INFECTIOUS DISEASE
ONTOGOLOGY 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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Wednesday, December 8, 2010
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
<table>
<thead>
<tr>
<th>Clinical Entities</th>
<th>Biological Entities</th>
<th>Investigational Entities</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ontology of General Medical Science</td>
<td>NCBI Taxonomy</td>
<td>Ontology of Biomedical Investigation</td>
</tr>
<tr>
<td>Vital Signs Ontology</td>
<td>Common Anatomy Reference Ontology</td>
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<tr>
<td>Symptom Ontology</td>
<td>Cell Ontology</td>
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<tr>
<td>Disease Ontology</td>
<td>Gene Ontology Cellular Component Ontology</td>
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<td></td>
<td>Protein Ontology</td>
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<td>Chemical Entities of Biological Interest</td>
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<td>Phenotype and Trait Ontology</td>
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<td>Environment Ontology</td>
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<td></td>
<td>Gene Ontology Biological Process Ontology</td>
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</tbody>
</table>
IDO CORE-EXTENSION APPROACH

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IDO AND OBO FOUNDRY ONTOLOGIES

IDO-Core

IDO-Asp
IDO-Sa
IDO-Bac
IDO-TB
IDO-Sch
IDO-Par
IDO-Flu
IDO-Mal
IDO-Flav
IDO-Virus
IDO-Fun
IDO-Cry

CL

OGMS

GO BP

OBI

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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
“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 ....”
• 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 REPRESENTATION OF INFECTIOUS DISEASE

**OGMS**
- disorder
- has_material_basis_in disease
- disease
- realizes disease course

**IDO-Core**
- Infectious disorder
- is_a infectious disease
- realizes infectious disease course
- infection

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IDO-CORE CENTRAL TERMS

TO: pathogen transmission process

- establishment in host
- become part of a disorder

pathogen

- has_disposition

pathogenic disposition

- is_a

infectious disposition

- has_disposition

infectious agent

- has_part

infectious disorder

- is_a

infection

OGMS: disease

- disposition_of

OGMS: disorder

A → B

means that the definition of A refers to B

infectious disease

- realizes

infectious disease course

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<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
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<tbody>
<tr>
<td>pathogenic disposition</td>
<td>A disposition to initiate processes that result in a disorder.</td>
</tr>
<tr>
<td>pathogen</td>
<td>A material entity with a pathogenic disposition.</td>
</tr>
<tr>
<td>infectious disposition</td>
<td>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.</td>
</tr>
<tr>
<td>infectious agent</td>
<td>An organism with an infectious disposition.</td>
</tr>
</tbody>
</table>
IDO-CORE CENTRAL DEFINITIONS

- infection $=_{\text{def}}$ 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

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<td>infectious disorder</td>
<td>An infection that is clinically abnormal.</td>
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<td>infectious disease</td>
<td>A disease whose physical basis is an infectious disorder.</td>
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<tr>
<td>infectious disease course</td>
<td>A disease course that is a realization of an infectious disease.</td>
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</table>
• *Staphylococcus aureus* infection $=_{\text{def}} \text{An infection that has as part organisms of type } Staphylococcus\text{ aureus.}$

• *Staphylococcus aureus* bacteremia $=_{\text{def}} \text{An infection that has as part organisms of type } Staphylococcus\text{ aureus located in the blood.}$
IDO EXTENSION:
TERMS AND DEFINITIONS

• drug resistance =_{def} A protective resistance that mitigates the damaging effects of a drug.

• antibiotic resistance =_{def} A drug resistance that mitigates the damaging effects of an antibiotic.

• resistance to beta-lactam antibiotic =_{def} An antibiotic resistance that mitigates the damaging effects of a beta-lactam antibiotic.
IDO EXTENSION:
TERMS AND DEFINITIONS

• methicillin-resistant \textit{Staphylococcus aureus} =_{\text{def}} \text{An organism of type } \textit{Staphylococcus aureus} \text{ 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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• 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

- *Staphylococcus aureus* infection
- *Staphylococcus aureus* bacteremia
- bacteremia
IDO-CORE AND EXTENSIONS: INFERRED HIERARCHY

- Staphylococcus aureus
- Bacteremia
- Infection
- Infectious disposition

- Staphylococcus aureus infection
- Bacteremia
IDO-CORE AND EXTENSIONS: INFERRED HIERARCHY

- Gram+ bacterial infection
- Staphylococcal infection
- Staphylococcus aureus infection
- USA300 hip infection
- MRSA infection
- Infection
- Infectious arthritis
- Bacterial arthritis

Wednesday, December 8, 2010
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