


The complexity of glycan metabolic pathways and the large numbers of genetic defects in these pathways make it difficult to deal with the broad range of diseases that are caused by mutations in glycan-related genes. The [Glyco-Disease Genes Database \(GDGDB\)](#) was created by [Research Center for Glycoscience \(RCMG\)](#) and released in [April 2010](#). This database is supported by Ministry of Education, Culture, Sports, Science and Technology (MEXT) as part of Life Science Integrated Database project. At present time [GDGDB](#) provides information on about [80 genetic diseases](#) of both glycan synthesis and degradation, and information on [mutant genes](#) that cause these diseases. In addition, we designed and created the “[Pathway](#)” and “[Phenotype](#)” ontologies for both types of these genetic diseases ([Congenital Disorders of Glycosylation \(CDG\)](#) and [Lysosomal Storage Diseases \(LSD\)](#)) and included them in the [GDGDB](#).

The Web-based user interface was created and released and now available at <http://jcggdb.jp/doc/ProjectTop.action?langType=1&projectId=2>.




JCGGDB

Glyco-Disease Genes Database

CMS TOP

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Disease	更新日
Alpha-Mannosidosis	2014/03/25
Aspartylglucosaminuria	2014/03/25
Beta-Mannosidosis	2014/03/25
Bruck syndrome	2014/03/25
Cholesteryl ester and triglyceride storage diseases	2014/03/28
Congenital Disorder of Glycosylation type Ia	2014/03/25
Congenital Disorder of Glycosylation type Ib	2014/03/25
Congenital Disorder of Glycosylation type Ic	2014/03/25
Congenital Disorder of Glycosylation type Id	2014/03/25
Congenital Disorder of Glycosylation type Ie	2014/03/25
Congenital Disorder of Glycosylation type If	2014/03/25
Congenital Disorder of Glycosylation type Ig	2014/03/25
Congenital Disorder of Glycosylation type Ih	2014/03/25
Congenital Disorder of Glycosylation type IIa	2014/03/25
Congenital Disorder of Glycosylation type IIb	2014/03/25
Congenital Disorder of Glycosylation type IIc	2014/03/25
Congenital Disorder of Glycosylation type IId	2014/03/25
Congenital Disorder of Glycosylation type IIe	2014/03/25
Congenital Disorder of Glycosylation type Ii	2014/03/25
Congenital Disorder of Glycosylation type Ij	2014/03/25
Congenital Disorder of Glycosylation type Ik	2014/03/25
Congenital Disorder of Glycosylation type Il	2014/03/25
Congenital Disorder of Glycosylation type Ix	2010/02/16
Congenital dyserythropoietic anemia type II	2014/03/25
Congenital muscular dystrophy type 1C (MDC1C)	2014/03/25
Dilated cardiomyopathy (CMD1X)	2014/03/25
Ehlers-Danlos syndrome type VIA	2014/03/25
Fucoxidosis	2014/03/25
Fukuyama congenital muscular dystrophy	2014/03/25
Galactosialidosis	2014/03/28
Glycogen storage disease type II	2014/03/25
Guillain-Barre syndrome	2014/03/25



JCGGDB

Glyco-Disease Genes Database

CMS TOP
> Glyco-Disease Genes Database

1. Disease
Congenital Disorder of Glycosylation type Ia
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2. Disease gene

3. Genetic Glyco-Diseases Ontologies

Disease

Disease

CDG Ia (Congenital Disorder of Glycosylation type Ia)

Pathosis

Some of these patients have been mistaken for having mitochondrial disorders. Hypotonia, variable psychomotor retardation, seizures, peripheral neuropathy, stroke-like episodes, strabismus, cardiomyopathy

Reference: Glycoforum -Beyond Glycogenes, Dr. Hudson H. Freeze

<http://www.glycoforum.gr.jp/science/glycogenes/08/08E.html>

OMIM

CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ia;
CDG1A

Edited by Shikana

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[illegible]

GDGDB & GGDonto ontologies - RDF representation of the GDGDB data and Ontology for Genetic Diseases known to be related to Glycan Metabolism

Glyco-Disease Genes Database (GDGDB) was developed by the Research Center for Medical Glycoscience (RCMG) and released in April 2010. Being the members of the “Life-Science Database Integration Project” of National Bioscience Database Center (NBDC) of Japan Science and Technology Agency (JST), we decided to organize GDGDB information and enrich its content by integration with other biomedical resources. We designed and developed the ontologies, called GDGDB ontology and GGDonto. In addition to these ontologies, we developed a system with the user interfaces, using which the users can retrieve and search information from GDGDB and GGDonto. These XML-based and SPARQL-based user interfaces and RDF files for GDGDB ontology and GGDonto are available at <http://acgg.asia/db/diseases/>. We hope that these GDGDB & GGDonto ontologies with GDGDB database will help the users to better understand the etiology, pathogenesis and manifestations of the genetic diseases known to be related to glycan metabolism.

Ontologies for genetic and infectious diseases known to be related to glycan metabolism and glycan binding

Glyco-Disease Genes Database (GDGDB) and Pathogen Adherence to Carbohydrate Database (PACDB) were developed by the Research Center for Medical Glycoscience. With the purpose to organize their information and enrich their contents with data from other resources we designed and created GGDonto and PAConto ontologies, respectively. These ontologies are based on the semantic web technologies and represented in RDF. We hope that these ontologies and databases will help the users to better understand the etiology, pathogenesis and manifestations of the diseases known to be related to glycan metabolism and glycan binding.

GGDonto ontology

Genetic Glyco-Diseases Ontology (GGDonto): Ontology of Genetic Diseases related to Glycans

GDGDB ontology

RDF representation of the data from the Glyco-Disease Genes Database

PAConto ontology

Ontology and RDF representation of the data from the Pathogen Adherence to Carbohydrate Database

[Introduction](#) | [GGDonto & GDGDB ontologies](#) | [PAConto ontology](#) | [Link to databases](#)

GGDonto & GDGDB ontologies

User Interfaces

GGDonto and GDGDB ontologies

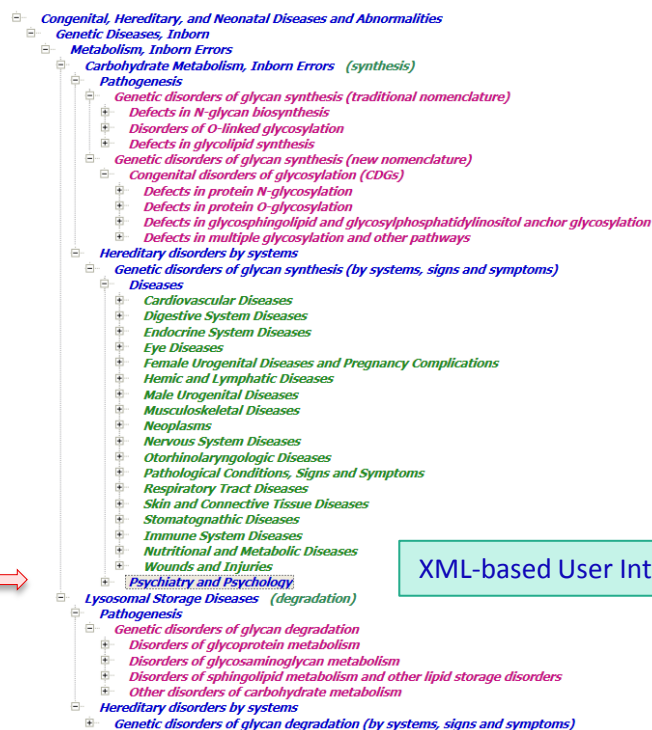
- SPARQL-based
- XML-based

RDF and documentation files

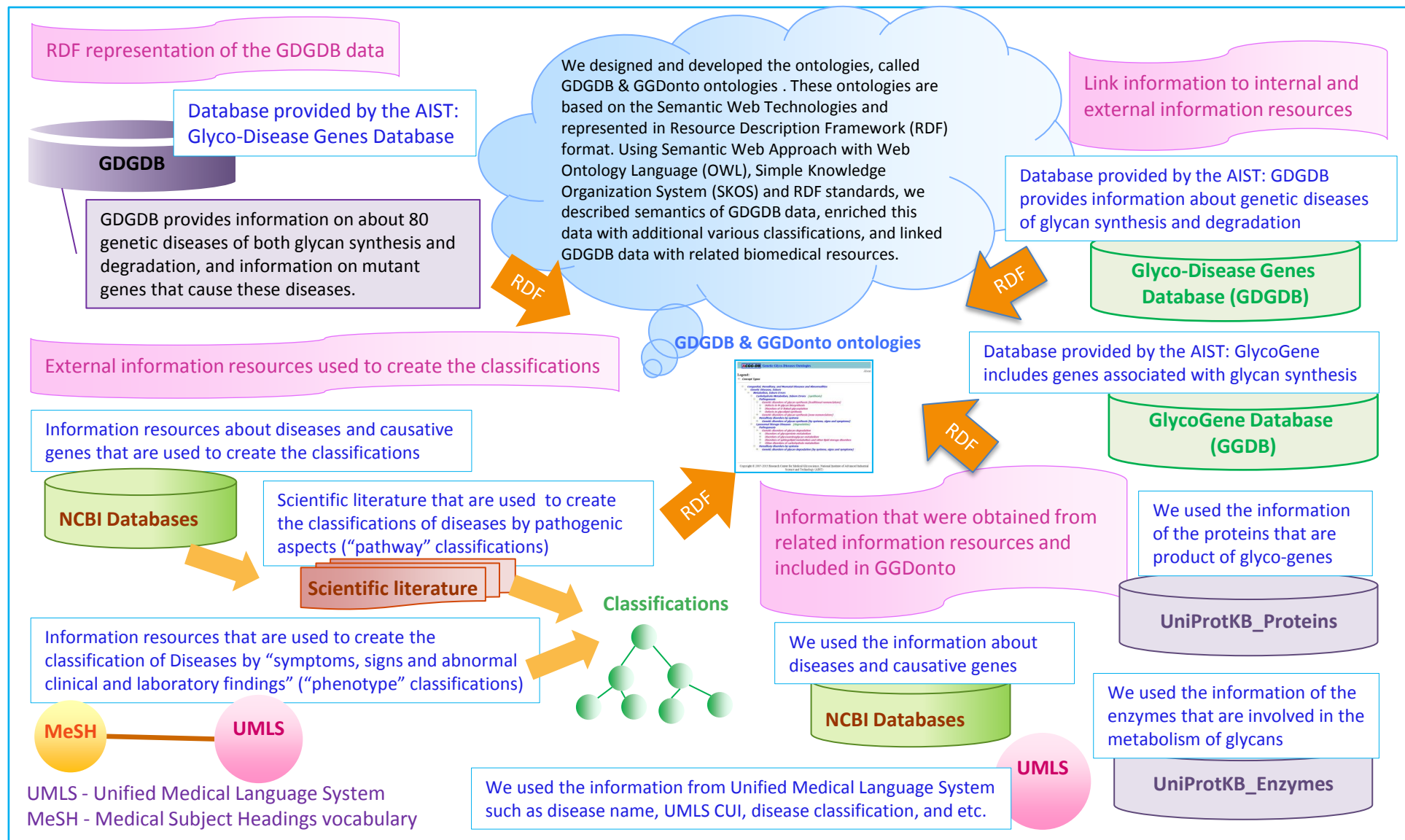
GGDonto ontology

- [Introduction](#)
- Ontology description and definition of classes and properties in [RDF/XML](#).
- Ontology description and definition of classes and properties in [RDF/Turtle](#)
- Data in [RDF/XML](#)

<http://acgg.asia/db/diseases/>
Web page in that the RDF files and the User Interfaces are provided

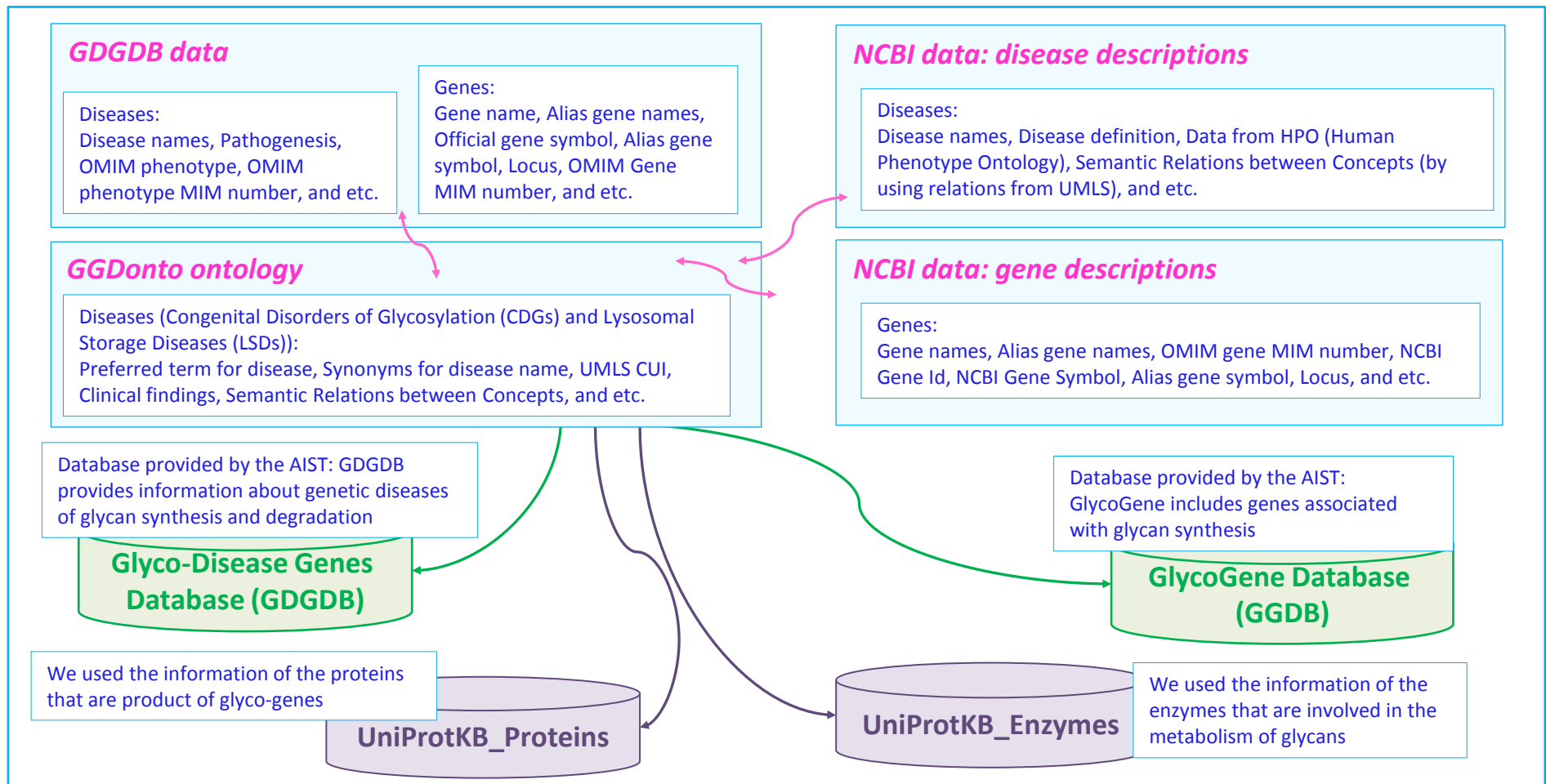


XML-based User Interface



Features of this ontology

- The proposed ontology has the advantage of representing comprehensible knowledge related to genetic defects in glycan metabolism, because it integrates the pathogenic aspects with the clinical features associated with each of these genetic disorders.
- The objects of these database and ontology are genetic disorders which are caused by inherent defects in single gene (single-gene disorders). Since the information about these diseases and they causative genes have been published in a variety of medical-related information resources, the relations between this ontology and external information resources are diverse.



Changing the Layout of the list of Diseases
Menu for changing the layout of the list

List of Diseases

Facets sets: Faceted browsing and filtering

- Database
- Disease Types by Metabolic Pathways
- Manifestation
- Ontology Tree

ACGG-DB Genetic Glyco-Diseases Ontology (GGDonto) and Glyco-Disease Genes DataBase (GDGDB)

Genetic Diseases of Glycan Metabolism

120 List of Diseases

sorted by: labels, then by...

1. Achondrogenesis type IB
[Aliases] ACG1B; Achondrogenesis, Fraccaro type
2. ALG11-CDG
[Aliases] Congenital Disorder of Glycosylation, Type Ip; CDG-Ip
3. **ALG12-CDG**
[Aliases] Congenital disorder of glycosylation, type Ig; CDG-Ig
4. ALG13-CDG
[Aliases] Congenital Disorder of Glycosylation, Type Is; CDG-Is
5. ALG1-CDG
[Aliases] Congenital disorder of glycosylation, type Ik; CDG-Ik
6. ALG2-CDG
[Aliases] CDG-Ii; Congenital disorder of glycosylation, type Ii
7. ALG3-CDG
[Aliases] Congenital disorder of glycosylation, type Id; CDG-Id
8. ALG6-CDG
[Aliases] Congenital disorder of glycosylation, type Ic; CDG-Ic
9. ALG8-CDG
[Aliases] Congenital disorder of glycosylation, type Ih; CDG-Ih
10. ALG9-CDG
[Aliases] Congenital disorder of glycosylation, type IL; CDG-IL
11. Alpha-mannosidosis

To detailed Information about Diseases and their Causative Genes

Text searching

TOP

Search

Databases

76 GDGDB
36 GGDB

Disease Types by Metabolic Pathways

74 Congenital Disorders of Glycosylation (CDGs)
46 Lysosomal Storage Diseases (LSDs)

Manifestation

1 Abdominal Pain
1 Adrenal Gland Diseases
9 Airway Obstruction

Ontology Tree

46 Genetic disorders of glycan degradation
46 Genetic disorders of glycan degradation (by systems, signs and symptoms)
74 Genetic disorders of glycan synthesis (by systems, signs and symptoms)
70 Genetic disorders of glycan synthesis (new nomenclature)
57 Genetic disorders of glycan synthesis (traditional nomenclature)

(FW_LSD) Ontology for Disorders of Glycan Degradation (Lysosomal Storage Diseases); classified by pathway

Ontology Tree

- 46 Genetic disorders of glycan degradation ▶
- 46 Genetic disorders of glycan degradation (by systems, signs and symptoms) ▶
- 74 Genetic disorders of glycan synthesis (by systems, signs and symptoms) ▶
- 70 Genetic disorders of glycan synthesis (new nomenclature) ▶
- 57 Genetic disorders of glycan synthesis (traditional nomenclature) ▶

Open the “Genetic disorders of glycan syntethesis (by systems, signs and symptoms)”

Ontology Tree

- 1 (PhT_GD)Heart Defects, Congenital
- 10 (PhT_GD)Heart Diseases ▼
 - 1 (PhT_GD)Cardiomegaly
 - 6 (PhT_GD)Cardiomyopathies ▼
 - 1 (PhT_GD)Endomyocardial Fibrosis
 - 5 (others)
 - 1 (PhT_GD)Heart Failure
 - 1 (PhT_GD)Heart Valve Diseases
 - 2 (PhT_GD)Pericardial Effusion

Select the “(PhT_GD)*Heart Diseases”

Genetic Diseases of Glycan Metabolism

10 List of Diseases filtered from 120 originally (Reset All Filters)

sorted by: labels; then by... ☒ grouped as sorted

1. **ALG1-CDG**
[Aliases] Congenital disorder of glycosylation, type Ik; CDG-Ik
2. **ALG12-CDG**
[Aliases] Congenital disorder of glycosylation, type Ig; CDG-Ig
3. **ALG8-CDG**
[Aliases] Congenital disorder of glycosylation, type Ih; CDG-Ih
4. **ALG9-CDG**
[Aliases] Congenital disorder of glycosylation, type IL; CDG-IL
5. **Cardiomyopathy, dilated, 1X**
[Aliases] Dilated cardiomyopathy with mild or no proximal muscle weakness; Cardiomyopathy, dilated, 1X (CMD1X); FKT N-CDG (cong. muscular dystrophy spectrum)
6. **Ehlers-Danlos syndrome, type VI**
[Aliases] Nevo syndrome; Ehlers-Danlos syndrome, kyphoscoliotic type
7. **Fukuyama congenital muscular dystrophy**
[Aliases] Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 (MDDGA4); Fukuyama congenital muscular dystrophy (FCMD); FKTN-CDG (cong. muscular dystrophy spectrum)
8. **Muscular dystrophy, limb-girdle, type 2I**
[Aliases] Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 (MDDGC5); Limb-girdle muscular dystrophy type 2I (LGMD2I); FKRP-CDG (cong. muscular dystrophy spectrum)
9. **Muscular dystrophy, limb-girdle, type 2M**
[Aliases] Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4 (MDDGC4); Limb-girdle muscular dystrophy type 2M (LGMD2M); FKTN-CDG (cong. muscular dystrophy spectrum)
10. **PMM2-CDG**
[Aliases] Congenital disorder of glycosylation, type Ia; CDG-Ia; Phosphomannomutase 2 deficiency; Jaeken syndrome

Search

Database

- 10 GDGDB ☒
- 12 GGDB ☐

Disease Types by Metabolic Pathways

- 10 Congenital Disorders of Glycosylation (CDGs)

Manifestation

- 1 Anemia
- 1 Aneurysm
- 1 Aneurysm, Dissecting
- 1 Aneurysm, Ruptured
- 1 Asthma

Ontology Tree

- 11 (PhT_GD)Growth Disorders ▶
- 3 (PhT_GD)Hearing Loss
- 1 (PhT_GD)Heart Defects, Congenital
- 10 (PhT_GD)Heart Diseases ▼
 - 1 (PhT_GD)Cardiomegaly
 - 6 (PhT_GD)Cardiomyopathies ▼
 - 1 (PhT_GD)Endomyocardial Fibrosis

List of diseases that are related to “(PhT_GD)*Heart Diseases”

※(PhT_GD): Ontology for Disorders of Glycosylation; classified by phenotype

ACGG-DB Genetic Glyco-Diseases Ontology (GGDonto) and Glyco-Disease Genes DataBase (GDGDB)

Top	GGDonto	GDGDB	Disease Desc.	Gene Desc.	Protein Desc.	ENZYME
ALG12-CDG						
Genetic Glyco-Diseases Ontology (GGDonto)						
Concept UI	CON00349 (Tree)					
Name	ALG12-CDG					
Aliases	CDG-Ig Congenital disorder of glycosylation, type Ig					
Disease name: Preferred Term	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ig					
Disease name: Preferred Term ABBR	CDG1G					
Disease name: Synonyms (from OMIM and MeSH)	CDGIg Congenital disorder of glycosylation type 1G CDG Ig					
UMLS CUI	C2931001					
OMIM DATA: Gene	ALG12					
OMIM DATA: Gene Number	607144					

Menu items for selecting related information

Stanza:GGDonto: <div data-stanza=«<http://acgg.asia/db/diseases/ggdonto>» data-stanza-umls_cui="C2931001"></div>GDGDB: <div data-stanza=«<http://acgg.asia/db/diseases/gdgdb>» data-stanza-omim_id="607143"></div>

Disease(NCBI): <div data-stanza="http://acgg.asia/db/diseases/gmncbi" data-stanza-umls_cui="C2931001"></div>

Gene(NCBI): <div data-stanza="http://acgg.asia/db/diseases/gincbi" data-stanza-gene_id="79087"></div>

Protein(UniprotKB): <div data-stanza="http://acgg.asia/db/diseases/uniprot/uniprot" data-stanza-uniprot_id="Q9BV10"></div>

ENZYME(Swiss-Prot): <div data-stanza="http://acgg.asia/db/diseases/uniprot/enzyme" data-stanza-ec_number="2.4.1.260"></div>

※Stanza: TogoStanza (<http://www.togostanza.org/>) is a generic Web framework which enables the development of reusable Web components that are embeddable into any Web applications.

Genetic Glyco-Diseases Ontology (GGDonto)	
Concept UI	CON00349 (Tree)
Name	ALG12-CDG
Aliases	CDG-Ig Congenital disorder of glycosylation, type Ig
Disease name: Preferred Term	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ig
Disease name: Preferred Term ABBR	CDG1G
Disease name: Synonyms (from OMIM and MeSH)	CDG1g Congenital disorder of glycosylation type 1G CDG Ig
UMLS CUI	C2931001
OMIM DATA: Gene	ALG12
OMIM DATA: Gene Number	607144
OMIM DATA: Phenotype Number	607143
Symptoms, signs and abnormal clinical and laboratory findings	Blindness (Vision loss) Bone Diseases, Developmental Developmental Disabilities Psychomotor Disorders
References	<ul style="list-style-type: none"> H.H. Freeze and H. Schachter. Genetic Disorders of Glycosylation. 2009:585-600. Table 42.1 Genetic defects of glycan synthesis in humans H.H. Freeze. Genetic defects in the human glycome. Nat Rev Genet. 2006;7(7):537-51. Table 2. Human diseases caused by genetic defects in N-glycosylation pathways T. Hennet. Diseases of glycosylation beyond classical congenital disorders of glycosylation. Review. Biochim Biophys Acta. 2012;1820(9):1306-17. Table 1. Diseases of glycosylation
GGDDB (ID)	41
Link to the Glyco-Disease Genes Database	
GGDB (Gene Symbol)	ALG12
Link to the GlycoGene Database	

GGDonto

Disease Description from NCBI

OMIM phenotype MIM number	607143
OMIM disease name	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ig
Disease name (from UMLS sources)	Congenital disorder of glycosylation type 1G (MSH)
Symptoms, signs and abnormal clinical and laboratory findings (from MedGen sources)	Abnormality of immune system physiology (HPO) link Cognitive impairment (HPO) link Muscular hypotonia (HPO) link Genital abnormalities (OMIM) link

Information about Disease from NCBI

Gene Description from NCBI

OMIM Gene MIM number	607144
Gene name	ALG12, alpha-1,6-mannosyltransferase
Gene Symbol	ALG12
Alais Gene Name	asparagine-linked glycosylation 12 homolog (S. cerevisiae, alpha-1,6-mannosyltransferase); asparagine-linked glycosylation 12 homolog (yeast, alpha-1,6-mannosyltransferase); asparagine-linked glycosylation 12, alpha-1,6-mannosyltransferase homolog; asparagine-linked glycosylation protein 1.2 homolog; dol-P-Man dependent alpha-1,6-mannosyltransferase; dol-P-Man:Man(7)GlcNAc(2)-PP-Dol alpha-1,6-mannosyltransferase; dolichyl-P-Man:Man(7)GlcNAc(2)-PP-dolichol alpha-1,6-mannosyltransferase; dolichyl-P-Man:Man(7)GlcNAc(2)-PP-dolichyl-alpha-1,6-mannosyltransferase; dolichyl-P-mannose:Man-7-GlcNAc-2-PP-dolichyl-alpha-6-mannosyltransferase; mannosyltransferase ALG12 homolog; membrane protein SB87
Alias Gene Symbol	CDG1G; ECM39; PP14673; hALG12
Locus	22q13.33
Gene Id	79087

Information about Gene from NCBI

Protein Description from UniProtKB

UniProt Accession number	Q9BV10
UniProt Entry name	ALG12_HUMAN
Protein Recommended Name	Dol-P-Man:Man(7)GlcNAc(2)-PP-Dol alpha-1,6-mannosyltransferase
Protein Function	Adds the eighth mannose residue in an alpha-1,6 linkage onto the dolichol-PP-oligosaccharide precursor (dolichol-PP-Man(7)GlcNAc(2)) required for protein glycosylation.
Catalytic Activity	Dolichyl beta-D-mannosyl phosphate + D-Man-alpha-(1->2)-D-Man-alpha-(1->2)-D-Man-alpha-(1->3)-(D-Man-alpha-(1->2)-D-Man-alpha-(1->3))-D-Man-alpha-(1->6))-D-Man-beta-(1->4)-D-GlcNAc-diphosphodolichol = D-Man-alpha-(1->2)-D-Man-alpha-(1->2)-D-Man-alpha-(1->3)-(D-Man-alpha-(1->2)-D-Man-alpha-(1->3))-D-Man-alpha-(1->6))-D-Man-beta-(1->4)-D-GlcNAc-beta-(1->4)-D-GlcNAc-diphosphodolichol + dolichyl phosphate.

Information about Protein from UniProtKB

Enzyme Description from Swiss-Prot/ENZYME

EC Number	2.4.1.260
Accepted Name	Dolichyl-P-Man
Reaction catalysed	Dolichyl beta-D-mannosyl phosphate + D-Man-alpha-(1->2)-D-Man-alpha-(1->2)-D-Man-alpha-(1->3)-(D-Man-alpha-(1->2)-D-Man-alpha-(1->3))-D-Man-alpha-(1->6))-D-Man-beta-(1->4)-D-GlcNAc-diphosphodolichol = D-Man-alpha-(1->2)-D-Man-alpha-(1->2)-D-Man-alpha-(1->3)-(D-Man-alpha-(1->2)-D-Man-alpha-(1->3))-D-Man-alpha-(1->6))-D-Man-beta-(1->4)-D-GlcNAc-beta-(1->4)-D-GlcNAc-diphosphodolichol + dolichyl phosphate.

Information about Enzyme from Swiss-Prot

Glyco-Diseases Gene Database (GDGDB)

GDGDB ID	41
Disease names	CDG Ig (Congenital Disorder of Glycosylation type Ig)
Pathogenesis	Hypotonia, severe psychomotor retardation, seizures, feeding difficulties, facial dysmorphism, coagulopathy Glycoforum - Beyond Glycogenes, Dr. Hudson H. Freeze http://www.glycoforum.gr.jp/science/glycogenes/08/08E.html
OMIM phenotype	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ig; CDG1G
OMIM phenotype mim number	607143
Gene names	asparagine-linked glycosylation 12, alpha-1,6-mannosyltransferase homolog (S. cerevisiae)
Official gene symbol	ALG12
Alias gene symbol	ECM39; hALG12; MGC3136; PP14673; MGC111358
Locus	22q13.33
Link	NCBI Gene : http://www.ncbi.nlm.nih.gov/gene/79087 GGDB : http://jcgddb.jp/rcmg/ggdb/Homolog?cat=symbol&symbol=ALG12

GDGDB