



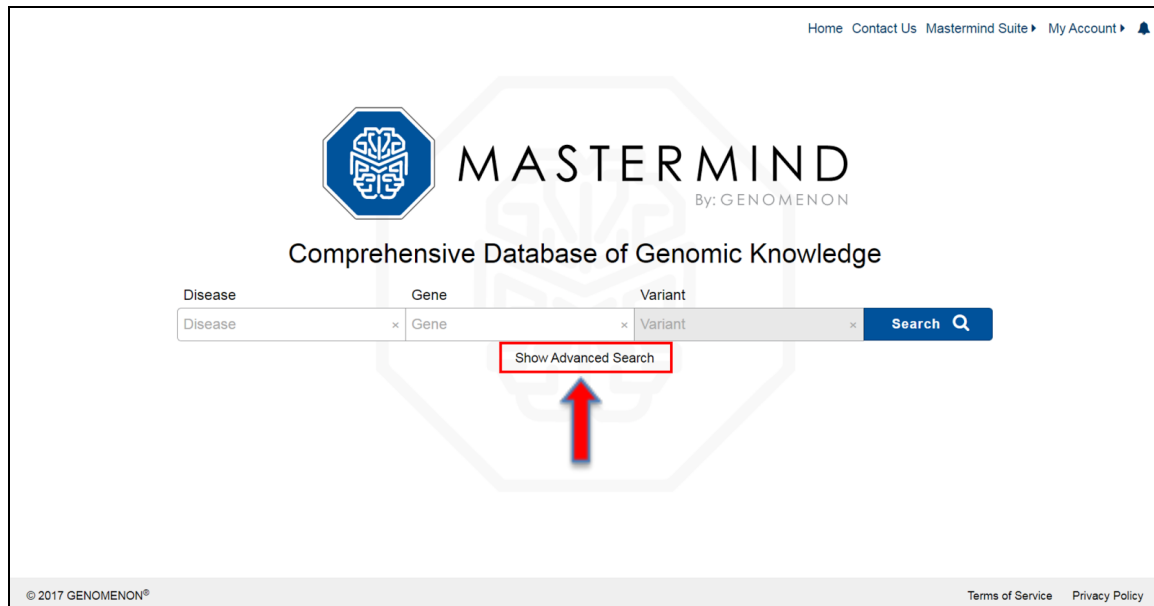
MASTERMIND®

Use Case Scenario: Advanced Search Capabilities of Mastermind

Advanced Search capabilities of Mastermind

Mastermind includes advanced search capabilities and which can be used to quickly refine your search results using your own custom keywords. It is also an especially powerful utility to find articles of interest when the expected search terms do not explicitly appear in the abstract and/or title or a described using different terminologies.

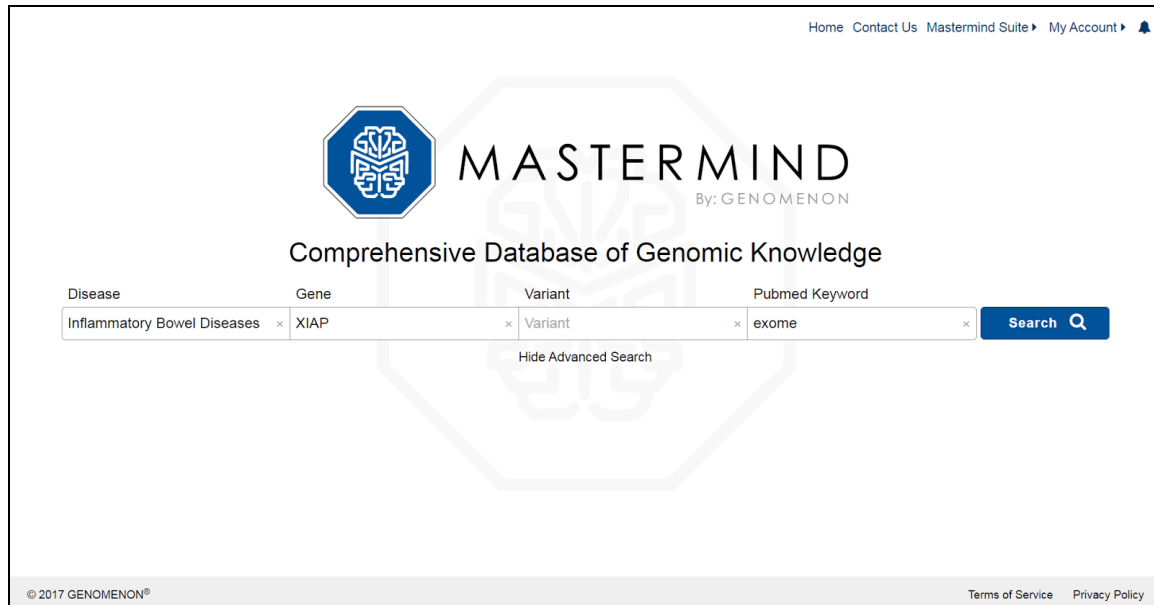
To activate the advanced search features, click on “Show Advanced Search” on the Mastermind home page.



The advanced search capabilities (PubMed Keyword field) are invoked when entering either a 1) Disease term, 2) Gene term or 3) Gene-Mutation keywords.

To illustrate the advanced search capabilities of Mastermind, we will search for all publications that have used exome sequencing to identify variants in the *XIAP* gene and their role in the development of inflammatory bowel disease.

To begin, enter the search term “inflammatory bowel diseases”, “XIAP” and “exome” in the Disease, Gene and PubMed Keyword text boxes, respectively. Click “Search”.



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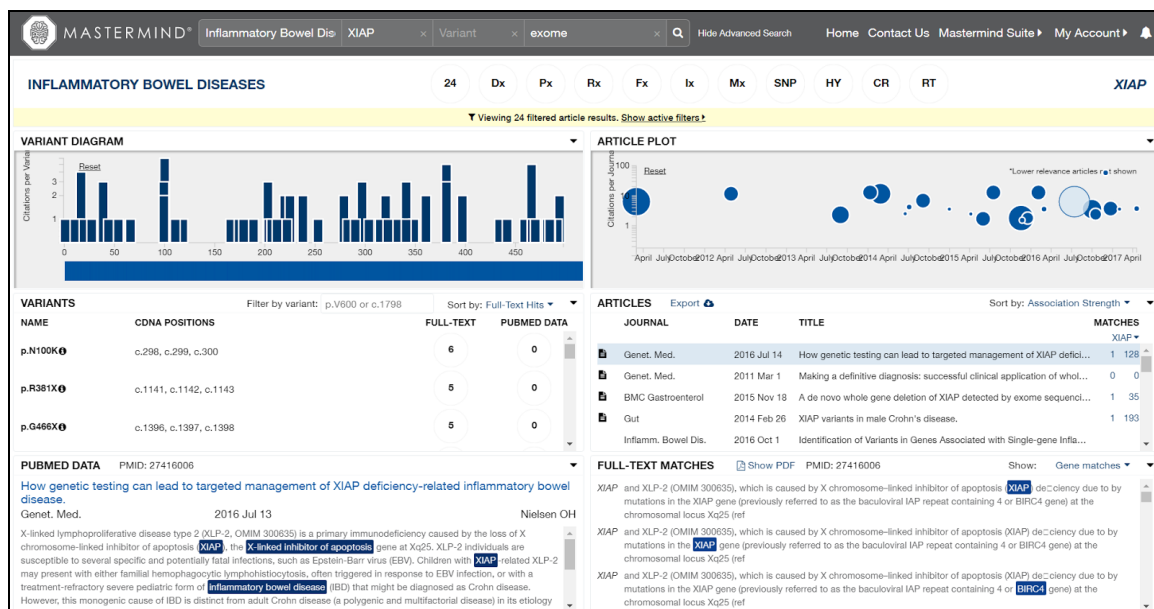
Comprehensive Database of Genomic Knowledge

Disease: Inflammatory Bowel Diseases x Gene: XIAP x Variant: Variant x PubMed Keyword: exome x **Search**

Hide Advanced Search

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This search leaps directly to a detail page with only 24 articles that satisfy the search criteria.



INFLAMMATORY BOWEL DISEASES 24 Dx Px Rx Fx Ix Mx SNP HY CR RT **XIAP**

Viewing 24 filtered article results. Show active filters

VARIANT DIAGRAM

Citations per Variant

ARTICLE PLOT

Citations per Article

VARIANTS Filter by variant: p.V600 or c.1798 Sort by: Full-Text Hits

NAME	CDNA POSITIONS	FULL-TEXT	PUBMED DATA
p.N100K	c.298, c.299, c.300	6	0
p.R381X	c.1141, c.1142, c.1143	5	0
p.G466X	c.1396, c.1397, c.1398	5	0

PUBMED DATA PMID: 27416006

How genetic testing can lead to targeted management of XIAP deficiency-related inflammatory bowel disease.

Genet. Med. 2016 Jul 13 Nielsen OH

X-linked lymphoproliferative disease type 2 (XLP-2, OMIM 300635) is a primary immunodeficiency caused by the loss of X chromosome-linked inhibitor of apoptosis (XIAP, the X-linked inhibitor of apoptosis) gene at Xq25. XLP-2 individuals are susceptible to several specific and potentially fatal infections, such as Epstein-Barr virus (EBV). Children with XIAP-related XLP-2 may present with either familial hemophagocytic lymphohistiocytosis, often triggered in response to EBV infection, or with a treatment-refractory severe pediatric form of inflammatory bowel disease (IBD) that might be diagnosed as Crohn disease. However, this monogenic cause of IBD is distinct from adult Crohn disease (a polygenic and multifactorial disease) in its etiology.

ARTICLES Export Sort by: Association Strength

JOURNAL	DATE	TITLE	MATCHES
Genet. Med.	2016 Jul 14	How genetic testing can lead to targeted management of XIAP deficiency-related inflammatory bowel disease.	1 128
Genet. Med.	2011 Mar 1	Making a definitive diagnosis: successful clinical application of whole-exome sequencing in a patient with X-linked lymphoproliferative disease type 2.	0 0
BMC Gastroenterol	2015 Nov 18	A de novo whole gene deletion of XIAP detected by exome sequencing in a patient with X-linked lymphoproliferative disease type 2.	1 35
Gut	2014 Feb 26	XIAP variants in male Crohn's disease.	1 193
Inflamm. Bowel Dis.	2016 Oct 1	Identification of Variants in Genes Associated with Single-gene Inflammatory Bowel Disease.	

FULL-TEXT MATCHES Show PDF PMID: 27416006 Show: Gene matches

XIAP and XLP-2 (OMIM 300635), which is caused by X chromosome-linked inhibitor of apoptosis (XIAP) deficiency due to by mutations in the XIAP gene (previously referred to as the baculoviral IAP repeat containing 4 or BIRC4 gene) at the chromosomal locus Xq25 (ref)

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The first article, “How genetic testing can lead to targeted management of XIAP deficiency-related inflammatory bowel disease” (Genet. Med) appears at the top of the list of publications, which are sorted by default into association strength. You can also tell by the size of bubble in “Article Plot” that it has the highest association strength with your keywords. The “Variant Diagram” panel highlights those variants which have been reported in these publications, and which have been mapped along the length of the protein.

To see the PDF of this article, click on the "Show PDF" button in "Full-Text Matches" panel, the lowest-right panel in the detail page. If the paper is freely available online, or if your institute has an online subscription the journal, the PDF will automatically load in the viewer.

The screenshot displays the Mastermind search results for 'Inflammatory Bowel Disease' and 'XIAP'. The top navigation bar includes 'MASTERMIND', search filters, and user account options. The main section is titled 'INFLAMMATORY BOWEL DISEASES' and shows '24' results. Below this, a 'VARIANT DIAGRAM' shows a bar chart of variant frequencies. A table of 'VARIANTS' lists three specific variants with their CDNA POSITIONS, FULL-TEXT counts, and PUBMED DATA counts. The 'PUBMED DATA' section provides a link to a full-text PDF of an article titled 'How genetic testing can lead to targeted management of XIAP deficiency-related inflammatory bowel disease'. The article preview on the right shows the title, authors (Ole Haagen Nielsen DMSc & Eric Charles LaCasse PhD), and a brief abstract.

VARIANTS	CDNA POSITIONS	FULL-TEXT	PUBMED DATA
p.N100K	c.298, c.299, c.300	6	0
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p.G466X	c.1396, c.1397, c.1398	5	0

You may notice that the both search term "XIAP" and the full gene name of "X-linked inhibitor of apoptosis" is highlighted in the abstract: Mastermind is capable of capturing all synonyms of any gene in our listings.

In the "Variants" panel, you will see all of the reported variants that have been described in the 24 publications. The variants can be sorted by their location in the article (Title/Abstract or Full-text) or their position along the linear axis of the protein. The quick search feature of "Filter by variant" will allow you to quickly find any variant in the list using standard variant syntax.

In summary, the Advanced Features of Mastermind can be used to 1) quickly filter publication by keyword and 2) find publications where non-standard terminologies may have been used by the corresponding author of the publication.



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We are pleased that you are interested in our software and we look forward to learning from your experience.

If any questions arise, please do not hesitate to contact us.

info@genomenon.com