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

→ NCBIs Variation Viewer

→ ClinGen (Allele identifier)

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- **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

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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) : -

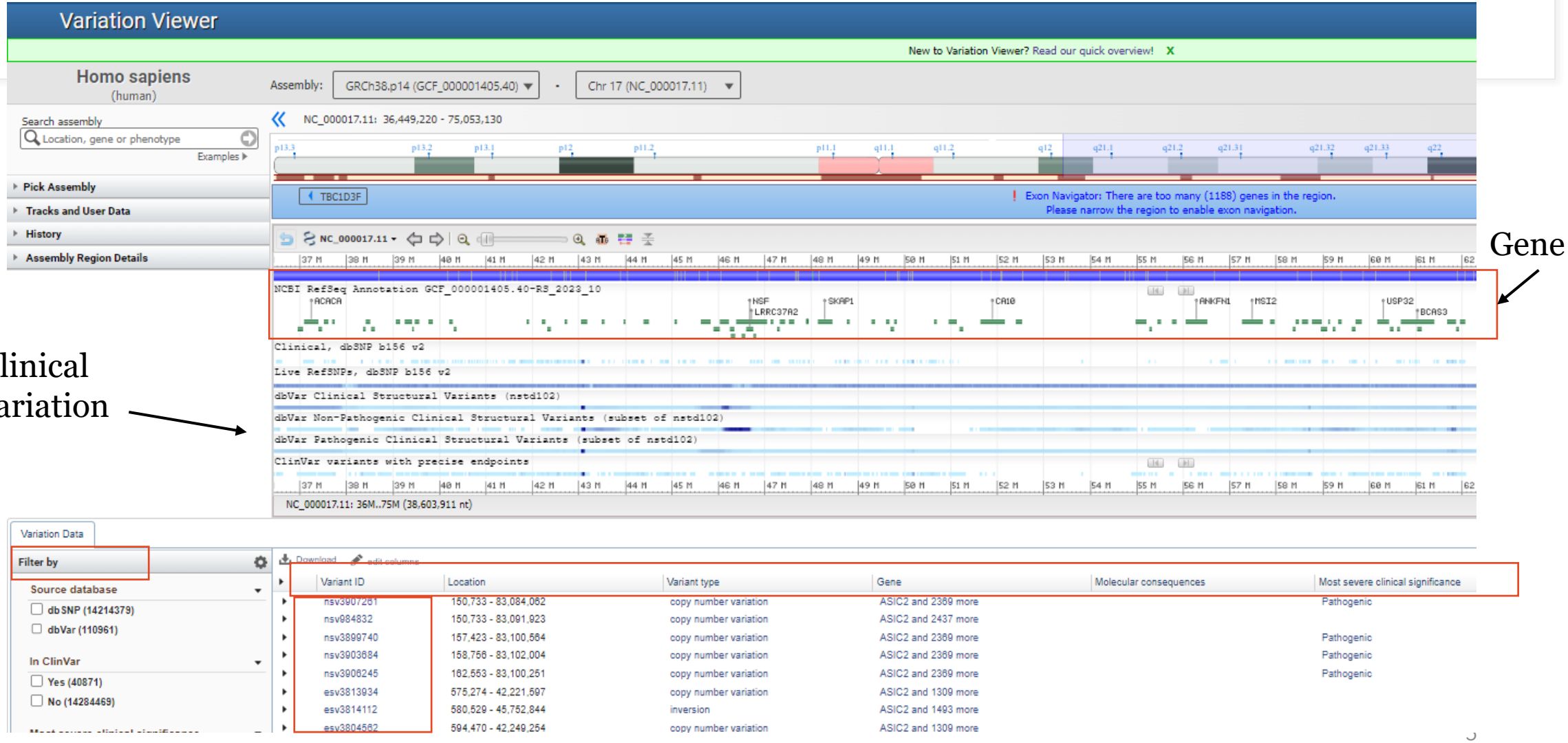
Allele frequency : -

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