INS insulin [*Homo sapiens* (human)]

Download Datasets

Gene ID: 3630, updated on 28-Aug-2022

Summary

Official Symbol	INS provided by HGNC
Official Full Name	insulin provided by HGNC
Primary source	HGNC:HGNC:6081
See related	Ensembl:ENSG00000254647 MIM:176730; AllianceGenome:HGNC:6081
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	IDDM; ILPR; IRDN; IDDM1; IDDM2; PNDM4; MODY10
Summary	This gene encodes insulin, a peptide hormone that plays a vital role in the regulation of carbohydrate and lipid metabolism. After removal of the precursor signal peptide, proinsulin is post-translationally cleaved into three peptides: the B chain and A chain peptides, which are covalently linked via two disulfide bonds to form insulin, and C-peptide. Binding of insulin to the insulin receptor (INSR) stimulates glucose uptake. A multitude of mutant alleles with phenotypic effects have been identified, including insulin-dependent diabetes mellitus, permanent neonatal diabetes diabetes mellitus, maturity-onset diabetes of the young type 10 and hyperproinsulinemia. There is a read-through gene, INS-IGF2, which overlaps with this gene at the 5' region and with the IGF2 gene at the 3' region. [provided by RefSeq, May 2020]
Expression	Restricted expression toward pancreas (RPKM 671.7) See more
Orthologs	mouse all
NEW	**Try the new Gene table** **Try the new Transcript table**

Genomic context

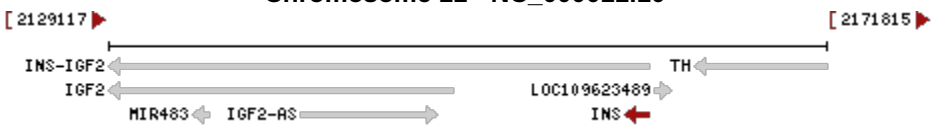
See INS in Genome Data Viewer

Location: 11p15.5

Exon count: 3

Annotation release	Status	Assembly	Chr	Location
110	current	GRCh38.p14 (GCF_000001405.40)	11	NC_000011.10 (2159779..2161209, complement)
110	current	T2T-CHM13v2.0 (GCF_009914755.1)	11	NC_060935.1 (2247427..2248857, complement)
105.20220307	previous assembly	GRCh37.p13 (GCF_000001405.25)	11	NC_000011.9 (2181009..2182439, complement)

Chromosome 11 - NC_000011.10

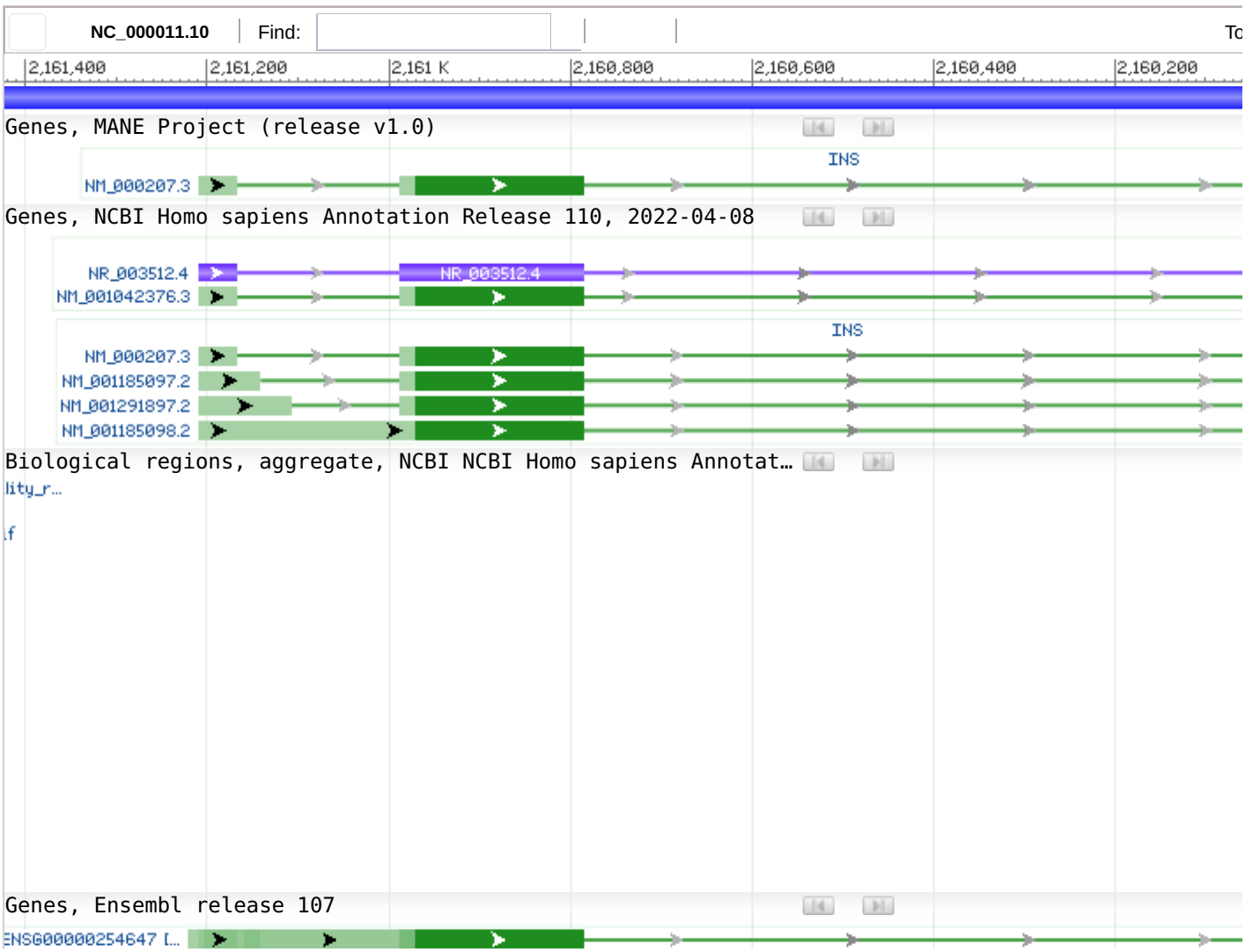


Genomic regions, transcripts, and products

Go to reference sequence details

Genomic Sequence: NC_000011.10 Chromosome 11 Reference GRCh38.p14 Primary Assembly

Go to nucleotide: Graphics FASTA GenBank



Expression

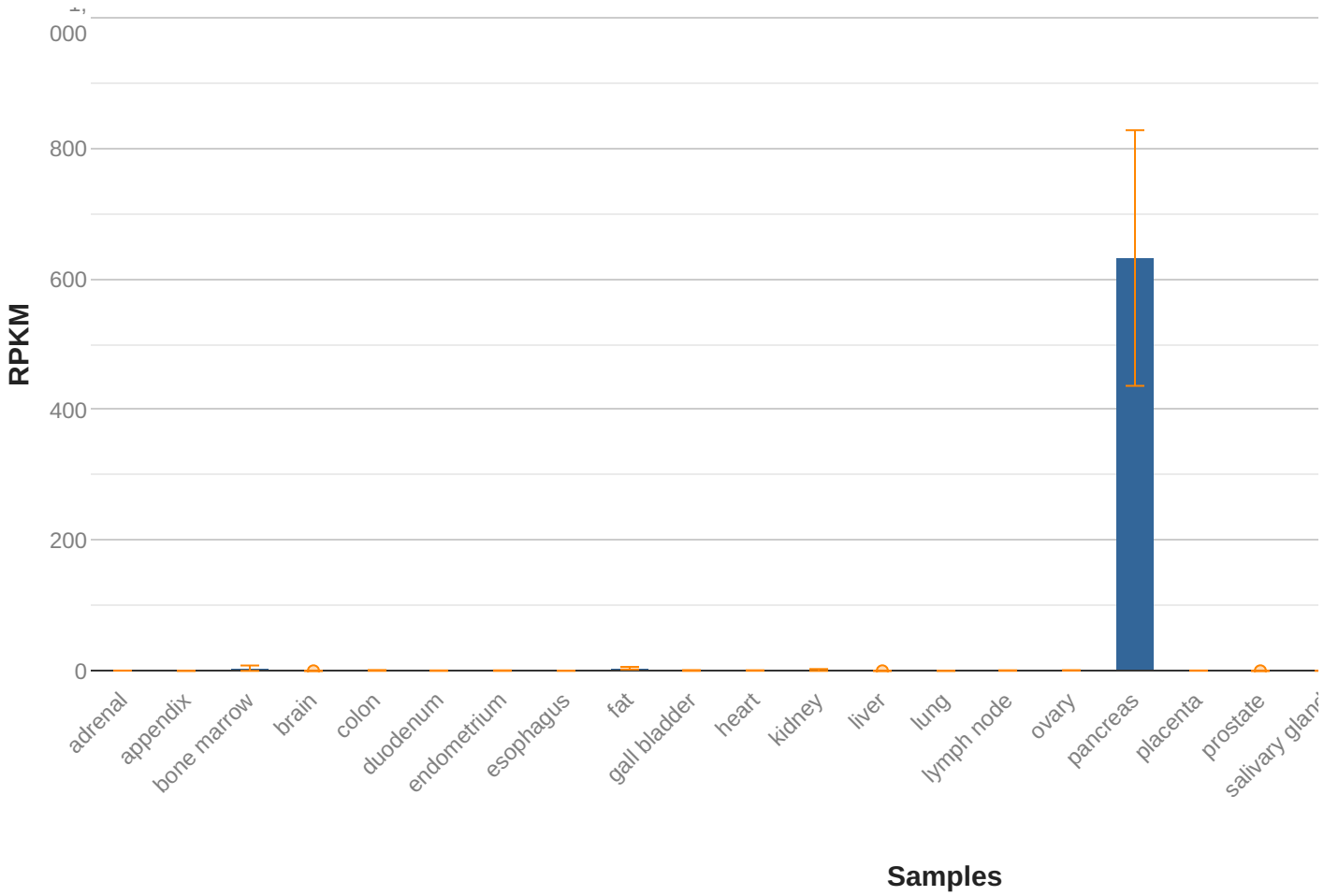
See details

HPA RNA-seq normal tissues

- 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



Bibliography

Related articles in PubMed

1. Insulin Directs Dichotomous Translational Regulation to Control Human Pluripotent Stem Cell Survival, Proliferation and Pluripotency.
   Zhou X, *et al*. Int J Biol Sci, 2022. PMID 35813470, **Free PMC Article**
2. Comparison of Quality of Carbohydrate Metrics Related to Fasting Insulin, Glycosylated Hemoglobin and HOMA-IR in Brazilian Adolescents.
   da Rocha CMM, *et al*. Nutrients, 2022 Jun 19. PMID 35745274, **Free PMC Article**
3. Participation of Magnesium in the Secretion and Signaling Pathways of Insulin: an Updated Review.
   de Sousa Melo SR, *et al*. Biol Trace Elem Res, 2022 Aug. PMID 35666386
4. Association between single nucleotide polymorphisms in non-coding regions of the insulin (INS) gene and schizophrenia.
   Melkersson K, *et al*. Neuro Endocrinol Lett, 2022 Apr. PMID 35490421
5. A Novel Nonsense *INS* Mutation Causes Inefficient Preproinsulin Translocation Into the Endoplasmic Reticulum.

Yang Y, *et al*. Front Endocrinol (Lausanne), 2021. PMID 35069438, **Free PMC Article**

See all (970) citations in PubMed

See citations in PubMed for homologs of this gene provided by HomoloGene

GeneRIFs: Gene References Into Functions

What's a GeneRIF?

1. Aberrant crosstalk between insulin signaling and mTOR in young Down syndrome individuals revealed by neuronal-derived extracellular vesicles.
2. Association between single nucleotide polymorphisms in non-coding regions of the insulin (INS) gene and schizophrenia.
3. Insulin Directs Dichotomous Translational Regulation to Control Human Pluripotent Stem Cell Survival, Proliferation and Pluripotency.
4. Comparison of Quality of Carbohydrate Metrics Related to Fasting Insulin, Glycosylated Hemoglobin and HOMA-IR in Brazilian Adolescents.
5. Participation of Magnesium in the Secretion and Signaling Pathways of Insulin: an Updated Review.
6. Progressive endoplasmic reticulum stress over time due to human insulin gene mutation contributes to pancreatic beta cell dysfunction.
7. In celebration of a century with insulin - Update of insulin gene mutations in diabetes.
8. Nicotinamide mononucleotide: a potential effective natural compound against insulin resistance.
9. A Novel Nonsense INS Mutation Causes Inefficient Preproinsulin Translocation Into the Endoplasmic Reticulum.
10. Evaluation of serum insulin-like growth factor-1, insulin, glucose levels in patients with adolescent and post-adolescent acne.

Submit: New GeneRIF Correction See all GeneRIFs (734)

Phenotypes

BioGRID CRISPR Screen Phenotypes (382 hits/1121 screens)

Find tests for this gene in the NIH Genetic Testing Registry (GTR)

Review eQTL and phenotype association data in this region using PheGenI

Associated conditions

Description	Tests
Diabetes mellitus, permanent neonatal 4 MedGen: C5394307, OMIM: 618858, GeneReviews: Not available	Compare labs
Hyperproinsulinemia MedGen: C0342283, OMIM: 616214, GeneReviews: Not available	Compare labs

Description	Tests
Maturity-onset diabetes of the young type 10 MedGen: C3150617, OMIM: 613370, GeneReviews: Maturity-Onset Diabetes of the Young Overview	Compare labs
Permanent neonatal diabetes mellitus MedGen: C1833104, GeneReviews: Permanent Neonatal Diabetes Mellitus	Compare labs
Type 1 diabetes mellitus 2 MedGen: C1852092, OMIM: 125852, GeneReviews: Not available	Compare labs

Copy number response

Description
Copy number response Haploinsufficency No evidence available (Last evaluated 2012-03-22) ClinGen Genome Curation Page Triplosensitivity No evidence available (Last evaluated 2012-03-22) ClinGen Genome Curation Page

EBI GWAS Catalog

Description
A genome-wide association study identifies KIAA0350 as a type 1 diabetes gene. EBI GWAS Catalog EBI GWAS Catalog, PubMed
A possible mechanism behind autoimmune disorders discovered by genome-wide linkage and association analysis in celiac disease. EBI GWAS Catalog EBI GWAS Catalog, PubMed
Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases. EBI GWAS Catalog EBI GWAS Catalog, PubMed
Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. EBI GWAS Catalog EBI GWAS Catalog, PubMed
Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. EBI GWAS Catalog EBI GWAS Catalog, PubMed

Description
Robust associations of four new chromosome regions from genome-wide analyses of type 1 diabetes. EBI GWAS Catalog EBI GWAS Catalog, PubMed

Variation

See variants in ClinVar

See studies and variants in dbVar

See Variation Viewer (GRCh37.p13)

See Variation Viewer (GRCh38)

Genotypes

See SNP Geneview Report
See 1000 Genomes Browser (GRCh37.p13)

HIV-1 interactions

Protein interactions

Protein	Gene	Interaction	Pubs
Vpr	vpr	HIV-1 Vpr antagonizes insulin's effect on the expression of glucose 6-phosphatase, manganese superoxide dismutase, and sterol carrier protein 2 genes in HepG2 cells	PubMed
	vpr	HIV-1 Vpr inhibits insulin-induced association of 14-3-3 and Foxo3a in HeLa cells	PubMed
	vpr	HIV-1 Vpr inhibits insulin-induced cytoplasmic translocation of Foxo3a, a subtype of the forkhead transcription factors	PubMed

Go to the HIV-1, Human Interaction Database

Pathways from PubChem

61 items View More Rows & Details

SORT BY Taxonomy

Pathway	Source	External ID	Taxonomy
Developmental Biology	Reactome	R-HSA-1266738	Homo sapiens (human)
Regulation of beta-cell development	Reactome	R-HSA-186712	Homo sapiens (human)
Regulation of gene expression in beta cells	Reactome	R-HSA-210745	Homo sapiens (human)
Metabolism	Reactome	R-HSA-1430728	Homo sapiens (human)
Integration of energy metabolism	Reactome	R-HSA-163685	Homo sapiens (human)
Regulation of insulin secretion	Reactome	R-HSA-422356	Homo sapiens (human)
Signaling Pathways	Reactome	R-HSA-162582	Homo sapiens (human)
Signaling by Receptor Tyrosine Kinases	Reactome	R-HSA-9006934	Homo sapiens (human)
Signaling by Insulin receptor	Reactome	R-HSA-74752	Homo sapiens (human)
Insulin receptor signalling cascade	Reactome	R-HSA-74751	Homo sapiens (human)
IRS activation	Reactome	R-HSA-74713	Homo sapiens (human)
Signal attenuation	Reactome	R-HSA-74749	Homo sapiens (human)
Insulin receptor recycling	Reactome	R-HSA-77387	Homo sapiens (human)
Metabolism of proteins	Reactome	R-HSA-392499	Homo sapiens (human)
Peptide hormone metabolism	Reactome	R-HSA-2980736	Homo sapiens (human)
Insulin processing	Reactome	R-HSA-264876	Homo sapiens (human)
Synthesis, secretion, and deacylation of Ghrelin	Reactome	R-HSA-422085	Homo sapiens (human)
Gene expression (Transcription)	Reactome	R-HSA-74160	Homo sapiens (human)
RNA Polymerase II Transcription	Reactome	R-HSA-73857	Homo sapiens (human)
Generic Transcription Pathway	Reactome	R-HSA-212436	Homo sapiens (human)
Cori cycle	WikiPathways	WP1946	Homo sapiens (human)
Sterol regulatory element-binding proteins (SREBP) signaling	WikiPathways	WP1982	Homo sapiens (human)
Alzheimer's disease and miRNA effects	WikiPathways	WP2059	Homo sapiens (human)
Adipogenesis	WikiPathways	WP236	Homo sapiens (human)
Cardiac progenitor differentiation	WikiPathways	WP2406	Homo sapiens (human)

1 2 3 Next

PubChem

Interactions

Items 1 - 25 of 525 << First < Prev Page 1 of 21 Next > Last >>

Products	Interactant	Other Gene	Complex	Source	Pubs	Description
P01308	P48745	CCN3		HPRD	PubMed	
P01308	P16870	CPE		HPRD	PubMed	
P01308	P07858	CTSB		HPRD	PubMed	
P01308	P07339	CTSD		HPRD	PubMed	
P01308	P14091	CTSE		HPRD	PubMed	
P01308	P35557	GCK		HPRD	PubMed	
P01308	P01906	HLA-DQA2		HPRD	PubMed	
P01308	P01918	HLA-DQB1		HPRD	PubMed	
P01308	P14735	IDE		HPRD	PubMed	
P01308	P08069	IGF1R		HPRD	PubMed	
P01308	Q16270	IGFBP7		HPRD	PubMed	
P01308	P01308	INS		HPRD	PubMed	
P01308	P06213	INSR		HPRD	PubMed	
P01308	P98164	LRP2		HPRD	PubMed	
P01308	P06400	RB1		HPRD	PubMed	
P01308	Q96C24	SYTL4		HPRD	PubMed	
P01308	Q9BRA2	TXNDC17		HPRD	PubMed	
BioGRID:109842	BioGRID:119004	A1CF		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109842	BioGRID:116520	AAK1		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109842	BioGRID:106540	ABCB7		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109842	BioGRID:112700	ABCC8		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109842	BioGRID:111783	ABCD3		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109842	BioGRID:106541	ABCF1		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109842	BioGRID:115372	ABCF2		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109842	BioGRID:117541	ABHD12		BioGRID	PubMed	Affinity Capture-MS

Items 1 - 25 of 525 << First < Prev Page 1 of 21 Next > Last >>

General gene information

Markers

Readthrough INS-IGF2

Readthrough gene: INS-IGF2, Included gene: IGF2

Homology

Homologs of the INS gene: The INS gene is conserved in chimpanzee, dog, mouse, rat, chicken, zebrafish, and frog.
Orthologs from Annotation Pipeline: 269 organisms have orthologs with human gene INS
Orthologs

Gene Ontology Provided by GOA

Function	Evidence Code	Pubs
enables hormone activity	IC	PubMed
enables hormone activity	IMP	PubMed
enables hormone activity	NAS	PubMed
enables identical protein binding	IPI	PubMed
enables insulin receptor binding	IDA	PubMed
enables insulin receptor binding	IPI	PubMed
enables insulin-like growth factor receptor binding	IPI	PubMed
enables protease binding	IPI	PubMed
enables protein binding	IPI	PubMed

Items 1 - 25 of 69 << First < Prev Page 1 of 3 Next > Last >>

Process	Evidence Code	Pubs
involved_in G protein-coupled receptor signaling pathway	IDA	PubMed
involved_in activation of protein kinase B activity	IDA	PubMed
involved_in acute-phase response	IDA	PubMed
involved_in alpha-beta T cell activation	IDA	PubMed
involved_in cell-cell signaling	IC	PubMed
involved_in cognition	TAS	PubMed
involved_in fatty acid homeostasis	IMP	PubMed
involved_in glucose homeostasis	IBA	PubMed
involved_in glucose homeostasis	IMP	PubMed
involved_in glucose metabolic process	IEA	
involved_in insulin receptor signaling pathway	IDA	PubMed
involved_in negative regulation of NAD(P)H oxidase activity	IDA	PubMed
involved_in negative regulation of acute inflammatory response	IDA	PubMed
involved_in negative regulation of fatty acid metabolic process	IMP	PubMed

Process	Evidence Code	Pubs
involved_in negative regulation of feeding behavior	IDA	PubMed
acts_upstream_of negative regulation of gene expression	IDA	PubMed
involved_in negative regulation of gluconeogenesis	NAS	PubMed
involved_in negative regulation of glycogen catabolic process	IMP	PubMed
involved_in negative regulation of lipid catabolic process	IMP	PubMed
involved_in negative regulation of lipid catabolic process	NAS	PubMed
involved_in negative regulation of oxidative stress-induced intrinsic apoptotic signaling pathway	NAS	PubMed
involved_in negative regulation of protein catabolic process	IDA	PubMed
involved_in negative regulation of protein secretion	IDA	PubMed
involved_in negative regulation of proteolysis	IMP	PubMed
involved_in negative regulation of reactive oxygen species biosynthetic process	IGI	PubMed

Items 1 - 25 of 69 << First < Prev Page 1 of 3 Next > Last >>

Component	Evidence Code	Pubs
located_in Golgi lumen	TAS	
located_in Golgi membrane	TAS	
located_in endoplasmic reticulum lumen	TAS	
located_in endoplasmic reticulum-Golgi intermediate compartment membrane	TAS	
located_in endosome lumen	TAS	
located_in extracellular region	IC	PubMed
located_in extracellular region	TAS	
is_active_in extracellular space	IBA	PubMed
located_in extracellular space	IDA	PubMed
located_in secretory granule lumen	TAS	
located_in transport vesicle	TAS	

General protein information

Preferred Names
insulin

Names
preproinsulin
proinsulin

NCBI Reference Sequences (RefSeq)

NEW **Try the new Transcript table**

RefSeqs maintained independently of Annotated Genomes

These reference sequences exist independently of genome builds. Explain

Genomic

1. **NG_007114.1 RefSeqGene**

Range	4986..6416
Download	GenBank, FASTA, Sequence Viewer (Graphics)

mRNA and Protein(s)

1. **NM_000207.3 → NP_000198.1 insulin preproprotein**

   See identical proteins and their annotated locations for NP_000198.1

   Status: REVIEWED

Description	Transcript Variant: This variant (1) represents the shortest variant. All variants encode the same protein.
Source sequence(s)	BC005255, BM510748
Consensus CDS	CCDS7729.1
UniProtKB/Swiss-Prot	P01308, Q5EEX2
UniProtKB/TrEMBL	I3WAC9
Related	ENSP00000370731.5, ENST00000381330.5

Conserved Domains (1) summary

	cd04367 **Location:26 → 110**	IIGF_insulin_like; IIGF_like family, insulin_like subgroup, specific to vertebrates. Members include a number of peptides including insulin and insulin-like growth factors I and II, which play a variety of roles in controlling processes such as metabolism, growth and ...

2. **NM_001185097.2 → NP_001172026.1 insulin preproprotein**

   See identical proteins and their annotated locations for NP_001172026.1

   Status: REVIEWED

Description	Transcript Variant: This variant (2) differs in the 5' UTR, compared to variant 1. All variants encode the same protein.
Source sequence(s)	AY899304, BM510347, BP322143
Consensus CDS	CCDS7729.1
UniProtKB/Swiss-Prot	P01308, Q5EEX2
UniProtKB/TrEMBL	I3WAC9
Related	ENSP00000250971.3, ENST00000250971.7

Conserved Domains (1) summary

	cd04367 **Location:26 → 110**	IIGF_insulin_like; IIGF_like family, insulin_like subgroup, specific to vertebrates. Members include a number of peptides including insulin and insulin-like growth factors I and II, which play a variety of roles in controlling processes such as metabolism, growth and ...

3. **NM_001185098.2 → NP_001172027.1 insulin preproprotein**

   See identical proteins and their annotated locations for NP_001172027.1

   Status: REVIEWED

Description	Transcript Variant: This variant (3) differs in the 5' UTR, compared to variant 1. All variants encode the same protein.
Source sequence(s)	AC132217, BM510347, BP322143
Consensus CDS	CCDS7729.1
UniProtKB/Swiss-Prot	P01308, Q5EEX2
UniProtKB/TrEMBL	I3WAC9
Related	ENSP00000380432.1, ENST00000397262.5

Conserved Domains (1) summary

cd04367 **Location:26 → 110**	IIGF_insulin_like; IIGF_like family, insulin_like subgroup, specific to vertebrates. Members include a number of peptides including insulin and insulin-like growth factors I and II, which play a variety of roles in controlling processes such as metabolism, growth and ...

4. **NM_001291897.2 → NP_001278826.1 insulin preproprotein**

   See identical proteins and their annotated locations for NP_001278826.1

   Status: REVIEWED

Description	Transcript Variant: This variant (4) differs in the 5' UTR, compared to variant 1. All variants encode the same protein.
Source sequence(s)	AC132217, BM510347
Consensus CDS	CCDS7729.1
UniProtKB/Swiss-Prot	P01308, Q5EEX2
UniProtKB/TrEMBL	I3WAC9

Conserved Domains (1) summary

cd04367 **Location:26 → 110**	IIGF_insulin_like; IIGF_like family, insulin_like subgroup, specific to vertebrates. Members include a number of peptides including insulin and insulin-like growth factors I and II, which play a variety of roles in controlling processes such as metabolism, growth and ...

RefSeqs of Annotated Genomes: Homo sapiens Annotation Release 110 details...

The following sections contain reference sequences that belong to a specific genome build. Explain

Reference GRCh38.p14 Primary Assembly

Genomic

1. **NC_000011.10 Reference GRCh38.p14 Primary Assembly**

Range	2159779..2161209 complement
Download	GenBank, FASTA, Sequence Viewer (Graphics)

Alternate T2T-CHM13v2.0

Genomic

1. **NC_060935.1 Alternate T2T-CHM13v2.0**

Range	2247427..2248857 complement
Download	GenBank, FASTA, Sequence Viewer (Graphics)

Related sequences

Items 1 - 25 of 26 < Prev Page 1 of 2 Next >

Nucleotide		Protein
Heading	Accession and Version	
genomic	AC132217.15 (86419..87850)	None
genomic	AH002844.2	AAA59172.1
genomic	AJ009655.1	CAA08766.1
genomic	CH471158.1	EAX02488.1
		EAX02489.1
genomic	CP068267.2 (2247427..2248857)	None
genomic	L15440.1	AAA59179.1
genomic	M10039.1	AAA59173.1
genomic	S99616.1	None
genomic	V00565.1	CAA23828.1
mRNA	AY899304.1	AAW83741.1
mRNA	BC005255.1	AAH05255.1
mRNA	BM510347.1	None
mRNA	BM510748.1	None
mRNA	BP322143.1	None
mRNA	BT006808.1	AAP35454.1
mRNA	DQ778082.1	ABI63346.1
mRNA	JF909299.1	AEG19452.1
mRNA	JQ951950.1	AFK93533.1
mRNA	MT335687.1	QMS45321.1
mRNA	MT335688.1	QMS45322.1
mRNA	MT335689.1	QMS45323.1
mRNA	MT335690.1	QMS45324.1
mRNA	MT335691.1	QMS45325.1
mRNA	MT335692.1	QMS45326.1

Nucleotide		Protein
Heading	Accession and Version	
mRNA	MT501209.1	None

Items 1 - 25 of 26 < Prev Page 1 of 2 Next >

Protein Accession	Links	
	GenPept Link	UniProtKB Link
P01308.1	GenPept	UniProtKB/Swiss-Prot:P01308

Additional links

Locus-specific Databases

INS database

Gene LinkOut