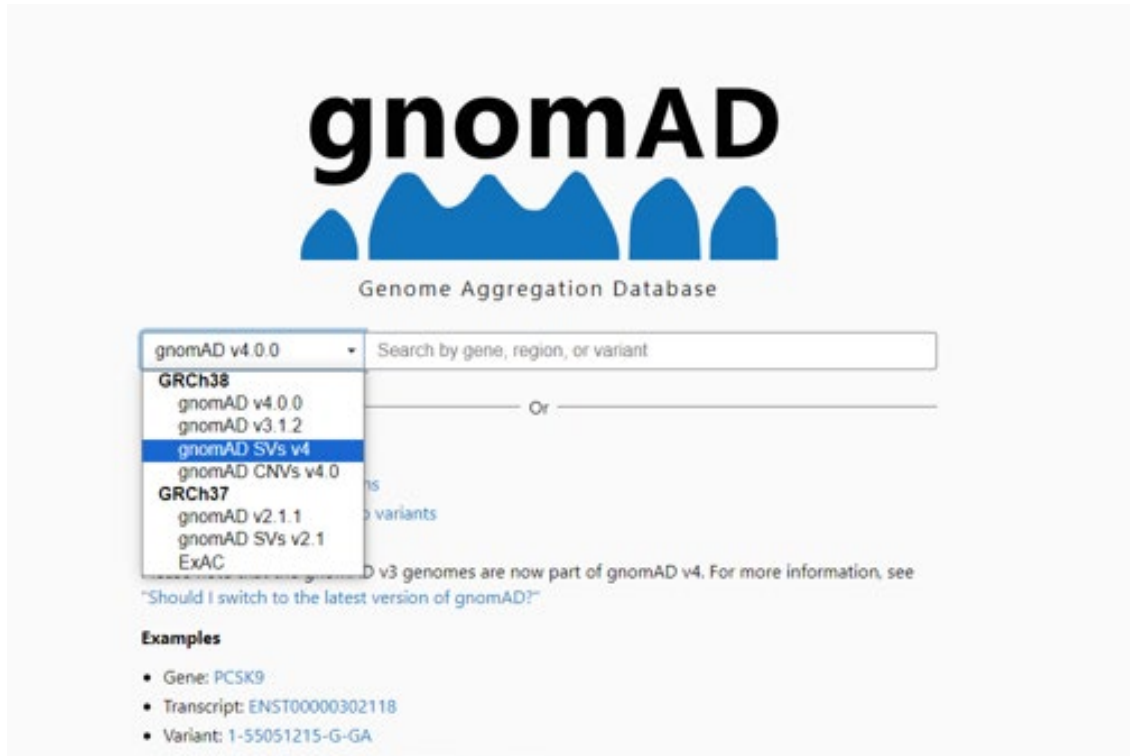


1. Go to <https://gnomad.broadinstitute.org/>
2. Click “gnomAD v4.0.0”
3. Click “gnomAD SVs v4” or “gnomAD SVs v2.1”



The screenshot shows the gnomAD website interface. At the top, the logo "gnomAD" is displayed in large black letters, with a blue wave-like graphic underneath. Below the logo, the text "Genome Aggregation Database" is visible. A search bar is present, with a dropdown menu open. The dropdown menu lists several options: "gnomAD v4.0.0", "GRCh38", "gnomAD v4.0.0", "gnomAD v3.1.2", "gnomAD SVs v4" (which is highlighted in blue), "gnomAD CNVs v4.0", "GRCh37", "gnomAD v2.1.1", "gnomAD SVs v2.1", and "ExAC". Below the search bar, there is a section titled "Examples" with a list of items: "Gene: PCSK9", "Transcript: ENST00000302118", and "Variant: 1-55051215-G-GA".

4. Enter the genomic coordinates

gnomAD
Genome Aggregation Database

gnomAD SVs v4 | 16-14688342-14711143

Or

- [Download gnomAD data](#)
- [Read gnomAD publications](#)
- [Find co-occurrence of two variants](#)

Please note that the gnomAD v3 genomes are now part of gnomAD v4. For more information, see

5. Structural variants (SVs) within this region are displayed including gains, losses and rearrangements of at least 50 nucleotides.

16-14688342-14711143 Change Dataset: gnomAD v4.0.0 | gnomAD SVs v4.0

Genome build: GRCh38 / hg38
Region size: 22,802 BP
External resources: [UCSC Browser](#)

Zoom in: 1.5x 3x 10x | Zoom out: 1.5x 3x 10x

Metric: Over 20 | Save plot

Include non-coding genes:

Z Score: not constrained (2.16) to constrained (4.0)

Color variants by: Consequence | Class

Consequences

pLoF only | Int. exon duplication only | Copy gain only | Other only | all

Classes

DEL only | DUP only | MCNV only | INS only | INV only | CPX only | OTH only | all

Include filtered variants

[Export variants to CSV](#) | [Configure table](#) | [Search variant table](#)

Variant ID	Source	Consequence	Class	Position	Size	Allele Count	Allele Number	Allele Frequency	Number of Homozygotes
DEL_CHR16_0A24707A	g	loss of function	deletion	14688000 - 14713450	25.4 kb	18177	126092	1.44e-1	8
DEL_CHR16_2C8C9DCF	g	loss of function	deletion	14544000 - 14772000	228 kb	2	125802	1.60e-5	0
DUP_CHR16_1010F44F	g	promoter	duplication	14695000 - 14705000	10.0 kb	3950	83454	4.73e-2	772
DUP_CHR16_AB38A33A	g	copy gain	duplication	14688000 - 14786000	98.0 kb	170	107948	1.59e-3	4
DUP_CHR16_B9198C6A	g	TSS duplication	duplication	14689000 - 14696000	7.00 kb	2830	76372	3.71e-2	487
DUP_CHR16_C29A3847	g	copy gain	duplication	14540000 - 15030100	490 kb	5	124612	4.00e-5	1
DUP_CHR16_D5227FFF	g	copy gain	duplication	14688000 - 14767500	79.5 kb	4422	87042	5.08e-2	833
INV_CHR16_63DFF50A	g	loss of function	inversion	8192848 - 17114213	8.92 Mb	2	126092	1.60e-5	1

Structural variants in gnomAD can be displayed in UCSC browser. For more information, go to [XXX \(link to Casey's document\)](#).

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