



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

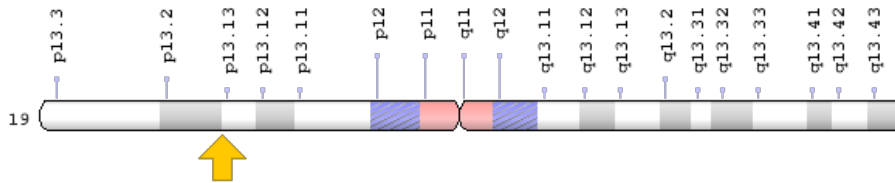
More than 120 mutations in the *MAN2B1* gene have been identified in people with alpha-mannosidosis, a rare inherited disorder that causes problems in many organs and tissues of the body. Affected individuals may have intellectual disability, distinctive facial features, and skeletal abnormalities. Some 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.

Chromosomal Location

Cytogenetic Location: 19p13.13, which is the short (p) arm of chromosome 19 at position 13.13

Molecular Location: base pairs 12,646,508 to 12,666,777 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Glycoprotein Degradation
<https://www.ncbi.nlm.nih.gov/books/NBK1934/#ch41.s3>

Clinical Information from GeneReviews

- Alpha-Mannosidosis
<https://www.ncbi.nlm.nih.gov/books/NBK1396>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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>

Catalog of Genes and Diseases from OMIM

- MANNOSIDASE, ALPHA, CLASS 2B, MEMBER 1
<http://omim.org/entry/609458>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MAN2B1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:6826
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:4125>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4125>
- UniProt
<https://www.uniprot.org/uniprot/O00754>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/MAN2B1>

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