The Landscape of Disease and Phenotype Ontologies

Compiled by Nicole Vasilevsky. Feel free to make pull requests to suggest edits here: https://github.com/OBOAcademy/obook/blob/master/archive_original_cpath_course/09-DiseasesAndPhenotypes/ontologylandscape.md.

Disease Ontologies & Terminologies

Disease Summary Table

Name	Disease Area
Artificial Intelligence Rheumatology Consultant System Ontology (AI-RHEUM)	Rheumatic diseases
Autism DSM-ADI-R Ontology (ADAR)	Autism
Autism Spectrum Disorder Phenotype Ontology (ASDPTO)	Autism
Brucellosis Ontology (IDOBRU)	brucellosis
Cardiovascular Disease Ontology (CVDO)	Cardiovascular
Chronic Kidney Disease Ontology (CKDO)	Chronic kidney disease
Chronic Obstructive Pulmonary Disease Ontology (COPDO)	Chronic obstructive pulmonary disease (COPD)
Coronavirus Infectious Disease Ontology (CIDO)	Coronavirus infectious diseases
Diagnostic and Statistical Manual of Mental Disorders (DSM)	Mental disorders
Dispedia Core Ontology (DCO)	Rare diseases
Experimental Factor Ontology (EFO)	Broad disease coverage
Fibrotic Interstitial Lung Disease Ontology (FILDO)	Fibrotic interstitial lung disease
Genetic and Rare Diseases Information Center (GARD)	Rare diseases
Holistic Ontology of Rare Diseases (HORD)	Rare disease
Human Dermatological Disease Ontology (DERMO)	Dermatology (skin)
Human Disease Ontology (DO)	Human disease
Infectious Disease Ontology (IDO)	Infectious disease
International Classification of Functioning, Disability and Health (ICF)	Cross-discipline, focuses disabilities
International Statistical Classification of Diseases and Related Health Problems (ICD-11)	Broad coverage
International Classification of Diseases for Oncology (ICD-O)	Cancer
Logical Observation Identifier Names and Codes (LOINC)	Broad coverage
Medical Subject Headings (MeSH)	Broad coverage
MedGen	Human medical genetics
Medical Dictionary for Regulatory Activities (MedDRA)	Broad coverage
Mental Disease Ontology (MDO)	Mental functioning
Mondo Disease Ontology (Mondo)	Broad coverage, Cross species
National Cancer Institute Thesaurus (NCIT)	Humam cancer and neoplasms

Name	Disease Area
Neurological Disease Ontology (ND)	Neurology
Online Mendelian Inheritance in Man (OMIM)	Mendelian, genetic diseases.
Ontology of Cardiovascular Drug Adverse Events (OCVDAE)	Cardiovascular
Ontology for General Medical Science (OGMS)	Broad coverage
Ontology for Genetic Susceptibility Factor (OGSF)	Genetic disease
Ontology of Glucose Metabolism Disorder (OGMD)	Metabolic disorders
Ontology of Language Disorder in Autism (LDA)	Austism
The Oral Health and Disease Ontology (OHD)	Oral health and disease
Orphanet (ORDO)	Rare diseases
Parkinson Disease Ontology (PDO)	Parkinson disease
Pathogenic Disease Ontology (PDO)	Pathogenic diseases
PolyCystic Ovary Syndrome Knowledgebase (PCOSKB)	Polycystic ovary syndrome
Rat Disease Ontology (RDO)	Broad coverage
Removable Partial Denture Ontology (RPDO)	Oral health
Resource of Asian Primary Immunodeficiency Diseases (RPO)	Immunodeficiencies
Sickle Cell Disease Ontology (SCDO)	Sickle Cell Disease
SNOMED Clinical Terminology (SNOMED CT)	Broad disease representation for human diseases.
Symptom Ontology	Human diseases
Unified Medical Language System	Broad coverage

Artificial Intelligence Rheumatology Consultant System ontology (AI-RHEUM)

Description: Contains findings, such as clinical signs, symptoms, laboratory test results, radiologic observations, tissue biopsy results, and intermediate diagnosis hypotheses, for the diagnosis of rheumatic diseases.
Disease area: Rheumatic diseases
Use Cases: Used by clinicians and informatics researchers.
Website: https://bioportal.bioontology.org/ontologies/AI-RHEUM
Open: Yes

Autism DSM-ADI-R Ontology (ADAR)

Description: An ontology of autism spectrum disorder (ASD) and related neurodevelopmental disorders.
Disease area: Autism
Use Cases: It extends an existing autism ontology to allow automatic inference of ASD phenotypes and Diagnostic and Statistical Manual of Mental Disorders (DSM) criteria based on subjects' Autism Diagnostic Interview–Revised (ADI-R) assessment data.
Website: https://bioportal.bioontology.org/ontologies/ADAR
Open: Yes

Autism Spectrum Disorder Phenotype Ontology (ASDPTO)

Description: Encapsulates the ASD behavioral phenotype, informed by the standard ASD assessment instruments and the currently known characteristics of this disorder.
Disease area: Autism
Use Cases: Intended for use in research settings where extensive phenotypic data have been collected, allowing a concept-based approach to identifying behavioral features of importance and for correlating these with genotypic data.
Website: https://bioportal.bioontology.org/ontologies/ASDPTO
Open: Yes

Brucellosis Ontology (IDOBRU)

Description: Describes the most common zoonotic disease, brucellosis, which is caused by Brucella, a type of facultative intracellular bacteria.

Disease area: brucellosis bacteria
Use Cases: An extension ontology of the core Infectious Disease Ontology (IDO-core). This project appears to be inactive.
Website: https://github.com/biomedontology/idobru **Open:** Yes

Cardiovascular Disease Ontology (CVDO)

Description: An ontology to describe entities related to cardiovascular diseases.
Disease area: Cardiovascular
Use Cases: Describes entities related to cardiovascular diseases including the diseases themselves, the underlying disorders, and the related pathological processes. Imports upper level terms from OGMS and imports some terms from Disease Ontology (DO).
GitHub repo: https://github.com/OpenLHS/CVDO/
Website: https://github.com/OpenLHS/CVDO
OBO Foundry webpage: http://obofoundry.org/ontology/cvdo.html
Open: Yes

Chronic Kidney Disease Ontology (CKDO)

Description: An ontology of chronic kidney disease in primary care.
Disease area: Chronic kidney disease
Use Cases: CKDDO was developed to assist routine data studies and case identification of CKD in primary care.
Website: http://purl.bioontology.org/ontology/CKDO
Open: Yes

Chronic Obstructive Pulmonary Disease Ontology (COPDO)

Description: Models concepts associated with chronic obstructive pulmonary disease in routine clinical databases.
Disease area: Chronic obstructive pulmonary disease (COPD)
Use Cases: Clinical use.
Website: https://bioportal.bioontology.org/ontologies/COPDO
Open: Yes

Coronavirus Infectious Disease Ontology (CIDO)

Description: Aims to ontologically represent and standardize various aspects of coronavirus infectious diseases, including their etiology, transmission, epidemiology, pathogenesis, diagnosis, prevention, and treatment.
Disease area: Coronavirus infectious diseases, including COVID-19, SARS, MERS; covers etiology, transmission, epidemiology, pathogenesis, diagnosis, prevention, and treatment.
Use Cases: Used for disease annotations related to coronavirus infections.
GitHub repo: https://github.com/cido-ontology/cido
OBO Foundry webpage: http://obofoundry.org/ontology/cido.html
Open: Yes

Diagnostic and Statistical Manual of Mental Disorders (DSM)

Description: Authoritative source to define and classify mental disorders to improve diagnoses, treatment, and research.
Disease area: Mental disorders
Use Cases: Used in clinical healthcare and research by pyschiatrists and psychologists.
Website: https://www.psychiatry.org/psychiatrists/practice/dsm
Open: No, must be purchased

Dispedia Core Ontology (DCO)

Description: A schema for information brokering and knowledge management in the complex field of rare diseases. DCO describes patients affected by rare diseases and records expertise about diseases in machine-readable form.
Disease area: Rare disease
Use Cases: DCO was initially created with amyotrophic lateral sclerosis as a use case.
Website: http://purl.bioontology.org/ontology/DCO
Open: Yes

Experimental Factor Ontology (EFO)

Description: Provides a systematic description of many experimental variables available in EBI databases, and for projects such as the GWAS catalog.
Disease area: Broad disease coverage, integrates the Mondo disease ontology.
Use Cases: Application ontology build for European Bioinformatics (EBI) tools and databases and Open Targets Genetics Portal.
Website: https://www.ebi.ac.uk/efo/
Open: Yes

Fibrotic Interstitial Lung Disease Ontology (FILDO)

Description: An in-progress, four-tiered ontology proposed to standardize the diagnostic classification of patients with fibrotic interstitial lung disease.
Disease area: Fibrotic interstitial lung disease
Use Cases: Goal is to standardize the diagnostic classification of patients with fibrotic ILD. A paper was published in 2017 and an ontology is not publicly available.
Publication: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5803648/
Open: No

Genetic and Rare Diseases Information Center (GARD)

Description: NIH resource that provides the public with access to current, reliable, and easy-to-understand information about rare or genetic diseases in English or Spanish.
Disease area: Rare diseases

Use Cases: Patient portal. Integrates defintions and synonyms from Orphanet, maps to HPO phenotypes, and is integrated by Mondo.
Website: https://rarediseases.info.nih.gov/
Open: Yes

Holistic Ontology of Rare Diseases (HORD)

Description: Describes the biopsychosocial state (i.e., disease, psychological, social, and environmental state) of persons with rare diseases in a holistic way.
Disease area: Rare disease
Use Cases: Rehabilita, Disruptive Technologies for the Rehabilitation of the Future, a project that aims to enhance rehabilitation transforming it to a more personalized, ubiquitous and evidence-based rehabilitation.
Website: http://purl.bioontology.org/ontology/HORD
Open: Yes

Human Dermatological Disease Ontology (DERMO)

Description: The most comprehensive dermatological disease ontology available, with over 3,500 classes available. There are 20 upper-level disease entities, with features such as anatomical location, heritability, and affected cell or tissue type.
Disease area: Dermatology (skin)
Use Cases: DermO can be used to extract data from patient electronic health records using text mining, or to translate existing variable-granularity coding such as ICD-10 to allow capture and standardization of patient/disease annotations.
Website: https://bioportal.bioontology.org/ontologies/DERMO
Open: Yes

Human Disease Ontology (DO)

Description: An ontology for describing the classification of human diseases organized by etiology.
Disease area: Human disease terms, phenotype characteristics and related medical vocabulary disease concepts.
Use Cases: Used by Model Organism Databases (MOD), such as Mouse Genome Informatics disease model for diseae annotations, and Alliance for Genome Resources for disease annotations. In 2018, DO tracked over 300 DO project citations suggesting wide adoption and usage for disease annotations.
GitHub repo: https://github.com/DiseaseOntology/HumanDiseaseOntology/
Website: http://www.disease-ontology.org/
OBO Foundry webpage: http://obofoundry.org/ontology/doid.html
Open: Yes

Infectious Disease Ontology (IDO)

Description: A set of interoperable ontologies that will together provide coverage of the infectious disease domain. IDO core is the upper-level ontology that hosts terms of general relevance across the domain, while extension ontologies host terms to specific to a particular part of the domain.
Disease area: Infectious disease features, such as acute, primary, secondary infection, and chronic, hospital acquired and local infection.
Use Cases: Does not seem active, has not been released since 2017.
GitHub repo: https://github.com/infectious-disease-ontology/infectious-disease-ontology/
Website: http://www.bioontology.org/wiki/index.php/Infectious_Disease_Ontology
OBO Foundry webpage: http://obofoundry.org/ontology/ido.html
Open: Yes

International Classification of Functioning, Disability and Health (ICF)

Description: Represents diseases and provides a conceptual basis for the definition and measurement of health and disability as organized by patient-oriented outcomes of function and disability. ICF considers environmental factors as well as the relevance of associated health conditions in recognizing major models of disability.
Disease area: Cross-discipline, focuses on health and disability
Use Cases: ICF is the World Health Organization (WHO) framework for measuring health and disability at both individual and population levels. ICF was officially endorsed by the WHO as the international standard to describe and measure health and disability.
Website: https://www.who.int/standards/classifications/international-classification-of-functioning-disability-and-health
Open: Yes

International Statistical Classification of Diseases and Related Health Problems (ICD-11)

Description: A medical classification list by the World Health Organization (WHO) that contains codes for diseases, signs and symptoms, abnormal findings, complaints, social circumstances, and external causes of injury or diseases.
Disease area: Broad coverage of human disease features, such as disease of anatomical systems, infectious diseases, injuries, external causes of morbidity and mortality.
Use Cases: The main purpose of ICD-11 is for clinical care, billing and coding for insurance companies.
Website: https://www.who.int/standards/classifications/classification-of-diseases
Open: Yes

International Classification of Diseases for Oncology (ICD-O)

Description: A domain-specific extension of the International Statistical Classification of Diseases and Related Health Problems for tumor diseases.
Disease area: A multi-axial classification of the site, morphology, behaviour, and grading of neoplasms.
Use Cases: Used principally in tumour or cancer registries for coding the site (topography) and the histology (morphology) of neoplasms, usually obtained from a pathology report.
Website: https://www.who.int/standards/classifications/other-classifications/international-classification-of-diseases-for-oncology
Open: Yes

Logical Observation Identifier Names and Codes (LOINC)

Description: Identifies medical laboratory observations.
Disease area: Broad coverage
Use Cases: The Regenstrief Institute first developed LOINC in 1994 in response to the demand for an electronic database for clinical care and management. LOINC is

publicly available at no cost and is endorsed by the American Clinical Laboratory Association and the College of American Pathologists. Since its inception, LOINC has expanded to include not just medical laboratory code names but also nursing diagnoses, nursing interventions, outcome classifications, and patient care data sets.
Website: https://loinc.org/
Open: Yes, registration is required.

Medical Subject Headings (MeSH)

Description: Medical Subject Headings (MeSH) thesaurus is a controlled and hierarchically-organized vocabulary produced by the National Library of Medicine.
Disease area: Broad coverage
Use Cases: It is used for indexing, cataloging, and searching of biomedical and health-related information. Integrated into Mondo.
Website: https://meshb.nlm.nih.gov/search
Open: Yes

MedGen

Description: Organizes information related to human medical genetics, such as attributes of conditions and phenotypes of genetic contributions.
Disease area: Human medical genetics
Use Cases: MedGen is NCBI's portal to information about conditions and phenotypes related to Medical Genetics. Terms from the NIH Genetic Testing Registry (GTR), UMLS, HPO, Orphanet, ClinVar and other sources are aggregated into concepts, each of which is assigned a unique identifier and a preferred name and symbol. The core content of the record may include names, identifiers used by other databases, mode of inheritance, clinical features, and map location of the loci affecting the disorder. The concept identifier (CUI) is used to aggregate information about that concept, similar to the way NCBI Gene serves as a gateway to gene-related information.
Website: https://www.ncbi.nlm.nih.gov/medgen/
Open: Yes

Medical Dictionary for Regulatory Activities (MedDRA)

Description: Provides a standardized international medical terminology to be used for regulatory communication and evaluation of data about medicinal products for human use.
Disease area: Broad coverage
Use Cases: Mainly targeted towards industry and regulatory users.
Website: https://www.meddra.org/
Open: Yes

Mental Disease Ontology (MDO)

Description: An ontology to describe and classify mental diseases such as schizophrenia, annotated with DSM-IV and ICD codes where applicable.
Disease area: Mental functioning, including mental processes such as cognition and traits such as intelligence.
Use Cases: The ontology has been partially aligned with the related projects Cognitive Atlas, knowledge base on cognitive science and the Cognitive Paradigm Ontology, which is used in the Brainmap, a database of neuroimaging experiments.
GitHub repo: https://github.com/jannahastings/mental-functioning-ontology
OBO Foundry webpage: http://obofoundry.org/ontology/mfomd.html
Open: yes

Mondo Disease Ontology (Mondo)

Description: An integrated disease ontology that provides precise mappings between source ontologies that comprehensively covers cross-species diseases, from common to rare diseases.
Disease area: Cross species, intended to cover all areas of diseases, integrating source ontologies that cover Mendelian diseases (OMIM), rare diseases (Orphanet), neoplasms (NCIt), human diseases (DO), and others. See all sources here.
Use Cases: Mondo was developed for usage in the Monarch Initiative, a discovery system that allows navigation of similarities between phenotypes, organisms, and human diseases across many data sources and organisms. Mondo is also used by ClinGen for disease curations, the Kids First Data Resource Portal for disease annotations and others, see an extensive list here.
GitHub repo: https://github.com/monarch-initiative/mondo
Website: https://mondo.monarchinitiative.org/
OBO Foundry webpage: http://obofoundry.org/ontology/mondo.html
Open: yes

National Cancer Institute Thesaurus (NCIT)

Description: NCI Thesaurus (NCIt)is a reference terminology that includes broad coverage of the cancer domain, including cancer related diseases, findings and abnormalities. The NCIt OBO Edition aims to increase integration of the NCIt with OBO Library ontologies. NCIt OBO Edition releases should be considered experimental.
Disease area: Cancer and neoplasms
Use Cases: NCI Thesaurus (NCIt) provides reference terminology for many National Cancer Institute and other systems. It is used by the Clinical Data Interchange Standards Consortium Terminology (CDISC), the U.S. Food and Drug Administration (FDA), the Federal Medication Terminologies (FMT), and the National Council for Prescription Drug Programs (NCPDP). It provides extensive coverage of neoplasms and cancers.
GitHub repo: https://github.com/NCI-Thesaurus/thesaurus-obo-edition/issues
Website: https://ncithesaurus.nci.nih.gov/ncitbrowser/pages/home.jsf?version=20.11e
OBO Foundry webpage: http://obofoundry.org/ontology/ncit.html
Open: Yes

Neurological Disease Ontology (ND)

Description: A framework for the representation of key aspects of neurological disease.
Disease area: Neurology
Use Cases: Goal is to provide a framework to enable representation of aspects of neurological diseases that are relevant to their treatment and study. This project may be inactive, the last commit to GitHub was in 2016.
GitHub repo: https://github.com/addiehl/neurological-disease-ontology
Open: Yes

Online Mendelian Inheritance in Man (OMIM)

Description: a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily.
Disease area: Mendelian, genetic diseases.
Use Cases: Integrated into the disease ontology, used by the Human Phenotype Ontology for disease annotations, patients and researchers.
Website: https://omim.org/
Open: yes

Ontology of Cardiovascular Drug Adverse Events (OCVDAE)

Description: A biomedical ontology of cardiovascular drug–associated adverse events.
Disease area: Cardiovascular
Use Cases: One novel study of the OCVDAE project is the development of the PCR method. Specifically, an AE-specific drug class effect is defined to exist when all the drugs (drug chemical ingredients or drug products) in a drug class are associated with an AE, which is formulated as a proportional class level ratio ("PCR") = 1. See more information in the paper: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5653862/. This project may be inactive, the last GitHub commit was in 2019.
GitHub repo: https://github.com/OCVDAE/OCVDAE
Website: https://bioportal.bioontology.org/ontologies/OCVDAE
Open: yes

Ontology for General Medical Science (OGMS)

Description: An ontology of entities involved in a clinical encounter.
Use Cases: Provides a formal theory of disease that can be further elaborated by specific disease ontologies. It is intended to be used as a upper level ontology for other disease ontologies. Used by Cardiovascular Disease Ontology.
GitHub repo: https://github.com/OGMS/ogms
OBO Foundry webpage: http://obofoundry.org/ontology/ogms.html
Open: Yes

Ontology for Genetic Susceptibility Factor (OGSF)

Description: An application ontology to represent genetic susceptibility to a specific disease, adverse event, or a pathological process.
Use Cases: Modeling genetic susceptibility to vaccine adverse events.
GitHub repo: https://github.com/linikujp/OGSF
OBO Foundry webpage: http://obofoundry.org/ontology/ogsf.html
Open: Yes

Ontology of Glucose Metabolism Disorder (OGMD)

Description: Represents glucose metabolism disorder and diabetes disease names, phenotypes, and their classifications.
Disease area: Metabolic disorders
Use Cases: Still under development (last verssion released in BioPortal was in 2021) but there is little information about its usage online.
Website: https://bioportal.bioontology.org/ontologies/OGMD
Open: Yes

Ontology of Language Disorder in Autism (LDA)

Description: An ontology assembled from a set of language terms mined from the autism literature.
Disease area: Austism
Use Cases: This has not been released since 2008 and looks like it is inactive.
Website: https://bioportal.bioontology.org/ontologies/LDA
Open: Yes

The Oral Health and Disease Ontology (OHD)

Description: Represents the content of dental practice health records and is intended to be further developed for use in translational medicine. OHD is structured using BFO (Basic Formal Ontology) and uses terms from many ontologies, NCBITaxon, and a subset of terms from the CDT (Current Dental Terminology).
Disease area: Oral health and disease
Use Cases: Used to represent the content of dental practice health records and is intended to be further developed for use in translation medicine. Appears to be inactive.
OBO Foundry webpage: http://www.obofoundry.org/ontology/ohd.html
Open: Yes

Orphanet (ORDO)

Description: The portal for rare diseases and orphan drugs. Contains a structured vocabulary for rare diseases capturing relationships between diseases, genes, and other relevant features, jointly developed by Orphanet and the EBI. It contains information on nearly 10,000 cancers and related diseases, 8,000 single agents and combination therapies, and a wide range of other topics related to cancer and biomedical research.
Disease area: Rare diseases
Use Cases: Used by rare disease research and clinical community. Integrated into the Mondo disease ontology, aligned with OMIM.
Website: https://www.orpha.net/consor/cgi-bin/index.php
Open: Yes

Parkinson Disease ontology (PDO)

Description: A comprehensive semantic framework with a subclass-based taxonomic hierarchy, covering the whole breadth of the Parkinson disease knowledge domain from major biomedical concepts to different views on disease features held by molecular biologists, clinicians, and drug developers.
Disease area: Parkinson disease
Use Cases: This resource has been created for use in the IMI-funded AETIONOMY project. Last release was in 2015, may be inactive. **Website:** https://bioportal.bioontology.org/ontologies/PDON
Open: Yes

Pathogenic Disease Ontology (PDO)

Description: Provides information on infectious diseases, disease synonyms, transmission pathways, disease agents, affected populations, and disease properties. Diseases are grouped into syndromic disease categories, organisms are structured hierarchically, and both disease transmission and relevant disease properties are searchable.
Disease area: human infectious diseases caused by microbes and the diseases that is related to microbial infection.
Use Cases: Has not been released since 2016 and may be inactive.
Website: https://bioportal.bioontology.org/ontologies/PDO
Open: Yes.

PolyCystic Ovary Syndrome Knowledgebase (PCOSKB)

Description: Comprises genes, single nucleotide polymorphisms, diseases, gene ontology terms, and biochemical pathways associated with polycystic ovary syndrome, a major cause of female subfertility worldwide.
Disease area: polycystic ovary syndrome **Use Cases:** Ontology underlying the Polycystic Ovary Syndrome Knowledgebase, a manually curated knowledgebase on PCOS.
Website: http://pcoskb.bicnirrh.res.in/go_d.php
Open: Yes

Rat Disease Ontology (RDO)

Description: Provides the foundation for ten comprehensive disease area–related data sets at the Rat Genome Database Disease Portals.
Disease area: Broad coverage including animal diseases, infectious diseases, chemically-induced disorders, occupational diseases, wounds and injuries and more.
Use Cases: Developed for use with the Rat Genome Database Disease Portals.
Website: https://rgd.mcw.edu/rgdweb/ontology/view.html?acc_id=DOID:4
Open: Yes

Removable Partial Denture Ontology (RPDO)

Description: Represents knowledge of a patient's oral conditions and denture component parts, originally developed to create a clinician decision support model.
Disease area: Oral health and dentures
Use Cases: A paper was published on this in 2016 but it does not appear any other information is available about this ontology on the website, presumably it is an inactive project.
Publication: https://www.nature.com/articles/srep27855
Open: No

Resource of Asian Primary Immunodeficiency Diseases (RPO)

Description: Represents observed phenotypic terms, sequence variations, and messenger RNA and protein expression levels of all genes involved in primary immunodeficiency diseases.
Disease area: Primary immunodeficiency diseases
Use Cases: This terminology is used in a freely accessible, dynamic and integrated database for primary immunodeficiency diseases (PID) called Resource of Asian Primary Immunodeficiency Diseases (RAPID), which is available here.
Open: Yes

Sickle Cell Disease Ontology (SCDO)

Description: SCDO establishes (a) community-standardized sickle cell disease terms and descriptions, (b) canonical and hierarchical representation of knowledge on sickle cell disease, and (c) links to other ontologies and bodies of work.
Disease area: Sickle Cell Disease (SCD). **Use Cases:** SCDO is intended to be a comprehensive collection of knowledge on SCD, facilitate exploration of new scientific questions and ideas, facilitate seamless data sharing and collaborations including meta-analysis within the SCD community, support the building of databasing and clinical informatics in SCD.
GitHub repo: https://github.com/scdodev/scdo-ontology/issues
Website: https://scdontology.h3abionet.org/
OBO Foundry webpage: http://obofoundry.org/ontology/scdo.html
Open: Yes

SNOMED Clinical Terminology (SNOMED CT)

Description: A comprehensive clinical terminology/ontology used in healthcare settings.
Disease area: Broad disease representation for human diseases.
Use Cases: Main coding system used in Electronic Health Records (EHRs).
Website: https://browser.ihtsdotools.org/?
Open: No, requires a license for usage.

Symptom Ontology

Description: An ontology of disease symptoms, with symptoms encompasing perceived changes in function, sensations or appearance reported by a patient indicative of a disease.
Disease area: Human diseases
Use Cases: Developed by the Disease Ontology (DO) team and used for describing symptoms of human diseases in the DO.
Website: http://symptomontologywiki.igs.umaryland.edu/mediawiki/index.php/Main_Page
OBO Foundry webpage: http://obofoundry.org/ontology/symp.html
Open: Yes

Unified Medical Language System

Description: The UMLS integrates and distributes key terminology, classification and coding standards, and associated resources to promote creation of more effective and interoperable biomedical information systems and services.
Disease area: Broad coverage

Use Cases: Healthcare settings including electronic health records and HL7.
Website: https://www.nlm.nih.gov/research/umls/index.html
Open: Yes

Phenotype ontologies

Phenotype Summary Table

Name	Species Area
Ascomycete phenotype ontology (APO)	Ascomycota
C. elegans phenotype (wbphenotype)	C elegans
Dictyostelium discoideum phenotype ontology (ddpheno)	Dictyostelium discoideum
Drosophila Phenotype Ontology (DPO)	Drosophila
Flora Phenotype Ontology (FLOPO)	Viridiplantae
Fission Yeast Phenotype Ontology (FYPO)	S. pombe
Human Phenotype Ontology (HPO)	Human
HPO - ORDO Ontological Module (HOOM)	Human
Mammalian Phenotype Ontology (MP)	Mammals
Ontology of Microbial Phenotypes (OMP)	Microbe
Ontology of Prokaryotic Phenotypic and Metabolic Characters	Prokaryotes
Pathogen Host Interaction Phenotype Ontology	pathogens
Planarian Phenotype Ontology (PLANP)	Schmidtea mediterranea
Plant Trait Ontology (TO)	Viridiplantae
Plant Phenology Ontology	Plants
Unified Phenotype Ontology (uPheno)	Cross-species coverage
Xenopus Phenotype Ontology (XPO)	Xenopus
Zebrafish Phenotype Ontology (ZP)	Zebrafish

Ascomycete phenotype ontology (APO)

Description: A structured controlled vocabulary for the phenotypes of Ascomycete fungi.
Species: Ascomycota
GitHub repo: https://github.com/obophenotype/ascomycete-phenotype-ontology/
Webpage: http://www.yeastgenome.org/ **OBO Foundry webpage:** http://obofoundry.org/ontology/wbphenotype.html
Open: Yes

C. elegans phenotype (wbphenotype)

Description: A structured controlled vocabulary of Caenorhabditis elegans phenotypes.
Species: C elegans
GitHub repo: https://github.com/obophenotype/c-elegans-phenotype-ontology
OBO Foundry webpage: http://obofoundry.org/ontology/wbphenotype.html
Open: Yes

Dictyostelium discoideum phenotype ontology (ddpheno)

Description: A structured controlled vocabulary of phenotypes of the slime-mould Dictyostelium discoideum.
Species: Dictyostelium discoideum

GitHub repo: https://github.com/obophenotype/dicty-phenotype-ontology/issues
Webpage: http://dictybase.org/
OBO Foundry webpage: http://obofoundry.org/ontology/ddpheno.html
Open: Yes

Drosophila Phenotype Ontology (DPO)

Description: An ontology of commonly encountered and/or high level Drosophila phenotypes.
Species: Drosophila
GitHub repo: https://github.com/obophenotype/c-elegans-phenotype-ontology
Webpage: http://purl.obolibrary.org/obo/fbcv
OBO Foundry webpage: http://obofoundry.org/ontology/dpo.html
Open: Yes

Flora Phenotype Ontology (FLOPO)

Description: Traits and phenotypes of flowering plants occurring in digitized Floras.
Species: Viridiplantae
GitHub repo: https://github.com/flora-phenotype-ontology/flopoontology/
OBO Foundry webpage: http://obofoundry.org/ontology/flopo.html
Open: Yes

Fission Yeast Phenotype Ontology (FYPO)

Description: FYPO is a formal ontology of phenotypes observed in fission yeast.
Species: S. pombe
GitHub repo: https://github.com/pombase/fypo
OBO Foundry webpage: http://obofoundry.org/ontology/fypo.html
Open: Yes

Human Phenotype Ontology (HPO)

Description: HPO provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality.
Species: Human
GitHub repo: https://github.com/obophenotype/human-phenotype-ontology
Website: https://hpo.jax.org/app/
OBO Foundry webpage: http://obofoundry.org/ontology/hp.html
Open: yes

HPO - ORDO Ontological Module (HOOM)

Description: Orphanet provides phenotypic annotations of the rare diseases in the Orphanet nomenclature using the Human Phenotype Ontology (HPO). HOOM is a module that qualifies the annotation between a clinical entity and phenotypic abnormalities according to a frequency and by integrating the notion of diagnostic criterion. In ORDO a clinical entity is either a group of rare disorders, a rare disorder or a subtype of disorder. The phenomes branch of ORDO has been refactored as a logical import of HPO, and the HPO-ORDO phenotype disease-annotations have been provided in a series of triples in OBAN format in which associations, frequency and provenance are modeled. HOOM is provided as an OWL (Ontologies Web Languages) file, using OBAN, the Orphanet Rare Disease Ontology (ORDO), and HPO ontological models. HOOM provides extra possibilities for researchers, pharmaceutical companies and others wishing to co-analyse rare and common disease phenotype associations, or re-use the integrated ontologies in genomic variants repositories or match-making tools.
Species: Human
Website: http://www.orphadata.org/cgi-bin/img/PDF/WhatIsHOOM.pdf
BioPortal: https://bioportal.bioontology.org/ontologies/HOOM
Open: yes

Mammalian Phenotype Ontology (MP)

Description: Standard terms for annotating mammalian phenotypic data.
Species: Mammals (main focus is on mouse and rodents)
GitHub repo: https://github.com/obophenotype/mammalian-phenotype-ontology
Website: http://www.informatics.jax.org/searches/MP_form.shtml
OBO Foundry webpage: http://obofoundry.org/ontology/mp.html
Open: Yes

Ontology of Microbial Phenotypes (OMP)

Description: An ontology of phenotypes covering microbes.
Species: microbes
GitHub repo: https://github.com/microbialphenotypes/OMP-ontology **Website:** http://microbialphenotypes.org **OBO Foundry webpage:** http://obofoundry.org/ontology/omp.html **Open:** Yes

Ontology of Prokaryotic Phenotypic and Metabolic Characters

Description: An ontology of phenotypes covering microbes.
Species: Prokaryotes **GitHub repo:** https://github.com/microbialphenotypes/OMP-ontology/issues
Website: http://microbialphenotypes.org/
OBO Foundry webpage: http://obofoundry.org/ontology/omp.html
Open: Yes

Pathogen Host Interaction Phenotype Ontology

Description: PHIPO is a formal ontology of species-neutral phenotypes observed in pathogen-host interactions.
Species: pathogens
GitHub repo: https://github.com/PHI-base/phipo
Website: http://www.phi-base.org
OBO Foundry webpage: http://obofoundry.org/ontology/phipo.html
Open: Yes

Planarian Phenotype Ontology (PLANP)

Description: Planarian Phenotype Ontology is an ontology of phenotypes observed in the planarian Schmidtea mediterranea.
Species: Schmidtea mediterranea
GitHub repo: https://github.com/obophenotype/planarian-phenotype-ontology
OBO Foundry webpage: http://obofoundry.org/ontology/planp.html
Open: Yes

Plant Trait Ontology (TO)

Description: A controlled vocabulary of describe phenotypic traits in plants.
Species: Viridiplantae
GitHub repo: https://github.com/Planteome/plant-trait-ontology/
OBO Foundry webpage: http://obofoundry.org/ontology/to.html
Open: Yes

Plant Phenology Ontology

Description: An ontology for describing the phenology of individual plants and populations of plants, and for integrating plant phenological data across sources and scales.
Species: Plants
GitHub repo: https://github.com/PlantPhenoOntology/PPO
OBO Foundry webpage: http://obofoundry.org/ontology/ppo.html
Open: Yes

Unified Phenotype Ontology (uPheno)

Description: The uPheno ontology integrates multiple phenotype ontologies into a unified cross-species phenotype ontology.
Species: Cross-species coverage
GitHub repo: https://github.com/obophenotype/upheno
OBO Foundry webpage: http://obofoundry.org/ontology/upheno.html
Open: Yes

Xenopus Phenotype Ontology (XPO)

Description: XPO represents anatomical, cellular, and gene function phenotypes occurring throughout the development of the African frogs Xenopus laevis and tropicalis.
Species: Xenopus
GitHub repo: https://github.com/obophenotype/xenopus-phenotype-ontology
OBO Foundry webpage: http://obofoundry.org/ontology/xpo.html
Open: Yes

Zebrafish Phenotype Ontology (ZP)

Description: The Zebrafish Phenotype Ontology formally defines all phenotypes of the Zebrafish model organism.
Species: Zebrafish
GitHub repo: https://github.com/obophenotype/zebrafish-phenotype-ontology
OBO Foundry webpage: http://obofoundry.org/ontology/zp.html
Open: Yes

References

- A Census of Disease Ontologies Melissa A. Haendel, Julie A. McMurry, Rose Relevo, Christopher J. Mungall, Peter N. Robinson, Christopher G. Chute Annual Review of Biomedical Data Science 2018 1:1, 305-331
- OMOP2OBO repository