GENOMENON®

Mastermind Integration

Technical Documentation

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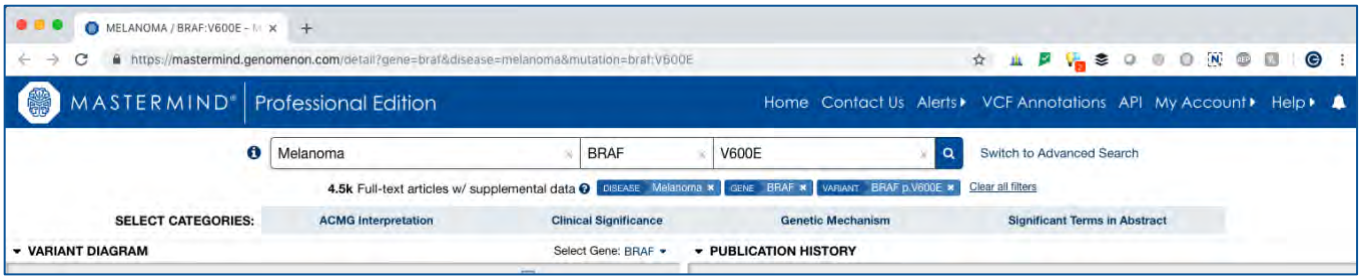
Linking to Mastermind

Link Structure

The Mastermind Genomic Search Engine provides structured URLs which allow easy deep-linking into search results for a given disease, gene, and/or variant. For example, here is a URL which takes the user directly to articles for Melanoma associated with the BRAF:V600E variant:

https://mastermind.genomenon.com/detail?gene=braf&disease=melanoma&mutation=braf:V600E



This URL format can be used to dynamically generate deep-links into Mastermind searches by simply replacing the disease, gene, and variant in the URL.

https://mastermind.genomenon.com/detail?gene={gene_symbol}&disease={disease_name}&mutation={gene_symbol:variant_key}

Any of the parameters may be omitted, with the following constraints:

- For Free Edition users, at least the disease *and* gene must be specified. The variant parameter may be omitted.
- For Professional Edition users, either one of at least the disease *or* gene must be specified. The other parameter may be omitted for a list of genes associated with the specified disease, or diseases associated with the specified gene.
- For variant searches, the gene parameter must also be specified for the variant.

Parameter values and formats

The disease, gene, and variant parameters should follow these rules:

Gene

The value for the gene parameter should be the desired HGNC gene symbol in lowercase.

Variant

The value for the variant parameter should include the same gene symbol from the gene parameter followed by a colon, followed by the variant key in Mastermind gene variant nomenclature, which is based on HGVS protein (effect) level nomenclature and optimized for search sensitivity.
For most variants, this is simply the protein nomenclature. However, to maximize sensitivity for specific types of deletions, insertions, duplications, frame-shifts, and non-coding variants, there is a simple transformation to Mastermind variant nomenclature, outlined in the FAQ.

Disease

The value for the disease should be the canonical MeSH term for the disease, in lower-case and URL-encoded (e.g. spaces are encoded as "%20").

To view results irrespective of citations of any specific disease, the value of "all%20diseases" may also be encoded. This will show results for all diseases together.

For help with any specific disease, gene, or variant, try typing the desired value into the appropriate field in the homepage search interface to see which valid values are suggested as part of the auto-complete drop-down.

Advanced Search

In addition to the standard disease/gene/variant searches described above, Mastermind Professional Edition also offers advanced search functionality for:

- searching multiple diseases, genes, and/or variants;
- ACMG/AMP guideline-based searching;
- filters for clinical significance and genetic mechanisms, such as amplifications, CNVs, and more;
- exporting of PMID lists with supporting information for any search.

All of these search features are able to be used, combined, and patterned directly within the URL of the links into Mastermind, provided that the users clicking the links have access to Mastermind Professional Edition. If the user has access only to Mastermind Free Edition, upon clicking, they will be shown a message that the linked search requires Professional Edition to view.

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Cited Variants Reference

The Mastermind Cited Variants Reference file is a VCF file providing a snapshot of variant evidence available in Mastermind. Below is a truncated excerpt:

HGVS	MMCNT1	MMCNT2	MMCNT3	MMURL3
g.3209075C>T	4	4	4	https://mastermind.genomenon.com/…mutation=SLC4A11:Q812sa
g.865595A>G	1	1	1	https://mastermind.genomenon.com/…mutation=SAMD11:K45E
g.874491_874492delinsTA	0	0	1	https://mastermind.genomenon.com/…mutation=SAMD11:R168X

`MMCNT1` column (most specific)

cDNA-level exact matches. This is the number of articles that mention the variant at the nucleotide level in either the title/abstract or the full-text.

`MMCNT2` column

cDNA-level possible matches. This is the number of articles with nucleotide-level matches (from 1) plus articles with protein-level matches in which the publication did not specify the cDNA-level change, meaning they could be referring to this nucleotide-level variant but there is insufficient data in these articles to determine conclusively.

`MMCNT3` column (most sensitive)

This is the number of articles citing any variant resulting in the same biological effect as this variant. This includes the articles from MMCNT1 and MMCNT2 plus articles with alternative cDNA-level variants that result in the same protein effect.

`MMURL3` column

This is a deep-link into Mastermind for the selected variant, which shows all articles from MMCNT3, in order to investigate and explore the evidence in the literature.

Mastermind API Endpoints

The Mastermind API is a set of programmatic endpoints available to interact with the Mastermind data at a more detailed and granular level.

API Endpoints

`/suggestions` API endpoint BASE

Returns the canonical name(s) to query in Mastermind for the given disease, gene, or variant.

`/counts` API endpoint BASE

Returns the number of articles that match the given query for disease, gene, and variant.

`/articles` API endpoint ADVANCED

Returns the first 5 PMIDs, ranked by relevance, for the given query of disease, gene, and variant.

`/genes` API endpoint ADVANCED

Returns the top 5 genes associated with the given disease, ranked by relevance.

`/diseases` API endpoint ADVANCED

Returns the top 5 diseases associated with the given gene and variant, ranked by relevance.

`/variants` API endpoint ADVANCED

Returns the top 5 variants associated with the given gene and disease, ranked by the number of articles matched.

`/article_info` API endpoint ADVANCED

Returns the matched diseases, genes, variants, and article meta-data for a given article.

The `/suggestions` API endpoint returns the canonical name(s) to query in Mastermind for the given disease, gene, or variant.

Query Parameters

Name	Description	Example
`disease`	The disease name associated with the articles of interest, specified in lowercased MeSH terminology.	melanoma
`gene`	The gene name associated with the articles of interest, specified in lowercase by their HGNC gene symbol.	braf
`mutation`	The variant associated with the articles of interest, specified in lowercase by their HGVS protein-level nomenclature for SNVs, or modified nomenclature for del/ins variants.	v600e

Response Format

```
{
  "type": [[disease, gene, or variant]],
  "matched": [[synonym or nomenclature matched]],
  "canonical": [[canonical name to use for Mastermind queries]],
  "url": [[link to Mastermind query using the canonical name]]
}
```

Example Response

```
{
  "type": "disease",
  "matched": "acute myeloid leukemia",
  "canonical": "Leukemia, Myeloid, Acute",
  "url": "https://mastermind.genomenon.com/search?disease=leukemia,%20myeloid
}
```

The `/counts` API endpoint returns the number of articles that match the given query for disease, gene, and variant.

Query Parameters

Name	Description	Example
`disease`	The disease name associated with the articles of interest, specified in lowercased MeSH terminology.	melanoma
`gene`	The gene name associated with the articles of interest, specified in lowercase by their HGNC gene symbol.	braf
`mutation`	The variant associated with the articles of interest, specified in lowercase by their HGVS protein-level nomenclature for SNVs, or modified nomenclature for del/ins variants.	v600e
`source`	Search within PubMed data and the full-text literature, or search PubMed data only. *This requires Mastermind Professional Edition to view articles in Mastermind.*	full-text (default) or pubmed
`categories`	An array of categories and/or keywords from our category/keyword list, which includes ACMG criteria and other clinical relevance indicators. *This requires Mastermind Professional Edition to view articles in Mastermind.*	["in vivo", "de novo"]

Response Format

```
{
  "count": [[total number of articles]]
  "url": [[link to Mastermind results]]
}
```

Example Response

```
{
  "count": 4347,
  "url": "https://mastermind.genomenon.com/detail/?disease=melanoma&gene=braf
}
```

The `/articles` API endpoint returns the first 5 PMIDs, ranked by relevance, for the given query of disease, gene, and variant.

Query Parameters

See "Counts" endpoint

Response Format

```
{
  "count": [[total number of articles]],
  "articles": [
    {
      "pmid": [[pmid]],
      "title": [[title]],
      "publication_date": [[normalized publication date]],
      "journal": [[journal ISO code]],
      "url": [[link to view article in Mastermind]]
    }
  ],
  "url": [link to Mastermind results]
}
```

Example Response

```
curl -H "Content-type: application/json" -H "X-API-TOKEN: ABC123XYZ" -X GET "
```

```
{
  "count": 7751,
  "articles": [
    {"pmid": 23543365, "title": "Gossypin as a novel selective dual inhibitor
    {"pmid": 12447372, "title": "High frequency of BRAF mutations in nevi.",
    {"pmid": 24145418, "title": "Phenformin enhances the therapeutic benefit
    {"pmid": 25624498, "title": "BRAF inhibitor resistance mediated by the AK
    {"pmid": 24192036, "title": "BRAF inhibitors suppress apoptosis through o
  ],
  "url": "https://mastermind.genomenon.com/detail?gene=braf&disease=melanoma"
}
```

The `/genes` API endpoint returns the top 5 genes associated with the given disease, ranked by relevance.

Query Parameters

Name	Description	Example
`disease`	The disease name associated with the articles of interest, specified in lowercased MeSH terminology.	melanoma

Response Format

```
{
  "genes": [
    {
      "symbol": [[gene symbol]],
      "url": [[link to Mastermind for disease and gene]]
    }
  ],
  "url": [[link to Mastermind query for the given disease]]
}
```

Example Response

```
{
  "genes": [
    {"symbol": "IL2", "url": "https://mastermind.genomenon.com/detail?gene=il
    {"symbol": "CD8A", "url": "https://mastermind.genomenon.com/detail?gene=c
    {"symbol": "CES2", "url": "https://mastermind.genomenon.com/detail?gene=c
    {"symbol": "TNF", "url": "https://mastermind.genomenon.com/detail?gene=tn
    {"symbol": "BRAF", "url": "https://mastermind.genomenon.com/detail?gene=b
  ],
  "url": "https://mastermind.genomenon.com/search?disease=melanoma"
}
```

The `/diseases` API endpoint returns the top 5 diseases associated with the given gene and variant, ranked by relevance.

Query Parameters

Name	Description	Example
`gene`	The gene name associated with the articles of interest, specified in lowercase by their HGNC gene symbol..	braf
`mutation`	The variant associated with the articles of interest, specified in lowercase by their HGVS protein-level nomenclature for SNVs, or modified nomenclature for del/ins variants.	v600e

Response Format

```
{
  "diseases": [
    {
      "name": [[MeSH disease name]],
      "url": [[link to Mastermind for disease with gene and variant]]
    }
  ],
  "url": [[link to Mastermind query for the given gene and variant]]
}
```

Example Response

```
curl -H "Content-type: application/json" -H "X-API-TOKEN: ABC123XYZ" -X GET "
```

```
{
  "diseases": [
    {"name": "Neoplasms", "url": "https://mastermind.genomenon.com/detail?gen
    {"name": "Melanoma", "url": "https://mastermind.genomenon.com/detail?gene
    {"name": "Carcinoma", "url": "https://mastermind.genomenon.com/detail?gen
    {"name": "Neoplasm Metastisis", "url": "https://mastermind.genomenon.com/
    {"name": "Thyroiditis", "url": "https://mastermind.genomenon.com/detail?g
  ],
  "url": "https://mastermind.genomenon.com/search?gene=braf&mutation=braf:V60
}
```

The `/variants` API endpoint returns the top 5 variants associated with the given gene and disease, ranked by the number of articles matched.

Query Parameters

Name	Description	Example
`gene`	The gene name associated with the articles of interest, specified in lowercase by their HGNC gene symbol.	braf
`disease`	The disease name associated with the articles of interest, specified in lowercased MeSH terminology.	melanoma

Response Format

```
{
  "variants": [
    {
      "key": [[Mastermind variant key name]]},
      "gene": [[HGVS gene symbol]],
      "url": [[link to Mastermind for disease, gene, and variant]],
      "articles_count": [[total number of articles]]
    }
  ],
  "url": [[link to Mastermind query for the given disease]]
}
```

Example Response (truncated to first two results)

```
{
  "variants": [
    {
      "key": "R408W",
      "gene": "PAH",
      "articles_count": 358,
      "url": "https://mastermind.genomenon.com/detail?gene=pah&disease=phenyl
    },
    {
      "key": "Y414C",
      "gene": "PAH",
      "articles_count": 225,
      "url": "https://mastermind.genomenon.com/detail?gene=pah&disease=phenyl
    }
  ],
  "url": "https://mastermind.genomenon.com/detail?gene=pah&disease=phenylketo
}
```

The `/article_info` API endpoint returns the matched diseases, genes, variants, and article meta-data for a given article.

Query Parameters

Name	Description	Example
`pmid`	The PubMed ID for the desired article.	23543365

Response Format

```
{
  "pmid": [[PMID of article]],
  "title": [[PubMed title]],
  "publication_date": [[Normalized publication date]],
  "journal": [[Journal ISO abbreviation]],
  "diseases": [
    {"name": [[MeSH disease name]]}
  ],
  "genes": [
    {
      "symbol": [[HGVS gene symbol]],
      "variants": [
        {"key": [[Mastermind variant key name]]}
      ]
    }
  ],
  "url": [[link to Mastermind query for the article]]
}
```

Example Response (truncated results)

```
{
  "pmid": 23543365,
  "title": "Gossypin as a novel selective dual inhibitor of V-RAF murine sarc
  "publication_date": "2017-12-01",
  "journal": "Mol. Cancer Ther.",
  "diseases": [
    {"name": "Melanoma"},
    {"name": "Retinoblastoma"},
    {"name": "Sarcoma"}
  ],
  "genes": [
    {
      "symbol": "BRAF",
      "variants": [
        {"key": "V600E"}
      ],
      "url": "https://mastermind.genomenon.com/detail?gene=braf&disease=all%2
    }, {
      "symbol": "CDK4",
      "variants": [],
      "url": "https://mastermind.genomenon.com/detail?gene=cdk4&disease=all%2
    }
  ]
}
```