ANK1 gene
ankyrin 1

Normal Function

The ANK1 gene provides instruction for making a protein called ankyrin-1. This protein is primarily active (expressed) in red blood cells, but it is also found in muscle and brain cells. In red blood cells, ankyrin-1 is located at the cell membrane, where it attaches (binds) to other membrane proteins. The binding of membrane proteins to one another maintains the stability and structure of red blood cells but also allows for their flexibility. The proteins allow the cell to change shape without breaking when passing through narrow blood vessels.

In muscle and brain cells, ankyrin-1 performs similar functions, binding to other membrane proteins to play a role in cell stability, cell movement, and other cell functions.

Health Conditions Related to Genetic Changes

Hereditary spherocytosis

At least 55 mutations in the ANK1 gene have been found to cause hereditary spherocytosis. Some of these mutations delete small pieces of genetic material, and others change single DNA building blocks (nucleotides) in the ANK1 gene. These mutations lead to the production of an ankyrin-1 protein that does not function normally and does not bind to other proteins within the red blood cell membrane. A lack of normal ankyrin-1 at the cell membrane also leads to a lack of another protein called spectrin because ankyrin-1 is not available to bind to spectrin. The shortage (deficiency) of these two proteins interferes with the structure and flexibility of the red blood cell membrane, causing red blood cells to be misshapen. These misshapen cells, called spherocytes, are removed from circulation and taken to the spleen for destruction. The shortage of red blood cells in circulation and the abundance of cells in the spleen are responsible for the signs and symptoms of hereditary spherocytosis.
Chromosomal Location

Cytogenetic Location: 8p11.21, which is the short (p) arm of chromosome 8 at position 11.21

Molecular Location: base pairs 41,653,225 to 41,896,741 on chromosome 8 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

Other Names for This Gene

• ANK
• ANK-1
• ANK1_HUMAN
• ankyrin-1
• ankyrin 1, erythrocytic
• ankyrin-R
• erythrocyte ankyrin

Additional Information & Resources

Educational Resources

• Molecular Cell Biology (fourth edition, 2000): Erythrocyte Cytoskeleton
  https://www.ncbi.nlm.nih.gov/books/NBK21493/

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ANK1%5BTIAB%5D%29+OR+%28ankyrin-R%29+OR+%28ankyrin-1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- ANKYRIN 1
  http://omim.org/entry/612641

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ANK1.html

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ANK1%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:286

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P16157

Sources for This Summary

- OMIM: ANKYRIN 1
  http://omim.org/entry/612641

- Bennett V, Healy J. Organizing the fluid membrane bilayer: diseases linked to spectrin and ankyrin.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18083066

- Eber S, Lux SE. Hereditary spherocytosis--defects in proteins that connect the membrane skeleton to the lipid bilayer.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15071790

  Clinical and hematologic features of 300 patients affected by hereditary spherocytosis grouped according to the type of the membrane protein defect.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18641031

- Pasini EM, Lutz HU, Mann M, Thomas AW.
  Red blood cell (RBC) membrane proteomics--Part I: Proteomics and RBC physiology.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19540949

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/ANK1

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