

# GGDonto - Ontology of Genetic Diseases known to be related to Glycan Metabolism

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## Namespaces

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

Prefix	URI
rdf	<a href="http://www.w3.org/1999/02/22-rdf-syntax-ns#">http://www.w3.org/1999/02/22-rdf-syntax-ns#</a>
prism	<a href="http://prismstandard.org/namespaces/basic/2.0/">http://prismstandard.org/namespaces/basic/2.0/</a>
rdfs	<a href="http://www.w3.org/2000/01/rdf-schema#">http://www.w3.org/2000/01/rdf-schema#</a>
uniprot	<a href="http://purl.uniprot.org/core/">http://purl.uniprot.org/core/</a>
ggdsch	<a href="http://jcggdb.jp/rdf/diseases/ggdonto-schema#">http://jcggdb.jp/rdf/diseases/ggdonto-schema#</a>
dct	<a href="http://purl.org/dc/terms/">http://purl.org/dc/terms/</a>
gmsch	<a href="http://jcggdb.jp/rdf/diseases/gmncbi-schema#">http://jcggdb.jp/rdf/diseases/gmncbi-schema#</a>
gdgsch	<a href="http://jcggdb.jp/rdf/diseases/gdgdbschema#">http://jcggdb.jp/rdf/diseases/gdgdbschema#</a>
owl	<a href="http://www.w3.org/2002/07/owl#">http://www.w3.org/2002/07/owl#</a>
pav	<a href="http://purl.org/pav/">http://purl.org/pav/</a>

skos	<a href="http://www.w3.org/2004/02/skos/core#">http://www.w3.org/2004/02/skos/core#</a>
bibo	<a href="http://purl.org/ontology/bibo/">http://purl.org/ontology/bibo/</a>
foaf	<a href="http://xmlns.com/foaf/0.1/">http://xmlns.com/foaf/0.1/</a>
ggdonto	<a href="http://jcggdb.jp/rdf/diseases/ggdonto#">http://jcggdb.jp/rdf/diseases/ggdonto#</a>

## 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](http://ncicb.nci.nih.gov/xml/owl/EVS/Thesaurus.owl#Diseases_and_Disorders)

**Predicates that have this class as domain:**

Predicate	Range	Functional	Description
ggdsch:diseaseIncluded	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		Other entities represented in this disease in OMIM, MeSH and other resources
ggdsch:diseasePreferredTerm	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		Preferred term for disease (from OMIM, MeSH and other resources)
ggdsch:diseasePreferredTermAbbr	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		Abbreviation for "Preferred term for disease" (from OMIM, MeSH and other resources)

ggdsch:diseaseSynonyms	http://www.w3.org/2001/XMLSchema#string		Synonyms for disease name (from OMIM, MeSH and other resources)
ggdsch:umlsCODE	http://www.w3.org/2001/XMLSchema#string		UMLS: Unique Identifier or code for string in source
ggdsch:umlsCUI	http://www.w3.org/2001/XMLSchema#string		UMLS: Unique identifier for concept
ggdsch:umlsSAB	http://www.w3.org/2001/XMLSchema#string		UMLS: Source abbreviation; ICD-10-CM: International Classification of Diseases, Tenth Revision, Clinical Modification; MeSH: Medical Subject Headings; MeSH (SCR): Supplemental Concept Record from MeSH; SNOMED CT: SNOMED Clinical Terms;

## GGDonto Disease Description

For describing information about diseases that are recorded in GGDonto

**URI:** <http://jcgddb.jp/rdf/diseases/ggdonto-schema#DiseaseDescriptionGGDonto>

**Superclass:** gdgsch:DiseaseDescription

**Predicates that have this class as domain:**

Predicate	Range	Functional	Description
ggdsch:diseaseType	ggdsch:DiseaseType	yes	Property for describing types of diseases

ggdsch:geneSymbolGGDB	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		For link to the GlycoGene DataBase
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## Clinical Findings Description

For describing clinical findings of diseases

**URI:** <http://jcgddb.jp/rdf/diseases/ggdonto-schema#ClinicalFindingsDescription>

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

**Predicates that have this class as domain:**

Predicate	Range	Functional	Description
ggdsch:umlsCODE	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		UMLS: Unique Identifier or code for string in source
ggdsch:umlsCUI	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		UMLS: Unique identifier for concept
ggdsch:umlsSAB	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		UMLS: Source abbreviation; ICD-10-CM: International Classification of Diseases, Tenth Revision, Clinical Modification; MeSH: Medical Subject Headings; MeSH (SCR): Supplemental Concept Record from MeSH; SNOMED CT: SNOMED Clinical Terms;

## Classification Description

For describing classification of diseases

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

**Superclass:** uniprot:Annotation

**Predicates that have this class as domain:**

Predicate	Range	Functional	Description
ggdsch:umlsCODE	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		UMLS: Unique Identifier or code for string in source
ggdsch:umlsCUI	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		UMLS: Unique identifier for concept
ggdsch:umlsSAB	<a href="http://www.w3.org/2001/XMLSchema#string">http://www.w3.org/2001/XMLSchema#string</a>		UMLS: Source abbreviation; ICD-10-CM: International Classification of Diseases, Tenth Revision, Clinical Modification; MeSH: Medical Subject Headings; MeSH (SCR): Supplemental Concept Record from MeSH; SNOMED CT: SNOMED Clinical Terms;

## ggdsch:DiseaseTypeClass

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

**Superclass:** owl:Thing

## Semantic Relations GGDonto

For describing information about semantic relations between concepts in GGDonto

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

**Superclass:** owl:ObjectProperty

**Predicates that have this class as domain:**

Predicate	Range	Functional	Description
ggdsch:ontologyType	ggdsch:OntologyType	yes	For describing ontology type for new semantic relations properties
ggdsch:parentOntology	ggdsch:TopLevelOntologies	yes	For describing parent ontology for new semantic relations properties
ggdsch:topOntology	ggdsch:TopLevelOntologies	yes	For describing top ontology for new semantic relations properties

## Top Level Ontologies

For describing Top Level Ontologies

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

**Superclass:** owl:Thing

**Instances:**

URI	Label	Description
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ggdsch:PhenotypeLSD	Ontology for Disorders of Glycan Degradation (Lysosomal Storage Diseases); classified by phenotype	
ggdsch:PhenotypeGD	Ontology for Disorders of Glycosylation; classified by phenotype	
ggdsch:PathwayLSD	Ontology for Disorders of Glycan Degradation (Lysosomal Storage Diseases); classified by pathway	
ggdsch:PathwayGDtradi	Traditional nomenclature for Disorders of Glycosylation; classified by pathway	
ggdsch:PathwayGDnew	New nomenclature for Disorders of Glycosylation; classified by pathway	
ggdsch:MultiTypesDis	Classification of disease or disorder with multiple types	
ggdsch:InbornErrGlycans	Classification of Inborn Errors of Glycan Metabolism	

## Ontology Type

For describing types of ontologies

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

**Superclass:** owl:Thing

**Instances:**

URI	Label	Description
ggdsch:TopOntology	Top Ontology	
ggdsch:SubOntology	Sub Ontology	

## Concept Type

For describing types of concepts

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

**Superclass:** owl:Thing

**Instances:**

URI	Label	Description
ggdsch:SingleDisease	Single Disease	
ggdsch:MultipleTypesDisease	Multiple Types Disease	Disease with multiple types
ggdsch:DiseaseType	Disease Type	
ggdsch:ClinicalFindings	Clinical Findings	
ggdsch:ClassificationTerm	Classification Term	

## Glyco-related Databases

For describing linkages between concepts and resources in related glycomics databases

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



**Superclass:** uniprot:Database

**Instances:**

URI	Label	Description
ggdsch:GGDB	GlycoGene Database (GGDB)	GGDB was created by Research Center for Medical Glycoscience (RCMG). GGDB provides necessary information for the analysis of glycogenes.
ggdsch:GDGDB	Glyco-Disease Genes Database (GDGDB)	GDGDB was created by Research Center for Medical Glycoscience (RCMG) and released in April 2010. At present time GDGDB provides information on about 80 genetic diseases of both glycan synthesis and degradation.

## 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
ggdsch:ConceptSchemePhenotypeLSD	Concept Scheme about Genetic disorders of glycan degradation (by systems, signs and symptoms)	

ggdsch:ConceptSchemePhenotypeGD	Concept Scheme about Genetic disorders of glycan synthesis (by systems, signs and symptoms)	
ggdsch:ConceptSchemePathwayLSD	Concept Scheme about Genetic disorders of glycan degradation	
ggdsch:ConceptSchemePathwayGDtradi	Concept Scheme about Genetic disorders of glycan synthesis (traditional nomenclature)	
ggdsch:ConceptSchemePathwayGDnew	Concept Scheme about Genetic disorders of glycan synthesis (new nomenclature)	
ggdsch:ConceptSchemeMultiTypesDis	Concept Scheme about Multiple Types Diseases	
ggdsch:ConceptSchemeInbornErrGlycans	Concept Scheme about Congenital, Hereditary, and Neonatal Diseases and Abnormalities related to Glycans	