



DIU Génétique et Reproduction

Banques de données et serveurs biomoléculaires et génétiques

18 Novembre 2022
Philippe Dessen
Institut Gustave Roussy, Villejuif

dessen@igr.fr

<http://pdessen.free.fr/KB>

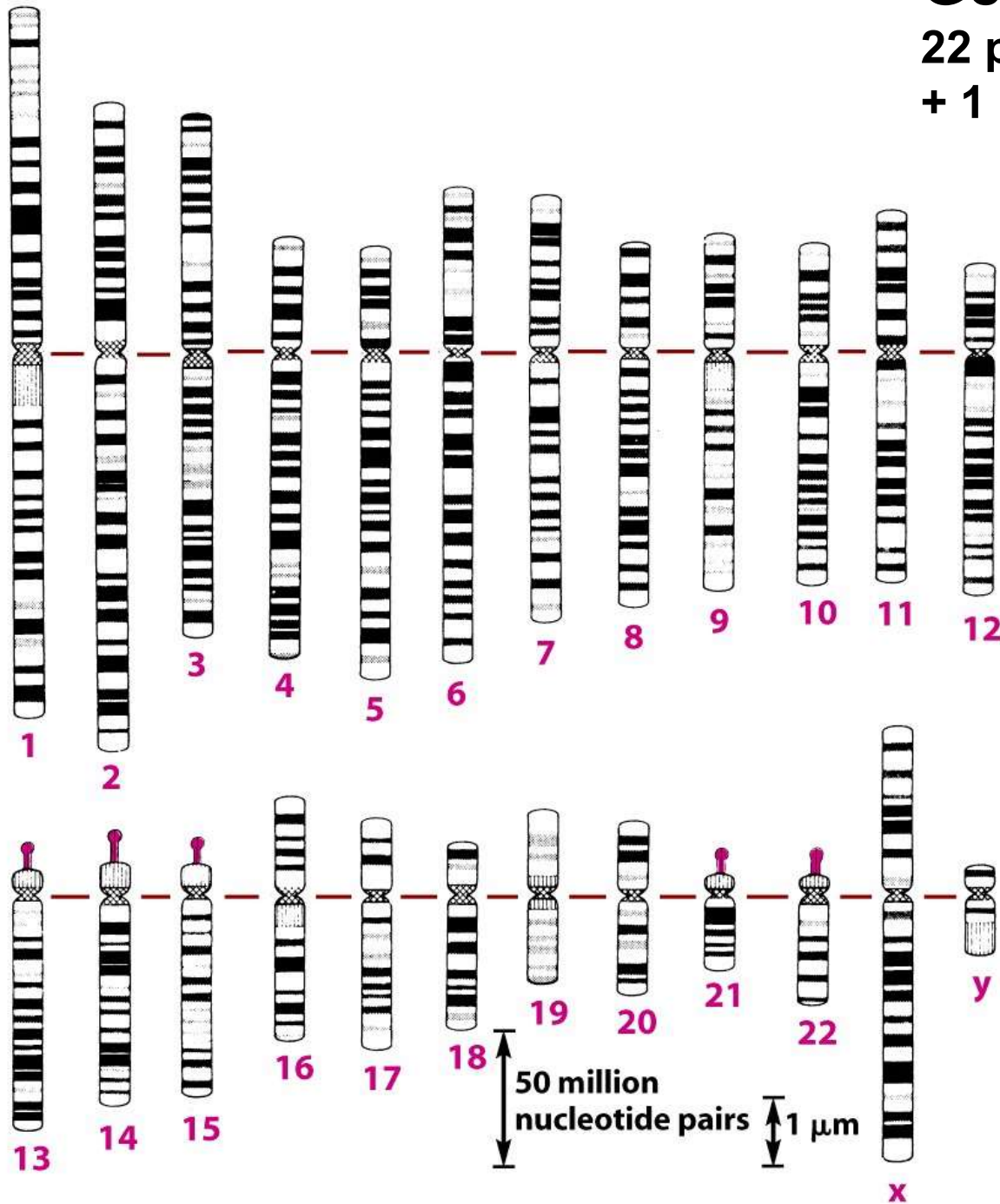
Abdelkader Heddar
Hôpital Bicêtre

abdelkader.heddar@aphp.fr

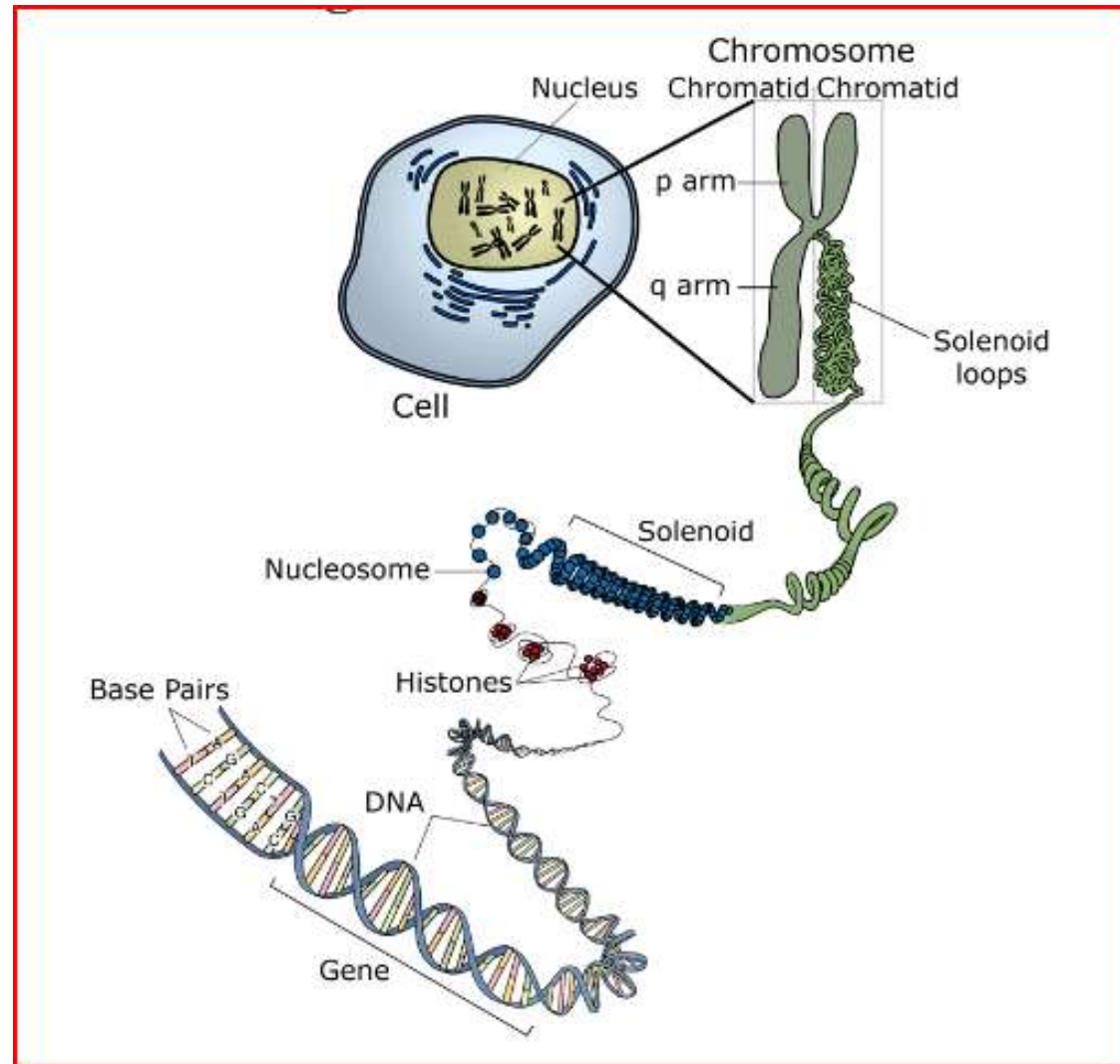
Génome humain

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

The International Nucleotide Sequence Database Collaboration

Guy Cochrane^{1,*}, Ilene Karsch-Mizrachi², Toshihisa Takagi³ and International Nucleotide Sequence Database Collaboration

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

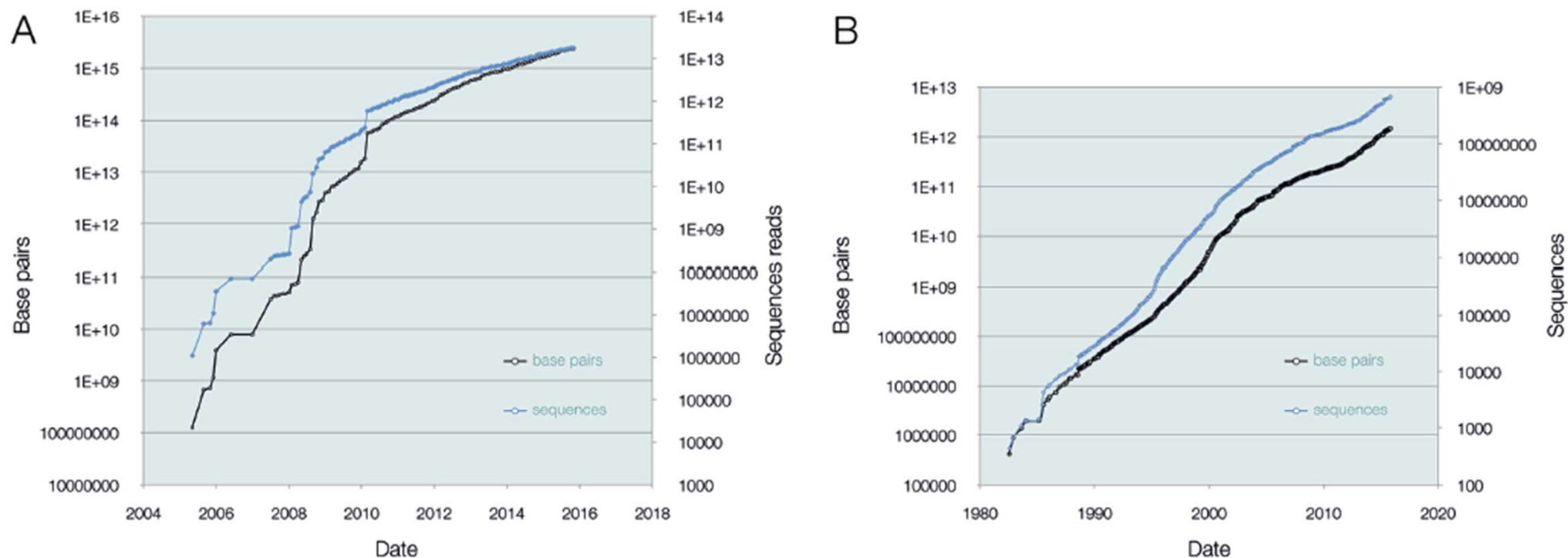
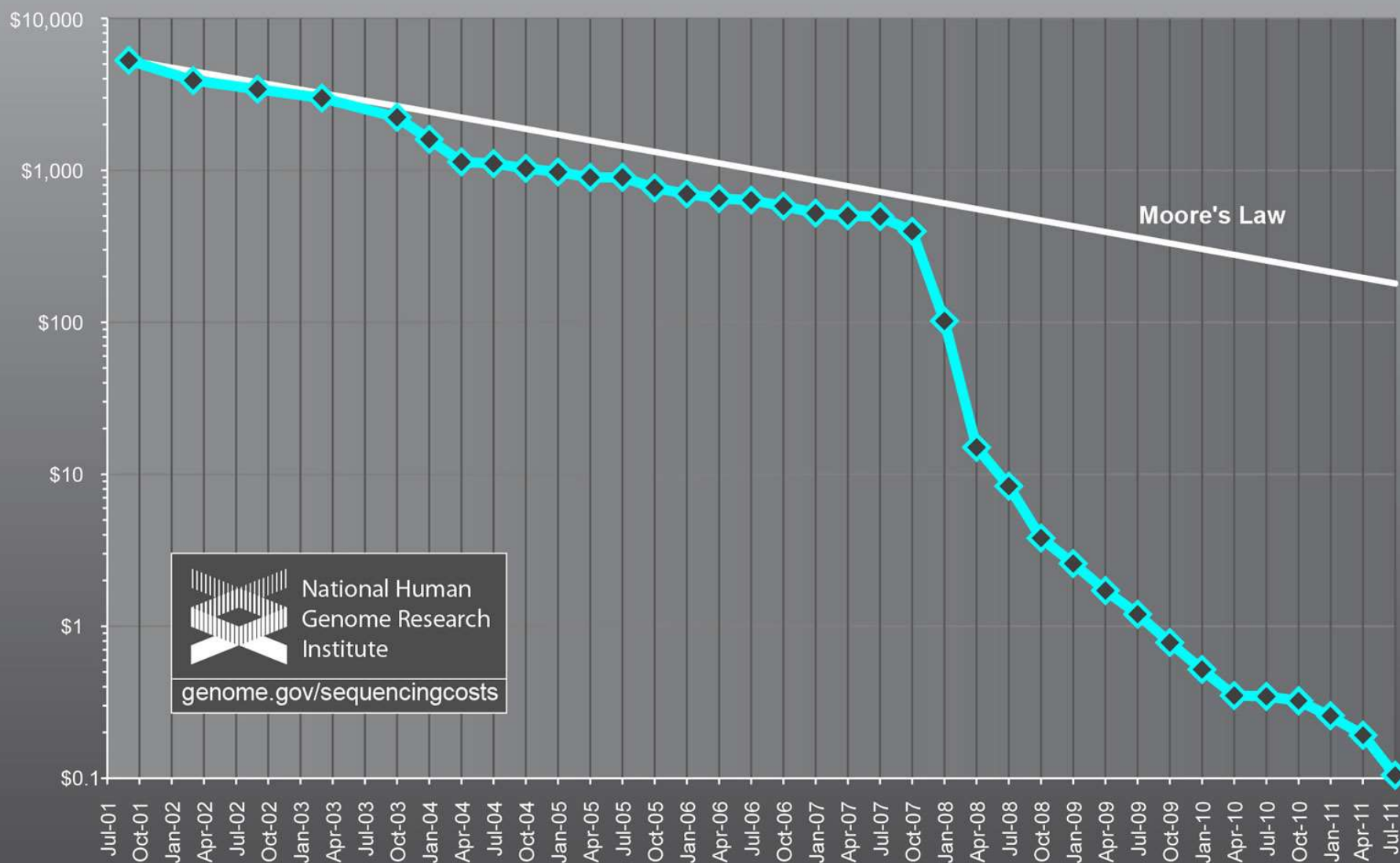


Figure 1. Cumulative growth in INSDC. (A) Base pairs (black, 2365.5 trillion) and sequence reads (blue, 17.8 trillion) for INSDC raw data. (B) Base pairs (black 1449 billion) and sequences (blue, 651.5 million) in INSDC assembled/annotated data.

Cost per Megabase of DNA Sequence



La complexité et les relations entre banques

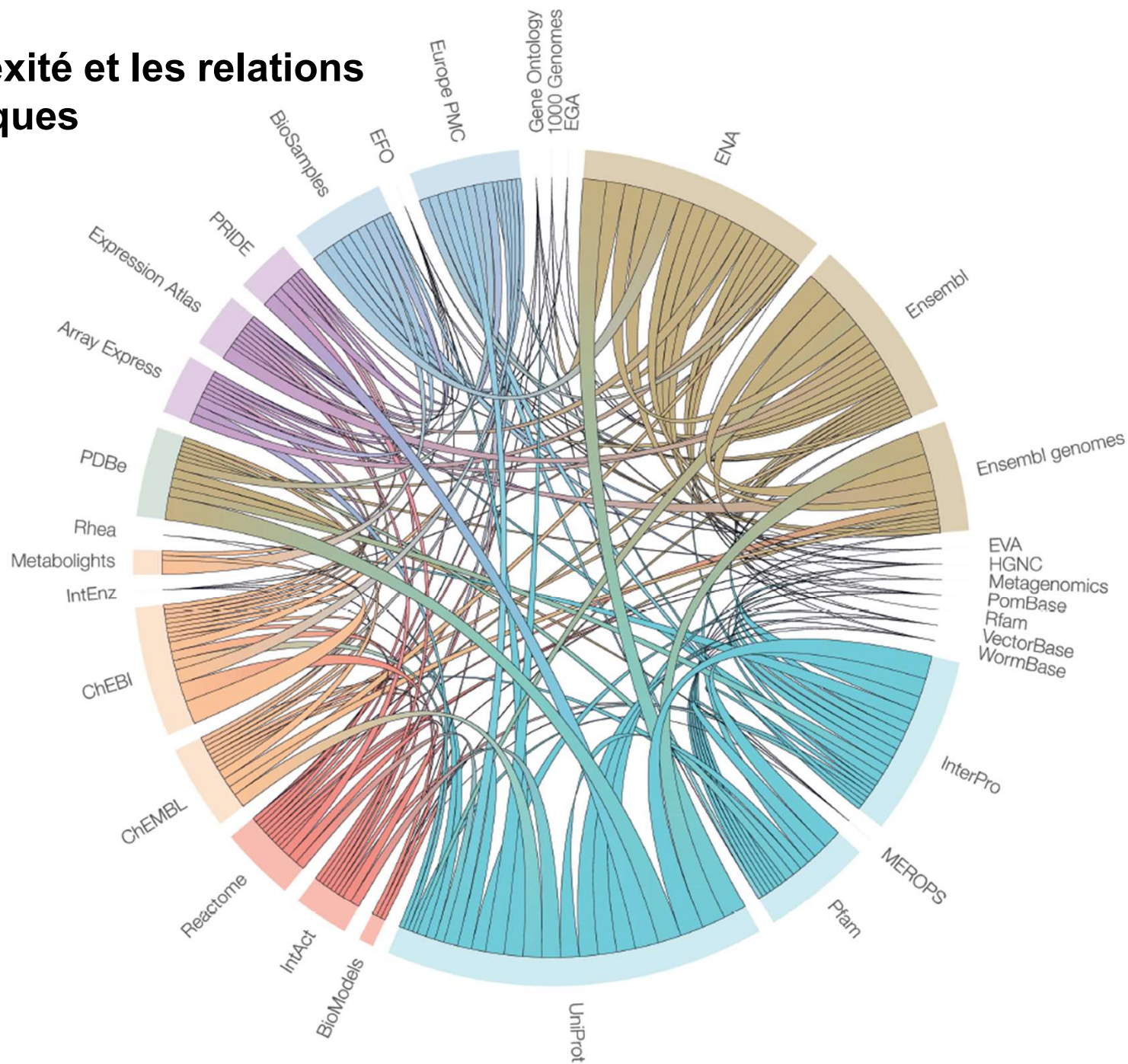


Figure 3. Representation of the internal interactions between different databases and resources at the EMBL-EBI, as determined by the exchange of data. All resources are placed on the circumference of the circle, with each resource represented by an arc proportional to the total number of interactions. 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 exchanged 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 (Jan 2015).

Ressources Internet et Données de génomes

French sites

[Bioinfo GR](#) [Gustave Roussy Github](#)

[Institut Français de bioinformatique](#) [Plateformes](#) [Outils](#) [Données](#) [IBiSA](#) [ReNaBI](#) [AVIESAN](#) [France-Genomique](#) [Plan France Médecine génomique 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

[EMBNNet](#) [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](#)
[Discover](#) [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 Médecine génomique2025](#) [St-Jude-cloud](#) [Cohorts](#) [Regulome](#)
[Harmonizome](#) [Oncogenomic landscape](#) [CancerTool](#) [Pedcbioportal](#) [ITCC-P4](#) [OncoSG](#) [Synapse](#) [Cancer genetic commons](#) [VICC](#) [ALPHA](#) [Pediatric genomic data](#)
[HuVarBase](#)

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)

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

**GOLD**

GENOMES ONLINE DATABASE

[JGI HOME](#) [LOG IN](#)[Home](#) [Search](#) [Distribution Graphs](#) [Biogeographical Metadata](#) [Statistics](#) [References](#) [Team](#) [Help](#) [News](#)

Studies ⁱ	22 232
Biosamples ⁱ	69 152
Sequencing Projects ⁱ	69 468
Analysis Projects ⁱ	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 sequencing projects, and their associated metadata, around the world.

1. Register	2. Annotate	3. Publish
 <p>Register your project information and Metadata in the Genomes Online Database</p> <p>Register</p>	 <p>Annotate your microbial genome or metagenome with IMG/ER or IMG/MER</p> <p>Annotate</p>	 Standards in Genomic Sciences <p>Publish your genome or metagenome in open access standards-supportive journal.</p> <p>Publish</p>

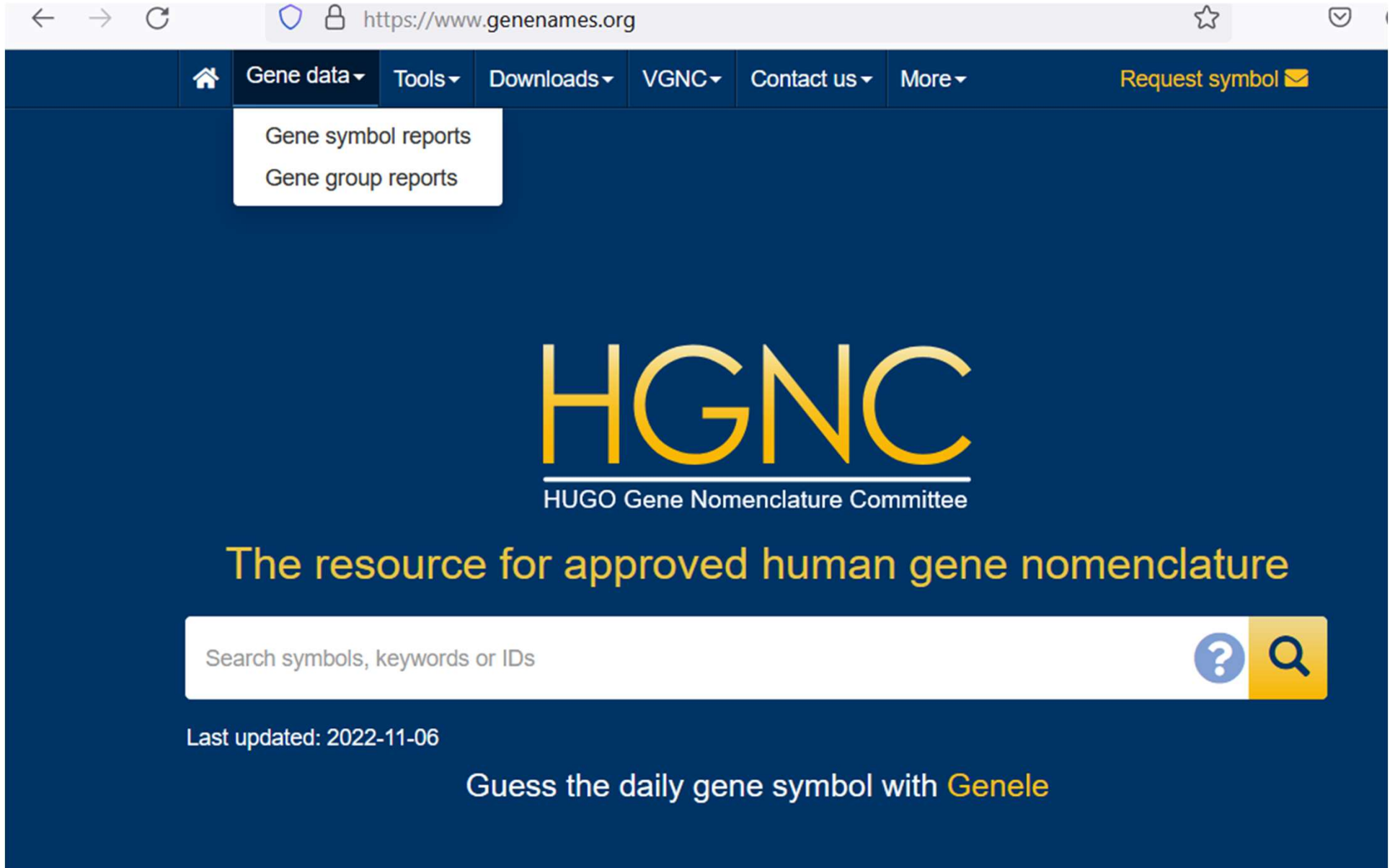
Studies Metagenomic 604 Non-Metagenomic 21 628	Biosamples  Classification Ecosystems Host-associated 12 732 Engineered 2 070 Environmental 8 516	Sequencing Projects  Complete Projects 7 435  Permanent Drafts 25 944  Incomplete Projects 32 976  Targeted Projects 2 122	Analysis Projects Genome Analysis 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 ⁱ 2 239 HMP Projects ⁱ 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)



The screenshot shows the HGNC website homepage. At the top is a dark blue navigation bar with a home icon, 'Gene data' (with a dropdown menu showing 'Gene symbol reports' and 'Gene group reports'), 'Tools', 'Downloads', 'VGNC', 'Contact us', and 'More'. A 'Request symbol' button with an envelope icon is on the right. The main content area has a dark blue background. In the center is the 'HGNC' logo in large yellow letters, with 'HUGO Gene Nomenclature Committee' in white text below it. Underneath the logo is the tagline 'The resource for approved human gene nomenclature' in yellow. A white search bar with the placeholder text 'Search symbols, keywords or IDs' is positioned below the tagline, featuring a question mark icon and a magnifying glass icon. At the bottom left, it says 'Last updated: 2022-11-06'. At the bottom center, it says 'Guess the daily gene symbol with Genele'.

← → ↻ <https://www.genenames.org> ☆

Home Gene data ▾ Tools ▾ Downloads ▾ VGNC ▾ Contact us ▾ More ▾ Request symbol ✉

Gene symbol reports
Gene group reports

HGNC

HUGO Gene Nomenclature Committee

The resource for approved human gene nomenclature

Search symbols, keywords or IDs ? 🔍

Last updated: 2022-11-06

Guess the daily gene symbol with Genele

<http://www.genenames.org/>



Gene Symbol Report ⁱ

INSR

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

GENE FAMILY ⁱ

[CD molecules](#)
[Fibronectin type III domain containing](#)

SPECIALIST DATABASE ⁱ

[CD](#) ^C

HOMOLOGS ⁱ

[MGI:96575](#) ^C Mouse Symbol: **Insr**
[RGD:2917](#) ^D Rat Symbol: **Insr**
[HCOP](#) ^D
[TreeFam](#) ^D

NUCLEOTIDE SEQUENCES ⁱ

[GenBank:M10051](#) [EMBL-Bank](#) [DDBJ](#) ^C
[RefSeq:XM_005259552](#) ^D
[CCDS:CCDS12176.1](#) ^C

GENE RESOURCES ⁱ

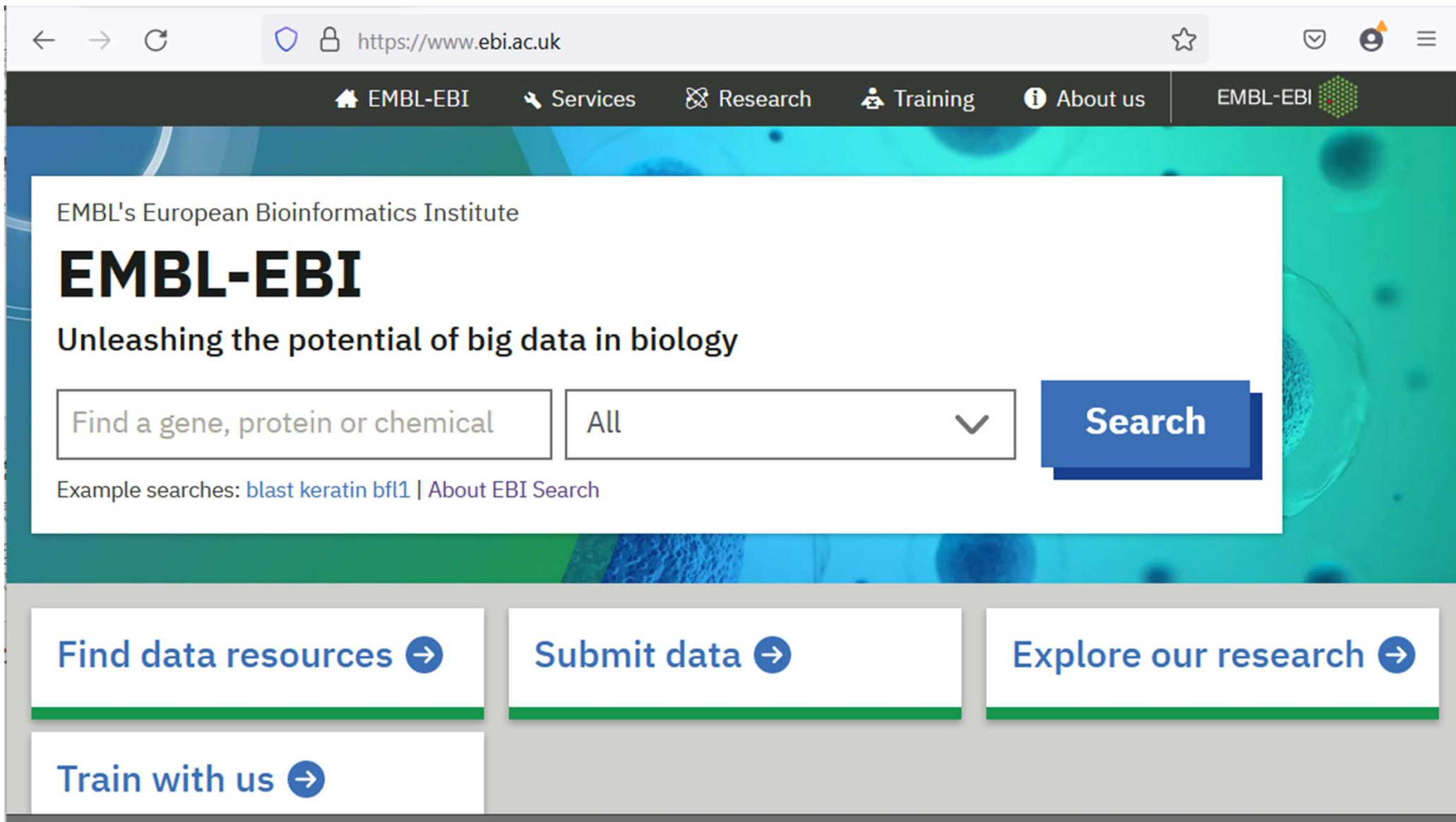
[Entrez Gene:3643](#) ^D [NCBI Sequence Viewer](#)
[Ensembl:ENSG00000171105](#) [Ensembl Genome Browser](#)
^D [UCSC Genome Browser](#)
[UCSC:uc002mgd.1](#) ^D

PROTEIN RESOURCES ⁱ

[UniProtKB:P06213](#) ^D
[InterPro](#) ^D

[OMIM](#) ^D
[GeneTests](#) ^D

European Bioinformatic Institute (EBI)



The image shows the homepage of the European Bioinformatics Institute (EBI). The browser address bar displays <https://www.ebi.ac.uk>. The navigation bar includes links for EMBL-EBI, Services, Research, Training, and About us. The main content area features the EMBL-EBI logo and the tagline "Unleashing the potential of big data in biology". A search bar is present with the placeholder text "Find a gene, protein or chemical" and a dropdown menu set to "All". A blue "Search" button is located to the right of the search bar. Below the search bar, example searches are listed: "blast keratin bfl1" and "About EBI Search". At the bottom of the page, there are four buttons: "Find data resources", "Submit data", "Explore our research", and "Train with us", each with a right-pointing arrow icon.

EMBL's European Bioinformatics Institute

EMBL-EBI

Unleashing the potential of big data in biology

Find a gene, protein or chemical

All

Search

Example searches: [blast keratin bfl1](#) | [About EBI Search](#)






Find data resources →








Submit data →

Explore our research →

Train with us →

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

← → ↻  <https://www.ebi.ac.uk/services>     ☰

 EMBL-EBI  Services  Research  Training  About us  EMBL-EBI 

The EMBL-EBI website has been redesigned. Please [send us feedback](#) about this page.

Services

Data resources and analysis tools to support life science research

EMBL's European Bioinformatics Institute (EMBL-EBI) maintains the world's most comprehensive range of freely available and up-to-date molecular data resources.

Find a data resource or a tool

Search

Explore all our data resources and tools →

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

Type

- ☐ Data resources
- ☐ Tools

Category

- ☐ Chemical biology
- ☐ Cross domain
- ☒ 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.



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

insulin receptor

Examples: histone, BN000065

Search 🔍

Enter accession

Examples: Taxon:9606, BN000065, PRJEB402

View 📄

Home

Submit ▼

Search ▼

Rulespace

About ▼

Support ▼

We recommend that you subscribe to the [ENA-announce mailing list](#) for updates on services.

For SARS-CoV-2 data submissions, users should contact us in advance of submission at virus-dataflow@ebi.ac.uk for specific advice on options and to access the highest levels of support.

We have also launched a [Drag-and-Drop Data Submission Service](#) (currently in Beta) suitable for certain SARS-Cov-2 submissions. We are inviting submitters to try this out. Please contact us at the email above for details.

Examples: histone, BN000065

Search 

Examples: Taxon:9606, BN000065, PRJEB402

View 

Home

Submit ▼

Search ▼

Rulespace

About ▼

Support ▼

Text Search

Uses [EBI Search](#) to perform a free text search across ENA data. For more detailed usage please refer to the [help & documentation section](#).

Search term:

Search 

Search results for **insulin receptor**

• Sequence

- [Sequence](#) (3,140)
- [Sequence \(Standard\)](#) (3,140)

Sequence [View all 3,140 results.](#)

[J05043](#)

Human insulin receptor (IR) gene, exon 1.

• Coding

- [Coding](#) (3,675)
- [Coding \(CON\)](#) (464)

Sequence (Standard) [View all 3,140 results.](#)

[J05043](#)

Human insulin receptor (IR) gene, exon 1.

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

UniProt

← → ↻ <https://www.uniprot.org>

UniProt [BLAST](#) [Align](#) [Peptide search](#) [ID mapping](#) [SPARQL](#) Release 2022_04 | [Statistics](#) [Help](#)

Find your protein

UniProtKB ▾ | Advanced | List [Search](#)

Examples: Insulin, APP, Human, P05067, organism_id:9606

UniProt is the world's leading high-quality, comprehensive and freely accessible resource of protein sequence and functional information. [Cite UniProt](#)”

[Feedback](#)

Proteins

UniProt Knowledgebase

Reviewed
(Swiss-Prot)
568,363

Unreviewed
(TrEMBL)
229,928,140

Species

Proteomes

Protein sets for species with sequenced genomes from across the tree of life

Protein Clusters

UniRef

Clusters of protein sequences at 100%, 90% & 50% identity

Sequence Archive

UniParc

Non-redundant archive of publicly available protein sequences seen across different databases

[Help](#)

<http://www.uniprot.org/>



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 ▴	Length
<input type="checkbox"/> P06213 📁	INSR_HUMAN	Insulin receptor[...]	INSR	Homo sapiens (Human)	1,380
<input type="checkbox"/> Q9PVZ4 📁	INSR_XENLA	Insulin receptor[...]	insr	Xenopus laevis (African clawed frog)	1,360
<input type="checkbox"/> P15208 📁	INSR_MOUSE	Insulin receptor[...]	Insr	Mus musculus (Mouse)	1,380
<input type="checkbox"/> P15127 📁	INSR_RAT	Insulin receptor[...]	Insr	Rattus norvegicus (Rat)	1,380
<input type="checkbox"/> Q28516 📁	INSR_MACMU	Insulin receptor[...]	INSR	Macaca mulatta (Rhesus macaque)	2,100
<input type="checkbox"/> P09208 📁	INSR_DROME	Insulin-like receptor[...]	InR, dinr, Dir-a,	Drosophila	2,140

Feedback

Help

UniProtKB - P06213 (INSR_HUMAN)

Protein **Insulin receptor**

Gene **INSR**

Organism *Homo sapiens (Human)*

Sequence features View only features (sites, domains, PTMs ...)

Status Annotation score: Experimental evidence at protein levelⁱ

Display

Function

- ☒ Function
- ☒ Names & Taxonomy
- ☒ Subcellular location
- ☒ Pathology & Biotech
- ☒ PTM / Processing
- ☒ Expression
- ☒ Interaction
- ☒ Structure
- ☒ Family & Domains
- ☒ Sequences (2)
- ☒ Cross-references
- ☒ Publications
- ☒ Entry information
- ☒ Miscellaneous
- ☒ Similar proteins

[▲ Top](#)

Functionⁱ

Receptor tyrosine kinase which mediates the pleiotropic actions of insulin. Binding of insulin leads to phosphorylation of several intracellular substrates, including, insulin receptor substrates (IRS1, 2, 3, 4), SHC, GAB1, CBL and other signaling intermediates. Each of these phosphorylated proteins serve as docking proteins for other signaling proteins that contain Src-homology-2 domains (SH2 domain) that specifically recognize different phosphotyrosines residues, including the p85 regulatory subunit of PI3K and SHP2. Phosphorylation of IRSs proteins lead to the activation of two main signaling pathways: the PI3K-AKT/PKB pathway, which is responsible for most of the metabolic actions of insulin, and the Ras-MAPK pathway, which regulates expression of some genes and cooperates with the PI3K pathway to control cell growth and differentiation. Binding of the SH2 domains of PI3K to phosphotyrosines on IRS1 leads to the activation of PI3K and the generation of phosphatidylinositol-(3, 4, 5)-trisphosphate (PIP3), a lipid second messenger, which activates several PIP3-dependent serine/threonine kinases, such as PDK1 and subsequently AKT/PKB. The net effect of this pathway is to produce a translocation of the glucose transporter SLC2A4/GLUT4 from cytoplasmic vesicles to the cell membrane to facilitate glucose transport. Moreover, upon insulin stimulation, activated AKT/PKB is responsible for: anti-apoptotic effect of insulin by inducing phosphorylation of BAD; regulates the expression of gluconeogenic and lipogenic enzymes by controlling the activity of the winged helix or forkhead (FOX) class of transcription factors. Another pathway regulated by PI3K-AKT/PKB activation is mTORC1 signaling pathway which regulates cell growth and metabolism and integrates signals from insulin. AKT mediates insulin-stimulated protein synthesis by phosphorylating TSC2 thereby activating mTORC1 pathway. The Ras/RAF/MAP2K/MAPK pathway is mainly involved in mediating cell growth, survival and cellular differentiation of insulin. Phosphorylated IRS1 recruits GRB2/SOS complex, which triggers the activation of the Ras/RAF/MAP2K/MAPK pathway. In addition to binding insulin, the insulin receptor can bind insulin-like growth factors (IGF1 and IGFII). Isoform **Short** has a higher affinity for IGFII binding. When present in a hybrid receptor with IGF1R, binds IGF1. PubMed:12138094 shows that hybrid receptors composed of IGF1R and INSR isoform **Long** are activated with a high affinity by IGF1, with low affinity by IGF2 and not significantly activated by insulin, and that hybrid receptors composed of IGF1R and INSR isoform **Short** are activated by IGF1, IGF2 and insulin. In contrast, PubMed:16831875 shows that hybrid receptors composed of IGF1R and INSR isoform **Long** and hybrid receptors composed of IGF1R and INSR isoform **Short** have similar binding characteristics, both bind IGF1 and have a low affinity for insulin. 7 Publications

Catalytic activityⁱ

ATP + a [protein]-L-tyrosine = ADP + a [protein]-L-tyrosine phosphate. PROSITE-ProRule annotation 8 Publications

Enzyme regulationⁱ

Activated in response to insulin. Autophosphorylation activates the kinase activity. PTPN1, PTPRE and PTPRF dephosphorylate important tyrosine residues, thereby reducing INSR activity. Inhibited by ENPP1. GRB10 and GRB14 inhibit the catalytic activity of the INSR, they block access of substrates to the activated receptor. SOCS1 and SOCS3 act as negative regulators of INSR activity, they bind to the activated INRS and interfere with the phosphorylation of INSR substrates. 5 Publications

Sites

Feature key	Position(s)	Length	Description	Graphical view	Feature identifier	Actions
Site ⁱ	66 – 66		1 Insulin-binding			
Binding site ⁱ	1033 – 1033		1 ATP PROSITE-ProRule annotation 1 Publication			
Binding site ⁱ	1057 – 1057		1 ATP PROSITE-ProRule annotation 1 Publication			
Active site ⁱ	1159 – 1159		1 Proton donor/acceptor 1 Publication			
Binding site ⁱ	1177 – 1177		1 ATP PROSITE-ProRule annotation 1 Publication			

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

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

NCBI

[NCBI](#) [Resources](#) [How To](#) [Sign in to NCBI](#)

Genome [Limits](#) [Advanced](#) [Help](#)



Genome

This resource organizes information on genomes including sequences, maps, chromosomes, assemblies, and annotations.

Using Genome

- [Help](#)
- [Browse by Organism](#)
- [Download / FTP](#)
- [Download FAQ](#)
- [Submit a genome](#)

Genome Tools

- [BLAST the Human Genome](#)
- [Microbial Nucleotide BLAST](#)
- [TaxPlot \(3-way Genome Comparison\)](#)

Custom resources

- [Human Genome](#)
- [Microbes](#)
- [Organelles](#)
- [Viruses](#)
- [Prokaryotic reference genomes](#)

Genome Annotation and Analysis

- [Eukaryotic Genome Annotation](#)
- [Prokaryotic Genome Annotation](#)
- [PASC \(Pairwise Sequence Comparison\)](#)

Other Resources

- [Assembly](#)
- [BioProject](#)
- [BioSample](#)
- [Map Viewer](#)
- [Protein Clusters](#)

External Resources

- [GOLD - Genomes Online Database](#)
- [Ensembl Genome Browser](#)
- [Bacteria Genomes at Sanger](#)
- [Large-Scale Genome Sequencing \(NHGRI\)](#)

You are here: [NCBI](#) > [Genomes & Maps](#) > [Genome](#) [Write to the Help Desk](#)

GETTING STARTED <ul style="list-style-type: none">NCBI EducationNCBI Help ManualNCBI HandbookTraining & Tutorials	RESOURCES <ul style="list-style-type: none">Chemicals & BioassaysData & SoftwareDNA & RNADomains & Structures	POPULAR <ul style="list-style-type: none">PubMedBookshelfPubMed CentralPubMed Health	FEATURED <ul style="list-style-type: none">Genetic Testing RegistryPubMed HealthGenBankReference Sequences	NCBI INFORMATION <ul style="list-style-type: none">About NCBIResearch at NCBINCBI NewsNCBI FTP Site
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<http://www.ncbi.nlm.nih.gov/genome>

Gene sources
Genomic

Categories
Alternatively spliced
Annotated genes
Non-coding
Protein-coding

Sequence content
CCDS
Ensembl
RefSeq
RefSeqGene

Status clear
☒ **Current**

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[Show additional filters](#)

Tabular ▾ 20 per page ▾ Sort by Relevance ▾ Send to: ▾ Hide sidebar >>

Filters: [Manage Filters](#)

Results by taxon

Top Organisms [\[Tree\]](#)
Homo sapiens (211)
Mus musculus (49)
Rattus norvegicus (28)
Salmo trutta (7)
Oncorhynchus mykiss (7)
All other taxa (1393)
[More...](#)

Find related data

Database: ▾

Search details

INSR[All Fields] AND

GENE Was this helpful?

[INSR – insulin receptor](#)

[Homo sapiens \(human\)](#)

Also known as: CD220, HHF5

Gene ID: 3643

[RefSeq transcripts \(4\)](#) [RefSeq proteins \(4\)](#) [RefSeqGene \(1\)](#) [PubMed \(712\)](#)

RefSeq Sequences

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

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):

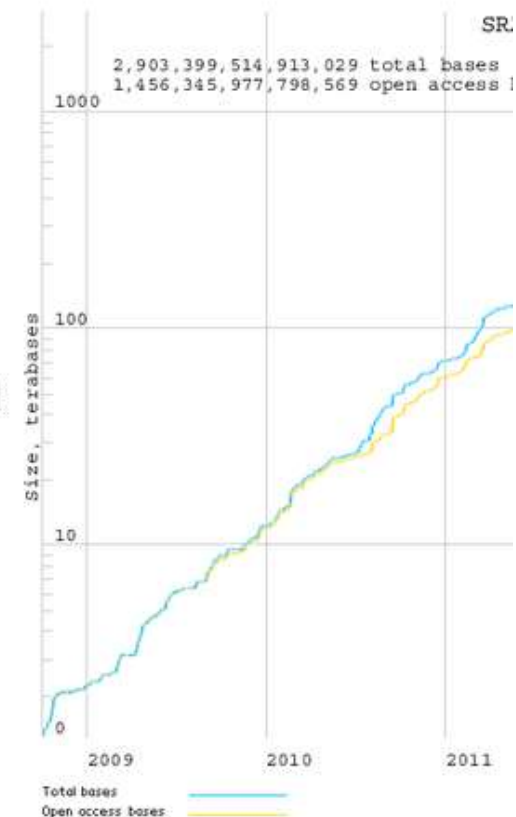
- [Email the Toolkit team for more information](#)

Search in SRA Documentation

Enter text or search terms to search: Go

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

Repository NGS sequencing



<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.

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

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

Scope: Format: Amount: GEO accession:

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

Relations

BioProject [PRJNA197343](#)

Analyze with GEO2R

Download family

[SOFT formatted family file\(s\)](#)
[MINiML formatted family file\(s\)](#)
[Series Matrix File\(s\)](#)

Format

[SOFT](#) [?](#)
[MINiML](#) [?](#)
[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/>



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/>



Browse/Select Species

POPULAR SPECIES



Human



Mouse



Rat



Zebrafish



Fruitfly



Worm



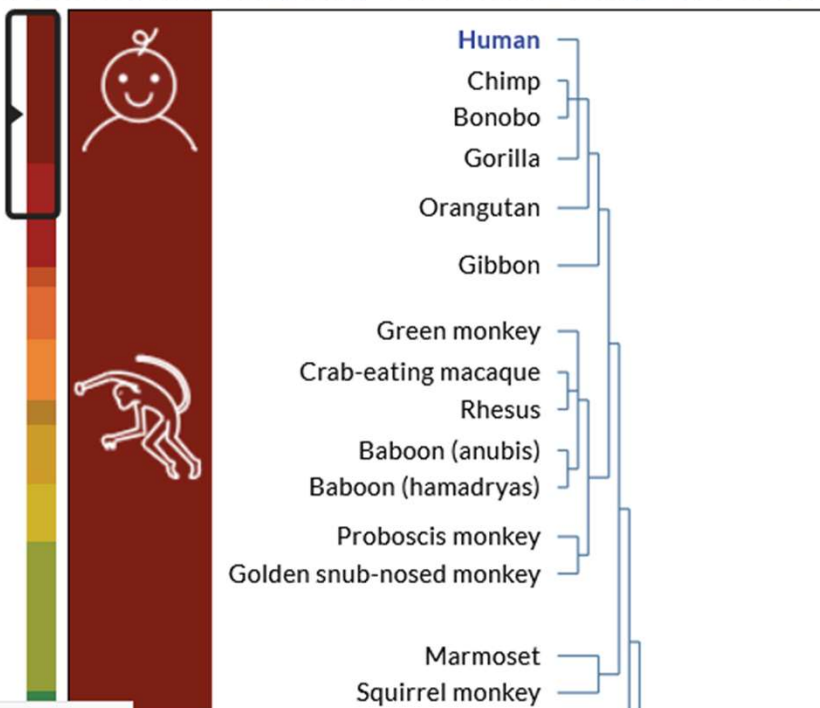
Yeast

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)

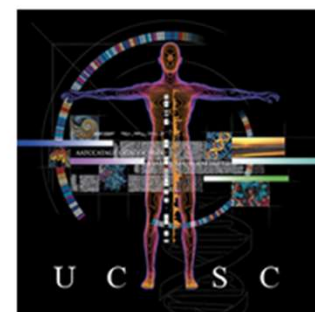
Assembly date: Dec. 2013 initial release; Dec. 2017 patch release 13

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)

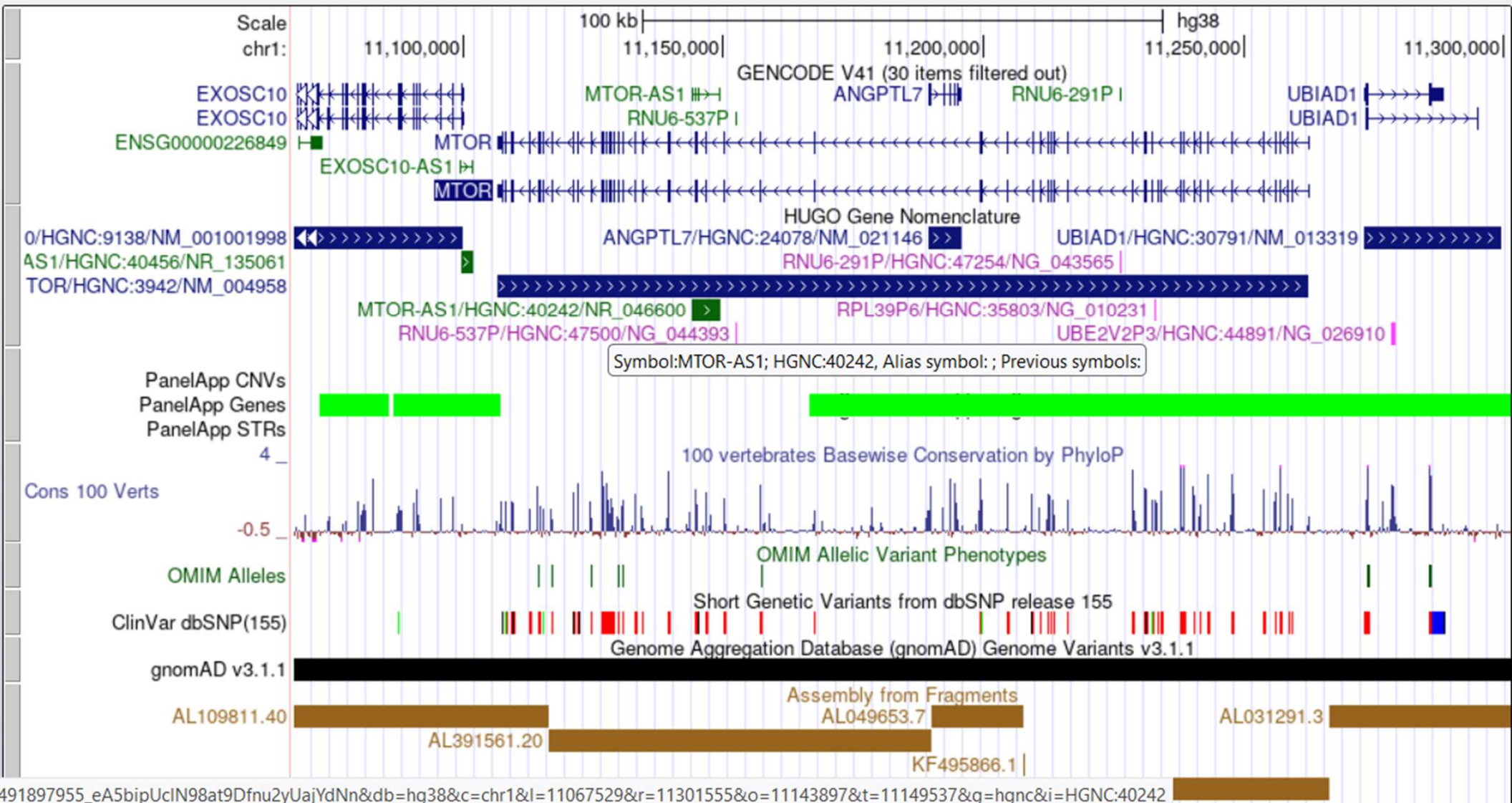
Search the assembly:

- **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

UCSC Genome Browser on Human (GRCh38/hg38)

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

multi-region chr1:11,067,530-11,301,555 234,026 bp. gene, chromosome range, or other position, see exampl go [examples](#)



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 is 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 carbohydrates, 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 receptor

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 bases
- ☒ 5' UTR Exons
- ☒ CDS Exons
- ☒ 3' UTR Exons
- ☒ Introns
- ☐ Downstream by bases
- ☒ One FASTA record per gene.
- ☐ One FASTA record per region (exon, intron, etc.) with extra bases upstream (5') and 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 truncated to 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

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

GAGAAAGGACGCGCGGCCCCAGCGCCTCTTGGGTGGCCGCCTCGGAGCAT
GACCCCCGCGGGCCAGCGCCGCGCTCTGATCCGAGGAGACCCCGCGCT
CCCGCAGCCATGGCCACCGGGGGCCGGCGGGGGGCGGCGCCGCGCCGCT
GCTGGTGGCGGTGGCCGCGCTGCTACTGGGCGCCGCGGGCCACCTGTACC
CCGGAAGAGGgtgagtctggggggcgggcggtggggcggggagcgccgcgat
gggggagaggacccccacccaagccaaaatcgagcccccgcttgtggactga
gaaccctccccaggggcgggggggcggtggccaggacggtagctcctgcat
cgcgtagggggagcggaagcctctgaccttggcctttgcccggccgggc
tcgcgcctccgcgccctgcgtgccccgacctgagccccgaggaaccttgccc

1er exon 5'UTR

--- - - - -
aagaaaacaccatctctcagcacctctagcaatgctatttctgccttgac
aattgtccacacacacagccagcatctgatgtgagcacatgtggtcggtg
ggtgtcgggtgcagggacaagagtgggggtttgggaggatgcgtggcagg
gccccagactcaccaggaagtgctccttctgccccgcagCACTGACCTC
ATGCGCATGTGCTGGCAATTCAACCCCAAGATGAGGCCAACCTTCCTGGA
GATTGTCAAACCTGCTCAAAGGACGACCTGCACCCCAGCTTTCCAGAGGTGT
CGTTCTTCCACAGCGAGGAGAACAAAGGCTCCCGAGAGTGAGGAGCTGGAG
ATGGAGTTTGAGGACATGGAGAATGTGCCCTGGACCGTTCTCTCGCACTG
TCAGAGGGAGGAGGCGGGGGCCGGGATGGAGGGTCTCTCGCTGGGTTTCA
AGCGGAGCTACGAGGAACACATCCCTTACACACACATGAACGGAGGCAAG
AAAAACGGGCGGATTCTGACCTTGCCTCGGTCCAATCCTTCCTAACAGTG
CCTACCGTGGCGGGGGCGGGCAGGGGTTCCCATTTTTCGCTTTCCTCTGGT
TTGAAAGCCTCTGGAAAACTCAGGATTCTCAGACTCTACCATGTCCAAT
GGAGTTCAGAGATCGTTTCTATACATTTCTGTTCATCTTAAGGTGGACTC
GTTTGTTTACCAATTTAACTAGTCCTGCAGAGGATTTAACTGTGAACCTG
GAGGGCAAGGGGTTTCCACAGTTGCTGCTCCTTTGGGGCAACGACGGTTT
CAAACCAGGATTTTGTGTTTTTTCGTTCCCCCACC CGCCCCCAGCAGAT
GGAAAGAAAGCACCTGTTTTTACAAATTCTTTTTTTTTTTTTTTTTTTT
GCTGGTGTCTGAGCTTCAGTATAAAAGACAAAACCTTCTGTGTGTGGAAC
AAAAGTTCGAAAGAAAAAACAAAAACAAAAACACCCAGCCCTGTTCCAGGA
GAATTTCAAAGTTTTTACAGGTTGAGCTTCAAAGATGGTTTTTTTTTGTTTTTT
TTTTTTCTCTCATCCAGGCTGAAGGATTTTTTTTTTTCTTTACAAAATGAG
TTCCTCAAATTGACCAATAGCTGCTGCTTTCATATTTTGGATAAGGGTCT
GTGGTCCCGCGTGTGCTCACGTGTGTATGCACGTGTGTGTGTCCATTAG
ACACGGCTGATGTGTGTGCAAAAGTATCCATGCGGAGTTGATGCTTTGGGA
ATTGGCTCATGAAGGTTCTTCTCAAAGGTGCGAGCTCATCCCCCTCTCTC
CTTCCTTCTTATTGACTGGGAGACTGTGCTCTCGACAGATTCTTCTTG
TCAGAAAGTCTAGCCTCAGGTTTCTACCCTCCCTTCACATTGGTGGCCAAAG
GGAGGAGCATTTTCATTTGGAGTGATTATGAATCTTTTCAAAGACCAAAACCA
AGCTAGGACATTAAAAAAAAAAAAAAAAAGAAAAAGAAAAAGAAAAACAAAATGG
AAAAAGGAAAAAAAAAAAAAGAACTGAGATGACAGAGTTTTGAGAATATATT
TGTACCATATTTAATTTTTTAAAGTCTCTGGTATTAGCCTCATAAGTTATT
GACTATTCCCCGGGTTTGGCGGGGAGTGGGGACATGAGTTGGTCTGCCTG
TTGTGGGGCCGGGAAGGGGAGGAGTCAGGCACAAGTGGCCTCTTTGTTT
GGTCTTAAAGGCATCCATTTCTGGGAATGAAGCCATGTTTCGCTGCTAAACA

dernier exon 3'UTR

Sequence à localiser

```
CGCGCGCTCTGATCCGAGGAGACCCCGCGCTCCCGCAGCCATGGCCACCG GGGGCCGGCGGGGGGCGGCGGCCGCGCCGCTGCTGGTGGCGGT  
GGCCGCG CTGCTACTGGGCGCCGCGGGCCACCTGTACCCCGGAGAGGgtgagtctgg gggcgcgggcggtggcggggagcgccgcatggggagaggacccaccca agcca  
aaatcgagcccccgcttgtggactgagaacctccccaggggcg gggcggtggccaggacggtagctcctcatcgcttagggggagcgggaa gcctctgaccttggccttggccgcccgggctcgcgctccgc  
gccctgc gtgccgacctgagcccaggaaccttggccggtgccgccccgcccgc ggctcctcttgagcgcgccctccgacctgtccccggccctccgagcc
```

Tools :: BLAT (hg38) : submit

← → ↻

🔒 genome.ucsc.edu/cgi-bin/hgBlat?hgsid=1491899397_aKaGbzsS4X40iS7mtG9oiLcResng&comr

📄 ☆ 📧 👤 ☰

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

Human BLAT Search

BLAT Search Genome

Genome: ☐ Search all
Human ▾

Assembly: Dec. 2013 (GRCh38/hg38) ▾

Query type: BLAT's guess ▾

Sort output: query,score ▾

Output type: hyperlink ▾

CGCGCGCTCTGATCCGAGGAGACCCCGCGCTCCCGCAGCCATGGCCACCG
GGGGCCGGCGGGGGGCGGCGGCCGCGCCGCTGCTGGTGGCGGTGGCCGCG
CTGCTACTGGGCGCCGCGGGCCACCTGTACCCCGGAGAGGgtgagtctgg
ggcgcgggcggtggcggggagcgccgcatggggagaggacccaccca
agccaaaatcgagcccccgcttgtggactgagaacctccccaggggcg
ggcgcggtggccaggacggtagctcctgcctgc
gcctctgaccttggccttggccgcccgggctc
gtgccgacctgagcccaggaaccttggccgc
ggctcctcttgagcgcgccctccgacctgtgc

BLAT Search Results

Go back to [chr19:7112255-7293931](#) on the Genome Browser.

Custom track name:

Custom track description:

☐ All Results (no minimum matches)

ACTIONS	QUERY	SCORE	START	END	QSIZE	IDENTITY	CHROM	STRAND	START	END	SPAN
browser details	YourSeq	450	1	450	450	100.0%	chr19	-	7293482	7293931	450
browser details	YourSeq	24	51	78	450	96.3%	chr10	+	21535070	21535101	32
browser details	YourSeq	23	322	347	450	96.0%	chr2	-	25982601	25982626	26
browser details	YourSeq	21	66	86	450	100.0%	chr6	-	13615177	13615197	21

Tools

[All tools](#)

BioMart >

Export custom datasets from Ensembl with this data-mining tool


BLAST/BLAT >

Search our genomes for your DNA or protein sequence

Variant Effect Predictor >

Analyse your own variants and predict the functional consequences of known and unknown variants


Search

All species  for

Go

e.g. [BRCA2](#) or [rat 5:62797383-63627669](#) or [rs699](#) or [coronary heart disease](#)

All genomes

-- Select a species -- 

- [View full list of all Ensembl species](#)
- [Edit your favourites](#)

Favourite genomes



Human
GRCh38.p12

[Still using GRCh37?](#)



Mouse
GRCm38.p6



Zebrafish
GRCz11

Human (GRCh37)

Location: 19:7,112,266-7,294,045

Gene: **INSR**

Gene-based displays

- Gene summary**
- Splice variants (7)
- Transcript comparison
- Supporting evidence
- Sequence
- External references
- Regulation
- Expression
- Comparative Genomics
 - Genomic alignments
 - Gene tree (image)
 - Gene tree (text)
 - Gene tree (alignment)
 - Gene gain/loss tree
- Orthologues (61)
- Paralogues (14)
- Protein families (1)
- Phenotype
- Genetic Variation
 - Variation table
 - Variation image
 - Structural variation
- External data
 - Personal annotation
- ID History
 - Gene history

Configure this page

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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

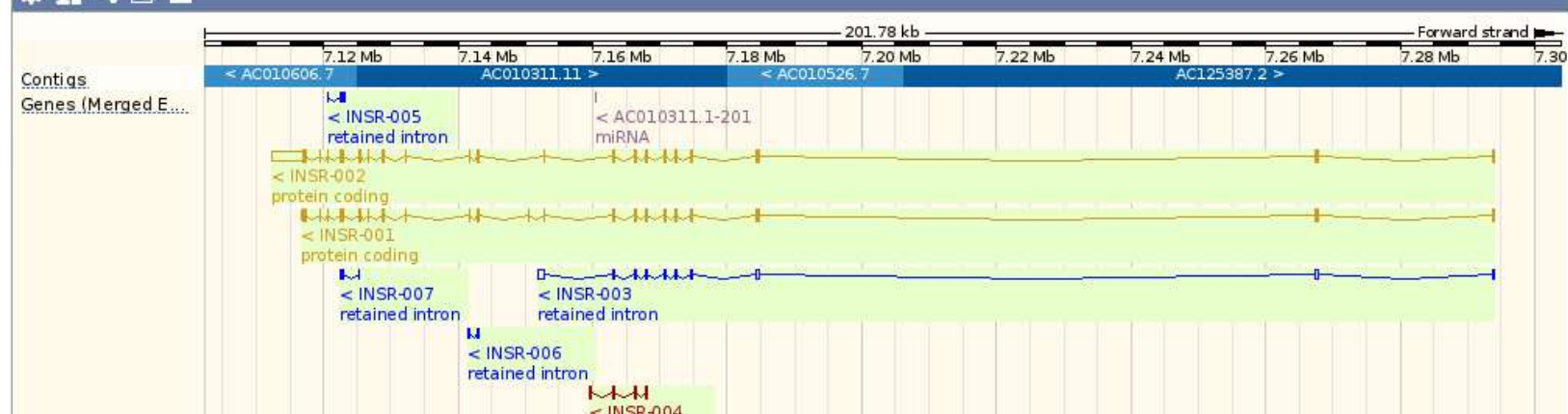
Annotation for this gene includes both automatic annotation from Ensembl and [Havana](#) manual curation, see [article](#).

Alternative genes

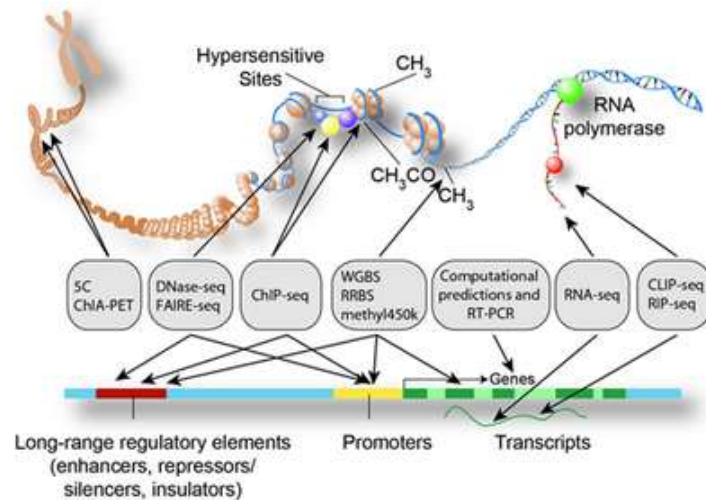
This gene corresponds to the following database identifiers:

Havana gene: [OTTHUMG00000181992](#) (version 2)


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



ENCODE: 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](#)

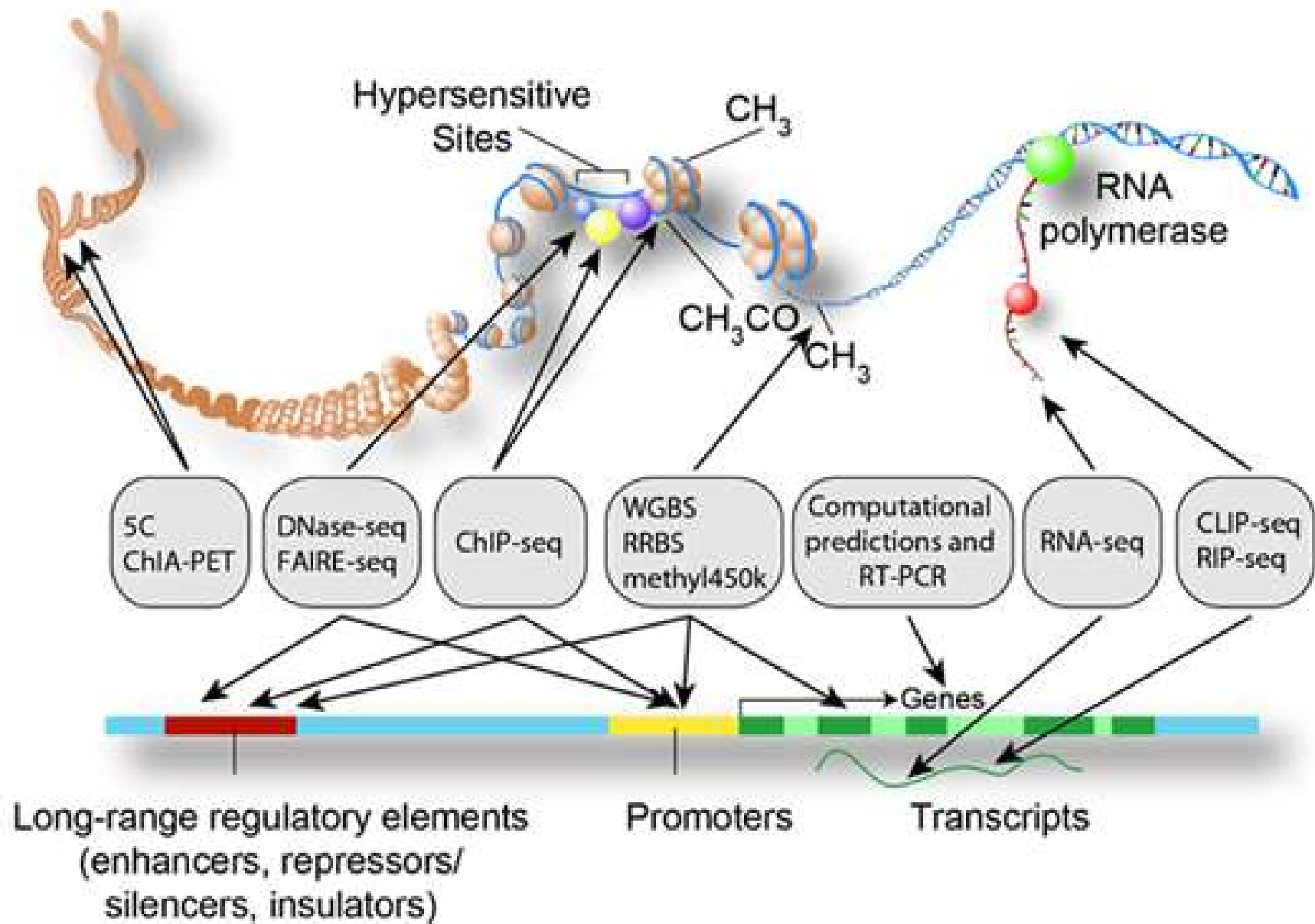
News

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>



applications du NGS

www.broadinstitute.org/software/igv/home

Google

igv Integrative Genomics Viewer

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 - IGV User Guide
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search

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Home

Integrative Genomics Viewer

What's New

September 2014. The IGV iPad app can now be installed from the Apple App Store. ***IGV for iPad*** is a lightweight genomic data viewer that provides some of the functionality available in our regular desktop IGV. See the [IGV for iPad documentation](#) for details.

June 2014. We're hiring! See the [job description](#) on the Broad Institute careers website.

Overview

The Integrative Genomics Viewer (IGV) is a high-performance visualization tool for interactive

Citing IGV

To cite your use of IGV in your publication:

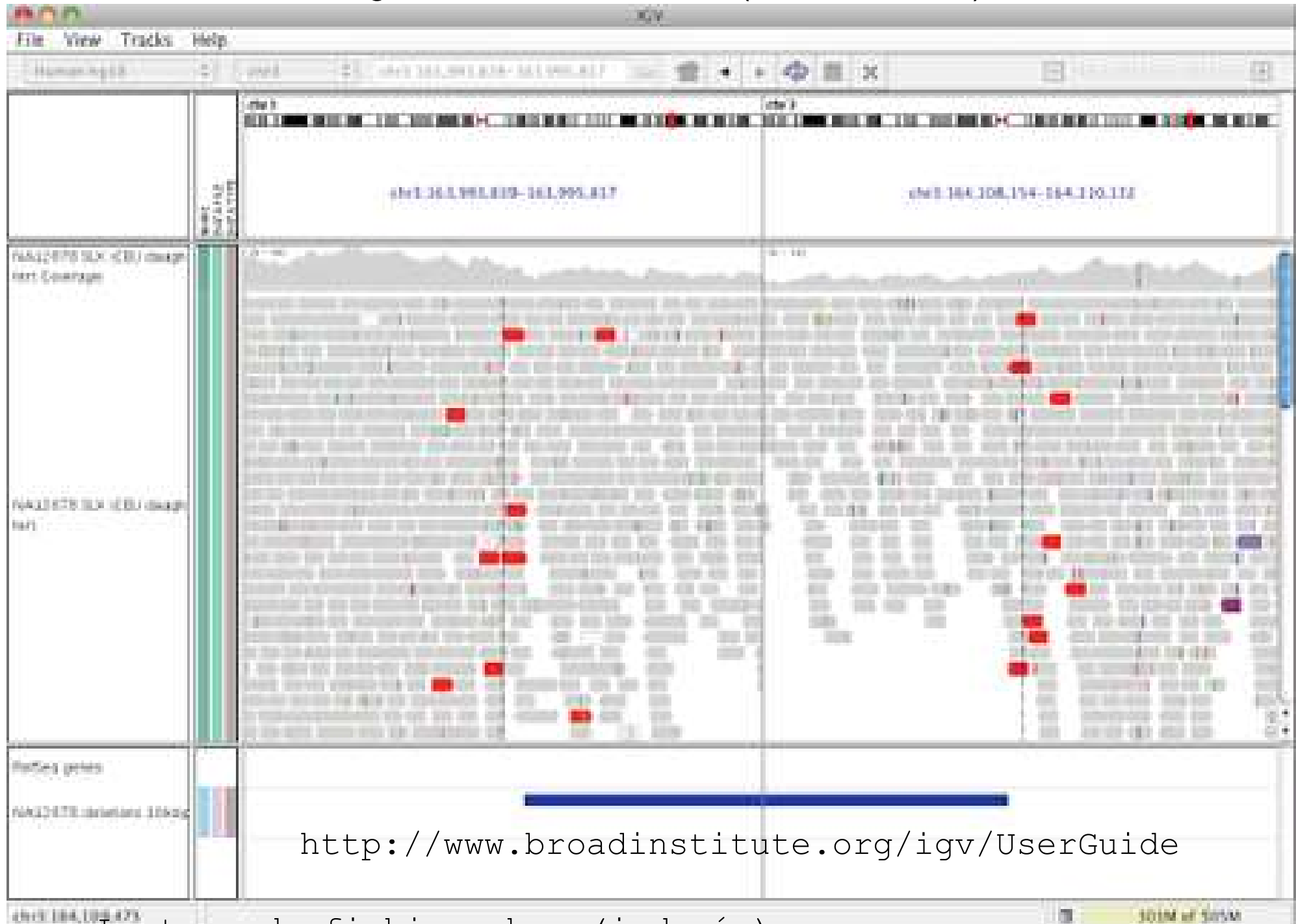
Helga Thorvaldsdóttir, James T. Robinson, Jill P. Mesirov. [Integrative Genomics Viewer \(IGV\): high-performance genomics data visualization and exploration](#). *Briefings in Bioinformatics* 2012.

James T. Robinson, Helga Thorvaldsdóttir, Wendy Winckler, Mitchell Guttman, Eric S. Lander, Gad Getz, Jill P. Mesirov. [Integrative Genomics Viewer](#). *Nature Biotechnology* 29, 24–26 (2011).

Funding

<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 types and/or subtypes which are of clinical and societal importance across the globe.

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



Genome Aggregation Database

gnomAD v2.1.1

Search by gene, region, or variant

Or

- [Find co-occurrence of two variants](#)
- [Download gnomAD data](#)
- [Read gnomAD publications](#)

Please note that gnomAD v2.1.1 and v3.1.2 have substantially different but overlapping sample compositions and are on different genome builds. For more information, see ["Should I switch to the latest version of gnomAD?"](#)

Examples

- Gene: [PCSK9](#)

<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)

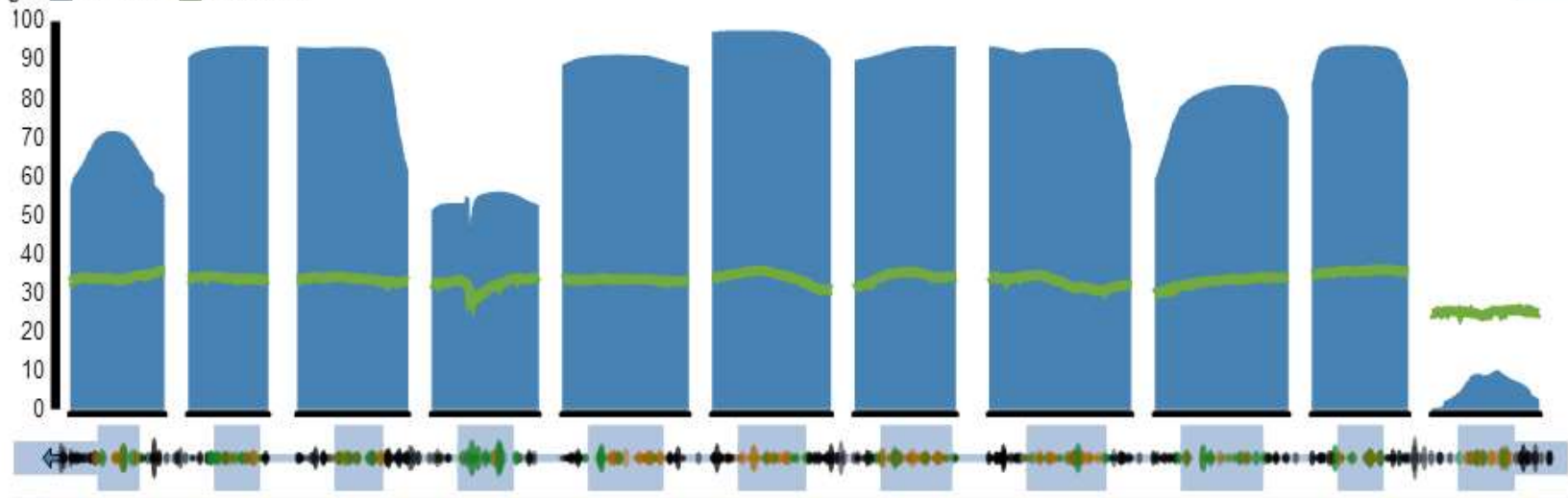
Mean coverage 77.80

Display: [Overview](#) [Detail](#) ☐ Include UTRs in plot

Coverage metric: [Average](#) [Individuals over X](#)

Metric: [mean](#) [↕](#)

Coverage: [Exomes](#) [Genomes](#)



All Missense + LoF LoF

include.

☒ Exomes

☒ SNPs

☒ Genomes

☒ Indels

☐ 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
15:90626207 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

Export table to CSV

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

include:

- ☒ Exomes ☒ SNPs ☒ Filtered (non-PASS) variants
☒ Genomes ☒ Indels

Variant	Source	Consequence	Annotation	Flags	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency	
15:90627498 CT / C (rs201015211)		p.Ter453Trp	frameshift	LC LoF	1	30964	0	3.23e-5	
15:90627506 TG / T (rs764173046)		p.Arg451GlyfsTer17	frameshift	LC LoF	1	245688	0	4.07e-6	
15:90628232 C / T		c.1178+1G>A	splice donor		1	246096	0	4.063e-6	
15:90628261 T / TC		p.Lys384GlufsTer96	frameshift		1	246090	0	4.064e-6	
15:90628278 CCA / C (rs770463292)		p.Gly378ProfsTer101	frameshift		1	246132	0	4.063e-6	
15:90628318 TG / T (rs773746533)		p.Ser365AlafsTer47	frameshift		1	245890	0	4.067e-6	
15:90628532 GT / G		p.Thr352ProfsTer60	frameshift		1	150162	0	6.659e-6	
15:90628534 GAC / G (rs765909746)		p.Val351HisfsTer128	frameshift		79	149086	0	0.0005299	
15:90630446 GC / G (rs752451868)		p.Leu289SerfsTer41	frameshift		1	246272	0	4.061e-6	
15:90630678 A / AG		p.Phe270LeufsTer2	frameshift		1	246272	0	4.061e-6	
15:90631653 C / A (rs761129118)		p.Glu206Ter	stop gained		1	246256	0	4.061e-6	
15:90631821 G / A (rs763369478)		p.Gln178Ter	stop gained		1	246132	0	4.063e-6	
15:90631917 T / TC (rs780120934)		 p.Thr146AspfsTer126	frameshift		2	277136	0	7.217e-6	
15:90631917 TC / T (rs780120934)		p.Thr146LeufsTer15	frameshift		1	246248	0	4.061e-6	
15:90633729 CA / C		p.Asp119MetfsTer10	frameshift		1	246100	0	4.063e-6	
15:90634850 TC / T		p.Lys48SerfsTer11	frameshift		1	246272	0	4.061e-6	
15:90643807 C / A (rs867541960)		c.-42+1G>T†	splice donor	LC LoF	1	30970	0	3.229e-5	
15:90645534 G / GT		p.Thr30AsnfsTer27	frameshift		1	29772	0	3.359e-5	
15:90645587 GC / G		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 information by providers. The scope includes the test's purpose, methodology, validity, evidence of the test's usefulness, 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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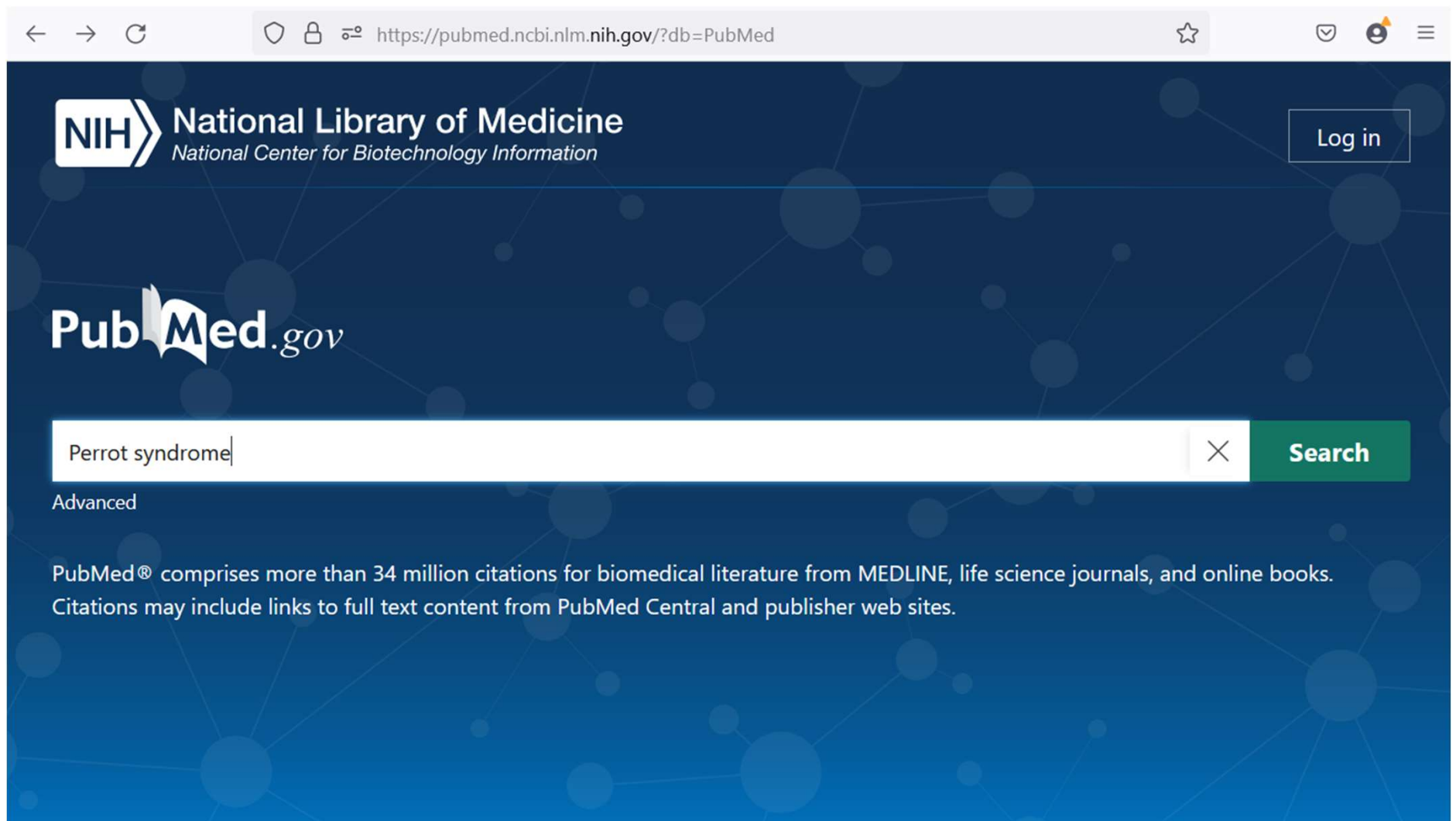
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- 4 Institute of Genetic Medicine, Newcastle University, Newcastle upon Tyne, UK; and Endocrine Department, Newcastle upon Tyne Hospitals, Newcastle upon Tyne, UK.
- 5 The Institute for Genetics, Ha'Emek Medical Centre, Afula, Israel.
- 6 Rapaport faculty of Medicine, Technion Haifa, Haifa, Israel.
- 7 Structural Genomics Consortium, Nuffield Department of Clinical Medicine, University of Oxford, Oxford, UK.
- 8 School of Psychological Sciences, University of Manchester, Manchester, UK.
- 9 Central Manchester University Hospitals NHS Foundation Trust, Manchester Academic Health Science Centre, Manchester, UK.
- 10 Clinical Genetics, St Michaels Hospital, Bristol Genetics Laboratory Pathology Sciences, Southmead Hospital Bristol, Bristol, UK.
- 11 Medical Genetics Center, Munich, Germany.
- 12 Department of Obstetrics and Gynecology, The Catholic University of Korea, Seoul, Korea.
- 13 Faculty of Life Sciences, University of Manchester, Manchester, UK.

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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- Ovarian Disease

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[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)

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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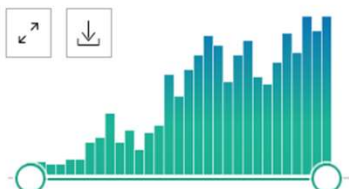
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Liu Y, Liu J, Xiong F, Sun Y, Luo J, He P, He F.

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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.

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Chinese.