MANBA gene
mannosidase beta

Normal Function
The MANBA gene provides instructions for making the enzyme beta-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). Beta-mannosidase is involved in the last step of this process, helping to break down complexes of two sugar molecules (disaccharides) containing a sugar molecule called mannose.

Health Conditions Related to Genetic Changes
Beta-mannosidosis
Approximately 12 mutations that cause beta-mannosidosis have been identified in the MANBA gene. The mutations result in a beta-mannosidase enzyme with little or no activity, and interfere with the ability of the enzyme to perform its role in breaking down mannose-containing disaccharides. These disaccharides gradually accumulate in the lysosomes and cause cells to malfunction, resulting in the signs and symptoms of beta-mannosidosis.

Chromosomal Location
Cytogenetic Location: 4q24, which is the long (q) arm of chromosome 4 at position 24
Molecular Location: base pairs 102,630,770 to 102,760,968 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

- MANB1
- MANBA_HUMAN
- mannanase
- mannase
- mannosidase, beta A, lysosomal

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (1999): Glycoprotein Degradation
  https://www.ncbi.nlm.nih.gov/books/NBK20729/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28MANBA%5BTIAB%5D%29+OR+%28MANB1%5BTIAB%5D%29+OR+%28mannase%5BTIAB%5D%29+OR+%28mannanase%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- MANNOSIDASE, BETA A, LYSOSOMAL
  http://omim.org/entry/609489

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MANBA.html
- ClinVar
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4126
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/O00462
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9384606

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12468273

- Essentials of Glycobiology (1999): Glycoprotein Degradation 
  https://www.ncbi.nlm.nih.gov/books/NBK20729/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16904924

- OMIM: MANNOSIDASE, BETA A, LYSOSOMAL 
  http://omim.org/entry/609489

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17420068

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12890191

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16377659

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