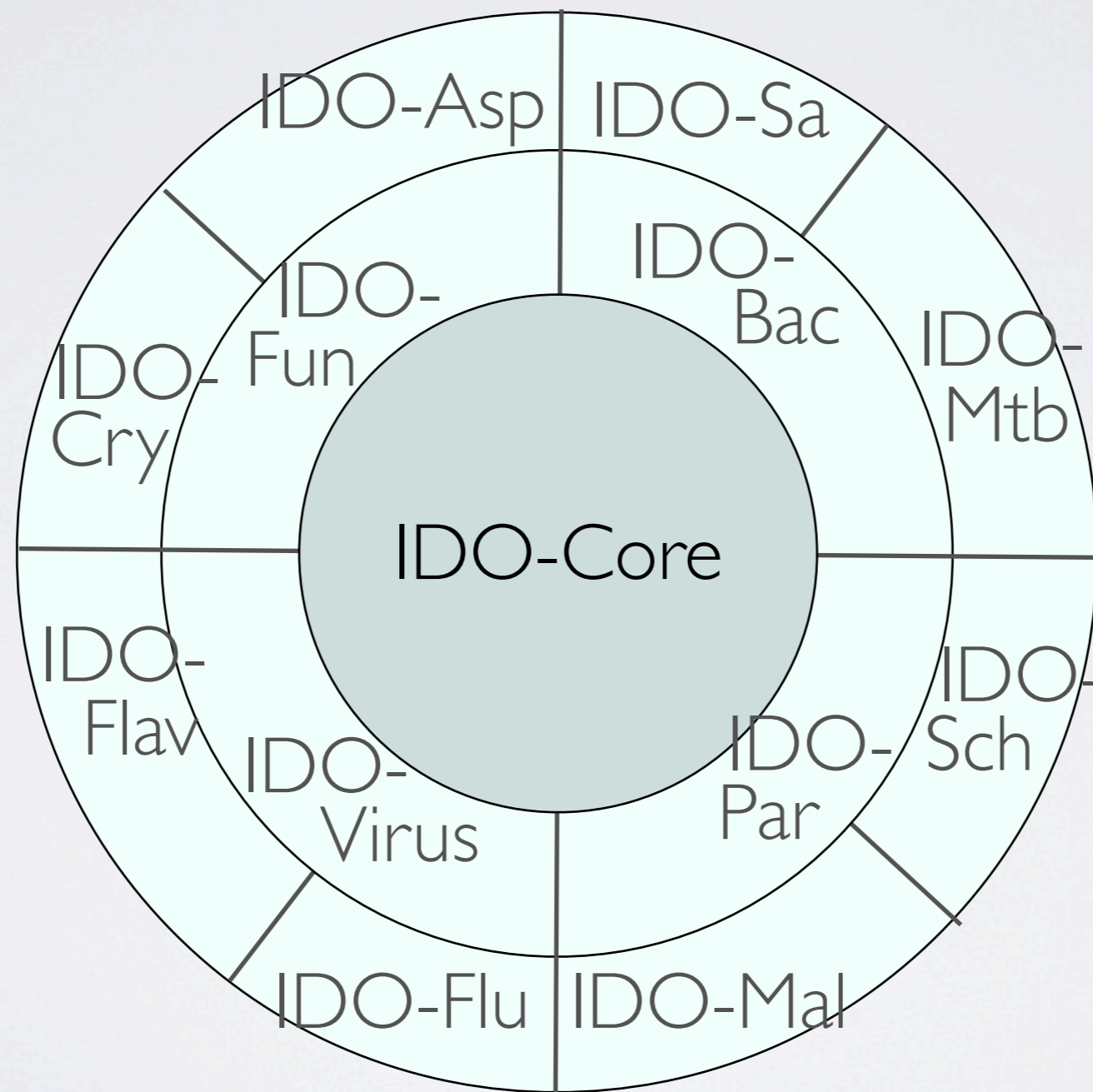
IDO AND OBO FOUNDRY ONTOLOGIES
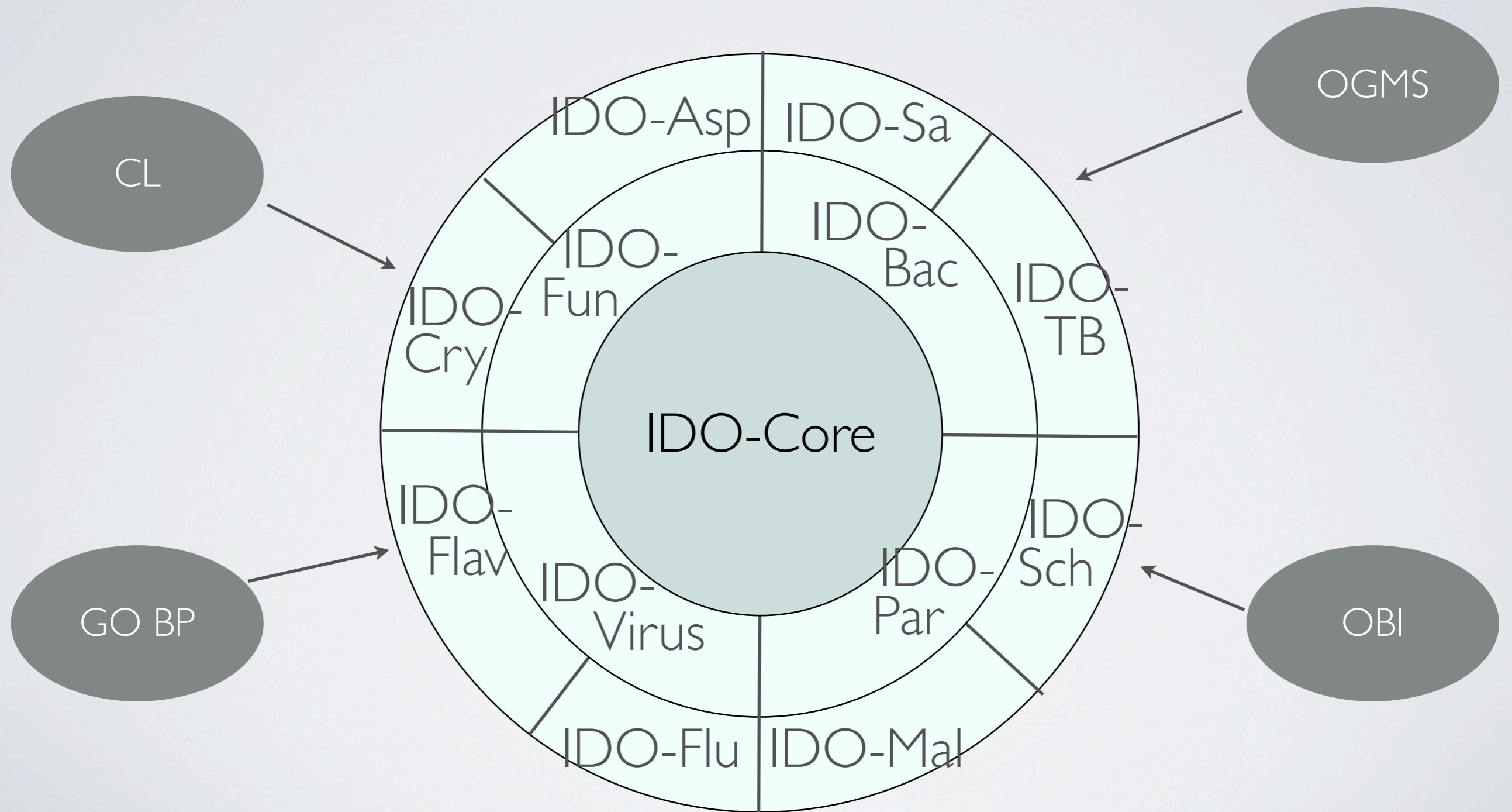
PURPOSE OF IDO-CORE

- Provide terms relevant to infectious diseases generally (e.g. infection, host, pathogen, vector)

- 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

SCOPE OF IDO-CORE

fomite
vector
reservoir

chain of
infection

pathogenicity
virulence
resistance

infectious
organism

intracellular
chronic
focal

infection

treatment
and
prevention

infected
host

antimicrobial
sterilizing immunity
antiseptic

carrier
long-term nonprogressor
symptomatic

infectious
disease

population
with infected
hosts

zoonotic
epidemic
endemic

incidence
seroprevalence
herd immunity

OGMS REPRESENTATION OF DISEASE

"We use <u>clinically abnormal</u> 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 =_{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.

OGMS

disorder

has_material_basis_in

disease

realizes

disease course
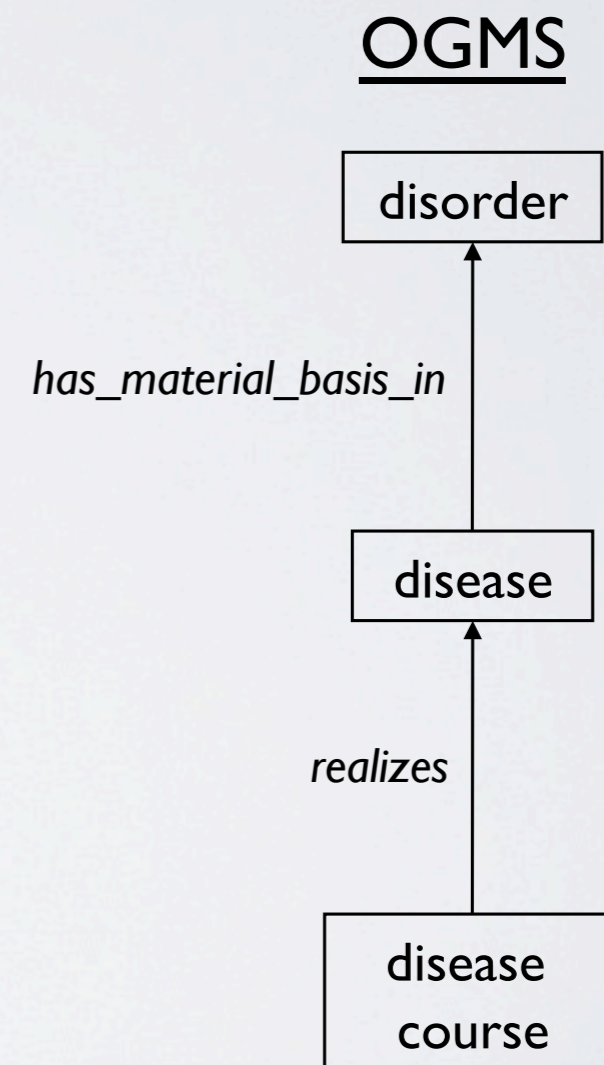
IDO-CORE REPRESENTATION OF INFECTIOUS DISEASE
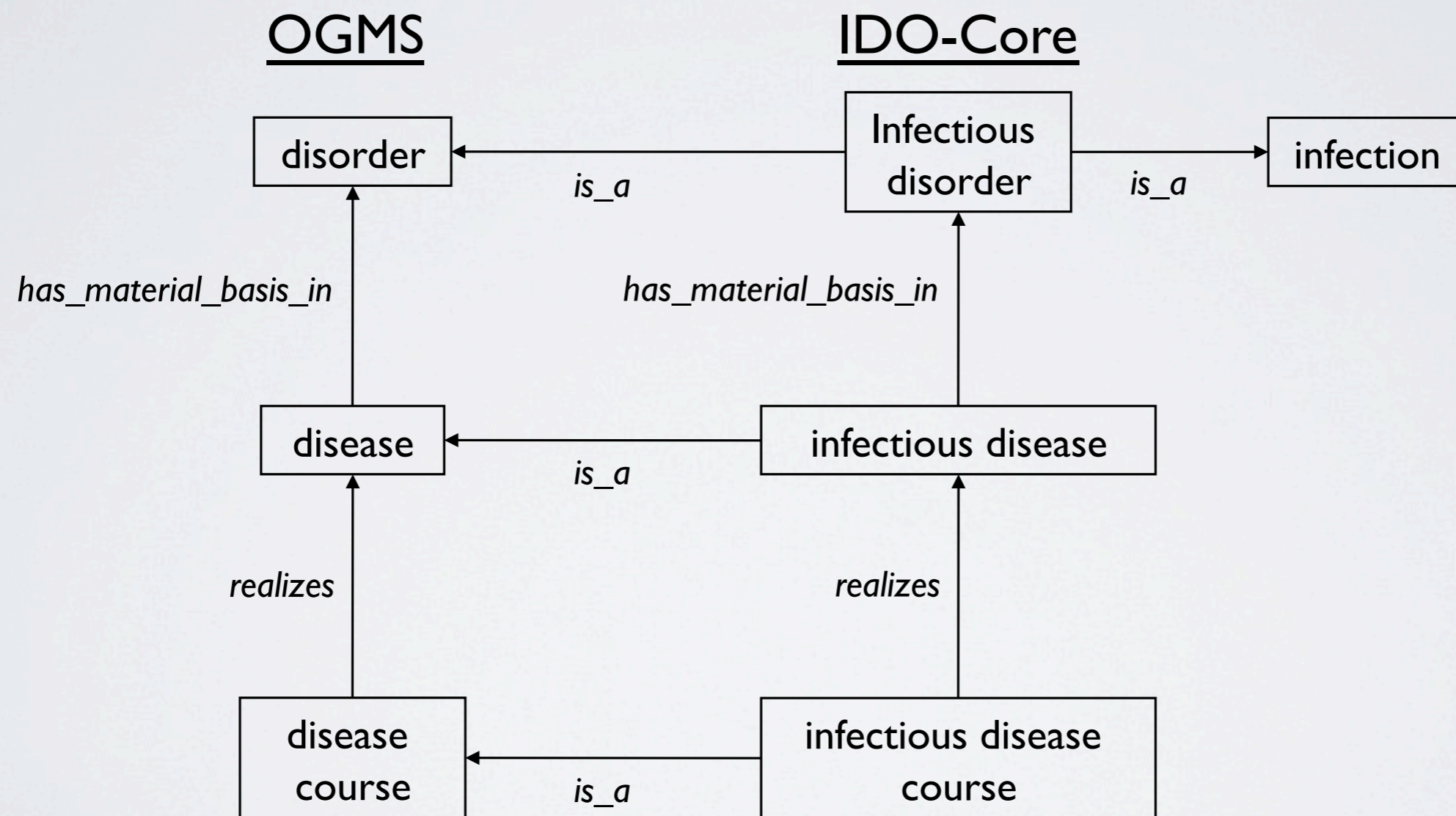
IDO-CORE CENTRAL TERMS
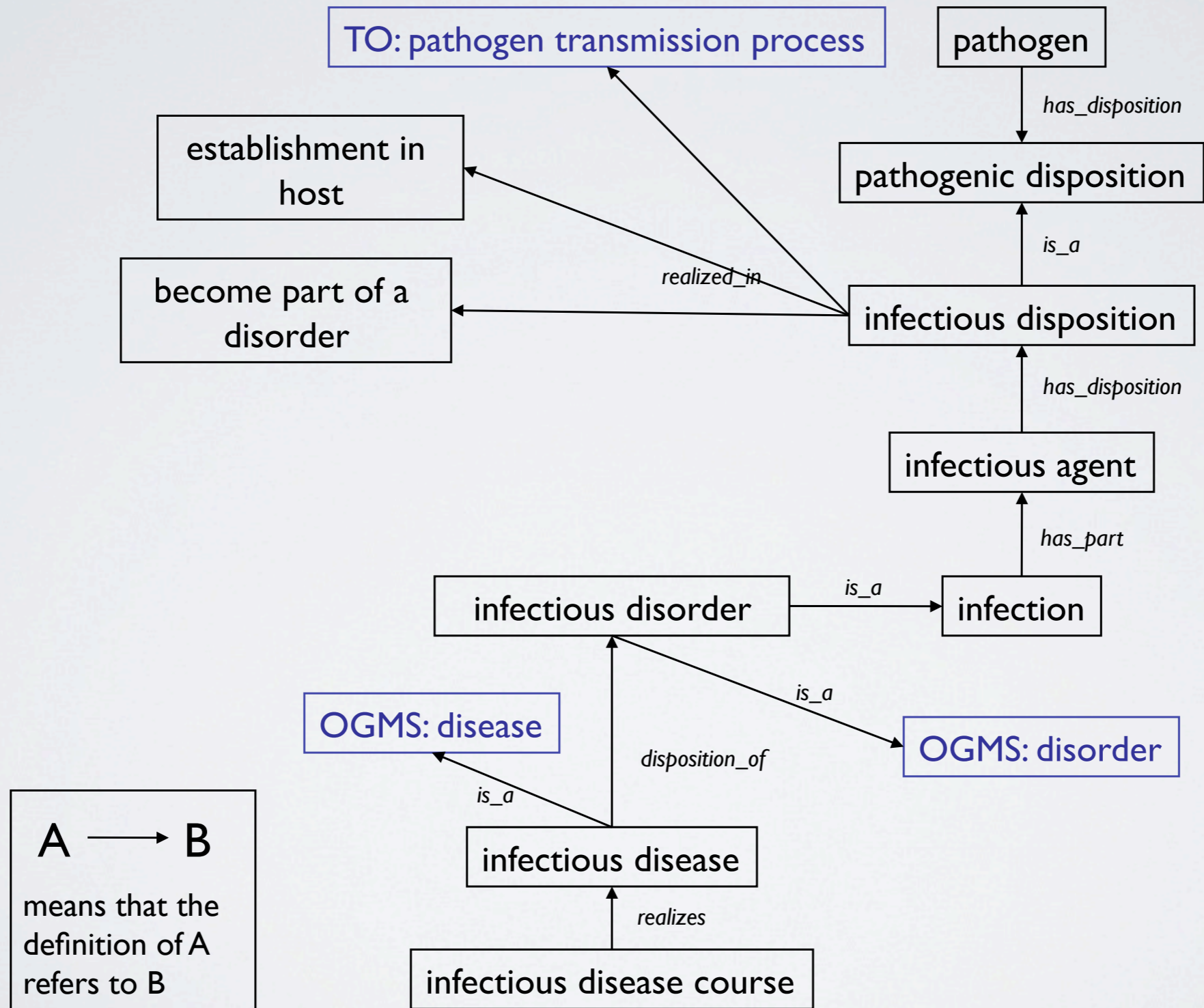
IDO-CORE CENTRAL DEFINITIONS

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 $=_{def}$ 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

- *Staphylococcus aureus* infection $=_{def}$ An infection that has as part organisms of type *Staphylococcus aureus*.

- *Staphylococcus aureus* bacteremia $=_{def}$ 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
        - has_disposition SOME 'infectious disposition') AND
        - located_in SOME blood]

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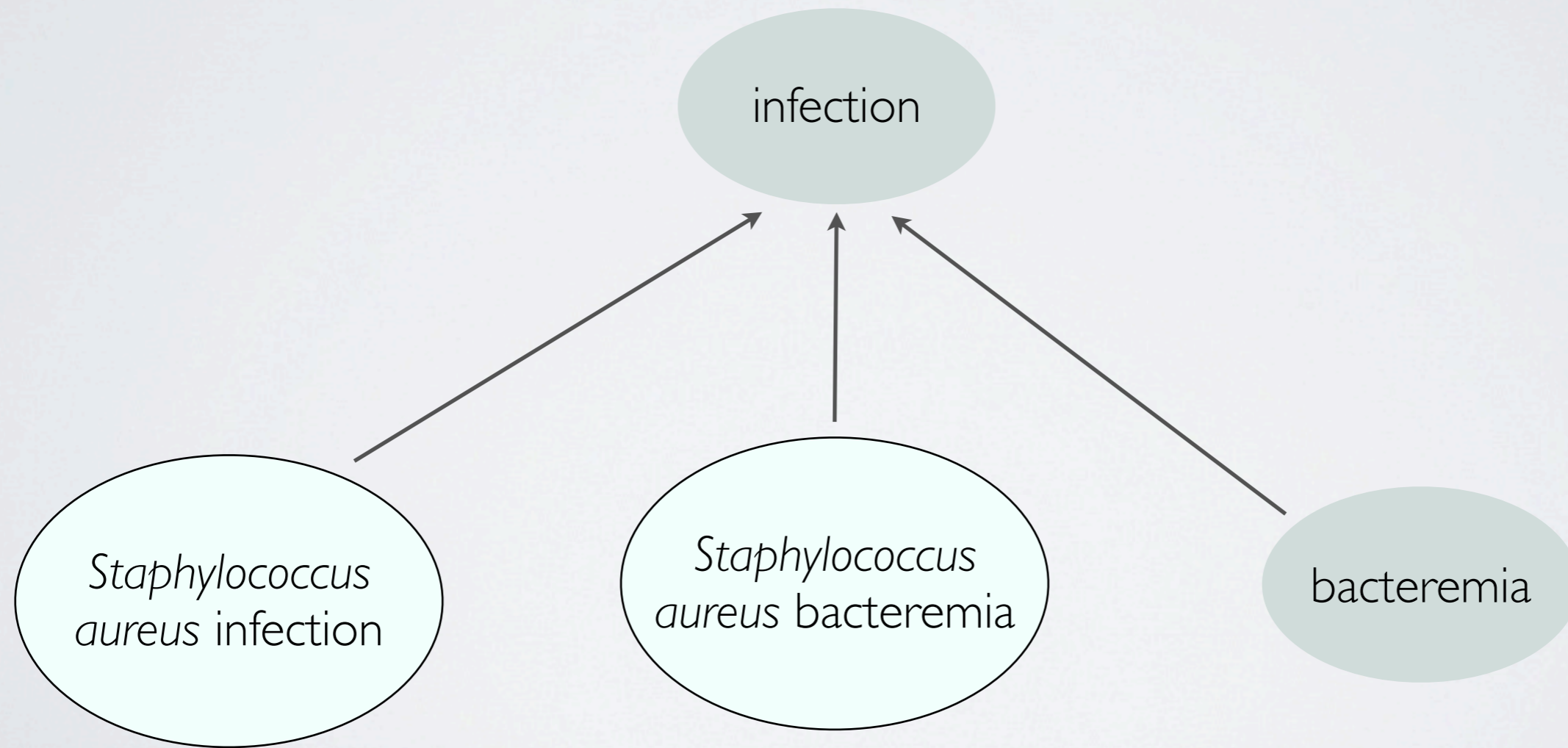   - located_in SOME blood]

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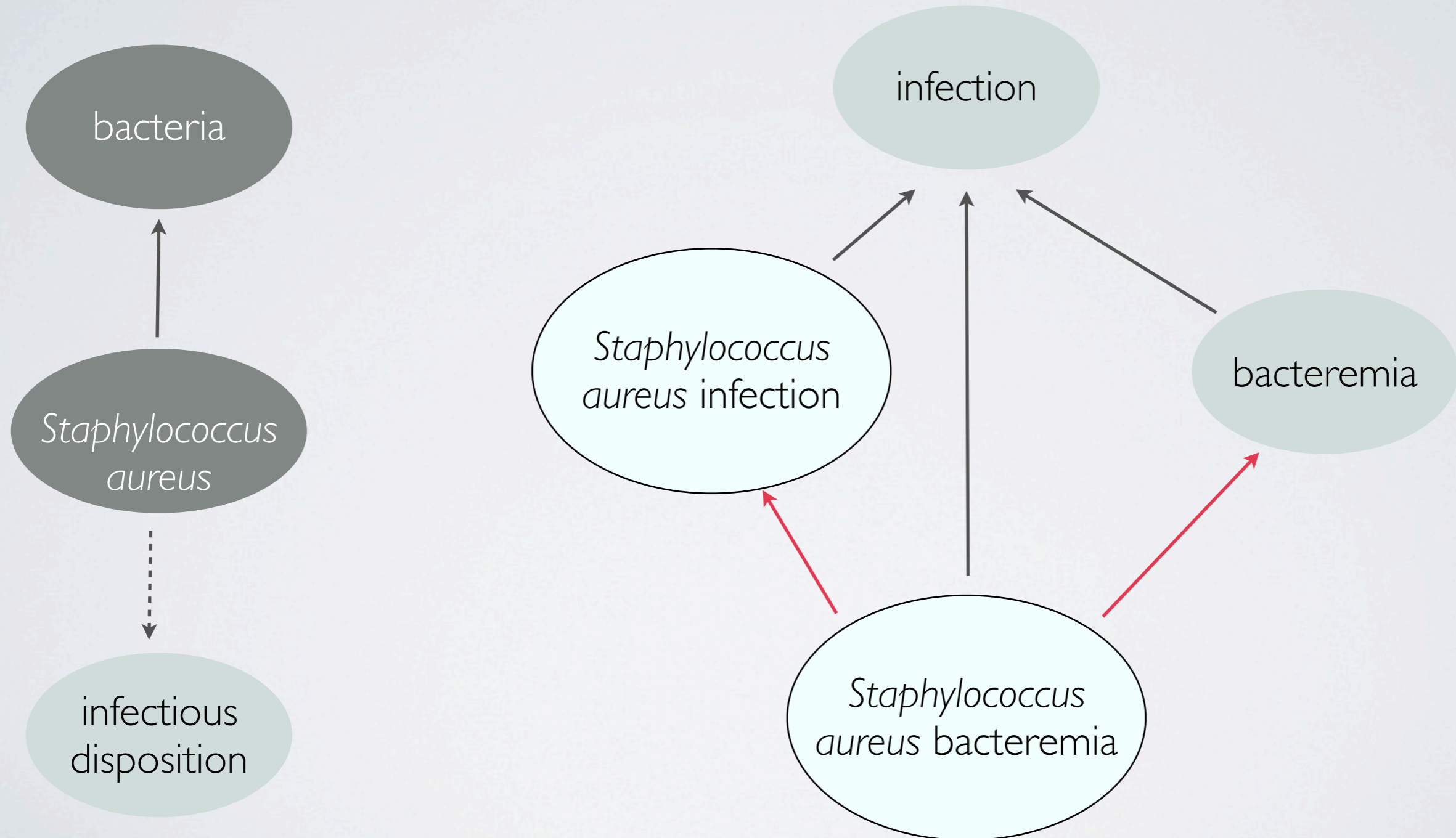   - located_in SOME blood]

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
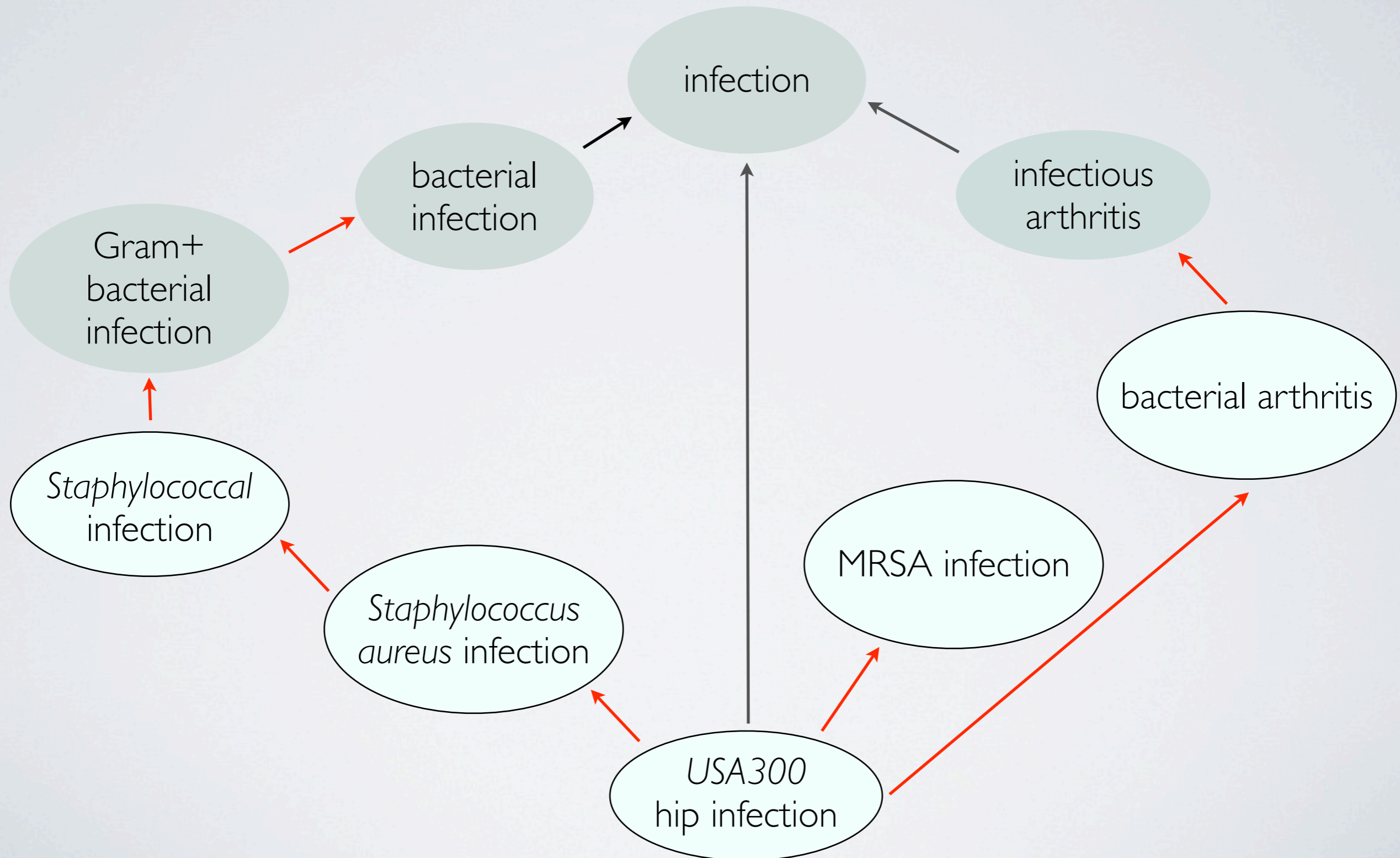 - has_part SOME [*Staphylococcus aureus* AND located_in SOME blood]

IDO-CORE AND EXTENSIONS: ASSERTED HIERARCHY

IDO-CORE AND EXTENSIONS: INFERRED HIERARCHY

IDO-CORE AND EXTENSIONS: INFERRED HIERARCHY

infection

bacterial infection

Gram+ bacterial infection

infectious arthritis

bacterial arthritis

Staphylococcal infection

Staphylococcus aureus infection

MRSA infection

USA300 hip infection

ACKNOWLEDGEMENTS

- Creators of IDO-Core

  - Lindsay Cowell
  - Alex Diehl
  - Albert Goldfain
  - Bjoern Peters
  - Alan Ruttenberg
  - Barry Smith

- IDO-Core Contributors

  - Melanie Courtot
  - Anna Maria Masci
  - Kitsos Louis
  - Richard Scheuermann
  - Burke Squires
  - Pantelis Topalis

BURROUGHS WELLCOME FUND

National Institute of Allergy and Infectious Diseases
National Institutes of Health