

## **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).

### **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

### Tests Listed in the Genetic Testing Registry

- Tests of XIAP ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=331\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=331[geneid]))

### Scientific Articles on PubMed

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

### Catalog of Genes and Diseases from OMIM

- INHIBITOR OF APOPTOSIS, X-LINKED; XIAP (<https://omim.org/entry/300079>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/331>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=XIAP\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=XIAP[gene]))

## References

- Holcik M, Gibson H, Korneluk RG. XIAP: apoptotic brake and promising therapeutic target. *Apoptosis*. 2001 Aug;6(4):253-61. doi:10.1023/a:1011379307472. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11445667>)
- Latour S. Natural killer T cells and X-linked lymphoproliferative syndrome. *Curr Opin Allergy Clin Immunol*. 2007 Dec;7(6):510-4. doi:10.1097/ACI.0b013e3282f1bad6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17989527>)
- Marsh RA, Madden L, Kitchen BJ, Mody R, McClimon B, Jordan MB, Bleesing JJ, Zhang K, Filipovich AH. XIAP deficiency: a unique primary immunodeficiency best classified as X-linked familial hemophagocytic lymphohistiocytosis and not as X-linked lymphoproliferative disease. *Blood*. 2010 Aug 19;116(7):1079-82. doi:10.1182/blood-2010-01-256099. Epub 2010 May 20. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20489057>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2938130/>)
- Marsh RA, Villanueva J, Kim MO, Zhang K, Marmer D, Risma KA, Jordan MB, Bleesing JJ, Filipovich AH. Patients with X-linked lymphoproliferative disease due to BIRC4 mutation have normal invariant natural killer T-cell populations. *Clin Immunol*. 2009 Jul;132(1):116-23. doi: 10.1016/j.clim.2009.03.517. Epub 2009 Apr 23. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19398375>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2729708/>)

- Rigaud S, Fondaneche MC, Lambert N, Pasquier B, Mateo V, Soulas P, Galicier L, Le Deist F, Rieux-Laucat F, Revy P, Fischer A, de Saint Basile G, Latour S. XIAP deficiency in humans causes an X-linked lymphoproliferative syndrome. *Nature*. 2006 Nov 2;444(7115):110-4. doi: 10.1038/nature05257. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17080092>)

## **Genomic Location**

The *XIAP* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

**Last updated August 1, 2010**