ImmPort Disease Ontology

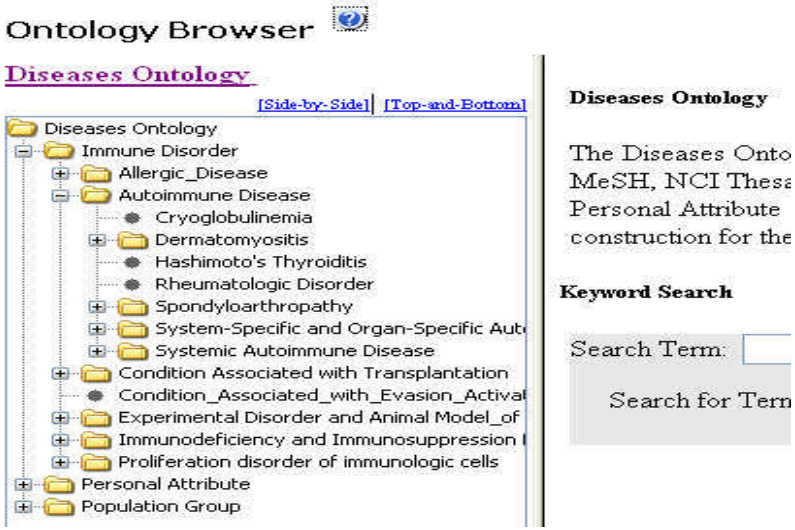
Diseases Ontology

The Diseases Ontology contains three taxonomies of classes (concepts or general things): Immune Disorder, Population, and Personal Attribute. The Immune Disorder tree is based on the National Library of Medicine (NLM) Medical Subject Headings; National Cancer Institute Thesaurus; International Classification of Diseases, Ninth Revision, Clinical Modification (ICD-9-CM); ICD-10; and other open source public databases. The subtrees of Race and Ethnicity under Personal Attribute are from the Centers for Disease Control and Prevention Race and Ethnicity Code Set 2000. The Diseases Ontology is part of the ImmPort Ontology, which will be available in a future ImmPort system release.

Java Runtime Environment Program Needed

To view the ontology, you need a Java Runtime Environment program on your local computer. This program is freely available for download. If your local computer does not have this program installed, the ImmPort system will automatically ask if you would like to download the program. Click "Yes." The download will proceed automatically to a wizard installation, after which the ontology tree view will appear (see Figure 1). The tree view display will enable you to browse the ontology.

Figure 1. Ontology Browser Tree View



ImmPort Disease Ontology

Disease Ontology, continued

If the download/installation process does not work (i.e., the ontology tree view display does not appear), manually download the Java Runtime Environment program at Java's Web site: http://www.java.com/en/download/manual.jsp. Installation instructions are provided on this Web site.

Tree Hierarchy

[**Side-by-Side**]
The browser displays class information side-by-side. When a class in the tree hierarchy is clicked, its Lineage(s), Annotations, Relationships (Object Properties), and Datatype Properties are shown to the right of the tree hierarchy.
[**Top-and-Bottom**]
The browser also displays class information top-to-bottom. When a class in the tree hierarchy is clicked, its Lineage(s), Annotations, Relationships (Object Properties), and Datatype Properties are shown below the tree hierarchy.
Click the plus symbol to expand the current view for more information. You can also tab to the image by pressing the right arrow key on your keyboard.
Click the minus symbol to collapse the current view to hide information. You can also tab to the image by pressing the left arrow key on your keyboard.

Class Information

Keyword Search

Terms in the ontology can be searched by entering a word or part of a word that appears in the name or attribute of a class or its property using Like, Exact Match, Starts With, or Ends With. Keyword searches are not case sensitive.

Like: Search terms by entering a string composed of a word, words, or part of a word that appears anywhere in a term or starts or ends this term. This search automatically uses wild cards (special symbols that stand for one or more characters) at the start and end of the string used. To ensure search results, make sure there is no hidden space at the start or end of your term.

Exact Match: Search terms by entering the exact spelling of concept/property name or value of concept/property attribute. To ensure search results, make sure there is no hidden space at the start or end of your term.
Starts With: Search terms by entering a string that matches the start of the term of interest. This search automatically uses a wild card at the end of the string used. To ensure search results, make sure there is no hidden space at the start or end of your string.

Ends With: Search terms by entering a string that matches the end of the term of interest. This search automatically uses a wild card at the start of the string used. To ensure search results, make sure there is no hidden space at the start or end of your string.

Disease Ontology, continued

Lineage(s)

A class may be related to its superclass through a true is_a relationship (e.g., Polymyositis is a Dermatomyositis, which is an Autoimmune_Disease, which is an Immune_Disorder). Based on description logic, a class inherits all the Relationships (Object Properties) and Datatype Properties (if any) assigned to its superclass. In a few occasions, a class may have more than one superclass and belongs to more than one lineage.

Annotations

Specific information may be available about a class, including Preferred_Name, DEFINITION, Synonym, etc. The following provides more detail on annotation information presented:

DEFINITION: A human-readable definition is provided for each class in the ontology and the displayed format is Owner|Definition (Source).

FULL_SYN: Synonym of a class in the ontology with type (PT: preferred term, SY: synonym) and source of this synonym indicated. Synonym|Type of Synonym|Source of Synonym is the format displayed.

ICD-10_Code: Code of International Classification of Diseases and Related Health Problems, 10th revision. ICD-10 is a detailed description of known diseases and injuries. It is published by the World Health Organization (WHO) and is used worldwide for mortality statistics.

ICD-9_Code: Code of International Classification of Diseases and Related Health Problems, 9th revision.

Class Name: Class_Name is used as a unique identifier by Protege-OWL, the environment where this ontology is being developed and therefore has to be OWL-compliant. Class_Name is composed of a string only made of digits, letters, underscores, and hyphens. A Class_Name cannot start with a digit.

Preferred_Name: A human-friendly and commonly used name in plain English.

Synonym: Synonym of a class in plain English, including those in plural form or British spelling. In contrast to FULL_SYN, no type nor source of synonym is indicated. Synonym is human-friendly and not OWL-compliant. A class may have multiple synonyms.

UMLS_CUI: Concept Unique Identifier in Unified Medical Language System that is produced and distributed by NLM. There are three UMLS Knowledge Sources: the Metathesaurus, the Semantic Network, and SPECIALIST Lexicon. The purpose of UMLS is to facilitate the development of computer systems that behave as if they "understand" the meaning of the language of biomedicine and health.

Relationships (Object Properties)

Relationships are properties by which one class is related to another class in the ontology. For example, "Population_Group" (a class) has_susceptibility_to_disease (a relationship) "Immune_Disorder" (a class). This box may be empty if no relationship has been used for modeling.

Datatype Properties

Datatype Properties are properties by which a class is related to a datatype. This box may be empty if no datatype property has been used for modeling.