MASTERMIND®

Use Case Scenario:
Searching Mastermind by Gene

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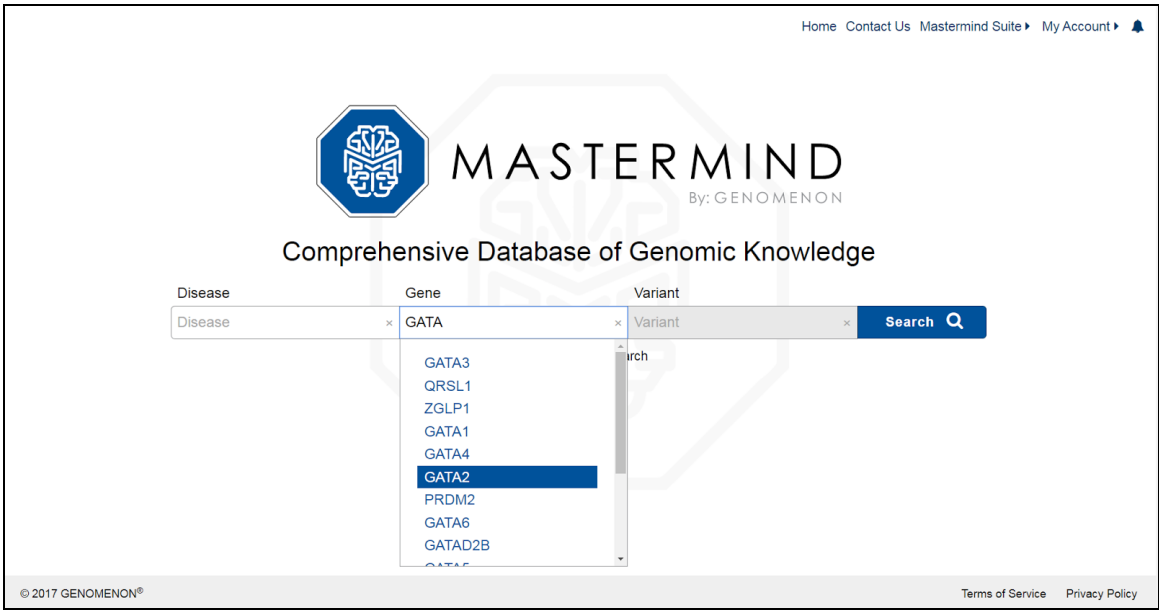
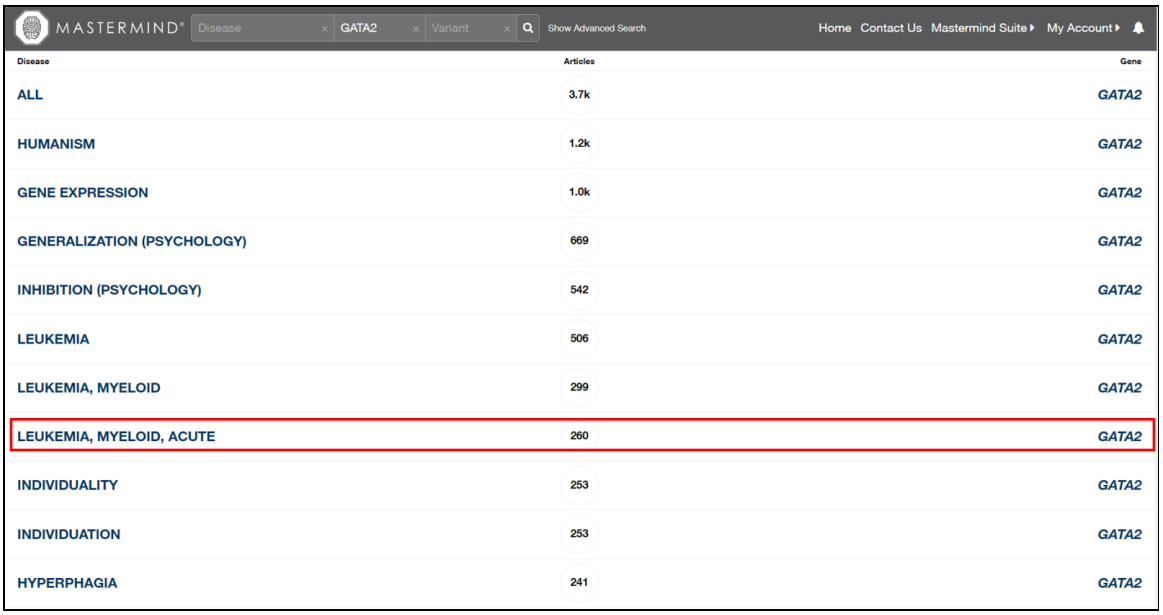
Searching Mastermind by Gene Name

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-phenotypecorrelation.

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.

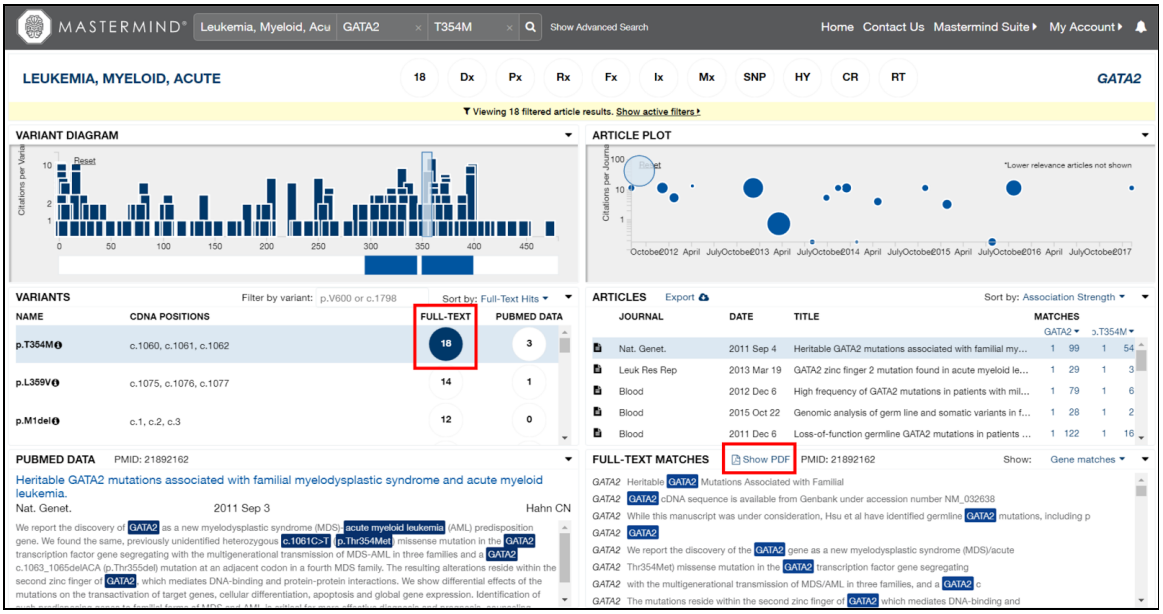


After clicking "Search" a results summary page will be shown. This list represents all of the Medical Subject Heading (MeSH) terms that are associated with the *GATA2* gene. Results are rank-ordered by the number of publications in each MeSH term. The "ALL" link will open a summary page where all of 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.



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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