Searching dbSNP in Entrez

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How to construct queries

dbSNP is part of NCBI's network of Entrez databases. As with these other databases, data of interest may be located simply by entering keywords into the <u>dbSNP</u> search box. The Advanced Search page, linked below the dbSNP search box, can assist in the construction of complex queries. To construct a complex query, specify the search terms, their fields, and the Boolean operations to perform on the terms using the following syntax:

term[field] OPERATOR term[field]

where term is the search terms, field is the search field, and OPERATOR is the Boolean operator ('AND', 'OR', 'NOT'; must be capitalized).

Field full name	Field aliases	Description	Search term values and rules	Example
All Fields	ALL, *	Search all searchable (indexed) fields	Asterisk (*) in the search term is not interpreted as a wildcard	SNV AND pathogenic
Base Position	POSITION, SNPPOS	Chromosome base position on GRCh38 (current)	A natural number representing the SNP's start coordinate on its chromosome on the latest assembly (ie. GRCh38). Most useful when search in combination with the CHR field.	<u>19956018[POSITI</u> ON] AND 8[CHR]
Base Position Previous	POSITION_GRCH 37, CHRPOS_PREV_ ASSM	Chromosome base position on GRCh37 (previous)	A natural number representing the SNP's start coordinate on its chromosome on the previous assembly (ie. GRCh37). Most useful when search in combination with the CHR field.	<u>19813529[POSITI</u> <u>ON_GRCH37]</u> <u>AND 8[CHR]</u>

Common Query fields and examples

Chromosome	CHR, CHRNUM	Chromosomes	One of 1-22, X, Y, MT	<u>7[CHR]</u>
Clinical Significance	CLIN	Variations with defined clinical effects or significances	16 search term values, defined for a relatively small subset of SNPs.	<u>"likely</u> pathogenic"[CLIN]
Filter	FILT, FLTR, SUBSET, SB, FIL	Limits the records returned	A variety of filters is available, including functional, positional, source, etc.	get all dbSNP records <u>"all[sb]"</u> or subsets <u>"splice 5</u> snp"[Filter]
Function Class	FXN, Function_class, FUNC, FUNCTION, FUNCTION_CLAS S	Function class	21 function classes are defined	<u>"frameshift"[Functio</u> <u>n Class]</u>
Gene Name	GENE, GENE_SYMBOL	Entrez Gene symbol	Corresponds to the Official Symbol field in the Entrez Gene resource	MAPK1[GENE]
Gene ID	GENE_ID	Entrez Gene UID	The numeric ID referencing the Entrez Gene ID	5594[GENE ID]
Global Minor Allele Frequency	GMAF	Minor Allele Frequency derived from global population (i.e., 1000G); can also be study-wide MAF that is not from global population	Most useful when entered as a range, as in the example	<u>(0.0[GMAF] :</u> 0.01[GMAF])
Project or Submitter Handle	HAN, PROJECT	Submitter Handle or Project Name	Submitter lab or project name including 1000Genomes, GnomAD, and DebNick	1000genomes[Sub mitter Handle] or 1000genomes[PR OJECT]
Reference SNP ID	RS, SNPID	Clustered SNP ID (rs)	The numeric ID must be prefixed with "rs". Also retrieves SNPs that have been merged into the specified SNP.	<u>rs328[RS]</u>
SNP Class	SCLS, SNPCLASS	SNP class	Possible values are: "del", "delins", "ins", "mnv", and "snv".	del[SNPCLASS]
Submitter SNP ID	SS, SSNUM	The ID assigned to each report of a SNP at submission time	Must be prefixed with "ss". Note that the query still returns Reference	<u>ss329[SS]</u>

			SNPs rather than Submitter SNPs.	
Validation Status	VALI, VALIDATE, VALIDATION	Validation status	Possible values	<u>"by</u>
			are: "by cluster" or	cluster"[Validation
			"by frequency"	Status]

Complex queries and others

Description	Query	Note
Variant allele with MAF = 0	"00000.0000"[Global Minor Allele Frequency]	variant allele is homozygous and may be due to differences between assembly versions
Pathogenic variants in BRCA1 with MAF < 0.01	<u>"pathogenic"[Clinical Significance]</u> AND BRCA1 AND 00000.0000:00000.00999[GLOBA L_MAF]	set GLOBAL_MAF range between 0 and 0.00999 for MAF < 0.01
Common variant (MAF => 0.01)	00000.0100[GLOBAL_MAF] : 00001.0000[GLOBAL_MAF]	set GLOBAL_MAF range from 0.0100 and 1.0000 for MAF => 0.01
1000Genomes common variant (MAF => 0.01) not found by TOPMED	"1000genomes"[Submitter Handle] NOT "topmed"[Submitter Handle] AND 00000.0100: 00001.0000[GLOBAL MAF]	set GLOBAL_MAF range from 0.0100 and 1.0000 for MAF => 0.01

HGVS Search

Users can search dbSNP with HGVS names, as shown with the example: $\underline{NM_000237.3:c.1421C>G~OR}$ $\underline{NG_013007.1:g.7147G>A}$