






Obtaining CVD variants from ClinVar database




Standard operating procedure
Nov 24, 2023

Download ClinVar disease names file from the project's github page:



Github: https://github.com/slavain/CVD_genomics






 **CVD_genomics** Public

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 **slavain** Added clinvar disease names table 0c789ab 5 minutes ago  **12** commits


	database_associations	Added GWAS associations obtained from GWAS catalog.	3 days ago
	CVDs_and_keywords.md	Update CVDs_and_keywords.md	last month
	disease_names	Added clinvar disease names table	5 minutes ago
	literature_master_list.csv	Add list of literature	2 weeks ago
	tasks.md	Updated tasks.md	3 days ago


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About

No description, website, or topics provided.

 Activity

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Releases

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Packages



Open the disease names file and search it for the term of interest, for example “hyperlipidemia”

29227	Hyperleucinemia	Human Phenotype Ontology	C0268576	HP:0010911	16 Feb 2016	Finding			
29228	Hyperlexia	MONDO	C1855928	MONDO:0009386	238350	16 Feb 2016	Disease		
29229	Hyperlipidemia	Human Phenotype Ontology	C0020473	HP:0003077		16 Feb 2016	Finding		
29230	Hyperlipidemia	MONDO	C0020473	MONDO:0021187		16 Feb 2016	Finding		
29231	Hyperlipidemia	associated with hepatomegaly				18 Feb 2020	Finding		
29232	Hyperlipidemia	due to hepatic triglyceride lipase deficiency			MONDO	C3151466	MONDO:0013533	614025	19 Apr 2022
29233	Hyperlipidemia, combined, 1	MONDO	C1865289	MONDO:0011237	602491	16 Feb 2016	Disease		
29234	Hyperlipidemia, combined, 2	MONDO	C1858308	MONDO:0011470	604499	16 Feb 2016	Disease		
29235	Hyperlipidemia, familial combined, LPL related	MONDO	C0020474	MONDO:0007759	144250	15 Jan 2021	Disease		
29236	Hyperlipidemia, familial combined, susceptibility to	NCBI curation			C4016424			28 Feb 2020	Disease
29237	Hyperlipoproteinemia	MONDO	C0020476	MONDO:0037748		16 Feb 2016	Disease		
29238	Hyperlipoproteinemia	Human Phenotype Ontology	C0020476	HP:0010980		16 Feb 2016	Disease		
29239	Hyperlipoproteinemia due to APOE1	NCBI curation				07 Sep 2023	Disease		
29240	Hyperlipoproteinemia type IV	NCBI curation			C0020480	144600	22 Apr 2020	Disease	
29241	Hyperlipoproteinemia, type 1D	MONDO	C4014767	MONDO:0014412	615947	19 Apr 2022	Disease		
29242	Hyperlipoproteinemia, type I		C0023817		238600	16 Feb 2016	Disease		

This step will give us an idea which relevant conditions are present in the database and which terms to use when searching ClinVar

Use the relevant terms found in the disease names files to search ClinVar database <https://www.ncbi.nlm.nih.gov/clinvar/> as shown on the screenshot. Here we are searching in phenotype/disease field

ClinVar

ClinVar  "hyperlipidemia"[Disease/Phenotype] 

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Hel

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Announcing changes to support somatic variant classifications

We have delayed changes to the ClinVar XML files and our submission spreadsheet templates until January 2024; these changes will improve support for classifications of somatic variants in ClinVar. To help our users and submitters prepare for this change, we are providing a preview of submission spreadsheet templates, updated XSDs, sample XMLs, and supporting documentation on [GitHub](#). Please share this information with your colleagues, including your bioinformatics team!

ClinVar is experiencing issues with search; some searches are failing and you may get inconsistent results at different times. We are working to get this resolved as quickly as possible. We apologize for the inconvenience, and we appreciate your patience.

Clinical
significance

Conflicting interpretations (17)

Benign (23)

Likely benign (14)

Uncertain significance (45)

Likely pathogenic (11)

Pathogenic (20)

Types of conflicts

P/LP vs LB/B (2)

P/LP vs VUS (8)

VUS vs LB/B (11)

Search results

[Display options](#) ▾ [Sort by Location](#) ▾ [Download](#) ▾

Items: 1 to 100 of 115

<< First < Prev Page **1** of 2 [Next >](#) [Last >>](#)

	Variation Location	Gene(s)	Protein change	Condition(s)	Clinical significance (Last reviewed)	Review status
<input type="checkbox"/>	NM_000157.4(GBA1):c.1093G>A (p.Glu365Lys)	GBA1, LOC106627981	E365K, E278K, E316K	not provided, not specified, Gaucher disease, Parkinson disease, late-onset Gaucher disease	Benign/Likely benign; risk factor (Oct 1, 2023)	criteria provided, multiple submitters, no conflicts
1.	GRCh37: Chr1:155206167 GRCh38: Chr1:155236376					

Select only pathogenic and likely pathogenic variants

Clinical significance

Conflicting interpretations (0)

Benign (0)

Likely benign (0)

Uncertain significance (0)

✓ Likely pathogenic (5)

→

✓ Pathogenic (10)

Molecular consequences

Frameshift (2)

Missense (9)

Nonsense (2)

Splice site (0)

ncRNA (1)

Near gene (0)

Search results

Display options ▾ Sort by Location ▾ Download ▾ items: 15

Filters activated: Pathogenic, Likely pathogenic. [Clear all](#) to show 26 items.

The following term was not found in ClinVar: clinsig established risk allele[Properties].

	Variation <i>Location</i>	Gene(s)	Protein change	Condition(s)	Clinical significance (Last reviewed)	
<input type="checkbox"/>	NM_000384.3(APOB):c.2249del (p.Met750fs) <i>GRCh37:</i> Chr2:21247992 <i>GRCh38:</i> Chr2:21025120	APOB	M750fs	Early-onset coronary artery disease	Likely pathogenic (Dec 29, 2022)	<div>Feedback</div> <div>subr</div>
<input type="checkbox"/>	NM_000384.3(APOB):c.1260del (p.Glu420fs)	APOB	E420fs	Early-onset coronary artery disease	Pathogenic (May 11, 2023)	

Download the results

Clinical
significance

Conflicting
interpretations (0)

Benign (0)

Likely benign (0)

Uncertain
significance (0)

✓ Likely
pathogenic (5)

✓ Pathogenic (10)

Molecular
consequen...

Frameshift (2)

Missense (9)

Nonsense (2)

Splice site (0)

ncRNA (1)

Search results

Display options Sort by Location Download Items: 15

Filters activated: P

The following term

Choose Destination

File

Download 15 items.

Format

Tabular (text)

Sort by

Location

Create File

	Variant	Location	Condition(s)	Clinical significance (Last reviewed)
<input type="checkbox"/>	NM_000384.3(APOR)			
1.	0fs)			
	GRCh37: Chr2:			
	GRCh38: Chr2:			
<input type="checkbox"/>	NM_000384.3(APOR):c.1260del (p.Glu42	APOR	Early-onset coronary artery disease	Likely pathogenic (Dec 29, 2022)
<input type="checkbox"/>	NM_000384.3(APOR):c.1260del (p.Glu42	APOR	Early-onset coronary artery disease	Pathogenic

Feedback