

## MAN2B1 gene

mannosidase alpha class 2B member 1

### Normal Function

The *MAN2B1* gene provides instructions for making the enzyme alpha-mannosidase. This enzyme works in the lysosomes, which are compartments that digest and recycle materials in the cell. Within lysosomes, the enzyme helps break down complexes of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins). In particular, alpha-mannosidase helps break down oligosaccharides containing a sugar molecule called mannose.

### Health Conditions Related to Genetic Changes

#### Alpha-mannosidosis

More than 120 mutations in the *MAN2B1* gene have been identified in people with alpha-mannosidosis, a rare inherited disorder that causes problems in many organs and tissues of the body. Affected individuals may have intellectual disability, distinctive facial features, and skeletal abnormalities. Some of the *MAN2B1* gene mutations that cause alpha-mannosidosis change one protein building block (amino acid) in the alpha-mannosidase enzyme. Other mutations result in an abnormally shortened enzyme, or cause the enzyme to be pieced together incorrectly.

These mutations interfere with the ability of the alpha-mannosidase enzyme to perform its role in breaking down mannose-containing oligosaccharides. These oligosaccharides accumulate in the lysosomes and cause the cells to malfunction and eventually die. Tissues and organs are damaged by the abnormal accumulation of oligosaccharides and the resulting cell death, leading to the characteristic features of alpha-mannosidosis.

### Other Names for This Gene

- LAMAN
- lysosomal acid alpha-mannosidase
- MA2B1\_HUMAN
- MANB
- mannosidase, alpha B, lysosomal

- mannosidase, alpha, class 2B, member 1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of MAN2B1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4125\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4125[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28MAN2B1%5BTIAB%5D%29+OR+%28%28LAMAN%5BTIAB%5D%29+OR+%28lysosomal+acid+alpha-mannosidase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

## Catalog of Genes and Diseases from OMIM

- MANNOSIDASE, ALPHA, CLASS 2B, MEMBER 1; MAN2B1 (<https://omim.org/entry/609458>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4125>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MAN2B1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MAN2B1[gene]))

## References

- Essentials of Glycobiology (second edition, 2009): Glycoprotein Degradation (<https://www.ncbi.nlm.nih.gov/books/NBK1934/#ch41.s3>)
- Hansen G, Berg T, Riise Stensland HM, Heikinheimo P, Klenow H, Evjen G, Nilssen O, Tollersrud OK. Intracellular transport of human lysosomal alpha-mannosidase and alpha-mannosidosis-related mutants. *Biochem J*. 2004 Jul 15; 381(Pt 2):537-46. doi: 10.1042/BJ20031499. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15035660>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1133862/>)
- Kuokkanen E, Riise Stensland HM, Smith W, Kjeldsen Buvang E, Van Nguyen L, Nilssen O, Heikinheimo P. Molecular and cellular characterization of novel alpha-mannosidosis mutations. *Hum Mol Genet*. 2011 Jul 1; 20(13):2651-61. doi:10.1093/hmg/ddr167. Epub 2011 Apr 19. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21505070>)
- Malm D, Nilssen O. Alpha-mannosidosis. *Orphanet J Rare Dis*. 2008 Jul 23; 3:21. doi:

10.1186/1750-1172-3-21. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18651971>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2515294/>)

- Pittis MG, Montalvo AL, Heikinheimo P, Sbaragli M, Balducci C, Persichetti E, Van Maldergem L, Filocamo M, Bembi B, Beccari T. Functional characterization of four novel MAN2B1 mutations causing juvenile onset alpha-mannosidosis. Clin Chim Acta. 2007 Jan;375(1-2):136-9. doi: 10.1016/j.cca.2006.06.034. Epub 2006 Jul 6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16919251>)
- Sbaragli M, Bibi L, Pittis MG, Balducci C, Heikinheimo P, Ricci R, Antuzzi D, Parini R, Spaccini L, Bembi B, Beccari T. Identification and characterization of five novel MAN2B1 mutations in Italian patients with alpha-mannosidosis. Hum Mutat. 2005 Mar;25(3):320. doi: 10.1002/humu.9310. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15712269>)
- Sun H, Wolfe JH. Recent progress in lysosomal alpha-mannosidase and its deficiency. Exp Mol Med. 2001 Mar 31;33(1):1-7. doi: 10.1038/emm.2001.1. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11322479>)

## Genomic Location

The *MAN2B1* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

**Last updated May 1, 2014**