



# Introduction to Bioinformatics

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Dr. Archana Upadhyia

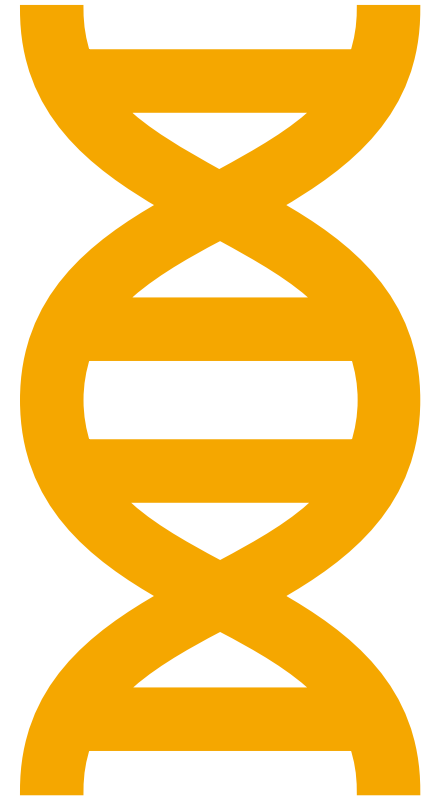
Associate Professor

Humera Khan College of Pharmacy

# What is a gene?

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- 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

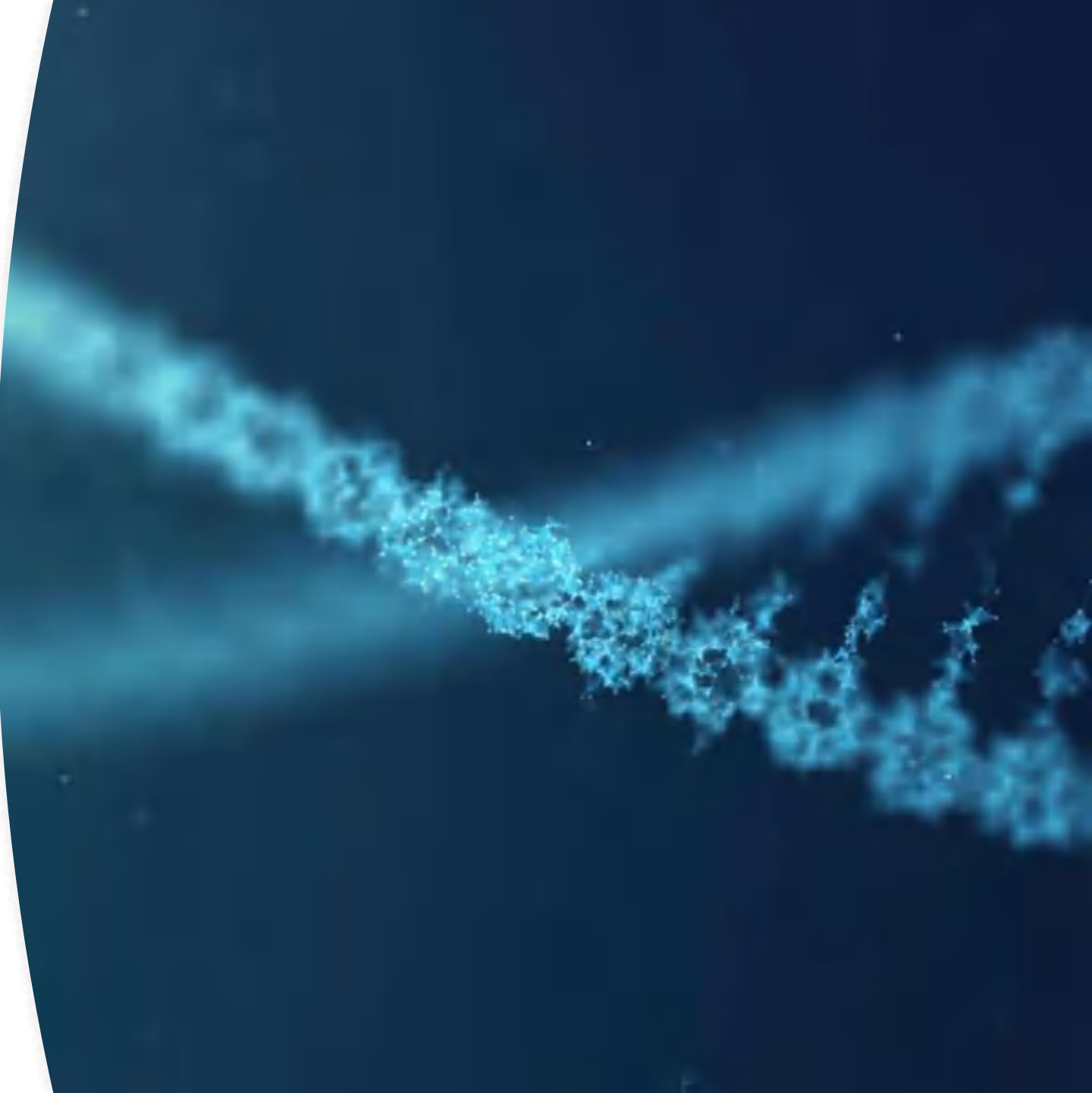
- Ubiquitin proteasome pathway
- Cellular proteases



# What is Bioinformatics?

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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





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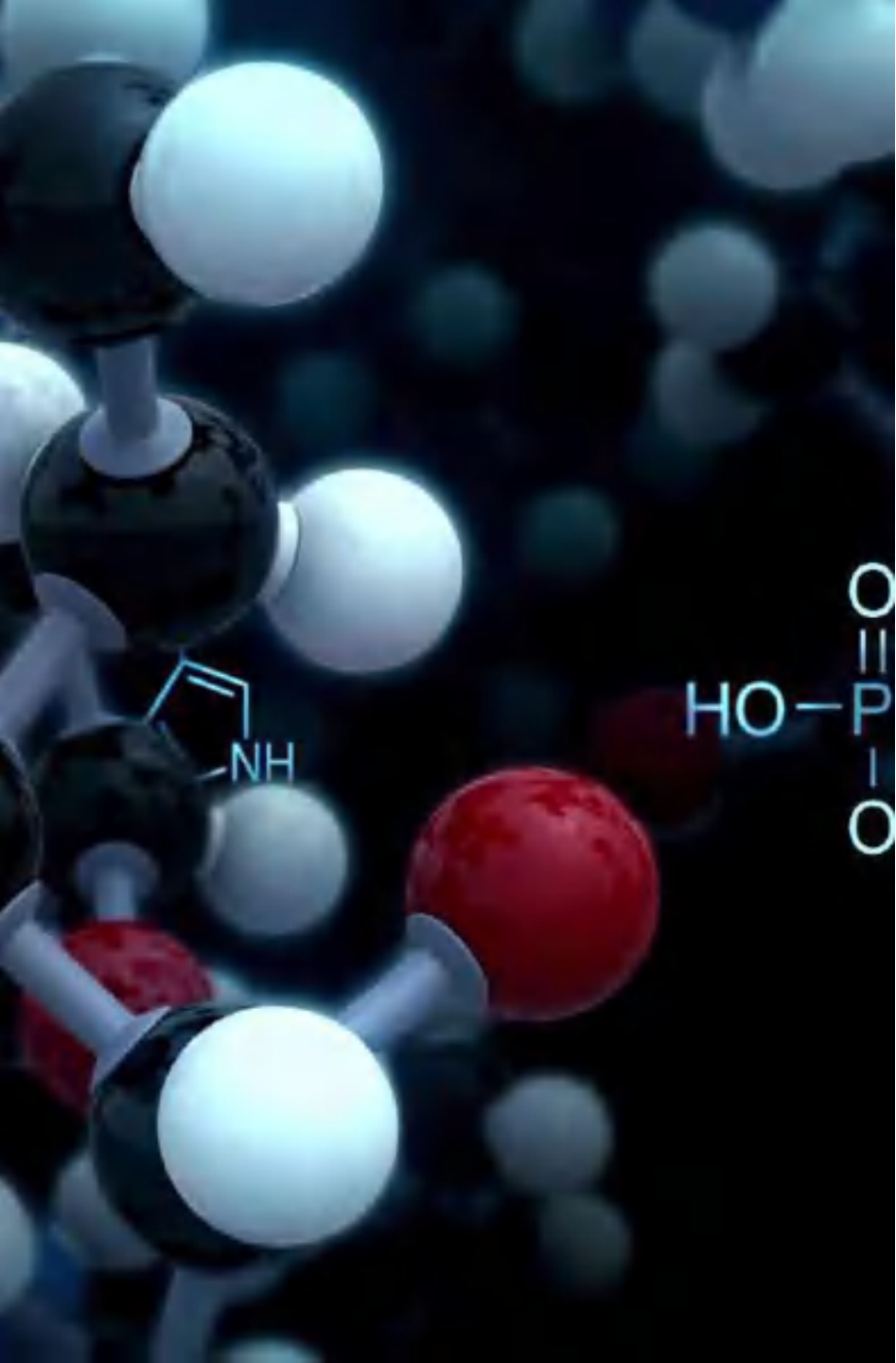
# Popular Databases

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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 Phosphatase I)
  - 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 I (G6PCI)

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





All Databases ▾

Search

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Genome

SNP

Gene

Protein

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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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Gene

Glucose 6 Phosphatase



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## Gene sources

Genomic

Plasmids

## Categories

Alternatively spliced

Annotated genes

Non-coding

Protein-coding

Pseudogene

## Sequence content

CCDS

Ensembl

RefSeq

RefSeqGene

## Status

✓ Current

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Tabular 20 per page Sort by Relevance

Send to:

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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
<input type="checkbox"/> <a href="#">G6PC1</a> ID: 2538	glucose-6-phosphatase catalytic subunit 1 [ <i>Homo sapiens</i> (human)]	Chromosome 17, NC_000017.11 (42900799..42914436)	G6PC, G6PT, G6Pase, GSD1, GSD1a	613742
<input type="checkbox"/> <a href="#">G6pc1</a> ID: 14577	glucose-6-phosphatase catalytic subunit 1 [ <i>Mus musculus</i> (house mouse)]	Chromosome 11, NC_000077.7 (101258542..101268729)	G6Pase, G6pc, G6pt, Glc-6-Pase	
<input type="checkbox"/> <a href="#">G6PC2</a> ID: 67619	glucose-6-phosphatase catalytic subunit 2 [ <i>Homo sapiens</i> (human)]	Chromosome 2, NC_000002.12 (168901291..168910000)	IGRP	608058
<input type="checkbox"/> <a href="#">G6PC3</a> ID: 67579	glucose-6-phosphatase catalytic subunit 3 [ <i>Homo sapiens</i> (human)]	Chromosome 17, NC_000017.11 (44070673..44076344)	SCN4, UGRP	611045
<input type="checkbox"/> <a href="#">G6pc1</a> ID: 25634	glucose-6-phosphatase catalytic subunit 1 [ <i>Rattus norvegicus</i> (Norway rat)]	Chromosome 10, NC_051345.1 (86307400..86318766)	G6Pase, G6pc, Psm63	
<input type="checkbox"/> <a href="#">G6pc2</a> ID: 14578	glucose-6-phosphatase catalytic, 2 [ <i>Mus musculus</i> (house mouse)]	Chromosome 2, NC_000068.8 (69041417..69058337)	G6pc-rs, IGRP	
<input type="checkbox"/> <a href="#">MUSG6</a>	Mus musculus G6Pase			

Filters: [Manage Filters](#)

## 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: 

## Search details

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

Search

[See more...](#)

# Information retrieved

- Catalytic 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)
  - Catalytic 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, GSDI, GSDIa
- 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	<a href="#">mRNA</a>
NR	<a href="#">ncRNA</a> (non coding RNA)
NP	<a href="#">Protein</a>
XM	predicted <a href="#">mRNA</a> model
XR	predicted <a href="#">ncRNA</a> model
XP	predicted <a href="#">Protein</a> model (eukaryotic sequences)
WP	predicted <a href="#">Protein</a> model (prokaryotic sequences)



**G6PC1** glucose-6-phosphatase catalytic subunit 1 [ *Homo sapiens* (human) ][Download Datasets](#)**Table of contents**

Summary

Genomic context

Genomic regions, transcripts, and products

Expression

Bibliography

Phenotypes

Variation

Pathways from PubChem

Interactions

General gene information

Markers, Clone Names, Homology, Gene Ontology

General protein information

NCBI Reference Sequences (RefSeq)

Related sequences

Additional links

Locus-specific Databases

**Genome Browsers**

Genome Data Viewer

Variation Viewer (GRCh37.p13)

**Summary**

<b>Official Symbol</b>	G6PC1 <small>provided by HGNC</small>
<b>Official Full Name</b>	glucose-6-phosphatase catalytic subunit 1 <small>provided by HGNC</small>
<b>Primary source</b>	<a href="#">HGNC:HGNC:4056</a>
<b>See related</b>	<a href="#">Ensembl:ENSG00000131482</a> <a href="#">MIM:613742</a> ; <a href="#">AllianceGenome:HGNC:4056</a>
<b>Gene type</b>	protein coding
<b>RefSeq status</b>	REVIEWED
<b>Organism</b>	<i>Homo sapiens</i>
<b>Lineage</b>	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorhini; Catarrhini; Hominidae; Homo
<b>Also known as</b>	G6PC; G6PT; GSD1; GSD1a; G6Pase
<b>Summary</b>	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]
<b>Expression</b>	Biased expression in liver (RPKM 48.0), kidney (RPKM 22.9) and 2 other tissues <a href="#">See more</a>
<b>Orthologs</b>	<a href="#">mouse</a> <a href="#">all</a>

**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

NEW Try the new Gene table  
Try the new Transcript table

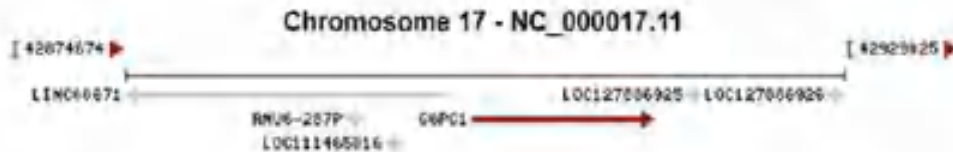
Genomic context

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 (41052818..41066455)

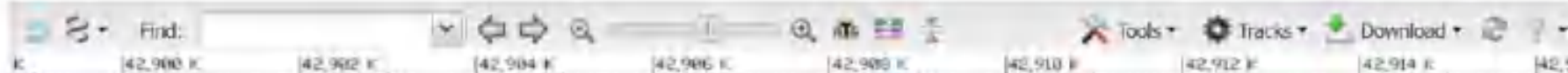


Genomic regions, transcripts, and products

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

The screenshot shows the NCBI Genome Data Viewer interface for gene G6PC1. The browser address bar shows the URL: [ncbi.nlm.nih.gov/genome/gdv/browser/gene/?id=2538](https://ncbi.nlm.nih.gov/genome/gdv/browser/gene/?id=2538). The main track view displays the gene structure for G6PC1, with the 'Gene' dropdown set to 'G6PC1' and the 'Transcript' dropdown set to 'NM\_000151.4'. A red box highlights these dropdown menus. The left sidebar shows the 'Ideogram View' with chromosome 17 highlighted by a red box. The main track view includes tracks for 'Genes, NCBI RefSeq Annotation GCF\_000001405.40-RS\_2023\_03', 'NCBI RefSeq Annotation GCF\_000001405.40-RS\_2023\_03', 'Genes, Ensembl release 109', and 'ClinVar Variations, dbSNP 6156 V2'. The ClinVar track shows several SNPs with their positions and alleles.

SNP ID	Position (approx.)	Alleles
rs60356479	42,900,000	CCC/CC
rs1057516674	42,900,000	GT/-
rs104694560	42,900,000	TG/G/T
rs1057517605	42,900,000	A/C/G
rs764920767	42,900,000	G/R
rs104694566	42,900,000	T/C
rs567776757	42,900,000	A/G
rs1001175	42,900,000	C/T
rs1001176	42,900,000	G/R
rs104694567	42,900,000	A/R
rs116797100	42,900,000	C/G/T
rs2592595	42,900,000	A/R
rs60356480	42,900,000	TAT/TAT/ATA
rs104694568	42,900,000	G/R
rs104694569	42,900,000	G/R
rs104694570	42,900,000	G/R
rs104694571	42,900,000	T/C/G
rs104694572	42,900,000	G/R/C
rs60356481	42,900,000	CTTCT/CT
rs60356482	42,900,000	G/R/T
rs60356483	42,900,000	G/R/T
rs60356484	42,900,000	G/R/T
rs60356485	42,900,000	C/T
rs60356486	42,900,000	G/T
rs104694563	42,900,000	C/T
rs760226142	42,900,000	C/R/T
rs367727229	42,900,000	G/R/T
rs387966805	42,900,000	T/R/C



# Gene next to G6PCI-RNY4P2

The screenshot displays the NCBI Gene browser for the G6PCI gene (Gene ID: 2538). The browser is set to the GRCh38.p14 assembly. The gene structure shows exons and introns, with the RNY4P2 transcript highlighted in green. The sequence viewer shows the DNA sequence and the corresponding protein sequence. The ideogram view shows the gene's location on chromosome 10. The tracks section includes various annotations such as biological regions, NCBI RefSeq Annotation, ClinVar variants, RNA-seq exon coverage, and RNA-seq intron-spanning reads.

Examples ▶

Assemblies

Gene: RNY4P2 Exons: GENE exons

Assembly: GCF\_00001405.10 (GRCh38.p14)

Locations for Gene G6PCI

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

Ideogram View

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

1 2 3 4 5 6 7 8 9 10 11 12 13

14 15 16 17 18 19 20 21 22 X Y MT

Tracks and User Data

biological regions, aggregate, NCBI RefSeq Annotation GCF\_00001405.10 GRCh38.p14

Warning: No track data found in this range

Genes, Ensembl release 105

Warning: No track data found in this range

ClinVar Variations, dbSNP b156 v2

Warning: No track data found in this range

Love RefSeq, dbSNP b156 v2

rs1591997063 T/A rs2056196733 G/A 606666/606666 rs205619655 T/A/T rs236815440 A/T rs139241388 G/A rs2056191167 T/C rs2056191168 T/C rs116772031 C/A rs1297997807 A/B rs252322815 T/T rs166155472 A/B rs117722917 G/C rs128021944 C/T rs97081128 C/T rs758943162 T/C rs131627546 A/G/T rs952112462 G/T rs1785862517 C/G rs2056194958 T/A/C rs2056191843 A/B rs2056191141 C/T rs190481798 C/T rs181126224 T/A/C rs2056190838 C/T rs2056190839 A/G rs1823550793 C/T rs69853817 C/T rs111769944 G/A rs2056190801 T/C rs1232666722 G/A/C rs1096190951 A/B rs142167968 T/C rs1443120985 G/B rs879877179 A/B rs37196716 C/T rs2056190878 T/C rs2056190785 G/C rs1700000244 A/B rs1258645385 C/A/T rs802964752 C/G/T rs979755172 T/C rs198228479 T/C rs2056190791 A/B rs1394926974 A/B rs205619021 C/B rs1597997976 C/A/W

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 Annotation Release 110

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

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110

# Gene previous to G6PCI-LINC00671

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

Examples ▾

Assemblies

Assembly Switch organism

GCF\_000001405.40 (GRCh38.p14) Select an assembly to change view

Locations for Gene G6PCI

Sequence	Location
NC_000017.11	42,600,707 - 42,914,433

Ideogram View

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

1 2 3 4 5 6 7 8 9 10 11 12 13  
14 15 16 17 18 19 20 21 22 X Y MT

Region: LINC00671  
Transcript: NR\_027254.1

NC\_000017.11  
Genes, MAME Project (release v1.2)  
NCBI RefSeq Annotation GCF\_000001405.40-BB\_2023\_03  
ENSG00000214822  
ENSG00000214821

Biological regions, aggregate, NCBI RefSeq Annotation GCF\_0000...  
enhancer  
transcription\_start\_site

Genes, Ensembl release 109  
ENSG00000214822  
ENSG00000214821

Cited Variations, dbSNP b156 v2



# Details of G6PC1

The screenshot displays the Ensembl genome browser interface for the G6PC1 gene. The browser shows the gene structure with exons and introns, and a detailed information panel for the G6PC1 gene is highlighted with a red box.

**Gene Information:**

- 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

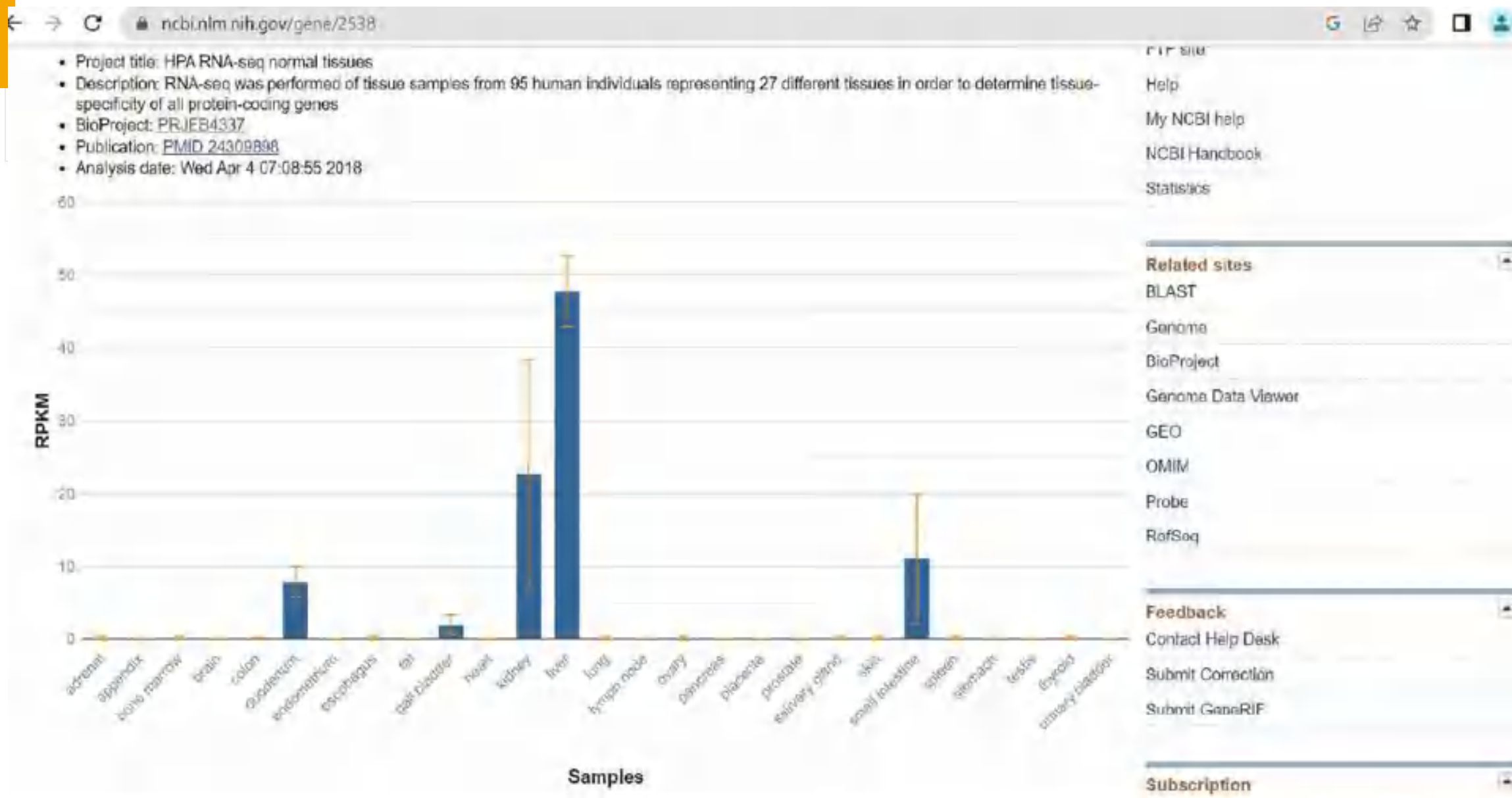
**BLAST & Records:**

- 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)

**Cited Variations, dbSNP b156 v2:**

rs80356479   C/C/C	rs1801175   C/T	rs80356480   T/T/T/T/T/T	rs60356486   C/T/C/T/C/T
rs1057516674   G/T/-	rs1801176   G/A	rs104894569   G/A	rs104894571   T/C/G
rs104894565   A/G/T	rs184894567   G/A	rs161628   G/A	rs80356494   G/A/T
rs1857517008   A/C/G	rs116797180   C/G/T	rs80356482   G/A/C	rs80356485   C/T
rs764928767   G/R	rs2593595   A/G	rs80356483   G/T	rs184894583   C/T
rs104894566   T/C		rs780226142   C/R/T	rs367727229   G/R/T
rs87776757   A/G		rs387906585   T/A/C	

# Go Back to webpage: <https://www.ncbi.nlm.nih.gov/gene/2538> Tissue Specific Expression



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

ncbi.nlm.nih.gov/gene/2538

Samples

### 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 30690478
3. [Glucose-6-phosphatase- \$\alpha\$  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 28502058
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](#)

### Subscription

RelSeq

### Recent activity

[Turn Off](#) [Clear](#)

- [G6PC1 glucose-6-phosphatase catalytic subunit 1 \(Homo sapiens\)](#) [Clear](#)
- [Glucose 6 Phosphatase \(109\)](#) [Clear](#)
- [Glucose 6 Phosphatase AND \(alive\[prop\]\) \(7276\)](#) [Clear](#)
- [Glucose 6-Phosphatase AND \(alive\[prop\]\) \(7276\)](#) [Clear](#)
- [Human Glucose 6 Phosphatase 1 AND \(alive\[prop\]\) \(1978\)](#) [Clear](#)

[See more...](#)

# 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)	<a href="#">AC016889</a> , <a href="#">AW614228</a> , <a href="#">U05120</a>
Consensus CDS	<a href="#">CCDS11446.1</a>
UniProtKB/Swiss-Prot	<a href="#">A1L4G0</a> , <a href="#">B4E1C3</a> , <a href="#">K7EL8</a> , <a href="#">P35575</a>
Related	<a href="#">ENSP00000253801.1</a> , <a href="#">ENST00000253801.7</a>

Conserved Domains (1) [summary](#)

[cd03381](#) PAP2\_glucose\_6\_phosphatase; PAP2\_like proteins, glucose-6-phosphatase subfamily.  
Location:42 → 280: 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)	<a href="#">AC016889</a> , <a href="#">AK303771</a> , <a href="#">AW614228</a> , <a href="#">BC130478</a>
Consensus CDS	<a href="#">CCDS3992.1.1</a>
UniProtKB/Swiss-Prot	<a href="#">P35575</a>
Related	<a href="#">ENSP00000465958.1</a> , <a href="#">ENST00000592383.5</a>

Conserved Domains (1) [summary](#)

[cd00474](#) 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, Phosphatidyglycerophosphatase 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

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB

## P35575 · G6PC1\_HUMAN

Protein<sup>1</sup> | Glucose-6-phosphatase catalytic subunit 1  
Gene<sup>1</sup> | G6PC1  
Status<sup>1</sup> | UniProtKB reviewed (Swiss-Prot)  
Organism<sup>1</sup> | Homo sapiens (Human)

Amino acids | 357 (go to sequence)

Protein existence<sup>1</sup> | Evidence at protein level

Annotation score<sup>1</sup> |

Entry Variant viewer Feature viewer Publications External links History

BLAST Align Download Add Add a publication Entry feedback

### Function<sup>1</sup>

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. 5 Publications

### Catalytic activity<sup>1</sup>

D-glucose 6-phosphate + H<sub>2</sub>O = D-glucose + phosphate 5 Publications  
EC:3.1.3.9 (UniProtKB | ENZYME | Rhea )  
Source: Rhea 16689

Hide Rhea reaction

D-glucose 6-phosphate      H<sub>2</sub>O      D-glucose      phosphate

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

## Amino acids in G6PCI: 357

### Function of G6PCI:

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<sub>2</sub>O = D-glucose + phosphate (5 Publications)

EC:3.1.3.9 (Enzyme)



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

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB Advanced | List Search Help

Entry Variant viewer Feature viewer Publications External links History

### GO annotations<sup>1</sup>

Slimming set:  
generic

all annotations  
all molecular function  
virus receptor activity  
dna binding  
rna binding  
cytoskeletal motor activity  
catalytic activity  
glucose activity  
structural activity  
transporter activity  
cytoskeletal protein activity  
lipid binding  
cyclic GMP activity  
structural protein binding  
transmembrane activity  
hydroxylase activity  
ligase activity  
transferase activity  
proteinase activity  
carboxylase activity  
nitrogenase activity  
protein binding  
transmembrane receptor activity  
nutrient transporter activity  
receptor ligand activity  
molecular transporter activity  
toxin activity  
collagen activity  
molecular mediator activity  
virus coreceptor activity  
catalytic activity  
catalytic activity  
molecular activity  
transmembrane activity  
gene-activated transcription factor activity  
small molecule sensor activity  
molecular sequestering activity  
other molecular function  
all biological process

Cell color: indicative of number of GO terms

ASPECT	TERM	Source	Publications
Cellular Component	<a href="#">endoplasmic reticulum membrane</a>	Source: UniProtKB	1 Publication
Cellular Component	<a href="#">membrane</a>	Source: UniProtKB	1 Publication
Molecular Function	<a href="#">glucose-6-phosphatase activity</a>	Source: UniProtKB	2 Publications
Molecular Function	<a href="#">phosphate ion binding</a>	Source: UniProtKB	1 Publication
Molecular Function	<a href="#">phosphotransferase activity, alcohol group as acceptor</a>	Source: Ensembl	

### Help

#### Gene Ontology (GO)

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

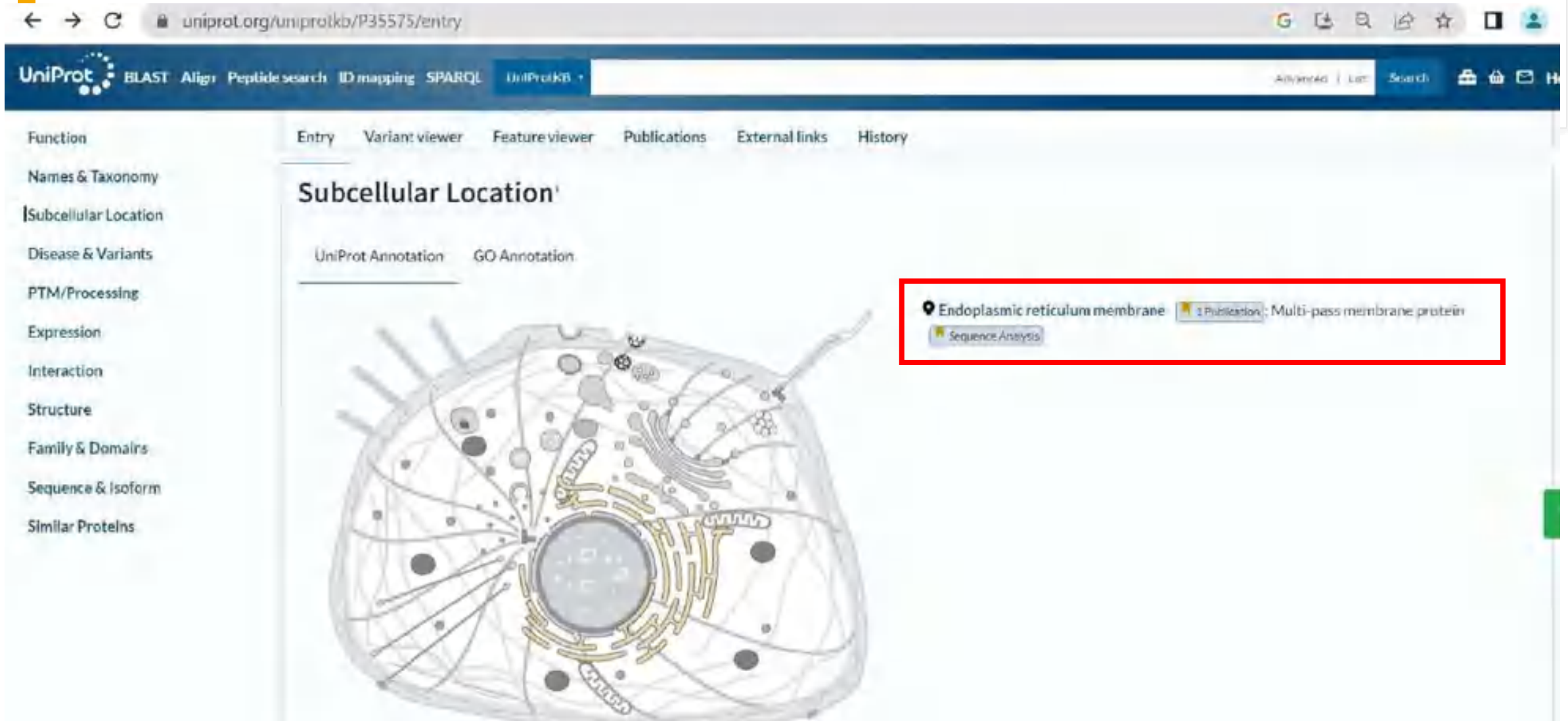
- Molecular function
- Biological process
- Cellular component

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



The screenshot displays the UniProt website interface for the protein entry P35575. The main content area is titled 'Subcellular Location' and includes a navigation menu with options like 'Entry', 'Variant viewer', 'Feature viewer', 'Publications', 'External links', and 'History'. Below the title, there are tabs for 'UniProt Annotation' and 'GO Annotation'. A diagram of a cell is shown, with various organelles labeled. A red box highlights the 'Endoplasmic reticulum membrane' annotation, which is linked to '1 Publication' and 'Multi-pass membrane protein'. A 'Sequence Analysis' button is also visible.

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB

Function  
Names & Taxonomy  
Subcellular Location  
Disease & Variants  
PTM/Processing  
Expression  
Interaction  
Structure  
Family & Domains  
Sequence & Isoform  
Similar Proteins

Entry Variant viewer Feature viewer Publications External links History

### Subcellular Location

UniProt Annotation GO Annotation

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

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB Search

Function  
Names & Taxonomy  
Subcellular Location  
**Disease & Variants**  
PTM/Processing  
Expression  
Interaction  
Structure  
Family & Domains  
Sequence & Isoform  
Similar Proteins

Entry Variant viewer Feature viewer Publications External links History

### Disease & Variants<sup>1</sup>

Involvement in disease<sup>1</sup>  
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	Publications
VAR_046249	5	M>R	in GSD1A: dbSNP:rs1250172816 <a href="#">↗</a>	1 Publication
VAR_046250	16	T>A	in GSD1A: dbSNP:rs761839506 <a href="#">↗</a>	1 Publication
VAR_046251	16	T>R	in GSD1A: complete loss of glucose-6-phosphatase activity and reduced enzyme stability; dbSNP:rs155558914 <a href="#">↗</a>	1 Publication
VAR_009202	20	Q>R	in GSD1A: dbSNP:rs2056021410 <a href="#">↗</a>	2 Publications
VAR_005937	22	D>V	in GSD1A: dbSNP:rs104801565 <a href="#">↗</a>	1 Publication



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

## MUTATION AND FUNCTIONS OF MUTANTS

The screenshot shows the UniProtKB entry for a natural variant (VAR\_005251) in the GSD1A gene. The variant is a C-to-R substitution at position 9. The page displays a sequence alignment with vertical bars indicating mutation positions and a table of features for mutagenesis.

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB

Function  
Names & Taxonomy  
Subcellular Location  
Disease & Variants  
PTM/Processing  
Expression  
Interaction  
Structure  
Family & Domains  
Sequence & Isoform  
Similar Proteins

Entry Variant Viewer Feature viewer Publications External links History

VAR\_005251 295 R>C in GSD1A: dbSNP:rs104894563 2 Publications

Expand Table

### Features

Showing features for natural variant (mutagenesis).

1 50 100 150 200 250 300 357

TYPE	ID	POSITION(S)	DESCRIPTION
Mutagenesis			
▶ 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

The screenshot shows the UniProt website interface for protein P35575. The left sidebar contains navigation links: Function, Names & Taxonomy, Subcellular Location, Disease & Variants, PTM/Processing (selected), Expression, Interaction, Structure, Family & Domains, Sequence & Isoform, and Similar Proteins. The main content area has tabs for Entry, Variant viewer, Feature viewer, Publications, External links, and History. The 'PTM/Processing' section is active, displaying 'Features' for chain 1, glycosylation. A sequence viewer shows a bar from position 1 to 357. Below it is a table of features:

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

The 'Glycosylation' row is highlighted with a red box. Below the table is a 'Keywords' section and an 'Expand table' button.

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

## 3D Predicted Structure of G6PC1 (Isoform I)

Function  
Names & Taxonomy  
Subcellular Location  
Disease & Variants  
PTM/Processing  
Expression  
Interaction  
**Structure**  
Family & Domains  
Sequence & Isoform  
Similar Proteins

Entry Variants viewer Feature viewer Publications External links History

### Structure<sup>1</sup>

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	IDENTIFIER	METHOD	RESOLUTION	CHAIN	POSITIONS	LINKS
<input type="text" value="- Select -"/>	AF-P35575-F1	<input type="text" value="- Select -"/>			1-357	AlphaFold

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

## 3D Predicted Structure of G6PCI (Isoform I)

The screenshot shows the UniProt entry for G6PCI (Isoform I). The 'Structure' section displays a 3D ribbon model of the protein. The model is color-coded by confidence: Very high (pLDDT > 90) in dark blue, Confident (70 > pLDDT > 70) in light blue, Low (50 > pLDDT > 50) in yellow, and Very low (pLDDT < 50) in red. The table below the structure lists the prediction method as AlphaFold.

SCORE	IDENTIFIER	METHOD	RESOLUTION	CHAIN	POSITIONS	LINKS
AlphaFold	AF_P03575_F1	Predicted			1-357	AlphaFold <a href="#">Download</a>

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 I of G6PCI

The screenshot shows the UniProt website interface. The top navigation bar includes the UniProt logo and various search options like BLAST, Align, Peptide search, ID mapping, SPARQL, and UniProtKB. The main content area is titled 'Entry' and includes tabs for 'Variant viewer', 'Feature viewer', 'Publications', 'External links', and 'History'. The 'Sequence & Isoform' section is highlighted with a red box, containing the text 'BLAST 2 isoforms' and 'Align 2 isoforms', and a note: 'This entry describes 2 isoforms<sup>1</sup> produced by **Alternative splicing**.' Below this, the entry 'P35575-1' is shown, with a note: '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.' The 'Name 1' section is highlighted with a green box, showing 'Length 357' and 'Mass (Da) 40,484'. The 'Last updated' and 'Checksum' information is also visible. The protein sequence is displayed in a grid format with residue numbers above each amino acid.

Function  
Names & Taxonomy  
Subcellular Location  
Disease & Variants  
PTM/Processing  
Expression  
Interaction  
Structure  
Family & Domains  
Sequence & Isoform  
Similar Proteins

Entry Variant viewer Feature viewer Publications External links History

## Sequence & Isoform<sup>1</sup>

BLAST 2 isoforms Align 2 isoforms

This entry describes 2 isoforms<sup>1</sup> 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

Tools + Download Add Highlight Copy sequence

Length 357  
Mass (Da) 40,484

Last updated 2008-09-23 v2  
Checksum<sup>1</sup> 2FEA1C78928A9919

MEEGMMVLHD FGIQSTHYLQ VNYQDSQDMF TLVSVIADLR NAFYVLFPIW FHLQEAVGTK LLWVAVIGDW LNLVFKWILF GQRPYMWVLD TDYYSNTSVP LIKQFPVTCE TGPGSPSGHA  
NGTAGVYYVM VTSTLSIFQG KIKPTYRFRC LNVILWLGFW AVQLNVCLSR IYLAHFPHQ VWAGVLSGIA VAETFSHIHS IYNASLKKYF LITFFLFSFA IGFYLLKGL GVDLLWTLEX  
AQRWCEQPEW VHIDITPFAS LLKNLGLTFG LGLALNSSMY RESCKGKLSK WLPFRLSSIV ASLVLLHVFD SLKPPSQVEL VFYVLSFCKS AVVPLASVSV IPYCLAQVLG QPHKKS



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

# SEQUENCE OF ISOFORM 2 of G6PCI

The screenshot shows the UniProt entry for P35575-2. The page includes a navigation bar with UniProt logo and search options. The main content area has tabs for Entry, Variant viewer, Feature viewer, Publications, External links, and History. The entry name 'P35575-2' is highlighted in a red box. Below it, the name 'Name 2' and a link to UniParc are shown. A 'Hide sequence' button is present. The sequence itself is displayed in a grey box with residue numbers 10-120 and 138-178. A 'Differences from canonical' section, also highlighted in a red box, lists differences at positions 115-175 and 176-356. The 'Length 176' and 'Mass (Da) 20,209' are highlighted in another red box. At the bottom, there is a section for 'Computationally mapped potential isoform sequences'.

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB + Advanced | Lite Search Help

Function  
Names & Taxonomy  
Subcellular Location  
Disease & Variants  
PTM/Processing  
Expression  
Interaction  
Structure  
Family & Domains  
Sequence & Isoform  
Similar Proteins

Entry Variant viewer Feature viewer Publications External links History

MDRRLEQPEW VHLDTTPAS LKRETEFEG LGLADRSSPT KESLAGKLSK MPPRSLSSIV ASEVELRVFD SERPPSQVEL VFYDLSPLRS AVVPLASVSV IPYCEAQVIG QPDKSL

**P35575-2**

Name 2  
See also [sequence in UniParc](#) or [sequence clusters in UniRef](#)

Hide sequence

Tools • Download Add Highlight Copy sequence

**Length 176**  
**Mass (Da) 20,209**

Checksum<sup>1</sup> 9185BD4F9911B02D

10 20 30 40 50 60 70 80 90 100 110 120  
MEEGMVLLHD FGTQSTHYLQ VNYQDSQWIF ILVSVIADLR NAFYVLEPTW FHLQFANGTE LILVAVIGDW LHLVFKWILF GQRFVWVWLD TDVYSNLSVP LIKQFPVTCE TGPGRKADL

138 140 150 160 170  
QISVLECHFV VGILGCAAEK LSVTNLPCCS FSSSSCCWSP VRHCCCRNFQ PHPQHL

Differences from canonical 115-175:  
SPSGHAMGTAGVYVYVMTSTLSTIFQGKIKPTVRFRCILNITLWLGFWAVQ  
LNVCLSRIVLAA +  
KDKADLQISVLECHFVVGILGCAAEKLSVTNLPCCSFSSSSCCWSPVRH  
CCCRNFQPHPQH [1 Publication](#)  
176-356: Missing [1 Publication](#)

Computationally mapped potential isoform sequences<sup>2</sup>  
There is 1 potential isoform mapped to this entry

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

The screenshot shows the UniProt 'Align results' page for protein P35575. The 'Overview' tab is selected. The alignment compares two isoforms: sp:P35575-1|G6PC1\_HUMAN and sp:P35575-2|G6PC1\_HUMAN. The sequences are shown in a grid format with a yellow highlight indicating the alignment. The alignment is 100% identical for the first 176 amino acids. The second isoform has a deletion of 10 amino acids (residues 177-186) compared to the first isoform. The UniProt logo and navigation menu are visible at the top.

UniProt BLAST Align Peptide search ID mapping SPARQL Tool results + Advanced | Log Search Help

## Align results

Overview Trees Percent Identity Matrix Text Output Input Parameters API Request

Download Resubmit

Highlight properties Select annotation View: Overview  Wrapped

sp:P35575-1 G6PC1_HUMAN	MEEGMNVLHDFGIQSTHYLQVNYQDSQDWFILVSVIADLRNAFYVLFPIWFHLQEA VG I KL	61
sp:P35575-2 G6PC1_HUMAN	MEEGMNVLHDFGIQSTHYLQVNYQDSQDWFILVSVIADLRNAFYVLFPIWFHLQEA VG I KL	61
<b>P35575:Chain</b>		
sp:P35575-1 G6PC1_HUMAN	LWVAVIGDWLNLVFKWILEGQRPYWWVLDTDYYSNTSVPLIKQFPVTCETGPGSPSGHAMG	122
sp:P35575-2 G6PC1_HUMAN	LWVAVIGDWLNLVFKWILEGQRPYWWVLDTDYYSNTSVPLIKQFPVTCETGPGKDKADLQI	122
<b>P35575:Chain</b>		
sp:P35575-1 G6PC1_HUMAN	TAG-VYYVMVTSTLSIFQGKIKPTYRFRCLNVILWLGFWAVQLNVCLSRIYLA AH FPHQVV	182
sp:P35575-2 G6PC1_HUMAN	SVLECHFV--VGI LGCA-AECLSVTNLPCCSFSSSSCCWSPVRHCCCRN-FQP-H-PQHL-	176
<b>P35575:Chain</b>		
sp:P35575-1 G6PC1_HUMAN	AGVLSGIAVAETFSHIHSIYNASLKKYFLITFFLFSFAIGFYLLLKGLGVDLLWTLEKAQR	243
sp:P35575-2 G6PC1_HUMAN	-----	176
<b>P35575:Chain</b>		
sp:P35575-1 G6PC1_HUMAN	WCEQPEWVHTDTPPFASLLKNLGTLEGLGLALNSSMYRESCCKGKLSKWLPFRLSSIVASLV	304
sp:P35575-2 G6PC1_HUMAN	-----	176
<b>P35575:Chain</b>		

CLICK ON BLAST

unirot.org/uniprotkb/P35575/entry

Google Chrome isn't your default browser [Set as default](#)

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB  [Advanced](#) | [List](#) [Search](#) [Print](#) [Share](#) [Help](#)

Function Entry Variant viewer Feature viewer Publications External links History

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

**Tools** [Download](#) [Add](#) [Highlight](#) [Copy sequence](#)

BLAST  
ProtParam  
ProtScale  
Compute pI/Mw  
PeptideMass  
PeptideCutter

**Last updated** 2008-08-23 v2  
**Checksum** 2FEA1C78928A9919

STHYLQ	VHYQDSQDMF	ILVSVIADLR	NAFYVLFPIW	FHLQEAVGIK	LLKVAVIGDM	LNLVFXWILF	GQXPYWVLD	TDYYSNTSVP	LKQFPVTC	TGPGSPSGMA
LSIFQG	KIKPTYRFRC	LWVILWLGFW	AVQLRVCLSR	IYLAAHFPHQ	VVAGVLSGIA	VAETFSHIH5	IYNASLKKYF	LITFFLF5FA	IGFYLLKGL	GVDLLWLEK
ITPFAS	LLKNLGTEFG	LGLALN55PY	RESCKGKLSK	WLPFRLSSIV	ASLVLLHVFQ	SLKPPSQVEL	VFYVLSFCKS	AVVPLASVSV	IPYCLAQVLG	QPHKKS

**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: 0.64 - 100

Score: <380 - 1800

**Status**

- Reviewed (Swiss-Prot) (7)
- Unreviewed (TrEMBL) (244)

**Popular organisms**

- Human (2)
- Rat (2)
- Ervine (1)
- Mouse (1)

### BLAST 250 results found in UniProtKB

Overview | Taxonomy | Hit Distribution | Text Output | Input Parameters | API Request

Download | Customize columns | Resubmit

Entry	Entry Name	Protein Names	Gene Names	Organism	Length	Identity
P35575	G6PC1_HUMAN	Glucose-6-phosphatase catalytic subunit 1 [...]	G6PC1, G6PC, G6PT	Homo sapiens (Human)	357 AA	100%
G3RX42	G3RX42_GORGO	Glucose-6-phosphatase [...]	G6PC1	Gorilla gorilla gorilla (Western lowland gorilla)	357 AA	99.4%
A0A2R9AIQ5	A0A2R9AIQ5_PANPA	Glucose-6-phosphatase [...]	G6PC1	Pan paniscus (Pygmy chimpanzee) (Bonobo)	357 AA	99.4%
A0A2I3SGU1	A0A2I3SGU1_PANTR	Glucose-6-phosphatase [...]	G6PC1	Pan troglodytes (Chimpanzee)	357 AA	99.4%
G1RMH0	G1RMH0_NOMLE	Glucose-6-phosphatase	G6PC1	Nomascus leucogenys	357 AA	97.8%

CLICK ON BLAST

UniProt BLAST Align Peptide search ID mapping SPARQL UniProtKB

Function  
Names & Taxonomy  
Subcellular Location  
Disease & Variants  
PTM/Processing  
Expression  
Interaction  
Structure  
Family & Domains  
| Sequence & Isoform  
Similar Proteins

Entry Variant viewer Feature viewer Publications External links History

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

Tools [Download](#) [Add](#) [Highlight](#) [Copy sequence](#)

**BLAST**  
**ProtParam**  
ProtScale  
Compute pI/Mw  
PeptideMass  
PeptideCutter

**Last updated** 2008-08-23 v2  
**Checksum** 2FEA1C78928A9919

STHYLQ VHYQDSQDMF ILVSVIADLR NAFYVLFPIW FHLQEAVGIK LLKVAVIGDM LNLVFXWILF GQXPYWVVLID TDYYSNTSVP LIKQFPVTCE TGPGPSGMA  
LSIFQG KIKPTYRFRC LHWILWLGFW AVQLRVCLSR IYLAAHFPHQ VVAGVLSGIA VAETFSHIH5 IYNASLKKYF LITFFLF5FA IGFYLLKGL GVDLLWLEK  
ITPFAS LLKNLGTEFG LGLALN55PY RESCKGRLSK WLPFRLSSIV ASLVLIHVFQ SLKPPSQVEL VFYVLSFCKS AVVPLASVSV IPIYCLAQVLG QPHKKS

**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	Luminal
FT	TRANSMEM	29-49	Helical
FT	TOPO_DOM	50-60	Cytoplasmic
FT	TRANSMEM	61-81	Helical
FT	TOPO_DOM	82-117	Luminal
FT	TRANSMEM	118-138	Helical
FT	TOPO_DOM	139-147	Cytoplasmic
FT	TRANSMEM	148-168	Helical
FT	TOPO_DOM	169-179	Luminal
FT	TRANSMEM	180-202	Helical
FT	TOPO_DOM	203-209	Cytoplasmic
FT	TRANSMEM	210-230	Helical
FT	TOPO_DOM	231-254	Luminal
FT	TRANSMEM	255-275	Helical
FT	TOPO_DOM	276-291	Cytoplasmic
FT	TRANSMEM	292-312	Helical
FT	TOPO_DOM	313-320	Luminal
FT	TRANSMEM	321-341	Helical
FT	TOPO_DOM	342-357	Cytoplasmic



# PROTSCALE

web.expasy.org/cgi-bin/protscale/protscale.pl?P35575



Expasy

ProtScale

Home | [Contact](#)

## 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**):

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.

- |  |   |
|--|---|
| <input type="radio"/> Molecular weight                         | <input type="radio"/> Number of codon(s)        |
| <input type="radio"/> Bulkiness                                | <input type="radio"/> Polarity / Zimmerman      |
| <input type="radio"/> Polarity / Grantham                      | <input type="radio"/> Refractivity              |
| <input type="radio"/> Recognition factors                      | <input type="radio"/> Hphob. / Eisenberg et al. |
| <input type="radio"/> Hphob. OMH / Sweet et al.                | <input type="radio"/> Hphob. / Hopp & Woods     |
| <input checked="" type="radio"/> Hydropath. / Kyte & Doolittle | <input type="radio"/> Hphob. / Manavalan et al. |
| <input type="radio"/> Hphob. / Abraham & Leo                   | <input type="radio"/> Hphob. / Black            |

# PROTSKALE...

web.expasy.org/cgi-bin/protscale/protscale.pl?P35575

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



# PROTSACLE...

The screenshot shows a web browser window with the URL `web.expasy.org/cgi-bin/protscale/protscale.pl?P35575`. The page displays a list of 30 protein analysis metrics, each with a radio button for selection. The metrics are arranged in two columns:

- 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

Below the list, there are several configuration options:

- Window size:
- Relative weight of the window edges compared to the window center (in %):
- 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

A link is provided: "If you need more information about how to set these parameters, please click here."

At the bottom, there are two buttons: "Submit" and "Reset".

# COMPUTE pI/Mw

Browser tabs: G6PC1 - Glucose-6-phosphatas- x Expasy x Expasy - ProtScale x Expasy - ProtParam +

Address bar: web.expasy.org/cgi-bin/compute\_pi/pi\_tool?P35575@noft@average

Expasy logo | Compute pI/Mw | Home | Contact

## 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 G6PC I Retrieved from UniProt

Location within the cell: Endoplasmic Reticulum Membrane

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

Variants that cause Glycogen Storage Disease IA (GSD IA): 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, 81 G>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/>

The screenshot shows the ENZYME database website. At the top, there is a navigation bar with the Expasy logo and links for Home, Browse, Search, Documentation, Downloads, and Contact. The main heading is "ENZYME - Enzyme nomenclature database". Below this, a paragraph explains that ENZYME is a repository of information based on the recommendations of the Nomenclature Committee of the International Union of Biochemistry and Molecular Biology (IUBMB). It also mentions that EC (Enzyme Commission) numbers are provided. A note states that a few EC numbers are preliminary and include an 'n' as part of the fourth digit. A release date of 28-Jun-23 with 8745 active entries is noted. The page is divided into four sections: "Access to ENZYME" with search options by EC number, enzyme class, description, chemical compound, and comments; "Documents" with links to the user manual and how to obtain the database; "Services" with links to report forms and FTP downloads; and "Related tools and databases" with links to Rhea, IUBMB ExplorEnz, and BRENDA.

ENZYME

Expasy [Home](#) [Browse](#) [Search](#) [Documentation](#) [Downloads](#) [Contact](#)

## 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 (8745 active entries)*

### Access to ENZYME

- by EC number:
- by enzyme class
- by description (official name) or alternative name(s):
- 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

## 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



# ENZYME entry: EC 3.1.3.9

### Accepted Name

**glucose-6-phosphatase**

### Reaction catalysed



### 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 2.7.1.78 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	O9NQR9, G6PC2_HUMAN
	Q9Z186, G6PC2_MOUSE	Q148G2, G6PC3_BOVIN	A1A5Z0, G6PC3_DANRE
	Q9BUM1, G6PC3_HUMAN	O6NSO9, G6PC3_MOUSE	O6AZ83, 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.**

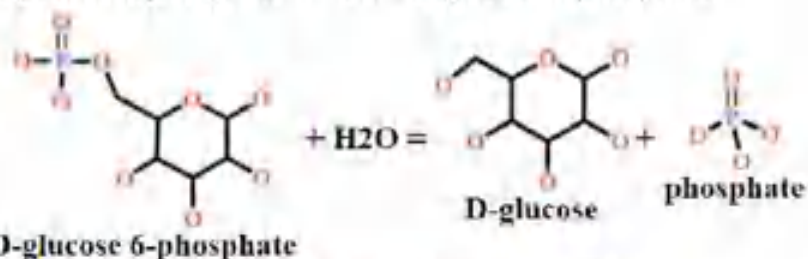
0 PDB entries

**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 + H<sub>2</sub>O = *D*-glucose + phosphate.

 Molecule diagrams generated from imol files obtained from the [KEGG](http://KEGG) [http site](http://ftp.KEGG).

**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:** [PDB](#) [IntEnz](#) [Expasy](#) [KEGG](#)

 There are no PDB entries in enzyme class [E.C.3.1.3.9](#)





## ENZYME: 3.1.3.9

[Help](#)

Entry	EC 3.1.3.9 Enzyme
Name	glucose-6-phosphatase; glucose 6-phosphate phosphatase
Class	Hydrolases; Acting on ester bonds; Phosphoric-monoester hydrolases <a href="#">BRITE hierarchy</a>
Sysname	D-glucose-5-phosphate phosphohydrolase
Reaction(IUBMB)	D-glucose 6-phosphate + H <sub>2</sub> O = D-glucose + phosphate [RN:R00303]
Reaction(KEGG)	R00303 > R01788 <a href="#">Reaction</a>
Substrate	D-glucose 6-phosphate [CPD:C00092]; H <sub>2</sub> O [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)  
   PDB (11)  
 DNA sequence (89)  
   RefSeq(nuc) (41)  
   GenBank (29)  
   EMBL (19)  
 Protein domain (4)  
   InterPro (3)  
   Pfam (1)  
 All databases (4744)

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	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)].
<b>History</b>	EC 3.1.3.9 created 1961
<b>Pathway</b>	ec00010 Glycolysis / Gluconeogenesis ec00052 Galactose metabolism ec00500 Starch and sucrose metabolism ec01100 Metabolic pathways ec01110 Biosynthesis of secondary metabolites
<b>Orthology</b>	K01084 glucose-6-phosphatase
<b>Genes</b>	HSA: 2538(G6PC1) 57818(G6PC2) 92579(G6PC3) PTR: 454703(G6PC) 454720(G6PC3) 741431(G6PC2) PPS: 100974757(G6PC) 100984489(G6PC2) 100993144(G6PC3) GGD: 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 <a href="#">show all</a> <a href="#">Taxonomy</a>
<b>Reference</b>	1 [PMID:169241]
<b>Authors</b>	Anchors JM, Karnovsky ML.
<b>Title</b>	Purification of cerebral glucose-6-phosphatase. An enzyme involved in sleep.
<b>Journal</b>	J Biol Chem 250:6408-16 (1975)
<b>Reference</b>	2 [PMID:154220]
<b>Authors</b>	Colilla W, Jorgenson RA, Nordlie RC.
<b>Reference</b>	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

The screenshot shows the OMIM website interface. At the top, there is a navigation bar with links for About, Statistics, Downloads, Contact Us, MIMmatch, Donate, and Help. Below the navigation bar, a search bar contains the text '613742', which is circled in red. To the right of the search bar, there are options for 'View Results as:' (Gene Map Table, Clinical Synopsis) and 'Display:' (Highlights). Below the search bar, the search results are displayed. The first result is highlighted in yellow and is '613742. GLUCOSE-6-PHOSPHATASE, CATALYTIC, G6PC'. The second result is '232200. GLYCOGEN STORAGE DISEASE Ia; GSD1A'. The third result is '604556. DUAL-SPECIFICITY TYROSINE PHOSPHORYLATION-REGULATED KINASE 1B; DYRK1B'. The fourth result is '608058. GLUCOSE-6-PHOSPHATASE, CATALYTIC, 2; G6PC2'. The fifth result is '232220. GLYCOGEN STORAGE DISEASE Ib; GSD1B'.

About Statistics Downloads Contact Us MIMmatch Donate Help

613742

Search: '613742'  
Results: 15 entries

Show 100 | Download As | First | Previous | Next | Last

- 1: **613742. GLUCOSE-6-PHOSPHATASE, CATALYTIC, G6PC**  
Cytogenetic location: 17q21.31, Genomic coordinates (GRCh38): 17,42,900,799-42,911,438  
Matching terms: 613742  
▶ Gene-Phenotype Relationships ▶ ICD4 ▶ Links
- 2: **232200. GLYCOGEN STORAGE DISEASE Ia; GSD1A**  
Cytogenetic location: 17q21.31  
Matching terms: 613742  
▶ Phenotype-Gene Relationships ▶ Phenotypic Series ▶ ICD4 ▶ 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
- 5: **232220. GLYCOGEN STORAGE DISEASE Ib; GSD1B**

Search OMIM...

Options Display: Highlights

- #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

# 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

[Clinical Synopsis](#)
[Phenotypic Series](#)
[PhenGene Graphics](#)

- ICD+
- External Links
    - Protein
    - Clinical Resources
      - Clinical Trials
      - EuroGentest
      - Gene Reviews
      - Genetic Alliance
      - GTR
      - GARD
      - Orphanet
      - POSSUM
    - Animal Models
    - Cell Lines



Search OMIM...

Options + Display: Highlights

#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

[Clinical Synopsis](#)
[Phenotypic Series](#)
[PhenGene Graphics](#)

#### 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