Molecular Biology Laboratory

Bioinformatics and Genomics Lab.

Week1. DNA / RNA Sequence Alignment (BLAST) & Genome Browser

- 1. Practice how to use UCSC Genome Browser
- Access to the UCSC Genome Browser and find human insulin gene
 - > Search "UCSC Genome Browser" in google and access to genome browser.



Hover the mouse pointer to "Genomes" and select genome version. We will use "Human GRC38/hg38" in our practical exercise.



> Type "INS" search window and select "Homo sapiens insulin".

UCSC Genome Bro	owser on Human (GRCh38/hg38)
move <<< < > >> >>> zoom	in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x
multi-region chr11:10,472,002-10,486,431 14,430 bp.	INS go examples
1 p14.3 p14.1 11p13 11p12 11p11.2 q12 ,000 10,476,000 5 kb	INS (Homo sapiens insulin (INS), transcript variant 2, mRNA. (from RefSeq NM_001185097)) INS-(GF2 (Homo sapiens INS-GF2 readthrough (INS-IGF2), transcript variant 2, mRNA. (from RefSeq NM_001042376)) INSC (Homo sapiens INSC spindle orientation adaptor protein (INSC), transcript variant 2, mRNA. (from RefSeq NM_001042376)) INSIG (Homo sapiens insulin induced gene 1 (INSIG1), transcript variant 1, mRNA. (from RefSeq NM_001042536)) INSIG (Homo sapiens insulin induced gene 1 (INSIG1), transcript variant 1, mRNA. (from RefSeq NM_00542)) INSIG (Homo sapiens insulin induced gene 2 (INSIG2), transcript variant 1, mRNA. (from RefSeq NM_00542)) INSIG (Homo sapiens insulin induced gene 2 (INSIG2), transcript variant 1, mRNA. (from RefSeq NM_016133)) INSIG (Homo sapiens insulin like 3 (INSL3), transcript variant 2, mRNA. (from RefSeq NM_005543)) INSIS (Homo sapiens insulin like 3 (INSL3), transcript variant 2, mRNA. (from RefSeq NM_005543)) INSIS (Homo sapiens insulin like 4 (INSL4), mRNA. (from RefSeq NM_002195)) INSIS (Homo sapiens insulin like 5 (INSL5), mRNA. (from RefSeq NM_005478)) INSIS (Homo sapiens insulin like 6 (INSIG) mRNA. (from RefSeq NM_005478))
	INSED (Homo sapiens insulin fine of (HSLD), IntRVA. (Holf HCB2W, WILDUT (19)) INSM1 (Homo sapiens INSMI transcriptional repressor 1 (INSM1), mRNA. (from RefSeq NM_002196)) INSM2 (Homo sapiens insulin receptor (INSR), transcript variant 2, mRNA. (from RefSeq NM_00129817)) INSRR (Homo sapiens insulin receptor related receptor (INSRR), mRNA. (from RefSeq NM_011215)) INSRYN1 (Homo sapiens inhibitory synaptic factor 1 (INSYN1), transcript variant 5, mRNA. (from RefSeq NM_001384353)) INSYN1 (Homo sapiens inhibitory synaptic factor 1 (INSYN1), transcript variant 2, long non-coding RNA. (from RefSeq NR_120353)) INSYN2 (Homo sapiens inhibitory synaptic factor 2A (INSYN2A), mRNA. (from RefSeq NM_001039762)) INSYN2B (Homo sapiens inhibitory synaptic factor family member 2B (INSYN2B), transcript variant 2, mRNA. (from RefSeq NM_001346304))
Gene Expression in 54 tis	INSYN1 (Homo sapiens inhibitory synaptic factor 1 (INSYN1), transcript variant 5, mRNA. (from RefSeq NM_001384353)) INSYN1-AS1 (Homo sapiens INSYN1 antisense RNA 1 (INSYN1-AS1), transcript variant 2, long non-coding RNA. (from RefSeq NR_120353)) INSYN2A (Homo sapiens inhibitory synaptic factor 2A (INSYN2A), mRNA. (from RefSeq NM_001039762)) INSYN2B (Homo sapiens inhibitory synaptic factor family member 2B (INSYN2B), transcript variant 2, mRNA. (from RefSeq NM_001346304))] sues from GTEx RNA-seq of 17382 samples, 948 donors (V8, Aug 2019)

> Compare Isoforms of the insulin gene and look through other information.

LICSC Conomo Browcor on Human (CBCh29/hg29)							
move <<< << > > >>> Zoom in 1.5x 3x 10x base Zoom out 1.5x 3x 10x base Zoom out 1.5x 3x 10x 100x							
multi-region Chr11:2,159,418-2,161,582 2,165 bp. gene, chromosome range, or other position, see examples go examples							
p15/211p15.1 p14.3 p14.1 11p13 11p12 11p11.2 p11p12 11p11.2 p11p13.1 p13.2 11p13.4 11p14.1 14.211p14.3 11p21 11p22.1 11p22.3 11p23.3 24.11 p24.2 24.3 11							
1 kb hg38 2,159,800 2,159,900 2,160,000 2,160,200 2,160,300 2,160,500 2,160,600 2,160,800 2,160,900 2,161,000 2,161,100 2,161,200 2,161,300 Reference Assembly Ritemate Haplotype Sequence Alignments GENCODE V41							
OMIM Allelic Variant Phenotypes							
Gene Expression in 54 tissues from GTEx RNA-seg of 17382 samples, 948 donors (V8, Aug 2019)							
INC							

- Make custom gene annotation and visualize
 - Click the marked insulin isoform, "ENST00000397262.5", and look through the information of the isoform.

Human Gene INS (ENS)	Human Gene INS (ENST00000397262.5) from GENCODE V41							
Description: Homo sapiens insulin (INS), transcript variant 4, mRNA. (from RefSeq NM_001291897) RefSeq Summary (NM_001185098): This gene encodes insulin, a peptide hormone that plays a vital role in the regulation of carbohydrate and lipid metabolism. Gencode Transcript: ENST00000397262.5 Gencode Gene: ENSG00000254647.7 Transcript (Including UTRs) Position: hg38 chr11:2,159,779-2,161,204 Size: 1,426 Total Exon Count: 2 Strand: - Coding Region Position: hg38 chr11:2,159,852-2,160,971 Size: 1,120 Coding Exon Count: 2								
Page Index Se	equence and Links	UniProtKB Comments	MalaCards	CTD	RNA-Seq Expression			
Microarray Expression RN	Microarray Expression RNA Structure Protein Structure Other Species GO Annotations mRNA Descriptions							
Pathways Oth	ther Names	GeneReviews	Methods					
Data last updated at UCS	C: 2022-05-14 09:5	57:26						

Open notepad and save it as "Custom_INS_Annotation.gtf". \succ



 \triangleright Fill the blank with reference to the below information.

- Cautions
 - ✓ Each row is arranged in ascending order based on the start position.
 - There is a difference of 1 between the end position of UTR and the end position of CDS. \checkmark
 - \checkmark Each column is separated by a tab, but gene id and transcript id are written in one column and they are separated by space.
- For uploading custom an annotation file to the genome browser, hover the mouse on "My \geq Data" and click "Custom Tracks".

Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	View	Help	About Us
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> Upload the custom annotation file and click "Submit".

Add Custom Tracks
clade Mammal v genome Human v assembly Dec. 2013 (GRCh38/hg38) v
Display your own data as custom annotation tracks in the browser. Data must be formatted in <u>bigBed</u> , <u>GTF</u> , <u>hic</u> , <u>interact</u> , <u>MAF</u> , <u>narrowPeak</u> , <u>Personal Genome SNP</u> , <u>PSL</u> , or <u>WIG</u> formats.
 You can paste just the URL to the file, without a "track" line, for bigBed, bigWig, bigGenePred, BAN To configure the display, set <u>track</u> and <u>browser</u> line attributes as described in the <u>User's Guide</u>. Examples are <u>here</u>. If you do not have web-accessible data storage available, please see the <u>Hosting</u>
Please note a much more efficient way to load data is to use <u>Track Hubs</u> , which are loaded from the <u>T</u>
Paste URLs or data: Or upload: 파일선택 Week1_Custon_Answer.gtf Submit
Clear

> After uploading, click "go" to visualize the custom annotation track.

Manage Cu	Manage Custom Tracks								
genome: Human assembly: Dec. 2013 (GRCh38/hg38) [hg38]									
Name	Description	Type Doc	Items	Pos	delete	view in	Genome Browser	•	go
User Track	User Supplied Track	gtf	1	<u>chr11:</u>			add custom tracks		
 Compare the custom annotation track and marked Insulin isoform. 									
	UCSC G	enome Brows	er on Hu	man (GR	Ch38/hq3	8)			

move <<< << > >>>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x							
multi-region chr11:2,159,421-2,161,561 2,141 bp. gene, chromosome range, or other position, see examples go examples							
b15.2 11p15.1 p14.3 p14.1 11p13 11p12 11p11.2 q12.1 113.1 13.2 11q13.4 11q14.1 14.2 11q14.8 11q21 11q22.1 11q22.3 11q23.3 24.1	q24.2						
1 kb 159,800 2,159,900 2,160,000 2,160,100 2,160,200 2,160,300 2,160,400 2,160,500 2,160,600 2,160,700 2,160,800 2,160,900 2,161,000 2,161,100 2,160,100 2,160,100 2,160,100 2,160,100 2,160,100 2,160,100 2,160,100 2,160,100 2,160,100 2,161,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,100 2,10	200 2						
Reference Assembly Fix Patch Sequence Alignments Reference Assembly Alternate Haplotype Sequence Alignments GENCODE V41							

2. Practice how to get sequence data and use BLASTN

- Find GFP gene sequence and align the sequence
 - > Search "NCBI" in google and access NCBI.

Google	ncbi	x 🔳 🌢 Q	
	Q 전체 ⊑ 이미지 ▮ 도서 ▶ 동영상 ⊘ 쇼핑 : 더보기	도구	
	검색결과 약 608,000,000개 (0.52초)		
	https://www.ncbi.nlm.nih.gov National Center for Biotechnology Information Welcome to NCBI. The National Center for Biotechnology Information adva health by providing access to biomedical and genomic information. BLAST BLAST finds regions of similarity between biological sequences.	미국 국립생물공 학정보센터 ^{회사}	
	PubMed PubMed® comprises more than 34 million citations for biomedical		미국 국립생물공학정보센터는 미국 보건성 산하 국립 의학도서관의 운영 분야 중 하나이다. NCBI는 1988년 미국 메릴랜드주에 설립되었다. 위키백과
	The Nucleotide database is a collection of sequences from		장립사: 글로느 페퍼 창립: 1988년 11월 4일
	Gene The .gov means it's official. Federal government websites often end		본사: 미국 메릴랜드 베서스다 모회사: 미국 국립 의학 도서관

Search "GFP Aequorea victoria" and click "Nucleotide".

Search NCBI	GFP Aequore	a victoria	X Sea	arch	
Results found in 13 databases					
Literature		Genes		Proteins	
Bookshelf	11	Gene	0	Conserved Domains	0
MeSH	0	GEO DataSets	56	Identical Protein Groups	6
NLM Catalog	2	GEO Profiles	0	Protein	715
PubMed	439	HomoloGene	0	Protein Family Models	0
PubMed Central	2,358	PopSet	2	Structure	307
Genomes		Clinical		PubChem	
Assembly	0	ClinicalTrials.gov	0	BioAssays	0
BioCollections	0	ClinVar	0	Compounds	0
BioProject	0	dbGaP	0	Pathways	0
BioSample	8	dbSNP	0	Substances	1
Genome	0	dbVar	0		
Nucleotide	146	GTR	0		
SRA	8	MedGen	0		
Taxonomy	0	OMIM	0		

Select one from the list. We will use 4th one, "Aequorea victoria green-fluorescent protein (GFP) mRNA, complete cds" with 922 bp linear mRNA in a practical exercise.

	<< First < Prev Page 1 of 8 Next > Last >>
	Synthetic construct for Aequorea victoria partial gfp gene for GFP
1.	735 bp linear other-genetic
	Accession: LN515608.1 GI: 690969141
	PubMed Taxonomy
	GenBank FASTA Graphics
	Aeguorea victoria clone EM1 GFP gene, partial cds
2.	135 bp linear DNA
	Accession: JN800726.1 GI: 353442080
	Protein Taxonomy
	GenBank FASTA Graphics
	A severe with the inclusion of ED 000 second fluence on the table (rfn) second particlised
3	Aequorea victoria isolate SGFP-206 green nuorescent protein (grp) gene, partial cus
σ.	Accession: 1X472005.1 GI: 506054254
	Protein PubMed Taxonomy
	GenBank FASTA Graphics PopSet
	Aequorea victoria green-fluorescent protein (GFP) mRNA, complete cds
4.	922 bp linear mRNA
	Accession: L29345.1 GI: 606383
	Protein PubMed laxonomy
	GenBank FASTA Graphics

Click "FASTA" to get the gene sequence of GFP.

AUTHORS

PUBMED

8082767

TITLE JOURNAL Inouye,S. and Tsuji,F.I.

FEBS Lett. 351 (2), 211-214 (1994)

Aequorea victoria green-fluorescent protein (GFP) mRNA, complete cds GenBank: L29345.1 FASTA Graphics Go to: 🖂 LOCUS AEVGFP 922 bp mBNA linear INV 30-DEC-1994 DEFINITION Aequorea victoria green-fluorescent protein (GFP) mRNA, complete cds. ACCESSI ON L29345 VERSION L29345.1 KEY#ORDS aequorin; bioluminescence; energy acceptor protein; green-fluorescent protein. SOURCE Aequorea victoria ORGANISM <u>Aequorea victoria</u> Eukaryota; Metazoa; Cnidaria; Hydrozoa; Hydroidolina; Leptothecata; Aequoreidae; Aequorea. REFERENCE 1 (bases 1 to 922) AUTHORS Inouye,S. and Tsuji,F.I. TITLE Aequorea green fluorescent protein. Expression of the gene and fluorescence characteristics of the recombinant protein JOURNAL FEBS Lett. 341 (2-3), 277-280 (1994) PUBMED 8137953 REFERENCE 2 (bases 1 to 922)

Evidence for redox forms of the Aequorea green fluorescent protein

Aequorea victoria green-fluorescent protein (GFP) mRNA, complete cds

GenBank: L29345.1

GenBank Graphics

 \triangleright

Search "BLAST" in google and access BLAST.



Basic Local Alignment Search Tool BLAST+ 2.13.0 is here! BLAST finds regions of similarity between biological sequences. The Starting with this release, we are including the program compares nucleotide or protein sequences to sequence NEW blastn_vdb and tblastn_vdb executables in the BLAST+ databases and calculates the statistical significance. Learn more distribution. Thu, 17 Mar 2022 12:00:00 EST More BLAST news Web BLAST blastx translated nucleotide > protein Protein BLAST Nucleotide BLAST tblastn protein translated nucleotide protein > protein ucleotide > nucleotide

Copy and paste the GFP sequence to the query sequence box, and set the job title. When pasting the sequence, we must paste the header part too. There are many options, but we will use the default setting in the practical experiment. After then, select the program "BLASTN" and click "BLAST" for alignment.

	BLAST [®] » blastn suite				
blastn bla	stp blastx tblastn tblastx Standard Nucleotide BLAST				
	BLASTN programs search nucleotide databases using a nucleotide query				
Enter Query S	equence				
Enter accession n	umber(s), gi(s), or FASTA sequence(s) 😮 clear 🗡 Query subrange 🖓				
>L29345.1 Aequorea complete cds TACACACGAATAAA TGGAGTTGTCCCA4	victoria green-fluorescent protein (GEP) mRNA, AGATAACAAAGATGAGTAAAGGAGAAGAACTTTTCAC T				
Or, upload file	파일 선택 선택된 파일 없음 🔺 📀				
Job Title	GFP BLAST				
	Enter a descriptive title for your BLAST search 😮				
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Choose Searc	h Sat				
Database					
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Optional	Enter organism name or id-completions will be suggested exclude Add organism				
	Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown 🤡				
Exclude Optional	☐ Models (XM/XP) ☐ Uncultured/environmental sample sequences				
Limit to	Sequences from type material				
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Optional	Enter an Entrez query to limit search ?				
Program Sele	ction				
Optimize for	O Highly similar sequences (megablast)				
	More dissimilar sequences (discontiguous megablast)				
	Somewhat similar sequences (blastn)				
	Choose a BLAST algorithm 😢				
BLAST	Search database Nucleotide collection (nr/nt) using Blastn (Optimize for somewhat similar sequences)				
	Show results in a new window				

We can see the alignment result. There are many similar sequences in the database, but the first one is the best-matched sequence. Select it and see "Graphic Summary" and "Alignments".

Job Title	GFP BLAST	Filter Results
RID	HD72483P013 Search expires on 09-07 09:03 am Download All V	
Program	BLASTN 😧 Citation 🗸	Organism only top 20 will appear exclude
Database	nt <u>See details</u> ✓	Type common name, binomial, taxid or group name
Query ID	Icl Query_20431	+ Add organism
Description	L29345.1 Aequorea victoria green-fluorescent protein (GF	Percent Identity E value Query Coverage
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Sequences	producing significant alignments	Download [×] Select columns [×] Show 100 ♥ €
select all	1 sequences selected	GenBank Graphics Distance tree of results MSA Viewer
	Description	Scientific Name Max Total Query Score Score Cover Value Idem Acc. Len Accession
Aequorea v	ictoria green-fluorescent protein (GFP) mRNA, complete cds	Aequorea victoria 1663 1663 100% 0.0 100.00% 922 L29345.1
Aequorea v	ictoria green-fluorescent protein mRNA, complete cds	Aequorea victoria 1558 1558 100% 0.0 97.51% 966 M62653.1
Synthetic co	onstruct from Aequorea victoria GFPS (gfps) mRNA, complete cds	synthetic construct 1498 1498 96% 0.0 97.21% 998 AF183395.1
Cloning vec	tor p6xHis-GFP, complete sequence	unidentified cloni 1494 1494 96% 0.0 97.09% 5271 U89936.1

Job Title	GEP BLAST	Filter Results
RID	HD72483P013 Search expires on 09-07 09:03 am Download All	
Program	BLASTN ? Citation >	Organism only top 20 will appear exclude
Database	nt See details ¥	Type common name, binomial, taxid or group name
		+ Add organism
Description	129345 1 Aeguorea victoria green-fluorescent protein (GE	Percent Identity E value Query Coverage
Molecule type	dna	
Query Length	922	
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Descriptions	Graphic Summary Alignments Taxonomy	
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	1 150	300 450 600 750 900
Descriptions	Graphic Summary Alignments Taxonomy	
Alignment viev	Pairwise CDS	feature 😯 Restore defaults Download ~
1 sequences sele	ctea 😗	
🕹 Downlo	ad v GenBank Graphics	▼ Next ▲ Previous ≪Descriptions
Aeguore		
Acquoie	a victoria green-fluorescent protein (GFP) mRNA, con	nplete cds
Sequence	a victoria green-fluorescent protein (GFP) mRNA, con D: <u>L29345.1</u> Length: 922 Number of Matches: 1	nplete cds
Sequence Range 1: 1	a victoria green-fluorescent protein (GFP) mRNA, con D: <u>L29345.1</u> Length: 922 Number of Matches: 1 to 922 <u>GenBank</u> <u>Graphics</u>	Next Match Previous Match
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Range 1: 1 Score 1663 bits(Query 1 Sbjct 1 Query 61 Sbjct 61 Query 12 Sbjct 12	a victoria green-fluorescent protein (GFP) mRNA, con D: <u>L29345.1</u> Length: 922 Number of Matches: 1 to 922 GenBank Graphics Expect Identifies Gaps 1844) 0.0 922/922(100%) 0/922(0%) TACACACGAATAAAAGATAACAAAGATGAGTAAAGGAGAAGAACTITICACTGGAGT 111111111111111111111111111111111111	Implete cds Strand Previous Match Strand Plus/Plus TGT 60 111 TGT AGA 120 GAA 180
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- Find insulin gene sequences of human and pig, and compare the two sequences
 - Search "human insulin" in NCBI.

Search NCBI	Human insulin	×	Searc	h		
Results found in 29 databases						
GENE INS - insulin Homo services (human)		Was this helpful?		RefSeq trans	cripts ranscripts	
Also known as: IDDM, IDDM1, IDDM2, ILPR, IRE Gene ID: 3630	DN, MODY10, PNDM4			Transcript	Isoform	Len (nt)
RefSeq transcripts (4) RefSeq proteins (4)	RefSeqGene (2) PubMed (970)			NM_001185097.2		403
BefSen Sequences	BLASI			NM_001291897.2 View full table		525 NCBI Datasets
Reisey sequences		т				

> Go below and click "FASTA" to get the gene sequence of INS.

A Canomic regions transportints and products	A 2
- Genome regions, nanscripts, and products	Go to reference seguence détails
Genomic Sequence: NC 000011.10 Chromosome 11 Reference GRCh38.p14 Primary Assembly 💙	
	Go to nucleotide: Graphics FASTA GenBank
SNC_000011.10 · Find:	🗙 Tools 🗸 🕸 Tracks 🔹 📩 Download 👻 🤶 🗸
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NH_DGUIDSD982 >>>>>>>>>>>>>>>>>>>>>>>>>>>>>>>>>>>>	- > > NP_0011720271
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NCBL Reference Sequence: NC: 000011-10	
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GGGCTCAGGATTCCAGGGTGGCTGGACCCCAGGCCCCAGCTCTGCAGCAGGAGGACGTGGCTGGGCTCG	
TGAAGCATGTGGGGGTGAGCCCAGGGGCCCCAAGGCAGGGCACCTGGCCTTCAGCCTGCCT	
CTGTCTCCCAGATCACTGTCCTTCTGCCATGGCCCTGTGGATGCGCCTCCTGCCCCTGCTGGCGCTGCTG	
GCCCTCTGGGGACCTGACCCAGCCGCAGCCTTTGTGAACCAACACCTGTGCGGCTCACACCTGGTGGAAG	
CTCTCTACCTAGTGTGCGGGGAACGAGGCTTCTTCTACACACCCCAAGACCCGCCGGGAGGCAGAGGACCT	
GCAGGGTGAGCCAACTGCCCATTGCTGCCCCTGGCCGCCCCAGCCACCCCCTGCTCCTGGCGCTCCCAC	
CCAGCATGGGCAGAAGGGGGCAGGAGGCTGCCACCCAGCAGGGGGTCAGGTGCACTTTTTAAAAAGAAG	
TTCTCTTGGTCACGTCCTAAAAGTGACCAGCTCCCTGTGGCCCAGTCAGAATCTCAGCCTGAGGACGGTG	
TTGGCTTCGGCAGCCCCGAGATACATCAGAGGGTGGGCACGCTCCTCCCTC	
TGCCCCGCAGCCCATTTCTCCACCCTCATTTGATGACCGCAGATTCAAGTGTTTTGTTAAGTAAAGTCCT	
GGGT GACCT GGGGT CACAGGGT GCCCCACGCT GCCT GC	
GGGCGTGGCTGCCTGACTGGGCCAGACCCCTGTCGCCAGGCCTCACGGCAGCTCCATAGTCAGGAG	
ATGGGGAAGATGCTGGGGACAGGCCCTGGGGAGAAGTACTGGGATCACCTGTTCAGGCTCCCACTGTGAC	
GCTGCCCCGGGGCGGGGAAGGAGGTGGGACATGTGGGCGTTGGGGCCTGTAGGTCCACACCCAGTGTGG	
GT GACCCT CCT CT A A CCT GGGT CCA GCCCGGCT GGA GA T GGGT GGG	
AGGCGGGCACTGTGTCTCCCTGACTGTGTCCTCCTGTGTCCCTCTGCCTCGCCGCTGTTCCGGAACCTGC	
T CT GCGCGGCA CGT CCT GGCA GT GGGGCA GGT GGA GCT GGGCGGGGGCCCT GGT GCA GGCA G	
CCTTGGCCCTGGAGGGGTCCCTGCAGAAGCGTGGCATTGTGGAACAATGCTGTACCAGCATCTGCTCCCT	
CTACCAGETGGAGAACTACTGCAACTAGACGCAGECCGCAGGCAGECCCACACCCGECCGCCTCCTGCACC	
GAGAGAGATGGAATAAAGCCCTTGAACCAGC	

> Click "all" to see the orthologs of the INS gene.

INS insulin [Homo	o sapiens (human)]
Gene ID: 3630, updated on 2	18-Aug-2022
Summary	8 2
Official Symbol Official Full Name Primary source	INS provided by <u>HONC</u> insulin provided by <u>HONC</u> H <u>GNC.HGNC.6081</u>
See related Gene type RefSeq status	EnsembLENSG0000254647 MIM.176730: AllianceGenome:HGNC:6081 protein coding REVIEWED
Organism Lineage Also known as	Homo sapiens Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo 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 Orthologs	Restricted experision toward pancreas (RPKM 671.7) See more mouse all
NEW	Try the new Gene table Try the new Transcript table

> Click "INS" of pig and get gene sequence in the same way.

Species	Å.	Gene	.≜ ∀	Architecture	÷	aa	÷
Homo sapiens human		INS insulin				110	~
Mus musculus house mouse		Ins2 insulin II				110	~
Rattus norvegicus Norway rat		Ins2 insulin 2				119	~
Bos taurus cattle		INS insulin				221	~
Sus scrofa pig		INS insulin				175	~
Gallus gallus chicken		INS insulin		—		107	~

Sus scrofa isolate TJ Tabasco breed Duroc chromosome 2, Sscrofa11.1, whole genome shotgun sequence

NCBI Reference Sequence: NC_010444.4

GenBank Graphics
>NC_010444.4:c1498052-1496842 Sus scrofa isolate TJ Tabasco breed Duroc chromosome 2,
Sscrofall.1, whole genome shotgun sequence
TGGCCCTCAGGCCGCTCGTTAAGACTCTAATGACCTCAAGGCCCCCAGAGGCGCTGATGACCCACGGAGA
TGATCCCGCAGGCCTGGCAGCAGGGAAATGATCCAGAAAGTGCCACCTCAGCCCCCAGCCATCTGCCACC
CACCT GGA GGCCCT CA GGGGCCGGGGGCGGGGGGGGGG
CAGCCCTCTGGGACCAGCTGCGTTCCCAGGCCGCCGGCAAGCAGGTCTGTCCCCCTGGGCTCCCGTCAGC
TGGGTCTGGGCTGTCCTGCTGGGGCCAGGGCATCTCGGCAGGAGGACGTGGGCTCCTCTCTGGAGCCCT
T GGGGGGT GAGGCT GGT GGGGGCT GCAGGT GCCCCT GGCT GG
CACCCCCCCCCCTGCGGCCCTGCGCCCCCCCCCCCCCCC
CCCGGCCCAGGCCTTCGTGAACCAGCACCTGTGCGGCTCCCACCTGGTGGAGGCGCTGTACCTGGTGTGC
GGGGAGCGCGGCTTCTTCTACACGCCCAAGGCCCGTCGGGAGGCGGAGAACCCTCAGGGTGAGCCGAGGG
GGCGTCCCGGGAGCGGTCGGGGGAGTTTTTAAGGAGGAAATTGGTAAAAGTGACCAACTCCCTGGGAGCT
GAGCCCAGAGACACCCCTCCCACGCCCCGGTCCCAGAAAGCCCCCCTTCCCTCCC
GCGGCTCCAGGGAGGAATCTTACGGAGTCAAGGCCCGGGTGCCGCTGGTCTCCGAGTGACATGGCCGTGG
T GT CCCGT CT GCCGGCCCA CA T GCCCGT GGGAGA T GCCCCA T CCCCCT GGGAGGGGCCCCCGT GCCGGGC
AGGCAGCGGGAGGCCCAGGACCGGCGGCTGCCGGCTTCCACTCCAGGGTGGGCGGGGTGGGGGTGGC
TGTCTCTGTGTGACCGGCTCTCCCCGCAGCAGGTGCCGTGGAGCTGGGCGGAGGCCTGGGCGGCCTGCAG
GCCTGGCGCTGGAGGGCCCCCGCAGAAGCGTGGCATCGTGGAGCAGTGCTGCACCAGCATCTGTTCCC
TCTACCAGCTGGAGAACTACTGCAACTAGGCCGCCCCTGAGGGCGCCCTGCTGCTCCCCGCACCCCAAAAC
CCAATAAAGTCUTGAATGAGU

Go to BLASTN and check the "Align two or more sequences". Paste the human sequence in \triangleright the query sequence and paste the pig's sequence in the subject sequence. After then, select "blastn" for the program, and click "BLAST".

	Align Sequences Nucleotide Bl	AST
blastn bla	asto blastx tblastn tblastx	
	BLASTN programs search nucleotide subjects using a nu	cleotide query.
Enter Query S	Sequence	
Enter accession n	number(s), gi(s), or FASTA sequence(s) 😯 Clear 🗡 Query subrange 😯	
>NC_000011.10:c21	161209-2159779 Homo sapiens chromosome 11,	
AGCCCTCCAGGAC	ITY Assembly CAGGCTCAGAAGAAGGCCATCAAGCAAGGTCTGTT	
CCAAGGGCCTTTG	GCGTCAGGT To	
Or. upload file		
	비배월 전력 전력된 배월 없음 🖌 🤨	
Job Title	Human INS vs. Pig INS	
×	Enter a descriptive title for your BLAST search 🚱	
🗹 Align two or mo	nore sequences 😮	
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>NC_010444.4:c149	98052-1496842 Sus scrofa isolate TJ Tabasco breed	
TGGCCCTCAGGCC	22, <u>Sector 11</u> Anote genome storgen sequence	
GGCGCTGATGACC	CCACGGAGA To	
Or, upload file		
Program Sele	ection	
Optimize for	O Highly similar sequences (megablast)	
	More dissimilar sequences (discontiguous megablast)	
	Somewhat similar sequences (blastn)	
¥		
BLAST	Search nucleotide sequence using Blastn (Optimize for somewhat similar sequences)	
	Show results in a new window	

- We can see how similar human's and pig's insulin gene sequence from the result.

Job Title	Human INS vs. Pig INS	Filter Results
RID	HD8JZJPW114 Search expires on 09-07 09:29 am Download All V	Percent Identity E value Query Coverage
Program	Blast 2 sequences Citation ~	
Query ID	Icl Query_43979 (dna)	
Query Descr	NC_000011.10:c2161209-2159779 Homo sapiens chromo	Filter Reset
Query Length	1431	
Subject ID	Icl Query_43981 (dna)	
Subject Descr	NC_010444.4:c1498052-1496842 Sus scrofa isolate TJ T ϵ	
Subject Length	1211	
Other reports	MSA viewer 🔞	
Descriptions	Graphic Summary Alignments Dot Plot	
Sequences p	oroducing significant alignments	Download Y Select columns Y Show 100 ♥ 0
🗹 select all	1 sequences selected	Graphics MSA Viewer
	Description	Scientific Max Total Query E Per. Acc. Name Score Score Cover value Ident Len
NC_010444	4:c1498052-1496842 Sus scrofa isolate TJ Tabasco breed Duroc chromosome 2,	Sscrofa11.1, whole genom 250 476 31% 8e-70 83.83% 1211 Query_43981

nover to see the title 🖌 click to show alignments	
Alignment Scores <a> < 40 <a> 40 - 50 <a> 50 - 80 <a> 80 - 200	>= 200 🕜
1 sequences selected 🔞	
Distribution of the top 4 Blast Hits on 1 subject sequences	
Ouerv	
1 1 1 1 1 1 250 500 750 1000 1250	
	some 2.
Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4	some 2,
Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4 Range 1: 392 to 625 Graphics	some 2,
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Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4 Range 1: 392 to 625 Graphics Score Expect Identities Gaps Strand Previous Mat	some 2,
Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4 Range 1: 392 to 625 Graphics Score Expect Identities Gaps 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAGCCCTGCCTGTCTCCCCAGATCACTGTCCTTCTGCCATGGCCTGTGGATGCGCC 25	some 2,
Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4 Range 1: 392 to 625 Graphics Score Expect Identities Gaps Strand 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAGCCCTGCCTGCCTGCCCGGCCCGGTCCCCCGGGCCCTGTGGGATGGCGCC 25 Sign 2 GCCTCAACGCCGGCCCGGTCCCCCAGGTC-CTCACCCCCGGCCATGGCCCTGTGGACGCGCC	some 2,
Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4 Range 1: 392 to 625 Graphics Score Expect Identities Gaps Strand 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAGCCCTGCCTGTCTCCCAGATCACTGTCCTTCTGCCATGGCCTGTGGATGGGCC 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAGCCGTGCTGCTCTCCCAGATCACTGTCCTTCTGCCATGGCCTGTGGATGGGCC 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAAGCGCCGCTGCTCCCCAGATCACTGTCCTTCTGCCATGGCCTGTGGATGGGCC 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAAGCGCCCCGCTGCCCCCAGGTC-CTCACCCCCGCCATGGCCCTGTGGAAGCGCC 250 197 258 TCCTGCCCCTGCTGCTGCGCCAGGTC-CTCACCCCCGCCCCG	some 2,
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Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4 Range 1: 392 to 625 Graphics Score Expect Identities Gaps Strand 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAACGCCGGCCGGCCGGCCGGGCCAGGCCCTGTGGGATGGCCC Sbj ct 392 GCCTCAACGCCGGCCGGCCGGCCGGGGGGGGCCCTGGGGGGGCCCGGCCCGGGCCCGG	some 2,
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Sscrofa11.1, whole genome shotgun sequence Sequence ID: Query_43981 Length: 1211 Number of Matches: 4 Range 1: 392 to 625 Graphics Score Expect Identities Gaps Strand 250 bits(276) 8e-70 197/235(84%) 1/235(0%) Plus/Plus Query 198 GCCTCAAGCCCIGCCIGCTIGTCICCCAGATCACIGTCCITCIGCCATGGCCCIGIGGAAGGCGC 25 111111111111111111111111111111111111	some 2,