

v1.0

GenomeTrax

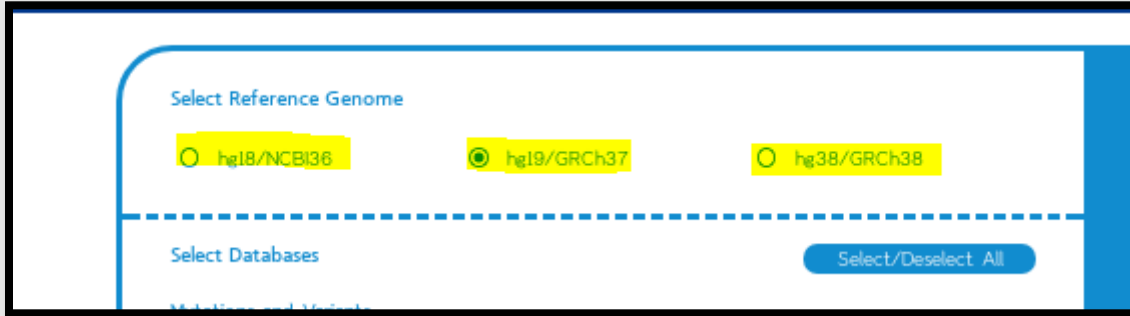
Manual



목차

- Search
- Result

Search



Select Reference Genome

☐ hg18/NCBI36 ☒ hg19/GRCh37 ☐ hg38/GRCh38

Select Databases Select/Deselect All

1. Genome을 선택합니다(hg18, hg19, hg38)

Search

Select Databases

Select/Deselect All

Mutations and Variants

☐ HGMD inherited disease mutations
☐ HGMD imputed inherited disease mutations
☐ PharmacoGenomic Mutation Database
☐ ClinVar Variants
☐ GWAS Catalogue
☐ EVS Exome Variations
☐ dbNSFP Nonsynonymous functional predictions
☐ dbSNP
☐ TCGA
☐ Complete Genomics

Regulatory Features

☐ TRANSFAC experimentally verified TFBS
☐ Predicted ChIP-Seq TFBS
☐ Predicted TFBSs in DNase hyper sensitivity regions
☐ CpG Islands
☐ Virtual Transcription Start Sites (TSSs)
☐ Microsatellites
☐ Post translational modifications
☐ miRNA
☐ dbSNV Variations in splice sites

Gene Functional Assignments

☐ Disease associations
☐ Drug Targets
☐ Pathway Membership
☐ HGMD disease genes
☐ Orphanet
☐ OMIM

2. 검색할 데이터베이스를 선택합니다.

* 'Select/Deselect All' 버튼을 통해 전 데이터베이스를 선택/해제할 수 있습니다.

Search(Coordinates)

Service Support

Search data on coordinates with conditions
(e.g. chr17:7154000-7764532 A>C TP53)

More Examples

chr17:7154000-7764532 A>C TP53

Submit

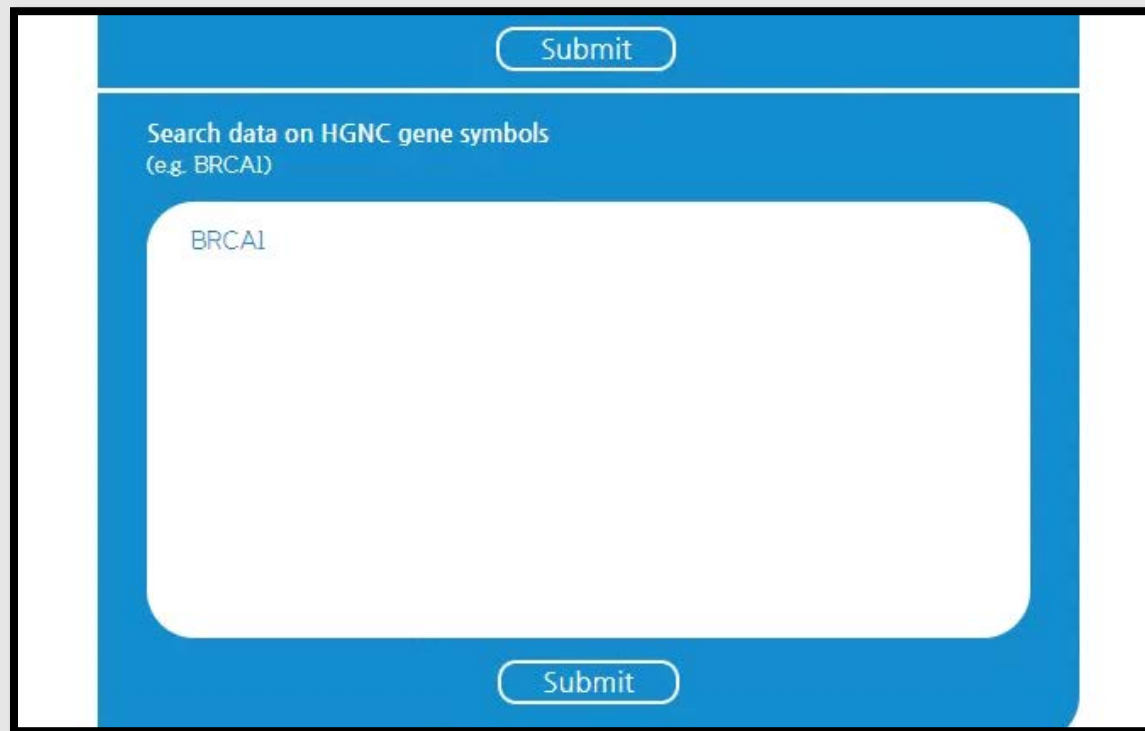
3. Coordinates 검색방법

chr[ver]:[featureStart]-[end][Nucleotide Change][Gene Name]

ex) chr17:7154000-7764532 A>C TP53

- 각 항목은 '띄어쓰기'로 구분하여야 합니다.
- chr[ver]:[featureStart]-[end]는 **무조건** 입력해주셔야 합니다.
- Nucleotide와 hgnc **없이** 검색이 가능합니다.
- 검색어 입력 후, 하단에 위치한 Submit버튼을 누르시면 됩니다.
- **검색 결과는 최대 1,000,000개로 제한됩니다.**

Search(Gene Name)



Submit

Search data on HGNC gene symbols
(e.g. BRCA1)

BRCA1

Submit

3-2. Gene Name(Hgnc) 검색 방법

[hgnc]

ex) BRCA1

- **Gene Name**만 입력해 주시면 됩니다.
- 검색 입력 후, 하단에 위치한 '**Submit**'버튼을 누르시면 됩니다.

Result

1) 데이터베이스 목록

2) Column별 필터링

3) All/Current 버튼

4) Download 버튼

Genometrax Result

1

HGMD inherited disease mutations

2

3

All ☒ Current

4

Download

Chromosome	Feature Start	Feature End	Type	Genome	SL	Hgnc	Description	Accession	Alt	Aminoacid Change
chr17	7571752	7571752	HGMD inherited disease mutations	hg19	-	TP53	Basal cell carcinoma (increased risk association with)	CM118782	G	N/A
chr17	7572846	7572846	HGMD inherited disease mutations	hg19	-	TP53	Breast and/or ovarian cancer	CM160112	G	E>A
chr17	7577081	7577081	HGMD inherited disease mutations	hg19	-	TP53	Adrenocortical carcinoma	CM920679	G	E>A
chr17	7577096	7577096	HGMD inherited disease mutations	hg19	-	TP53	Li-Fraumeni syndrome	CM114008	G	D>A
chr17	7577530	7577530	HGMD inherited disease mutations	hg19	-	TP53	Li-Fraumeni syndrome	CM114005	G	I>L
chr17	7578190	7578190	HGMD inherited disease mutations	hg19	-	TP53	Meningioma	CM015378	G	Y>S
chr17	7578271	7578271	HGMD inherited disease mutations	hg19	-	TP53	Li-Fraumeni syndrome	CM083194	G	H>P
chr17	7578291	7578291	HGMD inherited disease mutations	hg19	-	TP53	Breast cancer	CS941545	G	N/A
chr17	7578536	7578536	HGMD inherited disease mutations	hg19	-	TP53	Multiple cancer	CM086989	G	K>Q
chr17	7578914	7578914	HGMD inherited disease mutations	hg19	-	TP53	Li-Fraumeni syndrome	CM156966	G	T>P

Result(DB List)

1

HGMD inherited disease mutations	
PharmacoGenomic Mutation Database	
ClinVar Variants	Type
GWAS Catalogue	HGMD inherited disease mutations
EVS Exome Variations	HGMD inherited disease mutations
dbNSFP Nonsynonymous functional predictions	HGMD inherited disease mutations
dbSNP	HGMD inherited disease mutations
TCGA	HGMD inherited disease mutations
Complete Genomics	HGMD inherited disease mutations
TRANSFAC experimentally verified TFBS	HGMD inherited disease mutations
Predicted ChIP-Seq TFBS	HGMD inherited disease mutations
Predicted TFBSs in DNase hyper sensitivity regions	HGMD inherited disease mutations
CpG Islands	
Virtual Transcription Start Sites (TSSs)	
Microsatellites	
Post translational modifications	
miRNA	
dbSNV Variations in splice sites	
Disease associations	
Drug Targets	
Pathway Membership	
HGMD disease genes	
Orphanet	
OMIM	

4-1) 데이터베이스 목록

- 다종의 데이터베이스를 선택하여 검색한 경우, 특정 데이터베이스를 선택하여 결과를 확인할 수 있습니다.
- 해당 버튼 클릭하면 데이터 베이스 목록이 보여지며, 그 목록 중 하나를 선택하면 결과가 전환됩니다.

Result(Filtering)

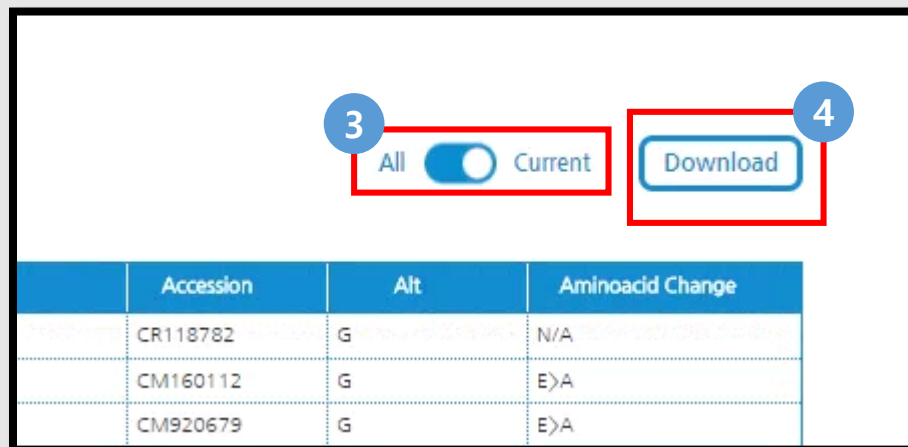
The screenshot shows the GenomeTrax interface. At the top, there is a search bar with the text "mutations". Below it is a table with columns: Feature End, Type, Genome, and Strand. A red box highlights the filter icon (three horizontal lines) in the Type column header, with a blue circle containing the number "2" next to it. A dropdown menu is open, showing the following options: Contains, Contains, Equals, Not equals, Starts with, and Ends with. The table contains several rows of data, all with "HGMD inherited disease mutations" in the Type column and "hg19" in the Genome column.

Feature End	Type	Genome	Strand
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-
7571752	HGMD inherited disease mutations	hg19	-

4-2) Column별 필터링

- 특정 결과를 필터링 하여 보고 싶을 때 사용합니다.
- 특정 열에 **마우스를 올리면** 메뉴(흰색 세줄)이 활성화 됩니다.
- 해당 버튼을 클릭하면 다양한 조건으로 검색이 가능합니다.
 - Contains : 해당 검색어를 포함
 - Equals : 검색어와 정확히 일치
 - Not equals : 해당 문자열과 불일치
 - Starts With : 검색어로 시작
 - Ends with : 검색어로 종결

Result(All/Current, Download)



	Accession	Alt	Aminoacid Change
	CR118782	G	N/A
	CM160112	G	E>A
	CM920679	G	E>A

4-3) All/Current 버튼

- 결과를 엑셀파일로 다운로드 할 때, 저장할 데이터베이스를 선택합니다.
- All** : 모든 데이터베이스가 **각각 시트별로 구분**되어 하나의 엑셀파일로 다운로드합니다.
- Current** : 현재 데이터베이스를 하나의 엑셀파일로 다운로드 받습니다.
 - 이때, **필터링 된 결과** 그대로 저장됩니다.

4-4) Download 버튼

- 검색한 결과를 **엑셀 파일로 다운로드** 받습니다.
- All/Current**에 따라 결과가 달라지며, Current의 경우 필터링 결과에 따라 저장되는 내용이 달라집니다.

E N D