



Infectious Disease Ontology

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Purpose of the Infectious Disease Ontology

- Serve as a standardized vocabulary
 - Facilitate communication
 - Enable precise data annotation, literature indexing, coding of patient records

Purpose of the Infectious Disease Ontology

- Serve as computable knowledge source
 - Computational analyses of high-throughput (and other) data
 - Text-mining of biomedical literature
 - Direct querying of the ontology
 - Automated reasoning - clinical decision support
 - Diagnosis
 - Prescribing
 - Biosurveillance
 - Vector management

Goals in Development

- Application Independence

Variety of Data Types in the Infectious Diseases Domain

- Biomedical Research (sequence data, cellular data, ...)
 - Pathogens, vectors, patients, model organisms
 - Microbiology, immunology, ...
- Vector Ecology Research
- Epidemiological Data for surveillance, prevention
- Clinical Care (case report data)
 - Clinical phenotypes, signs, symptoms
 - Treatments
 - Patient outcomes
- Clinical trial data for drugs, vaccines



Broad Scope

- **Scales:** molecules, cells, organisms, populations
- **Organisms:** host, pathogen, vector, model organisms, interactions between them
- **Domains:** biological, clinical care, public health
- **Diseases:** etiology, nature of pathogenesis, signs, symptoms, treatments

Goals in Development

- Application Independence
- Maximize use of Existing Ontology Resources



Broad Scope

- **Multiple Different Diseases and Pathogens**
 - Discoveries made in context of one disease can be applied to prevention and treatment of another
 - HIV - TB coinfection
 - Polymicrobial diseases

Goals in Development

- Application Independence
- Maximize use of Existing Ontology Resources
- Ensure interoperability across different diseases and pathogens

Maximize Use of Existing Ontology Resources

- Import or refer to terms contained in OBO Foundry reference ontologies
- Define new terms as cross-products from other Foundry ontologies
- Assert additional relations between terms

Benefits to Building from Foundry Ontologies

- Well-thought-out formalism
- Eliminating redundant effort
- Significant head-start
- Interoperability with other ontologies build within the Foundry or from Foundry ontologies
- Interoperability with information resources using Foundry ontologies for annotation
- Community acceptance

Independent Continuants in IDO

- **Anatomical location:** **FMA**: e.g. lung, kidney
- **Protein:** **PRO**: e.g. virulence factors such as Eap
- **Cell:** **CL**: e.g. macrophages
- **Pathological anatomical entity:** e.g. granuloma, sputum, pus

Occurrences in IDO

- Imported from **GO BP** when possible
e.g. [GO:0044406 : adhesion to host](#)
- **Population-level process:** e.g. emergence, epidemiological spread of disease
- **Pathological processes:** hematogenous seeding
- **Clinical process:** e.g. injection of PPD
- **Disease-specific process:**
 - Adhesion to host
 - S. aureus adhesion to host

Dependent Continuants in IDO

- **Quality: PATO:** e.g. attenuated, susceptible, co-infected, immunocompromised, drug resistant, zoonotic
- **Role:** e.g. host, pathogen, vector, carrier, reservoir, virulence factor, adhesin

Has_role

PRO

IDO

HBHA

has_role

biological adhesin

eap

has_role

biological adhesin

**Diphtheria
exotoxin**

has_role

virulence factor

**Protective
antigen**

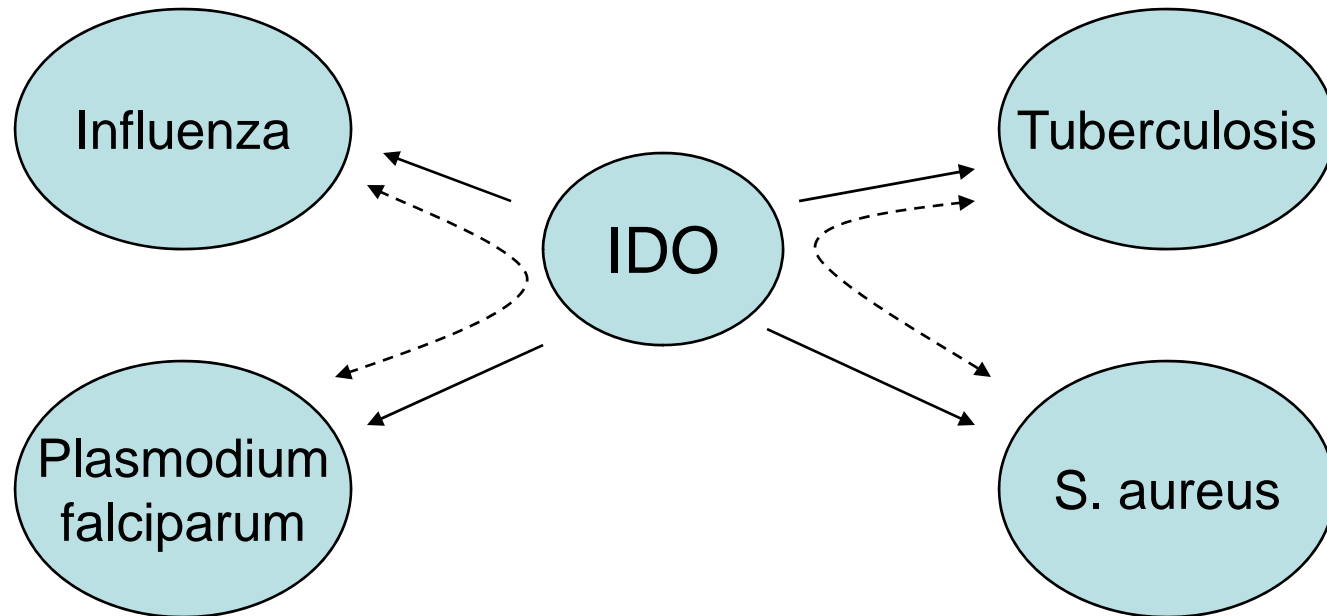
has_role

virulence factor



Cross-domain Interoperability

- Disease- and organism-specific ontologies
- Built as refinements to a template infectious disease ontology with terms relevant to a large number of infectious diseases





Benefits of the Template Ontology Approach

- Allows parallel development of multiple interoperable ontologies
 - Distributed development
 - rapid progress
 - curation by subdomain experts
 - Terminological consistency
 - term names and meanings
 - classification
- Prevent common mistakes

Disease-specific IDO test projects

- IMBB/VectorBase – *Vector borne diseases (A. gambiae, A. aegypti, I. scapularis, C. pipiens, P. humanus)*
 - Christos Louis
- Colorado State University – **Dengue Fever**
 - Saul Lozano-Fuentes
- Duke – **Tuberculosis, Staph. aureus, HIV**
 - Carol Dukes-Hamilton, Vance Fowler, Cliburn Chan
- Cleveland Clinic – **Infective Endocarditis**
 - Sivaram Arabandi
- MITRE, UT Southwestern, Maryland – **Influenza**
 - Joanne Luciano, Richard Scheuermann, Lynn Schriml
- University of Michigan – **Brucellosis**
 - Yongqun He

Disease-specific IDO test projects

- IMBB/VectorBase – Vector borne diseases (*A. gambiae*, *A. aegypti*, *I. scapularis*, *C. pipiens*, *P. humanus*)
 - Physiological processes of vectors that play a role in disease transmission
 - Decision Support
- Colorado State University – Dengue Fever
 - Dengue Decision Support System
- Duke – Tuberculosis, Staph. aureus, HIV
 - TB Trials Network: address the lack of interoperability between paper-based clinical trials data collection systems, health department systems and medical records systems by creating a system for electronic management of TB data
 - Candidate Disease Gene Prediction
 - CFAR, CHAVI - high-throughput data analysis; SIV - HIV interoperability

Disease-specific IDO test projects

- Cleveland Clinic – Infective Endocarditis
 - SemanticDB technology
- MITRE, UT Southwestern, Maryland – Influenza
 - Centers for Excellence in Influenza Research and Surveillance
 - Elucidate causes of influenza virulence
- University of Michigan – Brucellosis
 - Text-mining

Roles in IDO

- ▣ ← **i** role
 - ▣ ← **i** antibacterial role
 - ← **i** antimycobacterial role
 - ← **i** antifungal role
 - ← **i** antiparasitic role
 - ← **i** antiviral role
 - ▣ ← **i** biological factor
 - ← **i** adhesion factor
 - ← **i** colonization factor
 - ▣ ← **i** virulence factor
 - ← **i** invasin
 - ▣ ← **i** toxin of infectious agent
 - ← **i** endotoxin
 - ← **i** exotoxin
 - ← **i** portal of entry of infectious agent
 - ← **i** portal of exit of infectious agent
 - ▣ ← **i** reservoir of infectious agent
 - ← **i** end reservoir of infectious agent
- ← **i** source of infectious agent
- ▣ ← **i** symbiont
 - ← **i** commensal
- ▣ ← **i** host of parasite
 - ▣ ← **i** host of infectious agent
 - ▣ ← **i** carrier of infectious agent
 - ← **i** chronic carrier of infectious agent
 - ← **i** mutualist
 - ▣ ← **i** parasite
 - ▣ ← **i** infectious agent
 - ▣ ← **i** pathogen
 - ← **i** primary pathogen
 - ▣ ← **i** transporter of infectious agent
 - ← **i** vector of infectious agent
 - ▣ ← **i** vehicle of infectious agent
 - ← **i** fomite

Qualities in IDO

- ☐ ← ⓘ quality
 - ☐ ← ⓘ quality of continuant
 - ☐ ← ⓘ monadic quality of continuant
 - ← ⓘ immunodeficiency
 - ☐ ← ⓘ monadic quality of bearer of host role
 - ← ⓘ asymptomatic quality of host
 - ← ⓘ symptomatic quality of host
 - ☐ ← ⓘ monadic quality of bearer of infectious agent role
 - ← ⓘ active quality of infectious agent
 - ← ⓘ dormancy of infectious agent
 - ← ⓘ host range
 - ← ⓘ latency of infectious agent
 - ← ⓘ opportunistic quality of infectious agent
 - ← ⓘ tropism
 - ← ⓘ zoonosis
 - ☐ ← ⓘ monadic quality of infection
 - ← ⓘ acute quality of an infection
 - ← ⓘ chronic quality of an infection
 - ← ⓘ extracellular quality of an infection
 - ← ⓘ focal quality of an infection
 - ← ⓘ intracellular quality of an infection
 - ← ⓘ local quality of an infection
 - ← ⓘ polymicrobial quality of an infection
 - ← ⓘ primary quality of an infection
 - ← ⓘ secondary quality of an infection
 - ← ⓘ subacute quality of an infection
 - ← ⓘ subclinical quality of an infection
 - ← ⓘ systemic quality of an infection

Qualities in IDO

- ⊖ ← ⓘ relational quality of continuant
 - ⊕ ← ⓘ relational quality between at-risk population and bearers of the infectious agent role
 - ⊕ ← ⓘ relational quality between bearers of symbiont roles
 - ⊕ ← ⓘ relational quality between bearers of the infectious agent and drug roles
 - ⊕ ← ⓘ relational quality of an infectious disease and a host
 - ⊖ ← ⓘ relational quality of an infectious disease and a host population
 - ⊖ ← ⓘ elevated infectious disease incidence
 - ⊖ ← ⓘ infectious disease epidemic
 - ← ⓘ infectious disease pandemic
 - ← ⓘ endemic level of infectious disease
 - ⊖ ← ⓘ endemic quality of infectious disease
 - ← ⓘ hyper-endemic quality of infectious disease
 - ⊖ ← ⓘ epidemic quality of infectious disease
 - ← ⓘ pandemic quality of infectious disease
 - ⊖ ← ⓘ incidence of infectious disease
 - ← ⓘ incidence proportion of infectious disease
 - ← ⓘ incidence rate of infectious disease
 - ← ⓘ mortality rate of infectious disease
 - ⊖ ← ⓘ prevalence of infectious disease
 - ← ⓘ seroprevalence of infectious disease
 - ← ⓘ sporadicity of infectious disease

Processes in IDO

- ☐ ← ⓘ biological process
 - ← ⓘ colonization of host
 - ☐ ← ⓘ damaging host cells and tissues in subepithelial space
 - ← ⓘ induction of damaging host immune response
 - ← ⓘ symbiont-induced apoptosis
 - ← ⓘ symbiont-induced necrosis
 - ← ⓘ virus-induced cytopathogenesis
 - ☐ ← ⓘ epidemiological spread of disease
 - ← ⓘ horizontal transfer
 - ☐ ← ⓘ infection process
 - ← ⓘ infectious disease course
 - ☐ ← ⓘ initiation of infection process
 - ← ⓘ pathogenesis of infectious disease
 - ☐ ← ⓘ penetration of epithelial barrier
 - ☐ ← ⓘ penetration of host cell
 - ☐ ← ⓘ symbiont adherence to host cell
 - ☐ ← ⓘ symbiont evasion of host immune response
 - ☐ ← ⓘ antigenic variation
 - ← ⓘ genetic diversification
 - ← ⓘ phase variation
 - ← ⓘ resist phagocytosis in subepithelial space
 - ☐ ← ⓘ serum resistance
 - ☐ ← ⓘ symbiont spread through host
 - ☐ ← ⓘ transmission of infectious agent
- ☐ ← ⓘ immunization
 - ☐ ← ⓘ active immunization
 - ☐ ← ⓘ inoculation of host
 - ← ⓘ variolation
 - ← ⓘ vaccination
 - ← ⓘ passive immunization

Join the IDO Consortium

- <http://www.infectiousdiseaseontology.org>
- ido@duke.edu
- <http://lists.duke.edu/sympa>

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BURROUGHS
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FUND 

Ontology of S.a. - Human Interaction

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