MASTERMIND®

Use Case Scenario:
Searching Mastermind by Disease

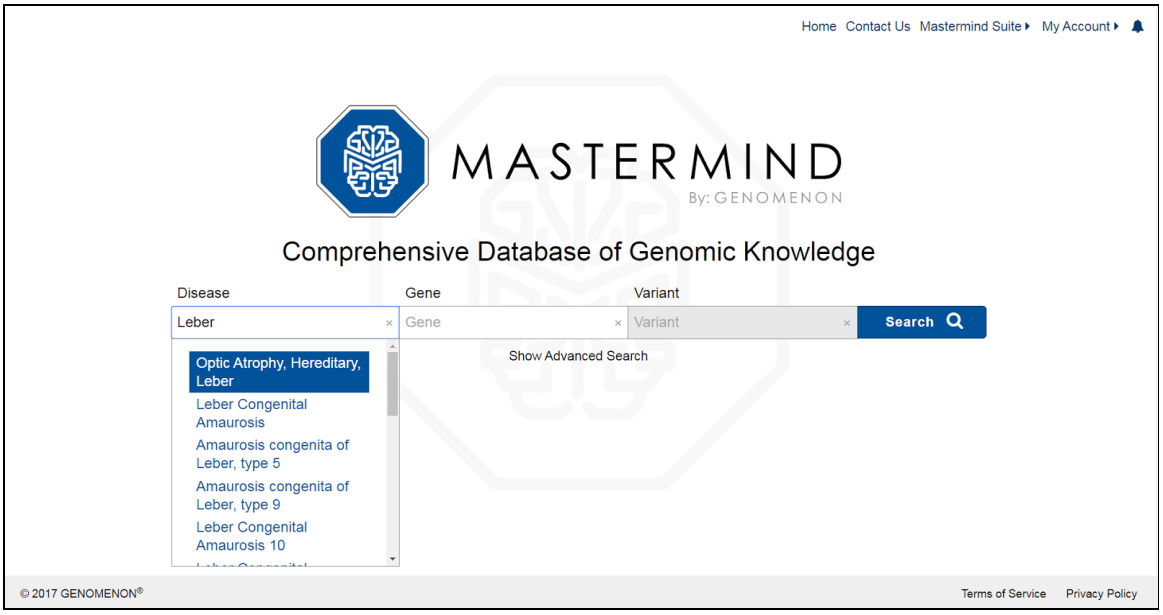
info@genomenon.com
https://www.genomenon.com

Searching Mastermind by Disease

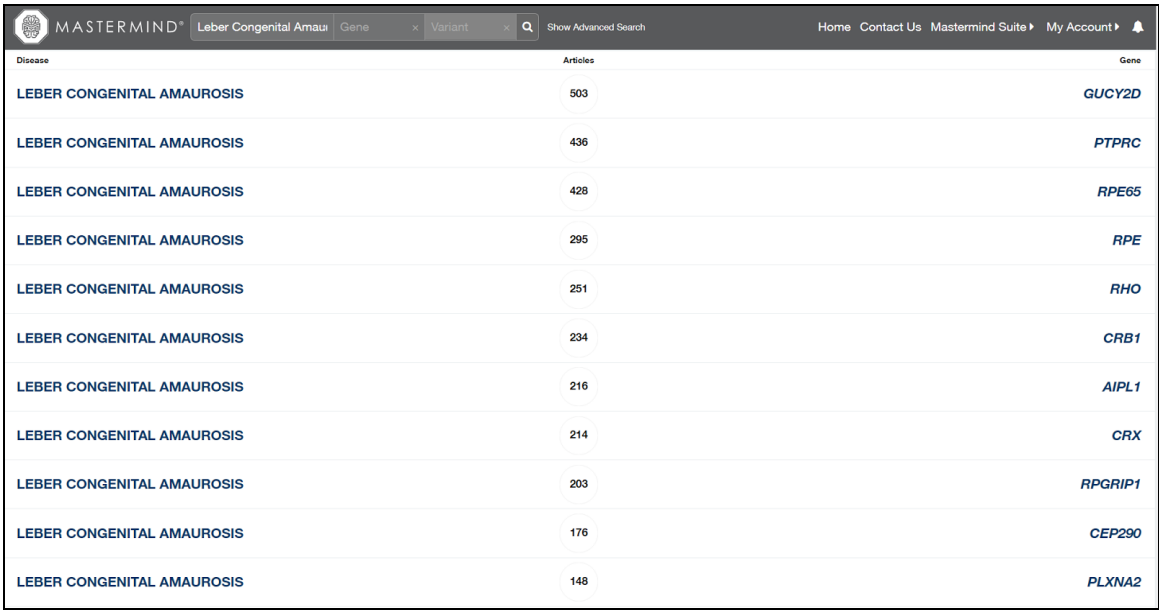
Mastermind can be used to obtain a comprehensive, up-to-date list of all of the genes associated with a given disease and their associated genetic variants. These results can be used to inform gene panel design by cataloguing genes and/or variants that are linked to a particular genetic disease.

To search by disease, enter your search term in the "Disease" search box on the Mastermind home page at http://mastermind.genomenon.com. Note that you will first need to login to the software with the username and password that was provided to you via email from GENOMENON at the start of your trial or license.

In this example, we will search for information on "Leber Congenital Amaurosis". 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.

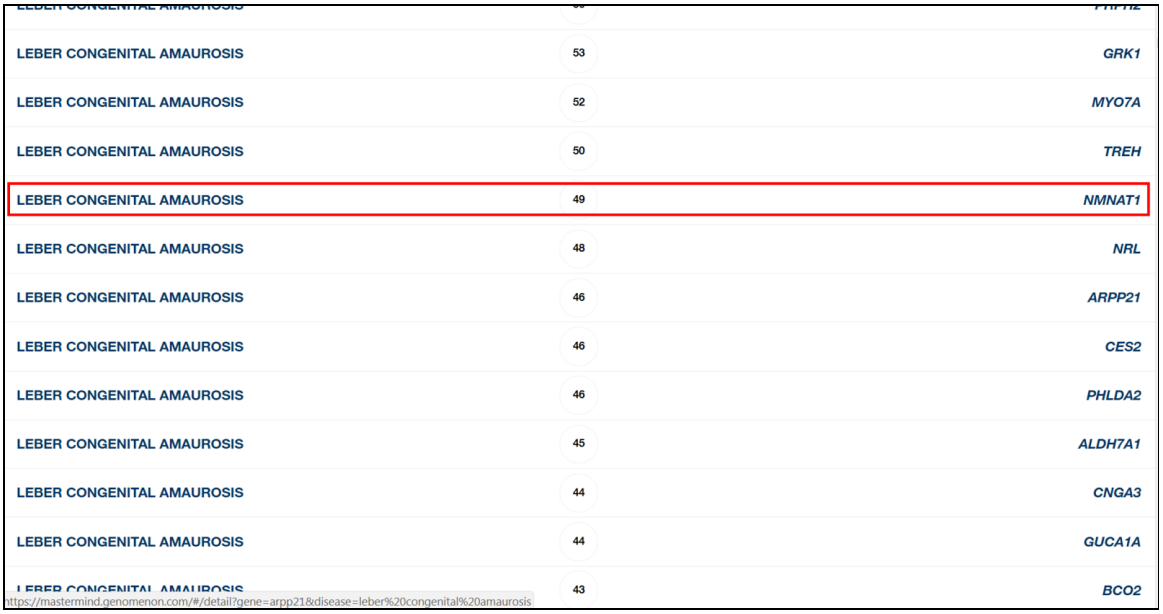


After clicking "Search" a results summary page will be shown. This list represents all genes that are associated with Leber Congenital Amaurosis (LCA) from the medical literature. Results are ordered by the number of publications citing the listed the gene-disease association. In this example, *GUCY2D*, which is associated with LCA type 1, has the highest number of publications showing an association between this gene and LCA. Less-documented or new/novel gene-disease associations are listed below in descending order of article count.

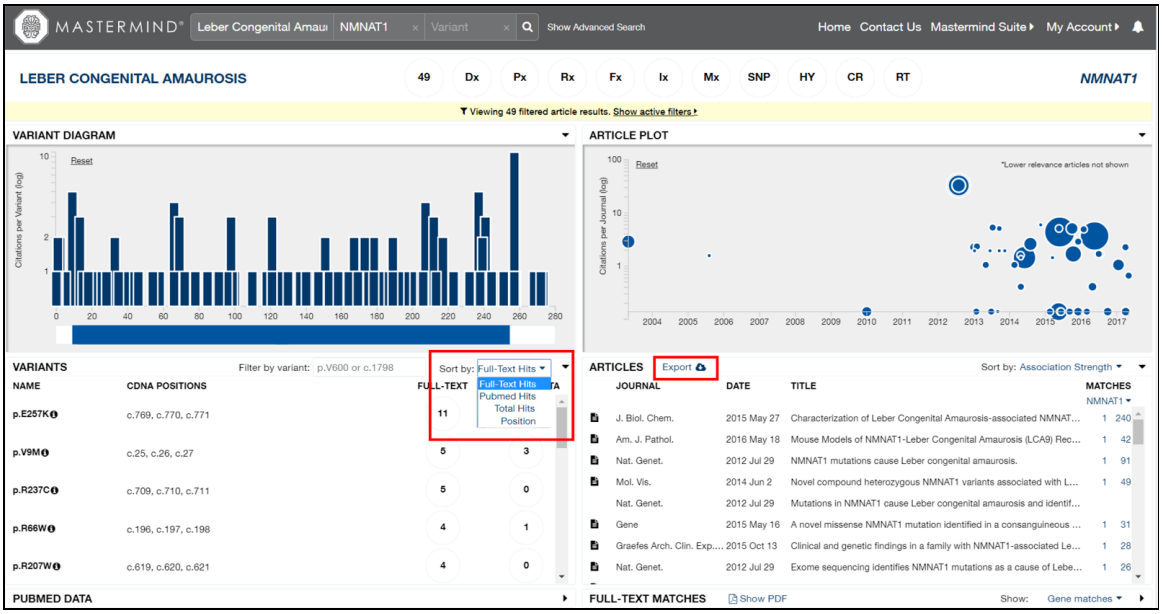


Clicking on the entry for *GUCY2D* allows you to see the full list of publications citing this association as well as all associated variants in *GUCY2D*. The "Variant Diagram" can be used to view the distribution of the reported variants along the linear access of the protein. In some instances, you may see a large pile-up of hits at a given location on the protein/cDNA, which indicates that multiple articles described the same variant. In this example, the range of reported genetic variants for *GUCY2D* span the entire length of the protein.

Scroll down towards the end of the list to the entry for *NMNAT1* and click on the gene name to see the list of publications and associated genetic variants. There are 49 publications with reported variants associated with *NMNAT1*. Mutations in *NMNAT1* are associated with LCA type 9 in affected individuals.



To find position at which the highest number of variants has been reported, move to the "Variants" panel of the report, which by default is sorted by "Full-Text Hits".



There are 11 full-text publications associated with the p.E257K variant. For future access, a file containing the PubMed Identification number, the title and the journal name for each article in the article list can be exported from Mastermind by clicking on the "Export" icon at the upper right of the Articles panel. To view the PDF of any publication, click on the title you are interested in, then click "Show PDF" header bar of the "Full-Text Matches" 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 Disease will enable you to: 1) see all genes associated with a given disease; 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.

info@genomenon.com