Genetics Home Reference

Your Guide to Understanding Genetic Conditions

XIAP gene

X-linked inhibitor of apoptosis

Normal Function

The *XIAP* gene provides instructions for making a protein that is found in many types of cells, including immune cells. It helps protect these cells from self-destructing (undergoing apoptosis) by blocking (inhibiting) the action of certain enzymes called caspases, which are necessary for apoptosis. Specifically, the XIAP protein inhibits caspase enzymes 3, 7, and 9. The XIAP protein also plays a role in several other signaling pathways that are involved in various functions in the body.

Health Conditions Related to Genetic Changes

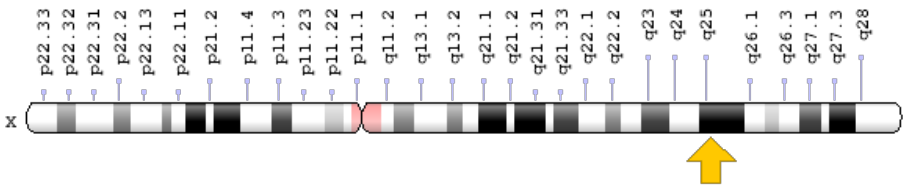
X-linked lymphoproliferative disease

XIAP gene mutations have been identified in some people with X-linked lymphoproliferative disease (XLP). These mutations reduce or eliminate production of the XIAP protein. It is unknown how a lack of XIAP protein results in the signs and symptoms of XLP, including a proliferation of lymphocytes that destroys blood-forming cells and damages the liver and other organs (hemophagocytic lymphohistiocytosis).

Chromosomal Location

Cytogenetic Location: Xq25, which is the long (q) arm of the X chromosome at position 25

Molecular Location: base pairs 123,859,812 to 123,913,979 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- API3
- apoptosis inhibitor 3
- baculoviral IAP repeat-containing protein 4
- BIRC4
- hILP
- IAP-like protein, human
- mammalian IAP homolog A
- MIHA
- X-linked inhibitor of apoptosis protein
- X-linked inhibitor of apoptosis, E3 ubiquitin protein ligase
- XIAP_HUMAN

Additional Information & Resources

GeneReviews

- Lymphoproliferative Disease, X-Linked
  https://www.ncbi.nlm.nih.gov/books/NBK1406

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28XIAP%5BTI%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- INHIBITOR OF APOPTOSIS, X-LINKED
  http://omim.org/entry/300079

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_XIAP.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=XIAP%5Bgene%5D
- HGNC Gene Family: Baculoviral IAP repeat containing
  http://www.genenames.org/cgi-bin/genefamilies/set/419

- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=592
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/331
- UniProt
  http://www.uniprot.org/uniprot/P98170

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