

RDF representation of biomedical and genomic information related to inherited diseases of glycan metabolism provided by NCBI and available from <ftp://ftp.ncbi.nlm.nih.gov/pub/medgen/>, <ftp://ftp.ncbi.nih.gov/gene/>, and <ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/>

Namespaces

The following RDF namespaces and prefixes will be used in the documentation.

Prefix	URI
rdf	http://www.w3.org/1999/02/22-rdf-syntax-ns#
rdfs	http://www.w3.org/2000/01/rdf-schema#
uniprot	http://purl.uniprot.org/core/
ggdsch	http://jcggdb.jp/rdf/diseases/ggdonto-schema#
gmonto	http://jcggdb.jp/rdf/diseases/gmnncbi#

dct	http://purl.org/dc/terms/
gdgsch	http://jcggdb.jp/rdf/diseases/gdgdb-schema#
gmsch	http://jcggdb.jp/rdf/diseases/gmncbi-schema#
owl	http://www.w3.org/2002/07/owl#
pav	http://purl.org/pav/
skos	http://www.w3.org/2004/02/skos/core#
bibo	http://purl.org/ontology/bibo/
foaf	http://xmlns.com/foaf/0.1/

Disease Description

For describing information about diseases that are known to be related to glycans

URI: <http://jcggdb.jp/rdf/diseases/gdgdb-schema#DiseaseDescription>

Superclass: uniprot:Annotation, http://ncicb.nci.nih.gov/xml/owl/EVS/Thesaurus.owl#Diseases_and_Disorders

Predicates that have this class as domain:

Predicate	Range	Functional	Description
gmsch:hpoName	http://www.w3.org/2001/XMLSchema#string		For description data from

			MedGen_HPO_OMIM_Mapping.txt file
gmsch:hpoUi	http://www.w3.org/2001/XMLSchema#string		For description data from MedGen_HPO_OMIM_Mapping.txt file
gmsch:medgenCui	http://www.w3.org/2001/XMLSchema#string		For description data from mim2gene_medgen and gene_condition_source_id files
gmsch:medgenName	http://www.w3.org/2001/XMLSchema#string		For description data from MedGen_HPO_OMIM_Mapping.txt file
gmsch:medgenSource	http://www.w3.org/2001/XMLSchema#string		For description data from MedGen_HPO_OMIM_Mapping.txt, mim2gene_medgen and gene_condition_source_id files
gmsch:nameInSource	http://www.w3.org/2001/XMLSchema#string		For description data from gene_condition_source_id file
gmsch:omimDisName	http://www.w3.org/2001/XMLSchema#string		For description data from MedGen_HPO_OMIM_Mapping.txt file
gmsch:omimMIMnumber	http://www.w3.org/2001/XMLSchema#string		For description data from mim2gene_medgen and gene_condition_source_id files
gmsch:omimType	http://www.w3.org/2001/XMLSchema#string		For description data from mim2gene_medgen file
gmsch:sourceConId	http://www.w3.org/2001/XMLSchema#string		For description data from gene_condition_source_id file

gmsch:umlsConDef	http://www.w3.org/2001/XMLSchema#string		For description of concepts in RDF representation of biomedical and genomic information provided in MedGen
gmsch:umlsConName	http://www.w3.org/2001/XMLSchema#string		For description of concepts in RDF representation of biomedical and genomic information provided in MedGen
gmsch:umlsHpoCui	http://www.w3.org/2001/XMLSchema#string		For description data from MedGen_HPO_OMIM_Mapping.txt file
gmsch:umlsRela	http://www.w3.org/2001/XMLSchema#string		For description data from MedGen_HPO_OMIM_Mapping.txt file
gmsch:umlsSource	http://www.w3.org/2001/XMLSchema#string		For description of concepts in RDF representation of biomedical and genomic information provided in MedGen
gmsch:umlsSty	http://www.w3.org/2001/XMLSchema#string		For description data from MedGen_HPO_OMIM_Mapping.txt file

gdg sch:DiseaseDescriptionNcbi

URI: <http://jcggdb.jp/rdf/diseases/gdgdb-schema#DiseaseDescriptionNcbi>

Superclass: owl:Thing

Gene Description

For describing information about glycan-related genes

URI: <http://jcggdb.jp/rdf/diseases/gdgdb-schema#GeneDescription>

Superclass: uniprot:Annotation, <http://ncicb.nci.nih.gov/xml/owl/EVS/Thesaurus.owl#Gene>

Predicates that have this class as domain:

Predicate	Range	Functional	Description
gmsch:medgenCui	http://www.w3.org/2001/XMLSchema#string		For description data from mim2gene_medgen and gene_condition_source_id files
gmsch:nameInSource	http://www.w3.org/2001/XMLSchema#string		For description data from gene_condition_source_id file
gmsch:ncbiGeneSymbol	http://www.w3.org/2001/XMLSchema#string		Preferred symbol corresponding to the GeneID.
gmsch:omimType	http://www.w3.org/2001/XMLSchema#string		For description data from mim2gene_medgen file
gmsch:sourceConId	http://www.w3.org/2001/XMLSchema#string		For description data from gene_condition_source_id file
gmsch:umlsConDef	http://www.w3.org/2001/XMLSchema#string		For description of concepts in RDF representation of biomedical and genomic information provided in MedGen
gmsch:umlsConName	http://www.w3.org/2001/XMLSchema#string		For description of concepts in RDF representation of biomedical and genomic information provided in MedGen
gmsch:umlsSource	http://www.w3.org/2001/XMLSchema#string		For description of concepts in RDF representation of

			biomedical and genomic information provided in MedGen
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PubMed Disease Data

For describing information about diseases from the PubMed.

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#DiseaseDataPubMed>

Superclass: gdgsch:DiseaseDescription

Disease and Gene Data from NCBI

For describing information about diseases and genes from the NCBI.

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#DiseaseGeneDataNcbi>

Superclass: gdgsch:GeneDescription, gdgsch:DiseaseDescription

Predicates that have this class as domain:

Predicate	Range	Functional	Description
gmsch:ncbiGeneData	gmsch:NcbiGeneData		Data from NCBI Gene and ClinVar.

Name from Source

Disease name from UMLS sources.

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#NameFromSource>

Superclass: gmsch:DiseaseDescriptionNcbi

Definition from Source

Disease definition from UMLS sources.

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#DefinitionFromSource>

Superclass: gmsch:DiseaseDescriptionNcbi

HPO Data

Data from HPO.

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#HpoData>

Superclass: gmsch:DiseaseDescriptionNcbi

NCBI Gene Data

Data from NCBI Gene and ClinVar.

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#NcbiGeneData>

Superclass: gmsch:DiseaseGeneDataNcbi

Information Sources

Medical and biological information sources that are created or used in NCBI

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#InfoSources>

Superclass: owl:Thing

Predicates that have this class as domain:

Predicate	Range	Functional	Description
gmsch:sourceAbbrNcbi	http://www.w3.org/2001/XMLSchema#string		Source Abbreviation in NCBI
gmsch:sourceOfficialAbbr	http://www.w3.org/2001/XMLSchema#string		Source Official Abbreviation
gmsch:sourceOfficialName	http://www.w3.org/2001/XMLSchema#string		Source Official Name
gmsch:sourceURI	http://www.w3.org/2001/XMLSchema#string		Source URI

Instances:

URI	Label	Description
gmsch:source_SNOMEDCT	SNOMEDCT	Systematized Nomenclature of Medicine - Clinical Terms
gmsch:source_MDR	MDR	Medical Dictionary for Regulatory Activities Terminology
gmsch:source_GHP	Genetics Home Reference	Genetics Home Reference
gmsch:source_MTH	MTH	UMLS Metathesaurus
gmsch:source_GTR	GTR	Genetic Testing Registry

gmsch:source_MSH	MeSH	Medical Subject Headings
gmsch:source_HPO	HPO	Human Phenotype Ontology
gmsch:source_NCI	NCI	National Cancer Institute Thesaurus
gmsch:source_GR	GeneReviews	GeneReviews
gmsch:source_OMIM	OMIM	

Semantic Relations UMLS

For describing information about semantic relations between concepts in this ontology, by using relations from UMLS.

URI: <http://jcggdb.jp/rdf/diseases/gmncbi-schema#SemanticRelationsUMLS>

Superclass: owl:ObjectProperty

Predicates that have this class as domain:

Predicate	Range	Functional	Description
gmsch:umlsAbr	http://www.w3.org/2001/XMLSchema#string		UMLS: Abbreviation of the Relation.
gmsch:umlsHI	http://www.w3.org/2001/XMLSchema#string		UMLS: Hierarchical links of the Relation to its parent ({isa}) and its children ({inverse_isa}). If there are no hierarchical links, then the value "none" is assigned.
gmsch:umlsRin	http://www.w3.org/2001/XMLSchema#string		UMLS: Name of the inverse of the Relation.

gmsch:umlsRI	http://www.w3.org/2001/XMLSchema#string		UMLS: Name of the Relation.
gmsch:umlsRIDef	http://www.w3.org/2001/XMLSchema#string		UMLS: Definition of the Relation.
gmsch:umlsRtn	http://www.w3.org/2001/XMLSchema#string		UMLS: Tree Number of the Relation.
gmsch:umlsUi	http://www.w3.org/2001/XMLSchema#string		UMLS: Unique Identifier of the Relation.

Instances:

URI	Label	Description
gmsch:causedBy	caused_by	This semantic relation is used for linking the Genetic Glyco-Diseases Ontologies (GGDonto) ontology to the RDF representation of biomedical and genomic information from NCBI.
gmsch:conceptualPartOf	conceptual_part_of	This semantic relation is used for linking the Genetic Glyco-Diseases Ontologies (GGDonto) ontology to the RDF representation of biomedical and genomic information from NCBI.
gmsch:hasManifestation	has_manifestation	This semantic relation is used for linking the Genetic Glyco-Diseases Ontologies (GGDonto) ontology to the RDF representation of biomedical and genomic information from NCBI.
gmsch:hasConceptualPart	has_conceptual_part	This semantic relation is used for linking the Genetic Glyco-Diseases Ontologies (GGDonto) ontology to the RDF representation of biomedical and genomic information from NCBI.
gmsch:conceptuallyRelatedTo	conceptually_related_to	This semantic relation is used for linking the Genetic Glyco-Diseases Ontologies (GGDonto) ontology to the RDF representation of biomedical and genomic

		information from NCBI.
gmsch:manifestationOf	manifestation_of	This semantic relation is used for linking the Genetic Glyco-Diseases Ontologies (GGDonto) ontology to the RDF representation of biomedical and genomic information from NCBI.
gmsch:causes	causes	This semantic relation is used for linking the Genetic Glyco-Diseases Ontologies (GGDonto) ontology to the RDF representation of biomedical and genomic information from NCBI.

Concept Type

For describing types of concepts

URI: <http://jcggdb.jp/rdf/diseases/ggdonto-schema#ConceptType>

Superclass: owl:Thing

Instances:

URI	Label	Description
gmsch:GeneInfoNcbi	NCBI gene info	
gmsch:DiseaseAndPubMedLink	Disease name and PubMed link information	
gmsch:DiseaseAndPhenotype	Disease and Phenotype description	
gmsch:DiseaseAndGeneNcbiOmim	Disease and gene data from NCBI and OMIM	

gmsch:DiseaseAndGeneNcbi	Disease name and NCBI gene data	
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Glyco-related Concept Schemes

For describing linkages between SKOS Concepts in different Concept Schemes that are defined in Ontologies developed by Glycoscience and Glycotechnology Research Group, AIST

URI: <http://jcggdb.jp/rdf/diseases/ggdonto-schema#GlycoRelatedConceptSchemes>

Superclass: skos:ConceptScheme

Instances:

URI	Label	Description
gmsch:ConceptSchemePubMed	This concept scheme includes the concepts that represent entries from MedGen, such as link information between UMLS Concept Unique Identifiers for inherited diseases of glycan metabolism and PubMed unique identifiers.	
gmsch:ConceptSchemeNcbiGeneInfo	This concept scheme includes the concepts that represent entries from NCBI (National Center for Biotechnology Information) Gene database.	
gmsch:ConceptSchemeNcbiGene	This concept scheme includes the concepts that represent entries from Gene and ClinVar, such as information about the relationships between MIM numbers (OMIM), GeneIDs, and Records in MedGen.	
gmsch:ConceptSchemeMedGen	This concept scheme includes the concepts that represent entries from MedGen related to	

	inherited diseases of glycan metabolism.	
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