



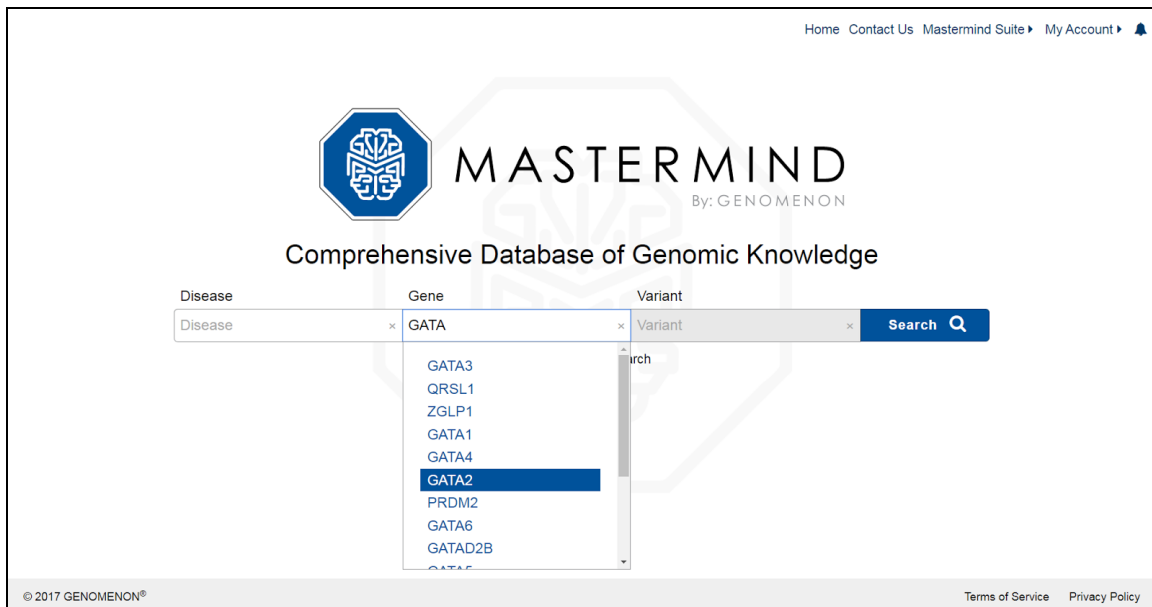
MASTERMIND®

Use Case Scenario:
Searching Mastermind by Gene

Searching Mastermind by Gene Name

Mastermind can be used to learn which diseases are associated with a given gene, and to obtain a comprehensive, up-to-date list of all of the published genetic variants associated with that gene. This is useful in clinical practice if an unfamiliar variant (often referred to as a Variant of Uncertain Significance) is encountered to help determine whether it has been published before and, if so, how many times it was described and in association with what diseases. Users can also use this capability to quickly identify new or novel mutations for targeted sequencing of the patient's genome to build a more accurate genotype-phenotype correlation.

To search by gene name, enter your search term in the "Gene" search box on the Mastermind home page. In this example, we will search for information on the *GATA2* gene of human. As you enter the search term in the text box, the auto-fill drop-down menu will allow you to select the desired search term.



The screenshot displays the Mastermind website's search interface. At the top right, there are navigation links: Home, Contact Us, Mastermind Suite, and My Account. The main header features the Mastermind logo (a blue octagon with a white geometric design) and the text "MASTERMIND By: GENOMENON". Below the header is the tagline "Comprehensive Database of Genomic Knowledge". The search interface consists of three input fields: "Disease", "Gene", and "Variant". The "Gene" field contains the text "GATA", and a dropdown menu is open below it, listing several gene names: GATA3, QRSL1, ZGLP1, GATA1, GATA4, GATA2 (highlighted in blue), PRDM2, GATA6, and GATAD2B. A "Search" button with a magnifying glass icon is located to the right of the input fields. At the bottom left, there is a copyright notice: "© 2017 GENOMENON®". At the bottom right, there are links for "Terms of Service" and "Privacy Policy".

After clicking "Search" a results summary page will be shown. This list represents all of the Medical Subject Heading (MeSH) terms that are associated with the GATA2 gene. Results are rank-ordered by the number of publications in each MeSH term. The "ALL" link will open a summary page where all of the publications and the reported variants associated with GATA2 can be viewed. One can use this search result to obtain a list of all the publications and associated variants for a specific disease-gene pairing, such as GATA2 and Acute Myeloid Leukemia (AML). Click on the line displaying "LEUKEMIA, MYELOID, ACUTE" to see the detail page for AML-GATA2.

Disease	Articles	Gene
ALL	3.7k	GATA2
HUMANISM	1.2k	GATA2
GENE EXPRESSION	1.0k	GATA2
GENERALIZATION (PSYCHOLOGY)	669	GATA2
INHIBITION (PSYCHOLOGY)	542	GATA2
LEUKEMIA	506	GATA2
LEUKEMIA, MYELOID	299	GATA2
LEUKEMIA, MYELOID, ACUTE	260	GATA2
INDIVIDUALITY	253	GATA2
INDIVIDUATION	253	GATA2
HYPERPHAGIA	241	GATA2

As there are a fairly large number of publications associated with both GATA2 and AML, a quick way to prioritize the search results is to investigate those variants with the highest number of citations in the medical literature. Navigate to the "Variants" panel, which will already be sorted by "Full-Text Hits", and selected "Full Text" for the variant that is most relevant to you.

The screenshot shows the Mastermind database interface for the gene GATA2. The search filters are set to 'Leukemia, Myeloid, Acute' and 'GATA2'. The interface displays several panels:

- VARIANT DIAGRAM:** A bar chart showing citations per variant across the GATA2 gene region.
- ARTICLE PLOT:** A scatter plot showing citations per article over time from 2012 to 2017.
- VARIANTS:** A table listing variants and their associated citations:

NAME	CDNA POSITIONS	FULL-TEXT	PUBMED DATA
p.T354M	c.1060, c.1061, c.1062	18	3
p.L359V	c.1075, c.1076, c.1077	14	1
p.M1del	c.1, c.2, c.3	12	0
- ARTICLES:** A table listing articles related to GATA2 mutations, including titles, dates, and match counts.
- FULL-TEXT MATCHES:** A section showing the full text of a specific article (PMID: 21892162) with a 'Show PDF' button highlighted in red.

As an example of the information made available in this association page, there are 18 citations where the p.T354M variant co-occurs in the full text with the terms GATA2 and AML. To view the PDF of any of these articles, first select a title from the "Articles" panel, and then click on the "Show PDF" from the "Full-Text" panel. In instances where the full-text article is not freely-available, you will need to either have an institutional subscription to the online journal, or pay a one-time fee to the Publisher to access and download the article directly from the Publisher's website.

In summary, searching Mastermind by Gene will enable you to 1) see all diseases associated with a given gene; 2) view the reported genetic variants for a given gene associated with a genetic disease; and 3) obtain (where applicable) the underlying, supporting publication from the biomedical literature.



MASTERMIND

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.

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