

Derived/Composite Nucleotide Sequence Databases

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ClinVar

- ❑ Is a freely accessible, public archive of reports of human variations classified for diseases and drug responses
- ❑ Processes submissions reporting variants found in patient samples
- ❑ The variants described in submissions are mapped to reference sequences, and reported according to the HGVS standard
- ❑ ClinVar works in collaboration with interested organizations to meet the needs of the medical genetics community
- ❑ How to search in ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar/docs/help/>)
- ❑ Linked to
 - dbVar
 - Graphics view of GenBank
 - NCBI's Variation Viewer
 - ClinGen (Allele identifier)

ClinVar

❑ Searching in ClinVar: (Filtering on left side based on variation type, size, clinical significance)

→ **Gene:** Variations related to that gene e.g., BRCA1, zeb2, BRCA2

→ **HGVS:** Variation ID (AA or nucleotide)

→ **Condition/Diseases and Phenotypes:** Celiac disease, cystic fibrosis, breast and ovarian cancer, phenylketonuria, sickle cell

Variation Report:

- Variation: Name of variants
- Gene: variant present in which gene
- Condition: conditions for which variants are interpreted
- Frequency: Allele frequency
- Clinical significance: Pathogenic, benign, non significant
- After exploring a particular gene:
 - Variant Details: Variation ID, Accession ID, Type of variation, Location, HGVS/Human Genome Variation Society. Provides variation in the Variation viewer from location and Graphics of GenBank format from HGVS
 - Genes: Other genes affected by the variation in searched gene and conditions associated with the variations can be explore

ClinVar

Variant Details

Identifiers:

GRCh38/hg38 17q23.1-25.1(chr17:36449220-75053130)x3

Variation ID: 148363 Accession: VCV000148363.2

Type and length:

copy number gain, 15,534,756 bp

Location:

Cytogenetic: 17q23.1-25.1 17: 36449220-75053130 (GRCh38) [NCBI UCSC] 17: 57595736-73049225 (GRCh37) [NCBI UCSC] 17: 54950518-70560820 (NCBI36) [NCBI UCSC]

Timeline in ClinVar:

	First in ClinVar ⓘ	Last submission ⓘ	Last evaluated ⓘ
Germline	Oct 26, 2017	Oct 26, 2017	Apr 21, 2011

HGVS:

Nucleotide	Protein	Molecular consequence
NC_000017.11:g.(?_36449220)_(75053130_?)dup		
NC_000017.10:g.(?_57595736)_(73049225_?)dup		
NC_000017.9:g.(?_54950518)_(70560820_?)dup		

Protein change:

-

Other names:

-

Canonical SPDI:

Functional consequence ⓘ: -

Global minor allele frequency (GMAF) ⓘ:

-








Allele frequency ⓘ:

-

Links :

dbVar: nssv1415284
dbVar: nsv817369
VarSome

Genes

Gene	OMIM	ClinGen Gene Dosage Sensitivity Curation		Variation Viewer 	Related variants	
		HI score 	TS score 		Within gene 	All 
AXIN2		Sufficient evidence for dosage pathogenicity	No evidence available	GRCh38 GRCh37	3638	3652
BPTF		Sufficient evidence for dosage pathogenicity	No evidence available	GRCh38 GRCh37	687	735

Variation Viewer

Variation Viewer

New to Variation Viewer? Read our quick overview! X

Homo sapiens (human)

Assembly: GRCh38.p14 (GCF_000001405.40) - Chr 17 (NC_000017.11)

Search assembly
Location, gene or phenotype
Examples ▶

▶ Pick Assembly
▶ Tracks and User Data
▶ History
▶ Assembly Region Details

NC_000017.11: 36,449,220 - 75,053,130

TBC1D3F

Exon Navigator: There are too many (1188) genes in the region. Please narrow the region to enable exon navigation.

NC_000017.11

NCBI RefSeq Annotation GCF_000001405.40-RS_2023_10

Clinical, dbSNP b156 v2

Live RefSNPs, dbSNP b156 v2

dbVar Clinical Structural Variants (nstd102)

dbVar Non-Pathogenic Clinical Structural Variants (subset of nstd102)

dbVar Pathogenic Clinical Structural Variants (subset of nstd102)

ClinVar variants with precise endpoints

NC_000017.11: 36M..75M (38,603,911 nt)

Clinical variation →

Gene →

Variation Data

Filter by

Source database

☐ dbSNP (14214379)

☐ dbVar (110961)

In ClinVar

☐ Yes (40871)

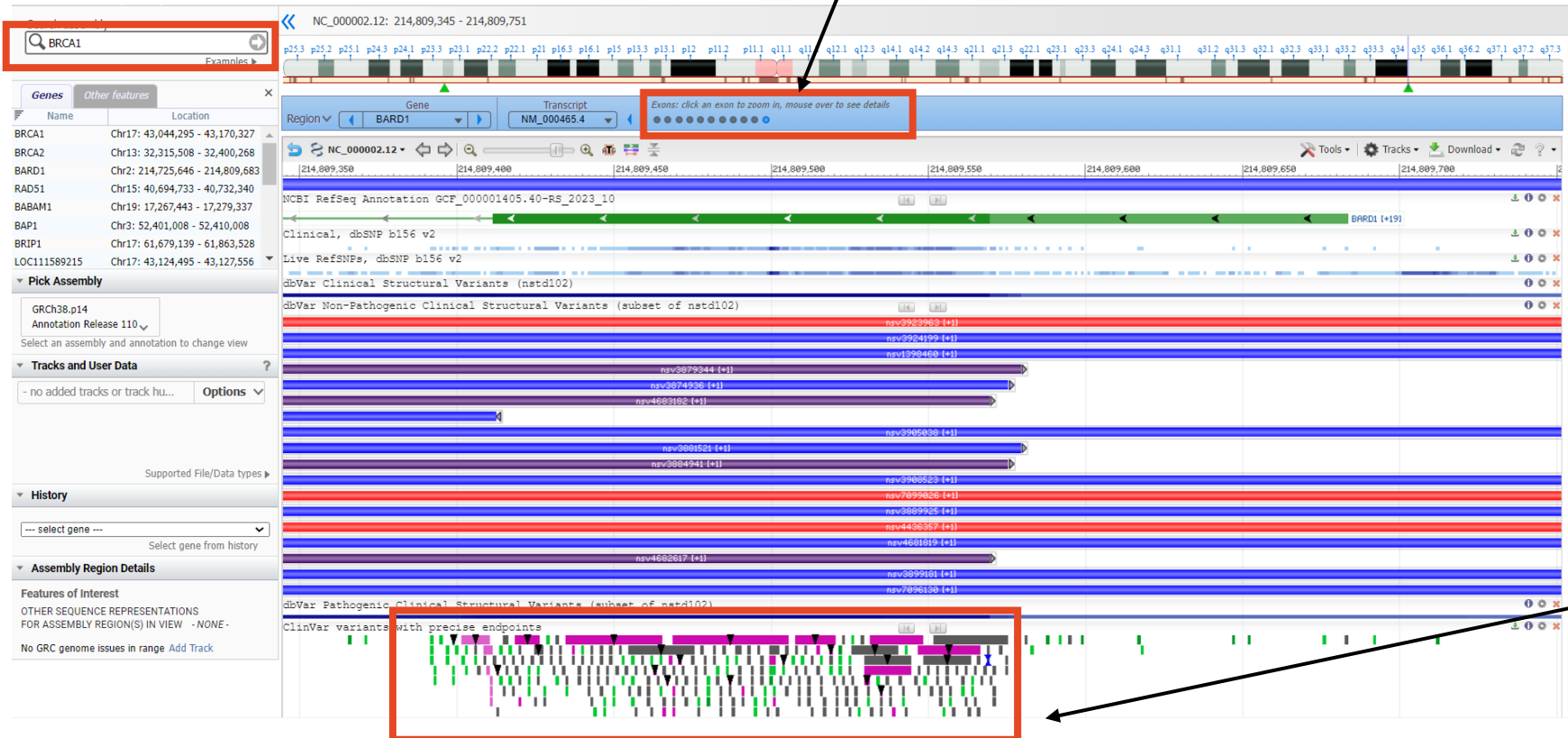
☐ No (14284469)

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Variant ID	Location	Variant type	Gene	Molecular consequences	Most severe clinical significance
nsv3907261	150,733 - 83,084,062	copy number variation	ASIC2 and 2369 more		Pathogenic
nsv984832	150,733 - 83,091,923	copy number variation	ASIC2 and 2437 more		
nsv3899740	157,423 - 83,100,564	copy number variation	ASIC2 and 2369 more		Pathogenic
nsv3903684	158,756 - 83,102,004	copy number variation	ASIC2 and 2369 more		Pathogenic
nsv3908245	162,553 - 83,100,251	copy number variation	ASIC2 and 2369 more		Pathogenic
esv3813934	575,274 - 42,221,597	copy number variation	ASIC2 and 1309 more		
esv3814112	580,529 - 45,752,844	inversion	ASIC2 and 1493 more		
esv3804582	594,470 - 42,249,254	copy number variation	ASIC2 and 1309 more		

Variation Viewer

Zoom



Variations in Purple