Introduction to Bioinformatics

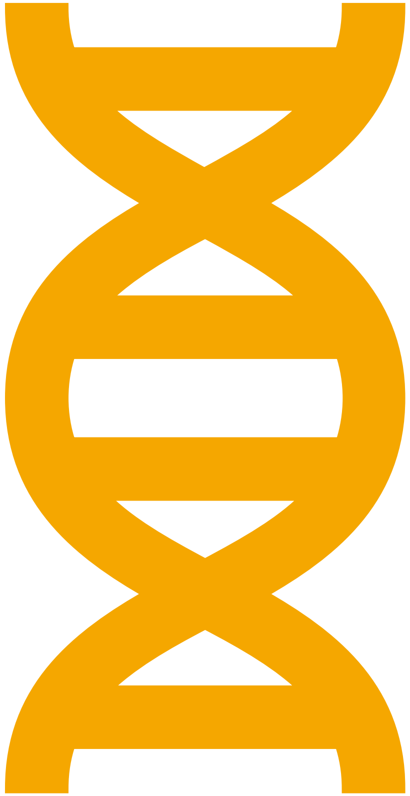
Dr. Archana Upadhya

Associate Professor

Humera Khan College of Pharmacy

What is a gene?

- The gene is considered the basic unit of inheritance.
- Genes are passed from parents to offspring and contain the information needed to specify physical and biological traits.
- Most genes code for specific proteins, or segments of proteins, which have differing functions within the body.
- Humans have approximately 20,000 protein-coding genes



What is a gene contd…

- Proteins are the brick and mortar that make up our cells and tissues. And genes are the part of our genome that encodes the information for making those proteins. For example, the human genome has roughly 20,000 protein-coding genes.
- Interestingly, all of the information for those 20,000 protein-coding genes is encoded by only 1.5% of the entire human genome.
- A more expansive definition of a gene includes those segments of DNA that encode information for making an RNA molecule that functions in some fashion other than directly coding for a protein; these are sometimes referred to as RNA genes.

What is a gene contd…

- Genes are made up of sequences of DNA and are arranged, one after another, at specific locations on chromosomes in the nucleus of cells.
- They contain information for making specific proteins that lead to the expression of a particular physical characteristic or trait, such as hair color or eye color, or to a particular function in a cell.

What controls the expression of a gene?

- Rate of transcription of gene to mRNA
- mRNA processing
- Rate of mRNA degradation
- Translation of mRNA to protein
- Post translational processing of protein
- Rate of protein degradation

Transcription

- Transcription of gene to mRNA
  - Interplay of transcription factors (factors that can bring about an increase in specific gene transcription)
  - Promoters (sequences to which the transcription factors and RNA polymerase bind)
  - Enhancers and Silencers (Regulatory elements (to which transcription factors bind) that can enhance or silence a given gene by promoting or inhibiting transcription of a gene

mRNA processing in the nucleus

- 5' cap
- 3' Poly A tail
- Alternative splicing
    - Certain tissues may have a different sequence of mRNA and therefore have different isoforms of the mRNA hence different isoforms of the protein.

Rate of mRNA degradation

- Sequences in the mRNA may bind to cytoplasmic factors to aid or reduce rate of mRNA degradation
- mRNA may be silenced by micro RNA (miRNA-non coding RNA) –protein translation may be inhibited.

Translation of mRNA to protein

- Presence of sequences on the mRNA aid in its translation to a protein
- Certain cytoplasmic factors may cause a reduction in translation

Post translation processing of protein

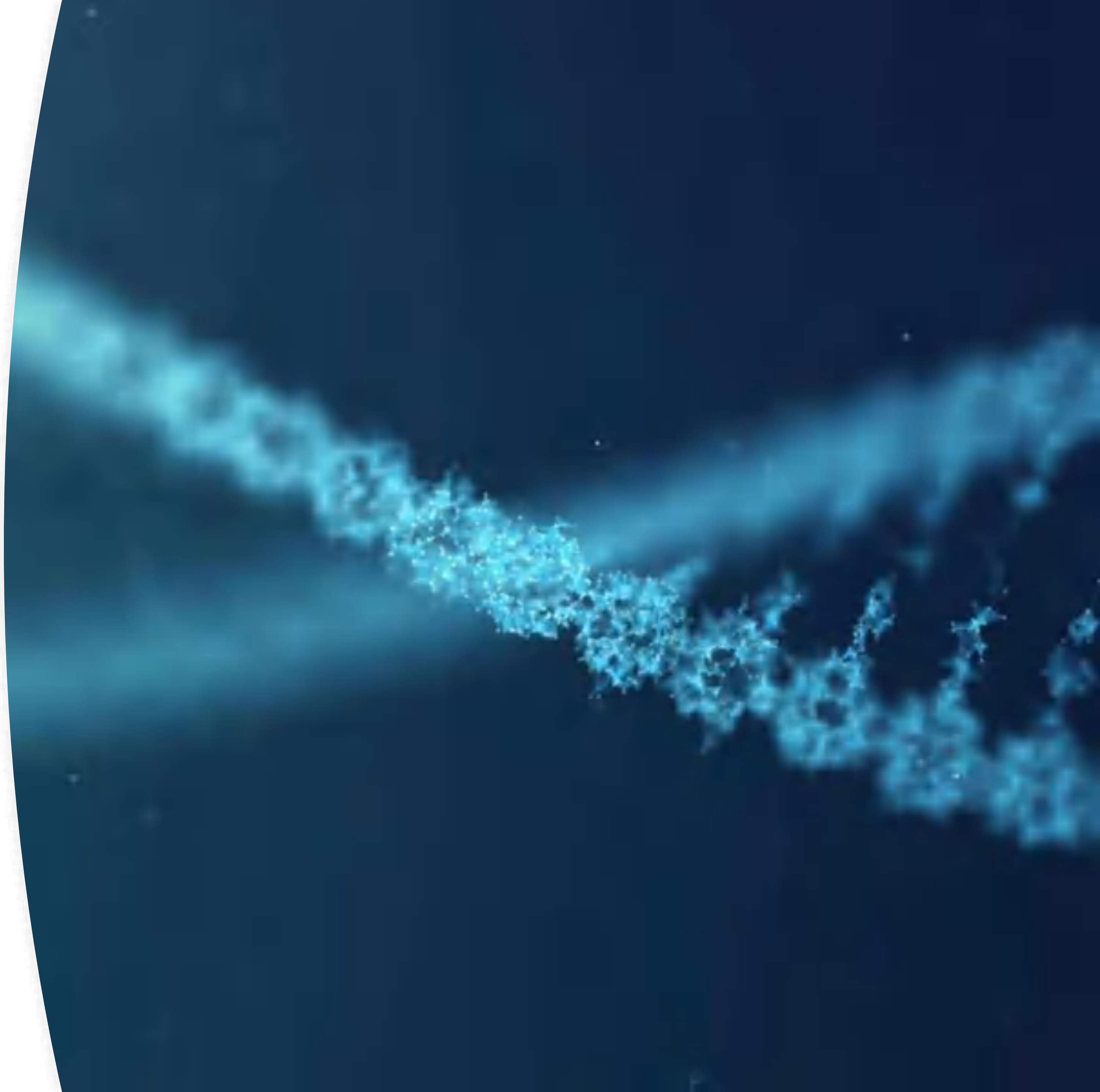
- Removal of signal peptide
- Addition of certain moieties to the protein
    - E.g. Acetylation, phosphorylation, glycosylation
- Formation of di-sulphide bonds
- Folding of the protein to the conformation most optimal for its activity

Protein Degradation

- Ubiquitin proteosome pathway
- Cellular proteases

What is Bioinformatics?

Bioinformatics, as related to genetics and genomics, is a scientific subdiscipline that involves using computer technology to collect, store, analyze and disseminate biological data and information, such as DNA and amino acid sequences or annotations about those sequences

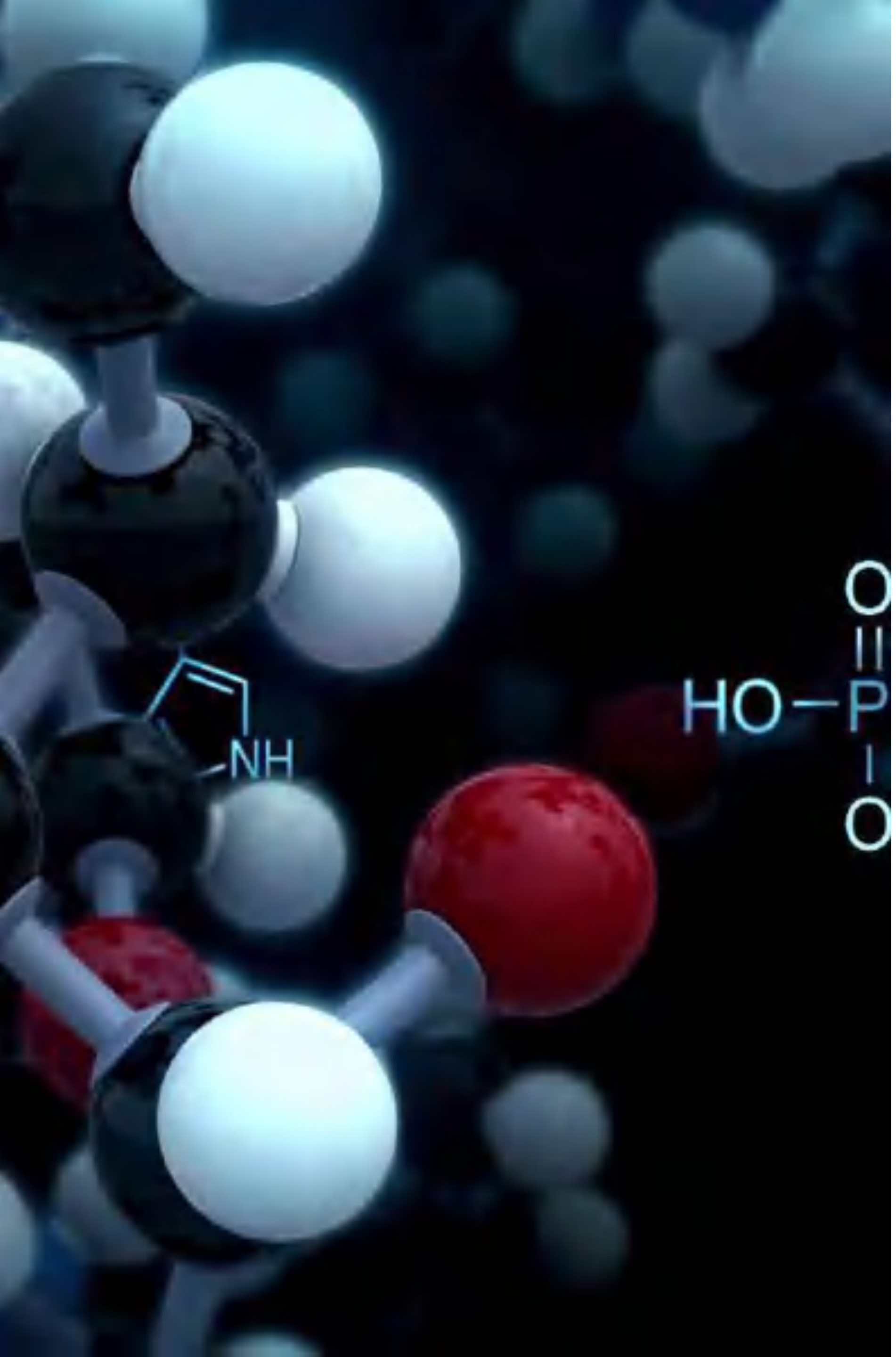


Popular Databases

1. NCBI Entrez: https://www.ncbi.nlm.nih.gov/
2. PubMed: https://pubmed.ncbi.nlm.nih.gov/
3. Genome Database: https://www.ncbi.nlm.nih.gov/genome/
4. Protein Data Bank: https://www.rcsb.org/
5. Enzyme Database: https://enzyme.expasy.org/
6. Pathway Database: https://www.genome.jp/kegg/pathway.html
7. OMIM: https://www.omim.org/

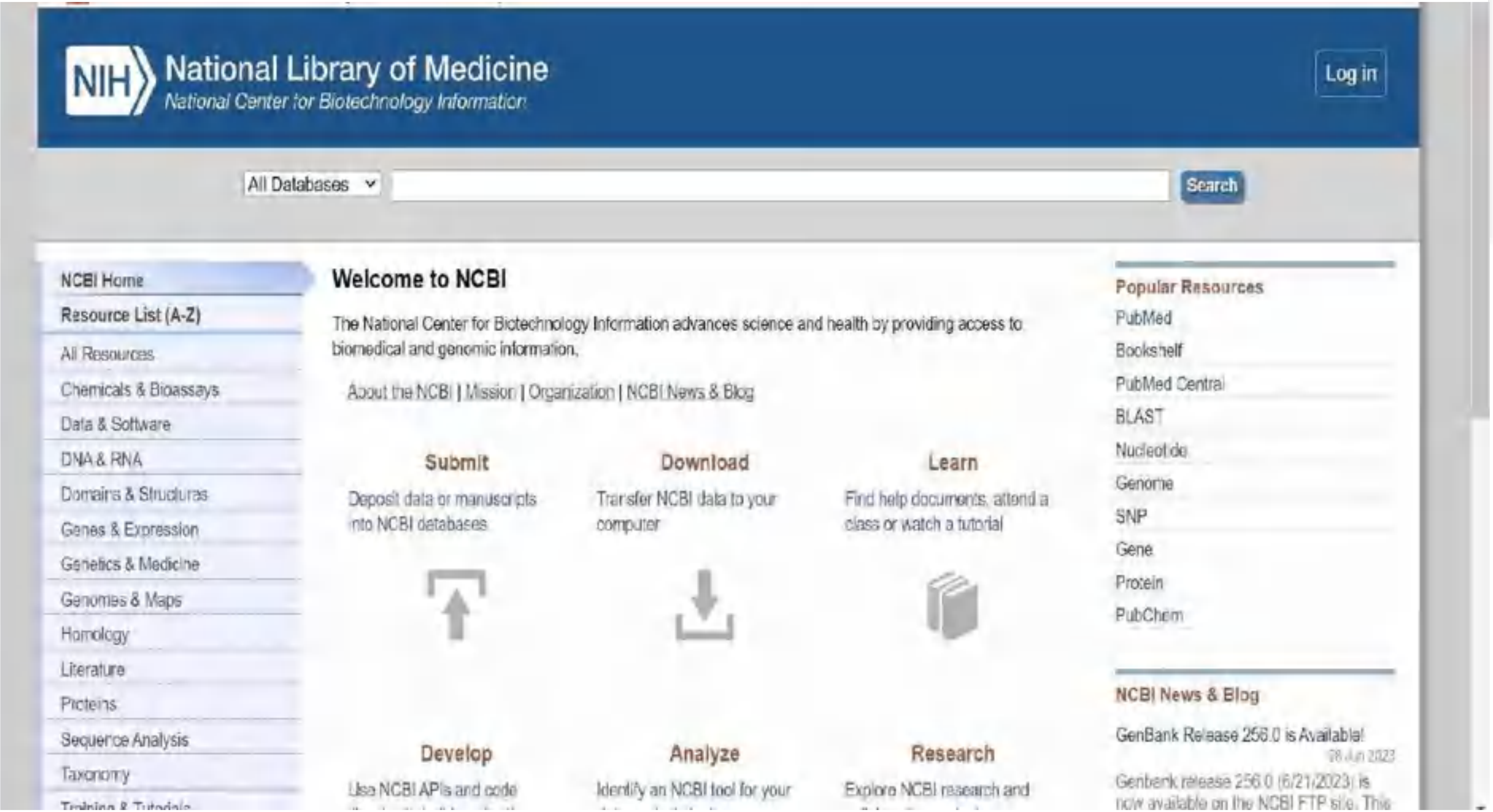
Organization of this session on Bioinformatics

- Theory (2h)
  - Model Gene (Glucose 6 Phosphatase1)
  - Information retrieval from the given databases
- Practical (4h)
  - Each student will be given a separate gene
  - Student has to retrieve information about the gene
  - A report has to be made on the given gene and submitted on the practical day itself.
  - The report will be part of the 25 page report you will be submitting as the Lab Practice School Report.



Human Glucose 6 Phosphatase 1(G6PC1)

- Type “ NCBI NLM” in Google
- Click on “NCBI - National Institutes of Health (NIH) “



National Library of Medicine
National Center for Biotechnology Information

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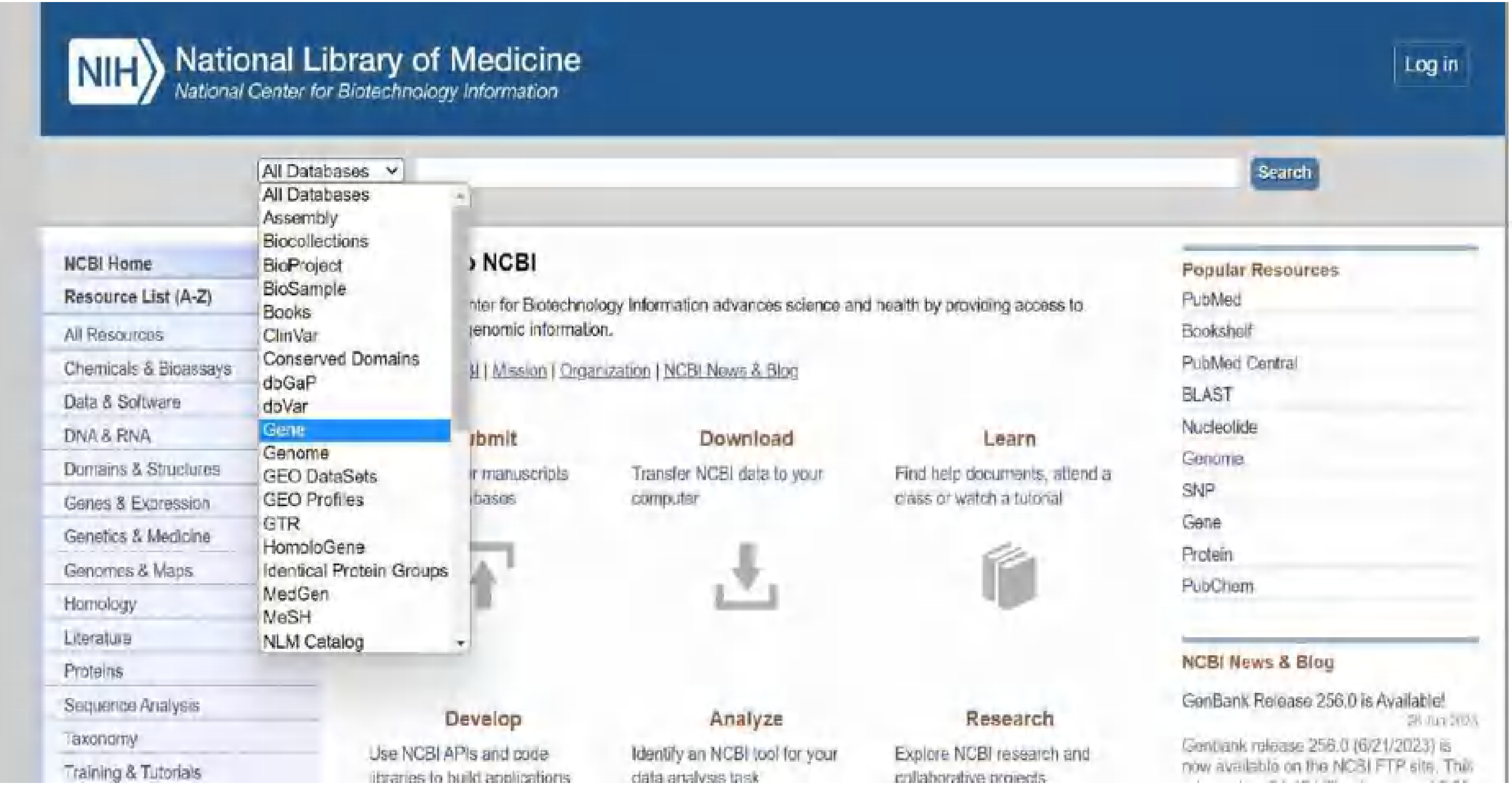
NCBI News & Blog

GenBank Release 256.0 is Available!
28 Jun 2023
Genbank release 256.0 (6/21/2023) is now available on the NCBI FTP site. This



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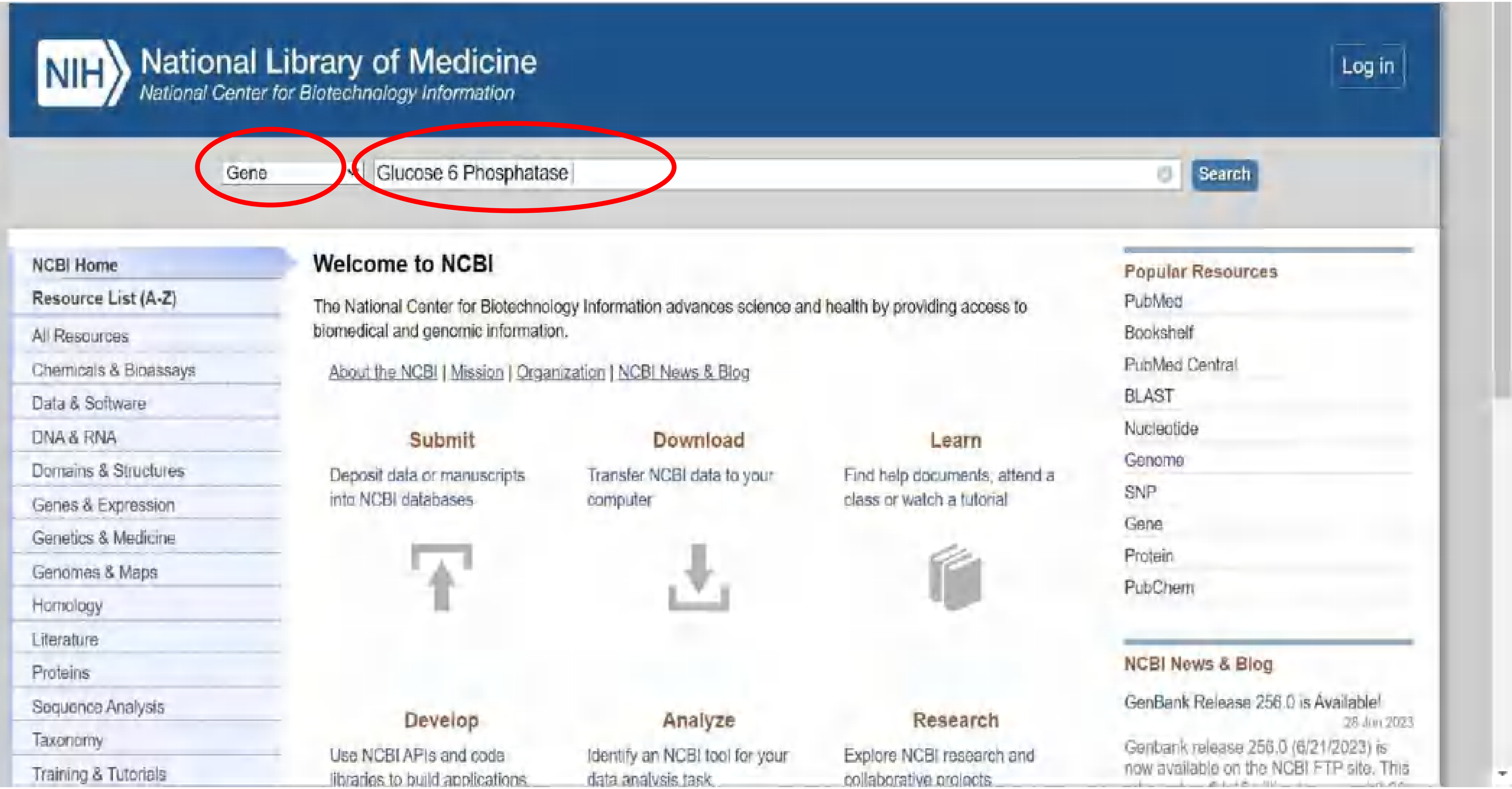

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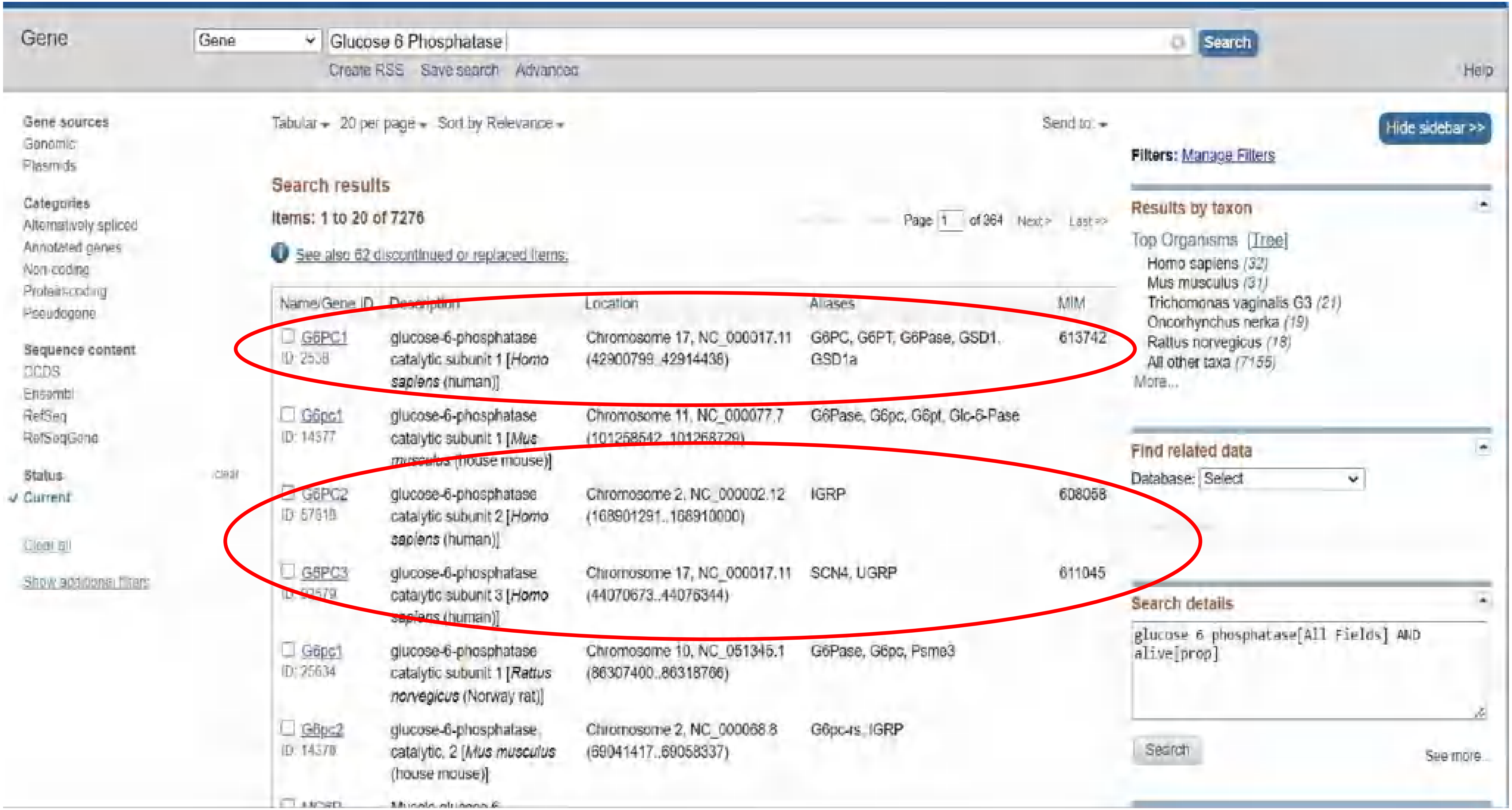


Search results

Items: 1 to 20 of 7276

Page 1 of 364 Next > Last >>

See also 62 discontinued or replaced items.

Name/Gene ID	Description	Location	Aliases	MIM
☐ G6PC1 ID: 2538	glucose-6-phosphatase catalytic subunit 1 [*Homo sapiens* (human)]	Chromosome 17, NC_000017.11 (42900799..42914436)	G6PC, G6PT, G6Pase, GSD1, GSD1a	613742
☐ G6pc1 ID: 14377	glucose-6-phosphatase catalytic subunit 1 [*Mus musculus* (house mouse)]	Chromosome 11, NC_000077.7 (101258542..101268729)	G6Pase, G6pc, G6pt, Glc-6-Pase	
☐ G6PC2 ID: 57818	glucose-6-phosphatase catalytic subunit 2 [*Homo sapiens* (human)]	Chromosome 2, NC_000002.12 (168901291..168910000)	IGRP	608058
☐ G6PC3 ID: 92579	glucose-6-phosphatase catalytic subunit 3 [*Homo sapiens* (human)]	Chromosome 17, NC_000017.11 (44070673..44076344)	SCN4, UGRP	611045
☐ G6pc1 ID: 25634	glucose-6-phosphatase catalytic subunit 1 [*Rattus norvegicus* (Norway rat)]	Chromosome 10, NC_051345.1 (86307400..86318766)	G6Pase, G6pc, Psme3	
☐ G6pc2 ID: 14378	glucose-6-phosphatase, catalytic, 2 [*Mus musculus* (house mouse)]	Chromosome 2, NC_000068.8 (69041417..69058337)	G6pc-rs, IGRP	

Results by taxon

Top Organisms [Tree]

Homo sapiens	(32)
Mus musculus	(31)
Trichomonas vaginalis G3	(21)
Oncorhynchus nerka	(19)
Rattus norvegicus	(18)
All other taxa	(7155)

More...

Find related data

Database: Select

Search details

```
glucose 6 phosphatase[All Fields] AND
alive[prop]
```

Information retrieved

- Catlaytic Subunits of Glucose 6-Phosphatase in Humans (Homo Sapiens): 3
- Chromosomal Locations of the three catalytic subunits-
  - Catalytic subunit 1 (G6PC1): Chromosome 17, NC_000017.11 (42900799..42914438)
  - Catalytic subunit 2 (G6PC2): Chromosome 2, NC_000002.12 (168901291..168910000)
  - Catlaytic subunit 3 (G6PC3): Chromosome 17, NC_000017.11 (44070673..44076344)

Gene IDs and Aliases of the three Catalytic Subunits

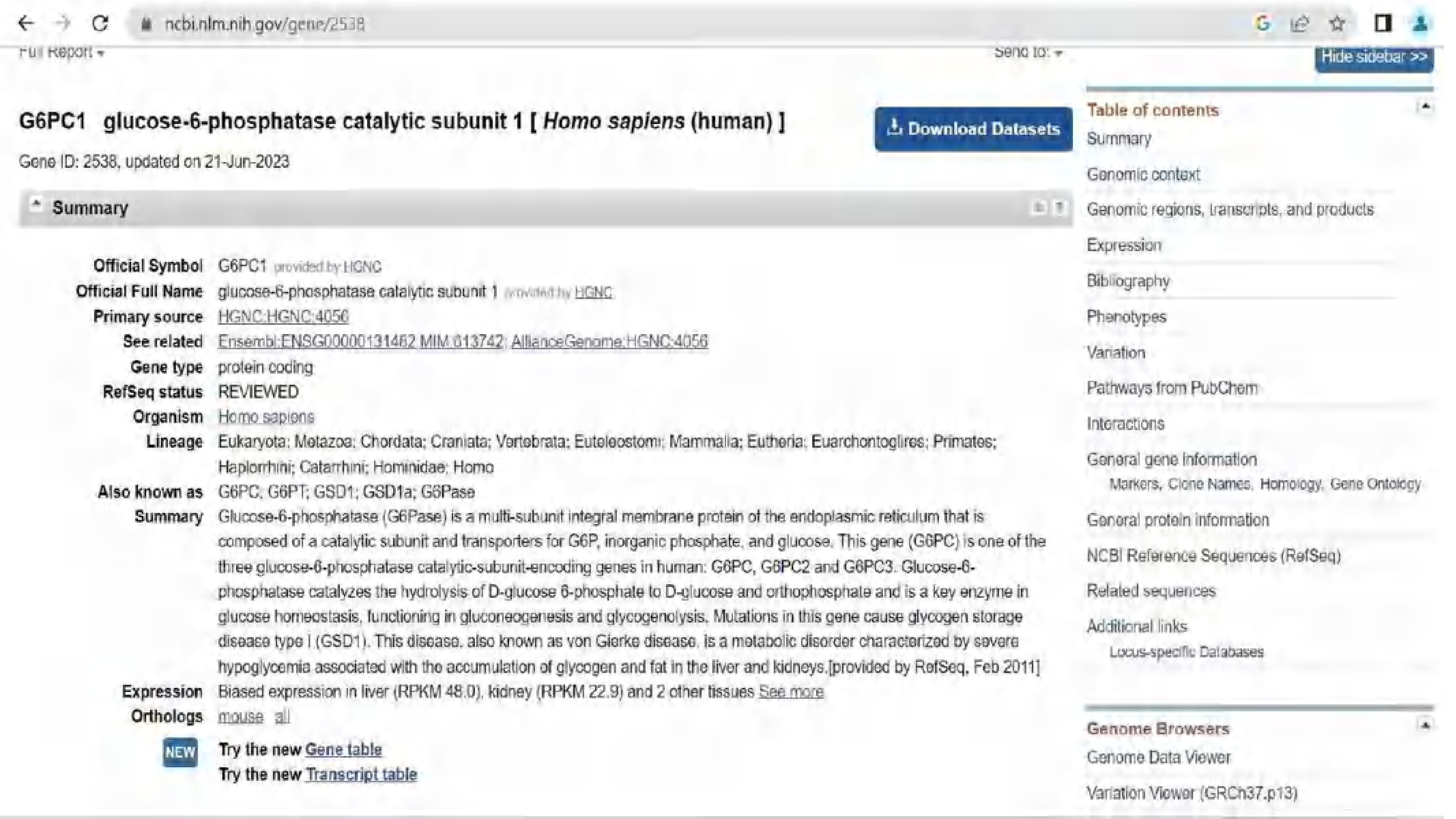
- G6PC1: Gene ID: 2538. Aliases: G6PC, G6PT, G6Pase, GSD1, GSD1a
- G6PC2: Gene ID: 57818 Aliases: IGRP
- G6PC3: Gene ID: 92579 Aliases: SCN4, UGRP

Online Mendelian Inheritance in Man (OMIM)- MIM numbers

- G6PC1: MIM (613742)
- G6PC2: MIM (608058)
- G6PC3: MIM (611045)

Accession categories and molecule types

Category	Description
NC	Complete genomic molecules
NG	Incomplete genomic region
NM	mRNA
NR	ncRNA (non coding RNA)
NP	Protein
XM	predicted mRNA model
XR	predicted ncRNA model
XP	predicted Protein model (eukaryotic sequences)
WP	predicted Protein model (prokaryotic sequences)



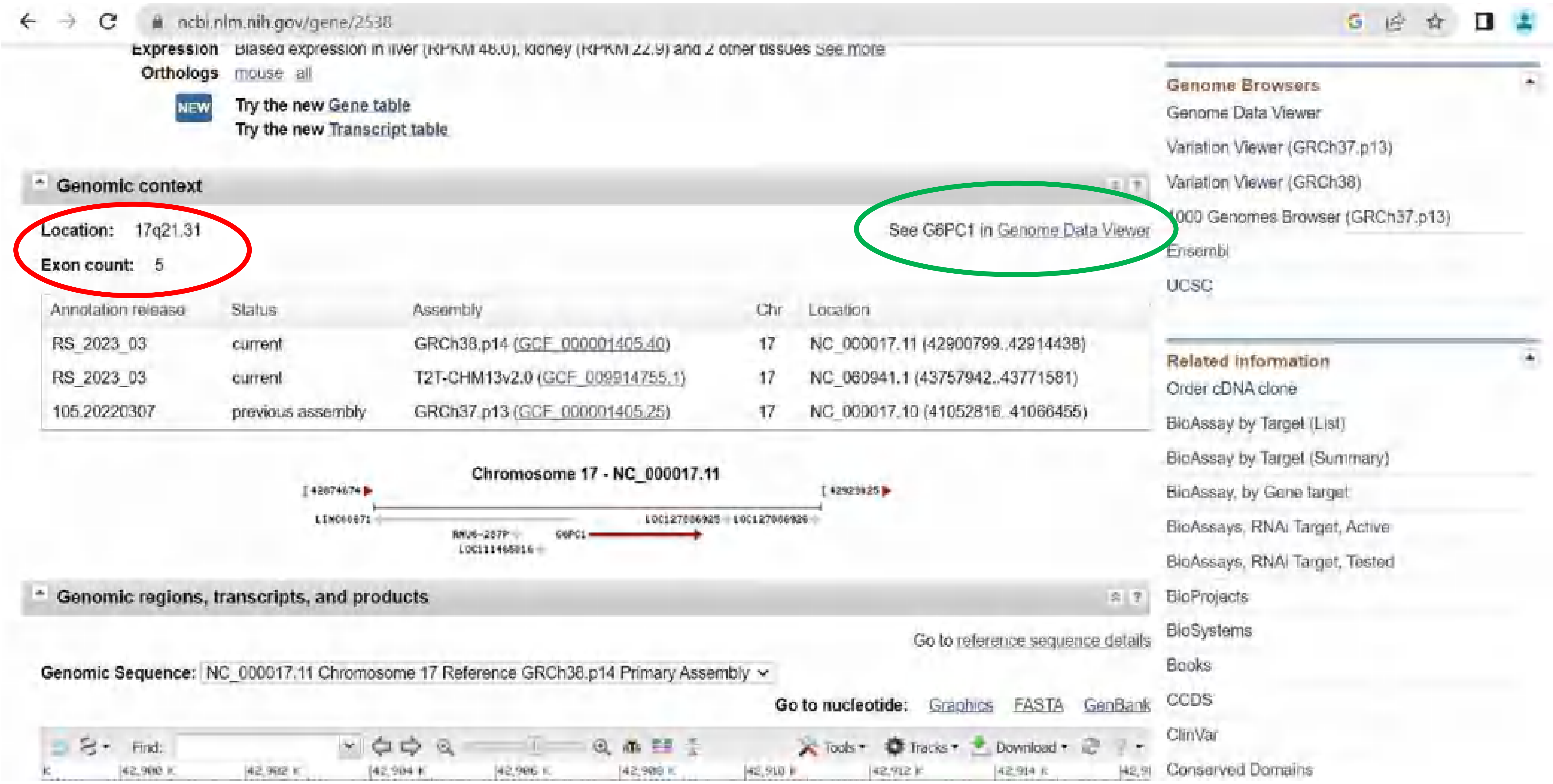
G6PC1 glucose-6-phosphatase catalytic subunit 1 [*Homo sapiens* (human)]

Gene ID: 2538, updated on 21-Jun-2023

Summary

Official Symbol	G6PC1 provided by HGNC
Official Full Name	glucose-6-phosphatase catalytic subunit 1 provided by HGNC
Primary source	HGNC:HGNC:4056
See related	Ensembl:ENSG00000131482 MIM:613742; AllianceGenome:HGNC:4056
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	G6PC; G6PT; GSD1; GSD1a; G6Pase
Summary	Glucose-6-phosphatase (G6Pase) is a multi-subunit integral membrane protein of the endoplasmic reticulum that is composed of a catalytic subunit and transporters for G6P, inorganic phosphate, and glucose. This gene (G6PC) is one of the three glucose-6-phosphatase catalytic-subunit-encoding genes in human: G6PC, G6PC2 and G6PC3. Glucose-6-phosphatase catalyzes the hydrolysis of D-glucose 6-phosphate to D-glucose and orthophosphate and is a key enzyme in glucose homeostasis, functioning in gluconeogenesis and glycogenolysis. Mutations in this gene cause glycogen storage disease type I (GSD1). This disease, also known as von Gierke disease, is a metabolic disorder characterized by severe hypoglycemia associated with the accumulation of glycogen and fat in the liver and kidneys.[provided by RefSeq, Feb 2011]
Expression	Biased expression in liver (RPKM 48.0), kidney (RPKM 22.9) and 2 other tissues See more
Orthologs	mouse all

NEW **Try the new** Gene table
Try the new Transcript table



Expression Biased expression in liver (RPKM 48.0), kidney (RPKM 22.9) and 2 other tissues See more

Orthologs mouse all

Try the new Gene table

Try the new Transcript table

Genomic context

Location: 17q21.31

Exon count: 5

See G6PC1 in Genome Data Viewer

Annotation release	Status	Assembly	Chr	Location
RS_2023_03	current	GRCh38.p14 (GCF_000001405.40)	17	NC_000017.11 (42900799..42914438)
RS_2023_03	current	T2T-CHM13v2.0 (GCF_009914755.1)	17	NC_060941.1 (43757942..43771581)
105.20220307	previous assembly	GRCh37.p13 (GCF_000001405.25)	17	NC_000017.10 (41052816..41066455)

Chromosome 17 - NC_000017.11

Genomic regions, transcripts, and products

Go to reference sequence details

Genomic Sequence: NC_000017.11 Chromosome 17 Reference GRCh38.p14 Primary Assembly

Go to nucleotide: Graphics FASTA GenBank

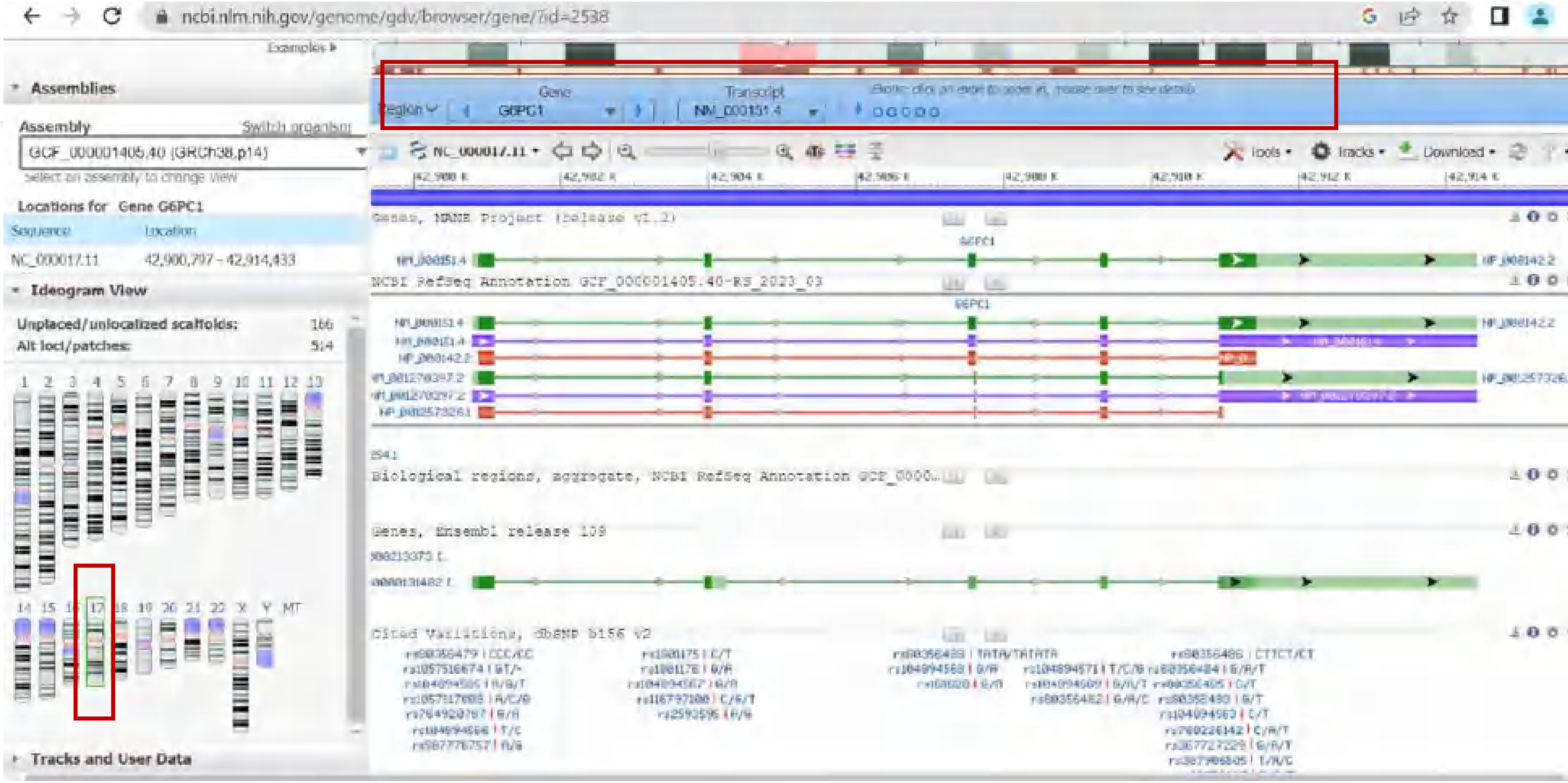
Genome Browsers

- Genome Data Viewer
- Variation Viewer (GRCh37.p13)
- Variation Viewer (GRCh38)
- 1000 Genomes Browser (GRCh37.p13)
- Ensembl
- UCSC

Related information

- Order cDNA clone
- BioAssay by Target (List)
- BioAssay by Target (Summary)
- BioAssay, by Gene target
- BioAssays, RNAi Target, Active
- BioAssays, RNAi Target, Tested
- BioProjects
- BioSystems
- Books
- CCDS
- ClinVar
- Conserved Domains

Click on Genome Data Viewer



ncbi.nlm.nih.gov/genome/gdv/browser/gene/?id=2538

Assemblies

Assembly: GCF_000001405.40 (GRCh38.p14)

Select an assembly to change view

Locations for Gene G6PC1

Sequence	Location
NC_000017.11	42,900,797 - 42,914,433

Ideogram View

Unplaced/unlocalized scaffolds:	166
Alt loci/patches:	514

Tracks and User Data

Region: Gene G6PC1 Transcript NM_000151.4

Genes, MANE Project (release v1.2)

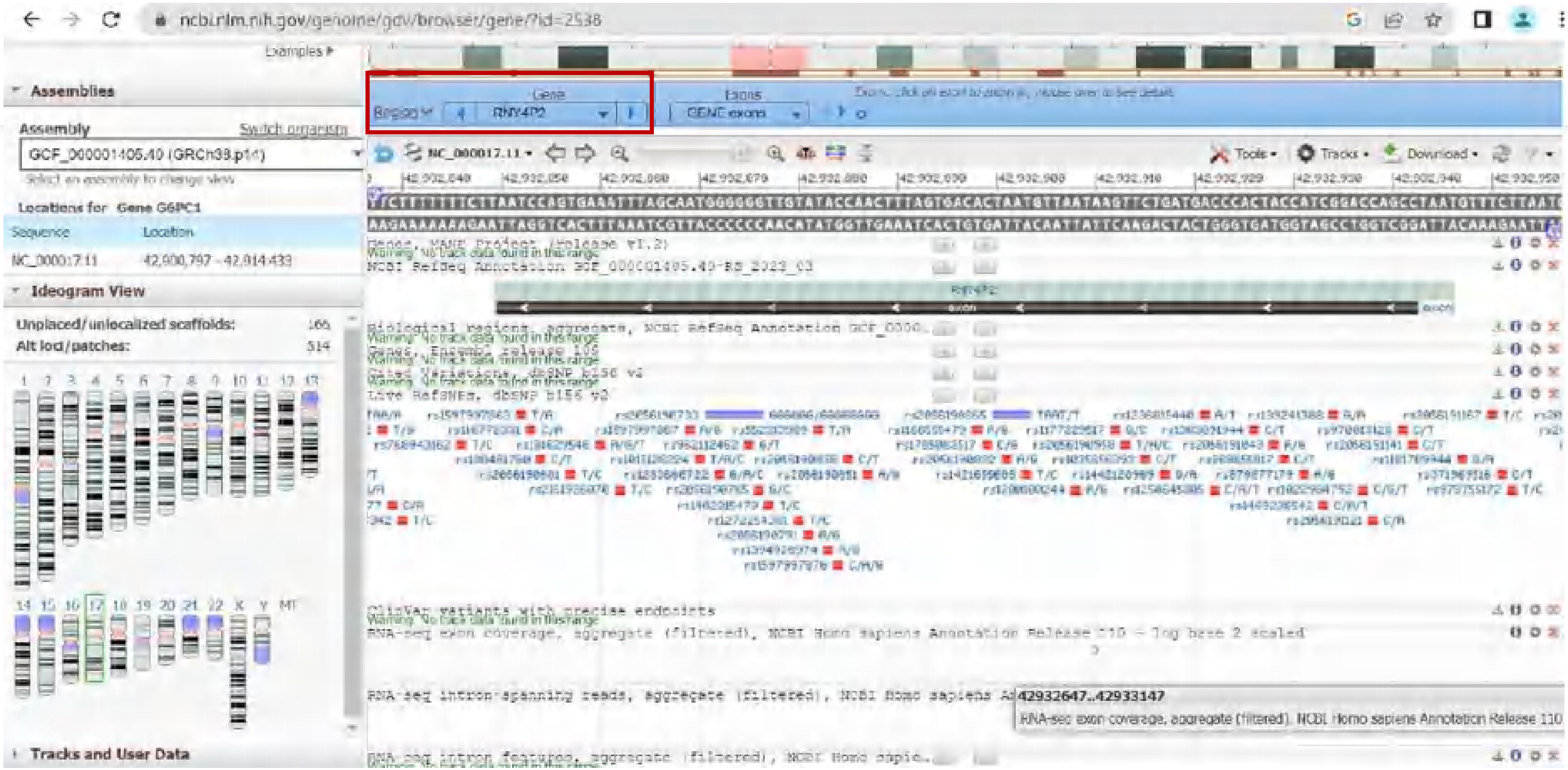
NCBI RefSeq Annotation GCF_000001405.40-RS_2023_03

Biological regions, aggregate, NCBI RefSeq Annotation GCF_0000...

Genes, Ensembl release 109

Cited Variations, dbSNP b156 v2

Gene next to G6PC1-RNY4P2



ncbi.nlm.nih.gov/genome/gdv/browser/gene/?id=2538

Assemblies

Assembly: GCF_000001405.40 (GRCh38.p14)

Locations for Gene G6PC1

Sequence	Location
NC_000017.11	42,900,797 - 42,914,433

Ideogram View

Unplaced/unlocalized scaffolds:	165
Alt loci/patches:	514

Gene: RNY4P2

Exons: GENE exons

Exons: click an exon to zoom in, mouse over to see details

NC_000017.11

Genes, MANE Project (Release v1.2)

Warning: No track data found in this range

NCBI RefSeq Annotation GCF_000001405.40-RS_2023_03

Biological regions, aggregate, NCBI RefSeq Annotation GCF_0000...

Warning: No track data found in this range

Genes, Ensembl release 109

Warning: No track data found in this range

Cited Variations, dbSNP b156 v2

Warning: No track data found in this range

Live RefSNPs, dbSNP b156 v2

ClinVar variants with precise endpoints

Warning: No track data found in this range

RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens A...

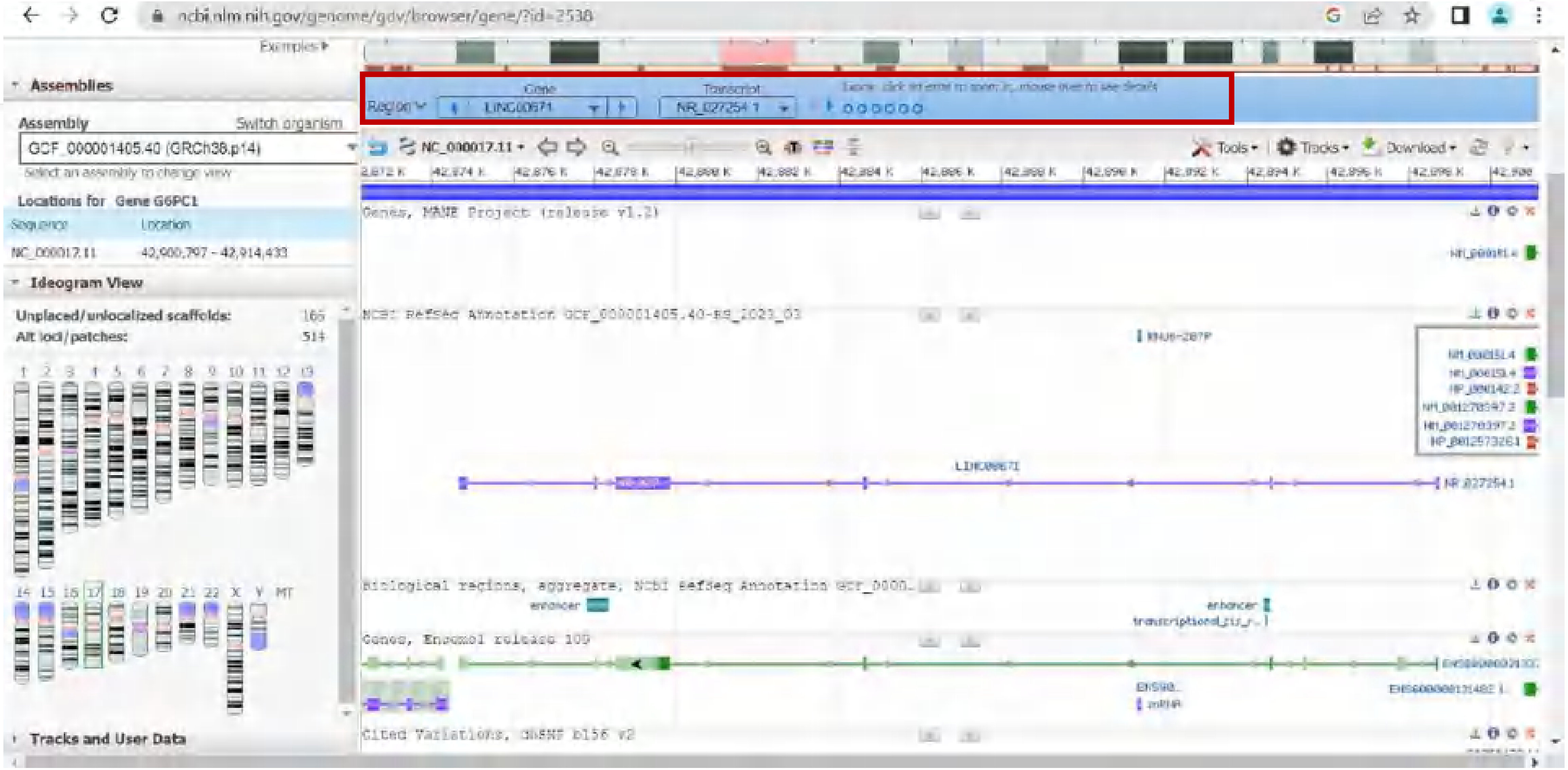
42932647..42933147

RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110

RNA-seq intron features, aggregate (filtered), NCBI Homo sapie...

Tracks and User Data

Gene previous to G6PC1-LINC00671



ncbi.nlm.nih.gov/genome/gdv/browser/gene/?id=2538

Assembly: GCF_000001405.40 (GRCh38.p14)

Locations for Gene G6PC1

Sequence	Location
NC_000017.11	42,900,797 - 42,914,433

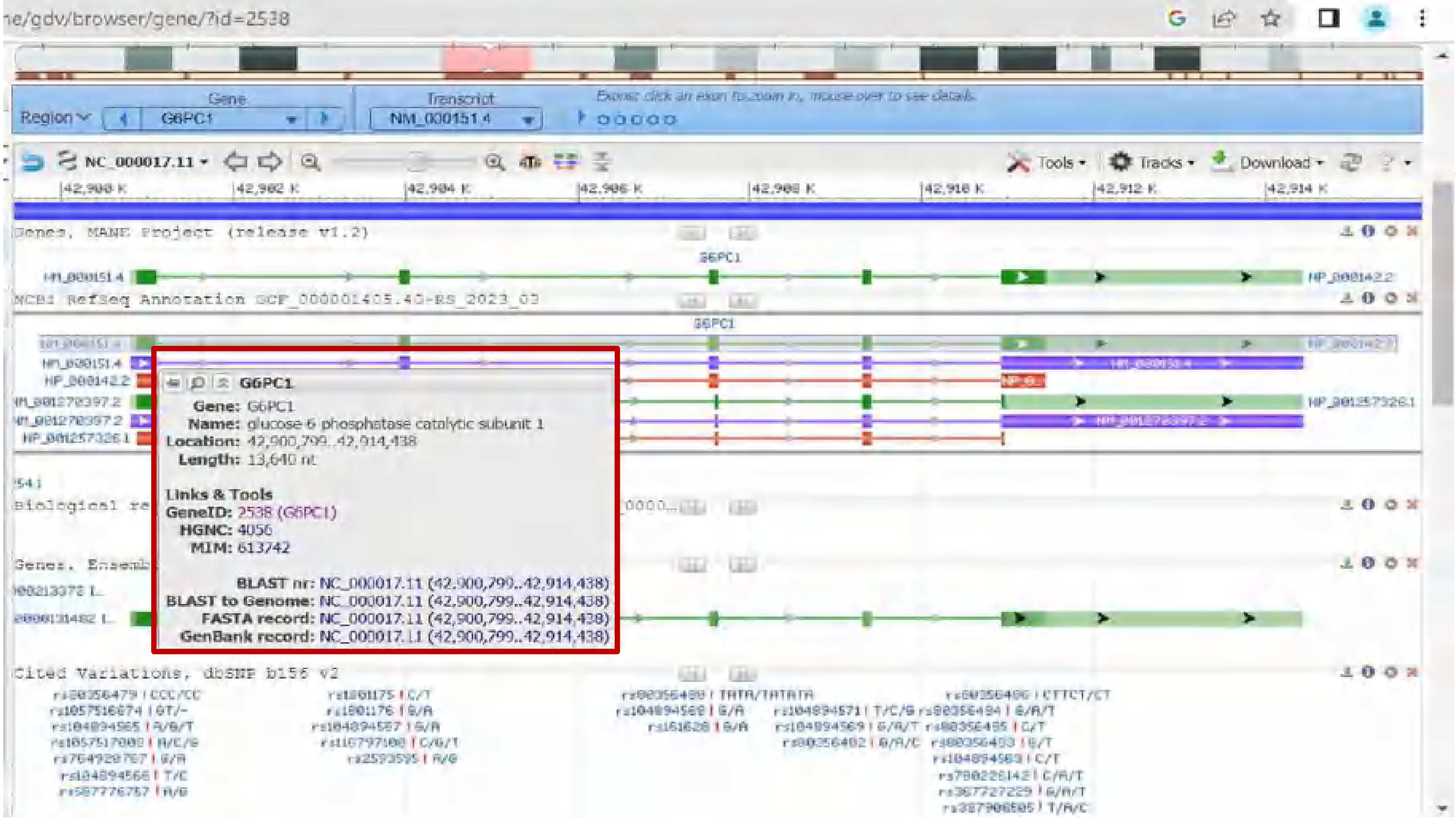
Unplaced/unlocalized scaffolds: 165

Alt loci/patches: 514

Gene: LINC00671

Transcript: NR_027254.1

Details of G6PC1



G6PC1

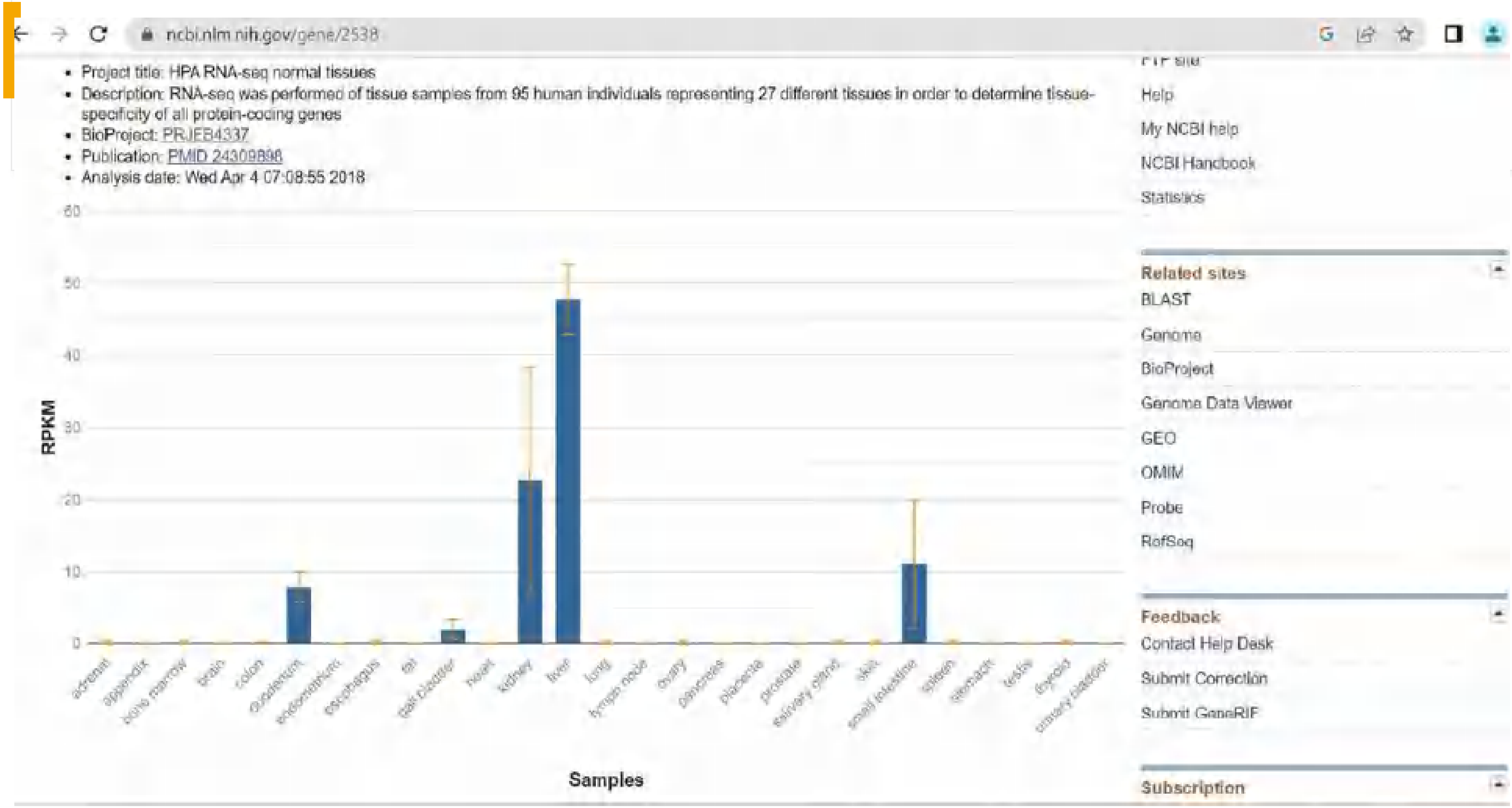
Gene: G6PC1
Name: glucose-6-phosphatase catalytic subunit 1
Location: 42,900,799..42,914,438
Length: 13,640 nt

Links & Tools
GeneID: 2538 (G6PC1)
HGNC: 4056
MIM: 613742

BLAST nr: NC_000017.11 (42,900,799..42,914,438)
BLAST to Genome: NC_000017.11 (42,900,799..42,914,438)
FASTA record: NC_000017.11 (42,900,799..42,914,438)
GenBank record: NC_000017.11 (42,900,799..42,914,438)

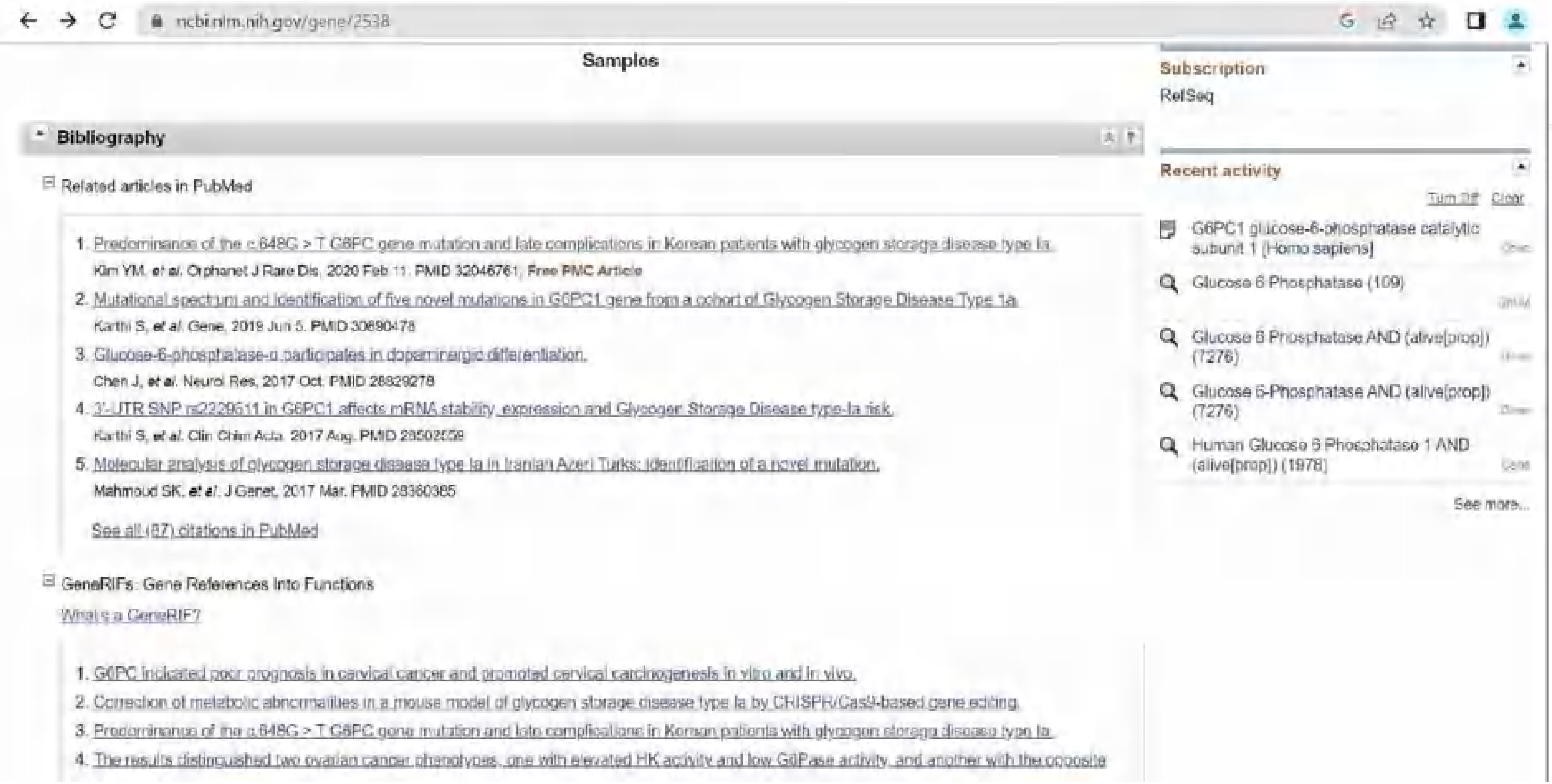
Go Back to webpage: https://www.ncbi.nlm.nih.gov/gene/2538

Tissue Specific Expression



- Project title: HPA RNA-seq normal tissues
- Description: RNA-seq was performed of tissue samples from 95 human individuals representing 27 different tissues in order to determine tissue-specificity of all protein-coding genes
- BioProject: PRJEB4337
- Publication: PMID 24309898
- Analysis date: Wed Apr 4 07:08:55 2018

Continue in https://www.ncbi.nlm.nih.gov/gene/2538 Bibliography-Related Articles



Bibliography

Related articles in PubMed

1. Predominance of the c.648G > T G6PC gene mutation and late complications in Korean patients with glycogen storage disease type Ia.
   Kim YM, *et al.* Orphanet J Rare Dis. 2020 Feb 11. PMID 32046761. **Free PMC Article**
2. Mutational spectrum and identification of five novel mutations in G6PC1 gene from a cohort of Glycogen Storage Disease Type 1a.
   Karthi S, *et al.* Gene. 2019 Jun 5. PMID 30890478
3. Glucose-6-phosphatase-α participates in dopaminergic differentiation.
   Chen J, *et al.* Neurol Res. 2017 Oct. PMID 28829278
4. 3'-UTR SNP rs2229611 in G6PC1 affects mRNA stability, expression and Glycogen Storage Disease type-Ia risk.
   Karthi S, *et al.* Clin Chim Acta. 2017 Aug. PMID 28502559
5. Molecular analysis of glycogen storage disease type Ia in Iranian Azeri Turks: identification of a novel mutation.
   Mahmoud SK, *et al.* J Genet. 2017 Mar. PMID 28360385

See all (87) citations in PubMed

GeneRIFs: Gene References Into Functions

What's a GeneRIF?

1. G6PC indicated poor prognosis in cervical cancer and promoted cervical carcinogenesis in vitro and in vivo.
2. Correction of metabolic abnormalities in a mouse model of glycogen storage disease type Ia by CRISPR/Cas9-based gene editing.
3. Predominance of the c.648G > T G6PC gene mutation and late complications in Korean patients with glycogen storage disease type Ia.
4. The results distinguished two ovarian cancer phenotypes, one with elevated HK activity and low G6Pase activity, and another with the opposite

G6PC1 glucose-6-phosphatase catalytic subunit 1 [Homo sapiens]

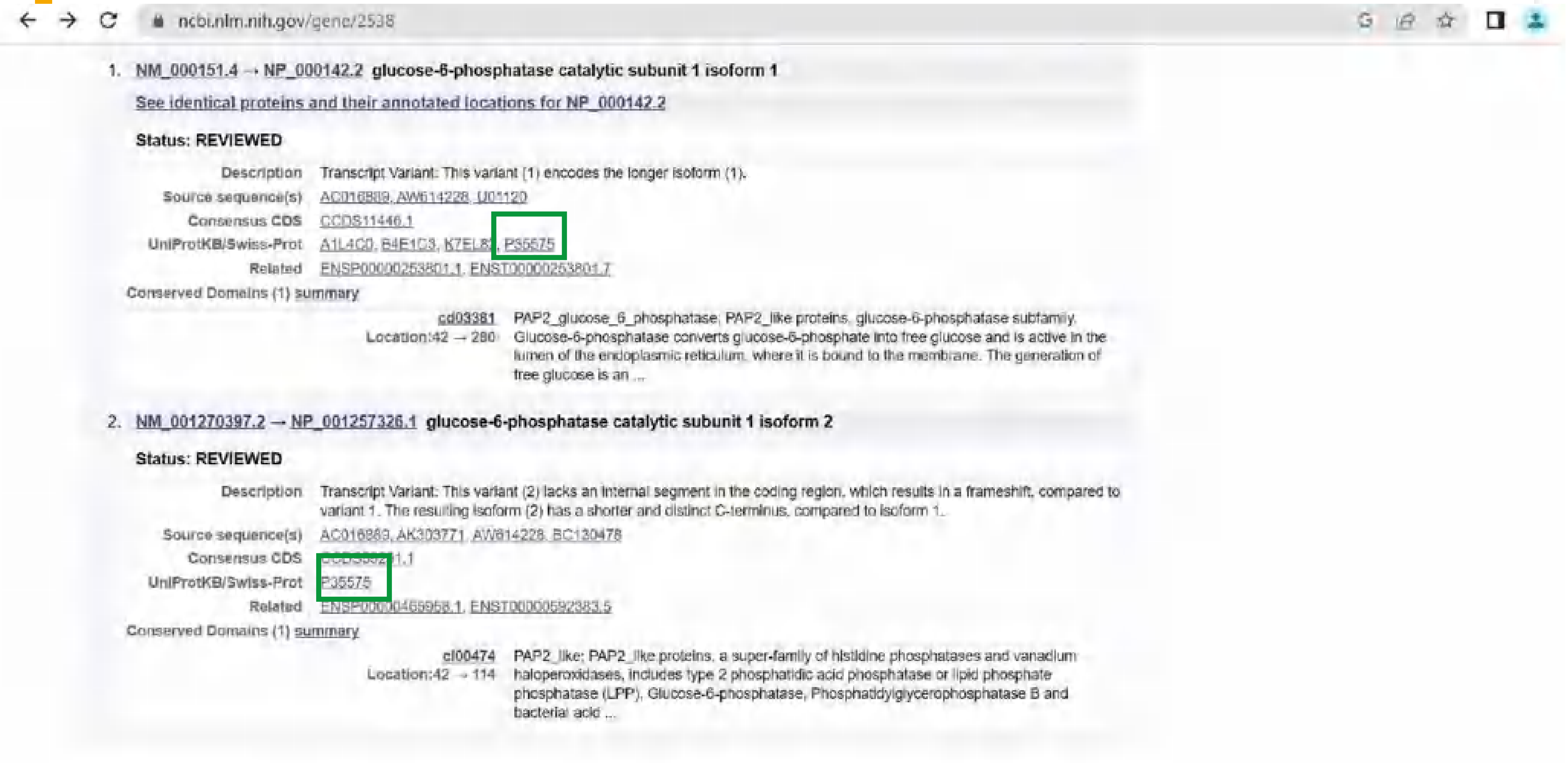
Glucose 6 Phosphatase (109)

Glucose 6 Phosphatase AND (alive[prop]) (7276)

Glucose 6-Phosphatase AND (alive[prop]) (7276)

Human Glucose 6 Phosphatase 1 AND (alive[prop]) (1978)

Continue in https://www.ncbi.nlm.nih.gov/gene/2538



ncbi.nlm.nih.gov/gene/2538

1. **NM_000151.4 → NP_000142.2 glucose-6-phosphatase catalytic subunit 1 isoform 1**

See identical proteins and their annotated locations for NP_000142.2

Status: REVIEWED

Description Transcript Variant: This variant (1) encodes the longer isoform (1).
Source sequence(s) AC016889, AW614228, U01120
Consensus CDS CCDS11446.1
UniProtKB/Swiss-Prot A1L4C0, B4E1C3, K7EL81, P35575
Related ENSP00000253801.1, ENST00000253801.7

Conserved Domains (1) summary

cd03381 Location:42 → 280 PAP2_glucose_6_phosphatase; PAP2_like proteins, glucose-6-phosphatase subfamily. Glucose-6-phosphatase converts glucose-6-phosphate into free glucose and is active in the lumen of the endoplasmic reticulum, where it is bound to the membrane. The generation of free glucose is an ...

2. **NM_001270397.2 → NP_001257326.1 glucose-6-phosphatase catalytic subunit 1 isoform 2**

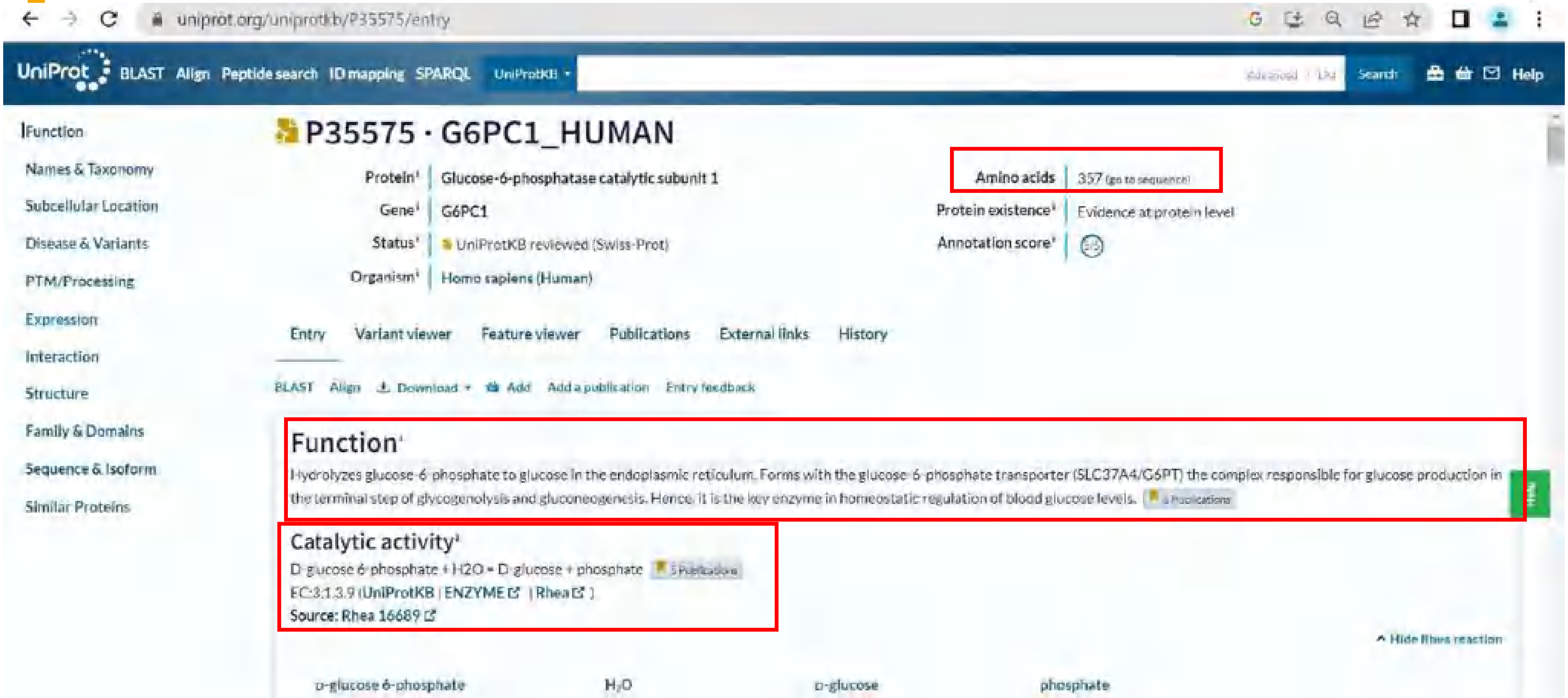
Status: REVIEWED

Description Transcript Variant: This variant (2) lacks an internal segment in the coding region, which results in a frameshift, compared to variant 1. The resulting isoform (2) has a shorter and distinct C-terminus, compared to isoform 1.
Source sequence(s) AC016889, AK303771, AW614228, BC130478
Consensus CDS [illegible]1.1
UniProtKB/Swiss-Prot P35575
Related ENSP00000465958.1, ENST00000592383.5

Conserved Domains (1) summary

cl00474 Location:42 → 114 PAP2_like; PAP2_like proteins, a super-family of histidine phosphatases and vanadium haloperoxidases, includes type 2 phosphatidic acid phosphatase or lipid phosphate phosphatase (LPP), Glucose-6-phosphatase, Phosphatidylglycerophosphatase B and bacterial acid ...

In the webpage: https://www.ncbi.nlm.nih.gov/gene/2538
Click on P35575 to open to the following UniProt webpage



P35575 · G6PC1_HUMAN

Protein: Glucose-6-phosphatase catalytic subunit 1
Gene: G6PC1
Status: UniProtKB reviewed (Swiss-Prot)
Organism: Homo sapiens (Human)
Amino acids: 357 (go to sequence)
Protein existence: Evidence at protein level
Annotation score: 5/5

Function

Hydrolyzes glucose-6-phosphate to glucose in the endoplasmic reticulum. Forms with the glucose-6-phosphate transporter (SLC37A4/G6PT) the complex responsible for glucose production in the terminal step of glycogenolysis and gluconeogenesis. Hence, it is the key enzyme in homeostatic regulation of blood glucose levels. 3 Publications

Catalytic activity

D-glucose 6-phosphate + H2O = D-glucose + phosphate 5 Publications
EC:3.1.3.9 (UniProtKB | ENZYME | Rhea)
Source: Rhea 16689

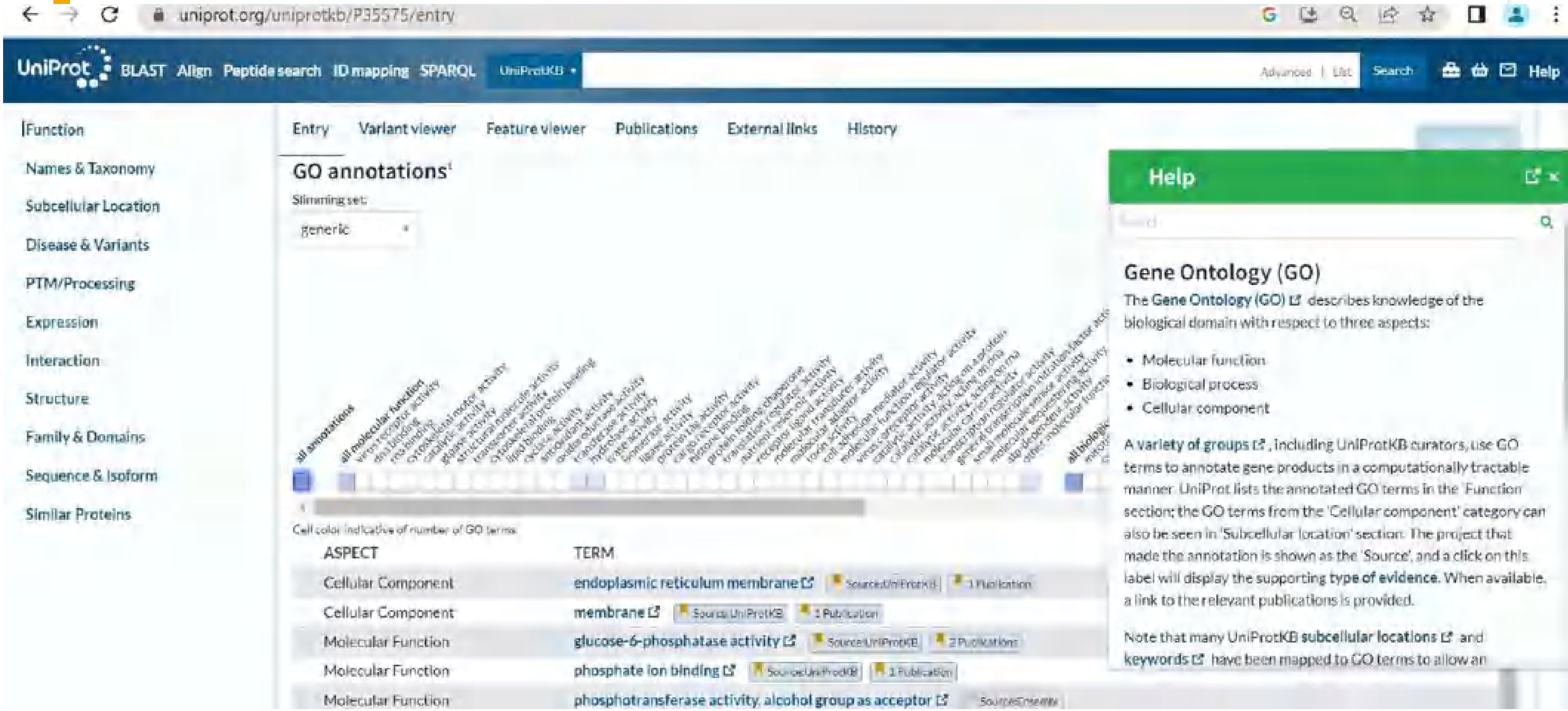
Information retrieved from previous slide from webpage
https://www.uniprot.org/uniprotkb/P35575/entry

Amino acids in G6PC1: 357

Function of G6PC1:
Hydrolyzes glucose-6-phosphate to glucose in the endoplasmic reticulum. Forms with the glucose-6-phosphate transporter (SLC37A4/G6PT) the complex responsible for glucose production in the terminal step of glycogenolysis and gluconeogenesis. Hence, it is the key enzyme in homeostatic regulation of blood glucose levels.

Catalytic activity
D-glucose 6-phosphate + H_2O = D-glucose + phosphate (5 Publications)
EC:3.1.3.9 (Enzyme)

Continue on https://www.uniprot.org/uniprotkb/P35575/entry



GO annotations

Slimming set: generic

Cell color indicative of number of GO terms

ASPECT	TERM
Cellular Component	endoplasmic reticulum membrane Source:UniProtKB 1 Publication
Cellular Component	membrane Source:UniProtKB 1 Publication
Molecular Function	glucose-6-phosphatase activity Source:UniProtKB 2 Publications
Molecular Function	phosphate ion binding Source:UniProtKB 1 Publication
Molecular Function	phosphotransferase activity, alcohol group as acceptor Source:Ensembl

Help

Gene Ontology (GO)

The Gene Ontology (GO) describes knowledge of the biological domain with respect to three aspects:

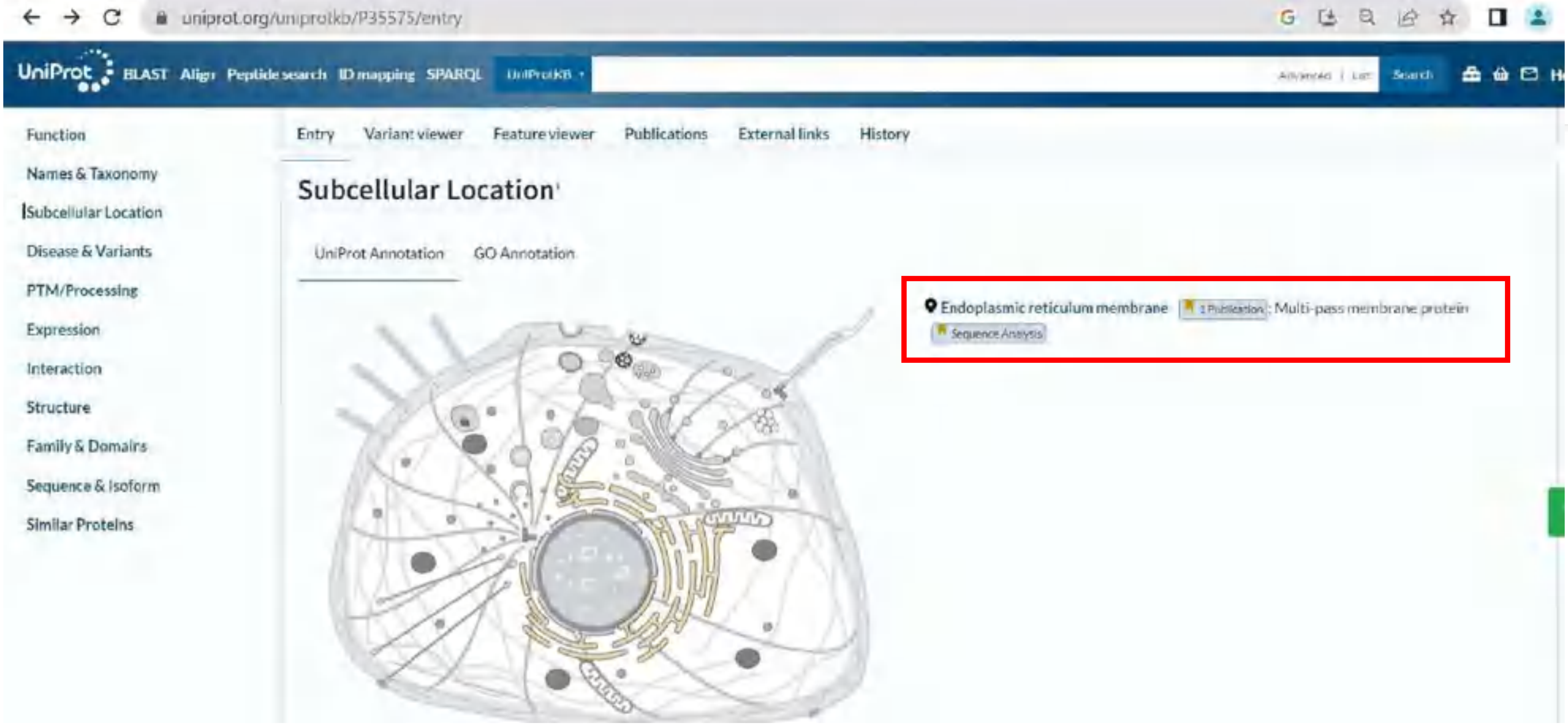
- Molecular function
- Biological process
- Cellular component

A variety of groups, including UniProtKB curators, use GO terms to annotate gene products in a computationally tractable manner. UniProt lists the annotated GO terms in the 'Function' section; the GO terms from the 'Cellular component' category can also be seen in 'Subcellular location' section. The project that made the annotation is shown as the 'Source', and a click on this label will display the supporting type of evidence. When available, a link to the relevant publications is provided.

Note that many UniProtKB subcellular locations and keywords have been mapped to GO terms to allow an

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

SUBCELLULAR LOCATION

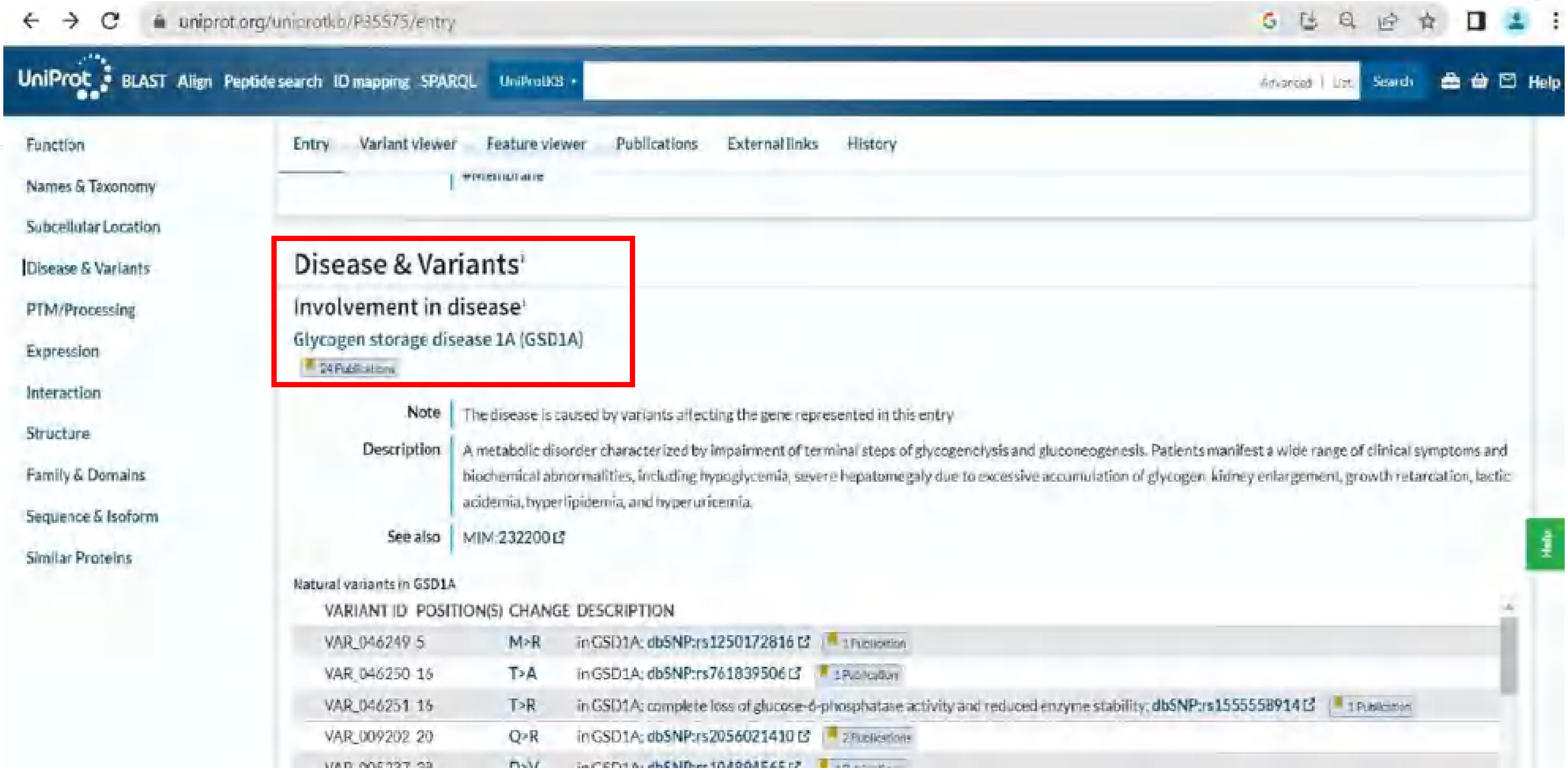


Subcellular Location

Endoplasmic reticulum membrane 1 Publication ; Multi-pass membrane protein Sequence Analysis

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

DISEASE AND VARIANTS OF THE ENZYME



Disease & Variants

Involvement in disease

Glycogen storage disease 1A (GSD1A)

24 Publications

Note The disease is caused by variants affecting the gene represented in this entry

Description A metabolic disorder characterized by impairment of terminal steps of glycogenolysis and gluconeogenesis. Patients manifest a wide range of clinical symptoms and biochemical abnormalities, including hypoglycemia, severe hepatomegaly due to excessive accumulation of glycogen, kidney enlargement, growth retardation, lactic acidemia, hyperlipidemia, and hyperuricemia.

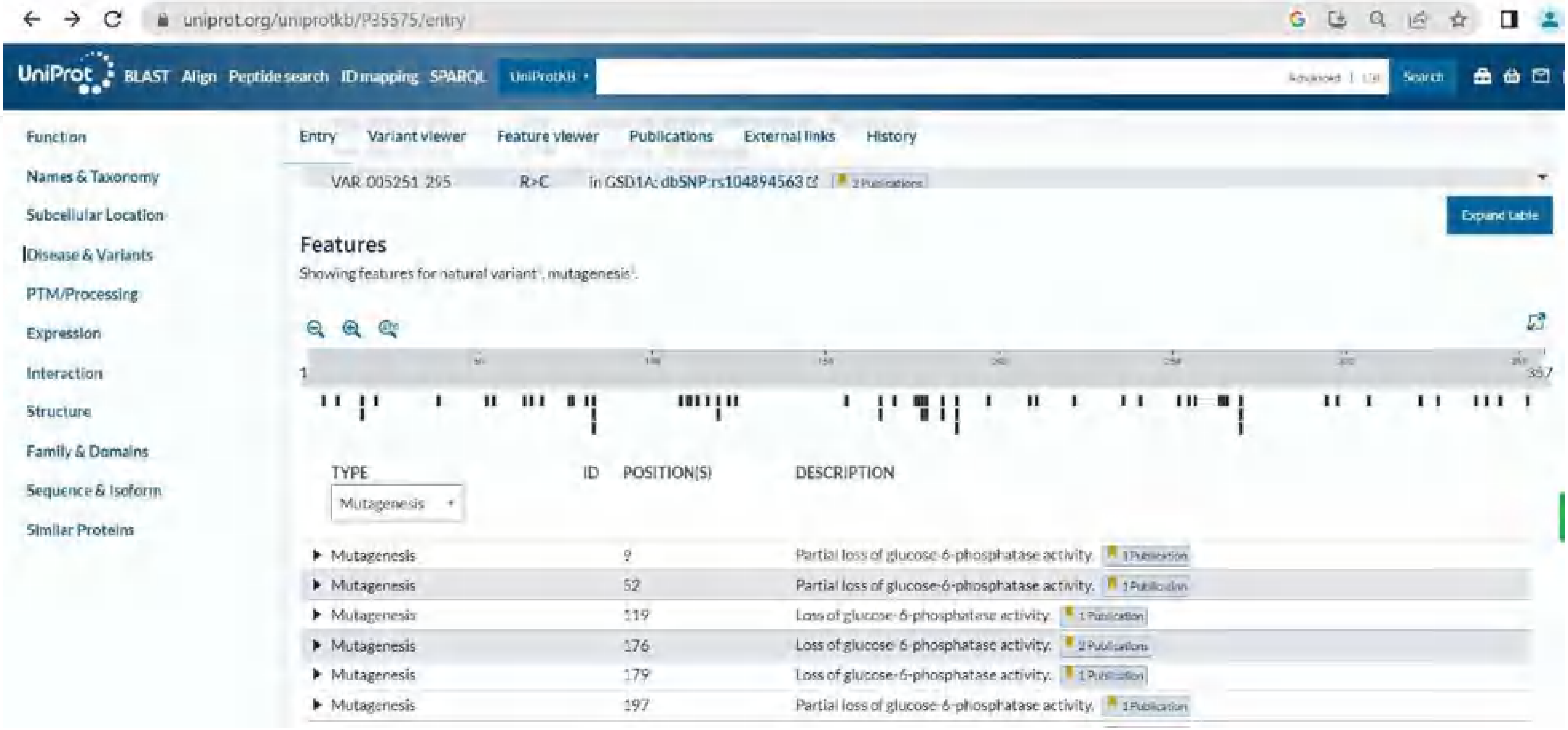
See also MIM:232200

Natural variants in GSD1A

VARIANT ID	POSITION(S)	CHANGE	DESCRIPTION
VAR_046249	5	M>R	in GSD1A; dbSNP:rs1250172816 1 Publication
VAR_046250	16	T>A	in GSD1A; dbSNP:rs761839506 1 Publication
VAR_046251	16	T>R	in GSD1A; complete loss of glucose-6-phosphatase activity and reduced enzyme stability; dbSNP:rs1555558914 1 Publication
VAR_009202	20	Q>R	in GSD1A; dbSNP:rs2056021410 2 Publications

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

MUTATION AND FUNCTIONS OF MUTANTS



VAR 005251 295 R>C in GSD1A; dbSNP:rs104894563 2 Publications

Features

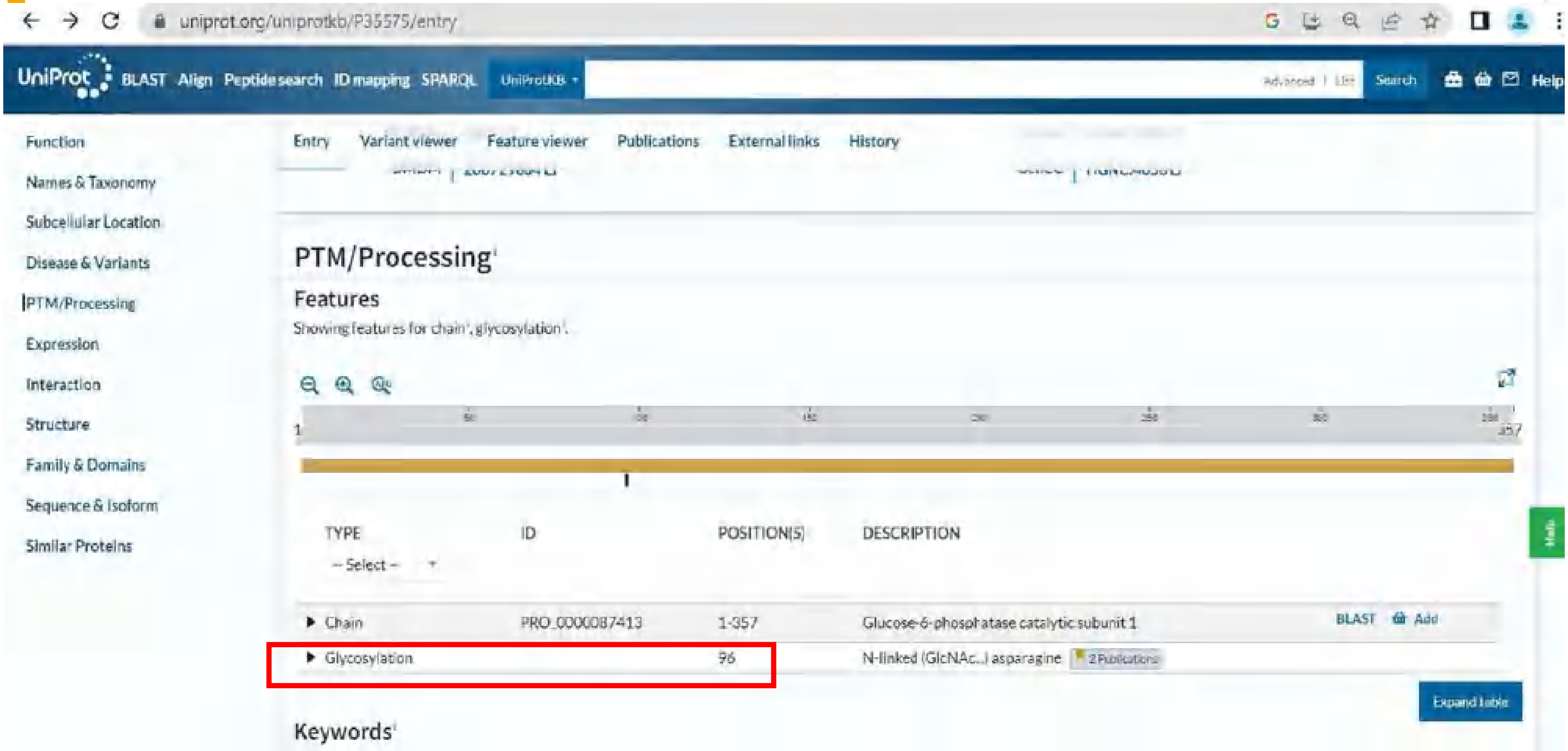
Showing features for natural variant, mutagenesis.

TYPE: Mutagenesis

TYPE	ID	POSITION(S)	DESCRIPTION
Mutagenesis		9	Partial loss of glucose-6-phosphatase activity. 1 Publication
Mutagenesis		52	Partial loss of glucose-6-phosphatase activity. 1 Publication
Mutagenesis		119	Loss of glucose-6-phosphatase activity. 1 Publication
Mutagenesis		176	Loss of glucose-6-phosphatase activity. 2 Publications
Mutagenesis		179	Loss of glucose-6-phosphatase activity. 1 Publication
Mutagenesis		197	Partial loss of glucose-6-phosphatase activity. 1 Publication

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

POST TRANSLATIONAL MODIFICATIONS-PROCESSING



PTM/Processing

Features

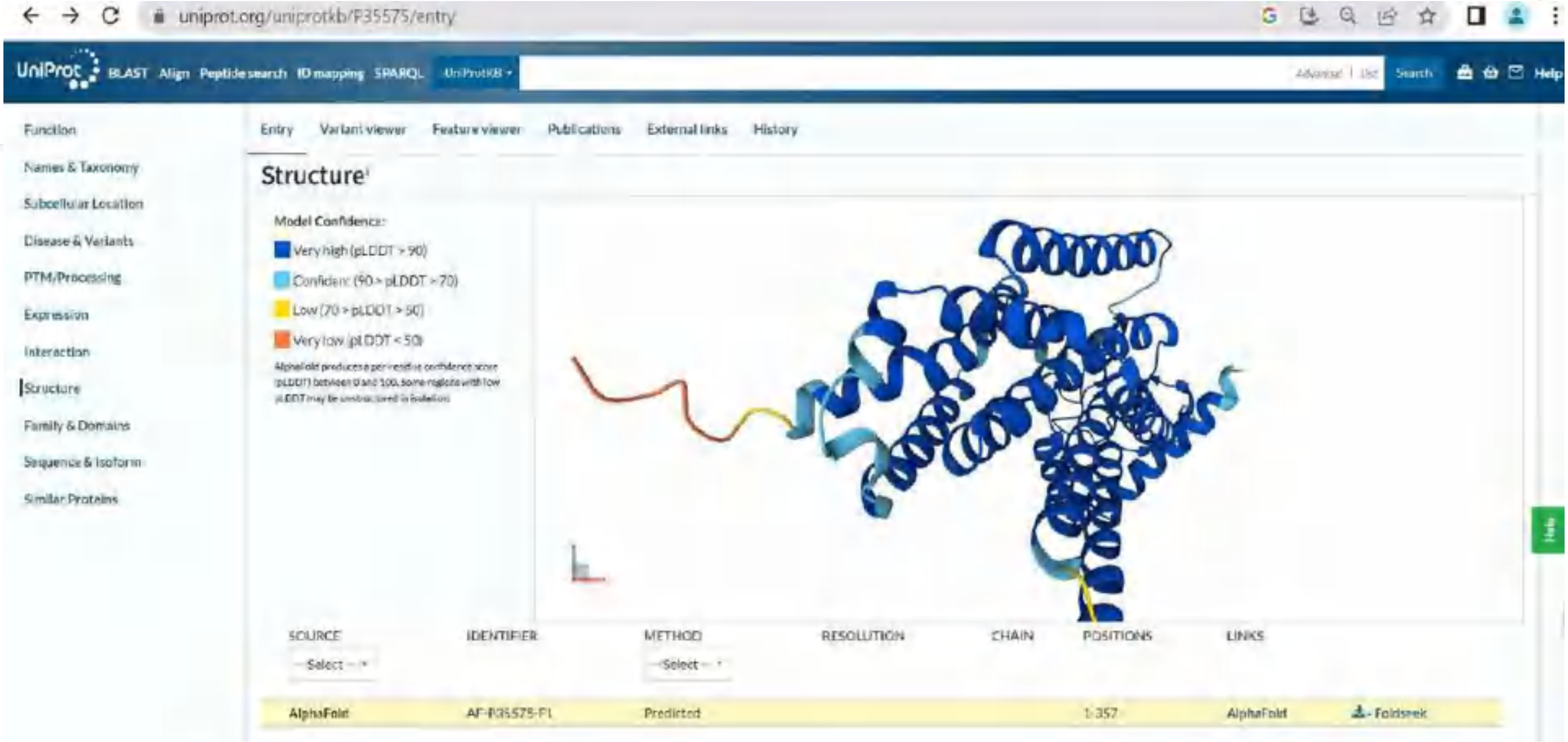
Showing features for chain, glycosylation.

TYPE	ID	POSITION(S)	DESCRIPTION
Chain	PRO_0000087413	1-357	Glucose-6-phosphatase catalytic subunit 1
Glycosylation		96	N-linked (GlcNAc...) asparagine 2 Publications

Keywords

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

3D Predicted Structure of G6PC1 (Isoform 1)



Structure

Model Confidence:

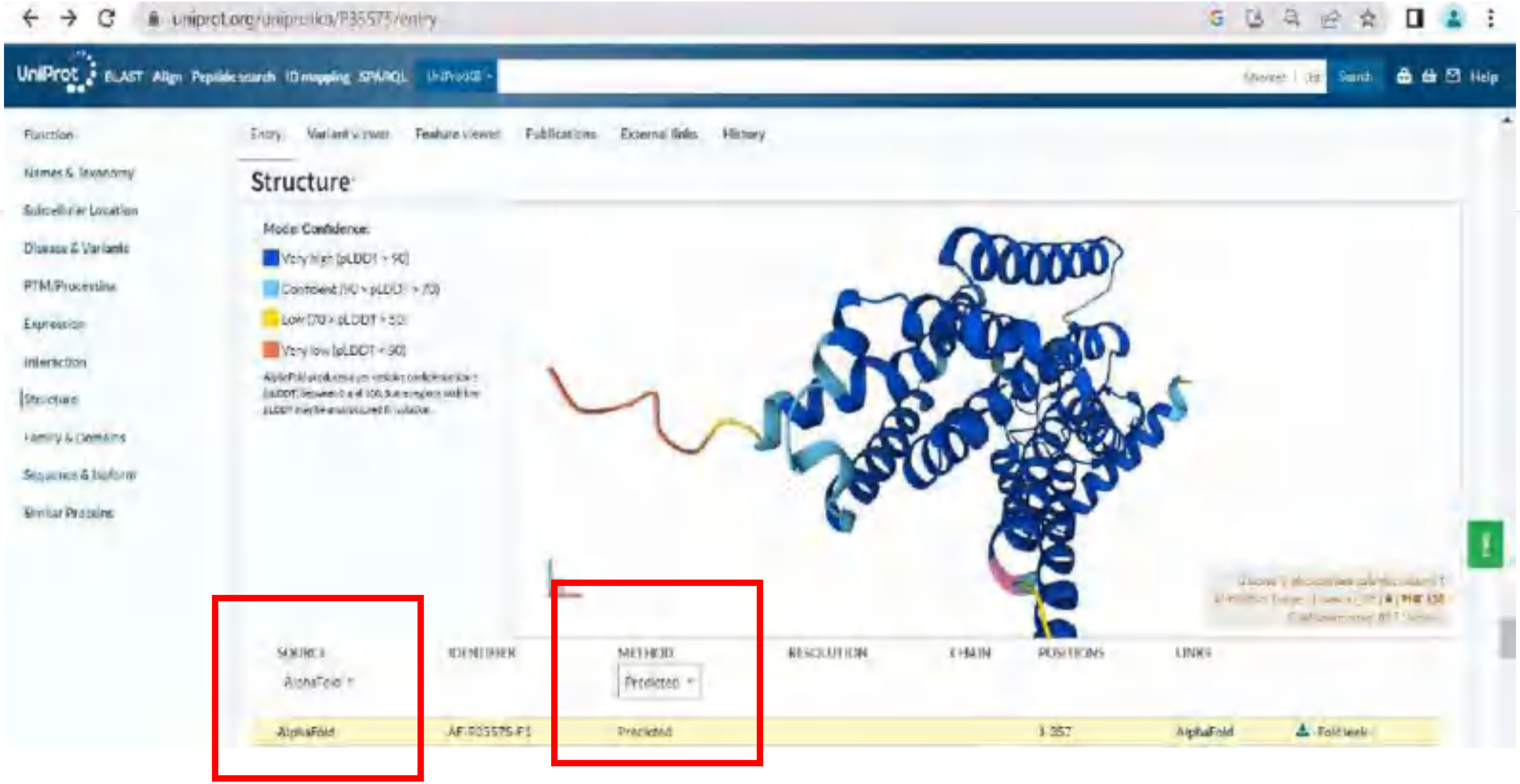
- 🟦 Very high (pLDDT > 90)
- 🟦 Confident (90 > pLDDT > 70)
- 🟨 Low (70 > pLDDT > 50)
- 🟧 Very low (pLDDT < 50)

AlphaFold produces a per-residue confidence score (pLDDT) between 0 and 100. Some regions with low pLDDT may be unstructured in isolation.

SOURCE --Select--	IDENTIFIER	METHOD --Select--	RESOLUTION	CHAIN	POSITIONS	LINKS	
AlphaFold	AF-P35575-F1	Predicted			1-357	AlphaFold	Foldseek

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

3D Predicted Structure of G6PC1 (Isoform 1)



Structure

Model Confidence:

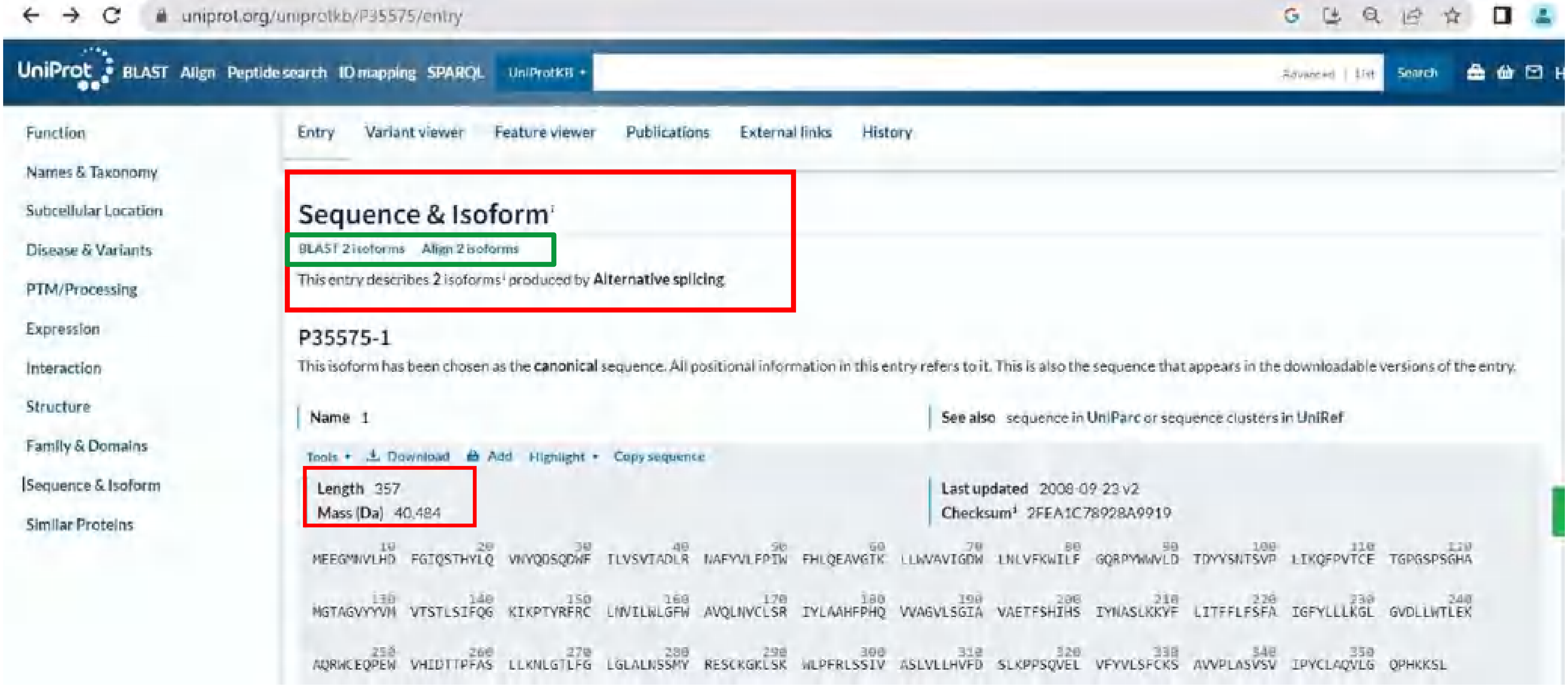
- 🟦 Very high (pLDDT > 90)
- 🟦 Confident (90 > pLDDT > 70)
- 🟨 Low (70 > pLDDT > 50)
- 🟧 Very low (pLDDT < 50)

SOURCE AlphaFold	IDENTIFIER	METHOD Predicted	RESOLUTION	CHAIN	POSITIONS	LINKS
AlphaFold	AF-P35575-F1	Predicted			1-357	AlphaFold Foldseek

AlphaFold is an AI system developed by DeepMind that predicts a protein's 3D structure from its amino acid sequence. It regularly achieves accuracy competitive with experiment.

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

SEQUENCE OF ISOFORM 1 of G6PC1



Sequence & Isoform

This entry describes 2 isoforms produced by **Alternative splicing**

P35575-1

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

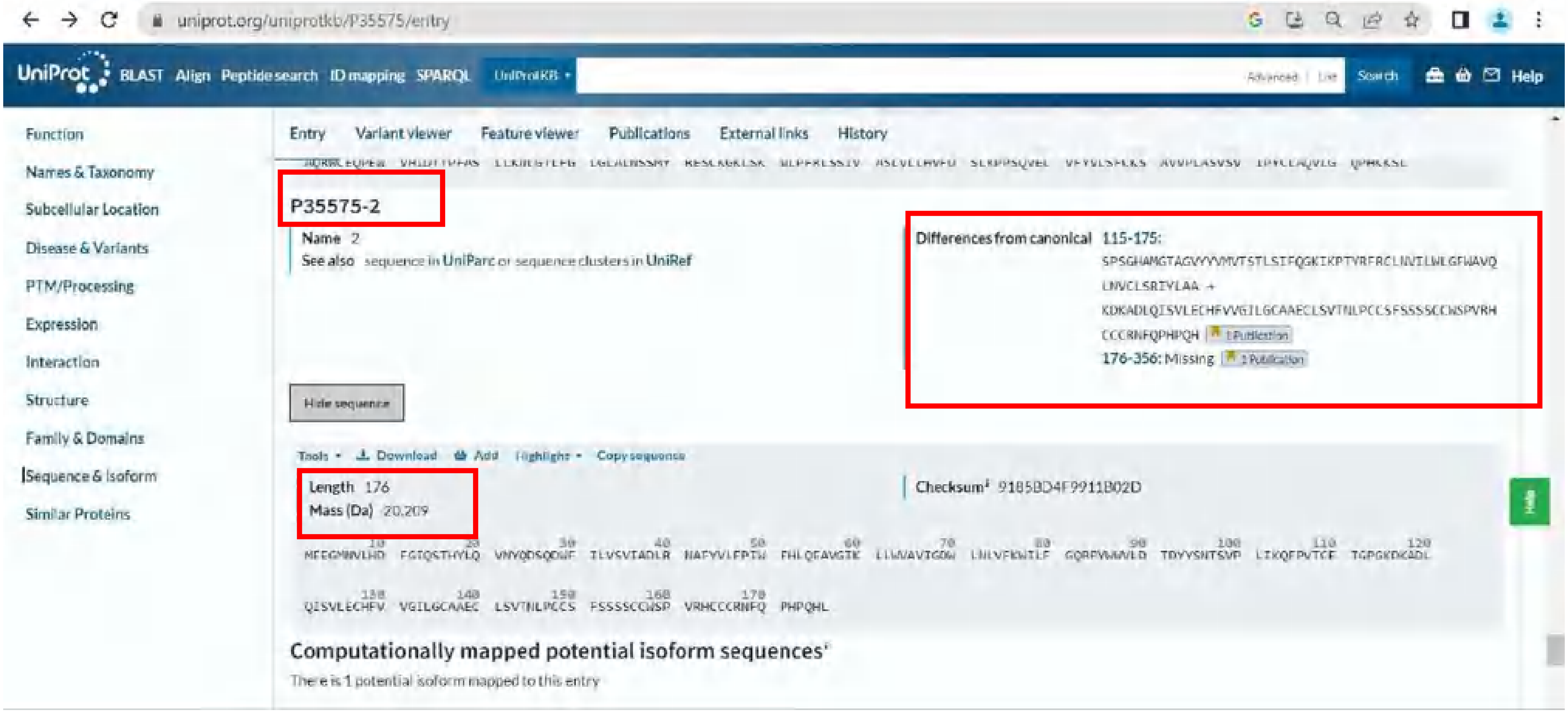
Length 357
Mass (Da) 40,484

Last updated 2008-09-23 v2
Checksum 2FEA1C78928A9919

```
        10         20         30         40         50         60         70         80         90        100        110        120
MEEGMNVLHD FGIQSTHYLQ VNYQDSQDWF ILVSVIADLR NAFYVLFPIW FHLQEAVGTK LLWVAVIGDW LNLVFKWILF GQRPYWWVLD TDYYSNTSVP LIKQFPVTCE TGPGSPSGHA

       130        140        150        160        170        180        190        200        210        220        230        240
MGTAGVYYVM VTSTLSIFQG KIKPTYRFRC LNVILWLGFW AVQLNVCLSR IYLAAHFPHQ VVAGVLSGIA VAETFSHIHS IYNASLKKYF LITFFLFSFA IGFYLLLKGL GVDLLWTLEK

       250        260        270        280        290        300        310        320        330        340        350
AQRWCEQPEW VHIDTTPFAS LLKNLGTLFG LGLALNSSMY RESCKGKLSK WLPFRLSSIV ASLVLLHVFD SLKPPSQVEL VFYVLSFCKS AVVPLASVSV IPYCLAQVLG QPHKKSL
```

Continue on https://www.uniprot.org/uniprotkb/P35575/entry

SEQUENCE OF ISOFORM 2 of G6PC1



```
AQRWCEQPEW VHIDTTPFAS LLKNLGTLFG LGLALNSSMY RESCKGKLSK WLPFRLSSIV ASLVLLHVFD SLKPPSQVEL VFYVLSFCKS AVVPLASVSV IPYCLAQVLG QPHKKSL
```

P35575-2

Name 2

See also sequence in UniParc or sequence clusters in UniRef

Differences from canonical 115-175:
SPSGHAMGTAGVYYVMVTSTLSIFQGKIKPTYRFRCLNVILWLGFWAVQ
LNVCLSRIYLAA →
KDKADLQISVLECHFVVGILGCAAECLSVTNLPCCSFSSSSCCWSPVRH
CCCRNFQPHPQH
176-356: Missing

Length 176
Mass (Da) 20,209

Checksum 9185BD4F9911B02D

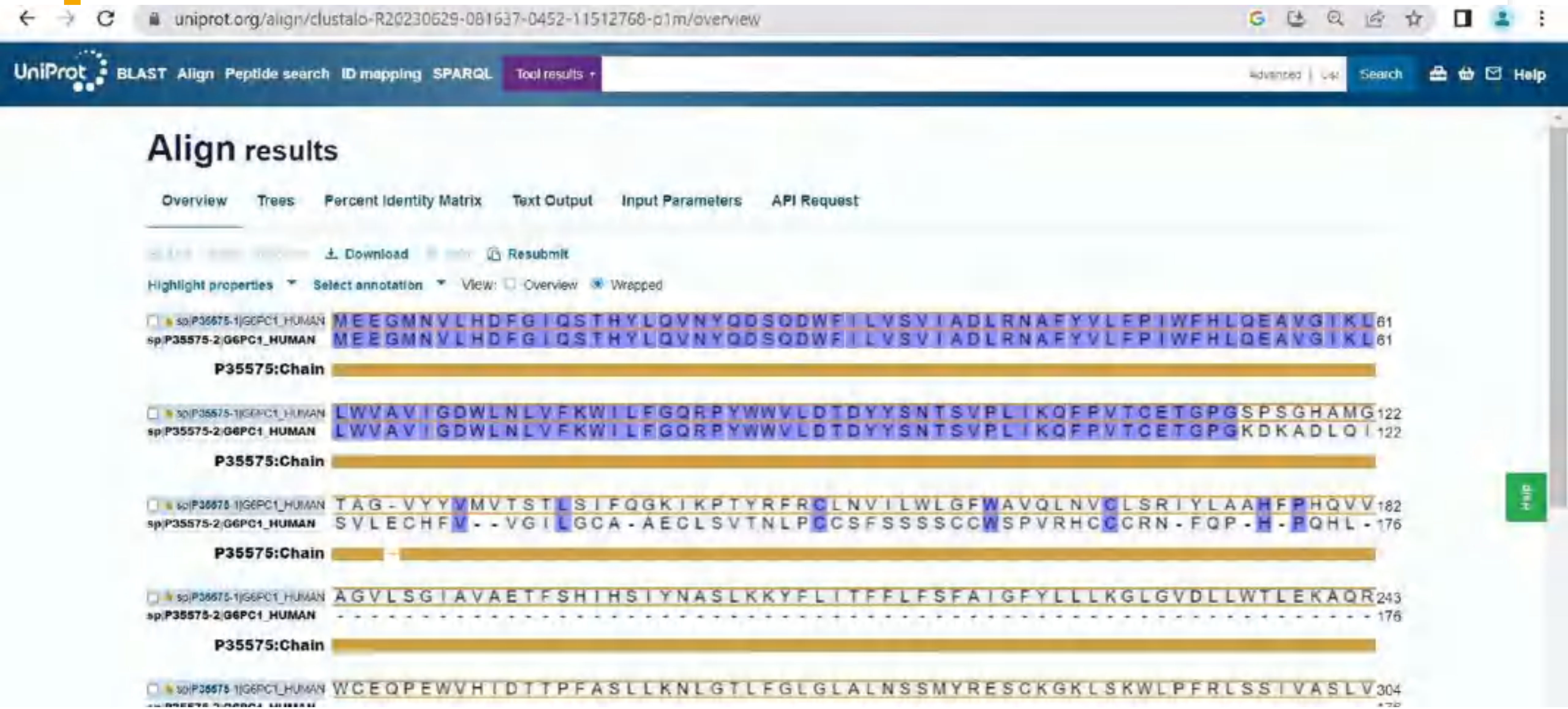
```
        10         20         30         40         50         60         70         80         90        100        110        120
MEEGMNVLHD FGIQSTHYLQ VNYQDSQDWF ILVSVIADLR NAFYVLFPIW FHLQEAVGIK LLWVAVIGDW LNLVFKWILF GQRPYWWVLD TDYYSNTSVP LIKQFPVTCE TGPGKDKADL

       130        140        150        160        170
QISVLECHFV VGILGCAAEC LSVTNLPCCS FSSSSCCWSP VRHCCCRNFQ PHPQHL
```

Computationally mapped potential isoform sequences

There is 1 potential isoform mapped to this entry

https://www.uniprot.org/uniprotkb/P35575/entry

CLICK ON ALIGN THE TWO ISOFORMS in the above webpage below SEQUENCE and ISOFORM



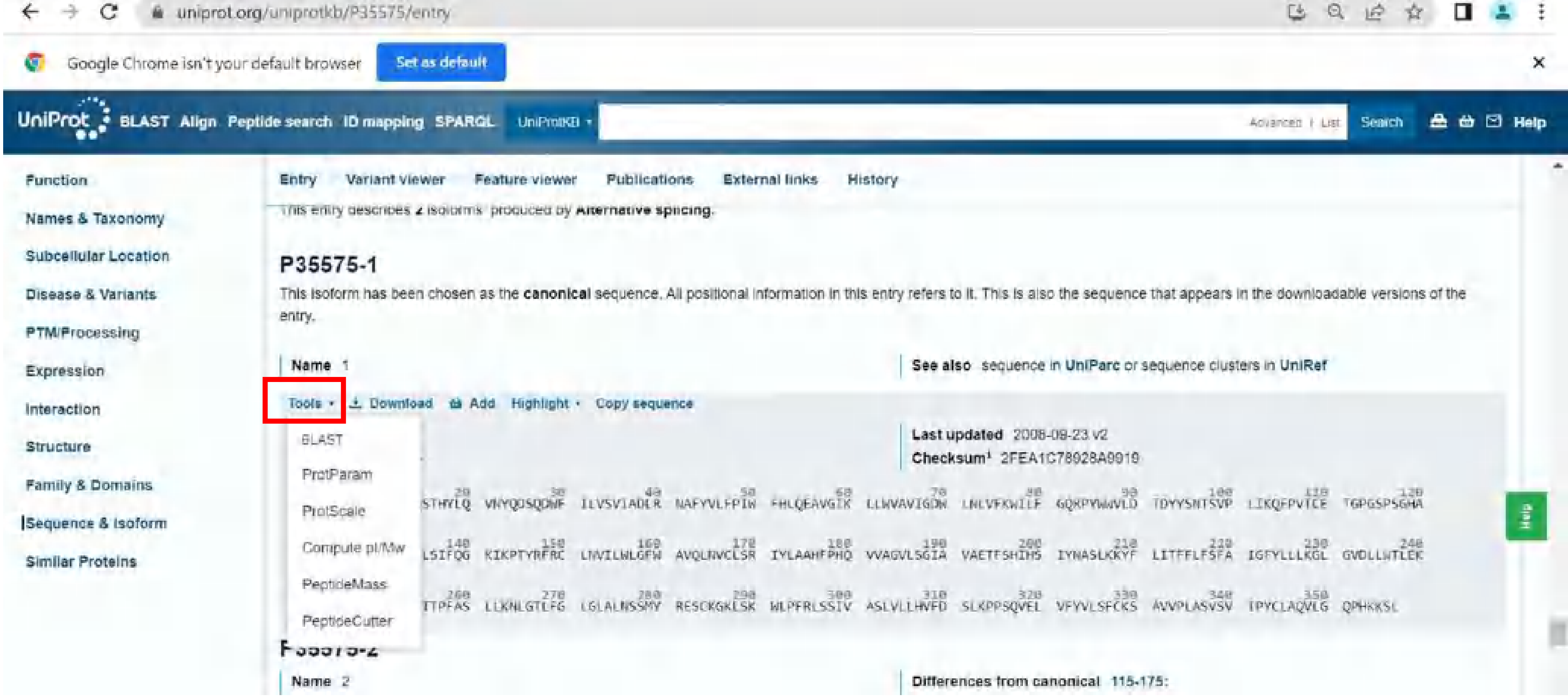
uniprot.org/align/clustalo-R20230629-081637-0452-11512768-p1m/overview

Align results

```
☐ sp|P35575-1|G6PC1_HUMAN  MEEGMNVLHDFGIQSTHYLQVNYQDSQDWFILVSVIADLRNAFYVLFPIWFHLQEAVGIKL  61
sp|P35575-2|G6PC1_HUMAN    MEEGMNVLHDFGIQSTHYLQVNYQDSQDWFILVSVIADLRNAFYVLFPIWFHLQEAVGIKL  61
P35575:Chain

☐ sp|P35575-1|G6PC1_HUMAN  LWVAVIGDWLNLVFKWILFGQRPYWWVLDTDYYSNTSVPLIKQFPVTCETGPGSPSGHAMG  122
sp|P35575-2|G6PC1_HUMAN    LWVAVIGDWLNLVFKWILFGQRPYWWVLDTDYYSNTSVPLIKQFPVTCETGPGKDKADLQI  122
P35575:Chain

☐ sp|P35575-1|G6PC1_HUMAN  TAG-VYYVMVTSTLSIFQGKIKPTYRFRCLNVILWLGFWAVQLNVCLSRIYLAAHFPHQVV  182
sp|P35575-2|G6PC1_HUMAN    SVLECHFV--VGILGCA-AECLSVTNLPCCSFSSSSCCWSPVRHCCCRN-FQP-H-PQHL-  176
P35575:Chain

☐ sp|P35575-1|G6PC1_HUMAN  AGVLSGIAVAETFSHIHSIYNASLKKYFLITFFLFSFAIGFYLLLKGLGVDLLWTLEKAQR  243
sp|P35575-2|G6PC1_HUMAN    -------------------------------------------------------------  176
P35575:Chain

☐ sp|P35575-1|G6PC1_HUMAN  WCEQPEWVHIDTTPFASLLKNLGTLFGLGLALNSSMYRESCKGKLSKWLPFRLSSIVASLV  304
```

https://www.uniprot.org/uniprotkb/P35575/entry

CLICK ON BLAST



This entry describes 2 isoforms produced by **Alternative splicing**.

P35575-1

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

Last updated 2008-09-23 v2

Checksum[1] 2FEA1C78928A0919

```
      20         30         40         50         60         70         80         90        100        110        120
STHYLQ VNYQDSQDWF ILVSVIADLR NAFYVLFPIW FHLQEAVGIK LLWVAVIGDW LNLVFKWILF GQRPYWWVLD TDYYSNTSVP LIKQFPVTCE TGPGSPSGHA

     140        150        160        170        180        190        200        210        220        230        240
LSIFQG KIKPTYRFRC LNVILWLGFW AVQLNVCLSR IYLAAHFPHQ VVAGVLSGIA VAETFSHIHS IYNASLKKYF LITFFLFSFA IGFYLLLKGL GVDLLWTLEK

     260        270        280        290        300        310        320        330        340        350
TTPFAS LLKNLGTLFG LGLALNSSMY RESCKGKLSK WLPFRLSSIV ASLVLLHVFD SLKPPSQVEL VFYVLSFCKS AVVPLASVSV IPYCLAQVLG QPHKKSL
```

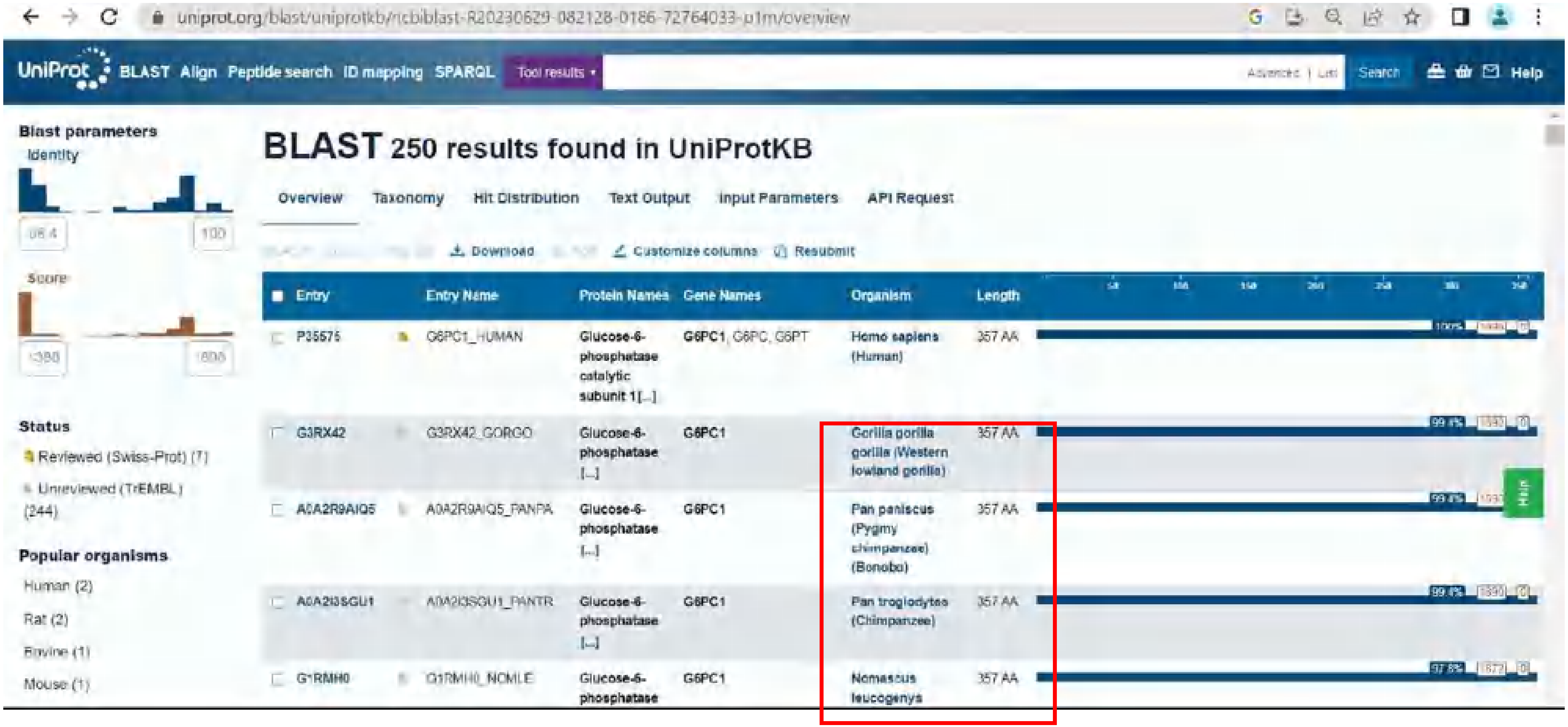
P35575-2

Name 2

Differences from canonical 115-175:

https://www.uniprot.org/uniprotkb/P35575/entry

BLAST RESULTS OF G6PC1 Enzyme against the UniProt Database



Blast parameters

Identity

Score

Status

Reviewed (Swiss-Prot) (7)

Unreviewed (TrEMBL) (244)

Popular organisms

Human (2)

Rat (2)

Bovine (1)

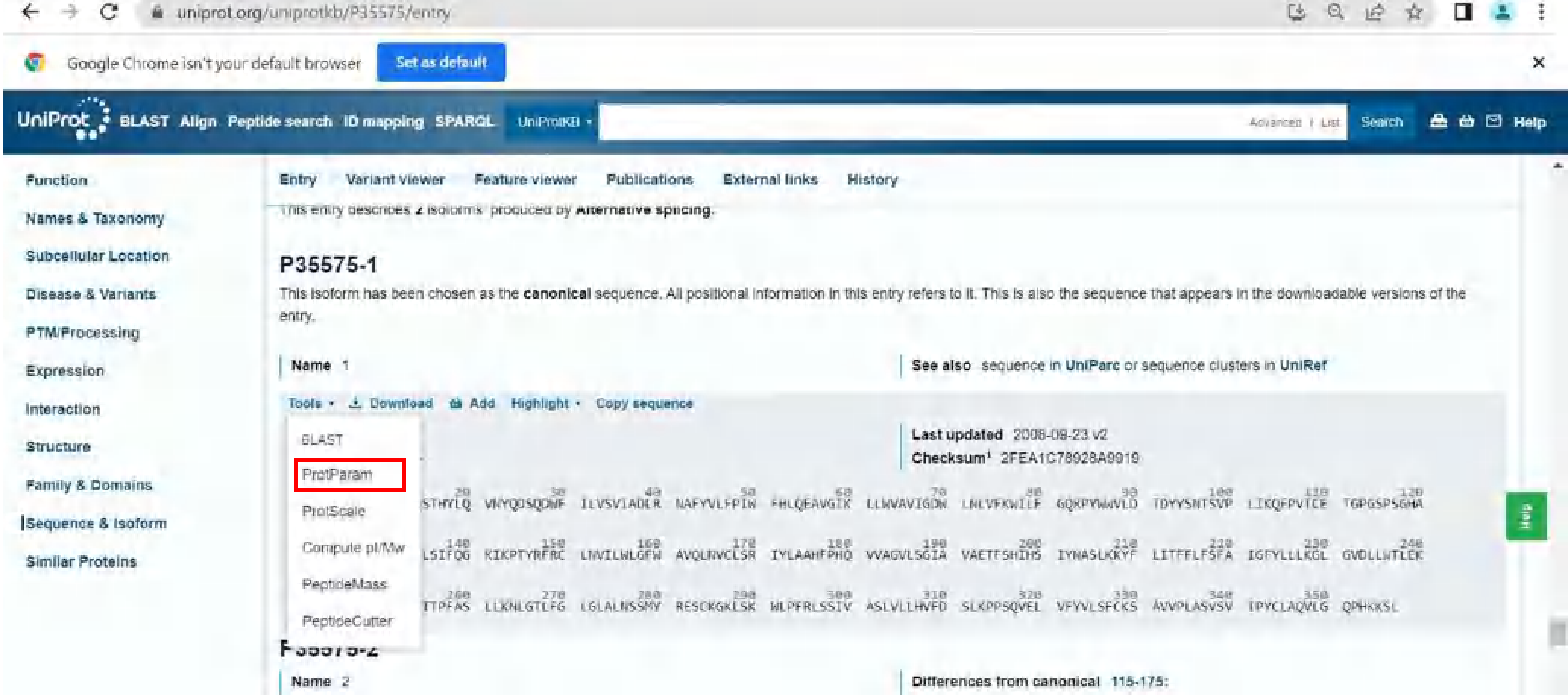
Mouse (1)

BLAST 250 results found in UniProtKB

Entry	Entry Name	Protein Names	Gene Names	Organism	Length
P35575	G6PC1_HUMAN	Glucose-6-phosphatase catalytic subunit 1[...]	G6PC1, G6PC, G6PT	Homo sapiens (Human)	357 AA
G3RX42	G3RX42_GORGO	Glucose-6-phosphatase [...]	G6PC1	Gorilla gorilla gorilla (Western lowland gorilla)	357 AA
A0A2R9AIQ5	A0A2R9AIQ5_PANPA	Glucose-6-phosphatase [...]	G6PC1	Pan paniscus (Pygmy chimpanzee) (Bonobo)	357 AA
A0A2I3SGU1	A0A2I3SGU1_PANTR	Glucose-6-phosphatase [...]	G6PC1	Pan troglodytes (Chimpanzee)	357 AA
G1RMH0	G1RMH0_NOMLE	Glucose-6-phosphatase	G6PC1	Nomascus leucogenys	357 AA

https://www.uniprot.org/uniprotkb/P35575/entry

CLICK ON BLAST



This entry describes 2 isoforms produced by **Alternative splicing**.

P35575-1

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

Last updated 2008-09-23 v2

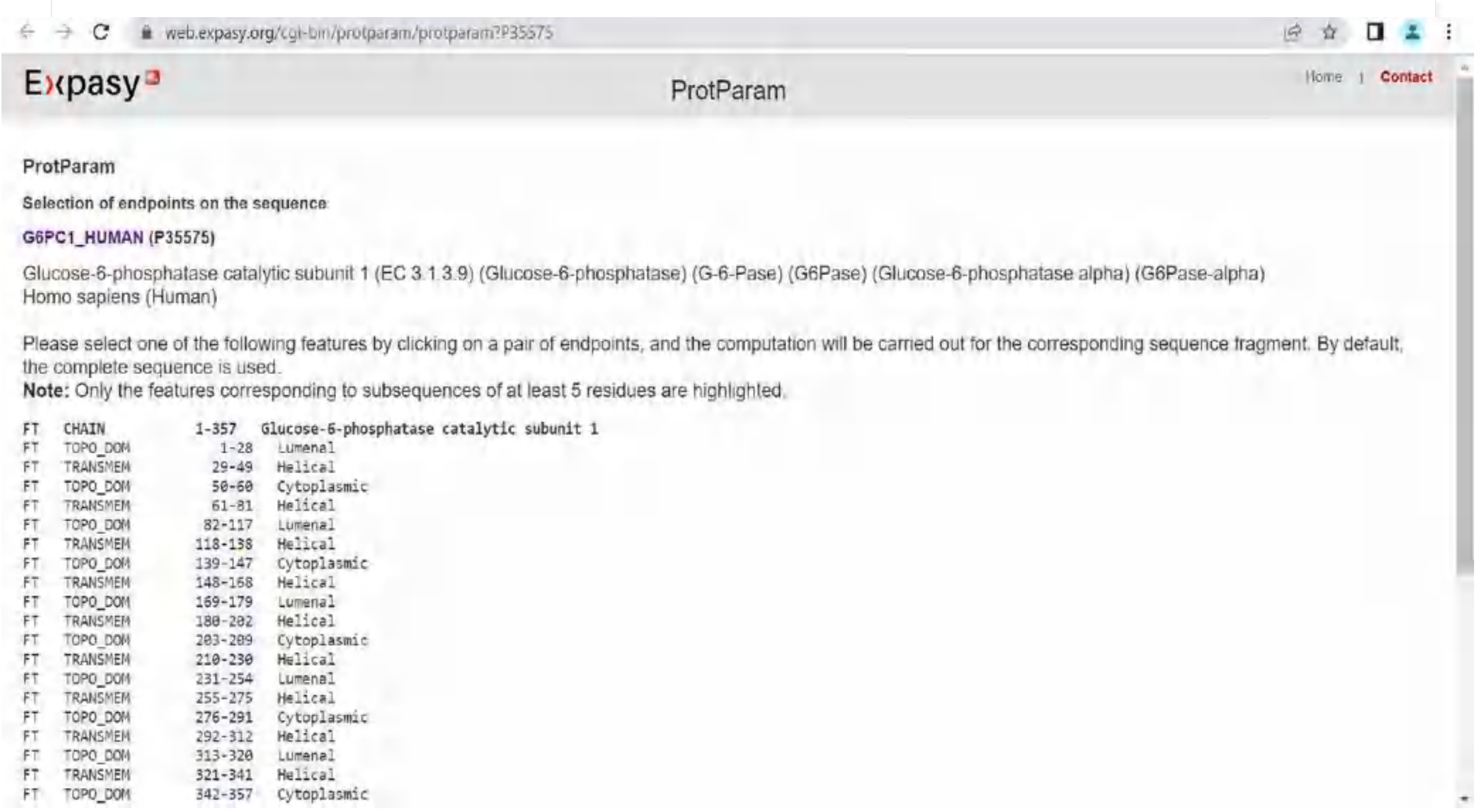
Checksum[i] 2FEA1C78928A0918

```
     20         30         40         50         60         70         80         90        100        110        120
STHYLQ VNYQDSQDWF ILVSVIADLR NAFYVLFPIW FHLQEAVGIK LLWVAVIGDW LNLVFKWILF GQRPYWWVLD TDYYSNTSVP LIKQFPVTCE TGPGSPSGHA
    140        150        160        170        180        190        200        210        220        230        240
LSIFQG KIKPTYRFRC LNVILWLGFW AVQLNVCLSR IYLAAHFPHQ VVAGVLSGIA VAETFSHIHS IYNASLKKYF LITFFLFSFA IGFYLLLKGL GVDLLWTLEK
    260        270        280        290        300        310        320        330        340        350
TTPFAS LLKNLGTLFG LGLALNSSMY RESCKGKLSK WLPFRLSSIV ASLVLLHVFD SLKPPSQVEL VFYVLSFCKS AVVPLASVSV IPYCLAQVLG QPHKKSL
```

P35575-2

Name 2

Differences from canonical 115-175:



ProtParam

Selection of endpoints on the sequence

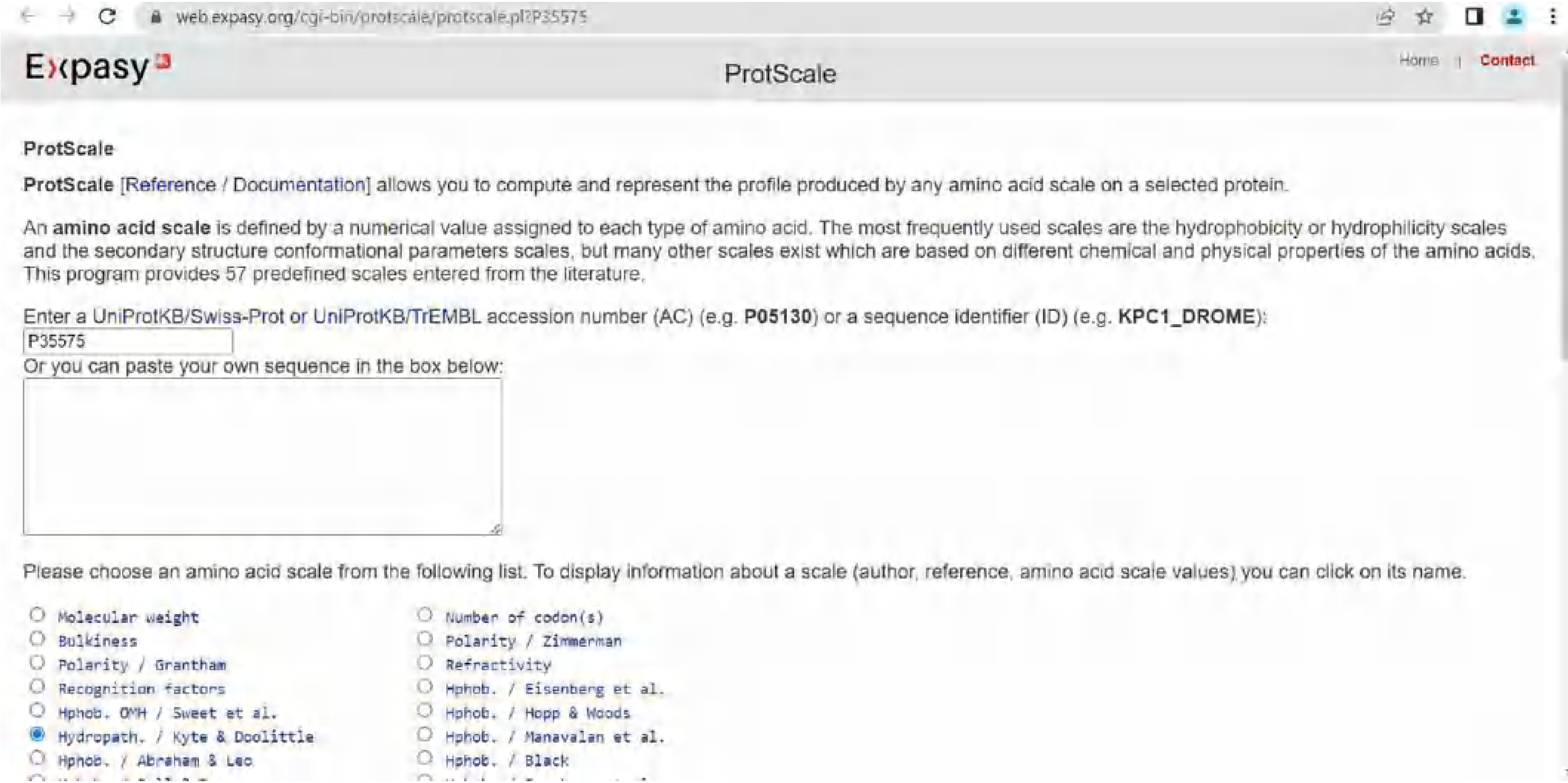
G6PC1_HUMAN (P35575)

Glucose-6-phosphatase catalytic subunit 1 (EC 3.1.3.9) (Glucose-6-phosphatase) (G-6-Pase) (G6Pase) (Glucose-6-phosphatase alpha) (G6Pase-alpha)
Homo sapiens (Human)

Please select one of the following features by clicking on a pair of endpoints, and the computation will be carried out for the corresponding sequence fragment. By default, the complete sequence is used.
Note: Only the features corresponding to subsequences of at least 5 residues are highlighted.

```
FT   CHAIN           1-357   Glucose-6-phosphatase catalytic subunit 1
FT   TOPO_DOM           1-28   Lumenal
FT   TRANSMEM          29-49   Helical
FT   TOPO_DOM          50-60   Cytoplasmic
FT   TRANSMEM          61-81   Helical
FT   TOPO_DOM         82-117   Lumenal
FT   TRANSMEM        118-138   Helical
FT   TOPO_DOM        139-147   Cytoplasmic
FT   TRANSMEM        148-168   Helical
FT   TOPO_DOM        169-179   Lumenal
FT   TRANSMEM        180-202   Helical
FT   TOPO_DOM        203-209   Cytoplasmic
FT   TRANSMEM        210-230   Helical
FT   TOPO_DOM        231-254   Lumenal
FT   TRANSMEM        255-275   Helical
FT   TOPO_DOM        276-291   Cytoplasmic
FT   TRANSMEM        292-312   Helical
FT   TOPO_DOM        313-320   Lumenal
FT   TRANSMEM        321-341   Helical
FT   TOPO_DOM        342-357   Cytoplasmic
```

PROTSCALE



ProtScale

ProtScale [Reference / Documentation] allows you to compute and represent the profile produced by any amino acid scale on a selected protein.

An **amino acid scale** is defined by a numerical value assigned to each type of amino acid. The most frequently used scales are the hydrophobicity or hydrophilicity scales and the secondary structure conformational parameters scales, but many other scales exist which are based on different chemical and physical properties of the amino acids. This program provides 57 predefined scales entered from the literature.

Enter a UniProtKB/Swiss-Prot or UniProtKB/TrEMBL accession number (AC) (e.g. **P05130**) or a sequence identifier (ID) (e.g. **KPC1_DROME**):

P35575

Or you can paste your own sequence in the box below:

Please choose an amino acid scale from the following list. To display information about a scale (author, reference, amino acid scale values) you can click on its name.

○ Molecular weight
○ Bulkiness
○ Polarity / Grantham
○ Recognition factors
○ Hphob. OMH / Sweet et al.
● Hydropath. / Kyte & Doolittle
○ Hphob. / Abraham & Leo

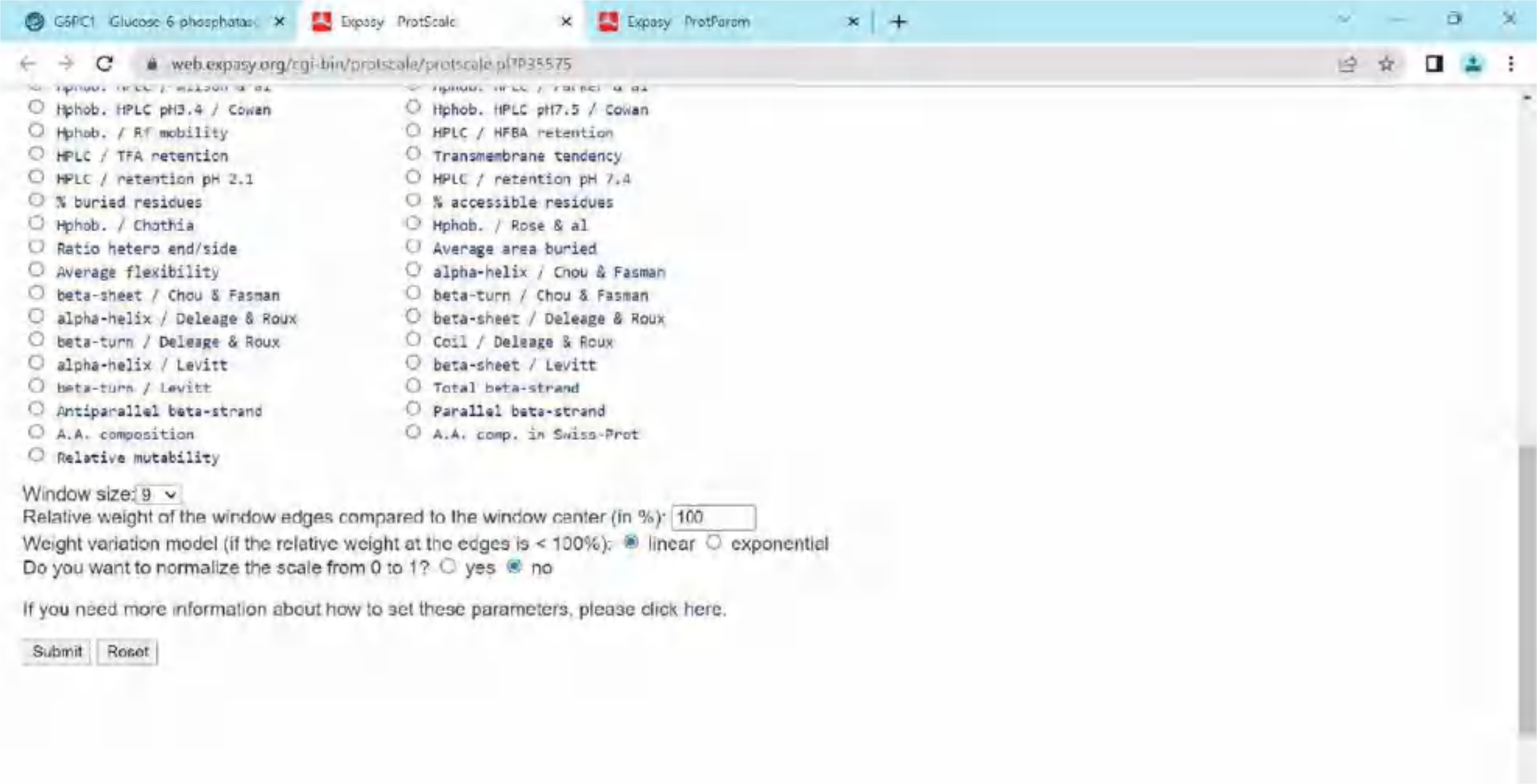
○ Number of codon(s)
○ Polarity / Zimmerman
○ Refractivity
○ Hphob. / Eisenberg et al.
○ Hphob. / Hopp & Woods
○ Hphob. / Manavalan et al.
○ Hphob. / Black

PROTSCALE…

Please choose an amino acid scale from the following list. To display information about a scale (author, reference, amino acid scale values) you can click on its name.

- ○ Molecular weight
- ○ Bulkiness
- ○ Polarity / Grantham
- ○ Recognition factors
- ○ Hphob. OMH / Sweet et al.
- ● Hydropath. / Kyte & Doolittle
- ○ Hphob. / Abraham & Leo
- ○ Hphob. / Bull & Breese
- ○ Hphob. / Guy
- ○ Hphob. / Miyazawa et al.
- ○ Hphob. / Roseman
- ○ Hphob. / Wolfenden et al.
- ○ Hphob. HPLC / Wilson & al
- ○ Hphob. HPLC pH3.4 / Cowan
- ○ Hphob. / Rf mobility
- ○ HPLC / TFA retention
- ○ HPLC / retention pH 2.1
- ○ % buried residues
- ○ Hphob. / Chothia
- ○ Ratio hetero end/side
- ○ Average flexibility
- ○ beta-sheet / Chou & Fasman
- ○ alpha-helix / Deleage & Roux
- ○ beta-turn / Deleage & Roux
- ○ alpha-helix / Levitt
- ○ beta-turn / Levitt
- ○ Antiparallel beta-strand
- ○ A.A. composition
- ○ Number of codon(s)
- ○ Polarity / Zimmerman
- ○ Refractivity
- ○ Hphob. / Eisenberg et al.
- ○ Hphob. / Hopp & Woods
- ○ Hphob. / Manavalan et al.
- ○ Hphob. / Black
- ○ Hphob. / Fauchere et al.
- ○ Hphob. / Janin
- ○ Hphob. / Rao & Argos
- ○ Hphob. / Tanford
- ○ Hphob. / Welling & al
- ○ Hphob. HPLC / Parker & al
- ○ Hphob. HPLC pH7.5 / Cowan
- ○ HPLC / HFBA retention
- ○ Transmembrane tendency
- ○ HPLC / retention pH 7.4
- ○ % accessible residues
- ○ Hphob. / Rose & al
- ○ Average area buried
- ○ alpha-helix / Chou & Fasman
- ○ beta-turn / Chou & Fasman
- ○ beta-sheet / Deleage & Roux
- ○ Coil / Deleage & Roux
- ○ beta-sheet / Levitt
- ○ Total beta-strand
- ○ Parallel beta-strand
- ○ A.A. comp. in Swiss-Prot

PROTSCALE…



○ Hphob. HPLC pH3.4 / Cowan
○ Hphob. / Rf mobility
○ HPLC / TFA retention
○ HPLC / retention pH 2.1
○ % buried residues
○ Hphob. / Chothia
○ Ratio hetero end/side
○ Average flexibility
○ beta-sheet / Chou & Fasman
○ alpha-helix / Deleage & Roux
○ beta-turn / Deleage & Roux
○ alpha-helix / Levitt
○ beta-turn / Levitt
○ Antiparallel beta-strand
○ A.A. composition
○ Relative mutability

○ Hphob. HPLC pH7.5 / Cowan
○ HPLC / HFBA retention
○ Transmembrane tendency
○ HPLC / retention pH 7.4
○ % accessible residues
○ Hphob. / Rose & al
○ Average area buried
○ alpha-helix / Chou & Fasman
○ beta-turn / Chou & Fasman
○ beta-sheet / Deleage & Roux
○ Coil / Deleage & Roux
○ beta-sheet / Levitt
○ Total beta-strand
○ Parallel beta-strand
○ A.A. comp. in Swiss-Prot

Window size: 9

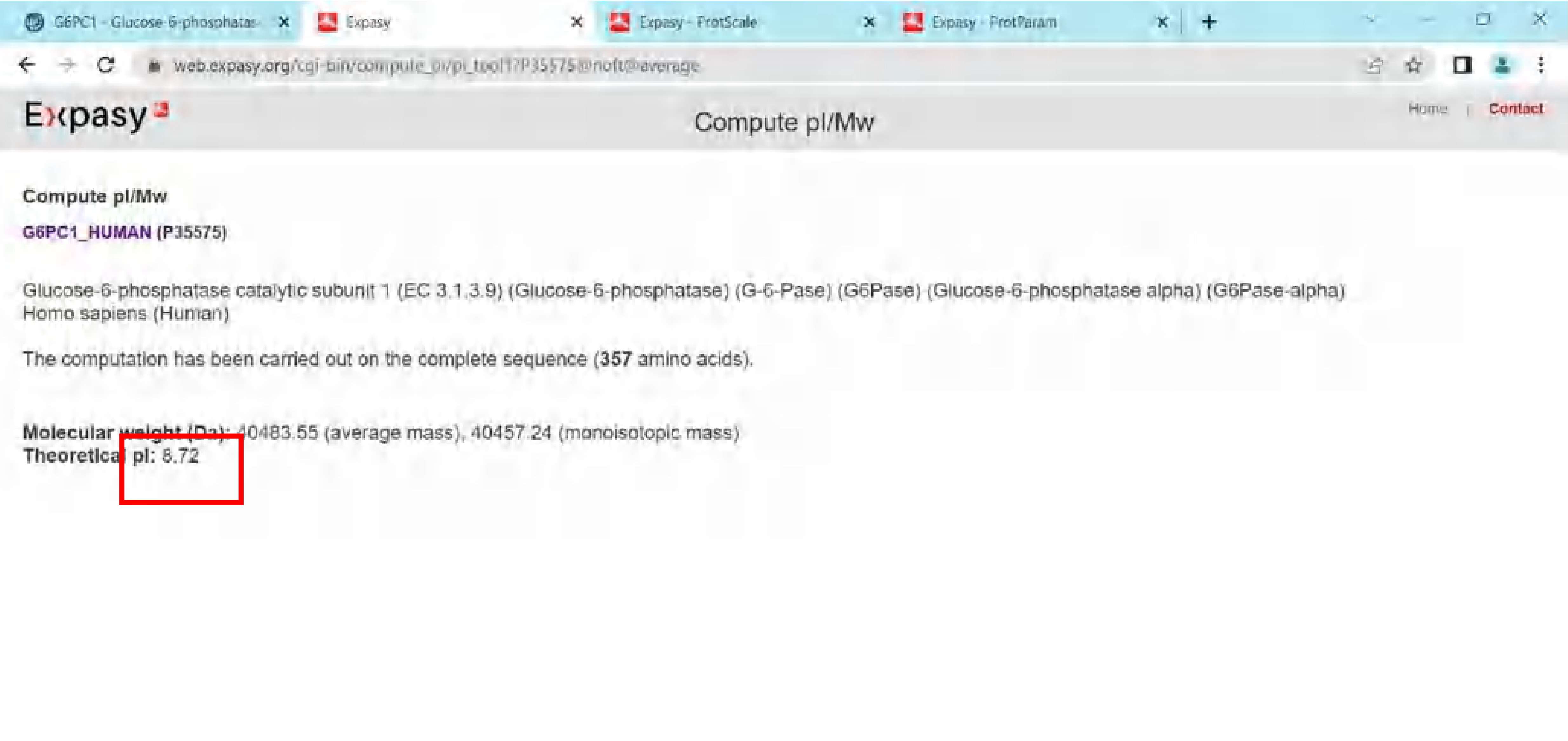
Relative weight of the window edges compared to the window center (in %): 100

Weight variation model (if the relative weight at the edges is < 100%): ● linear ○ exponential

Do you want to normalize the scale from 0 to 1? ○ yes ● no

If you need more information about how to set these parameters, please click here.

COMPUTE pI/Mw



Compute pI/Mw

Compute pI/Mw

G6PC1_HUMAN (P35575)

Glucose-6-phosphatase catalytic subunit 1 (EC 3.1.3.9) (Glucose-6-phosphatase) (G-6-Pase) (G6Pase) (Glucose-6-phosphatase alpha) (G6Pase-alpha)
Homo sapiens (Human)

The computation has been carried out on the complete sequence (**357** amino acids).

Molecular weight (Da): 40483.55 (average mass), 40457.24 (monoisotopic mass)
Theoretical pI: 8.72

Information of G6PC1 Retrieved from UniProt

Location within the cell: Endoplasmic Reticulum Membrane

Post Translational Modification – Gycosylation at position 96 (N) {N-Linked Glycosylation)

Variants that cause Glycogen Storage Disease 1A (GSD1A): 5M>R, 16 T>A, 16T>R, 20 Q>R, 38 D<V, 54Q>R, 63W>R, 65 A>P, 68 G>R, 76 K>N, 77 W>R, 81G>R.

Structure : Predicted by AlphaFold

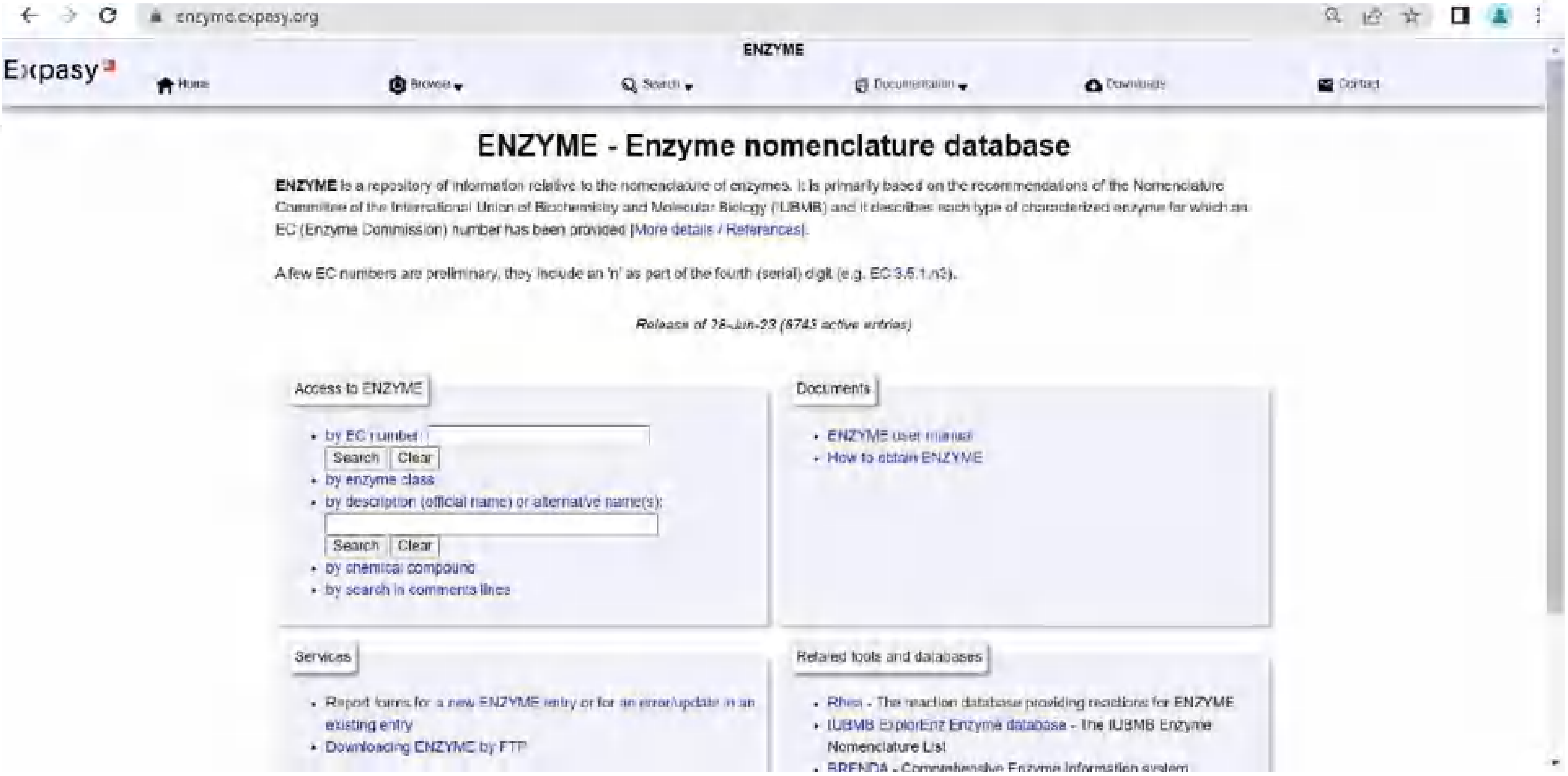
Isoforms 2 Created by Alternative Splicing. Isoform -1 (357 amino acids) , Isoform 2 (176 amino acids)

Amino acid Sequences of both isoforms obtained

Similarity of isoform -1 with glucose 6phosphatase with other species obtained by "BLAST"

Theoretical pI (isoelectric point of protein)=8.72

Go to Enzyme Database
https://enzyme.expasy.org/



ENZYME - Enzyme nomenclature database

ENZYME is a repository of information relative to the nomenclature of enzymes. It is primarily based on the recommendations of the Nomenclature Committee of the International Union of Biochemistry and Molecular Biology (IUBMB) and it describes each type of characterized enzyme for which an EC (Enzyme Commission) number has been provided [More details / References].

A few EC numbers are preliminary, they include an 'n' as part of the fourth (serial) digit (e.g. EC 3.5.1.n3).

Release of 28-Jun-23 (6743 active entries)

Access to ENZYME

- by EC number:
  Search Clear
- by enzyme class
- by description (official name) or alternative name(s):
  Search Clear
- by chemical compound
- by search in comments lines

Documents

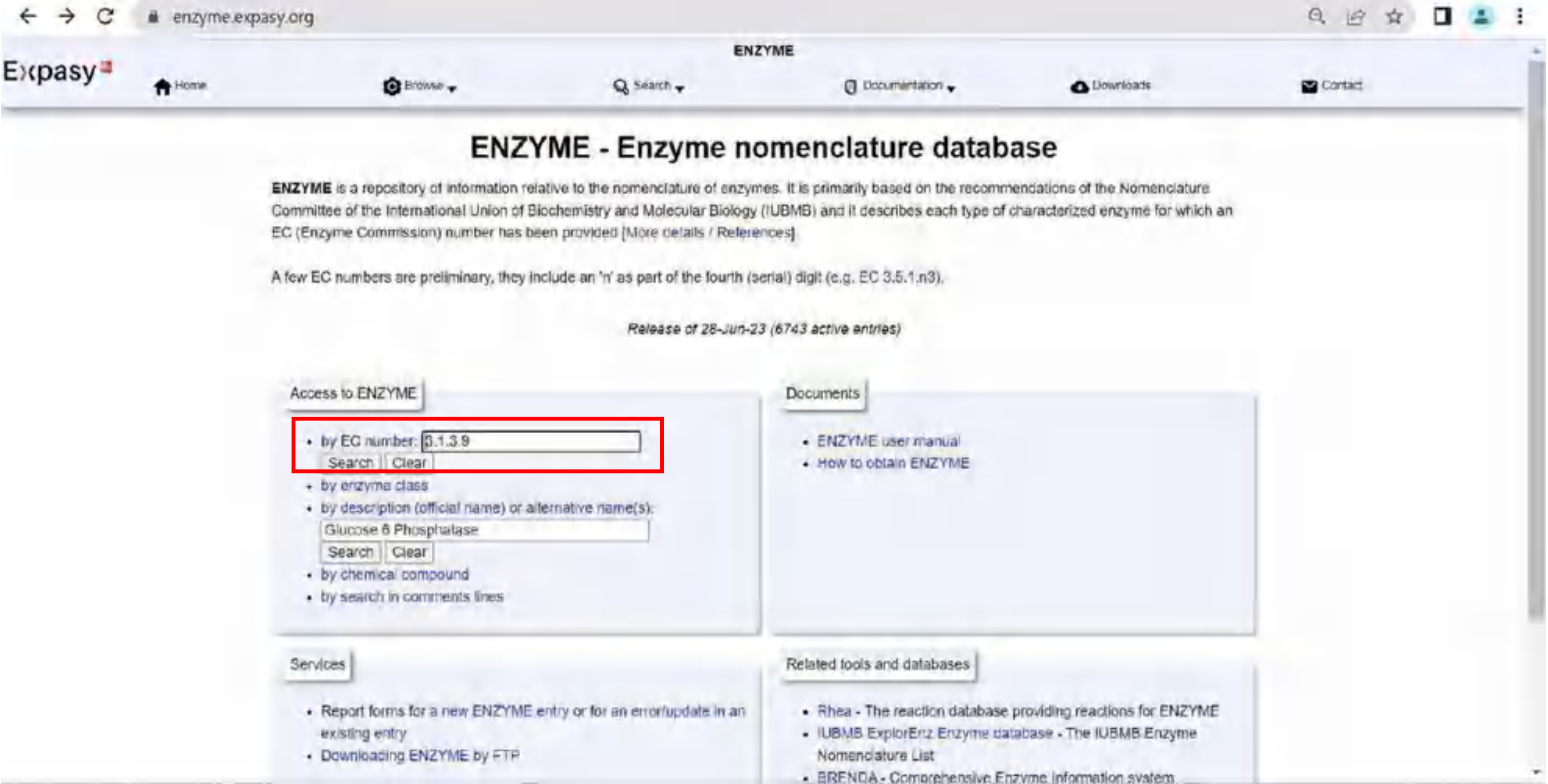
- ENZYME user manual
- How to obtain ENZYME

Services

- Report forms for a new ENZYME entry or for an error/update in an existing entry
- Downloading ENZYME by FTP

Related tools and databases

- Rhea - The reaction database providing reactions for ENZYME
- IUBMB ExplorEnz Enzyme database - The IUBMB Enzyme Nomenclature List
- BRENDA - Comprehensive Enzyme Information system



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A few EC numbers are preliminary, they include an 'n' as part of the fourth (serial) digit (e.g. EC 3.5.1.n3).

Release of 28-Jun-23 (6743 active entries)

Access to ENZYME

- by EC number: 3.1.3.9
  Search Clear
- by enzyme class
- by description (official name) or alternative name(s):
  Glucose 6 Phosphatase
  Search Clear
- by chemical compound
- by search in comments lines

Documents

- ENZYME user manual
- How to obtain ENZYME

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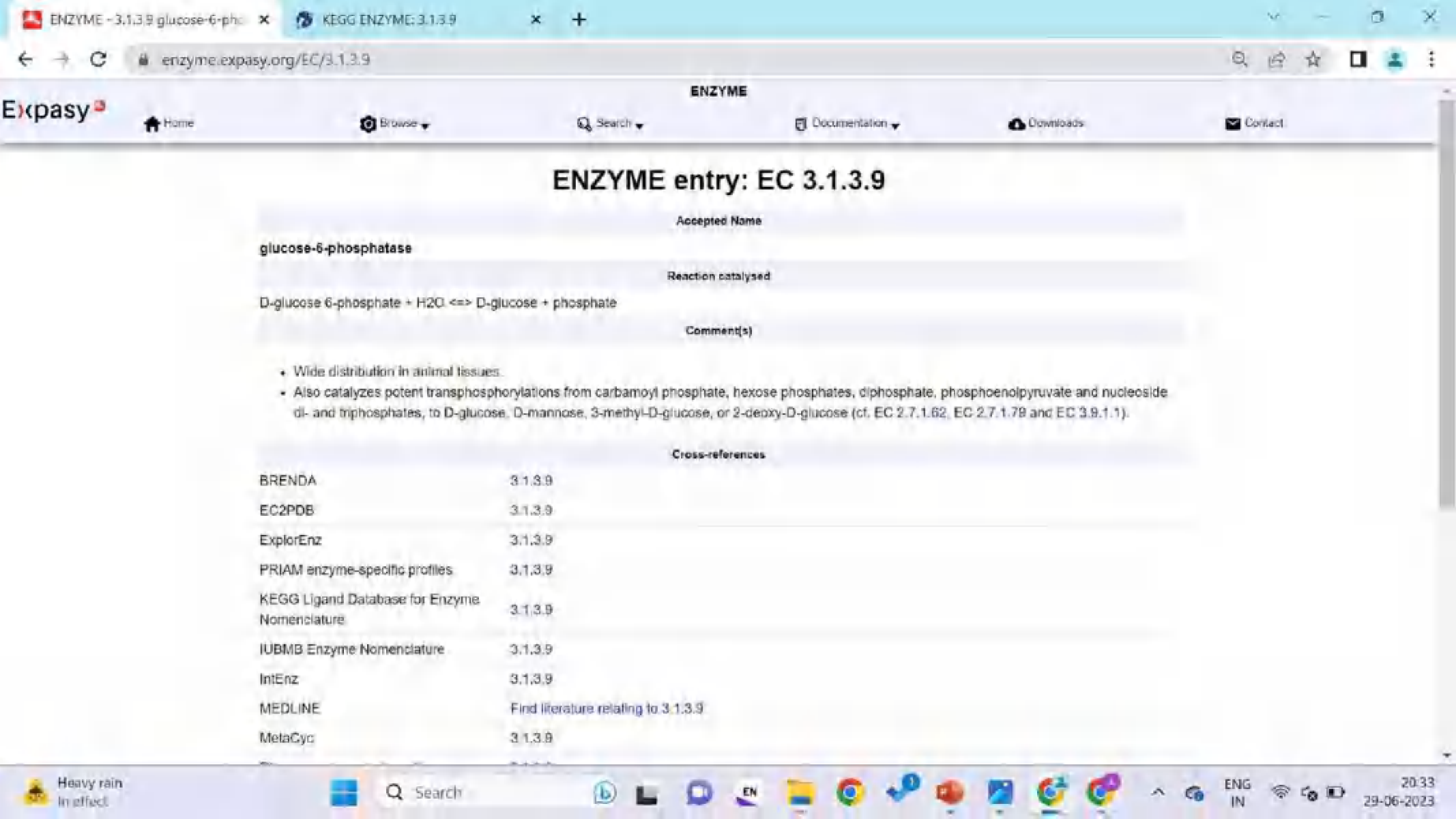


ENZYME entry: EC 3.1.3.9

Accepted Name

glucose-6-phosphatase

Reaction catalysed

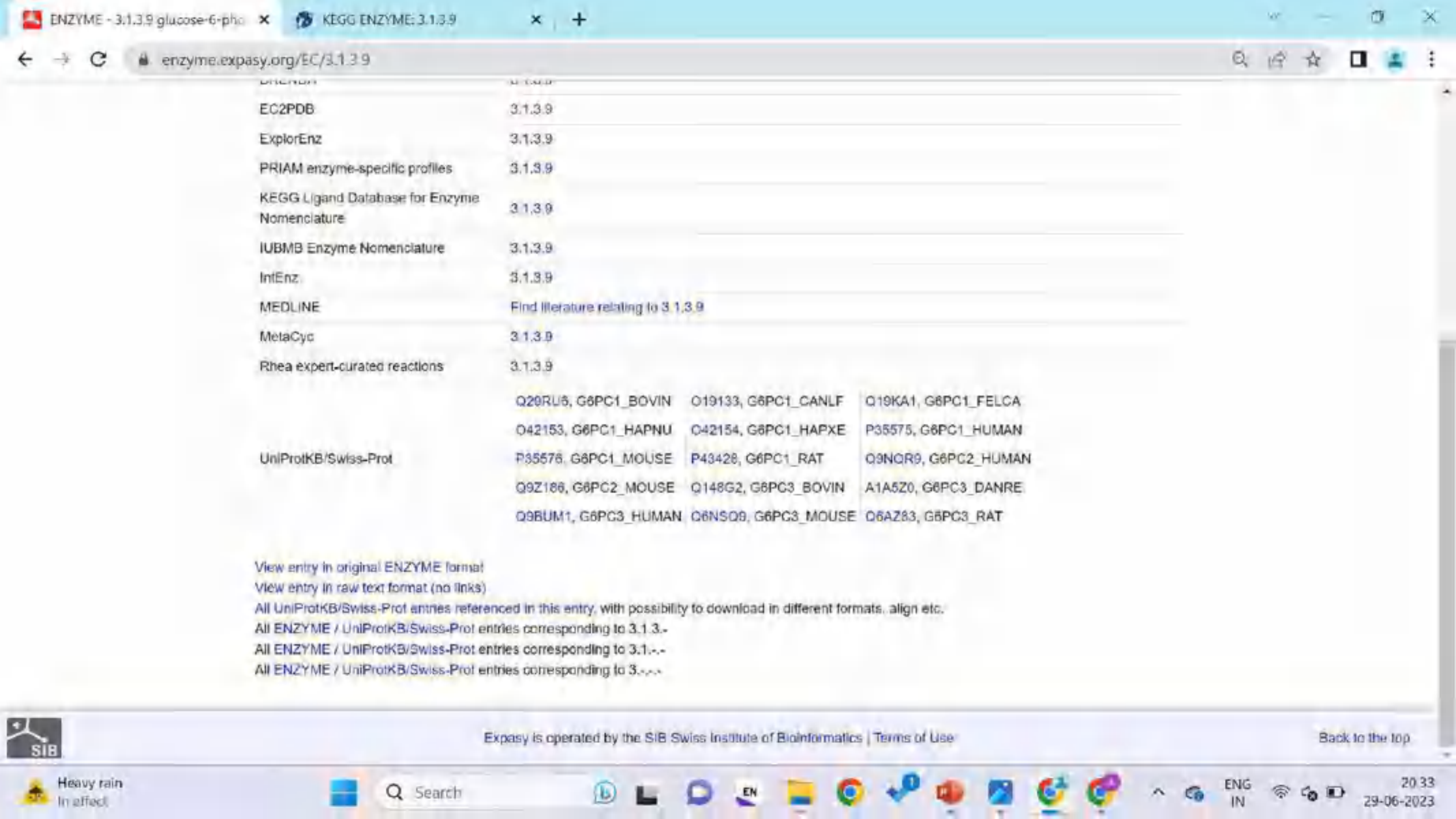
D-glucose 6-phosphate + H2O <=> D-glucose + phosphate

Comment(s)

- Wide distribution in animal tissues.
- Also catalyzes potent transphosphorylations from carbamoyl phosphate, hexose phosphates, diphosphate, phosphoenolpyruvate and nucleoside di- and triphosphates, to D-glucose, D-mannose, 3-methyl-D-glucose, or 2-deoxy-D-glucose (cf. EC 2.7.1.62, EC 2.7.1.79 and EC 3.9.1.1).

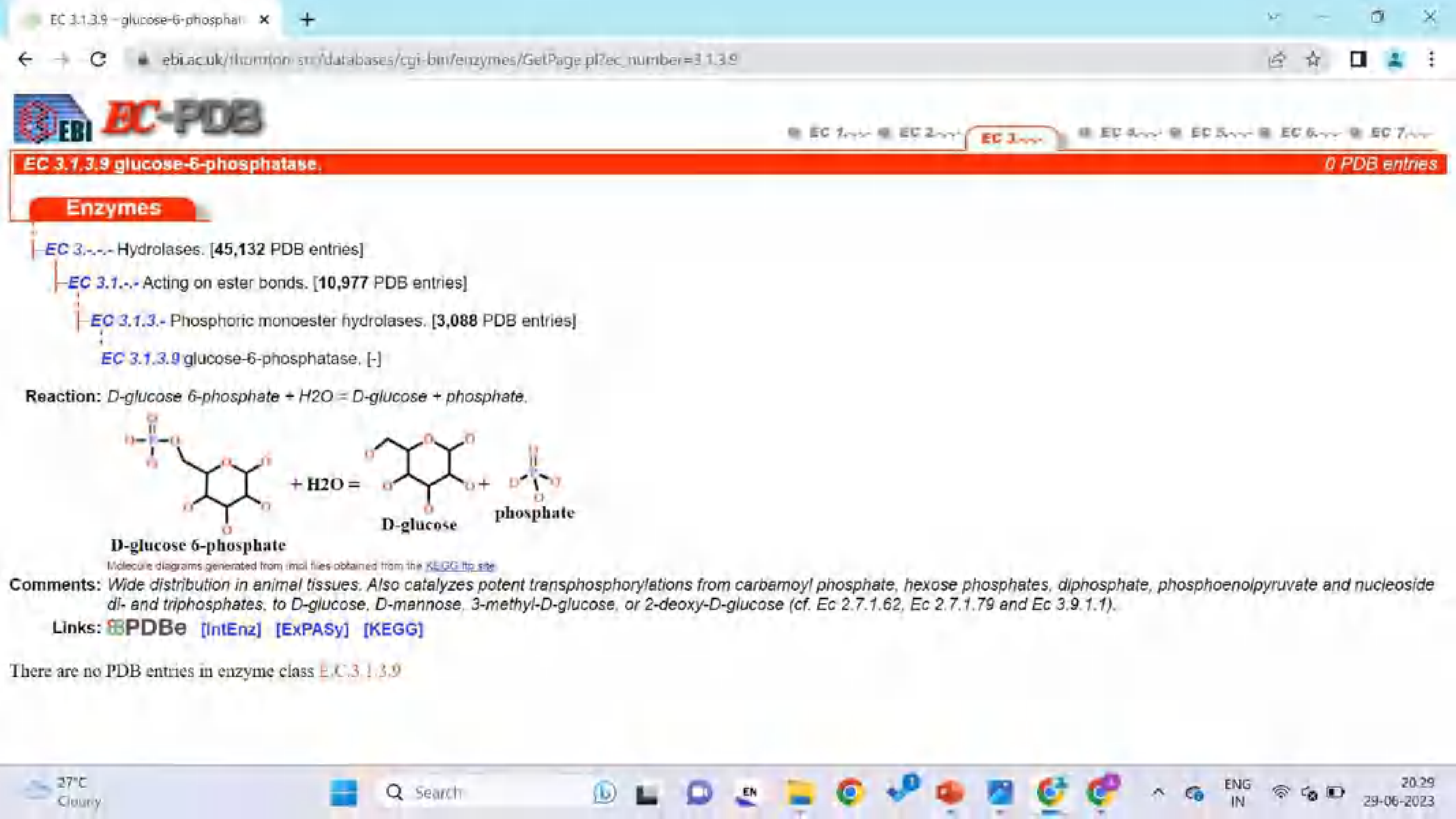
Cross-references

BRENDA	3.1.3.9
EC2PDB	3.1.3.9
ExplorEnz	3.1.3.9
PRIAM enzyme-specific profiles	3.1.3.9
KEGG Ligand Database for Enzyme Nomenclature	3.1.3.9
IUBMB Enzyme Nomenclature	3.1.3.9
IntEnz	3.1.3.9
MEDLINE	Find literature relating to 3.1.3.9
MetaCyc	3.1.3.9



EC2PDB	3.1.3.9
ExplorEnz	3.1.3.9
PRIAM enzyme-specific profiles	3.1.3.9
KEGG Ligand Database for Enzyme Nomenclature	3.1.3.9
IUBMB Enzyme Nomenclature	3.1.3.9
IntEnz	3.1.3.9
MEDLINE	Find literature relating to 3.1.3.9
MetaCyc	3.1.3.9
Rhea expert-curated reactions	3.1.3.9
UniProtKB/Swiss-Prot	Q29RU6, G6PC1_BOVIN O19133, G6PC1_CANLF Q19KA1, G6PC1_FELCA O42153, G6PC1_HAPNU O42154, G6PC1_HAPXE P35575, G6PC1_HUMAN P35576, G6PC1_MOUSE P43428, G6PC1_RAT Q9NQR9, G6PC2_HUMAN Q9Z186, G6PC2_MOUSE Q148G2, G6PC3_BOVIN A1A5Z0, G6PC3_DANRE Q9BUM1, G6PC3_HUMAN Q6NSQ9, G6PC3_MOUSE Q6AZ83, G6PC3_RAT

View entry in original ENZYME format
View entry in raw text format (no links)
All UniProtKB/Swiss-Prot entries referenced in this entry, with possibility to download in different formats, align etc.
All ENZYME / UniProtKB/Swiss-Prot entries corresponding to 3.1.3.-
All ENZYME / UniProtKB/Swiss-Prot entries corresponding to 3.1.-.-
All ENZYME / UniProtKB/Swiss-Prot entries corresponding to 3.-.-.-



EC 3.1.3.9 glucose-6-phosphatase. *0 PDB entries*

Enzymes

- EC 3.-.-.- Hydrolases. [**45,132** PDB entries]
  - EC 3.1.-.- Acting on ester bonds. [**10,977** PDB entries]
    - EC 3.1.3.- Phosphoric monoester hydrolases. [**3,088** PDB entries]
      - EC 3.1.3.9 glucose-6-phosphatase. [-]

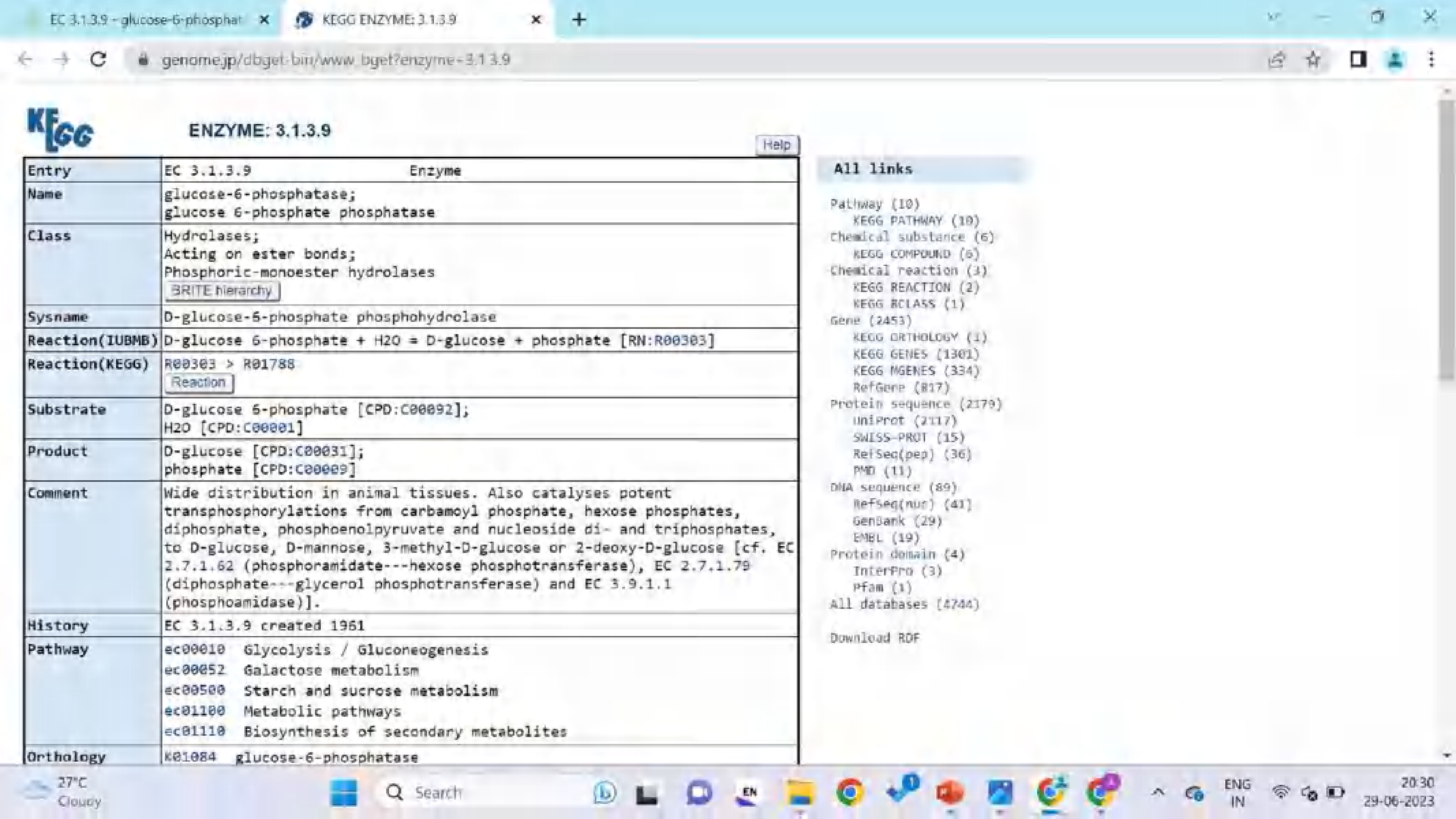
Reaction: *D-glucose 6-phosphate + H2O = D-glucose + phosphate.*

Molecule diagrams generated from mol files obtained from the KEGG ftp site

Comments: *Wide distribution in animal tissues. Also catalyzes potent transphosphorylations from carbamoyl phosphate, hexose phosphates, diphosphate, phosphoenolpyruvate and nucleoside di- and triphosphates, to D-glucose, D-mannose, 3-methyl-D-glucose, or 2-deoxy-D-glucose (cf. Ec 2.7.1.62, Ec 2.7.1.79 and Ec 3.9.1.1).*

Links: PDBe [IntEnz] [ExPASy] [KEGG]

There are no PDB entries in enzyme class E.C.3.1.3.9



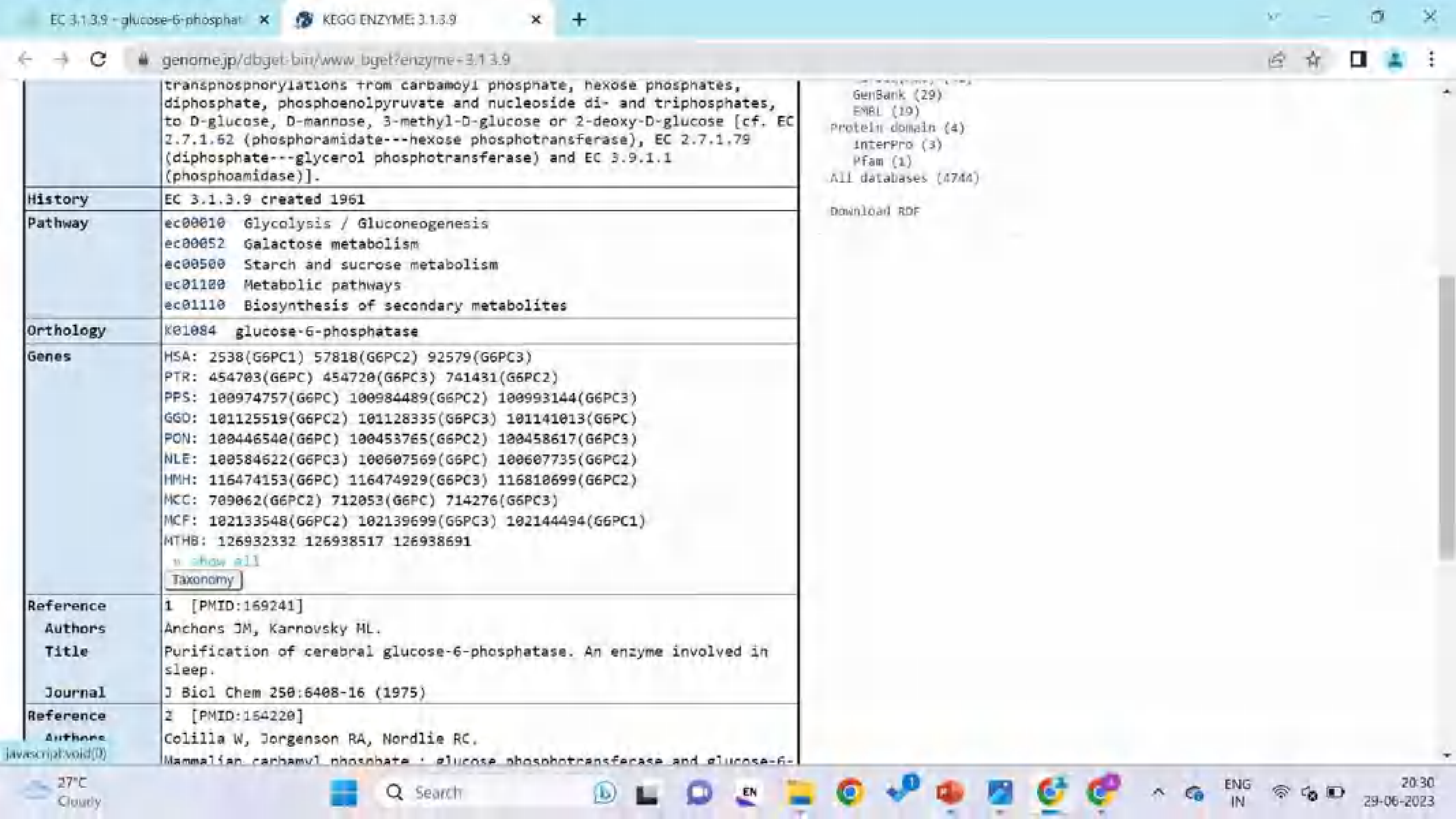
ENZYME: 3.1.3.9

Entry	EC 3.1.3.9 Enzyme
Name	glucose-6-phosphatase; glucose 6-phosphate phosphatase
Class	Hydrolases; Acting on ester bonds; Phosphoric-monoester hydrolases
Sysname	D-glucose-6-phosphate phosphohydrolase
Reaction(IUBMB)	D-glucose 6-phosphate + H2O = D-glucose + phosphate [RN:R00303]
Reaction(KEGG)	R00303 > R01788
Substrate	D-glucose 6-phosphate [CPD:C00092]; H2O [CPD:C00001]
Product	D-glucose [CPD:C00031]; phosphate [CPD:C00009]
Comment	Wide distribution in animal tissues. Also catalyses potent transphosphorylations from carbamoyl phosphate, hexose phosphates, diphosphate, phosphoenolpyruvate and nucleoside di- and triphosphates, to D-glucose, D-mannose, 3-methyl-D-glucose or 2-deoxy-D-glucose [cf. EC 2.7.1.62 (phosphoramidate---hexose phosphotransferase), EC 2.7.1.79 (diphosphate---glycerol phosphotransferase) and EC 3.9.1.1 (phosphoamidase)].
History	EC 3.1.3.9 created 1961
Pathway	ec00010 Glycolysis / Gluconeogenesis ec00052 Galactose metabolism ec00500 Starch and sucrose metabolism ec01100 Metabolic pathways ec01110 Biosynthesis of secondary metabolites
Orthology	K01084 glucose-6-phosphatase

All links

- Pathway (10)
  - KEGG PATHWAY (10)
- Chemical substance (6)
  - KEGG COMPOUND (6)
- Chemical reaction (3)
  - KEGG REACTION (2)
  - KEGG RCLASS (1)
- Gene (2453)
  - KEGG ORTHOLOGY (1)
  - KEGG GENES (1301)
  - KEGG MGENES (334)
  - RefGene (817)
- Protein sequence (2179)
  - UniProt (2117)
  - SWISS-PROT (15)
  - RefSeq(pep) (36)
  - PMD (11)
- DNA sequence (89)
  - RefSeq(nuc) (41)
  - GenBank (29)
  - EMBL (19)
- Protein domain (4)
  - InterPro (3)
  - Pfam (1)
- All databases (4744)

Download RDF



	transphosphorylations from carbamoyl phosphate, hexose phosphates, diphosphate, phosphoenolpyruvate and nucleoside di- and triphosphates, to D-glucose, D-mannose, 3-methyl-D-glucose or 2-deoxy-D-glucose [cf. EC 2.7.1.62 (phosphoramidate---hexose phosphotransferase), EC 2.7.1.79 (diphosphate---glycerol phosphotransferase) and EC 3.9.1.1 (phosphoamidase)].
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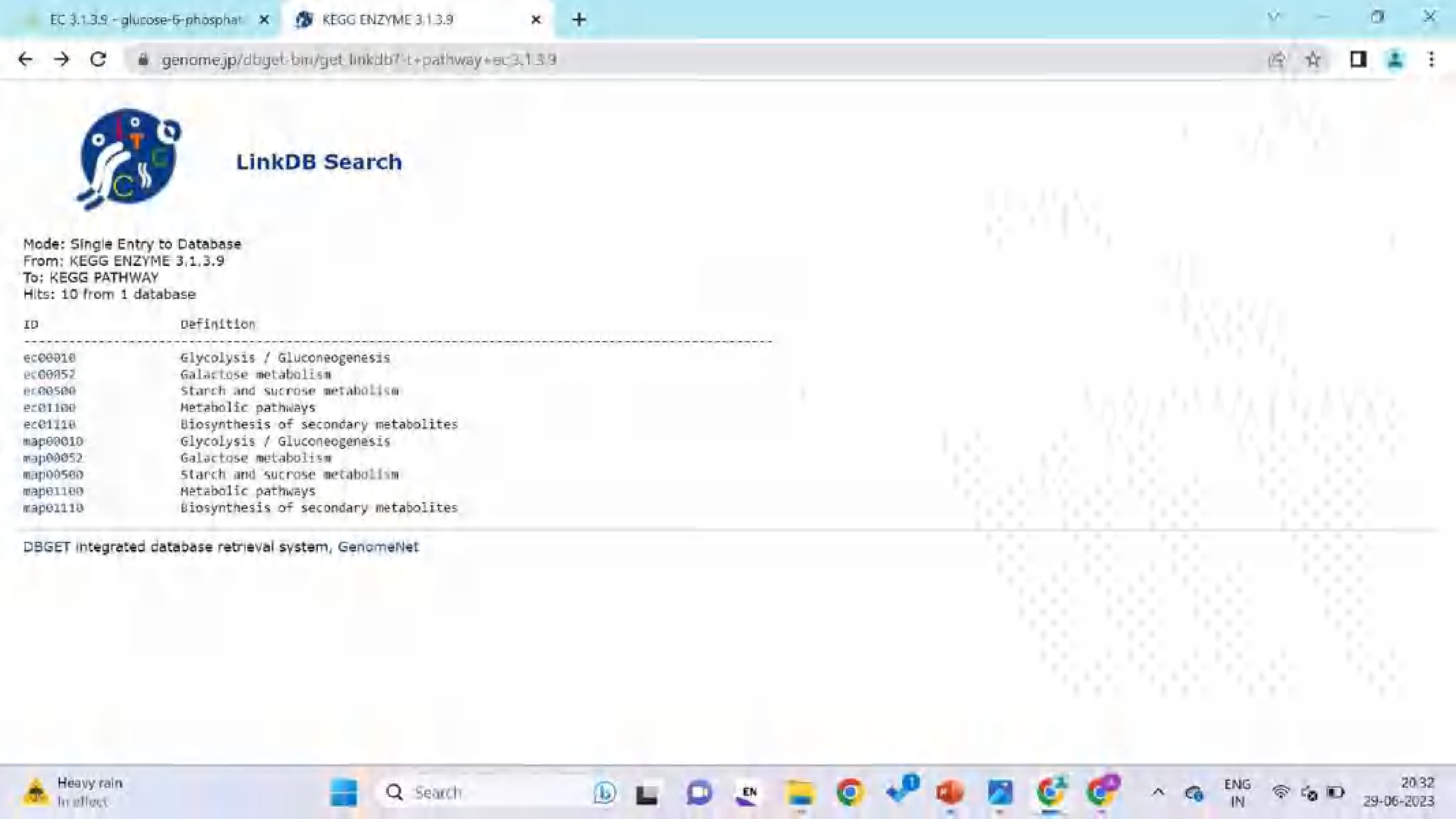
Pathway	ec00010 Glycolysis / Gluconeogenesis ec00052 Galactose metabolism ec00500 Starch and sucrose metabolism ec01100 Metabolic pathways ec01110 Biosynthesis of secondary metabolites
Orthology	K01084 glucose-6-phosphatase
Genes	HSA: 2538(G6PC1) 57818(G6PC2) 92579(G6PC3) PTR: 454703(G6PC) 454720(G6PC3) 741431(G6PC2) PPS: 100974757(G6PC) 100984489(G6PC2) 100993144(G6PC3) GGO: 101125519(G6PC2) 101128335(G6PC3) 101141013(G6PC) PON: 100446540(G6PC) 100453765(G6PC2) 100458617(G6PC3) NLE: 100584622(G6PC3) 100607569(G6PC) 100607735(G6PC2) HMH: 116474153(G6PC) 116474929(G6PC3) 116810699(G6PC2) MCC: 709062(G6PC2) 712053(G6PC) 714276(G6PC3) MCF: 102133548(G6PC2) 102139699(G6PC3) 102144494(G6PC1) MTHB: 126932332 126938517 126938691 » show all Taxonomy
Reference	1 [PMID:169241]
Authors	Anchors JM, Karnovsky ML.
Title	Purification of cerebral glucose-6-phosphatase. An enzyme involved in sleep.
Journal	J Biol Chem 250:6408-16 (1975)
Reference	2 [PMID:164220]
Authors	Colilla W, Jorgenson RA, Nordlie RC.
	Mammalian carbamyl phosphate : glucose phosphotransferase and glucose-6-

GenBank (29)
EMBL (19)
Protein domain (4)
InterPro (3)
Pfam (1)
All databases (4744)

Download RDF



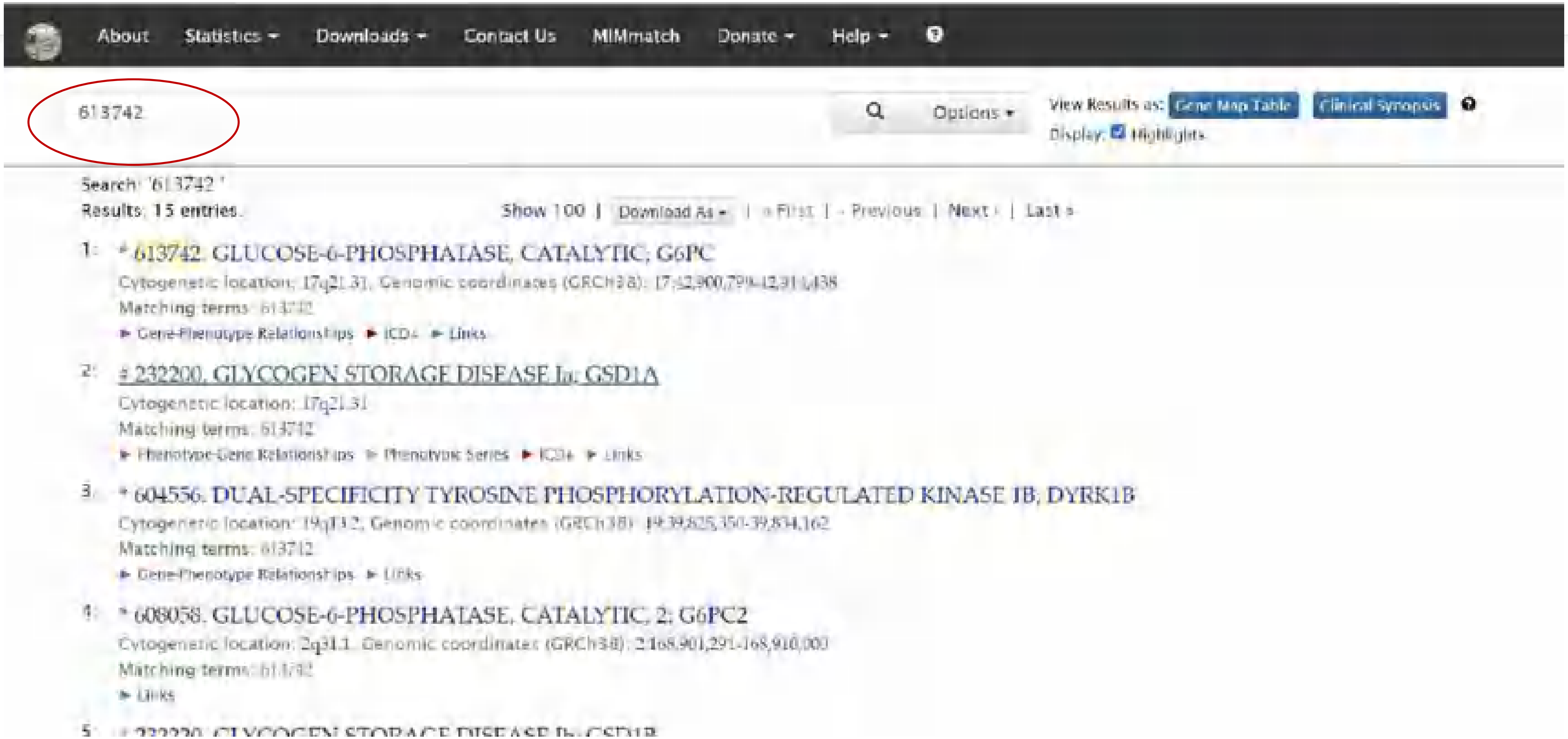
LinkDB Search

Mode: Single Entry to Database
From: KEGG ENZYME 3.1.3.9
To: KEGG PATHWAY
Hits: 10 from 1 database

```
ID                    Definition
----------------------------------------------------------------------------------------------------
ec00010               Glycolysis / Gluconeogenesis
ec00052               Galactose metabolism
ec00500               Starch and sucrose metabolism
ec01100               Metabolic pathways
ec01110               Biosynthesis of secondary metabolites
map00010              Glycolysis / Gluconeogenesis
map00052              Galactose metabolism
map00500              Starch and sucrose metabolism
map01100              Metabolic pathways
map01110              Biosynthesis of secondary metabolites
```

DBGET integrated database retrieval system, GenomeNet

Go to OMIM webpage



613742

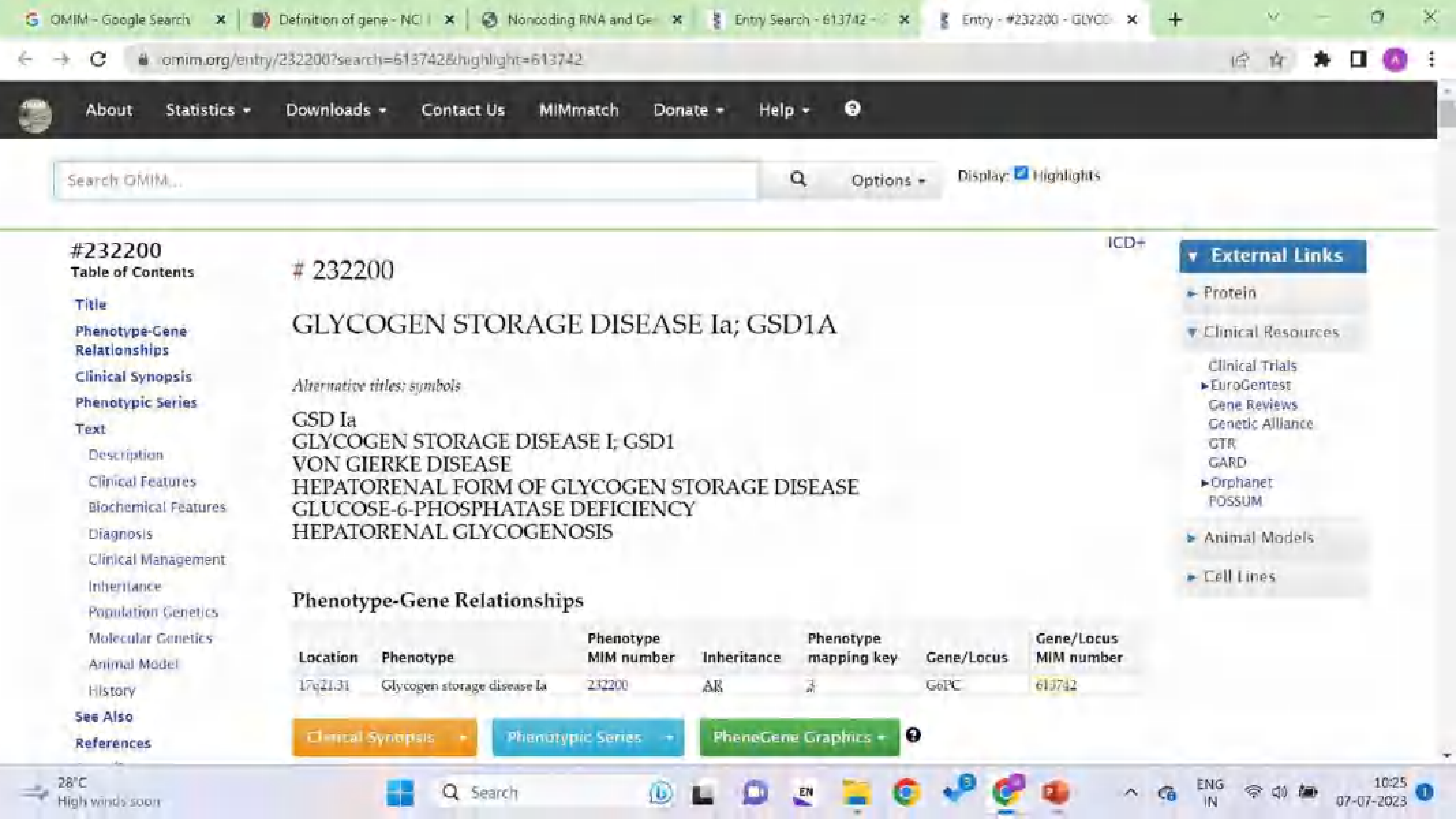
Search: '613742'
Results: 15 entries.

1: * 613742. GLUCOSE-6-PHOSPHATASE, CATALYTIC; G6PC
Cytogenetic location: 17q21.31, Genomic coordinates (GRCh38): 17:42,900,797-42,914,438
Matching terms: 613742
► Gene-Phenotype Relationships ► ICD+ ► Links

2: # 232200. GLYCOGEN STORAGE DISEASE Ia; GSD1A
Cytogenetic location: 17q21.31
Matching terms: 613742
► Phenotype-Gene Relationships ► Phenotypic Series ► ICD+ ► Links

3: * 604556. DUAL-SPECIFICITY TYROSINE PHOSPHORYLATION-REGULATED KINASE 1B; DYRK1B
Cytogenetic location: 19q13.2, Genomic coordinates (GRCh38): 19:39,825,350-39,834,162
Matching terms: 613742
► Gene-Phenotype Relationships ► Links

4: * 608058. GLUCOSE-6-PHOSPHATASE, CATALYTIC, 2; G6PC2
Cytogenetic location: 2q31.1, Genomic coordinates (GRCh38): 2:168,901,291-168,910,000
Matching terms: 613742
► Links



232200

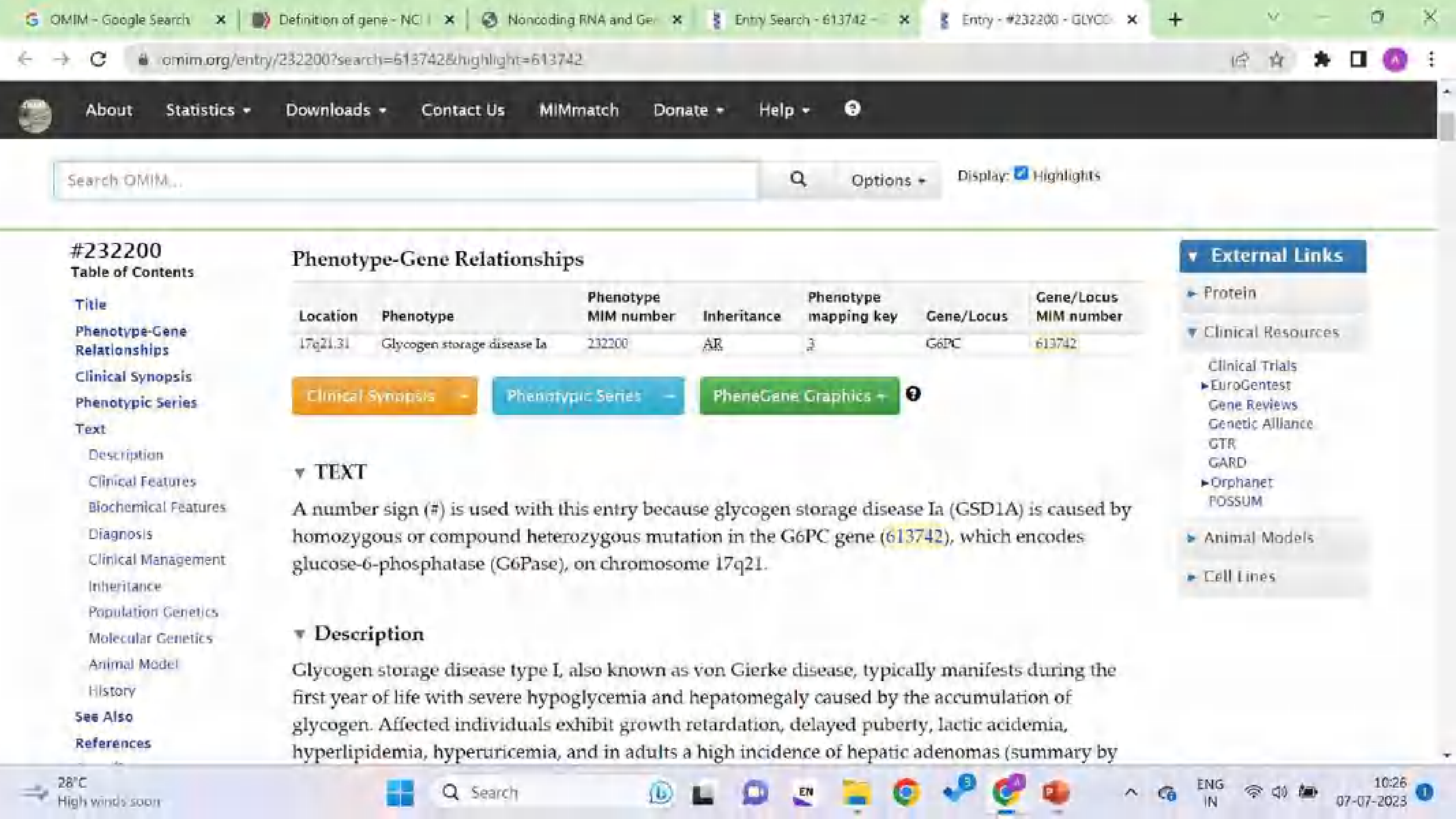
GLYCOGEN STORAGE DISEASE Ia; GSD1A

Alternative titles; symbols

GSD Ia
GLYCOGEN STORAGE DISEASE I; GSD1
VON GIERKE DISEASE
HEPATORENAL FORM OF GLYCOGEN STORAGE DISEASE
GLUCOSE-6-PHOSPHATASE DEFICIENCY
HEPATORENAL GLYCOGENOSIS

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
17q21.31	Glycogen storage disease Ia	232200	AR	3	G6PC	613742



#232200

Table of Contents

- Title
- Phenotype-Gene Relationships
- Clinical Synopsis
- Phenotypic Series
- Text
  - Description
  - Clinical Features
  - Biochemical Features
  - Diagnosis
  - Clinical Management
  - Inheritance
  - Population Genetics
  - Molecular Genetics
  - Animal Model
  - History
- See Also
- References

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
17q21.31	Glycogen storage disease Ia	232200	AR	3	G6PC	613742

▼ TEXT

A number sign (#) is used with this entry because glycogen storage disease Ia (GSD1A) is caused by homozygous or compound heterozygous mutation in the G6PC gene (613742), which encodes glucose-6-phosphatase (G6Pase), on chromosome 17q21.

▼ Description

Glycogen storage disease type I, also known as von Gierke disease, typically manifests during the first year of life with severe hypoglycemia and hepatomegaly caused by the accumulation of glycogen. Affected individuals exhibit growth retardation, delayed puberty, lactic acidemia, hyperlipidemia, hyperuricemia, and in adults a high incidence of hepatic adenomas (summary by

▼ External Links

- ► Protein
- ▼ Clinical Resources
  - Clinical Trials
  - ►EuroGentest
  - Gene Reviews
  - Genetic Alliance
  - GTR
  - GARD
  - ►Orphanet
  - POSSUM
- ► Animal Models
- ► Cell Lines