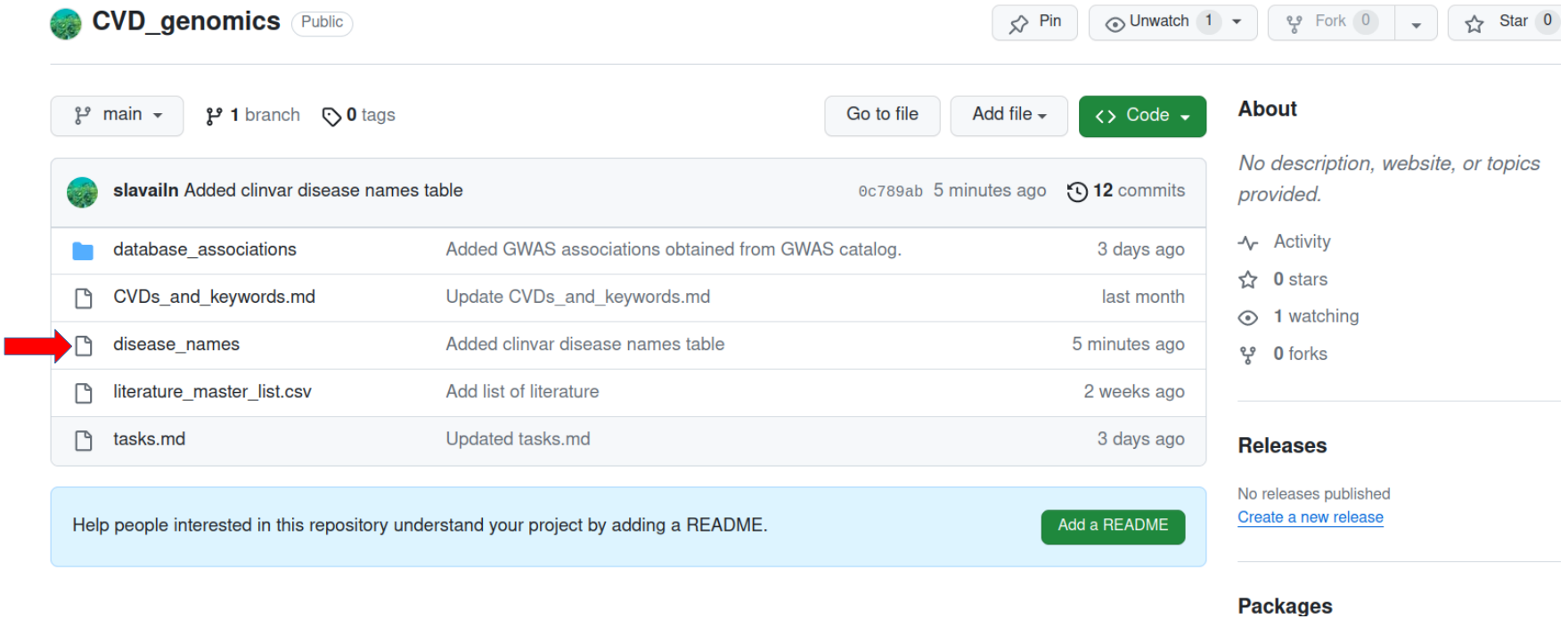
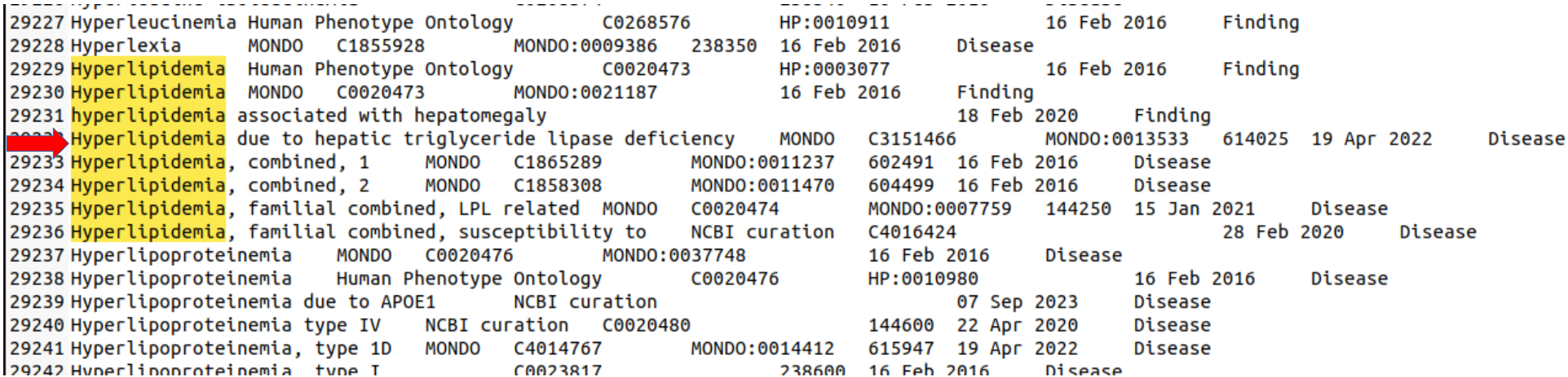
# Obtaining CVD variants from ClinVar database

Standard operating procedure
Nov 24, 2023

Download ClinVar disease names file from the project's github page:
Guthub: https://github.com/slavailn/CVD_genomics



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



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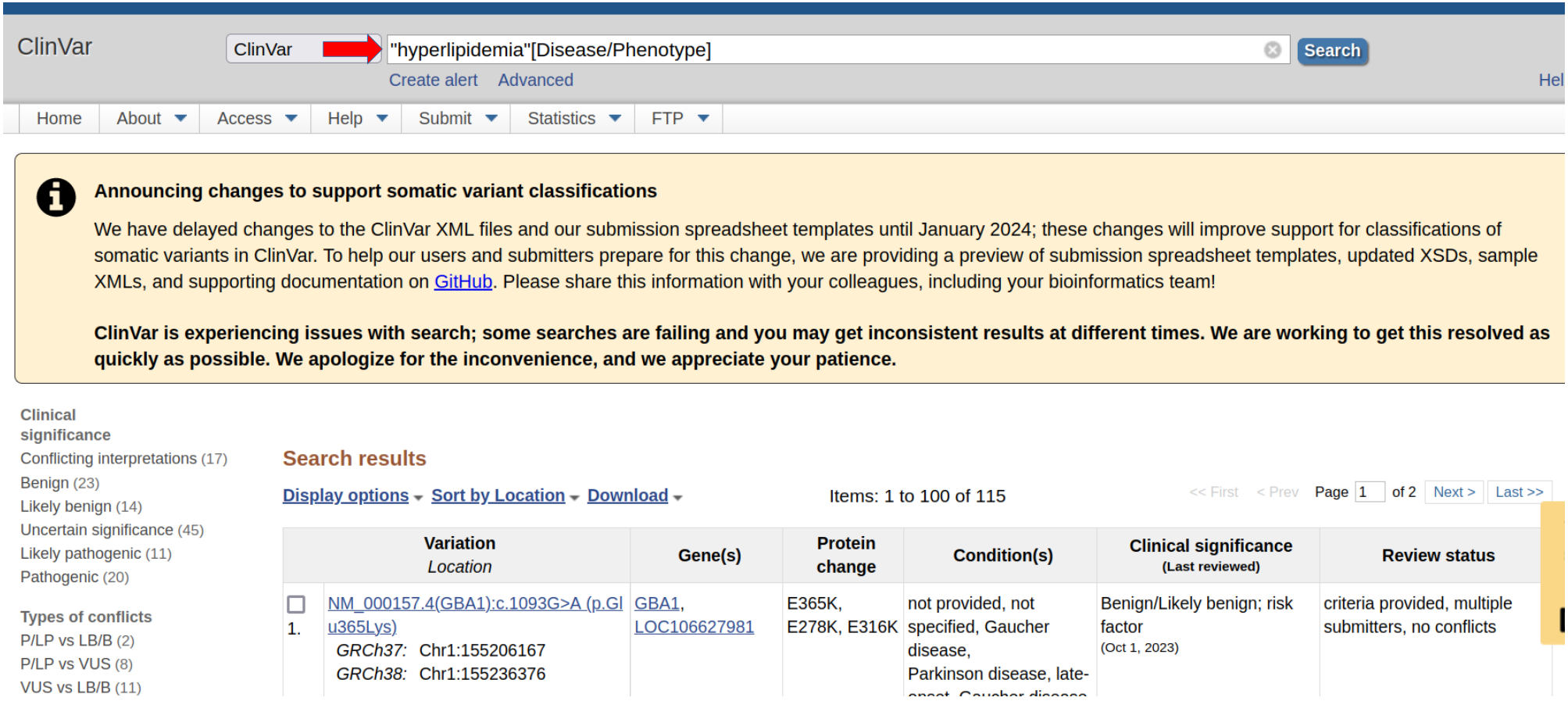
29227 Hyperleucinemia Human Phenotype Ontology        C0268576        HP:0010911              16 Feb 2016     Finding
29228 Hyperlexia      MONDO   C1855928        MONDO:0009386   238350  16 Feb 2016     Disease
29229 Hyperlipidemia  Human Phenotype Ontology        C0020473        HP:0003077              16 Feb 2016     Finding
29230 Hyperlipidemia  MONDO   C0020473        MONDO:0021187           16 Feb 2016     Finding
29231 hyperlipidemia associated with hepatomegaly                                       18 Feb 2020     Finding
      Hyperlipidemia due to hepatic triglyceride lipase deficiency    MONDO   C3151466        MONDO:0013533   614025  19 Apr 2022     Disease
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 whicn 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 searchin phenotype/disease field



# Select only pathogenic and likely pathogenic variants

**Clinical significance** clear

- 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)
- Near gene (0)

## Search results

Display options ▾ Sort by Location ▾ Download ▾ ems: 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 *Location* | Gene(s) | Protein change | Condition(s) | Clinical significance (Last reviewed) | |
|---|---|---|---|---|---|---|
| ☐ 1. | NM_000384.3(APOB):c.2249del (p.Met750fs) *GRCh37:* Chr2:21247992 *GRCh38:* Chr2:21025120 | APOB | M750fs | Early-onset **coronary artery disease** | Likely pathogenic (Dec 29, 2022) | |
| ☐ 2. | NM_000384.3(APOB):c.1260del (p.Glu420fs) | APOB | E420fs | Early-onset **coronary artery disease** | Pathogenic (May 11, 2023) | subr |

Feedback

# Download the results