



# MASTERMIND®

## Use Case Scenario: Searching Mastermind by Variant

## Searching Mastermind by Variant

Mastermind can be used to search for all publications associated with a known, previously reported genetic variant. As new articles describing a specific disease-causing variant are being published daily, using Mastermind to keep up-to-date with the latest information and clinical findings can help guide and accelerate precision medicine initiatives in the clinic.

In the following example, we will use Mastermind to search for the p.G551D variation in the *CFTR* gene. The p.G551D variant, in which the amino acid glycine is replaced with aspartic acid at position 551 in the protein, results in a dysfunctional cell surface protein that is unable to transport chloride through a channel.

From the Mastermind home page, enter "CFTR" in the Gene field to automatically enable the Variant query box.

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**MASTERMIND**  
By: GENOMENON

Comprehensive Database of Genomic Knowledge

Disease	Gene	Variant
Disease	CFTR	g

Show Advanced Search

- G551D
- G542X
- G85E
- G1349D
- G622D
- G550E
- G480C
- G551S
- G628R
- G64D

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The results page will show you a list of MeSH disease terms where this gene-variant pair has been described in the full text, title, and abstract of any corresponding publications. The results are also sorted with the highest number of publications appearing at the top.

Next, click on the disease term "CYSTIC FIBROSIS" to go to an overview page with information about this specific disease-gene-mutation association.

Disease	Articles	Gene
ALL	2.0k	CFTR
FIBROSIS	1.8k	CFTR
CYSTIC FIBROSIS	1.8k	CFTR
HUMANISM	326	CFTR
LUNG DISEASES	258	CFTR
INDIVIDUALITY	240	CFTR
INDIVIDUATION	240	CFTR
PANCREATITIS	220	CFTR
GENERALIZATION (PSYCHOLOGY)	217	CFTR
SWEATING	217	CFTR
INHIBITION (PSYCHOLOGY)	197	CFTR

In the bottom left panel, labeled "Variants", you will see the p.G551D variant, along with p.G551del, since our search term did not exclude this variant.

At the top of this detail page you will see a toolbar with abbreviated terms that further qualify the publications by various subcategories. This will be handy since even when searching by a Disease-Gene-Variant trio, there are still 1.7k articles to sort through. Mousing over each term will reveal the full names of each subcategory. The treatment category (Rx) itemizes those publications where a therapeutic treatment is likely to have been described. To view these publications, click on the "Rx" ico. This will open a menu with additional search terms that can be used to further filter your results.

The “drug” field will generate a list of publications (counted in parenthesis) that describe a specific drug therapy administered in CF patients harboring the *CFTR*-p.G551D variant. To generate this list, you will first need to click on the “Disable All” option to deselect all search terms, then click on “drug”.

The screenshot displays the Mastermind search results for Cystic Fibrosis with the p.G551D variant. The interface includes a navigation bar with tabs for Cystic Fibrosis, CFTR, and G551D. A filter menu at the top shows 'Rx' selected, with other options like Dx, Px, Fx, Ix, Mx, SNP, HY, CR, and RT. The main content area features 'Enable All' and 'Disable All' buttons. Below these are counts for various categories: treatment (413), pharmaceutical (30), dosage (3), drug resistance, drug (208), therapy (320), [a-z]\*ib (194), resistance mechanism, compound (176), dose (72), and [a-z]\*ab (32). A table lists variants and their associated PubMed data, and a section shows full-text matches for a specific article.

NAME	CDNA POSITIONS	FULL-TEXT	PUBMED DATA
p.G551D	c.1651, c.1652, c.1653	1.7k	266
p.G551del	c.1651, c.1652, c.1653	33	0

Publications can then be browsed in the “Articles” panel in the center right. The default view rank orders the publications by the strength of the term associations in the full text, title, and abstract, but they can also be sorted by Publication Date, Journal Name, and Impact Factor. Clicking on the “Export” icon will then export the list of filtered publications (PubMed ID, Title, and Journal in .csv format) which can be saved locally.

In summary, searching Mastermind with a gene name and known variant will enable you to 1) see all publications associated with a gene-variant-disease association and 2) filter the search results by subcategory to identify subsets of publications describing a particular treatment, therapy or biological outcome (among others).



# 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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