Diseases of glycosylation

Belaya, K., Hansen, L., Jassal, B., Joshi, HJ., Spillmann, D., Timson, DJ.
**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.

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**Literature references**


Reactome database release: 67

This document contains 5 pathways (see Table of Contents)
Diseases of glycosylation

Stable identifier: R-HSA-3781865

Diseases: congenital disorder of glycosylation

Diseases of glycosylation, usually referred to as congenital disorders of glycosylation (CDG), are rare inherited disorders ascribing defects of nucleotide-sugar biosynthesis and transport, glycosyltransfer events and vesicular transport. Most CDGs cause neurological impairment ranging from severe psychomotor retardation to mild intellectual disability. Defects in N-glycosylation are the main cause of CDGs (Marquardt & Denecke 2003, Grunewald et al. 2002, Hennet 2012, Goreta et al. 2012) and can be identified by a characteristic abnormal isoelectric focusing profile of plasma transferrin (Jaeken et al. 1984, Stibler & Jaeken 1990). Disorders of O-glycosylation, glycosaminoglycan and glycolipid metabolism have recently been discovered and, together with N-glycosylation, represent the major pathways affected by glycan biosynthetic disorders (Freeze 2006, Jaeken 2011). As the number of these disorders has increased, nomenclature has been simplified so that now, the name of the mutant gene is followed by the abbreviation CDG (Jaeken et al. 2009). Effective therapies for most types of CDGs are so far not available (Thiel & Korner 2013).

Literature references


Editions

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Diseases associated with glycosaminoglycan metabolism

**Location:** Diseases of glycosylation

**Stable identifier:** R-HSA-3560782

**Diseases:** congenital disorder of glycosylation

A number of genetic disorders are caused by mutations in the genes encoding glycosyltransferases and sulfotransferases, enzymes responsible for the synthesis of glycosaminoglycans (GAGs) as well as hexosaminidase degradation of GAGs (Mizumoto et al. 2013).

**Literature references**


**Editions**

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Diseases associated with N-glycosylation of proteins

Location: Diseases of glycosylation

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

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Diseases associated with O-glycosylation of proteins

Location: Diseases of glycosylation

Stable identifier: R-HSA-3906995

Diseases: congenital disorder of glycosylation

Glycosylation is the most abundant modification of proteins, variations of which occur in all living cells. Glycosylation can be further categorized into N-linked (where the oligosaccharide is conjugated to Asparagine residues) and O-linked glycosylation (where the oligosaccharide is conjugated to Serine, Threonine and possibly Tyrosine residues). Within the family of O-linked glycosylation, the oligosaccharides attached can be further categorized according to their reducing end residue: GalNAc (often described as mucin-type, due to the abundance of this type of glycosylation on mucins), Mannose and Fucose. This section reviews currently known congenital disorders of glycosylation associated with defects of protein O-glycosylation (Cylwik et al. 2013, Freeze et al. 2014).

Literature references


Editions

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Diseases associated with glycosylation precursor biosynthesis

Location: Diseases of glycosylation

Stable identifier: R-HSA-5609975

Diseases: congenital disorder of glycosylation

Glycosylation diseases associated with the enzymes that mediate the biosynthesis of glycosylation precursors are curated in this section (Jaeken & Matthijs 2007, Freeze et al. 2015).

Literature references


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