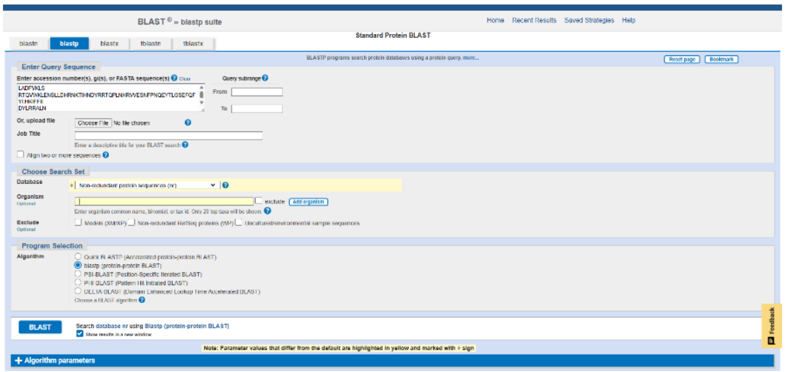
Rešitev seminarja:

1. **Navedi ime proteina in organizem iz katerega izvira.**
   Zaporedje vnesemo v pBLAST in podatkovno baro nastavimo na »non-redundant protein sequences (nr)«. Izberemo zadetek, ki se 100% ujema. Razberemo da je ime proteina **carbonic anhydrase 6 isoform 1 precursor-**karbonska anhidraza 6 organizem iz katerega izvira je **Homo sapiens-**človek.



Enter accession number(s), gi(s), or FASTA sequence(s): LADFVKLS RTQVWKLENSLLDHRNKTIHNDYRRTQPLNHRVVESNFPNQEYTLGSEFQF YIHKFFI DYLRRALN

Database: Non-redundant protein sequences (nr)

Algorithm: ● blastp (protein-protein BLAST)

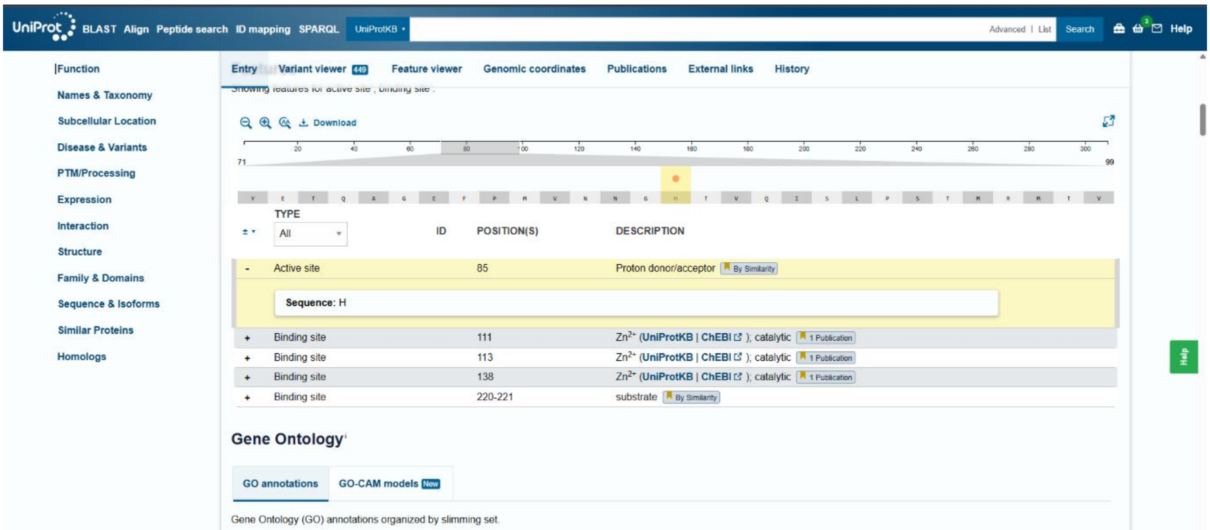
Search database nr using Blastp (protein-protein BLAST)

Note: Parameter values that differ from the default are highlighted in yellow and marked with ♦ sign

carbonic anhydrase 6 isoform 1 precursor [Homo sapiens]

NCBI Reference Sequence: NP_001206.2

2. **Ali ima protein aktivno mesto. Če ja navedi kateri aminokislinski ostanek to je in na katerem mestu se nahaja? Če bi želeli protein deaktivirati, v kateri aminokislinski ostanek bi zmutirali aktivno mesto?**
   Ime proteina vnesemo v UniProt in kot organizem izberemo Homo sapiens. Pod odsekom Function najdemo aktivno mesto, ki je H (histidin) 85. Da deaktiviramo aktivno mesto lahko uvedeno mutacijo npr. H85A.

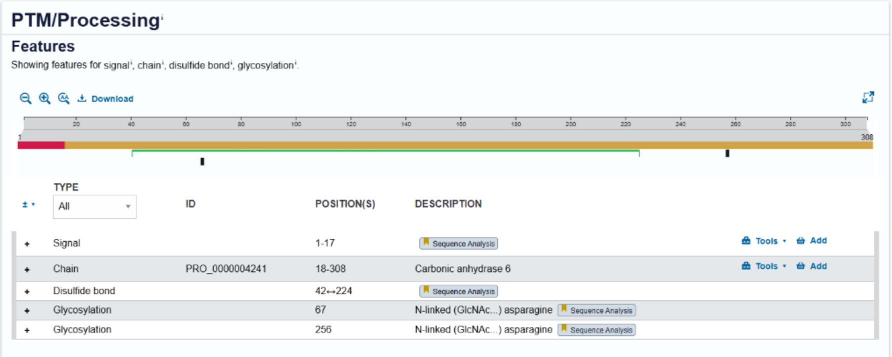


TYPE	ID	POSITION(S)	DESCRIPTION
Active site		85	Proton donor/acceptor By Similarity
Sequence: H			
Binding site		111	Zn^{2+} (UniProtKB | ChEBI); catalytic 1 Publication
Binding site		113	Zn^{2+} (UniProtKB | ChEBI); catalytic 1 Publication
Binding site		138	Zn^{2+} (UniProtKB | ChEBI); catalytic 1 Publication
Binding site		220-221	substrate By Similarity

Gene Ontology

Gene Ontology (GO) annotations organized by slimming set.

3. **Med katerima aminokislinskima ostankoma (ime in pozicija) se tvori disulfidna vez? Ali so prisotne tudi kakšne druge posttranslacijske modifikacije?**
   Gremo pod PTM/Processing. Vidimo da je disulfidna vez med C42-C224 (cistein). Omenjena posttranslacijska modifikacija je še glikozilacija.



PTM/Processing

Features

Showing features for signal, chain, disulfide bond, glycosylation.

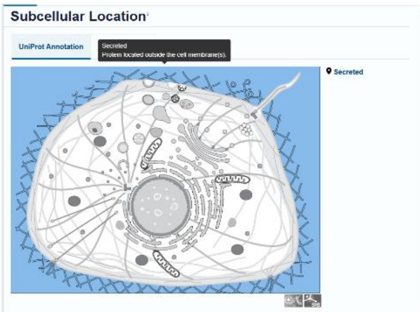
TYPE	ID	POSITION(S)	DESCRIPTION
Signal		1-17	Sequence Analysis
Chain	PRO_0000004241	18-308	Carbonic anhydrase 6
Disulfide bond		42↔224	Sequence Analysis
Glycosylation		67	N-linked (GlcNAc...) asparagine Sequence Analysis
Glycosylation		256	N-linked (GlcNAc...) asparagine Sequence Analysis

4. **Navedi kateri ioni so potrebni za delovanje.**
   Protein veže cinkove ione.

Cofactor

Zn^{2+} (UniProtKB | Rhea | CHEBI:29105) By Similarity

5. **Kje se ta protein nahaja?**
   Gremo pod razdelek Subcellular location. Vidimo, da je protein sekretorni protein in se nahaja v ekstracelularni regiji.

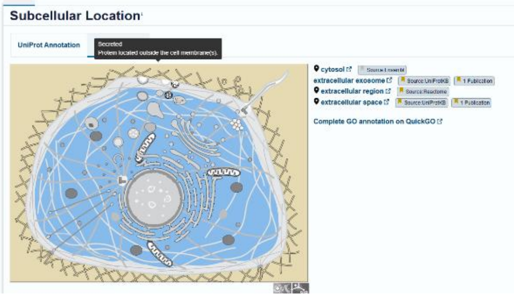


Subcellular Location

UniProt Annotation

Secreted
Protein located outside the cell membrane(s).

Secreted



Subcellular Location

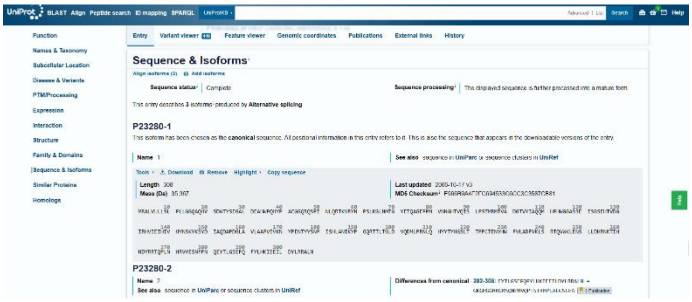
UniProt Annotation

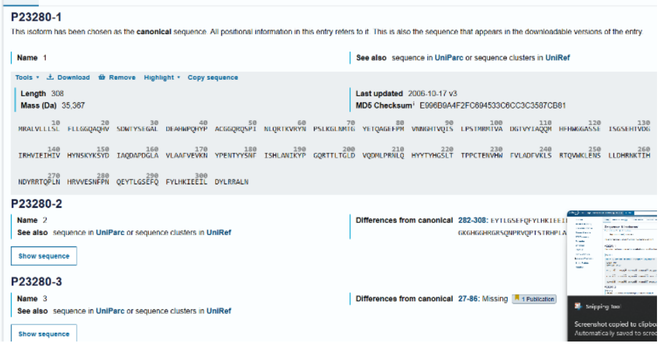
Secreted
Protein located outside the cell membrane(s).

cytosol Source:Ensembl
extracellular exosome Source:UniProtKB 1 Publication
extracellular region Source:Reactome
extracellular space Source:UniProtKB 1 Publication

Complete GO annotation on QuickGO

6. **Ali obstaja kakšen alterenativen "splicing"? Če ja primerjaj in komentiraj izooblike (naredi poravnavo).**
   Gremo pod razdelek Sequence & Isoforms. Vidimo da obstajajo tri izooblike. Vse damo v košarico izberemo in naredimo poravnavo. Opazimo da so zaporedja skoraj identična. Opazimo, da izoobliki 3 manjka odsek 27-86 in da pri izoobliki 2 pride do drugega zaporedja 282-313, ki je na koncu tudi daljše od ostalih dveh.





P23280-1

This isoform has been chosen as the **canonical** sequence. All positional information in this entry refers to it. This is also the sequence that appears in the downloadable versions of the entry.

Name 1

See also sequence in UniParc or sequence clusters in UniRef

Length 308
Mass (Da) 35,367

Last updated 2006-10-17 v3
MD5 Checksum E996B9A4F2FC694533C6CC3C3587CB81

```
        10         20         30         40         50         60         70         80         90        100        110        120        130
MRALVLLLSL FLLGGQAQHV SDWTYSEGAL DEAHWPQHYP ACGGQRQSPI NLQRTKVRYN PSLKGLNMTG YETQAGEFPM VNNGHTVQIS LPSTMRMTVA DGTVYIAQQM HFHWGGASSE ISGSEHTVDG

       140        150        160        170        180        190        200        210        220        230        240        250        260
IRHVIEIHIV HYNSKYKSYD IAQDAPDGLA VLAAFVEVKN YPENTYYSNF ISHLANIKYP GQRTTLTGLD VQDMLPRNLQ HYYTYHGSLT TPPCTENVHW FVLADFVKLS RTQVWKLENS LLDHRNKTIH

       270        280        290        300
NDYRRTQPLN HRVVESNFPN QEYTLGSEFQ FYLHKIEEIL DYLRRALN
```

P23280-2

Name 2
See also sequence in UniParc or sequence clusters in UniRef

Differences from canonical 282-308: EYTLGSEFQFYLHKIEEI
GKGHGGHRGRSQNPRVQPTSTRHPLA

Show sequence

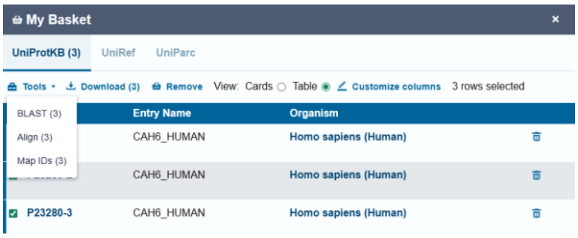
P23280-3

Name 3
See also sequence in UniParc or sequence clusters in UniRef

Differences from canonical 27-86: Missing 1 Publication

Show sequence

Snipping Tool
Screenshot copied to clipbo
Automatically saved to scre



	Entry Name	Organism
	CAH6_HUMAN	Homo sapiens (Human)
	CAH6_HUMAN	Homo sapiens (Human)
☑ P23280-3	CAH6_HUMAN	Homo sapiens (Human)

```
☐ sp|P23280-2|CAH6_HUMAN  MRALVLLLSLFLLGGQAQHVSDWTYSEGALDEAHWPQHYPACGGQRQSPINLQRTKVRYNPSLKGL  66
☐ sp|P23280|CAH6_HUMAN    MRALVLLLSLFLLGGQAQHVSDWTYSEGALDEAHWPQHYPACGGQRQSPINLQRTKVRYNPSLKGL  66
☐ sp|P23280-3|CAH6_HUMAN  MRALVLLLSLFLLGGQAQHVSDWTYS----------------------------------------  26

                P23280-2:Signal

☐ sp|P23280-2|CAH6_HUMAN  NMTGYETQAGEFPMVNNGHTVQISLPSTMRMTVADGTVYIAQQMHFHWGGASSEISGSEHTVDGIR  132
☐ sp|P23280|CAH6_HUMAN    NMTGYETQAGEFPMVNNGHTVQISLPSTMRMTVADGTVYIAQQMHFHWGGASSEISGSEHTVDGIR  132
☐ sp|P23280-3|CAH6_HUMAN  --------------------VQISLPSTMRMTVADGTVYIAQQMHFHWGGASSEISGSEHTVDGIR  72

                P23280-2:Signal

☐ sp|P23280-2|CAH6_HUMAN  HVIEIHIVHYNSKYKSYDIAQDAPDGLAVLAAFVEVKNYPENTYYSNFISHLANIKYPGQRTTLTG  198
☐ sp|P23280|CAH6_HUMAN    HVIEIHIVHYNSKYKSYDIAQDAPDGLAVLAAFVEVKNYPENTYYSNFISHLANIKYPGQRTTLTG  198
☐ sp|P23280-3|CAH6_HUMAN  HVIEIHIVHYNSKYKSYDIAQDAPDGLAVLAAFVEVKNYPENTYYSNFISHLANIKYPGQRTTLTG  138

                P23280-2:Signal

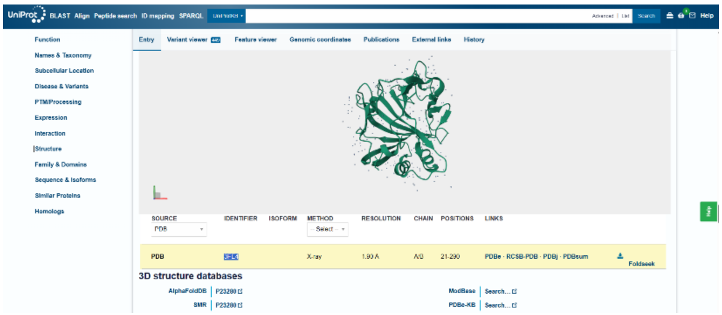
☐ sp|P23280-2|CAH6_HUMAN  LDVQDMLPRNLQHYYTYHGSLTTPPCTENVHWFVLADFVKLSRTQVWKLENSLLDHRNKTIHNDYR  264
☐ sp|P23280|CAH6_HUMAN    LDVQDMLPRNLQHYYTYHGSLTTPPCTENVHWFVLADFVKLSRTQVWKLENSLLDHRNKTIHNDYR  264
☐ sp|P23280-3|CAH6_HUMAN  LDVQDMLPRNLQHYYTYHGSLTTPPCTENVHWFVLADFVKLSRTQVWKLENSLLDHRNKTIHNDYR  204

                P23280-2:Signal

☐ sp|P23280-2|CAH6_HUMAN  RTQPLNHRVVESNFPNQGKGHGGHRGRSQNPRVQ-PTSTRHPLALGSLEA                 313
☐ sp|P23280|CAH6_HUMAN    RTQPLNHRVVESNFPNQEYTLGSEFQFYLHKIEEILDYLRRALN------                 308
☐ sp|P23280-3|CAH6_HUMAN  RTQPLNHRVVESNFPNQEYTLGSEFQFYLHKIEEILDYLRRALN------                 248

                P23280-2:Signal
```

7. **Zapiši pdb kodo tega proteina.**
   Gremo pod razdelek Structure in kot vir izberemo PDB. PDB koda je 3FE4.

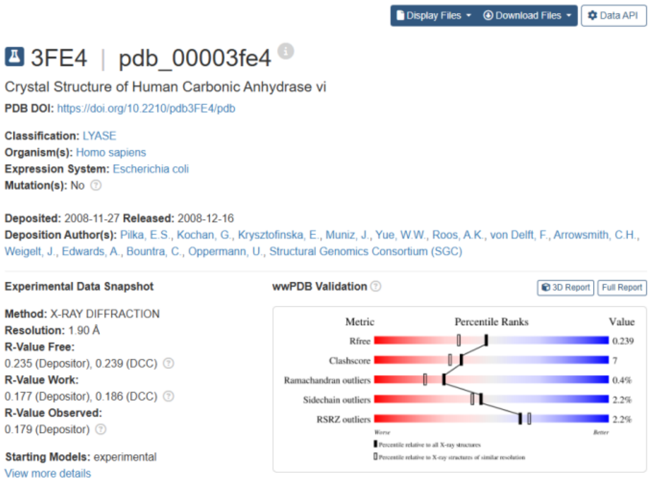


SOURCE	IDENTIFIER	ISOFORM	METHOD	RESOLUTION	CHAIN	POSITIONS	LINKS	
PDB			-- Select --					
PDB	[illegible]		X-ray	1.90 Å	A/B	21-290	PDBe · RCSB-PDB · PDBj · PDBsum	Foldseek

3D structure databases

AlphaFoldDB	P23280	ModBase	Search...
SMR	P23280	PDBe-KB	Search...

8. **Navedi ekspresijski sistem in datum izdaje.**
   ekspresijski sistem: Escherichia coli
   datum izdaje: 2008-12-16



3FE4 | pdb_00003fe4

Crystal Structure of Human Carbonic Anhydrase vi

PDB DOI: https://doi.org/10.2210/pdb3FE4/pdb

Classification: LYASE
Organism(s): Homo sapiens
Expression System: Escherichia coli
Mutation(s): No

Deposited: 2008-11-27 **Released:** 2008-12-16
Deposition Author(s): Pilka, E.S., Kochan, G., Krysztofinska, E., Muniz, J., Yue, W.W., Roos, A.K., von Delft, F., Arrowsmith, C.H., Weigelt, J., Edwards, A., Bountra, C., Oppermann, U., Structural Genomics Consortium (SGC)

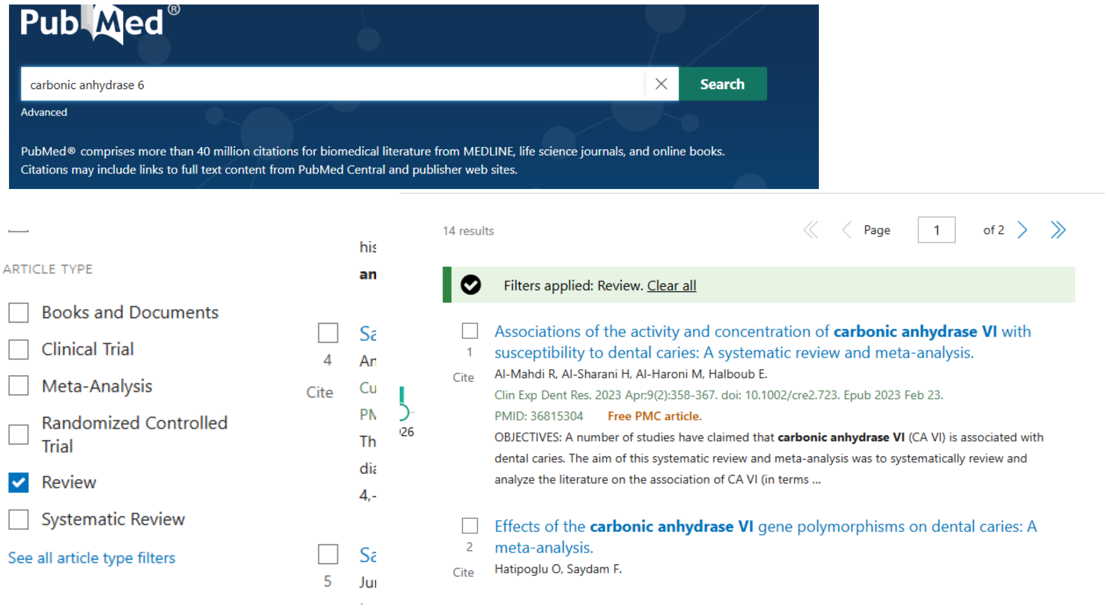
Experimental Data Snapshot

Method: X-RAY DIFFRACTION
Resolution: 1.90 Å
R-Value Free:
0.235 (Depositor), 0.239 (DCC)
R-Value Work:
0.177 (Depositor), 0.186 (DCC)
R-Value Observed:
0.179 (Depositor)

Starting Models: experimental
View more details

wwPDB Validation

9. **Pojdi na PubMed in išči zadetke, če v iskalno vrstico vneseš ime proteina (iz UniProta). Katero je bilo prvo leto, ko je bil objavljen članek na to temo? Koliko preglednih člankov je bilo napisno na to temo?**
   Prvo leto: 1964
   Pregledni čanki- nastavimo filter Review: 14
   Pripomba: do časa reševanja se ti podatki lahko spremenijo!



PubMed® comprises more than 40 million citations for biomedical literature from MEDLINE, life science journals, and online books. Citations may include links to full text content from PubMed Central and publisher web sites.

14 results — Page 1 of 2

Filters applied: Review. Clear all

ARTICLE TYPE

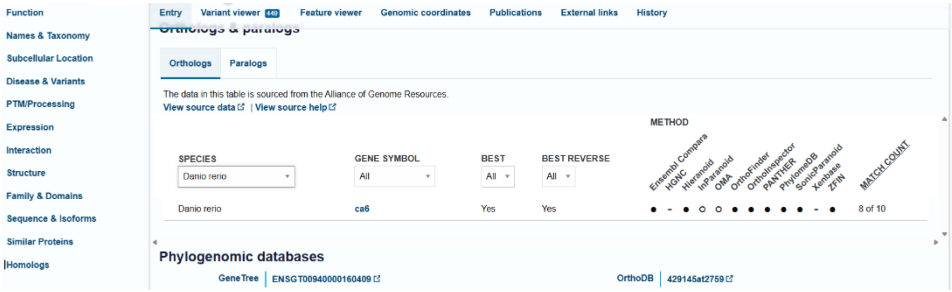
☐ Books and Documents
☐ Clinical Trial
☐ Meta-Analysis
☐ Randomized Controlled Trial
☑ Review
☐ Systematic Review

See all article type filters

☐ 1 Cite — Associations of the activity and concentration of **carbonic anhydrase VI** with susceptibility to dental caries: A systematic review and meta-analysis.
Al-Mahdi R, Al-Sharani H, Al-Haroni M, Halboub E.
Clin Exp Dent Res. 2023 Apr;9(2):358-367. doi: 10.1002/cre2.723. Epub 2023 Feb 23.
PMID: 36815304 **Free PMC article.**
OBJECTIVES: A number of studies have claimed that **carbonic anhydrase VI** (CA VI) is associated with dental caries. The aim of this systematic review and meta-analysis was to systematically review and analyze the literature on the association of CA VI (in terms ...

☐ 2 Cite — Effects of the **carbonic anhydrase VI** gene polymorphisms on dental caries: A meta-analysis.
Hatipoglu O, Saydam F.

10. **Poišči homolog *Danio rerio*. Izberi zadetek z daljšim aminokislinskim zaporedjem (538AA). Naredi poravnavo z našim izvornim proteinom. Komentiraj vezavna mesta in aktivno mesto homologa. Ali so enaka kot pri našem proteinu?**
    Gremo pod razdeleg Homologs in izberemo tega ki pripada organizu *Danio rerio*. Nato kot piše v navodilu izberemo tistega z 538AA dolgim zaporedjem. Naredimo poravnavo z človeškim proteinom. Pod select annotation za dobro anotiran protein (človeški) izberen najprej active site in nato binding site. Opazim, da se pri obeh vse ujema.

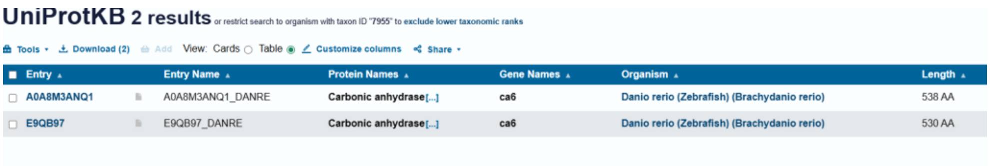


The data in this table is sourced from the Alliance of Genome Resources.
View source data | View source help

SPECIES	GENE SYMBOL	BEST	BEST REVERSE	Ensembl Compara	HGNC	Hieranoid	InParanoid	OMA	OrthoFinder	OrthoInspector	PANTHER	PhylomeDB	SonicParanoid	Xenbase	ZFIN	MATCH COUNT
Danio rerio	All	All	All													
Danio rerio	ca6	Yes	Yes	●	-	●	○	○	●	●	●	●	●	-	●	8 of 10

Phylogenomic databases

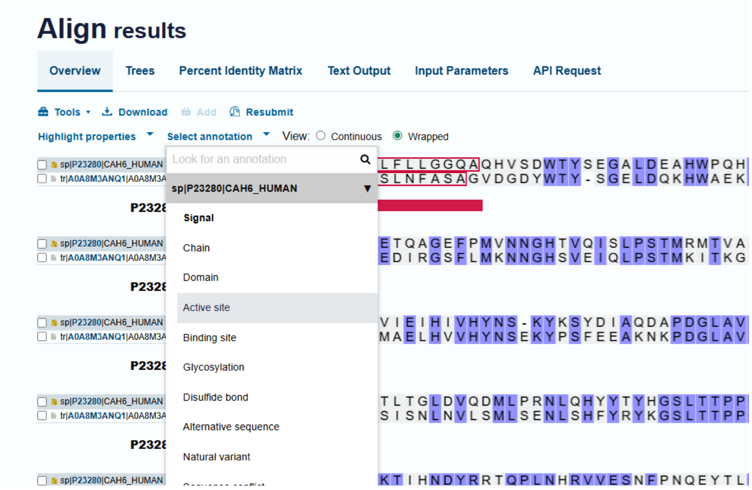
GeneTree | ENSGT00940000160409
OrthoDB | 429145at2759



UniProtKB 2 results or restrict search to organism with taxon ID "7955" to exclude lower taxonomic ranks

View: Cards ○ Table ●

Entry	Entry Name	Protein Names	Gene Names	Organism	Length
A0A8M3ANQ1	A0A8M3ANQ1_DANRE	Carbonic anhydrase[...]	ca6	Danio rerio (Zebrafish) (Brachydanio rerio)	538 AA
E9QB97	E9QB97_DANRE	Carbonic anhydrase[...]	ca6	Danio rerio (Zebrafish) (Brachydanio rerio)	530 AA



Align results

View: ○ Continuous ● Wrapped

```
☐ sp|P23280|CAH6_HUMAN   L F L L G G Q A Q H V S D W T Y S E G A L D E A H W P Q H
☐ tr|A0A8M3ANQ1|A0A8M3A  S L N F A S A G V D G D Y W T Y - S G E L D Q K H W A E K

P2328

☐ sp|P23280|CAH6_HUMAN   E T Q A G E F P M V N N G H T V Q I S L P S T M R M T V A
☐ tr|A0A8M3ANQ1|A0A8M3A  E D I R G S F L M K N N G H S V E I Q L P S T M K I T K G

P2328

☐ sp|P23280|CAH6_HUMAN   V I E I H I V H Y N S - K Y K S Y D I A Q D A P D G L A V
☐ tr|A0A8M3ANQ1|A0A8M3A  M A E L H V V H Y N S E K Y P S F E E A K N K P D G L A V

P2328

☐ sp|P23280|CAH6_HUMAN   T L T G L D V Q D M L P R N L Q H Y Y T Y H G S L T T P P
☐ tr|A0A8M3ANQ1|A0A8M3A  S I S N L N V L S M L S E N L S H F Y R Y K G S L T T P P

P2328

☐ sp|P23280|CAH6_HUMAN   K T I H N D Y R R T Q P L N H R V V E S N F P N Q E Y T L
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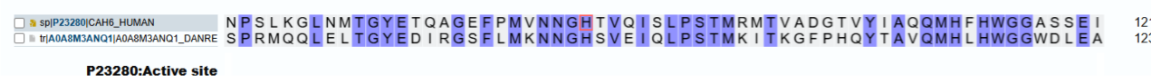


```
☐ sp|P23280|CAH6_HUMAN                N P S L K G L N M T G Y E T Q A G E F P M V N N G H T V Q I S L P S T M R M T V A D G T V Y I A Q Q M H F H W G G A S S E I   12
☐ tr|A0A8M3ANQ1|A0A8M3ANQ1_DANRE      S P R M Q Q L E L T G Y E D I R G S F L M K N N G H S V E I Q L P S T M K I T K G F P H Q Y T A V Q M H L H W G G W D L E A   12

        P23280:Active site
```

```
   N P S L K G L N M T G Y E T Q A G E F P M V N N G H T V Q I S L P S T M R M T V A D G T V Y I A Q Q M H F H W G G A S S E I   121
RE S P R M Q Q L E L T G Y E D I R G S F L M K N N G H S V E I Q L P S T M K I T K G F P H Q Y T A V Q M H L H W G G W D L E A   123

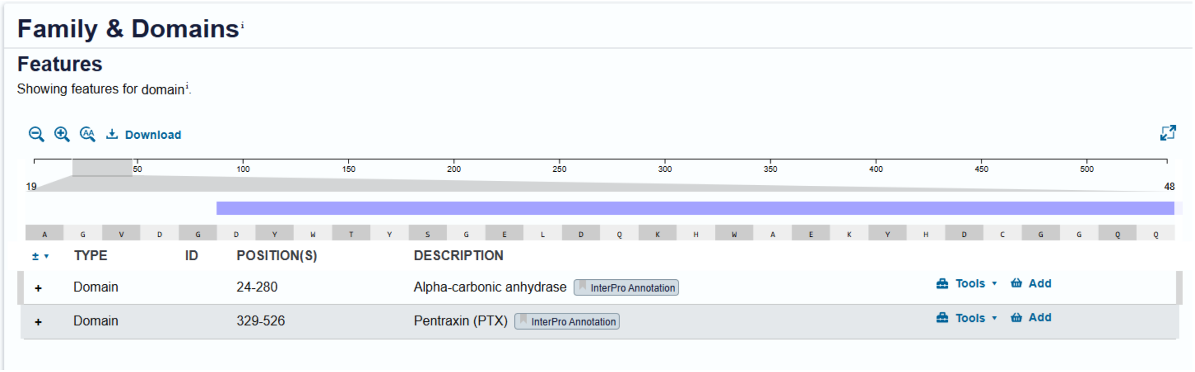
   S G S E H T V D G I R H V I E I H I V H Y N S - K Y K S Y D I A Q D A P D G L A V L A A F V E V K N Y P E N T Y Y S N F I S   182
RE S G S E H T M D G I R Y M A E L H V V H Y N S E K Y P S F E E A K N K P D G L A V L A F F F E D G H F - E N T Y Y S D F I S   184

   H L A N I K Y P G Q R T T L T G L D V Q D M L P R N L Q H Y Y T Y H G S L T T P P C T E N V H W F V L A D F V K L S R T Q V   244
RE N L A N I K Y V G Q S M S I S N L N V L S M L S E N L S H F Y R Y K G S L T T P P C F E S V M W T V F D T P I T L S H N Q I   246

   W K L E N S L L D H R N K T I H N D Y R R T Q P L N H R V V E S N F P N Q E Y T L G S - - - - E F Q F Y L H K I E E I L D Y   302
RE R K L E S T L M D H D N K T L W N D Y R M A Q P L N E R V V E S T F L P R L S K G G M C R Q E E I E A K L K R I E S L I L S   308
```

11. Katera je druga domena, ki se nahaja v homologu *Danio rerio*?

Grem na Family & Domains ter opazim dve domeni ena je Alpha-carbonic anhydrase-kar je domena, ki je v izvornem proteinu, druga domena pa je Pentraxin-pentraksin.



Family & Domains

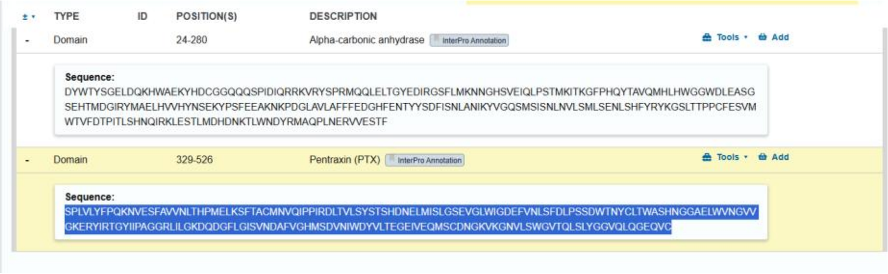
Features

Showing features for domain.

TYPE	ID	POSITION(S)	DESCRIPTION
+ Domain		24-280	Alpha-carbonic anhydrase InterPro Annotation
+ Domain		329-526	Pentraxin (PTX) InterPro Annotation

12. Kopiraj aminokislinsko zaporedje te domene in najdi človeški homolog. Navedi ime človeškega proteina.

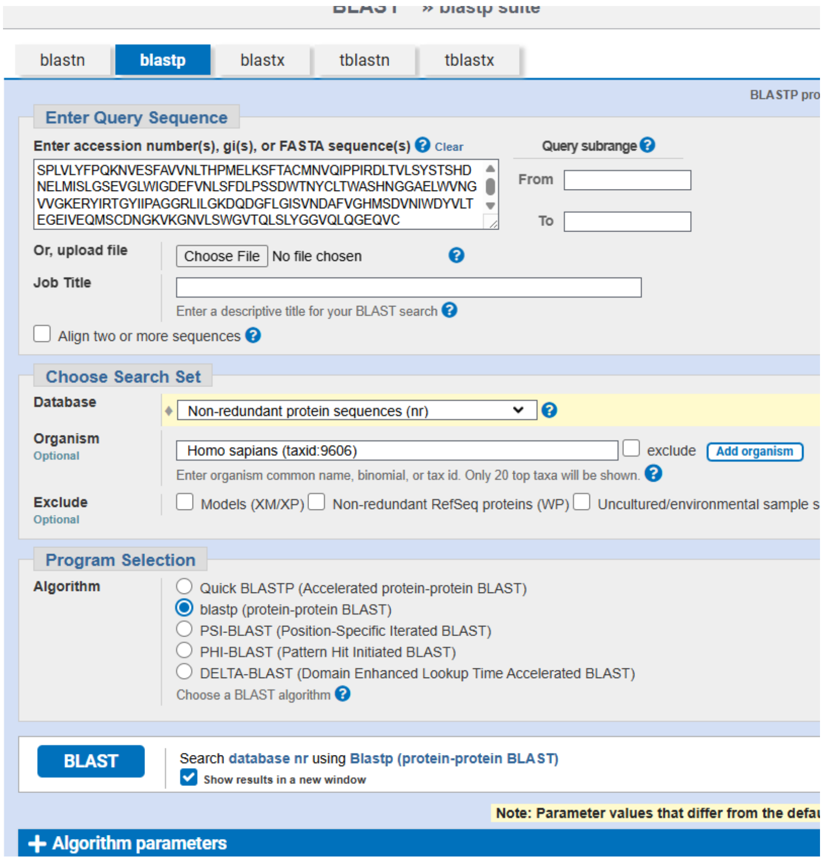
Kopiram aminokislinsko zaporedje te dome in jo priletim v pBLAST. Nastavim na nr database in ker piše da iščem človeški homolog nastavim organism na Homo sapiens. Izberem zadetek z največjim ujemanjem. Ime proteina je Chain A, Pentraxin-related protein PTX3.



TYPE	ID	POSITION(S)	DESCRIPTION
Domain		24-280	Alpha-carbonic anhydrase
Domain		329-526	Pentraxin (PTX)

Sequence (24-280):
```
DYWTYSGELDQKHWAEKYHDCGGQQQSPIDIQRRKVRYSPRMQQLELTGYEDIRGSFLMKNNGHSVEIQLPSTMKITKGFPHQYTAVQMHLHWGGWDLEASG
SEHTMDGIRYMAELHVVHYNSEKYPSFEEAKNKPDGLAVLAFFFEDGHFENTYYSDFISNLANIKYVGQSMSISNLNVLSMLSENLSHFYRYKGSLTTPPCFESVM
WTVFDTPITLSHNQIRKLESTLMDHDNKTLWNDYRMAQPLNERVVESTF
```

Sequence (329-526):
```
SPLVLYFPQKNVESFAVVNLTHPMELKSFTACMNVQIPPIRDLTVLSYSTSHDNELMISLGSEVGLWIGDEFVNLSFDLPSSDWTNYCLTWASHNGGAELWVNGVV
GKERYIRTGYIIPAGGRLILGKDQDGFLGISVNDAFVGHMSDVNIWDYVLTEGEIVEQMSCDNGKVKGNVLSWGVTQLSLYGGVQLQGEQVC
```



Enter Query Sequence

Enter accession number(s), gi(s), or FASTA sequence(s):
```
SPLVLYFPQKNVESFAVVNLTHPMELKSFTACMNVQIPPIRDLTVLSYSTSHD
NELMISLGSEVGLWIGDEFVNLSFDLPSSDWTNYCLTWASHNGGAELWVNG
VVGKERYIRTGYIIPAGGRLILGKDQDGFLGISVNDAFVGHMSDVNIWDYVLT
EGEIVEQMSCDNGKVKGNVLSWGVTQLSLYGGVQLQGEQVC
```

Query subrange: From / To

Or, upload file: Choose File No file chosen

Job Title: Enter a descriptive title for your BLAST search

☐ Align two or more sequences

Choose Search Set

Database: Non-redundant protein sequences (nr)

Organism (Optional): Homo sapiens (taxid:9606) ☐ exclude Add organism

Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown.

Exclude (Optional): ☐ Models (XM/XP) ☐ Non-redundant RefSeq proteins (WP) ☐ Uncultured/environmental sample s

Program Selection

Algorithm:
- ○ Quick BLASTP (Accelerated protein-protein BLAST)
- ● blastp (protein-protein BLAST)
- ○ PSI-BLAST (Position-Specific Iterated BLAST)
- ○ PHI-BLAST (Pattern Hit Initiated BLAST)
- ○ DELTA-BLAST (Domain Enhanced Lookup Time Accelerated BLAST)

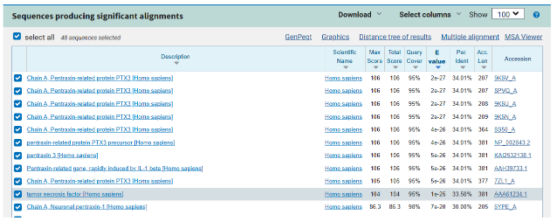
Choose a BLAST algorithm

BLAST — Search database nr using Blastp (protein-protein BLAST)

☑ Show results in a new window

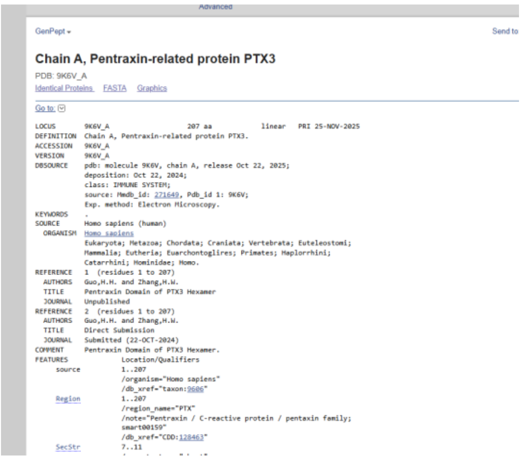
Note: Parameter values that differ from the defau

+ Algorithm parameters



☑ select all 48 sequences selected

	Description	Scientific Name	Max Score	Total Score	Query Cover	E value	Per. Ident	Acc. Len	Accession
☑	Chain A, Pentraxin-related protein PTX3 [Homo sapiens]	Homo sapiens	106	106	95%	2e-27	34.01%	207	9K6V_A
☑	Chain A, Pentraxin-related protein PTX3 [Homo sapiens]	Homo sapiens	106	106	95%	2e-27	34.01%	207	9PVQ_A
☑	Chain A, Pentraxin-related protein PTX3 [Homo sapiens]	Homo sapiens	106	106	95%	2e-27	34.01%	208	9K6U_A
☑	Chain A, Pentraxin-related protein PTX3 [Homo sapiens]	Homo sapiens	106	106	95%	2e-27	34.01%	209	9K6N_A
☑	Chain A, Pentraxin-related protein PTX3 [Homo sapiens]	Homo sapiens	106	106	95%	4e-26	34.01%	364	8S50_A
☑	pentraxin-related protein PTX3 precursor [Homo sapiens]	Homo sapiens	106	106	95%	4e-26	34.01%	381	NP_002843.2
☑	pentraxin 3 [Homo sapiens]	Homo sapiens	106	106	95%	5e-26	34.01%	381	KAI2532138.1
☑	Pentraxin-related gene, rapidly induced by IL-1 beta [Homo sapiens]	Homo sapiens	106	106	95%	5e-26	34.01%	381	AAH39733.1
☑	Chain A, Pentraxin-related protein PTX3 [Homo sapiens]	Homo sapiens	105	105	95%	5e-26	34.01%	377	7ZL1_A
☑	tumor necrosis factor [Homo sapiens]	Homo sapiens	104	104	95%	1e-25	33.50%	381	AAA61234.1
☑	Chain A, Neuronal pentraxin-1 [Homo sapiens]	Homo sapiens	86.3	86.3	98%	7e-20	30.00%	205	5YPE_A



Chain A, Pentraxin-related protein PTX3

PDB: 9K6V_A

```
LOCUS       9K6V_A                   207 aa            linear   PRI 25-NOV-2025
DEFINITION  Chain A, Pentraxin-related protein PTX3.
ACCESSION   9K6V_A
VERSION     9K6V_A
DBSOURCE    pdb: molecule 9K6V, chain A, release Oct 22, 2025;
            deposition: Oct 22, 2024;
            class: IMMUNE SYSTEM;
            source: Mmdb_id: 271649, Pdb_id 1: 9K6V;
            Exp. method: Electron Microscopy.
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE   1  (residues 1 to 207)
  AUTHORS   Guo,H.H. and Zhang,H.W.
  TITLE     Pentraxin Domain of PTX3 Hexamer
  JOURNAL   Unpublished
REFERENCE   2  (residues 1 to 207)
  AUTHORS   Guo,H.H. and Zhang,H.W.
  TITLE     Direct Submission
  JOURNAL   Submitted (22-OCT-2024)
COMMENT     Pentraxin Domain of PTX3 Hexamer.
FEATURES             Location/Qualifiers
     source          1..207
                     /organism="Homo sapiens"
                     /db_xref="taxon:9606"
     Region          1..207
                     /region_name="PTX"
                     /note="Pentraxin / C-reactive protein / pentaxin family;
                     smart00159"
                     /db_xref="CDD:128463"
     SecStr          7..11
```

Chain A, Pentraxin-related protein PTX3

Examples: Insulin, APP, Human, P05067, organism_id:9606

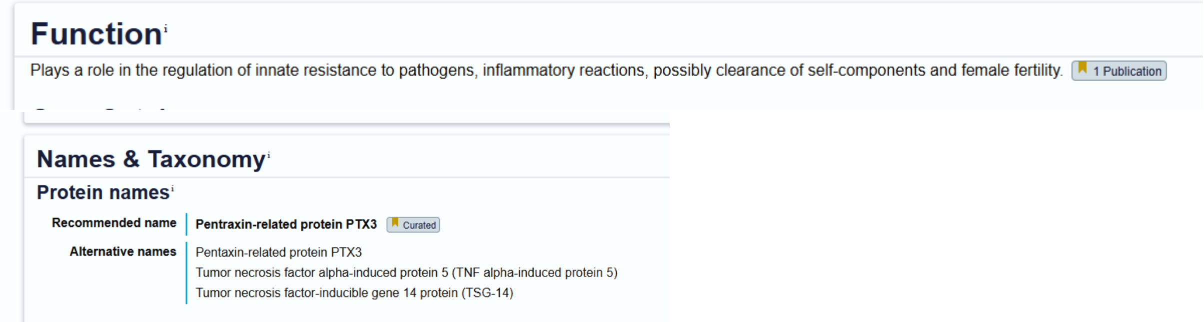
13. Navedi še dve alternativni imeni za ta protein. Kaj je funkcija tega proteina?

Funkcija: Igra vlogo pri uravnavanju prirojene odpornosti proti patogenom, vnetnih reakcijah, morebiti pri odstranjevanju lastnih komponent in ženski plodnosti.

Alternativna imena:

Tumor necrosis factor alpha-induced protein 5 (TNF alpha-induced protein 5)

Tumor necrosis factor-inducible gene 14 protein (TSG-14)



Function

Plays a role in the regulation of innate resistance to pathogens, inflammatory reactions, possibly clearance of self-components and female fertility.

Names & Taxonomy

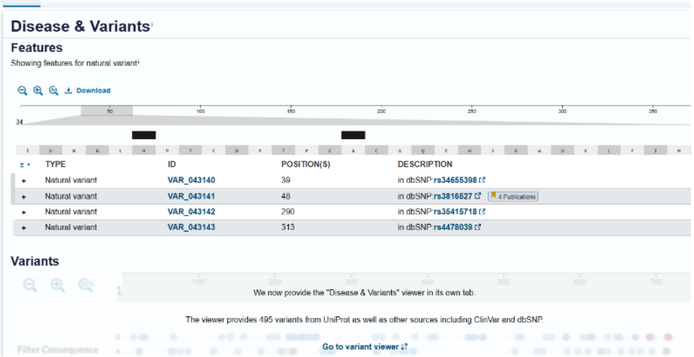
Protein names

Recommended name	**Pentraxin-related protein PTX3** Curated
Alternative names	Pentaxin-related protein PTX3 Tumor necrosis factor alpha-induced protein 5 (TNF alpha-induced protein 5) Tumor necrosis factor-inducible gene 14 protein (TSG-14)

14. V UniProt-u je omenjenih veliko mutacij tega proteina. Zamenjava katerega aminokislinskega ostanka v katerega povzroči nastanek raka/karcinoma? Navedi katera vrsta raka se pojavi (*Namig: dve vrsti raka*).

Gremo pod razdelek Disease & Variants→Go to variant viewer

Ugotovimo da nastanek raka povzroči mutacija Q167R (glutamin→arginin). Ta mutacija povzroči Serozni cistadenokarcinom jajčnikov in raka želodca.



Disease & Variants

Features

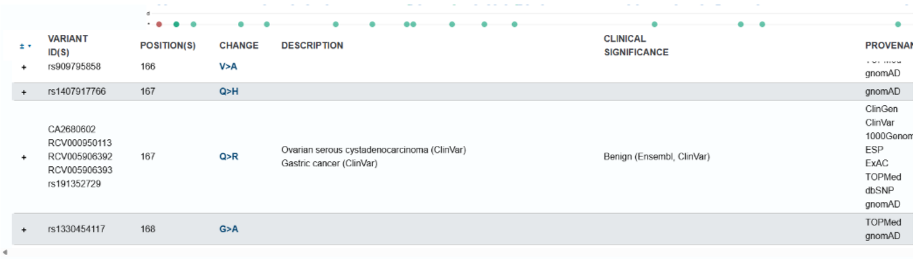
Showing features for natural variant.

TYPE	ID	POSITION(S)	DESCRIPTION
Natural variant	VAR_043140	39	in dbSNP:rs34655398
Natural variant	VAR_043141	48	in dbSNP:rs3816527 4 Publications
Natural variant	VAR_043142	290	in dbSNP:rs35415718
Natural variant	VAR_043143	313	in dbSNP:rs4478039

Variants

We now provide the "Disease & Variants" viewer in its own tab.

The viewer provides 495 variants from UniProt as well as other sources including ClinVar and dbSNP.



	VARIANT ID(S)	POSITION(S)	CHANGE	DESCRIPTION	CLINICAL SIGNIFICANCE	PROVENAN
+	rs909795858	166	V>A			gnomAD
+	rs1407917766	167	Q>H			gnomAD
+	CA2680602 RCV000950113 RCV005906392 RCV005906393 rs191352729	167	Q>R	Ovarian serous cystadenocarcinoma (ClinVar) Gastric cancer (ClinVar)	Benign (Ensembl, ClinVar)	ClinGen ClinVar 1000Genom ESP ExAC TOPMed dbSNP gnomAD
+	rs1330454117	168	G>A			TOPMed gnomAD