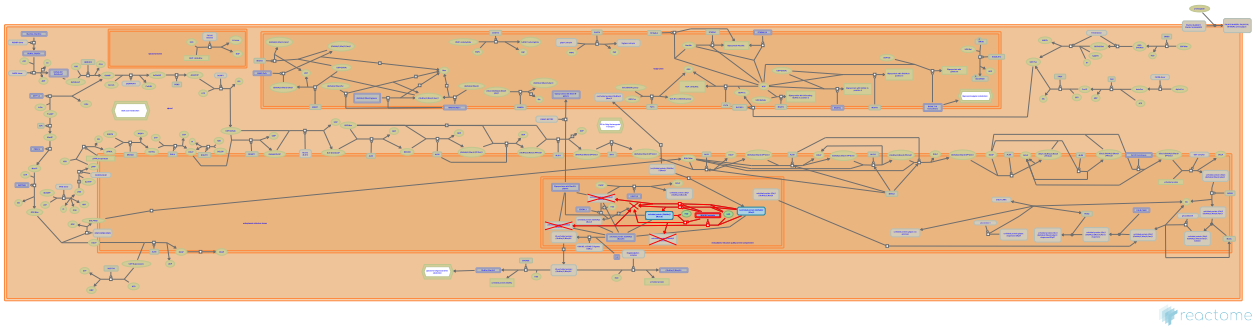
Defective MAN1B1 causes MRT15



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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

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics, 18*, 142. ↗

Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics, 33*, 3461-3467. ↗

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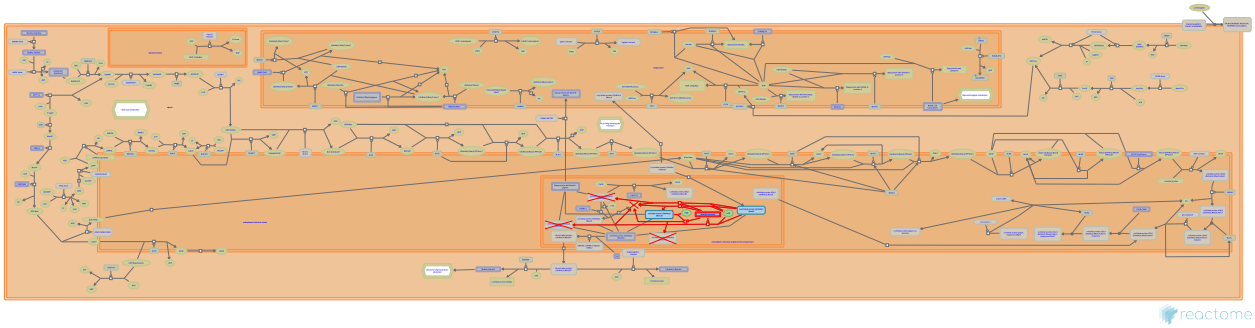
Defective MAN1B1 causes MRT15 ↗

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-glycoyslated 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 adaptative behaviour during the developmental period (Rafiq et al. 2010, Rafiq et al. 2011).

Literature references

Ullmann, R., Mahmood, K., Ramiah, A., Ropers, HH., Ishak, GE., Garshasbi, M. et al. (2011). Mutations in the alpha 1,2-mannosidase gene, MAN1B1, cause autosomal-recessive intellectual disability. *Am. J. Hum. Genet., 89*, 176-82. ↗

Shaheen, N., Noor, A., Scherer, SW., Amin-ud-Din, M., Ansar, M., Khan, MA. et al. (2010). Mapping of three novel loci for non-syndromic autosomal recessive mental retardation (NS-ARMR) in consanguineous families from Pakistan. *Clin. Genet., 78*, 478-83. ↗

Editions

2013-07-29	Authored	Jassal, B.
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Defective MAN1B1 does not hydrolyse 1,2-linked mannose (a branch) ↗

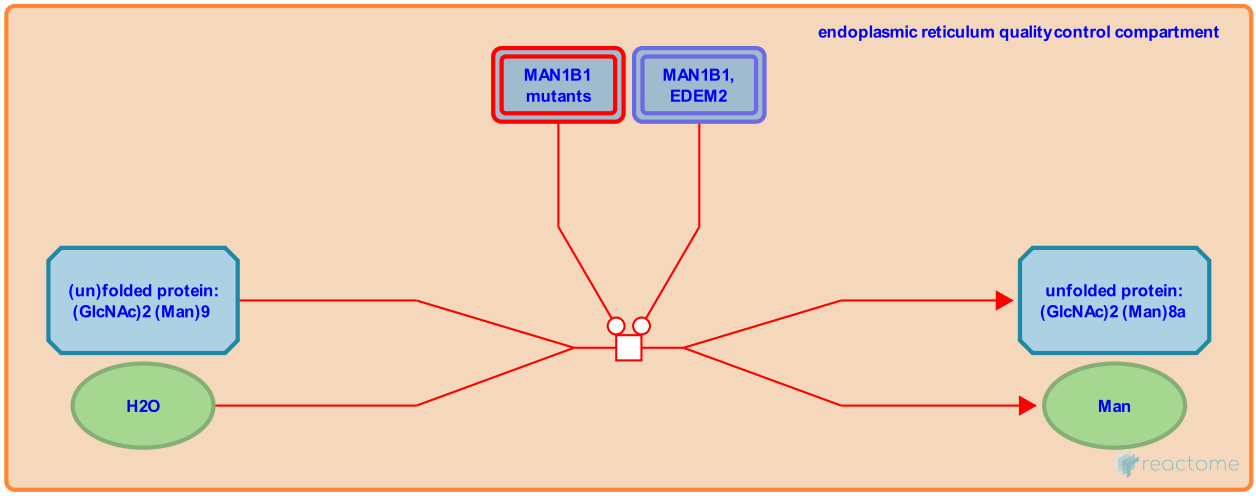
Location: Defective MAN1B1 causes MRT15

Stable identifier: R-HSA-4793949

Type: transition

Compartments: endoplasmic reticulum quality control compartment

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 adaptative behaviour during the developmental period (Rafiq et al. 2010, Rafiq et al. 2011). Mutations that can cause MRT15 are E397K, W473* and R334C (Rafiq et al. 2010, Rafiq et al. 2011).

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Defective MAN1B1 does not hydrolyse a second 1,2-linked mannose (a branch) ↗

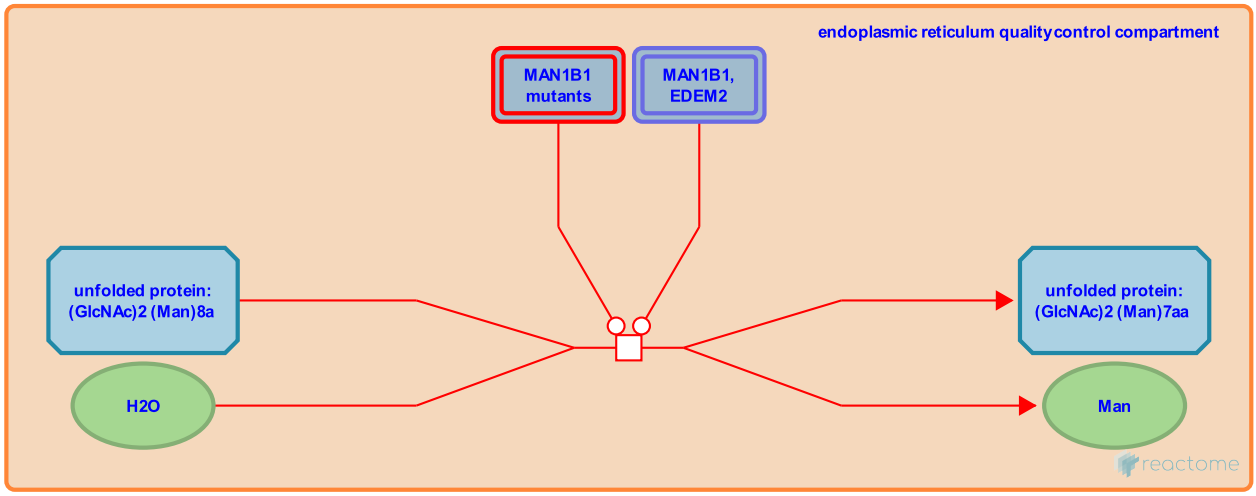
Location: Defective MAN1B1 causes MRT15

Stable identifier: R-HSA-9036008

Type: transition

Compartments: endoplasmic reticulum quality control compartment

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 adaptative behaviour during the developmental period (Rafiq et al. 2010, Rafiq et al. 2011). Mutations that can cause MRT15 are E397K, W473* and R334C (Rafiq et al. 2010, Rafiq et al. 2011).

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Defective MAN1B1 does not hydrolyse 1,2-linked mannose (b branch) ↗

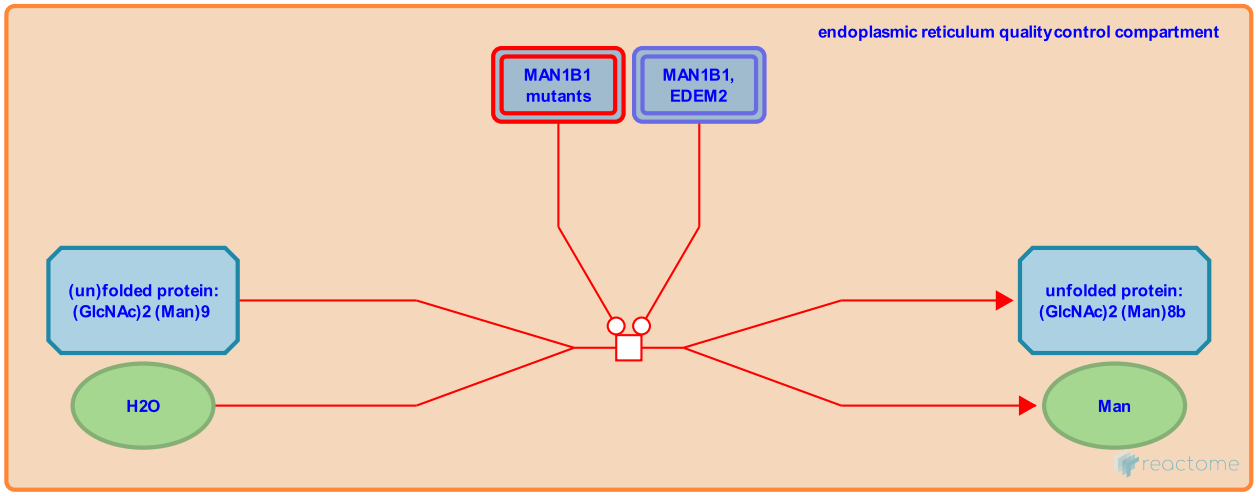
Location: Defective MAN1B1 causes MRT15

Stable identifier: R-HSA-9036011

Type: transition

Compartments: endoplasmic reticulum quality control compartment

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 adaptative behaviour during the developmental period (Rafiq et al. 2010, Rafiq et al. 2011). Mutations that can cause MRT15 are E397K, W473* and R334C (Rafiq et al. 2010, Rafiq et al. 2011).

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Defective MAN1B1 does not hydrolyse 1,2-linked mannose (c branch) ↗

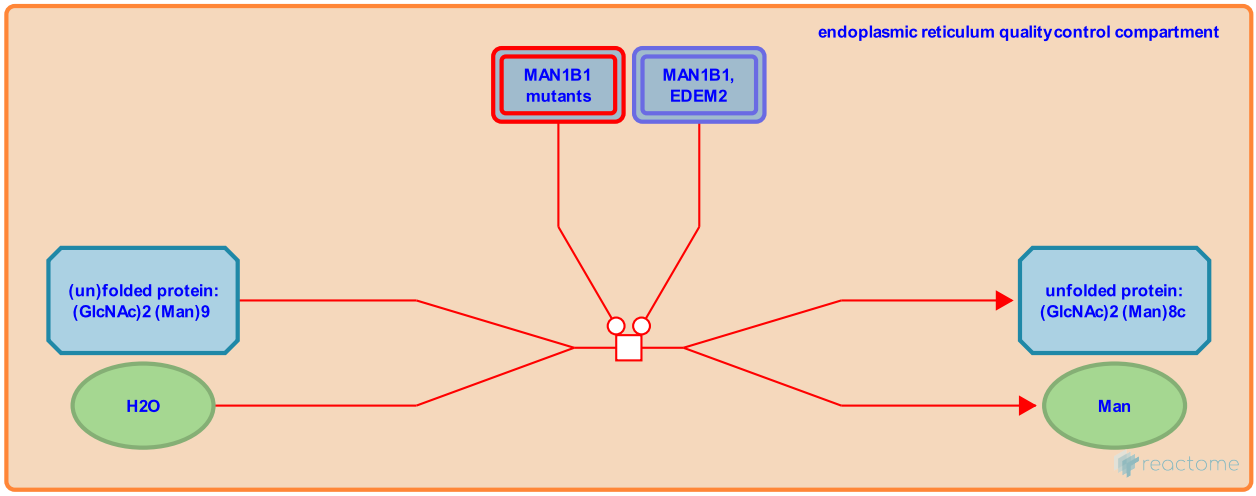
Location: Defective MAN1B1 causes MRT15

Stable identifier: R-HSA-9036012

Type: transition

Compartments: endoplasmic reticulum quality control compartment

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 adaptative behaviour during the developmental period (Rafiq et al. 2010, Rafiq et al. 2011). Mutations that can cause MRT15 are E397K, W473* and R334C (Rafiq et al. 2010, Rafiq et al. 2011).

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