DIU Génétique et Reproduction

Banques de données et serveurs biomoléculaires et génétiques

18 Novembre 2022

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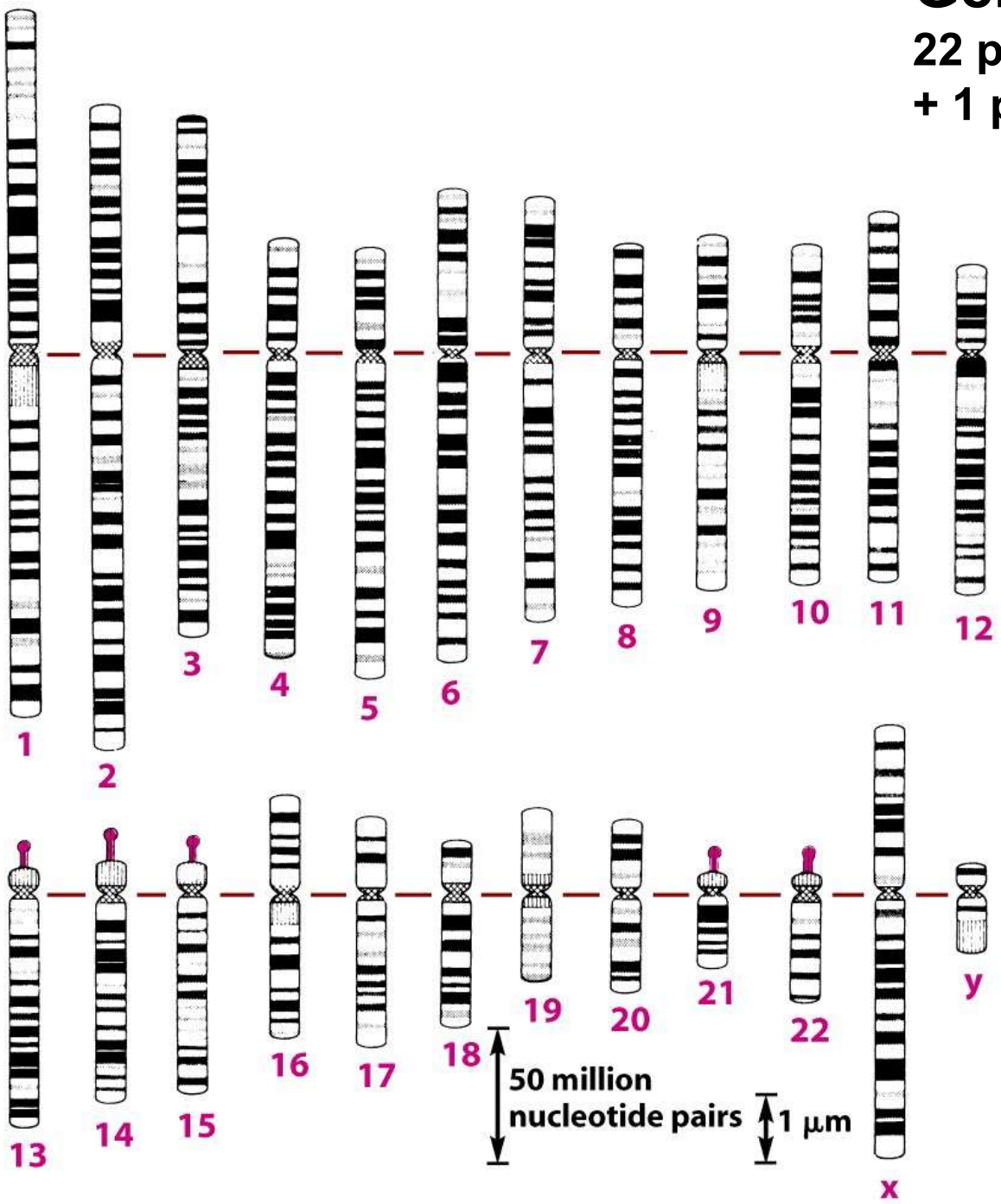
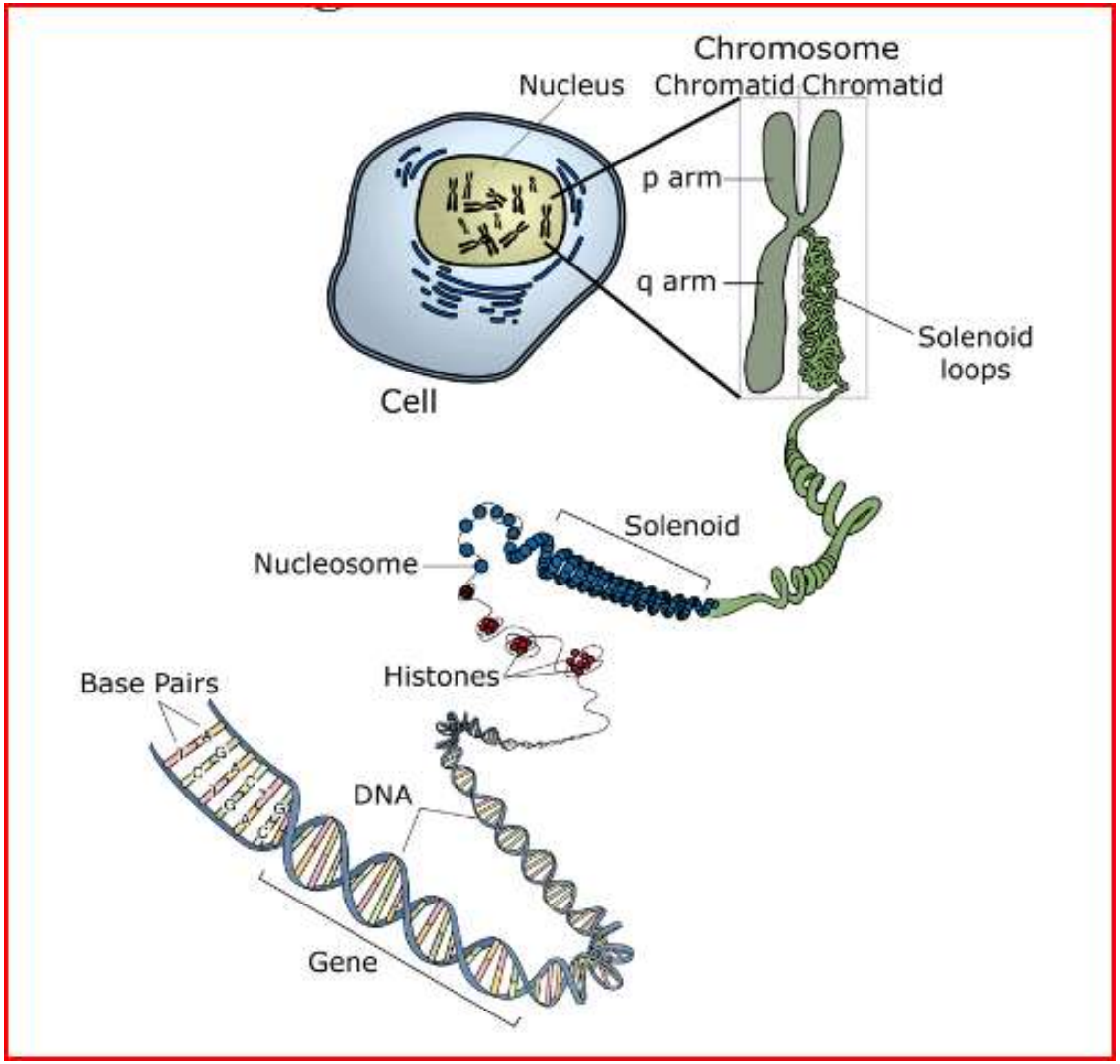
Génome humain

22 paires autosomes
+ 1 paire XX ou XY



Niveau d organisation de l'ADN dans un chromosome



Banques de séquences

Banques de séquences nucléotidiques EMBL, GenBank, RefSeq, Ensembl

séquences génomiques
séquences transcrites

Homme (Homo sapiens) et autres espèces (ex : Souris (Mus musculus)

Banques de séquences protéiques UNIPROT, SWISSPROT

déduction des séquences codantes par traduction (code génétique)
banque universelle : UNIPROT (non expertisée)
banque SWISSPROT (uniquement par expertise)

Banques de métadonnées

familles de gènes, domaines protéiques, facteur de transcription …

Banques génomiques

associées à la cartographie des gènes sur les génomes

Banques de polymorphisme et biomédicales

associées aux maladies (relations génotypes phénotypes)

Banques bibliographiques

D48–D50 Nucleic Acids Research, 2016, Vol. 44, Database issue *Published online 10 December 2015*
doi: 10.1093/nar/gkv1323

The International Nucleotide Sequence Database Collaboration

Guy Cochrane[1,*], Ilene Karsch-Mizrachi[2], Toshihisa Takagi[3] and International Nucleotide Sequence Database Collaboration

[1]European Molecular Biology Laboratory, European Bioinformatics Institute (EMBL-EBI), Wellcome Genome Campus, Hinxton, Cambridge CB10 1SD, UK, [2]National Center for Biotechnology Information, National Library of Medicine, National Institutes of Health, Bethesda, MD 20894, USA and [3]DDBJ Center, National Institute for Genetics, Mishima, Japan

The International Nucleotide Sequence Database Collaboration (INSDC; http://www.insdc.org) comprises three global partners committed to capturing, preserving and providing comprehensive public-domain nucleotide sequence information. The INSDC establishes standards, formats and protocols for data and metadata to make it easier for individuals and organisations to submit their nucleotide data reliably to public archives. This work enables the continuous, global exchange of information about living things. Here we present an update of the INSDC in 2015, including data growth and diversification, new standards and requirements by publishers for authors to submit their data to the public archives. The INSDC serves as a model for data sharing in the life sciences.

HIGH STANDARDS

The INSDC could not operate without the standardisation of all deposited data. The consortium's work in this area focuses on harmonising syntactical representation, supporting minimum information efforts and providing annotation style recommendations for consistency and clarity. Guidelines, data structures and systematic vocabularies developed by the INSDC include the Feature Table Definitions document (http://www.insdc.org/documents/feature-table), the INSDC country list (http://www.insdc.org/country.html) and conventions in the description of experimental support for annotated features (http://www.insdc.org/recommendations-vocabulary-insdc-experiment-qualifiers).

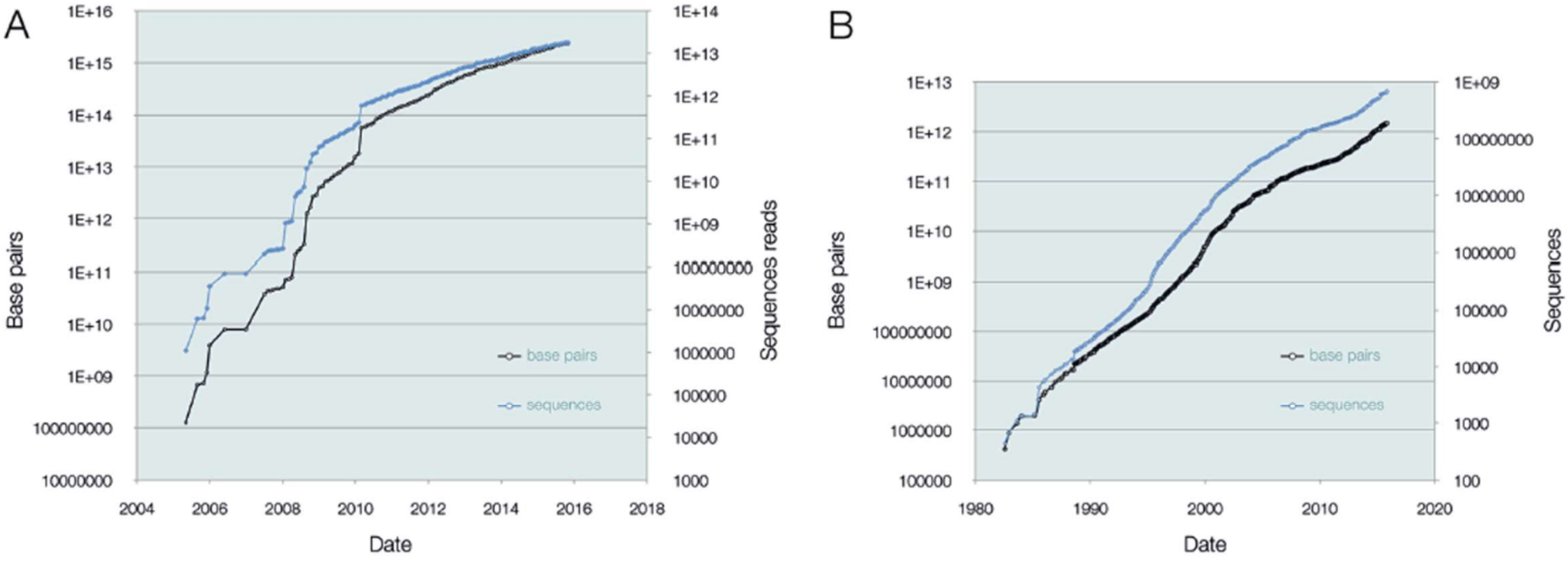
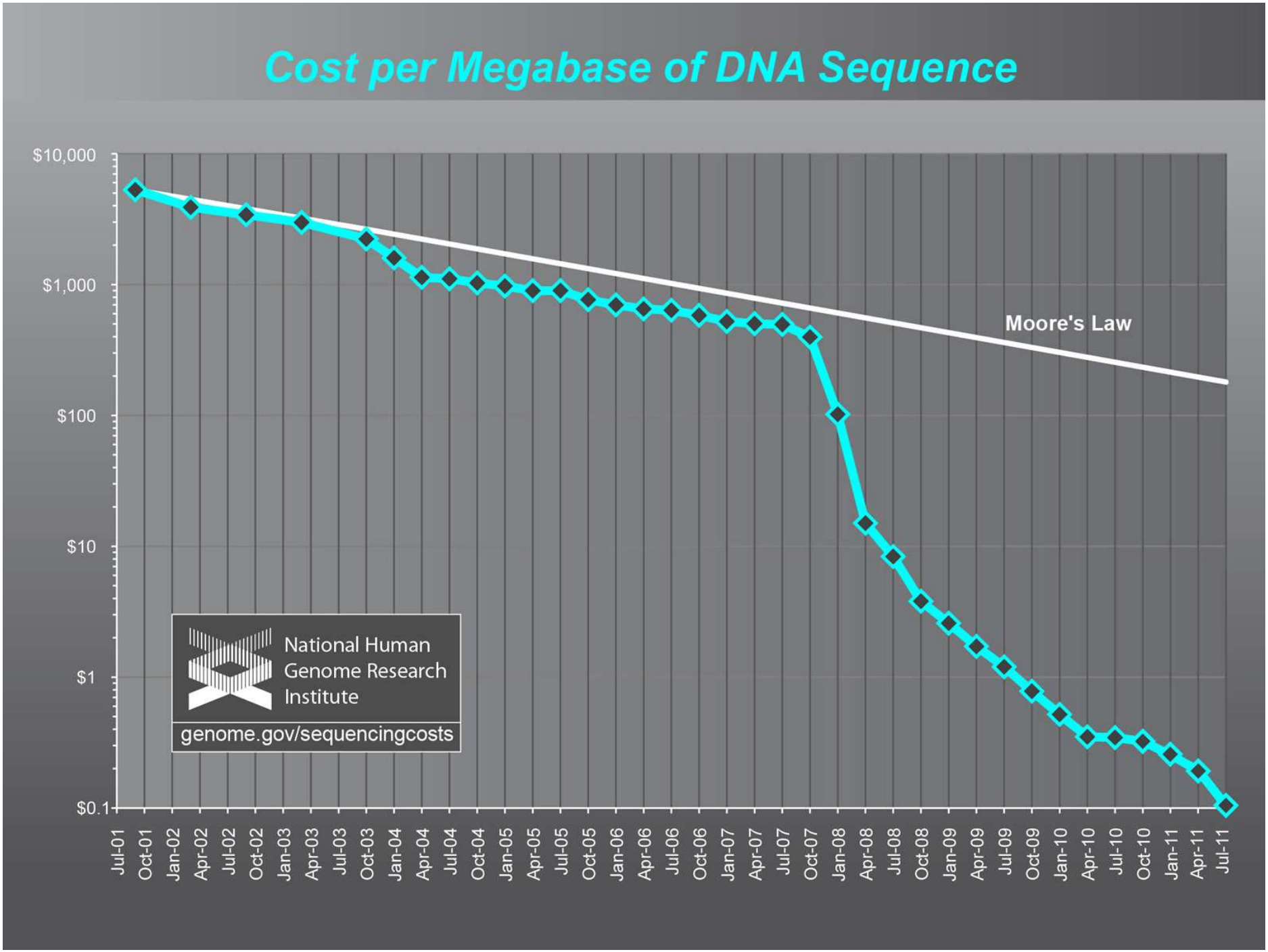


Figure 1. Cumulative growth in INSDC. (**A**) Base pairs (black, 2365.5 trillion) and sequence reads (blue, 17.8 trillion) for INSDC raw data. (**B**) Base pairs (black 1449 billion) and sequences (blue, 651.5 million) in INSDC assembled/annotated data.

Cost per Megabase of DNA Sequence



La complexité et les relations entre banques

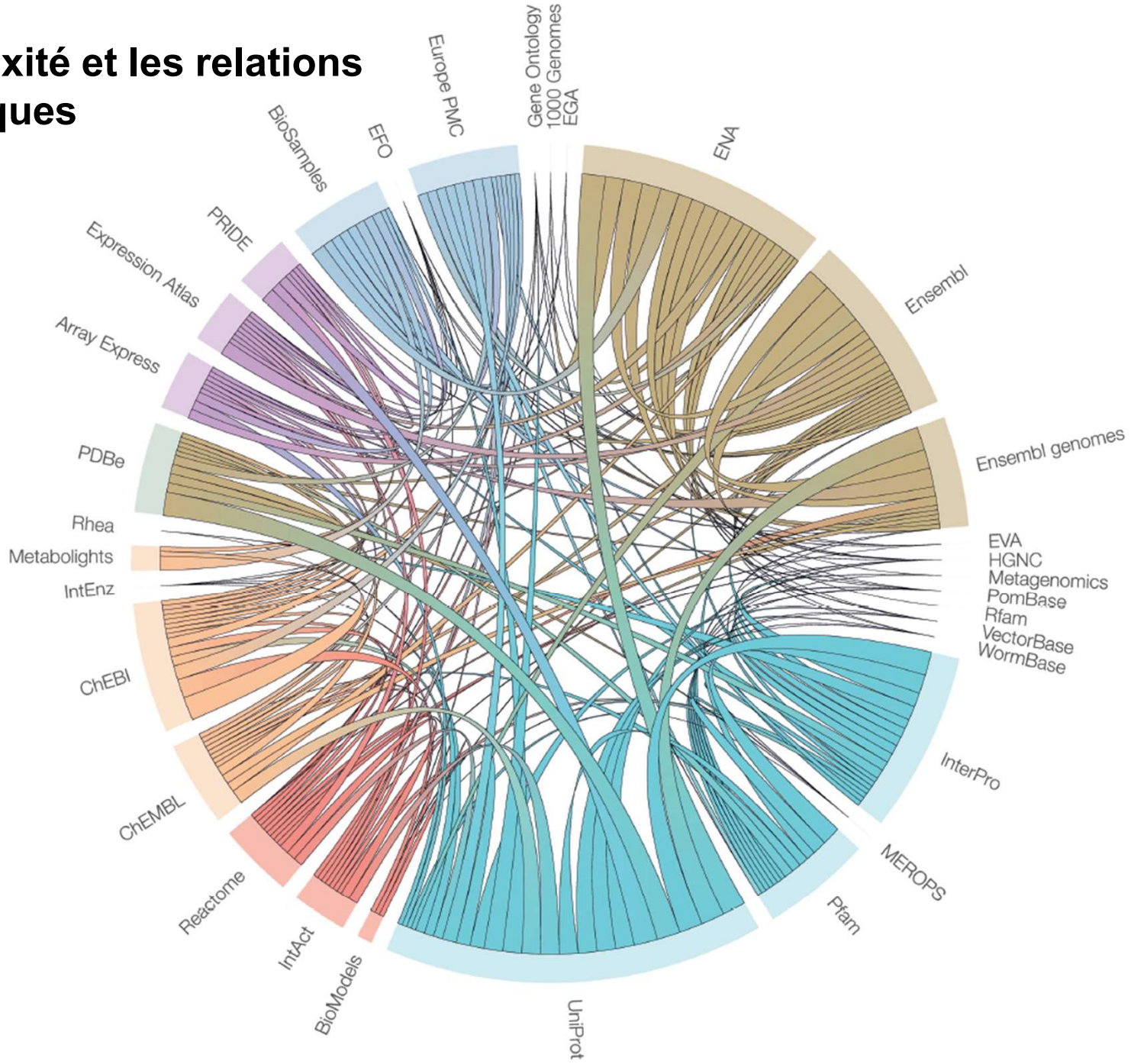


Figure 3. Representation of the internal interactions between different databases and resources at the EMBL-EBI, as determined by the exchange of data. All resources are placed on the circumference of the circle, with each resource represented by an arc proportional to the total number of interactions. The width of each internal arc, which transects the circle and connects two different resources, is weighted according to the number of different data types that are exchange between the two resources at the ends of the arc. The colouring of the internal arcs does not reflect the direction of data exchange. The graphic was generated using the D3 JavaScript library (http://d3js.org) and the data, gathered as part of an external review, were accurate at the time of acquisition

Ressources Internet et Données de génomes

French sites

Bioinfo_GR Gustave_Roussy_Github
Institut Francais de bioinformatique Plateformes Outils Donnees IBiSA ReNaBI AVIESAN France-Genomique Plan France Medecine genomique 2025
PBIL PBIL-IBCP
Genatlas Genoscope MicrosCope NGS-QC IMGT
Atlas of Genetics and Cytogenetics in Oncology and Haematology Atlas_Journal Atlas_links
Atlas of Genetics and Cytogenetics in Oncology and Haematology (old INIST release)
Institut Pasteur Galaxy (Pasteur)
RPBS (Mobyle)
Annotator - CIT

General sites

EMBNet ELIXIR
INSDC EBI Ensembl NCBI Entrez_gene Entrez_nuc Entrez_prot Entrez_GEO SRA GoldenPath_UCSC GDC_Cancer_Mutations GDC_Data_Analysis CGAP - NCI Sanger Center MSigDB COSMIC SRS DKFZ SRS NL HGNC Uniprot Expasy-SIB Pharos Harmonizome Bioinformatics.org Chipster SCIcrunch
Disqover GENCODE C CHESS DataMed Index CodeAlignView vizER IDDB The Telomere-to-Telomere (T2T) consortium UCSC assembly hub browser

Portals

GDC GDC Data Portal TARGET ICGC Data Portal CancerCoreEurope TCGA_cBIoPortal (Datasets) Broad Tumor Portal Firebrowse GDAC (Datasets) GTEx Portal Cancer_Imaging HPA TissGDB Integrative Onco Genomics (intOgen) OASIS Portal Cancer Browser (UCSC) UCSC_Xena UCSC Xena Browser
CancerResource canSAR BluePrint Portal genomicScape TCGA_Nature ENCODE_Dashboard_CRG GENIE_AACR GENIE_SageNet Pediatric_RNASeq_cancer
GnomAD browser Varsome UCSC_Variant_Integrator VEP PeCan (St Jude) PedPanCanc (DKFZ StJude_cloud Foundation_Medicine R2 Kidsfirstdrc Cancer Genomics Cloud MET500 Recount_mRNA AACR_GENIE cBIoPortal UALCAN France_Medecine_genomique2025 St-Jude-cloud Cohorts Regulome
Harmonizome Oncogenomic_landscape CancerTool Pedcbioportal ITCC-P4 OncoSG Synapse Cancer_genetic_commons VICC ALPHA Pediatric_genomic_data
HuVarBase

http://pdessen.free.fr/links.html

Foisonnement de logiciels et de ressources en bioinformatique

- Bio-Linux 7 (nov 2012) : plus de 500 logiciels

- Nucleic Acids Research Database Issue 2013 :
  1512 bases répertoriées par NAR

- Nucl. Acids Res. (1 January 2022) 50 (D1)
  https://academic.oup.com/nar/issue/50/1

EMBOSS Suite http://emboss.open-bio.org/

Wikipedia
https://en.wikipedia.org/wiki/List_of_open-source_bioinformatics_software

OMICS Tools http://omictools.com/

Bio_Tools https://bio.tools/

Bases de données de génomes

- Sommaire des BD NAR (Nucleic Acid Research)
- GOLD (Genome Online Database)
- NCBI Genomes
- EBI Genomes
- **UCSC (limité à des génomes modèles)**
- **enSembl (limité à des génomes modèles)**
- Sanger
- Genoscope

JGI GOLD GENOMES ONLINE DATABASE

JGI HOME LOG IN

Home Search Distribution Graphs Biogeographical Metadata Statistics References Team Help News

Studies	22 232
Biosamples	69 152
Sequencing Projects	69 468
Analysis Projects	57 258

Download Excel Data file
File last generated: 05 Oct, 2015

Welcome to the Genomes OnLine Database

GOLD Release v.5

GOLD:Genomes Online Database, is a World Wide Web resource for comprehensive access to information regarding genome and metagenome sequenci projects, and their associated metadata, around the world.

1. Register

GOLD Genomes Online Database

Register your project information and Metadata in the Genomes Online Database

Register

2. Annotate

Annotate your microbial genome or metagenome with IMG/ER or IMG/MER

Annotate

3. Publish

SIGS Standards in Genomic Sciences

Publish your genome or metagenome in open access standards-supportive journal.

Publish

Studies
- Metagenomic 604
- Non-Metagenomic 21 628

Biosamples
- Classification
- Ecosystems
  - Host-associated 12 732
  - Engineered 2 070
  - Environmental 8 516

Sequencing Projects
- Complete Projects 7 435
- Permanent Drafts 25 944
- Incomplete Projects 32 976
- Targeted Projects 2 122

Analysis Projects
- Genome Analysis 42 181
- Metagenome Analysis 6 145
- Single Cell (Screened) 1 379
- Single Cell (Unscreened) 214
- Genome from Metagenome 1 940
- Combined Assembly 79

JGI Projects
- JGI Studies 1 091
- JGI Biosamples 19 003
- JGI Sequencing Projects 27 805
- JGI Analysis Projects 19 586

Special Projects
- Type Strain Projects 4 826
- GEBA Projects 2 239
- HMP Projects 2 922

Organisms
- Organisms 64 487
  - Archaea 1 113
  - Bacteria 48 187
  - Eukarya 10 685
  - Viruses 4 469

Projects with Genbank Data
- Seq. Projects 35 493
  - Archaeal Projects 494
  - Bacterial Projects 29 326

Please cite:
Reddy TBK, Thomas A, Stamatis D, Bertsch J, Isbandi M, Jansson J, Mallajosyula J, Pagani I, Lobos E and Kyrpides N. The Genomes OnLine Database (GOLD) v.5: a metadata management system based on a four level (meta)genome project classification. *Nucl. Acids Res. (2014) doi: 10.1093/nar/gku950*
Full text

https://gold.jgi.doe.gov/

Nomenclature HGNC (gènes humains) (www.genenames.org)

http://www.genenames.org/

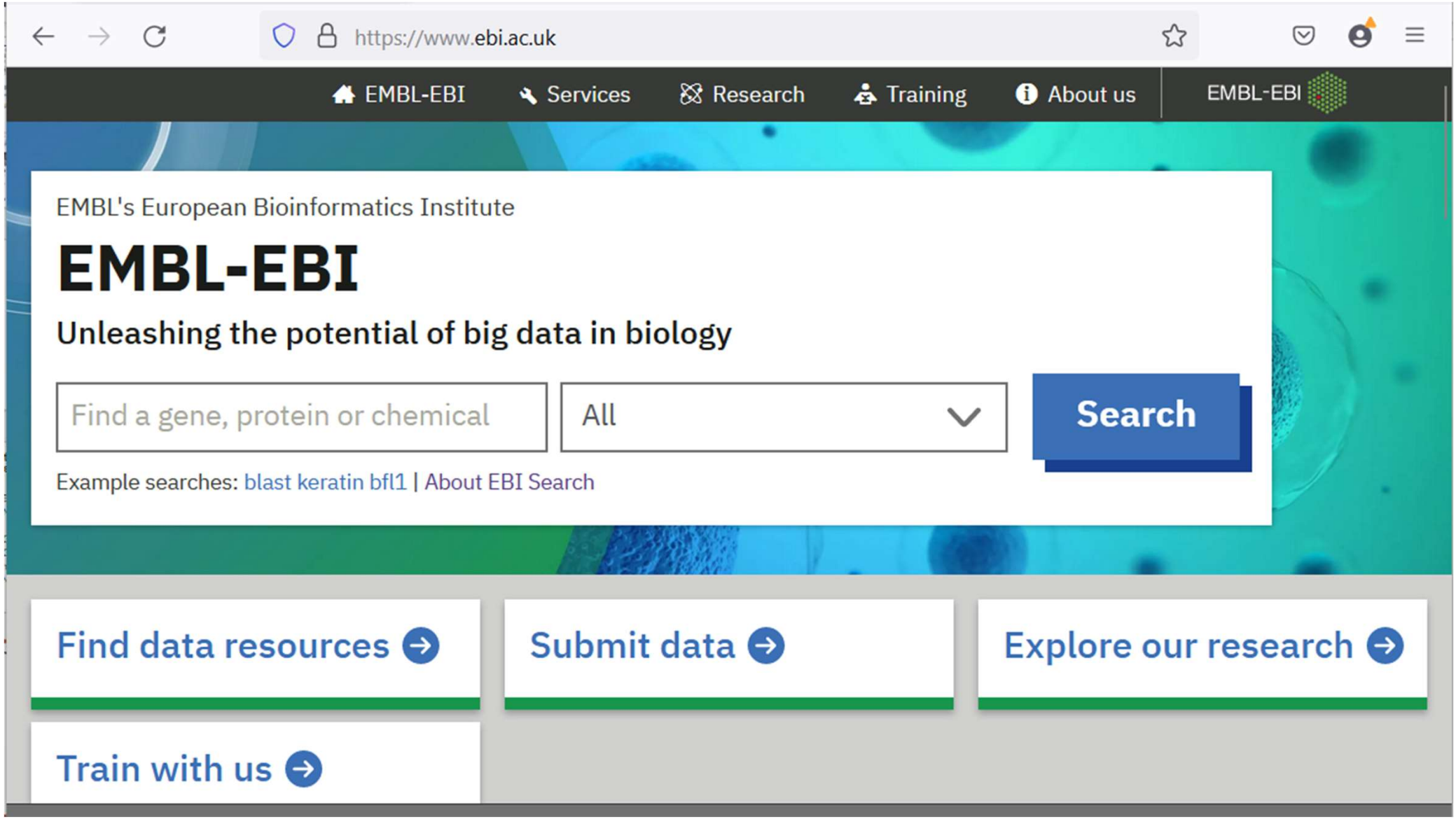
Gene Symbol Report

INSR

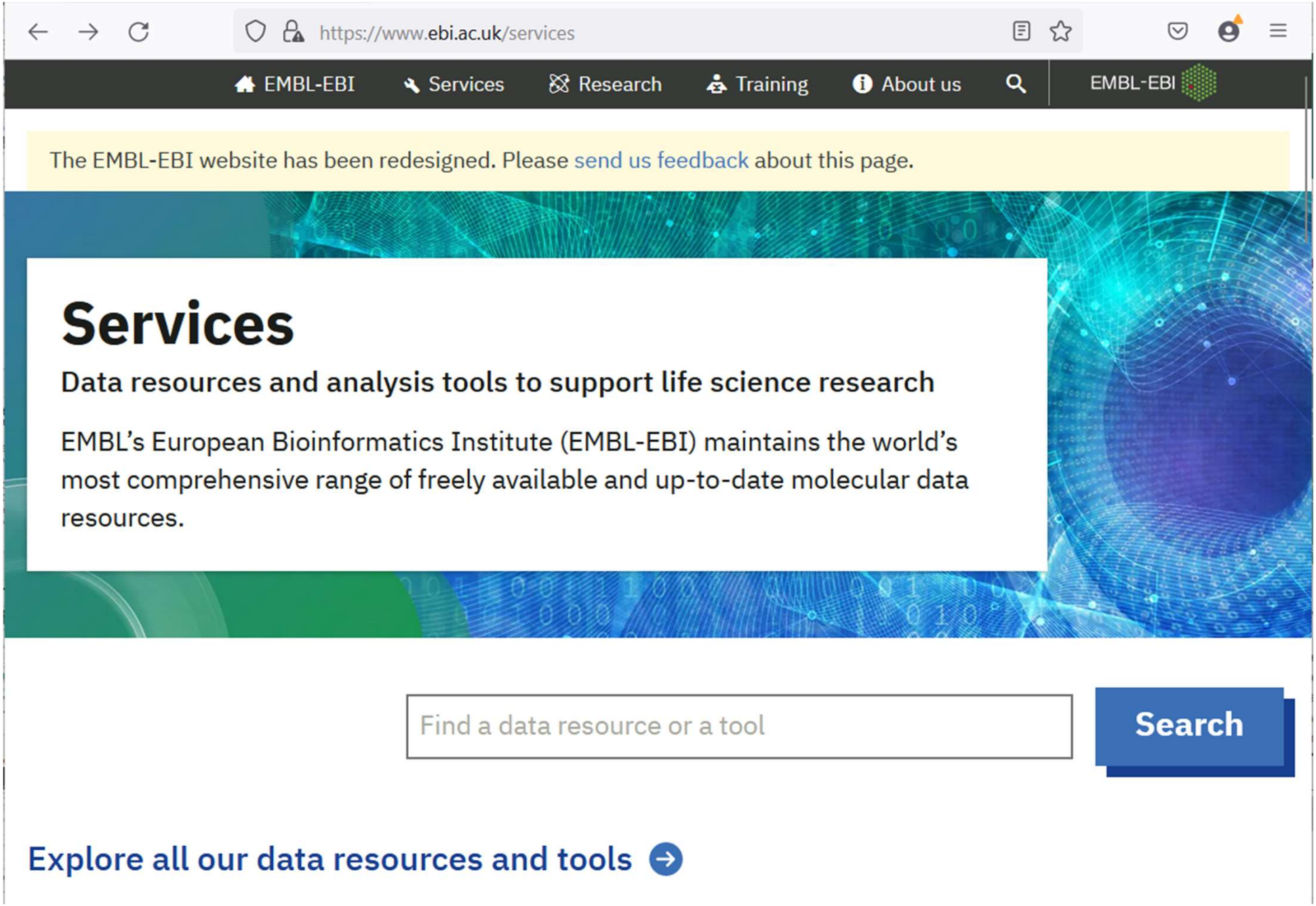
Approved Symbol	INSR
Approved Name	insulin receptor
HGNC ID	HGNC:6091
Previous Symbols & Names	-
Synonyms	CD220
Locus Type	gene with protein product
Chromosomal Location	19p13.3-p13.2

GENE FAMILY	CD molecules Fibronectin type III domain containing	
SPECIALIST DATABASE	CD C	
HOMOLOGS	MGI:96575 C Mouse Symbol: **Insr** RGD:2917 D Rat Symbol: **Insr** HCOP D TreeFam D	
NUCLEOTIDE SEQUENCES	GenBank:M10051 EMBL-Bank DDBJ C RefSeq:XM_005259552 D CCDS:CCDS12176.1 C	
GENE RESOURCES	Entrez Gene:3643 D Ensembl:ENSG00000171105 D UCSC:uc002mgd.1 D	NCBI Sequence Viewer Ensembl Genome Browser UCSC Genome Browser
PROTEIN RESOURCES	UniProtKB:P06213 D InterPro D	
	OMIM D GeneTests D	

European Bioinformatic Institute (EBI)



http://www.ebi.ac.uk/



http://www.ebi.ac.uk/services

https://www.ebi.ac.uk/services/data-resources-and-tools

Type

- [ ] Data resources
- [ ] Tools

Category

- [ ] Chemical biology
- [ ] Cross domain
- [x] DNA & RNA
- [ ] Gene expression
- [ ] Literature
- [ ] Ontologies
- [ ] Proteins
- [ ] Structures

Ensembl

Genome browser, API and database, providing access to reference genome annotation

DATA RESOURCE Web API | EMBL-EBI Terms of use

Ensembl Genomes

An Ensembl-style portal for the genomes of non-vertebrate species

DATA RESOURCE Web API | EMBL-EBI Terms of use

Clustal Omega

Multiple sequence alignment of DNA or protein sequences. Clustal Omega replaces the older ClustalW alignment tools.

https://www.ebi.ac.uk/services/data-resources-and-tools

ENA

A platform for the management, sharing, integration, archiving and dissemination of public-domain sequence data.

DATA RESOURCE Web API | EMBL-EBI Terms of use

International Genome Sample Resource

A deep catalog of shared human genetic variation in population groups worldwide that follows from the 1000 Genomes Project.

DATA RESOURCE EMBL-EBI Terms of use

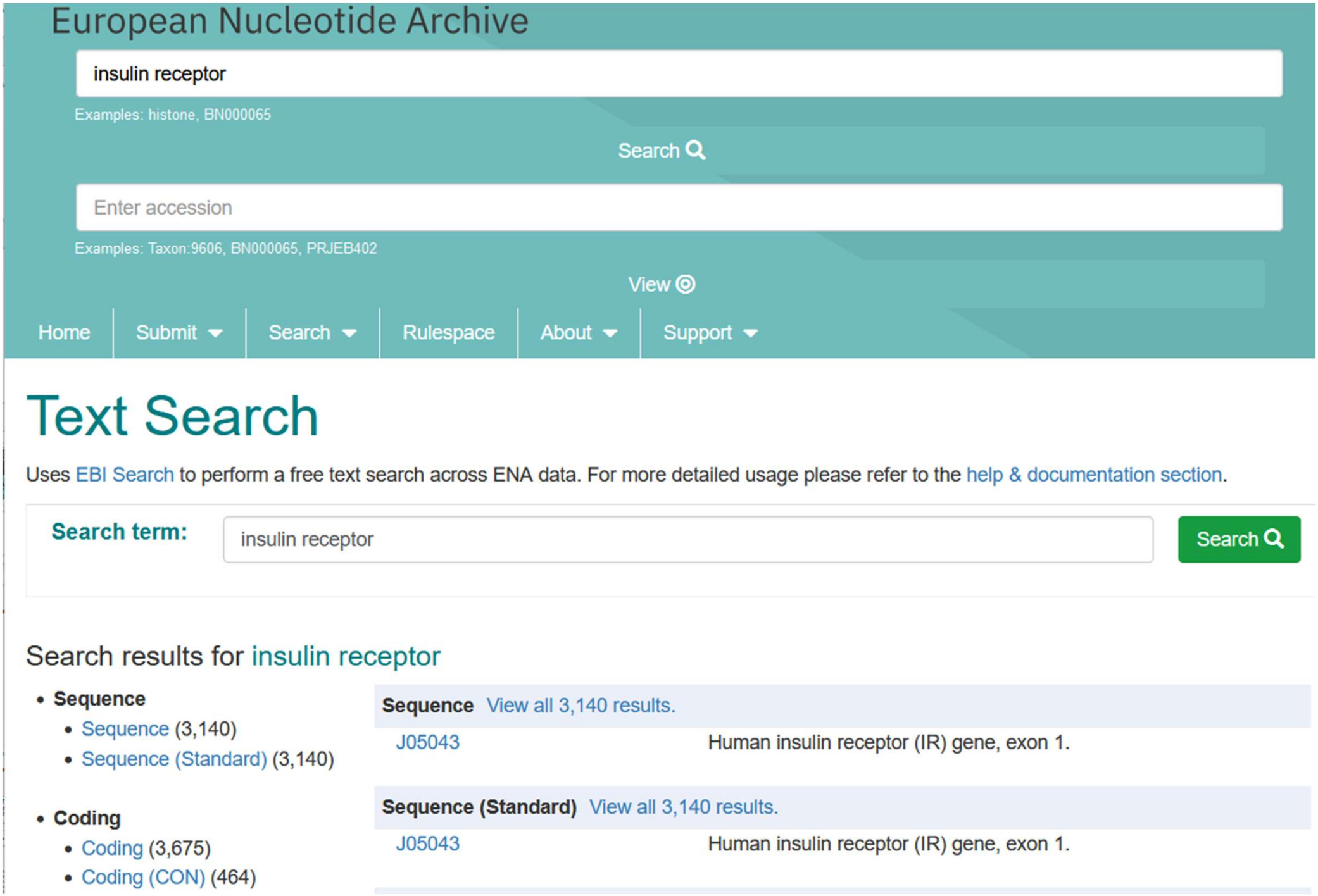
BLAST

Fast local similarity search tool for protein sequence databases.

TOOL Web API

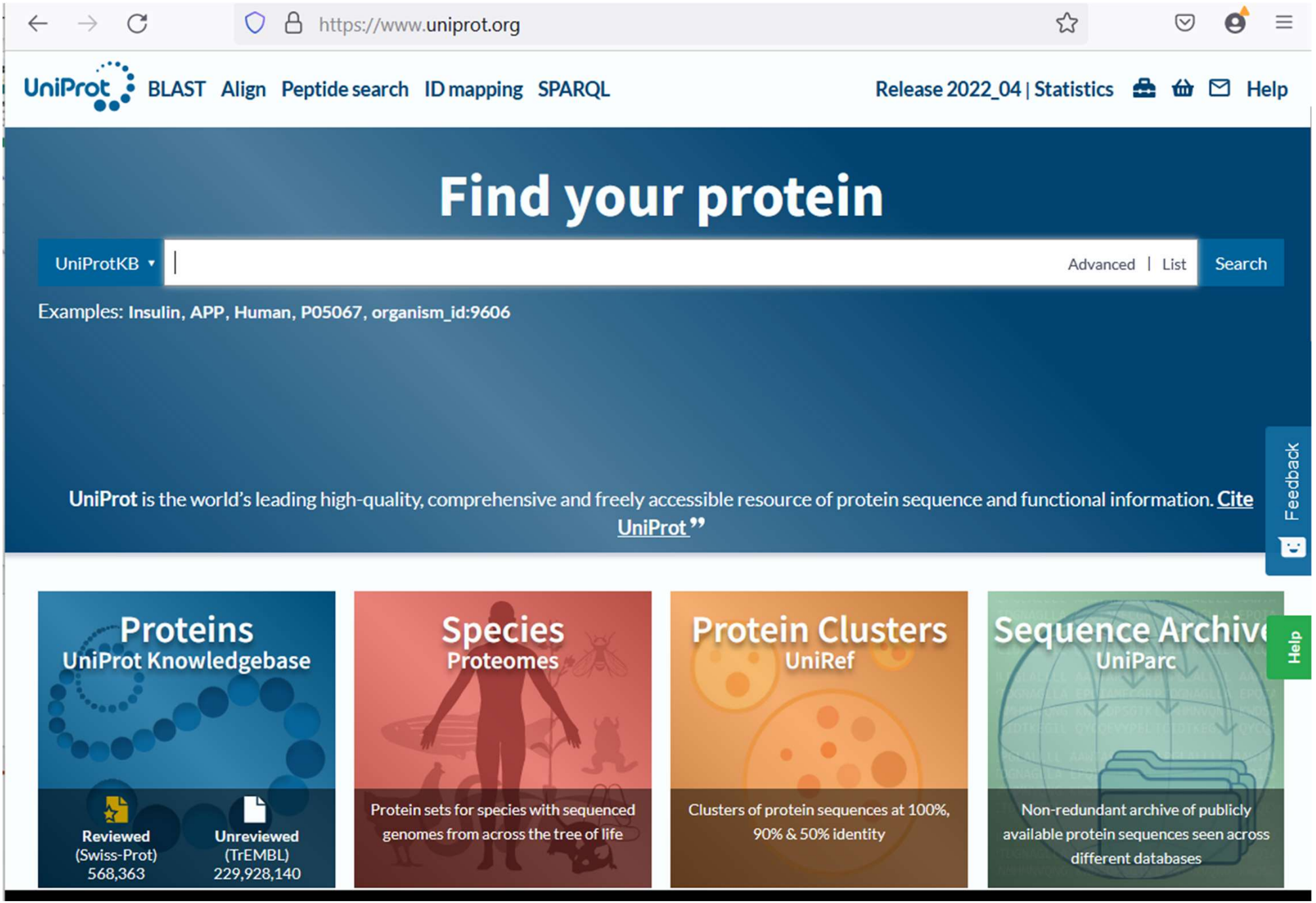
Tags: Sequence similarity search

https://www.ebi.ac.uk/ena/browser/home

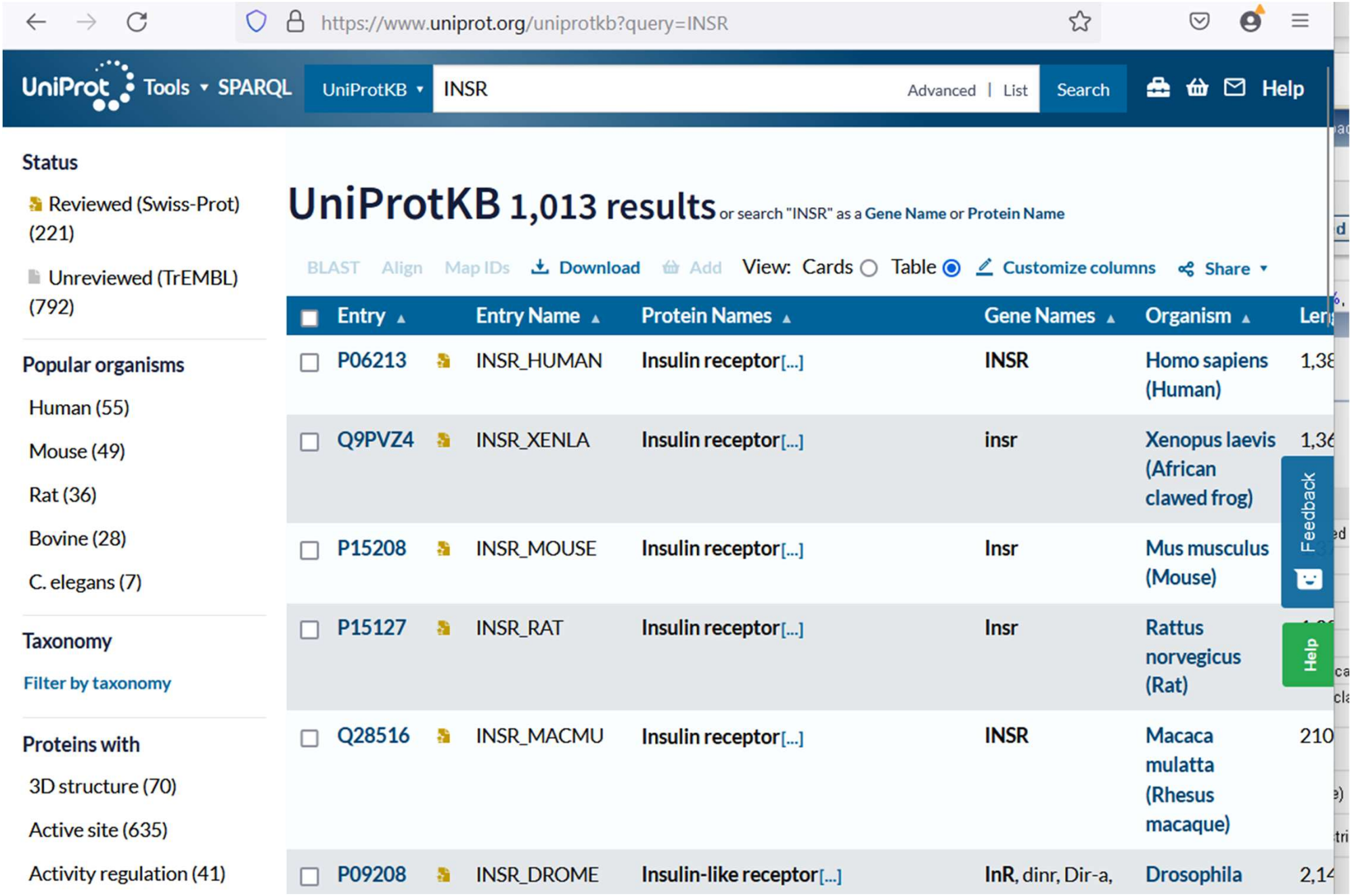


https://www.ebi.ac.uk/ena/browser/text-search?query=insulin%20receptor

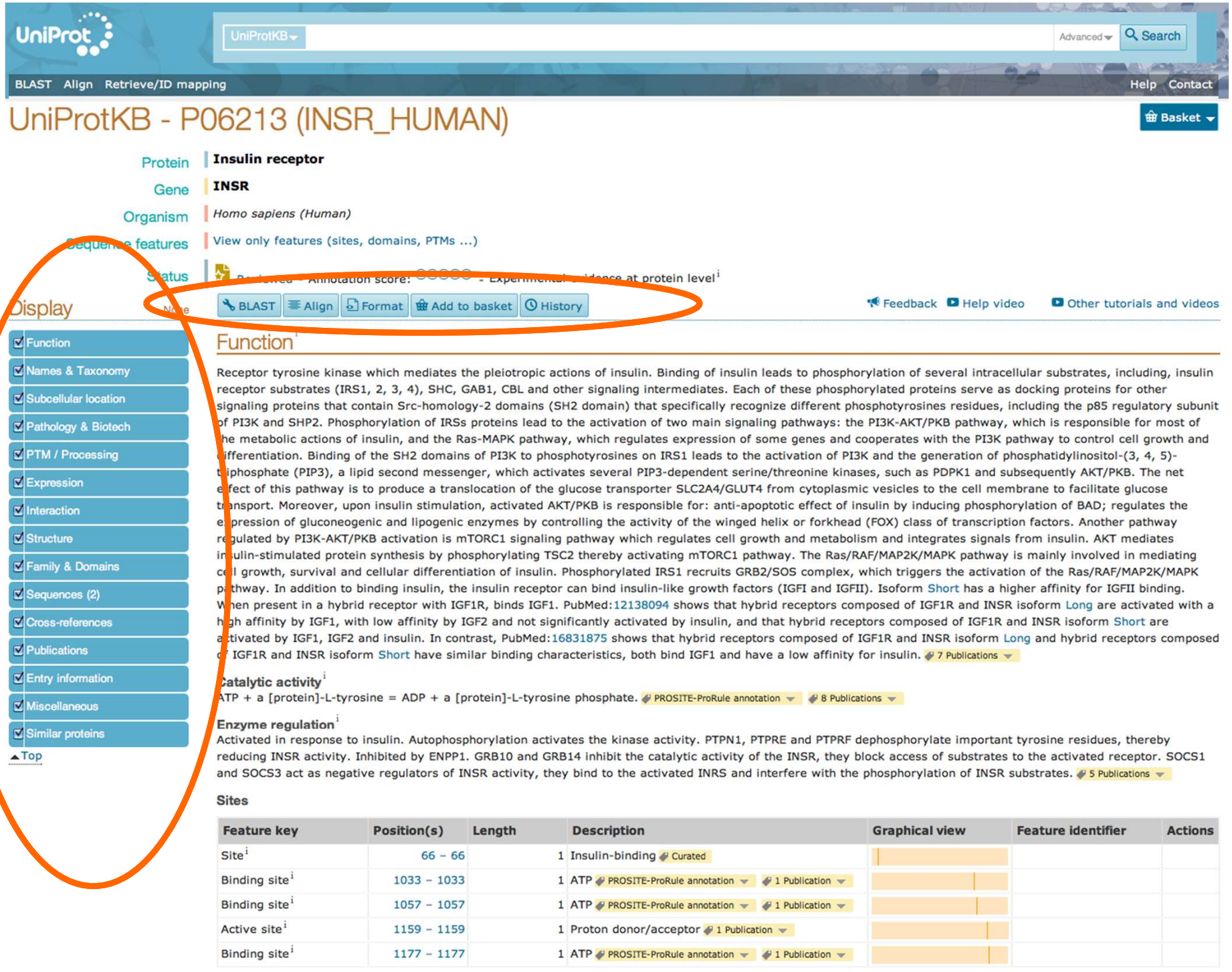
UniProt



http://www.uniprot.org/


https://www.uniprot.org/uniprotkb?query=INSR
UniProt
Tools
SPARQL
UniProtKB
INSR
Advanced | List
Search
Help
Status
Reviewed (Swiss-Prot) (221)
Unreviewed (TrEMBL) (792)
Popular organisms
Human (55)
Mouse (49)
Rat (36)
Bovine (28)
C. elegans (7)
Taxonomy
Filter by taxonomy
Proteins with
3D structure (70)
Active site (635)
Activity regulation (41)
UniProtKB 1,013 results or search "INSR" as a Gene Name or Protein Name
BLAST Align Map IDs Download Add View: Cards Table Customize columns Share
Entry
Entry Name
Protein Names
Gene Names
Organism
P06213 INSR_HUMAN Insulin receptor[...] INSR Homo sapiens (Human) 1,38
Q9PVZ4 INSR_XENLA Insulin receptor[...] insr Xenopus laevis (African clawed frog) 1,36
P15208 INSR_MOUSE Insulin receptor[...] Insr Mus musculus (Mouse)
P15127 INSR_RAT Insulin receptor[...] Insr Rattus norvegicus (Rat)
Q28516 INSR_MACMU Insulin receptor[...] INSR Macaca mulatta (Rhesus macaque) 210
P09208 INSR_DROME Insulin-like receptor[...] InR, dinr, Dir-a, Drosophila 2,14
Feedback
Help

http://www.uniprot.org/uniprot/P06213



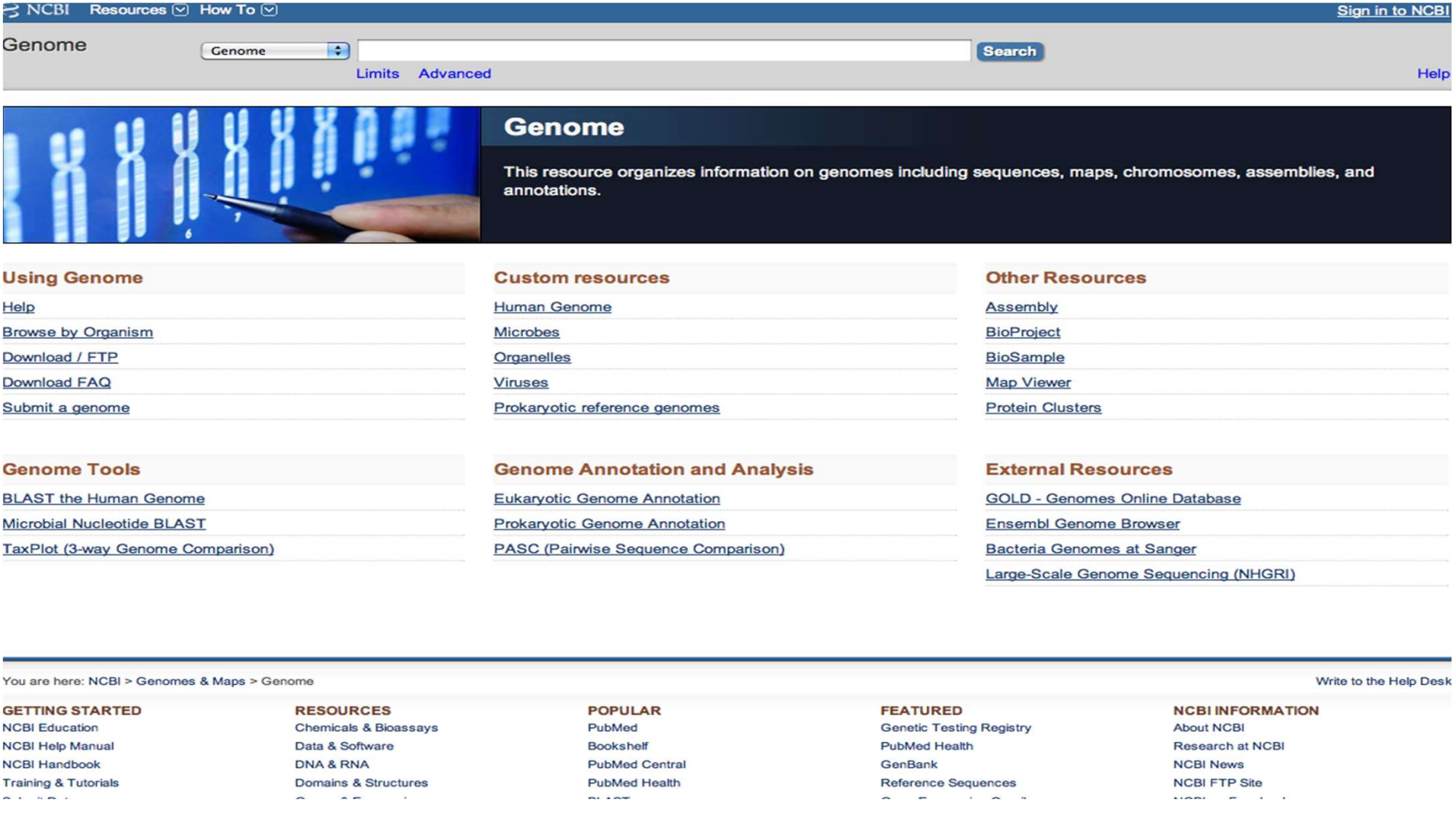
UNIPROT :
P06213 (INSR_HUMAN) Reviewed, UniProtKB/Swiss-Prot
Last modified September 18, 2013. Version 194

Sequence annotation (Features)

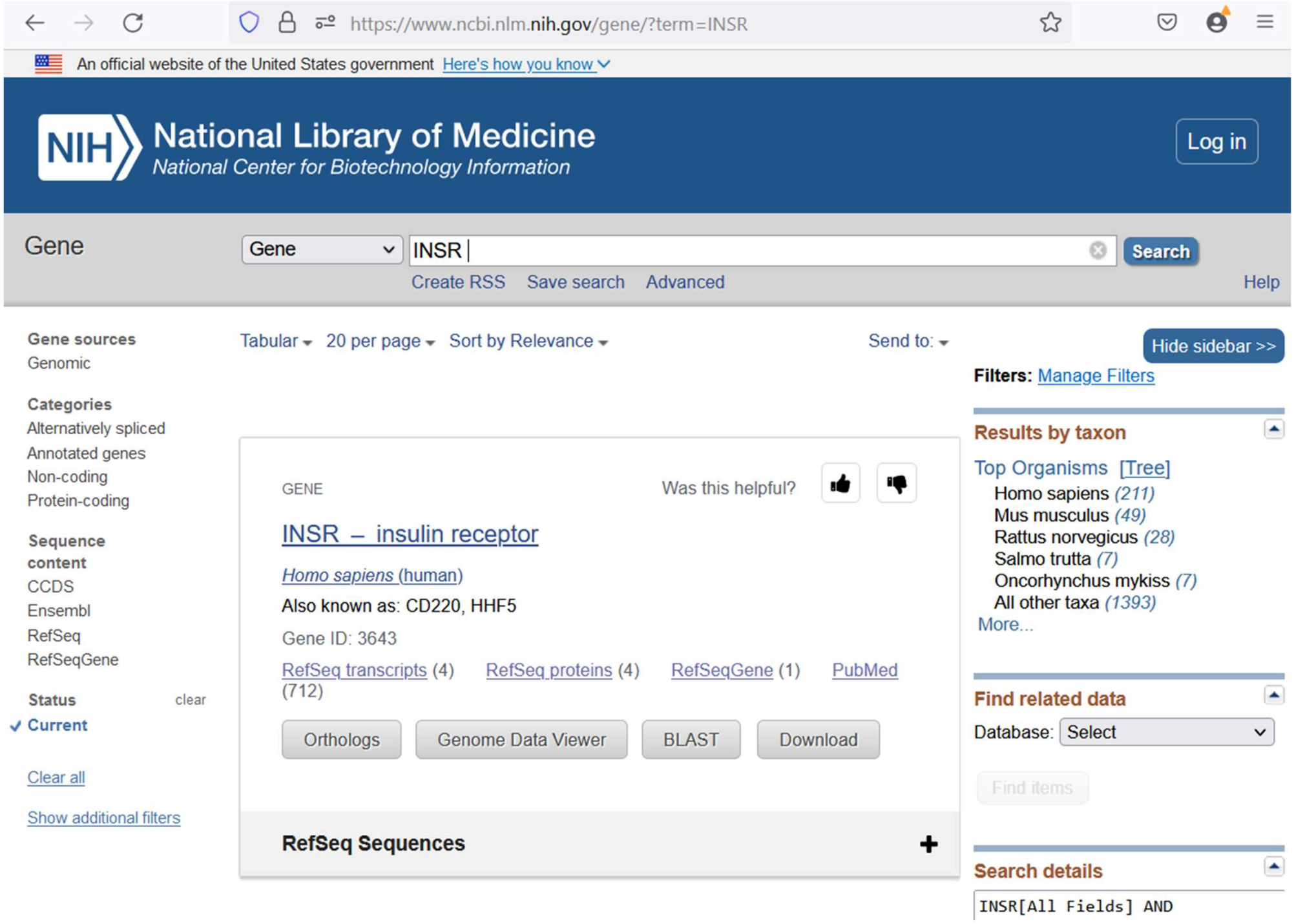
Feature key	Position(s)	Length	Description
Molecule processing			
Signal peptide	1 – 27	27	Ref.2 Ref.11 Ref.12
Chain	28 – 758	731	Insulin receptor subunit alpha
Chain	763 – 1382	620	Insulin receptor subunit beta
Regions			
Topological domain	28 – 758	731	Extracellular Probable
Topological domain	763 – 956	194	Extracellular Probable
Transmembrane	957 – 979	23	Helical; Potential
Topological domain	980 – 1382	403	Cytoplasmic Probable
Domain	622 – 695	74	Fibronectin type-III 1
Domain	757 – 842	86	Fibronectin type-III 2
Domain	850 – 946	97	Fibronectin type-III 3
Domain	1023 – 1298	276	Protein kinase
Nucleotide binding	1104 – 1110	7	ATP
Nucleotide binding	1163 – 1164	2	ATP
Region	733 – 741	9	Insulin-binding
Region	999	1	Important for interaction with IRS1, SHC1 and STAT5B
Region	1361 – 1364	4	PIK3R1-binding
Compositional bias	28 – 174	147	Leu-rich
Compositional bias	182 – 339	158	Cys-rich

https://www.uniprot.org/uniprot/P06213

NCBI



http://www.ncbi.nlm.nih.gov/genome



https://www.ncbi.nlm.nih.gov/gene/?term=INSR

SRA

NCBI Site map | All databases | Search

Sequence Read Archive

Main | Browse | Search | Download | Submit | Documentation | Software | Trace Archive | Trace Assembly | Trace Home | Trace BLAST

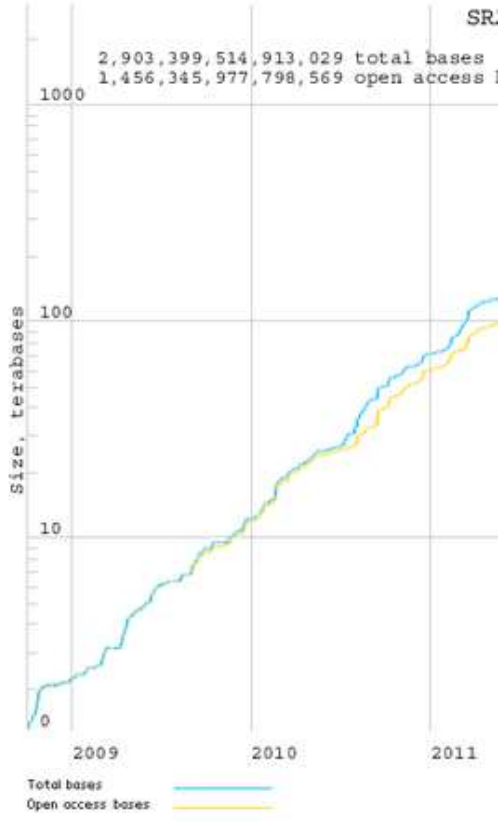
Overview | History | About

Repository NGS sequencing

Overview

The Sequence Read Archive (SRA) stores raw sequence data from "next-generation" sequencing technologies including 454, IonTorrent, Illumina, SOLiD, Helicos and Complete Genomics. In addition to raw sequence data, SRA now stores alignment information in the form of read placements on a reference sequence.

SRA is NIH's primary archive of high-throughput sequencing data and is part of the international partnership of archives (INSDC) at the NCBI, the European Bioinformatics Institute and the DNA Database of Japan. Data submitted to any of the three organizations are shared among them.



- **Submitting to SRA:**

  Making data available to the research community enhances reproducibility and allows for new discovery by comparing data sets.

  - Getting started
  - Quick start guide / FAQ
  - Submitter Login

- **Using SRA Data:**

  Use SRA data to validate experimental results, increase sample sizes, determine variance and open up new avenues of research.

  - SRA Toolkit: Download, Documentation
  - SRA Documentation

- **Coming Soon: SRA SDK (for developers using SRA):**

  - Email the Toolkit team for more information

Search in SRA Documentation

Enter text or search terms to search: [ ] Go

Scope: ☑SRA Handbook ☑SRA Application Notes ☑SRA Knowledge Base ☐Whole NCBI Bookshelf

http://trace.ncbi.nlm.nih.gov/Traces/sra/sra.cgi?view=announcement

GEO - Repository Gene Expression

Gene Expression Omnibus

GEO is a public functional genomics data repository supporting MIAME-compliant data submissions. Array- and sequence-based data are accepted. Tools are provided to help users query and download experiments and curated gene expression profiles.

Keyword or GEO Accession Search

Getting Started

- Overview
- FAQ
- About GEO DataSets
- About GEO Profiles
- About GEO2R Analysis
- How to Construct a Query
- How to Download Data

Tools

- Search for Studies at GEO DataSets
- Search for Gene Expression at GEO Profiles
- Search GEO Documentation
- Analyze a Study with GEO2R
- GEO BLAST
- Programmatic Access
- FTP Site

Browse Content

- Repository Browser

DataSets:	3847
Series:	51248
Platforms:	13435
Samples:	1247951

Information for Submitters

- Login to Submit
- Submission Guidelines
- Update Guidelines
- MIAME Standards
- Citing and Linking to GEO
- Guidelines for Reviewers
- GEO Publications

http://www.ncbi.nlm.nih.gov/geo/

Series GSE46139

Query DataSets for GSE46139

Status	Public on Oct 20, 2013
Title	Genome-wide analysis of E17.5 pituitary gland gene expression of control and Insm1 mutant mice
Organism	Mus musculus
Experiment type	Expression profiling by array
Summary	The Insm1 gene encodes a zinc finger factor expressed in many endocrine organs. We show here that Insm1 is required for differentiation of all endocrine cell types in the pituitary. Thus, in Insm1 mutant mice, hormones characteristic of the different pituitary cell types (thyroid, follicle and melanocyte stimulating hormone, adrenocorticotrope hormone, growth
Platforms (1)	GPL6885 Illumina MouseRef-8 v2.0 expression beadchip
Samples (16)	GSM1124851 Insm1 mutant #1
	GSM1124852 Insm1 mutant #2
	GSM1124853 Insm1 mutant #3

More...

Relations

BioProject PRJNA197343

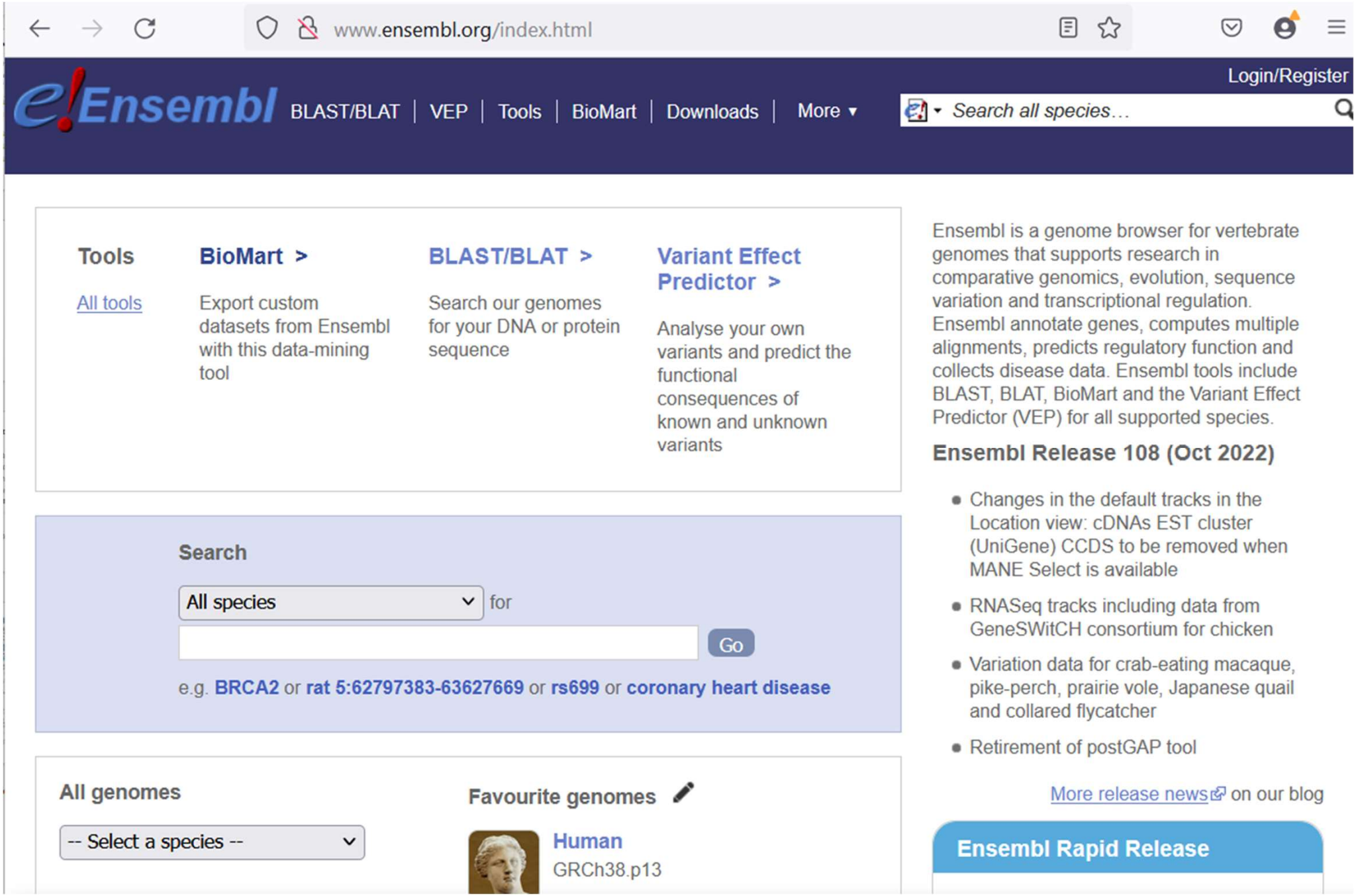
Analyze with GEO2R

Download family	Format
SOFT formatted family file(s)	SOFT
MINiML formatted family file(s)	MINiML
Series Matrix File(s)	TXT

Supplementary file	Size	Download	File type/resource
GSE46139_RAW.tar	3.1 Mb	(http)(custom)	TAR
GSE46139_non-normalized_data.txt.gz	2.5 Mb	(ftp)(http)	TXT

Raw data is available on Series record

Processed data included within Sample table



http://ensembl.org/

UCSC

genome.ucsc.edu

UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute

UCSC **Genome Browser**

Genomes | Genome Browser | Tools | Mirrors | Downloads | My Data | Projects | Help | About Us

genome.ucsc.edu/cgi-bin/hgTracks

Our tools

- **Genome Browser**
  interactively visualize genomic data
- **COVID-19 Research**
  use the SARS-CoV-2 genome browser and explore coronavirus datasets
- **BLAT**
  rapidly align sequences to the genome
- **Table Browser**
  download data from the Genome Browser database
- **Variant Annotation Integrator**
  get functional effect predictions for variant calls
- **Data Integrator**
  combine data sources from the Genome Browser database
- **Genome Browser in a Box (GBiB)**
  run the Genome Browser on your laptop or server
- **In-Silico PCR**
  rapidly align PCR primer pairs to the genome
- **LiftOver**
  convert genome coordinates between assemblies
- **Track Hubs**
  import and view external data tracks
- **REST API**
  returns data in JSON format

More tools...

http://genome.ucsc.edu/

UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute

UCSC Genome Browser Gateway

Browse/Select Species

POPULAR SPECIES

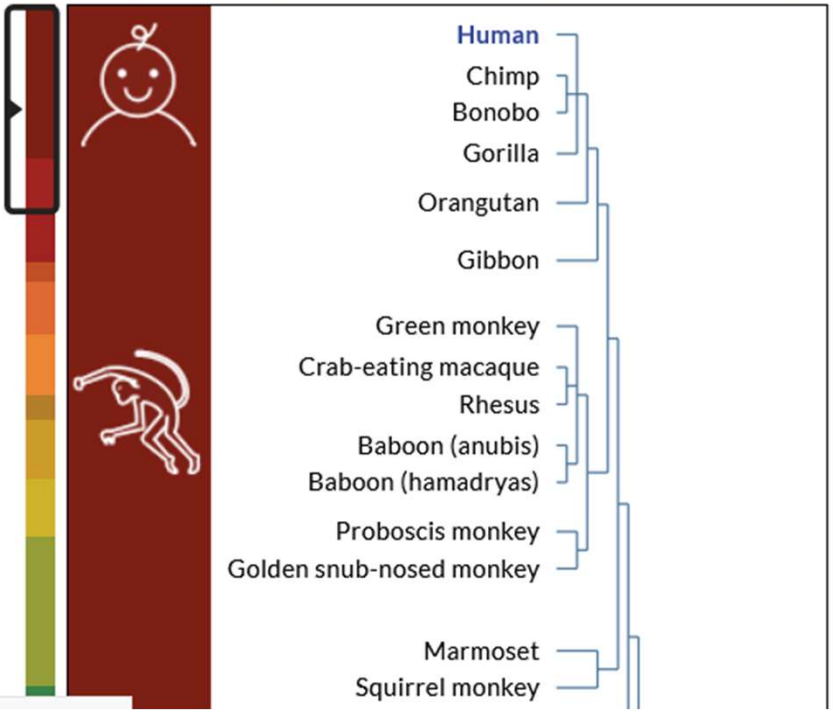
Human | Mouse | Rat | Zebrafish | Fruitfly | Worm | Yeast

Search through thousands of genome browsers

Enter species, common name or assembly ID

Unable to find a genome? Send us a request.

UCSC SPECIES TREE AND CONNECTED ASSEMBLY HUBS



Find Position

Human Assembly

Dec. 2013 (GRCh38/hg38)

GO

Position/Search Term

MTOR

Current position: chr1:11,106,535-11,262,551

Human Genome Browser - hg38 assembly

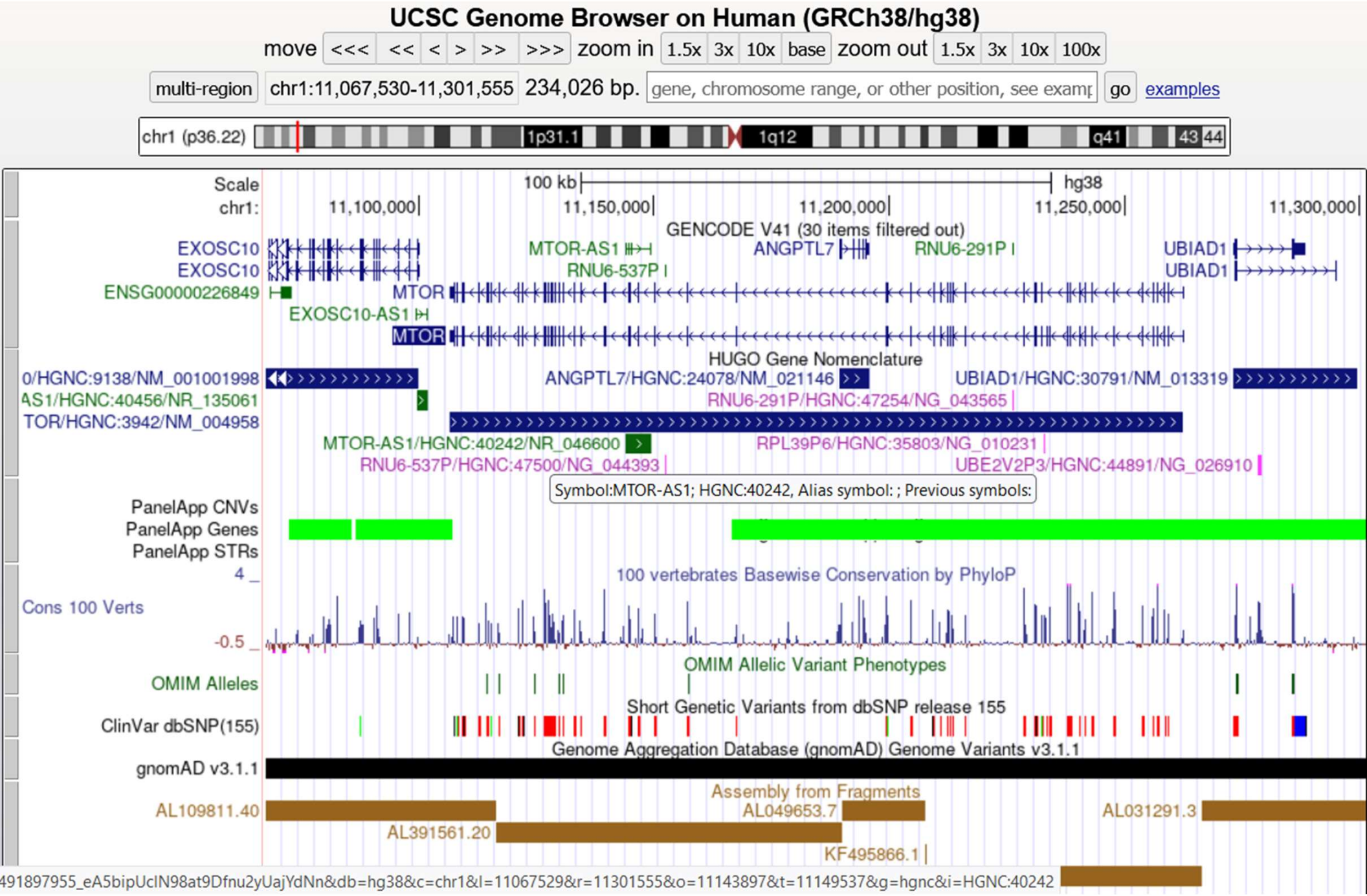
view sequences

UCSC Genome Browser assembly ID: hg38
Sequencing/Assembly provider ID: Genome Reference Consortium Human GRCh38.p13 (GCA_000001405.28)
Assembly date: Dec. 2013 initial release; Dec. 2017 patch release 13
Assembly accession: GCA_000001405.28
NCBI Genome ID: 51 (Homo sapiens (human))
NCBI Assembly ID: GCF_000001405.39 (GRCh38.p13, GCA_000001405.28)
BioProject ID: PRJNA31257

Homo sapiens
(Graphic courtesy of CBSE)

Search the assembly:

- **By position or search term:** Use the "position or search term" box to find areas of the genome associated with many different attributes, such as a specific chromosomal coordinate range; mRNA, EST, or STS marker names; or keywords from the GenBank description of an mRNA. More information, including



Browser de nombreux génomes : informations très complètes

Human Gene INSR (ENST00000341500.9) from GENCODE V41

Description: Homo sapiens insulin receptor (INSR), transcript variant 2, mRNA. (from RefSeq NM_001079817)
RefSeq Summary (NM_001079817): This gene encodes a member of the receptor tyrosine kinase family of proteins. The encoded preproprotein proteolytically processed to generate alpha and beta subunits that form a heterotetrameric receptor. Binding of insulin or other ligands to this receptor activates the insulin signaling pathway, which regulates glucose uptake and release, as well as the synthesis and storage of carbohydrate lipids and protein. Mutations in this gene underlie the inherited severe insulin resistance syndromes including type A insulin resistance syndrome, Donohue syndrome and Rabson-Mendenhall syndrome. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Oct 2015].
Gencode Transcript: ENST00000341500.9
Gencode Gene: ENSG00000171105.14
Transcript (Including UTRs)
Position: hg38 chr19:7,112,255-7,293,931 **Size:** 181,677 **Total Exon Count:** 21 **Strand:** -
Coding Region
Position: hg38 chr19:7,117,056-7,293,891 **Size:** 176,836 **Coding Exon Count:** 21

Page Index	Sequence and Links	UniProtKB Comments	MalaCards	CTD	RNA-Seq Expression
Microarray Expression	RNA Structure	Protein Structure	Other Species	GO Annotations	mRNA Descriptions
Pathways	Other Names	GeneReviews	Methods		

Data last updated at UCSC: 2022-05-14 09:57:26

− Sequence and Links to Tools and Databases

Genomic Sequence (chr19:7,112,255-7,293,931)			mRNA (may differ from genome)		Protein (1370 aa)
Gene Sorter	Genome Browser	Other Species FASTA	Gene interactions	Table Schema	BioGPS
CGAP	Ensembl	Entrez Gene	ExonPrimer	Gencode	GeneCards
HGNC	HPRD	Lynx	MGI	neXtProt	OMIM
PubMed	Reactome	UniProtKB	Wikipedia		

− Comments and Description Text from UniProtKB

ID: INSR_HUMAN
DESCRIPTION: RecName: Full=Insulin receptor; Short=IR; EC=2.7.10.1; AltName: CD_antigen=CD220; Contains: RecName: Full=Insulin recept

Genomic Sequence Near Gene

Get Genomic Sequence Near Gene

Note: if you would prefer to get DNA for more than one feature of this track at a time, try the Table Browser using the output format

Sequence Retrieval Region Options:

- ☐ Promoter/Upstream by 1000 bases
- ☑ 5' UTR Exons
- ☑ CDS Exons
- ☑ 3' UTR Exons
- ☑ Introns
- ☐ Downstream by 1000 bases
- ◉ One FASTA record per gene.
- ○ One FASTA record per region (exon, intron, etc.) with 0 extra bases upstream (5') and 0 extra downstream (3')
  - ☐ Split UTR and CDS parts of an exon into separate FASTA records

Note: if a feature is close to the beginning or end of a chromosome and upstream/downstream bases are added, they may be trur
avoid extending past the edge of the chromosome.

Sequence Formatting Options:

- ◉ Exons in upper case, everything else in lower case.
- ○ CDS in upper case, UTR in lower case.
- ○ All upper case.
- ○ All lower case.
- ☐ Mask repeats: ◉ to lower case ○ to N

submit

>hg19_knownGene_uc002mgd.1 range=chr19:7112266-7294011 5'pad=0 3'pad=0 strand=- repeatMasking=none

GAGAAGGACGCGCGGCCCCCAGCGCCTCTTGGGTGGCCGCCTCGGAGCAT
GACCCCCGCGGGCCAGCGCCGCGCGCTCTGATCCGAGGAGACCCCGCGCT
CCCGCAGCCATGGCCACCGGGGGCCGGCGGGGGGCGGCGGCCGCGCCGCT
GCTGGTGGCGGTGGCCGCGCTGCTACTGGGCGCCGCGGGCCACCTGTACC
CCGGAGAGGgtgagtctgggggcgcgggcgtgggcggggagcgccgcgat
ggggagaggaccccacccaagccaaaatcgagcccccgcttgtggactga
gaaccctccccaggggcgggggcggtggccaggacggtagctcctgcat
cgcgtaggggggagcgggaagcctctgaccttggcctttgcccgccccgggc
tcgcgcctccgcgccctgcgtgcccgacctgagcccgaggaaccttgccc

1er exon 5'UTR

--- - - - - - - - - - - - -
aagaaaacaccatctctcagcacctctagcaatgctatttctgccttgac
aattgtccacacacacagccagcatctgatgtgagcacatgtggtccgtg
ggtgtcggctgcagggacaagagtgggggtttgggaggatgcgtggcagg
gccccccagactcacccaggacgtgtccttctgccccgcagCACTGACCTC
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GATTGTCAACCTGCTCAAGGACGACCTGCACCCCAGCTTTCCAGAGGTGT
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GTTTGGTTACCAATTTAACTAGTCCTGCAGAGGATTTAACTGTGAACCTG
GAGGGCAAGGGGTTTCCACAGTTGCTGCTCCTTTGGGGCAACGACGGTTT
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GAATTTCAAGTTTTACAGGTTGAGCTTCAAGATGGTTTTTTTGGTTTTTT
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TTCCTCAAATTGACCAATAGCTGCTGCTTTCATATTTTGGATAAGGGTCT
GTGGTCCCGGCGTGTGCTCACGTGTGTATGCACGTGTGTGTGTCCATTAG
ACACGGCTGATGTGTGTGCAAAGTATCCATGCGGAGTTGATGCTTTGGGA
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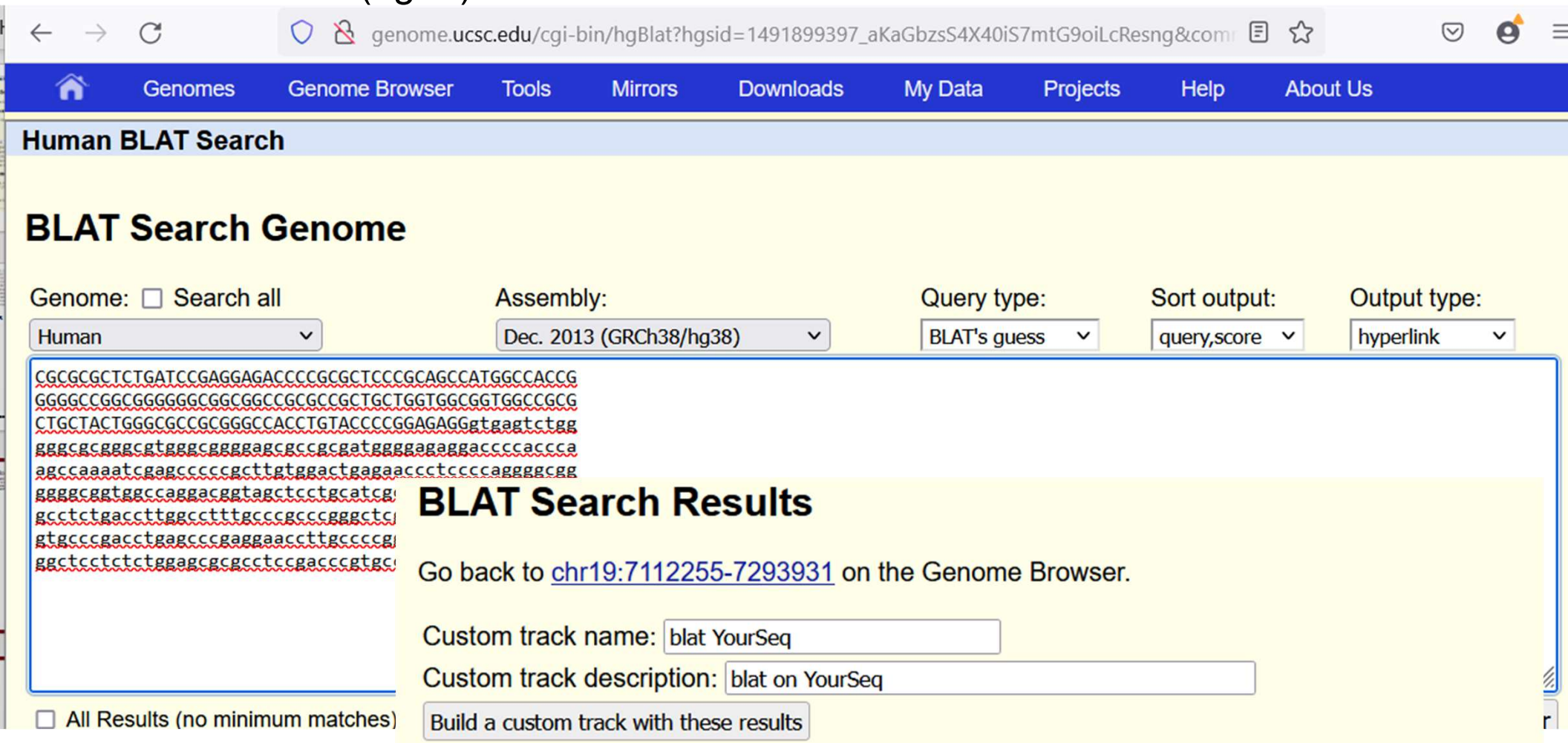
AAAAAGGAAAAAAAAAAAAGAACTGAGATGACAGAGTTTTGAGAATATATT
TGTACCATATTTAATTTTTAAAGTCTCTGGTATTAGCCTCATAAGTTATT
GACTATTCCCCGGGGTTGGCGGGGAGTGGGGACATGAGTTGGTCTGCCTG
TTGTGGGGCCGGGAAGGGGAGGGAGTCAGGCACAAGTGGCCTCTTTGTTT
GGTCTTAAAGGCATCCATTTCTGGGAATGAAGCCATGTTCGCTGCTAACA

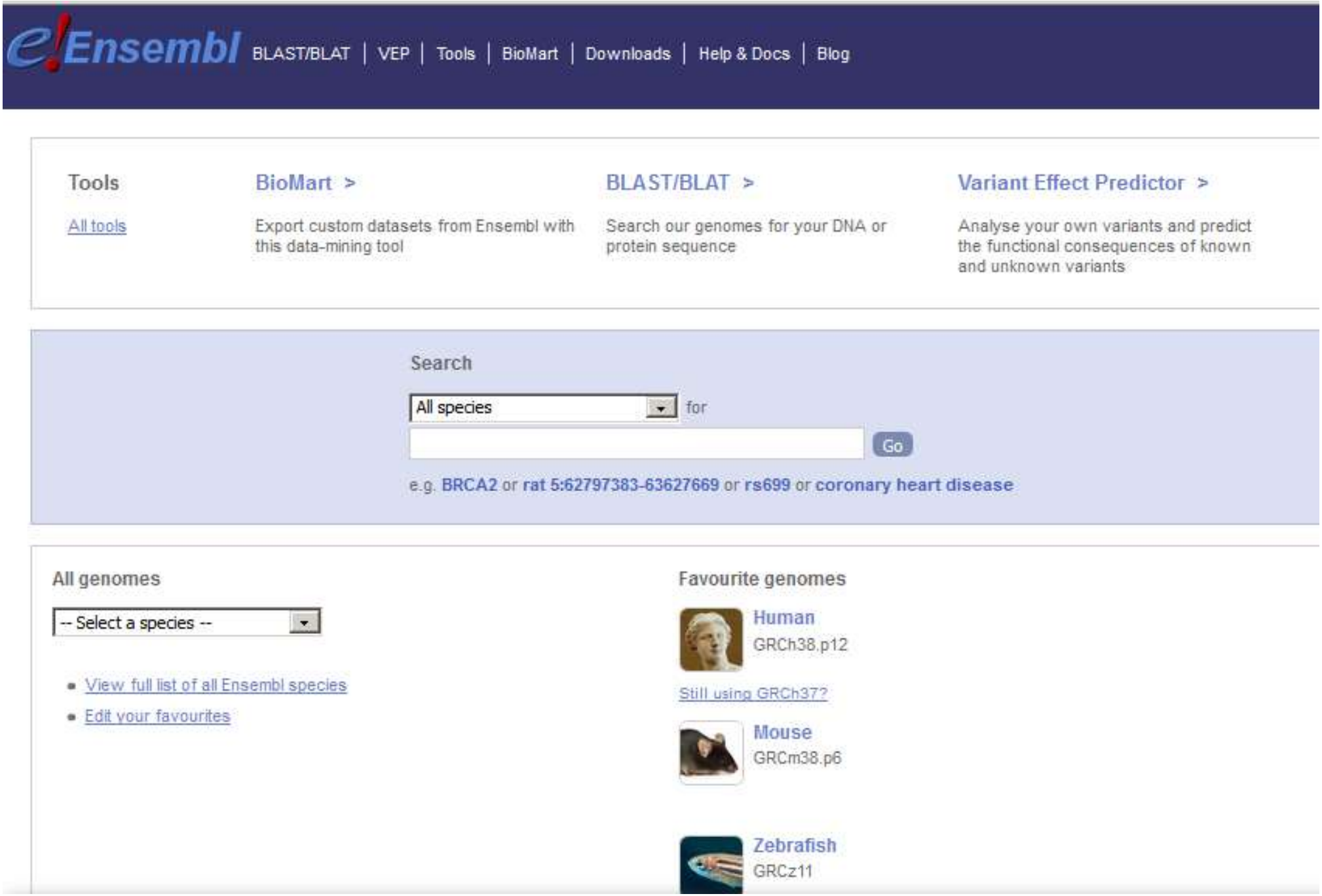
dernier exon 3'UTR

Sequence à localiser

CGCGCGCTCTGATCCGAGGAGACCCCGCGCTCCCGCAGCCATGGCCACCG GGGGCCGGCGGGGGGCGGCGGCCGCGCCGCTGCTGGTGGCGGT
GGCCGCG CTGCTACTGGGCGCCGCGGGCCACCTGTACCCCGGAGAGGgtgagtctgg gggcgcgggcgtgggcggggagcgccgcgatggggagaggaccccaccca agcca
aaatcgagcccccgcttgtggactgagaaccctccccaggggcgg ggggcggtggccaggacggtagctcctgcatcgcgtagggggagcgggaa gcctctgaccttggcctttgcccgcccgggctcgcgcctccgc
gccctgc gtgcccgacctgagcccgaggaaccttgccccggtgcccgccccgccgcg ggctcctctctggagcgcgcctccgacccgtgccccggcccctccgagcc

Tools :: BLAT (hg38) : submit





http://www.ensembl.org

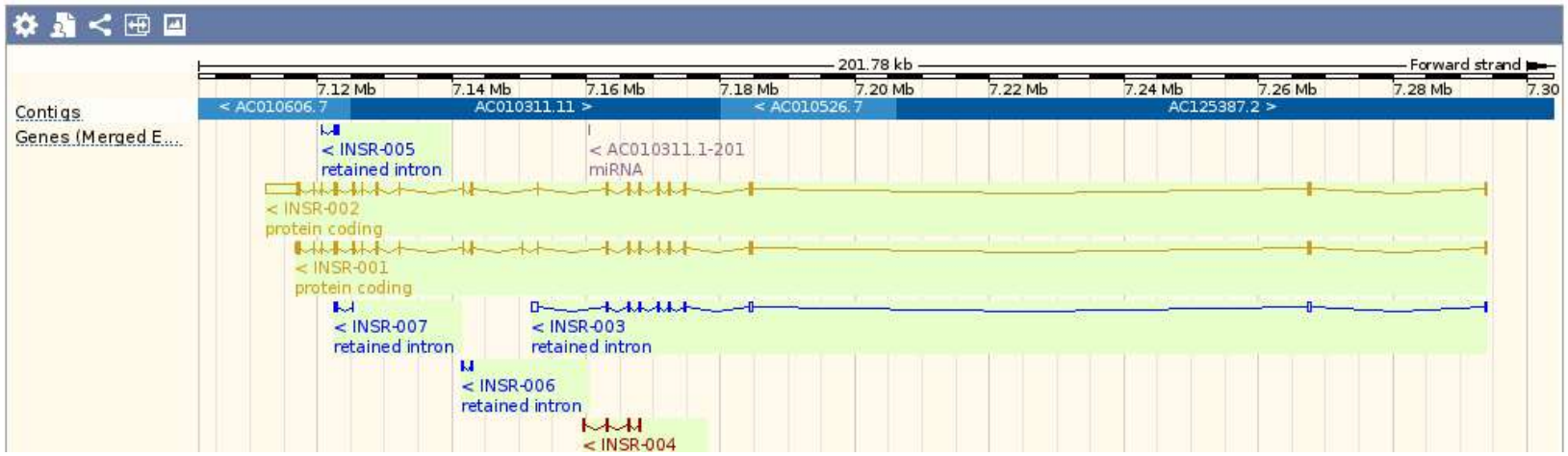
Gene: INSR ENSG00000171105

Description	insulin receptor [Source:HGNC Symbol;Acc:6091]
Location	Chromosome 19: 7,112,266-7,294,045 reverse strand.
INSDC coordinates	chromosome:GRCh37:CM000681.1:7112266:7294045:1
Transcripts	This gene has 7 transcripts (splice variants) Show transcript table

Gene summary

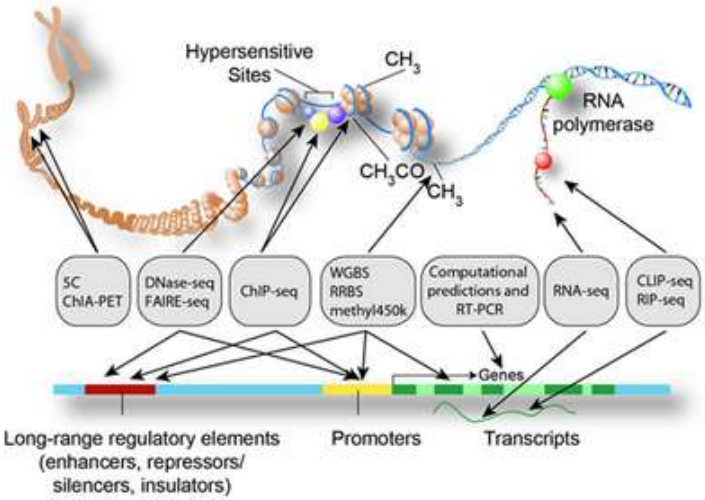
Name	INSR (HGNC Symbol)
Synonyms	CD220 [To view all Ensembl genes linked to the name click here.]
CCDS	This gene is a member of the Human CCDS set: CCDS12176, CCDS42487
Ensembl version	ENSG00000171105.9
Gene type	Known protein coding
Prediction Method	Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see article.
Alternative genes	**This gene corresponds to the following database identifiers:** **Havana gene:** OTTHUMG00000181992 (version 2)

Go to Region in Detail for more tracks and navigation options (e.g. zooming)



ENCODE Data Methods About ENCODE Help Search ENCODE Sign in

ENCODE: Encyclopedia of DNA Elements



The ENCODE (Encyclopedia of DNA Elements) Consortium is an international collaboration of research groups funded by the National Human Genome Research Institute (NHGRI). The goal of ENCODE is to build a comprehensive parts list of functional elements in the human genome, including elements that act at the protein and RNA levels, and regulatory elements that control cells and circumstances in which a gene is active.

Image credits: Darryl Leja (NHGRI), Ian Dunham (EBI), Michael Pazin (NHGRI)

Data

To find and download ENCODE Consortium data:

- Click the Data toolbar above and browse data
  - By assay

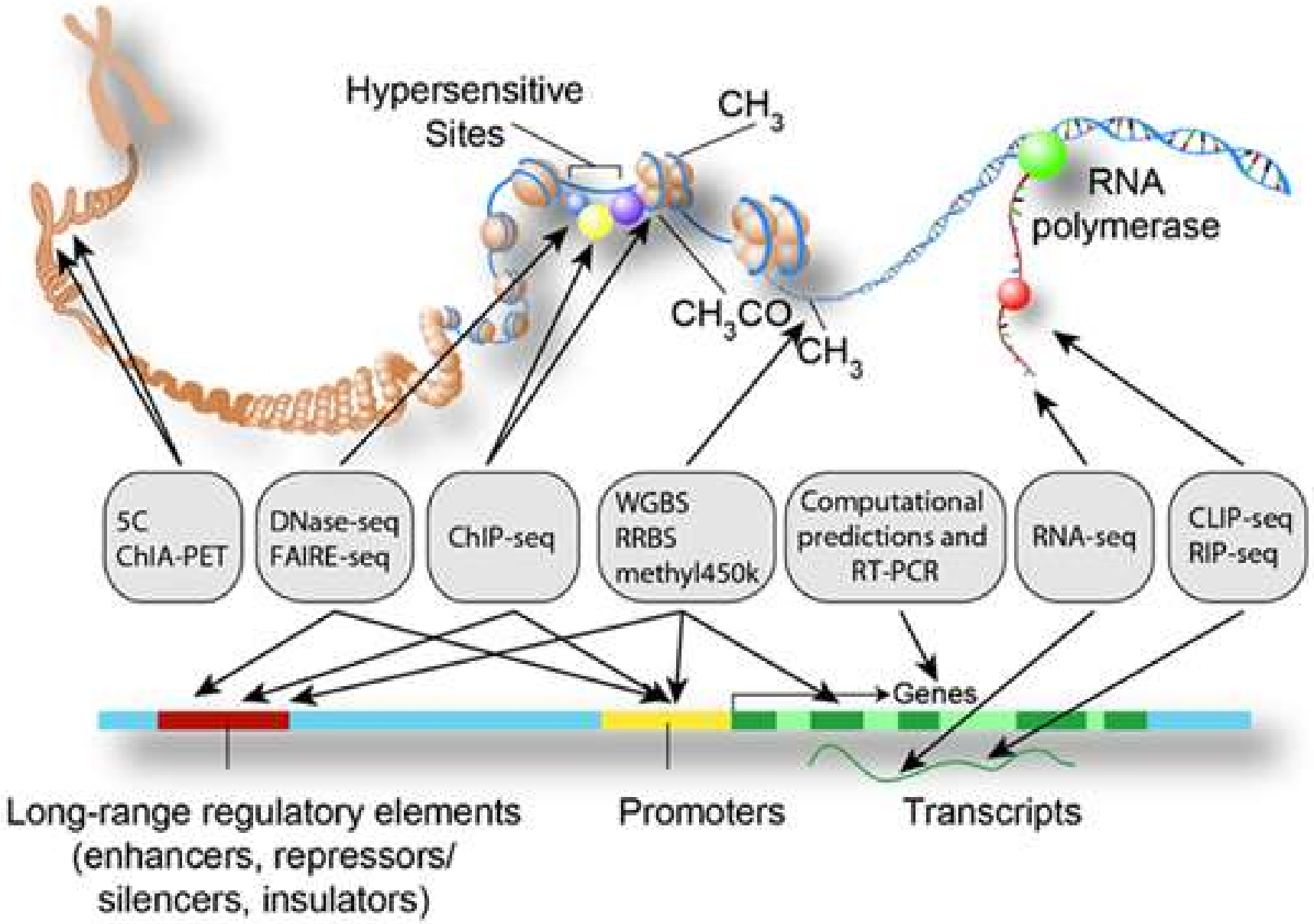
News

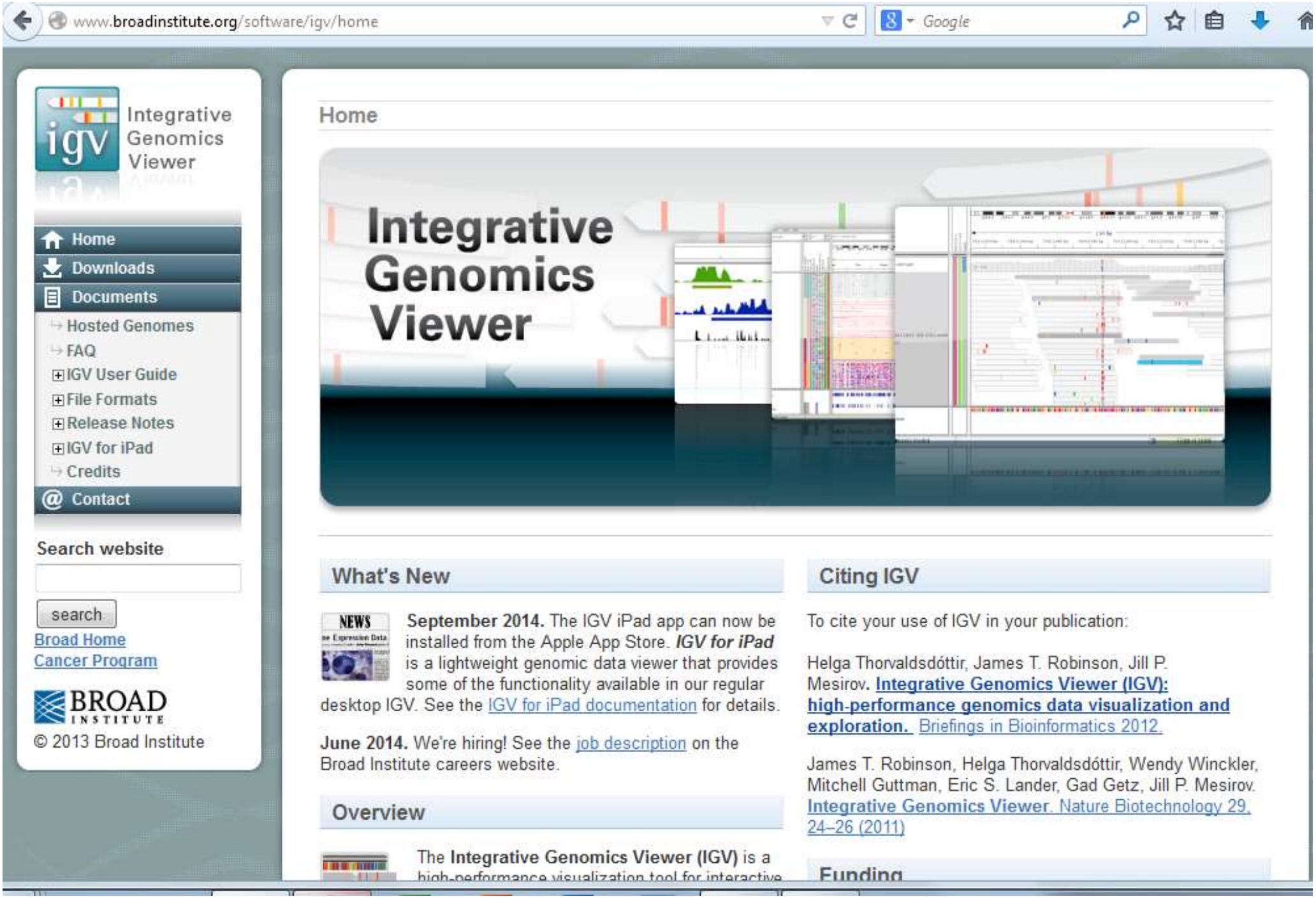
Sept 12, 2014: Data release: 23 human and 5 mouse datasets. [read more]

August 28, 2014: modENCODE and ENCODE comparison papers published. [read more]

https://www.encodeproject.org/

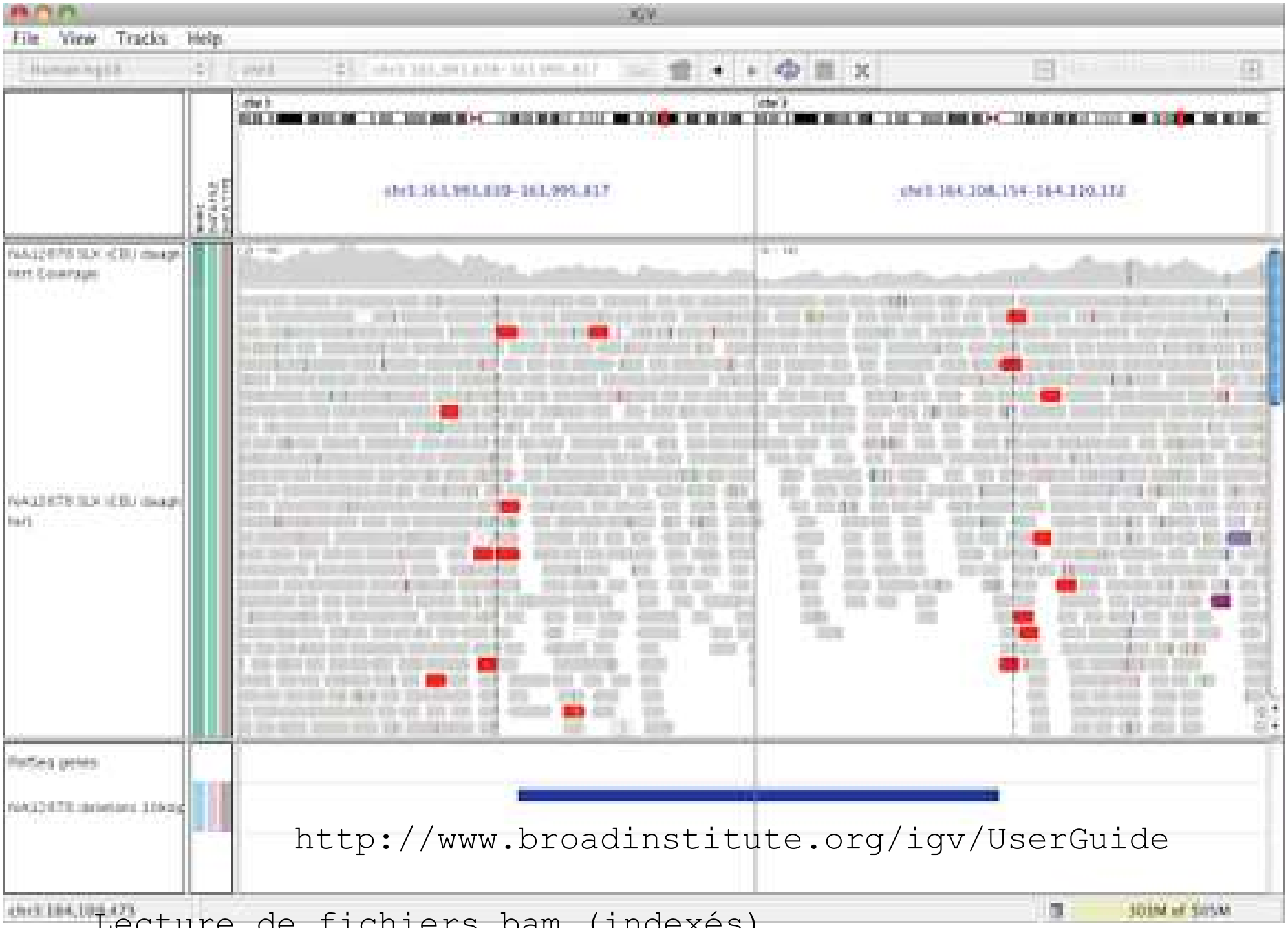
https://www.encodeproject.org/biosamples/ENCBS220I


Hypersensitive Sites
CH3
CH3CO
CH3
RNA polymerase
5C ChIA-PET
DNase-seq FAIRE-seq
ChIP-seq
WGBS RRBS methyl450k
Computational predictions and RT-PCR
RNA-seq
CLIP-seq RIP-seq
Genes
Long-range regulatory elements (enhancers, repressors/ silencers, insulators)
Promoters
Transcripts



http://www.broadinstitute.org/software/igv/home

IGV – Integrated Genome Viewer (Broad Institute)



http://www.broadinstitute.org/igv/UserGuide

Lecture de fichiers bam (indexés)

Links to polymorphism sites

Polymorphism : SNP, mutations

dbSNP Single Nucleotide Polymorphism (NCBI, Bethesda, Us)
A Database of Single Nucleotide Polymorphisms : A key aspect of research in genetics is associating sequence variations with heritable phenotypes. The most common variations are single nucleotide polymorphisms (SNPs), which occur approximately once every 100 to 300 bases. Because SNPs are expected to facilitate large-scale association genetics studies, there has recently been great interest in SNP discovery and detection.

HAPMAP (NCBI, Bethesda, Us)
The International HapMap Project is a partnership of scientists and funding agencies from Canada, China, Japan, Nigeria, the United Kingdom and the United States to develop a public resource that will help researchers find genes associated with human disease and response to pharmaceuticals. See "About the International HapMap Project" for more information.

Exome Variant server (EVS) (Washington, Us)
The goal of the NHLBI GO Exome Sequencing Project (ESP) is to discover novel genes and mechanisms contributing to heart, lung and blood disorders by pioneering the application of next-generation sequencing of the protein coding regions of the human genome across diverse, richly-phenotyped populations and to share these datasets and findings with the scientific community to extend and enrich the diagnosis, management and treatment of heart, lung and blood disorders.

gnomAD (Broad Institute, Boston, Us)
The Genome Aggregation Database (gnomAD) is a resource developed by an international coalition of investigators, with the goal of aggregating and harmonizing both exome and genome sequencing data from a wide variety of large-scale sequencing projects, and making summary data available for the wider scientific community. The data set provided on this website spans 123,136 exome sequences and 15,496 whole-genome sequences from unrelated individuals sequenced as part of various disease-specific and population genetic studies. The gnomAD Principal Investigators and groups that have contributed data to the current release are listed here.

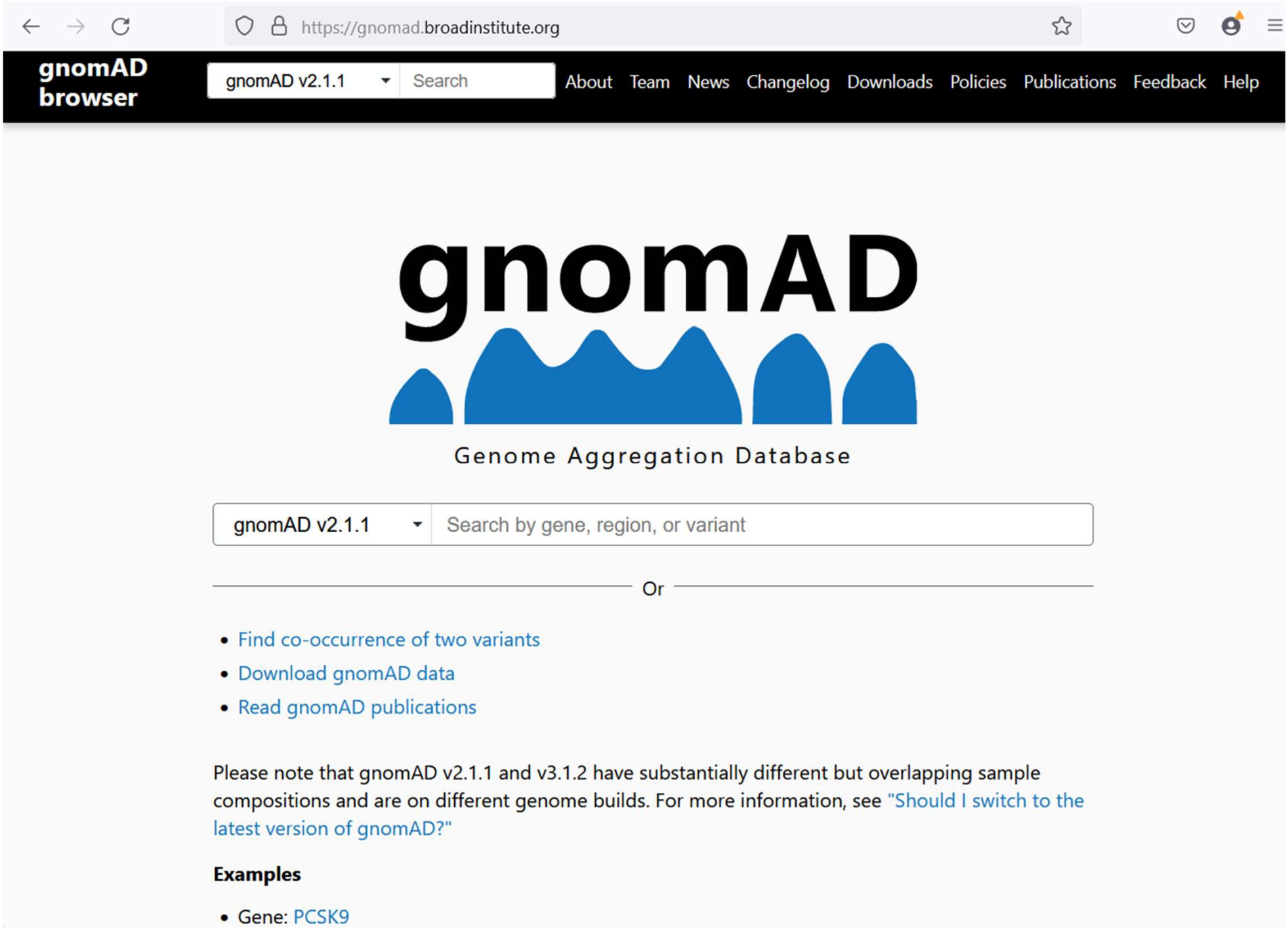
Varsome (US)
VarSome is a search engine, aggregator and impact analysis tool for human genetic variation and a community-driven project aiming at sharing global expertise on human variants. It renders and displays a detailed annotation of the queried variant, including multiple notations, predicted pathogenicity status from a variety of tools, genomic context, as well as information from 35+ public databases. It allows users to mark the pathogenicity of variants and to link variants to specific phenotypes, diseases and publications. Finally, it provides an automated pathogenicity assessment consistent with the widely accepted ACMG guidelines. It therefore provides a powerful analysis resource as well as a repository for the accumulated global knowledge of the genomics community. From a technical point of view, it allows convenient programmable single-point interface (API) for accessing all its data

M-CAP (US)
Mendelian Clinically Applicable Pathogenicity (M-CAP) Score M-CAP is the first pathogenicity classifier for rare missense variants in the human genome that is tuned to the high sensitivity required in the clinic (see Table). By combining previous pathogenicity scores (including SIFT, Polyphen-2 and CADD) with novel features and a powerful model, we attain the best classifier at all thresholds, reducing a typical exome/genome rare (<1%) missense variant (VUS) list from 300 to 120, while never mistaking 95% of known pathogenic variants as benign.

Varity (US)
VARITY (Improved pathogenicity prediction for rare human missense variants) Web Application User Guide. This web application provides: 1) Search and visualize VARITY predictions, features and feature contributions for all possible single nucleotide change missense variants for each of 18,239 human proteins. 2) Download VARITY predictions in one file for all 18,239 proteins. NOTE: All VARITY predictions are for research purpose and should be appropriately validated before clinical use

ICGC (OICR, Ontario, Ca)
ICGC Goal: To obtain a comprehensive description of genomic, transcriptomic and epigenomic changes in 50 different tumor typ es and/or subtypes which are of clinical and societal importance across the globe.

http://pdessen.free.fr/atlas_links.html



http://gnomad.broadinstitute.org/

Interested in working on the development of this resource? Apply here.

Gene: IDH2

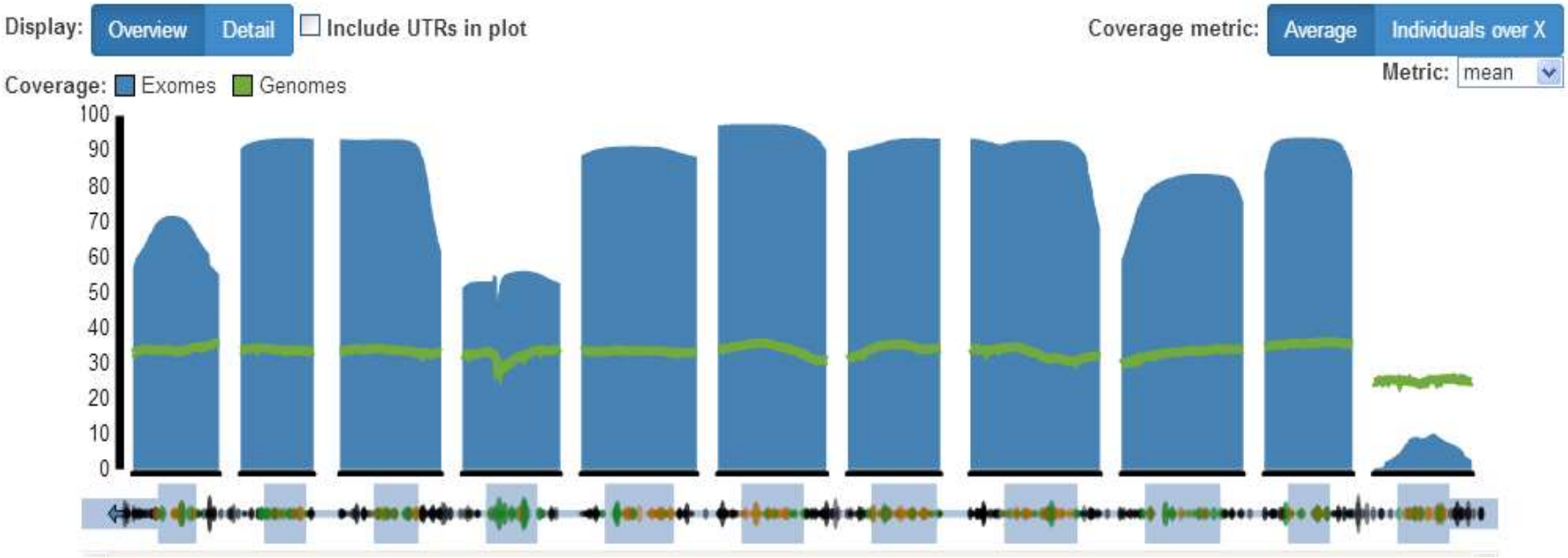
IDH2	isocitrate dehydrogenase 2 (NADP+), mitochondrial
Number of variants	992 (Including filtered: 1178)
UCSC Browser	15:90626277-90645736
GeneCards	IDH2
OMIM	147650
Other	External References

Transcripts

Gene summary

(Coverage shown for canonical transcript: ENST00000330062)

Mean coverage 77.80



All | Missense + LoF | LoF

Include:

Exomes | SNPs | Filtered (non-PASS) variants

Genomes | Indels

Export table to CSV

† denotes a consequence that is for a non-canonical transcript

Variant	Source	Consequence	Annotation	Flags	Allele Count	Allele Number	Number of Homozygotes	Allele Frequen
15:90626207 A / G (rs576677991)	G		downstream gene		4	30810	0	0.0001298
15:90626221 C / T (rs562171635)	G		downstream gene		3	30952	0	9.692e-5
15:90626228 A / G	G		downstream gene		6	30958	0	0.0001938
15:90626231 C / A (rs183887095)	G		downstream gene		1	30862	0	3.24e-5
15:90626231 C / G (rs183887095)	G		downstream gene		3	30862	0	9.721e-5
15:90626242 A / C (rs528412662)	G		downstream gene		3	30944	0	9.695e-5
15:90626248 T / C (rs541168105)	G		downstream gene		3	30946	0	9.694e-5
15:90626261 A / G (rs533103776)	G		downstream gene		3	30934	1	9.698e-5
15:90626269 G / A	G		downstream gene		1	30900	0	3.236e-5
15:90626281 G / A	G		3' UTR		1	30716	0	3.256e-5
15:90626286 T / C	G		3' UTR		2	30878	0	6.477e-5
15:90626286 T / G	G		3' UTR		2	30878	0	6.477e-5
15:90626315 C / G	G		3' UTR		3	27546	0	0.0001089
15:90626315 C / T	G		3' UTR		4	27546	0	0.0001452
15:90626316 C / G	G		3' UTR		1	26628	0	3.755e-5
15:90626323 C / T (rs546370045)	G		3' UTR		4	27958	0	0.0001431
15:90626324 A / G	G		3' UTR		1	27964	0	3.576e-5
15:90626325 T / C	G		3' UTR		1	30204	0	3.311e-5
15:90626426 G / A	G		3' UTR		2	28190	0	7.095e-5
15:90626433 C / T	G		3' UTR		3	28476	0	0.0001054
15:90626481 C / A	G		3' UTR		2	30516	0	6.554e-5

All | Missense + LoF | LoF

Include:

- [x] Exomes
- [x] Genomes
- [x] SNPs
- [x] Indels
- [x] Filtered (non-PASS) variants

Export table to CSV

† denotes a consequence that is for a non-canonical transcript

Variant	Source	Consequence	Annotation	Flags	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
15:90627498 CT / C (rs201015211)	G	p.Ter453Trp	frameshift	LC LoF	1	30964	0	3.23e-5
15:90627506 TG / T (rs764173046)	E	p.Arg451GlyfsTer17	frameshift	LC LoF	1	245688	0	4.07e-6
15:90628232 C / T	E	c.1178+1G>A	splice donor		1	246096	0	4.063e-6
15:90628261 T / TC	E	p.Lys384GlufsTer96	frameshift		1	246090	0	4.064e-6
15:90628278 CCA / C (rs770463292)	E	p.Gly378ProfsTer101	frameshift		1	246132	0	4.063e-6
15:90628318 TG / T (rs773746533)	E	p.Ser365AlafsTer47	frameshift		1	245890	0	4.067e-6
15:90628532 GT / G	E	p.Thr352ProfsTer60	frameshift		1	150162	0	6.659e-6
15:90628534 GAC / G (rs765909746)	E	p.Val351HisfsTer128	frameshift		79	149086	0	0.0005299
15:90630446 GC / G (rs752451868)	E	p.Leu289SerfsTer41	frameshift		1	246272	0	4.061e-6
15:90630678 A / AG	E	p.Phe270LeufsTer2	frameshift		1	246272	0	4.061e-6
15:90631653 C / A (rs761129118)	E	p.Glu206Ter	stop gained		1	246256	0	4.061e-6
15:90631821 G / A (rs763369478)	E	p.Gln178Ter	stop gained		1	246132	0	4.063e-6
15:90631917 T / TC (rs780120934)	G	p.Thr146AspfsTer126	frameshift		2	277136	0	7.217e-6
15:90631917 TC / T (rs780120934)	E	p.Thr146LeufsTer15	frameshift		1	246248	0	4.061e-6
15:90633729 CA / C	E	p.Asp119MetfsTer10	frameshift		1	246100	0	4.063e-6
15:90634850 TC / T	E	p.Lys48SerfsTer11	frameshift		1	246272	0	4.061e-6
15:90643807 C / A (rs867541960)	G	c.-42+1G>T†	splice donor	LC LoF	1	30970	0	3.229e-5
15:90645534 G / GT	G	p.Thr30AsnfsTer27	frameshift		1	29772	0	3.359e-5
15:90645587 GC / G	E	p.Cys12SerfsTer15	frameshift		0	74980	0	0

Links to disease sites

Diseases

OMIM Online Mendelian Inheritance in Man" (John Hopkins, Baltimore, Us)
OMIM is a catalog of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues at Johns Hopkins and elsewhere, and developed for the World Wide Web by NCBI, the National Center for Biotechnology Information. The database contains textual information, pictures, and reference information. It also contains copious links to NCBI's Entrez database of MEDLINE articles and sequence information.

MedGen (NCBI, Bethesda, Us)
MedGen is NCBI portal to information about human disorders and other phenotypes having a genetic component. MedGen is structured to serve health care professionals, the medical genetics community, and other interested parties by providing centralized access to diverse types of content. For example, because MedGen aggregates the plethora of terms used for particular disorders into a specific concept, it provides a Rosetta stone for stakeholders who may use different names for the same disorder. Maintaining a clearly defined set of concepts and terms for phenotypes is essential to support efforts to characterize genetic variation by its effects on specific phenotypes. The assignment of identifiers for those concepts allows computational access to phenotypic information, an essential requirement for the large-scale analysis of genomic data.

dbGap (NCBI, Bethesda, Us)
The database of Genotypes and Phenotypes (dbGaP) was developed to archive and distribute the data and results from studies that have investigated the interaction of genotype and phenotype in Humans.

ClinVar (NCBI, Bethesda, Us)
ClinVar is designed to provide a freely accessible, public archive of reports of the relationships among human variations and phenotypes, with supporting evidence. By so doing, ClinVar facilitates access to and communication about the relationships asserted between human variation and observed health status, and the history of that interpretation. ClinVar collects reports of variants found in patient samples, assertions made regarding their clinical significance, information about the submitter, and other supporting data. The alleles described in submissions are mapped to reference sequences, and reported according to the HGVS standard. ClinVar then presents the data for interactive users as well as those wishing to use ClinVar in daily workflows and other local applications. ClinVar works in collaboration with interested organizations to meet the needs of the medical genetics community as efficiently and effectively as possible. Information about using ClinVar.

GTR (The Genetic Testing Registry) (NIH, Bethesda, Us)
The Genetic Testing Registry (GTR) provides a central location for voluntary submission of genetic test informat ion by providers. The scope includes the test's purpose, methodology, validity, evidence of the test's usefulnes s, and laboratory contacts and credentials. The overarching goal of the GTR is to advance the public health and research into the genetic basis of health and disease.

Open Targets (Hinxton, Uk)
The Target Validation Platform (www.targetvalidation.org) aims to support researchers in identifying early drug targets faster and with more confidence. The platform integrates data from several public databases and is the result of a collaboration between the Sanger Institute, GlaxoSmithKline (GSK), the European Bioinformatics Institute (EBI) and Biogen.
As part of our ongoing efforts to improve this valuable public resource, we want to talk to experimental biology researchers who study associations of human genes with diseases. We are interested in understanding how well the platform meets your needs and what other information and features would make it more useful to you. A typical session takes about an hour of your time. Previous participants have found them to be a lot of fun!

http://pdessen.free.fr/atlas_links.html

HuGE Navigator

HuGE Navigator provides access to a continuously updated knowledge base in human genome epidemiology, including information on population prevalence of genetic variants, gene-disease associations, gene-gene and gene- environment interactions, and evaluation of genetic tests
The Office of Public Health Genomics (OPHG), CDC The Centers for Disease Control and Prevention (CDC) established the Office of Public Health Genomics (OPHG) in 1997. OPHG aims to integrate genomics into public health research, policy, and programs, which could improve interventions designed to prevent and control the country's leading chronic, infectious, environmental, and occupational diseases.
OPHG's efforts focus on conducting population-based genomic research, assessing the role of family health history in disease risk and prevention, supporting a systematic process for evaluating genetic tests, translating genomics into public health research and programs, and strengthening capacity for public health genomics in disease prevention programs. (Centers for Disease Control and Prevention (CDC)

ORPHANET : Database of rare diseases and orphan drugs (INSERM, Paris, Fr)

This project is the result of a commonly observed fact: rare diseases are difficult to deal with for medical practitioners. This is due to their restricted knowledge of the diseases' natural history, the patient care required, treatment, and sometimes even of its existence. Scientific knowledge exists, or at least partial knowlege, but it is scattered. Because of the physical media on which it is communicated, the information is difficult to access for the great majority of physicians, not to mention patients and their families. Only a very small number of doctors specialize in these diseases, and their practices are scarcely known, sometimes even totally unknown to other practitioners.
The fields currently covered are:

- rare diseases, that is to say, those diseases for which prevalence is inferior to 1/1000 in the population
- research projects related to these diseasess
- specialized practices related to these diseases
- laboratories specializing in their diagnosis
- research laboratories currently involved in the field
- patient organizations dealing with these diseases
- other national or international servers dedicated to these diseases.
- other similar or complementary national or international databases
- bibliographic references for these diseases

- a message-taking service that dispatches user's questions to an expert in the field.

DisGeNET (Es)

DisGeNET is a discovery platform containing one of the largest publicly available collections of genes and variants associated to human diseases (Piñero et al., 2016; Piñero et al., 2015). DisGeNET integrates data from expert curated repositories, GWAS catalogues, animal models and the scientific literature. DisGeNET data are homogeneously annotated with controlled vocabularies and community-driven ontologies. Additionally, several original metrics are provided to assist the prioritization of genotype¿phenotype relationships.

ClinGen : Clinical Genome resource (NIH, Bethesda, Us)

ClinGen is a National Institutes of Health (NIH)-funded resource dedicated to building an authoritative central resource that defines the clinical relevance of genes and variants for use in precision medicine and research.

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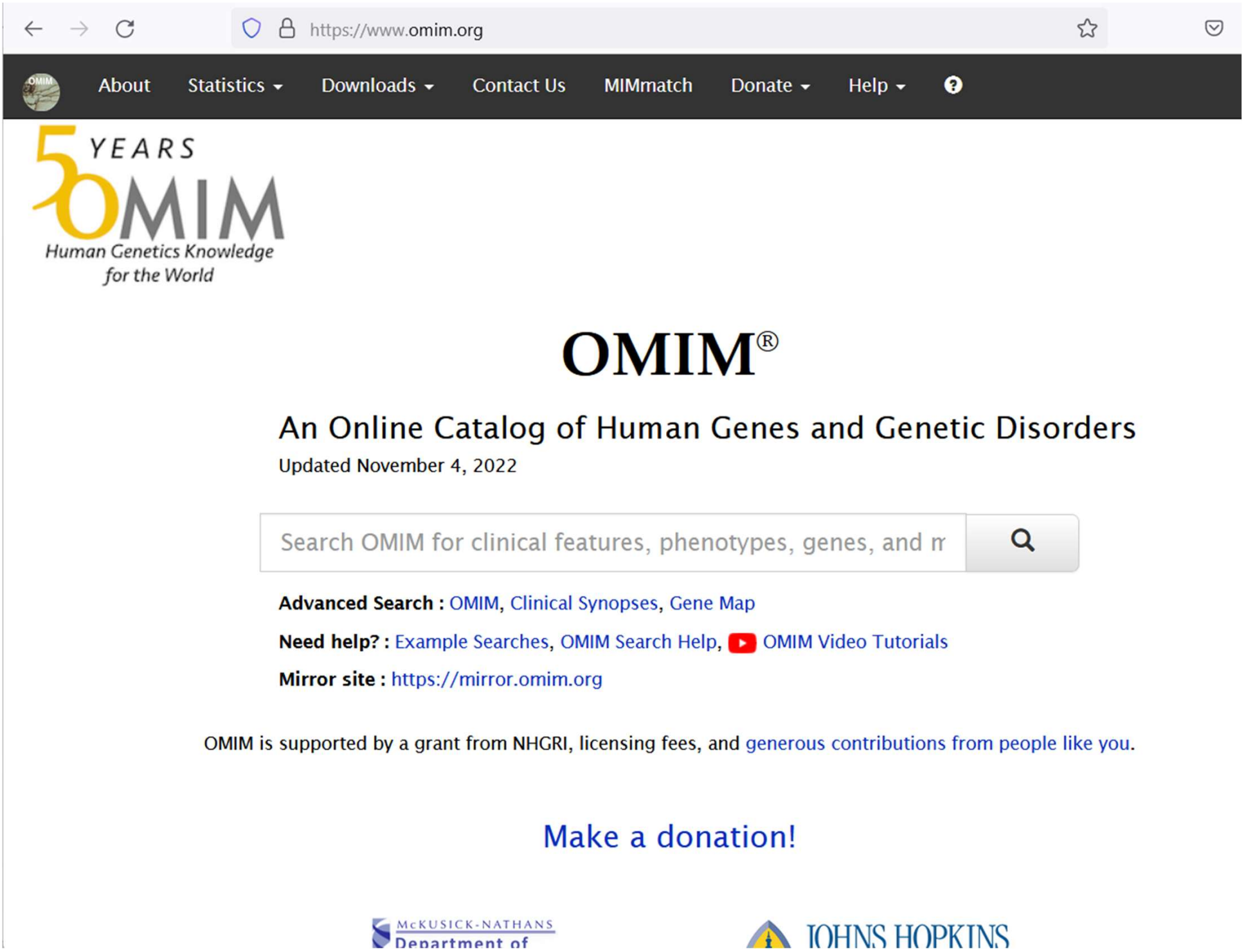

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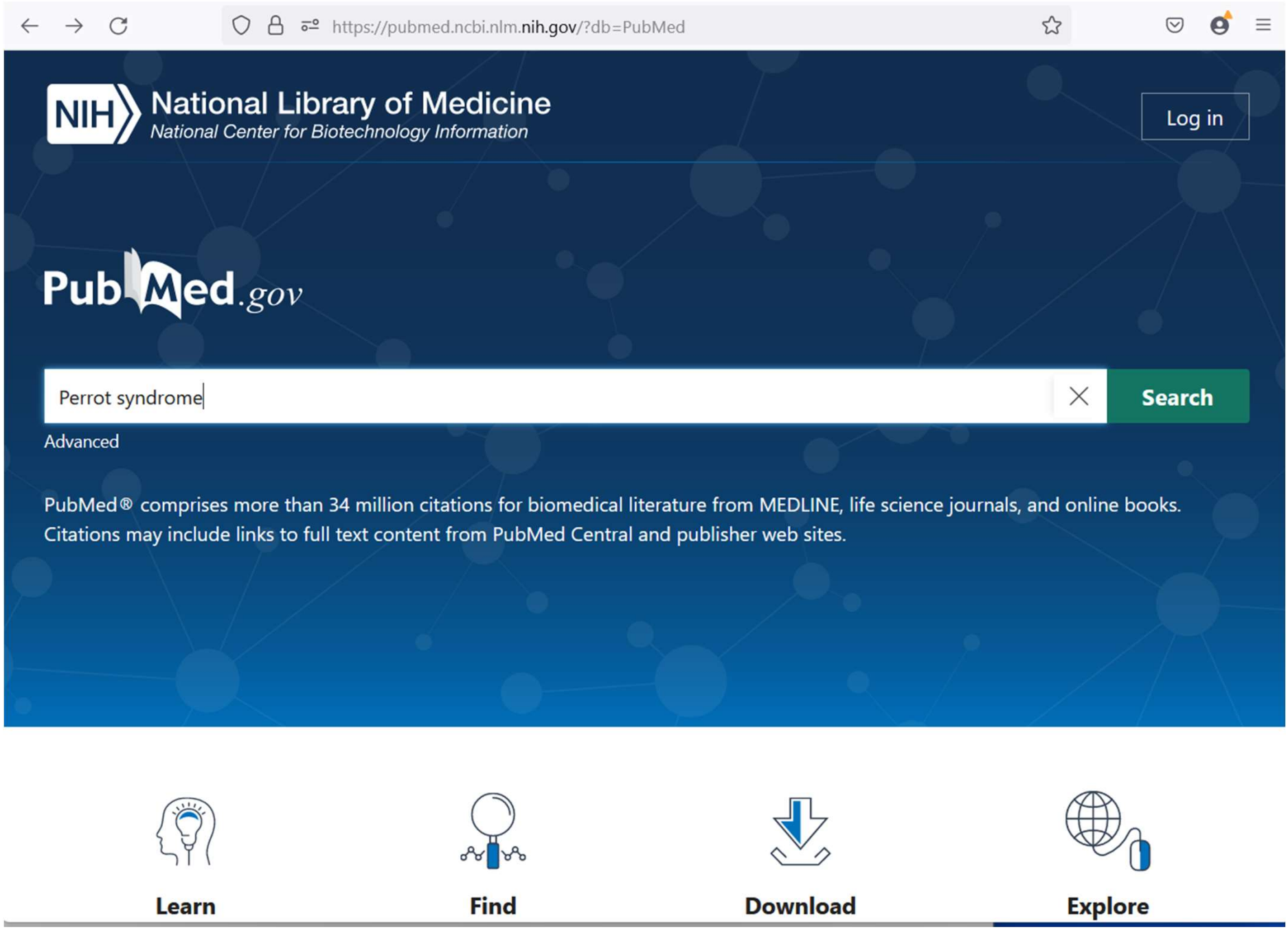

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Demain LA[1], Urquhart JE[1], O'Sullivan J[1], Williams SG[1], Bhaskar SS[1], Jenkinson EM[1], Lourenco CM[2], Heiberg A[3], Pearce SH[4], Shalev SA[5,6], Yue WW[7], Mackinnon S[7], Munro KJ[8,9], Newbury-Ecob R[10], Becker K[11], Kim MJ[12], O' Keefe RT[13], Newman WG[1,9].

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Abstract

Perrault syndrome is a rare autosomal recessive disorder characterized by sensorineural hearing loss (SNHL) in both sexes and primary ovarian insufficiency in 46, XX karyotype females. Biallelic variants in five genes are reported to be causative: HSD17B4, HARS2, LARS2, CLPP and C10orf2. Here we present eight families affected by Perrault syndrome. In five families we

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MeSH Unique ID: D010049

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- Diseases, Ovarian
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Brenner Tumor
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Hereditary Breast and Ovarian Cancer Syndrome
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Thecoma
Ovarian Torsion
Primary Ovarian Insufficiency

Primary Ovarian Insufficiency

Cessation of ovarian function after MENARCHE but before the age of 40, without or with OVARIAN FOLLICLE depletion. It is characterized by the presence of OLIGOMENORRHEA or AMENORRHEA, elevated GONADOTROPINS, and low ESTRADIOL levels. It is a state of female HYPERGONADOTROPIC HYPOGONADISM. Etiologies include genetic defects, autoimmune processes, chemotherapy, radiation, and infections. The most commonly known genetic cause is the expansion of a CGG repeat to 55 to 199 copies in the 5' untranslated region in the X-linked FMR1 gene.

Year introduced: 2011(1992)

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Primary Ovarian Insufficiency

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Year introduced: 2011(1992)

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- ☐ microbiology
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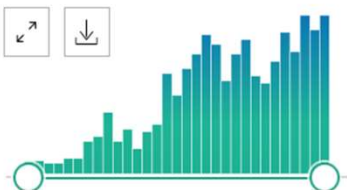

1 ☐ [Clinical significance of serum miRNA-146, OX-LDL and ROS expression in patients with primary ovarian insufficiency].

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Zhonghua Yi Xue Yi Chuan Xue Za Zhi. 2022 Nov 10;39(11):1211-1215. doi: 10.3760/cma.j.cn511374-20211018-00823.

PMID: 36317205 Chinese.