Diseases associated with N-glycosylation of proteins

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

The contents of this document may be freely copied and distributed in any media, provided the authors, plus the institutions, are credited, as stated under the terms of Creative Commons Attribution 4.0 International (CC BY 4.0) License. For more information see our license.

This is just an excerpt of a full-length report for this pathway. To access the complete report, please download it at the Reactome Textbook.

12/11/2022

https://reactome.org


**Introduction**

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

**Literature references**


Reactome database release: 82

This document contains 18 pathways (see Table of Contents)

[https://reactome.org](https://reactome.org)
**Diseases associated with N-glycosylation of proteins**

**Stable identifier:** R-HSA-3781860

**Diseases:** congenital disorder of glycosylation

Congenital disorders of glycosylation (CDGs) are a group of autosomal recessive disorders caused by enzymatic defects in the synthesis and processing of asparagine (N)-linked glycans or oligosaccharides on glycoproteins. These glycoconjugates play critical roles in processes such as metabolism, cell recognition and adhesion, cell migration, protease resistance, host defense, and antigenicity. CDGs are divided into 2 main groups: type I CDGs comprise defects in the assembly of the dolichol lipid-linked oligosaccharide (LLO) chain and its transfer to the nascent protein, whereas type II CDGs comprise defects in the trimming and processing of protein-bound glycans (Marquardt & Denecke 2003, Grunewald et al. 2002, Hennet 2012, Cylwik et al. 2013).

**Literature references**


**Editions**

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-06-28</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective DPAGT1 causes CDG-1j, CMSTA2

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4549356

Diseases: congenital disorder of glycosylation type I

UDP-N-acetylg glucosamine--dolichyl-phosphate N-acetylglucosaminephosphotransferase (DPAGT1) catalyses the initial committed step in the biosynthesis of dolichyl pyrophosphate-oligosaccharides. Defects in DPAGT1 cause congenital disorder of glycosylation 1j (DPAGT1-CDG, previously known as CDG-1j; MIM:608093), a multisystem disorder characterised by under-glycosylated serum glycoproteins (Wu et al. 2003, Timal et al. 2012). Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. Defects in DPAGT1 can also cause myasthenic syndrome, congenital, with tubular aggregates, 2 (CMSTA2; MIM:614750), characterised by muscle weakness of mainly the proximal limb muscles, with tubular aggregates present on muscle biopsy. Sufferers find walking difficult and fall frequently. Younger sufferers show hypotonia and poor head control. A disorder of neuromuscular transmission is detected on electromyography (Belaya et al. 2012, Finlayson et al. 2013).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Author/Editor</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>2015-09-12</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>

https://reactome.org
Defective ALG14 causes ALG14-CMS

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-5633231

Diseases: congenital myasthenic syndrome

UDP-N-acetylglucosamine transferase subunit ALG14 homolog (ALG14) forms a complex with ALG13 protein and is required for the addition of the second N-acetylglucosamine (GlcNAc) to the lipid linked oligosaccharide (LLO) intermediate (GlcNAcDOLDP) (Gao et al. 2005). Defects in ALG14 can cause congenital myasthenic syndrome (ALG14-CMS), which is due to a defect in neuromuscular signal transmission (Cossins et al. 2013). The most commonly affected muscles include proximal limb muscles. Mutations causing ALG14-CMS include p.P65L and p.R104* (Cossins et al. 2013).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
</tbody>
</table>
Chitobiosylphosphodolichol beta-mannosyltransferase (ALG1) normally transfers a mannose moiety to the lipid-linked oligosaccharide (LLO aka N-glycan precursor) which is required for subsequent N-glycosylation of proteins. Defects in ALG1 can cause congenital disorder of glycosylation 1k (ALG1-CDG, previously known as CDG1k; MIM:608540), a multisystem disorder characterised by under-glycosylated serum glycoproteins. CDG type 1 diseases result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. Compared to other CDGs, ALG1-CDG has a very severe phenotype, which can result in an early death (Schwarz et al. 2004, Kranz et al. 2004, Dupre et al. 2010).

**Literature references**


Defective ALG2 causes CDG-1i

**Location:** Diseases associated with N-glycosylation of proteins

**Stable identifier:** R-HSA-4549349

**Diseases:** congenital disorder of glycosylation type I, congenital myasthenic syndrome

Alpha 1,3/1,6 mannosyltransferase ALG2 (ALG2) is a bifunctional mannosyltransferase normally transfers a mannose moiety to the lipid linked oligosaccharide (LLO aka N glycan precursor) which is required for subsequent N glycosylation of proteins. Defects in ALG2 can cause congenital disorder of glycosylation 1i (ALG2-CDG, previously known as CDG1i; MIM:607906), a multisystem disorder characterised by under glycosylated serum glycoproteins. CDG type 1 diseases result in a wide phenotypic spectrum, from poor neurological development, psychomotor retardation and dysmorphic features to hypotonia, coagulation abnormalities and immunodeficiency (Thiel et al. 2003). Defect in ALG2 can also cause congenital myasthenic syndrome (ALG2-CMS), which is due to a defect in neuromuscular signal transmission (Cossins et al. 2013). The most commonly affected muscles include proximal limb muscles. Mutations causing ALG2-CMS include p.V68G and p.72_75delinsSPR (Cossins et al. 2013).

**Literature references**


**Editions**

<table>
<thead>
<tr>
<th>Date</th>
<th>Description</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-09-12</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>

https://reactome.org
Defective ALG11 causes CDG-1p

**Location:** Diseases associated with N-glycosylation of proteins

**Stable identifier:** R-HSA-4551295

**Diseases:** congenital disorder of glycosylation type I

GDP-Man:Man(3)GlcNAc(2)-PP-Dol alpha-1,2-mannosyltransferase (ALG11) transfers the fourth and fifth mannoses (Man) to the N-glycan precursor in an alpha-1,2 orientation. These additions are the last two on the cytosolic side of the ER membrane before the N-glycan is flipped to the luminal side of the membrane. Recently discovered defects in ALG11 have been linked to congenital disorder of glycosylation, type 1p (ALG11-CDG, CGD1p) (Rind et al. 2010, Thiel et al. 2012). The disease is a multi-system disorder characterised by under-glycosylated serum glycoproteins. Early-onset developmental retardation, dysmorphic features, hypotonia, coagulation disorders and immunodeficiency are reported features of this disorder (Rind et al. 2010, Thiel et al. 2012).

**Literature references**


**Editions**

<table>
<thead>
<tr>
<th>Edition</th>
<th>Author(s)</th>
<th>Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-09-13</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective RFT1 causes CDG-1n

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4570571

Diseases: congenital disorder of glycosylation type I

The N-glycan precursor is flipped across the ER membrane, moving it from the cytosolic side to the ER lumenal side. The exact mechanism of this translocation is not well understood but protein RFT1 homolog (RFT1) is known to be involved (Helenius et al. 2002). Defects in RFT1 are associated with congenital disorder of glycosylation 1n (RFT1-CDG, CDG-1n). The disease is a multi-system disorder characterised by under-glycosylated serum glycoproteins. Early-onset developmental retardation, dysmorphic features, hypotonia, coagulation disorders and immunodeficiency are reported features of this disorder (Haeuptle et al. 2008).

Literature references


Editions

<table>
<thead>
<tr>
<th>Edition</th>
<th>Authored, Edited</th>
<th>Reviewed</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-09-23</td>
<td>Jassal, B.</td>
<td></td>
</tr>
<tr>
<td>2014-10-31</td>
<td></td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective ALG3 causes CDG-1d

**Location:** Diseases associated with N-glycosylation of proteins

**Stable identifier:** R-HSA-4720475

**Diseases:** congenital disorder of glycosylation type I

Dol-P-Man:Man(5)GlcNAc(2)-PP-Dol alpha-1,3-mannosyltransferase (ALG3) adds the sixth mannose (although the first to be derived from dolichyl-phosphate-mannose, DOLPman) to the lipid-linked oligosaccharide (LLO) intermediate GlcNAc(2) Man(5) (PPDol)1 (Korner et al. 1999). Defects in ALG3 are associated with congenital disorder of glycosylation 1d (ALG3-CDG, CDG1d; MIM:601110), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins. CDG type 1 diseases result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency (Sun et al. 2005).

**Literature references**


**Editions**

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-10-21</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective ALG9 causes CDG-1l

**Location:** Diseases associated with N-glycosylation of proteins

**Stable identifier:** R-HSA-4720454

**Diseases:** congenital disorder of glycosylation type I

Alpha-1,2-mannosyltransferase ALG9 (ALG9) normally catalyses the transfer of mannose to the lipid-linked oligosaccharide (LLO) precursor. It adds the 7th and 9th mannose moieties to LLO. Defects in ALG9 are associated with congenital disorder of glycosylation 1l (ALG9-CDG, CDG1l; MIM:608776), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins. CDG type 1 diseases result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency (Frank et al. 2004, Weinstein et al. 2005). The LLO profile showed accumulation of (GlcNAc)2 (Man)6 (PP-Dol)1 and (GlcNAc)2 (Man)8 (PP-Dol)1 fragments, suggesting a defect in ALG9 and correlating with the normal function of ALG9 in adding the 7th and 9th mannose moieties (Frank et al. 2004).

**Literature references**


**Editions**

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-10-21</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective MPDU1 causes CDG-1f

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4687000

Diseases: congenital disorder of glycosylation type I

Mannose-P-dolichol utilisation defect 1 protein (MPDU1) is required for the efficient utilisation of the mannose donor dolichyl-phospho-mannose (DOLPman) in the synthesis of both lipid-linked oligosaccharides (LLOs) and glycosylphosphatidylinositol. Defects in MPDU1 can cause congenital disorder of glycosylation 1f (MPDU1-CDG, CDG-1f; MIM:609180), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins. CDG type 1 diseases result in a wide phenotypic spectrum, such as poor neurological development, psychomotor retardation, dysmorphic features, hypotonia, coagulation abnormalities and immunodeficiency. In this condition, DOLPman is no longer utilised in transferase reactions extending LLOs, even as substrate levels and transferase enzyme activities appear normal (Anand et al. 2001, Schenk et al. 2001).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-10-15</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective ALG12 causes CDG-1g

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4720489

Diseases: congenital disorder of glycosylation type I

Dol-P-Man:Man(7)GlcNAc(2)-PP-Dol alpha-1,6-mannosyltransferase (ALG12) (Chantret et al. 2002) normally transfers the 8th mannose moiety to the lipid-linked oligosaccharide (LLO aka N-glycan precursor) which is required for subsequent N-glycosylation of proteins. Defects in ALG12 are associated with congenital disorder of glycosylation 1g (ALG12-CDG, CDG1g; MIM:607143), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins (Chantret et al. 2002, Grubenmann et al. 2002). CDG type 1 diseases result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency.

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Author(s)</th>
<th>Editions</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-10-21</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective ALG6 causes CDG-1c

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4724289

Diseases: congenital disorder of glycosylation type I

Dolichyl pyrophosphate Man9GlcNAc2 α-1,3-glucosyltransferase (ALG6) normally adds the first glucose moiety to the lipid-linked oligosaccharide precursor (LLO aka N-glycan precursor) which is required for subsequent N-glycosylation of proteins (Imbach et al. 1999). Defects in ALG6 can cause congenital disorder of glycosylation 1c (ALG6-CDG, CDG-1c; MIM:603147), a multisystem disorder characterised by under-glycosylated serum glycoproteins (Imbach et al. 1999, Imbach et al. 2000, Westphal et al. 2000, Sun et al. 2005). ALG6 deficiency is accompanied by an accumulation of the N-glycan precursor (GlcNAc)2(Man)9(PP-Dol)1 and is the second most common CDG disease subtype after PMM2-CDG (CDG-1a) (Imbach et al. 1999). CDG type 1 diseases result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency.

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-10-23</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>

https://reactome.org
Defective ALG8 causes CDG-1h

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4724325

Diseases: congenital disorder of glycosylation type I

The probable dolichyl pyrophosphate Glc1Man9GlcNAc2 alpha-1,3-glucosyltransferase (ALG8) (Stanchi et al. 2001, Chantret et al. 2003) normally adds the second glucose moiety to the lipid-linked oligosaccharide precursor (LLO aka N-glycan precursor) which is required for subsequent N-glycosylation of proteins. Defects in ALG8 can cause congenital disorder of glycosylation 1h (ALG8-CDG, CDG-1h; MIM:608104), a multisystem disorder characterised by under-glycosylated serum glycoproteins (Chantret et al. 2003, Schollen et al. 2004). ALG8 deficiency is accompanied by an accumulation of the N-glycan precursor (Glc)1 (GlcNAc)2 (Man)9 (PP-Dol)1. CDG type 1 diseases result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency.

Literature references


Editions

<table>
<thead>
<tr>
<th>Editions</th>
<th>Authored, Edited</th>
<th>Reviewed</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-10-23</td>
<td>Jassal, B.</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective MGAT2 causes CDG-2a

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4793952

Diseases: congenital disorder of glycosylation type II

Alpha-1,6-mannosyl-glycoprotein 2-beta-N-acetylglucosaminyltransferase (MGAT2) normally catalyses the transfer of a GlcNAc moiety onto the alpha-1,6 mannose of an alpha-1,4 branch of oligomannose N-glycans to form complex N-glycans (Tan et al. 1995). Defects in MGAT2 are associated with congenital disorder of glycosylation type IIa (MGAT2-CDG, CDG-2a; MIM:212066), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins (Tan et al. 1996, Cormier-Daire et al. 2000, Alkuraya 2010, Alazami et al. 2012). Type II CDGs refer to defects in the trimming and processing of protein-bound glycans.

Literature references


Editions

<table>
<thead>
<tr>
<th>Edition</th>
<th>Date</th>
<th>Authors/Editors</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-10-29</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>

https://reactome.org
Defective MOGS causes CDG-2b

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4793954

Diseases: congenital disorder of glycosylation type II

After the lipid-linked oligosaccharide (LLO) precursor is attached to the protein, the outer alpha-1,2-linked glucose is removed by by mannosyl-oligosaccharide glucosidase (MOGS). This is a mandatory step for protein folding control and glycan extension. Defects in MOGS are associated with congenital disorder of glycosylation type IIb (CDGIIb), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins (De Praeter et al. 2000, Voelker et al. 2002). Type II CDGs refer to defects in the trimming and processing of protein-bound glycans.

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-07-29</td>
<td>Authored</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2013-10-29</td>
<td>Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective B4GALT1 causes CDG-2d

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4793953

Diseases: congenital disorder of glycosylation type II

Congenital disorders of glycosylation (CDG, previously called carbohydrate-deficient glycoprotein syndromes, CDGSs), are a group of hereditary multisystem disorders. They are characterized biochemically by hypoglycosylation of glycoproteins, diagnosed by isoelectric focusing (IEF) of serum transferrin. There are two types of CDG, types I and II. Type I CDG has defects in the assembly of lipid-linked oligosaccharides or their transfer onto nascent glycoproteins, whereas type II CDG comprises defects of trimming, elongation, and processing of protein-bound glycans. Clinical symptoms are dominated by severe psychomotor and mental retardation, as well as blood coagulation abnormalities (Jaeken 2013). B4GALT1-CDG (CDG type IId) is a multisystem disease, characterized by dysmorphic features, hydrocephalus, hypotonia and blood clotting abnormalities (Hansske et al. 2002).

Literature references


Editions

<table>
<thead>
<tr>
<th>Edition</th>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-07-29</td>
<td></td>
<td></td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2013-10-29</td>
<td></td>
<td>Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td></td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective MAN1B1 causes MRT15

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4793950

Diseases: non-specific X-linked mental retardation

Endoplasmic reticulum mannosyl-oligosaccharide 1,2-alpha-mannosidase (MAN1B1) normally trims single mannose residues from misfolded glycoproteins, targeting them for degradation and thus providing a quality control process for N-glycosylated proteins. Defects in MAN1B1 can cause mental retardation, autosomal recessive 15 (MRT15; MIM:614202), a disorder resulting in nonsyndromic moderate to severe mental retardation. It is characterised by significantly below average intellectual functioning associated with impaired adaptive behaviour during the developmental period (Rafiq et al. 2010, Rafiq et al. 2011).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-07-29</td>
<td>Authored</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2013-10-29</td>
<td>Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2014-10-31</td>
<td>Reviewed</td>
<td>Belaya, K.</td>
</tr>
</tbody>
</table>
Defective NEU1 causes sialidosis

Location: Diseases associated with N-glycosylation of proteins

Stable identifier: R-HSA-4341670

Diseases: lysosomal storage disease

Sialidases have important roles in the degradation of glycoconjugates by removing terminal sialic acid residues.

Defects in sialidase 1 (NEU1) cause sialidosis, a lysosomal storage disease characterised by the progressive lysosomal storage of sialidated glycopeptides and oligosaccharides and the accumulation and excretion of N-acetylneuraminic acid (Neu5Ac) covalently-linked ('bound') glycoconjugates (Lowden & O'Brien 1979). The sialidoses are distinct from the sialurias in which there is storage and excretion of 'free' Neu5Ac. Sialidosis manifests into types I and II forms. Type I is the milder form, also known as the 'normosomatic' type or the cherry red spot-myoclonus syndrome. Sialidosis type II is the more severe form with an earlier onset, and is also known as the 'dysmorphic' type.

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
<th>Edition</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013-08-21</td>
<td>Authored, Edited</td>
<td>Jassal,B.</td>
<td></td>
</tr>
<tr>
<td>2015-04-30</td>
<td>Reviewed</td>
<td>Spillmann,D.</td>
<td></td>
</tr>
</tbody>
</table>

https://reactome.org
Table of Contents

Introduction 1

- Diseases associated with N-glycosylation of proteins 2
  - Defective DPAGT1 causes CDG-1j, CMSTA2 3
  - Defective ALG14 causes ALG14-CMS 4
  - Defective ALG1 causes CDG-1k 5
  - Defective ALG2 causes CDG-1i 6
  - Defective ALG11 causes CDG-1p 7
  - Defective RFT1 causes CDG-1n 8
  - Defective ALG3 causes CDG-1d 9
  - Defective ALG9 causes CDG-1l 10
  - Defective MPDU1 causes CDG-1f 11
  - Defective ALG12 causes CDG-1g 12
  - Defective ALG6 causes CDG-1c 13
  - Defective ALG8 causes CDG-1h 14
  - Defective MGAT2 causes CDG-2a 15
  - Defective MOGS causes CDG-2b 16
  - Defective B4GALT1 causes CDG-2d 17
  - Defective MAN1B1 causes MRT15 18
  - Defective NEU1 causes sialidosis 19

Table of Contents 20