Introduction à Ensembl/Biomart

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Jean Muller

Objectifs

- Révision sur les banques/bases de données biologiques
- Connaitre l'existence et l'utilité des principaux "Genome browser"
- Comprendre comment fonctionne le "Genome browser : Ensembl"
- S'initier à
  - la navigation dans Ensembl
  - l'utilisation des outils d'Ensembl
  - l'utilisation de Biomart

Plan

- Introduction
  - Les banques/bases de données biologiques
  - Les “genome browsers”
- Le projet Ensembl
- Comprendre Ensembl
- Navigation dans le “genome browser” Ensembl
- Les outils intégrés à Ensembl
- Utilisation de Biomart

Les banques/Bases de données biologiques

De l'artisanat au haut débit…

1951 première séquence protéique

1967 construction d'arbres phylogénétiques

1970 algorithme de Needleman & Wunsch

1977 séquençage de l'ADN (Méthode Sanger)

premier package bioinformatique (Staden)

1978 bases de données Pir, EMBL, Genbank

1981 algorithme d'alignement local (Smith & Waterman)

1990 programme Blast

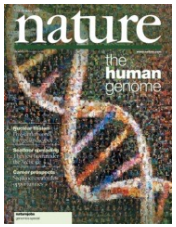
1991 étiquettes d'ADNc « EST »

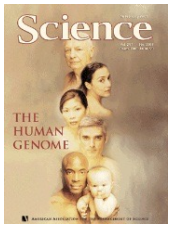
1995 séquençage du génome complet d'une bactérie

1996 séquençage complet du génome de la levure

2001 première version du génome humain

=> Début de l'ère post-génomique





L’ère post-génomique

- 2002 Séquence préliminaire du génome de la souris (Waterston et al., 2002)
- 2004 ENCODE, Identification de tous les éléments fonctionnels du génome humain
- 2005 Roche 454: Séquenceur auto. haut-débit de 2ème génération par pyroséquençage : GS20
- 2007 Illumina/Solexa NGS de 2ème génération par synthèse microfluidique : GAIIx

  Applied Biosystems NGS de 2ème génération par ligation : système SOLiD
- 2008 Helicos Séquenceur auto. de 2ème génération par synthèse sans pré-amplification
- 2012 ENCODE Encyclopédie des éléments fonctionnels du génome humain
- 2014 Génome à 1000$ 2 annonces Illumina et Life Technologies
- 2016- >40 000 génomes complets publiés (3 domaines du vivant)

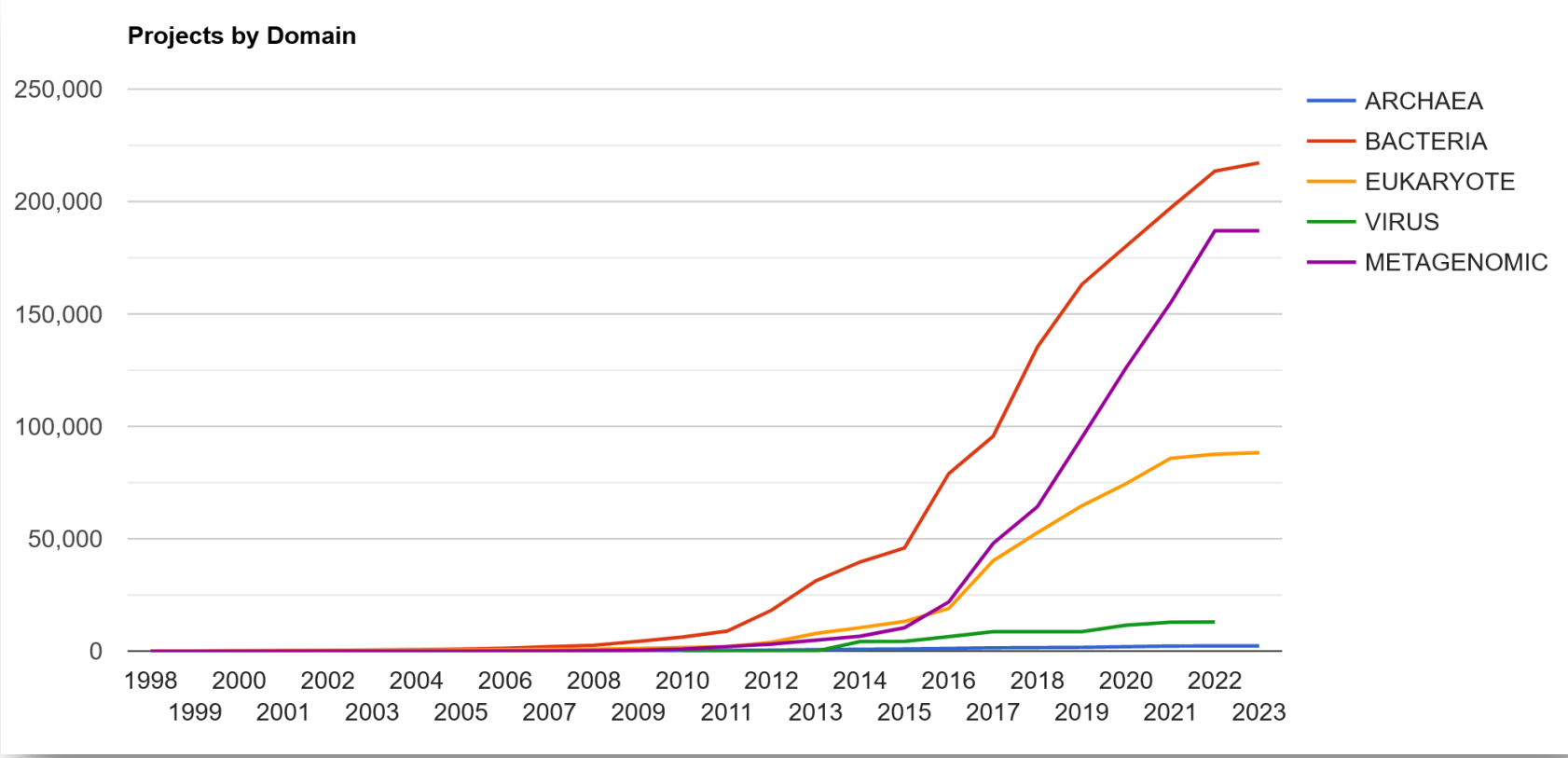
  4989 archées, 409995 bactéries, 47196 eukaryotes et 18327 virus

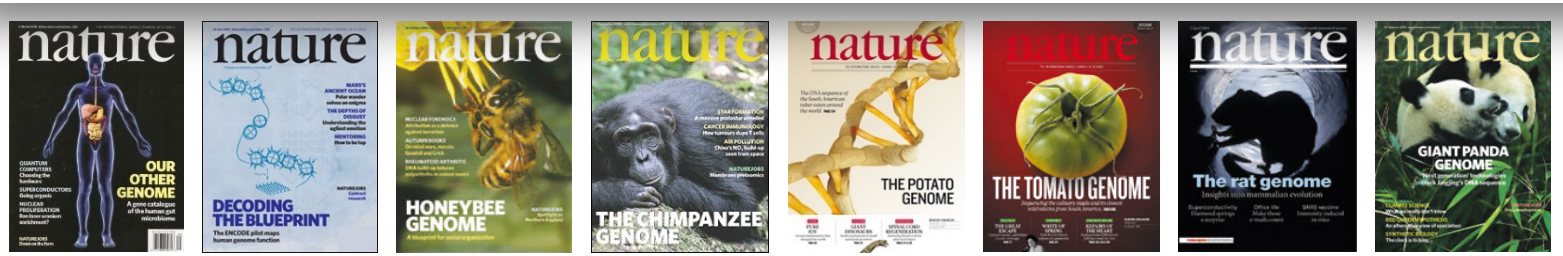
  (www.genomesonline.org, 01/2023)

  Exomes et génomes humains séquencés complètement (patients + pop. Générale)

L’ère post-génomique

- 2002 Séquence préliminaire du génome de la souris (Waterston et al., 2002)



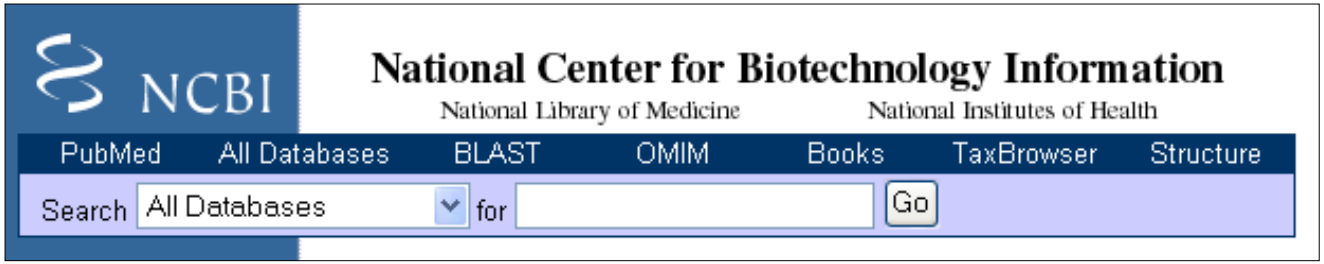


Centres de bioinformatique

- EBI (European Bioinformatics Institute)

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

- NCBI (National Center for Biotechnology Information)



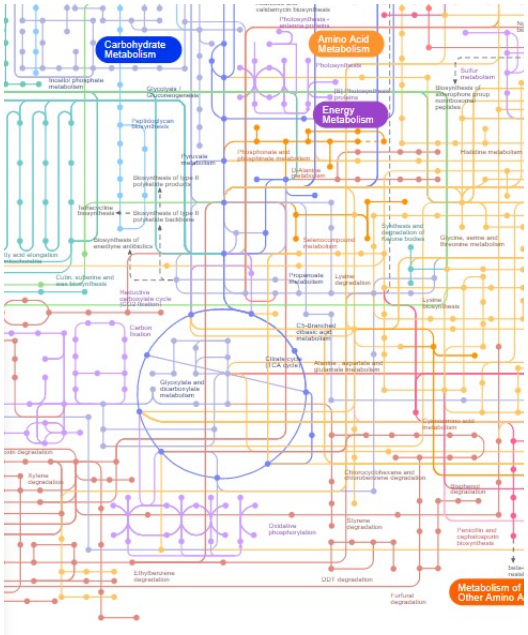
http://www.ncbi.nlm.nih.gov/

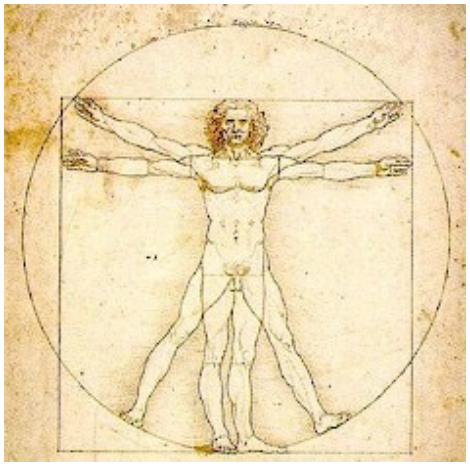
Banques de données en biologie moléculaire

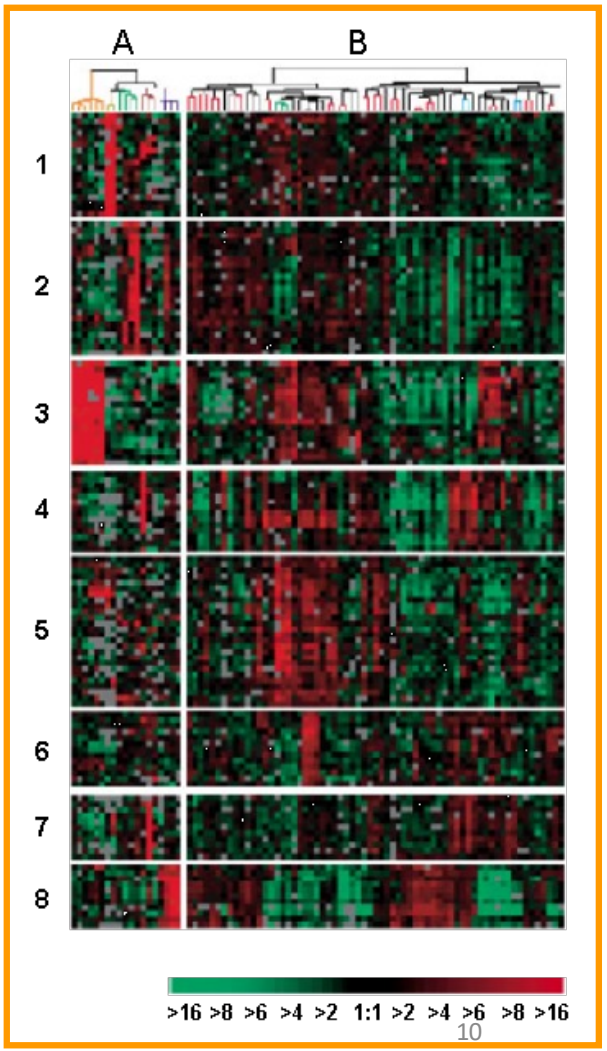
- Rôles des banques
  - Stockage
  - Diffusion (ftp, web…)
  - Organisation et standardisation des données
  - Connectivité avec autres banques
  - Actualisation

Multiplicité des banques

MALWTRLRPLLALLALWPPPPARAFVNQHLCGSHLVEALYLVCGERGFFYTPKARREVEGPQVGALELAGGPGA



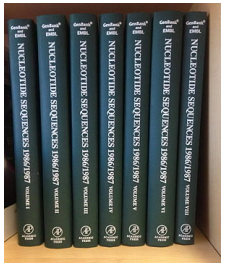




Banques de séquences nucléiques généralistes

- Des banques incontournables :
  - dépôt obligatoire dans une des 3 banques avant publication
  - unique moyen d'accès aux séquences
- Alimentation :
  - soumission directe par la communauté scientifique
    (associée ou non à une publication)
  - dépôts de brevets
- Conséquences
  - banques exhaustives
  - banques extrêmement redondantes
  - contiennent des erreurs

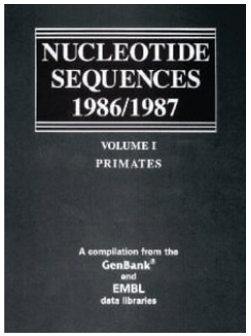
Banques de séquences nucléiques généralistes



wikipedia

NCBI → ENA European Nucleotide Archive

GenBank **EMBL**



DNA databank of Japan

- 3 banques
- Échanges quotidiens des séquences collectées
- Effort d'unification=> format
  - 1986: accord entre GenBank/EMBL
  - 1987: accord entre GenBank/EMBL/DDBJ

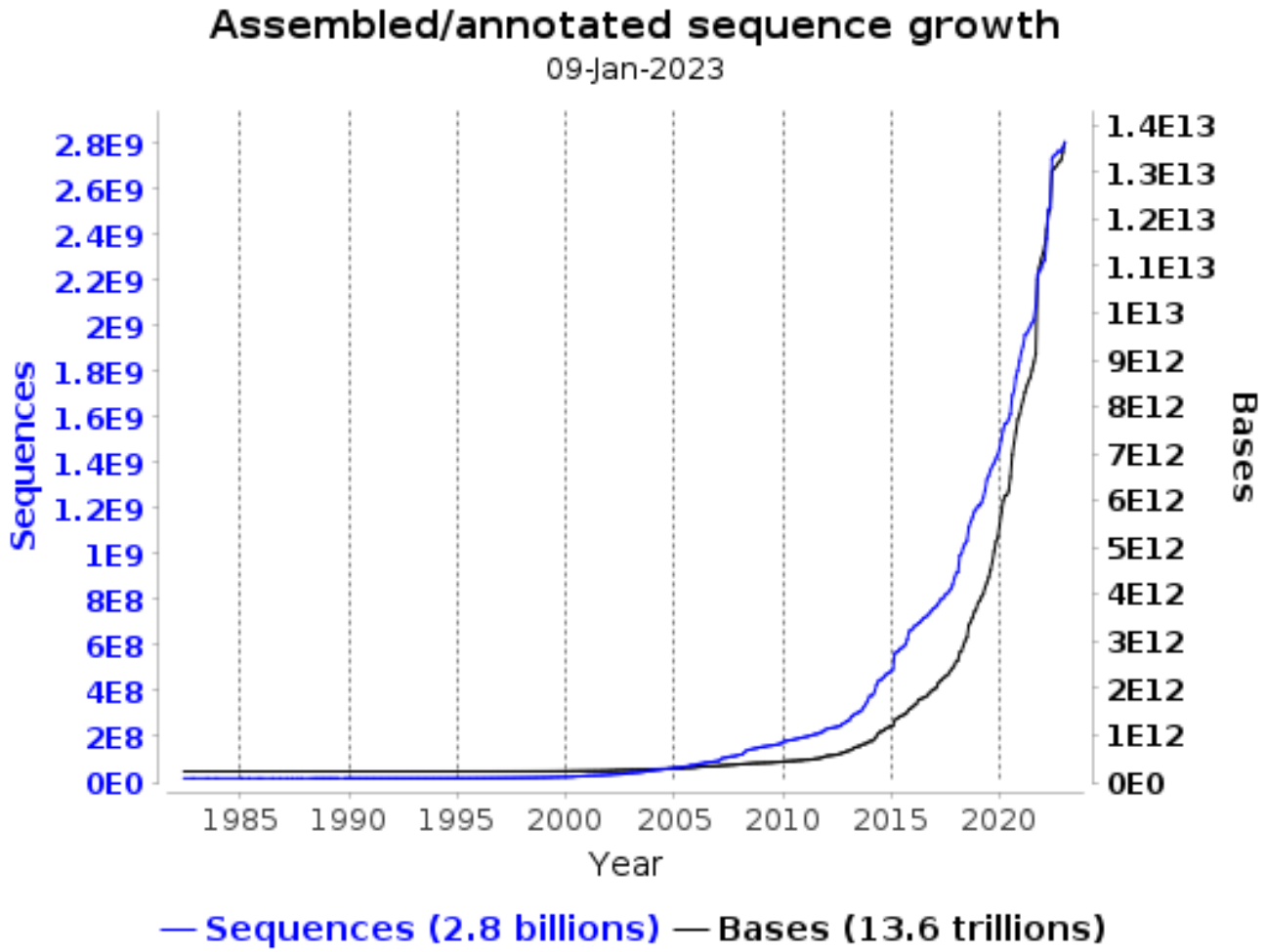
DDBJ DNA Data Bank of Japan

INSDC International Nucleotide Sequence Database Collaboration

doi:10.1126/science.1163399

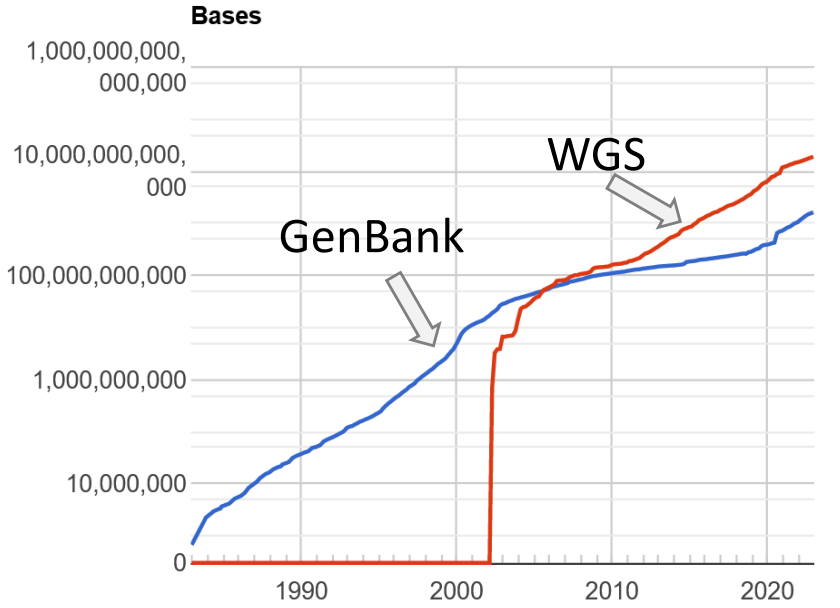
Evolution de la banque EMBL

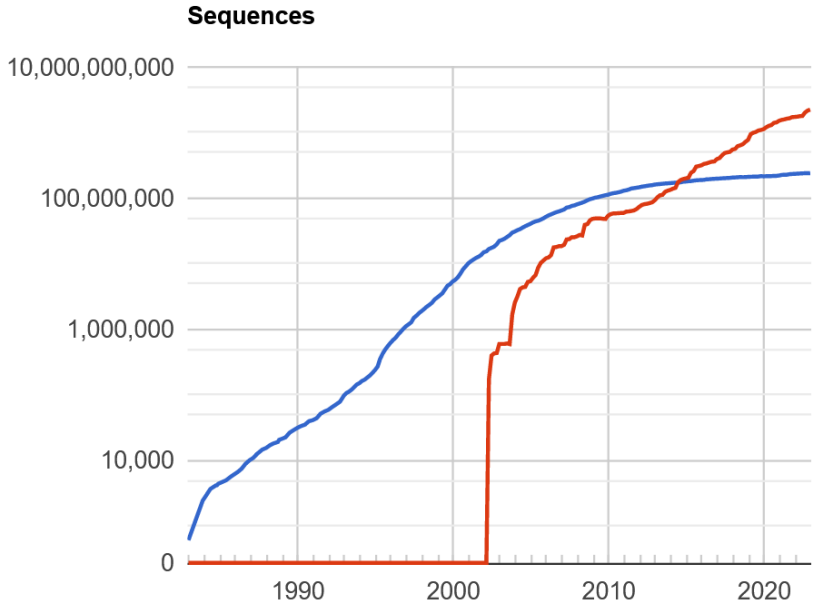
ENA
European Nucleotide Archive



http://www3.ebi.ac.uk/Services/DBStats/

Evolution de la banque GenBank





01/2023: 1635 milliards de nucléotides, 241 millions d'entrées
Doublement tous les 18 mois

https://www.ncbi.nlm.nih.gov/genbank/statistics/

Banques de séquences protéiques généralistes

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

09/2016	03/2018	**01/2019**	**02/2020**
70 427 238	106,245,682	**130,366,644**	**167,278,920**

Transcrits: **29,869,155**
Organismes: **99,842**

http://www.uniprot.org/

	10/2016	02/2018	**02/2020**
TrEMBL	68,493,254	109,414,541	**179,812,129**

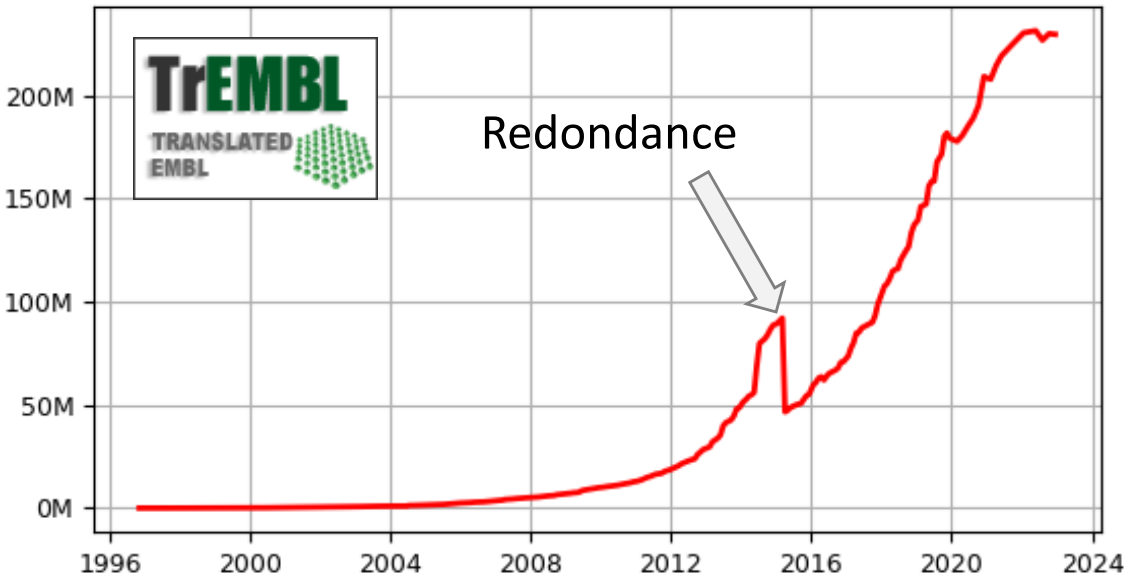
TrEMBL:
179,250,561 entrées

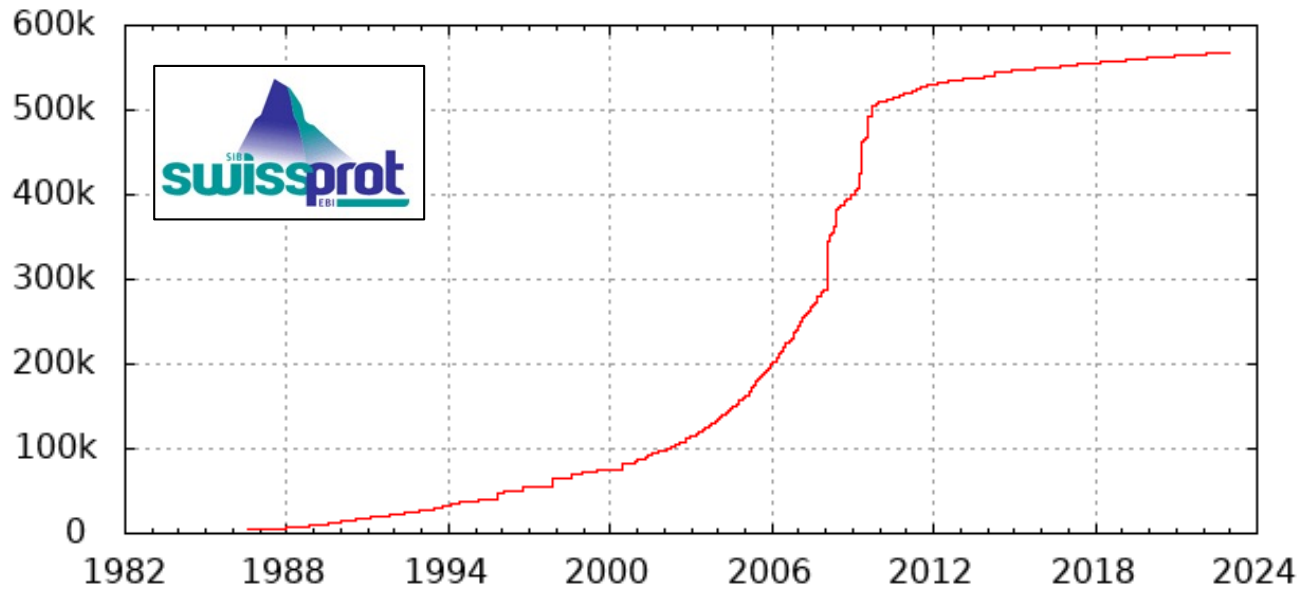
Swiss-Prot:
561,568 entrées

- 2 banques majeures
- Qualité variable/stabilisée
- Exhaustivité / Annotation

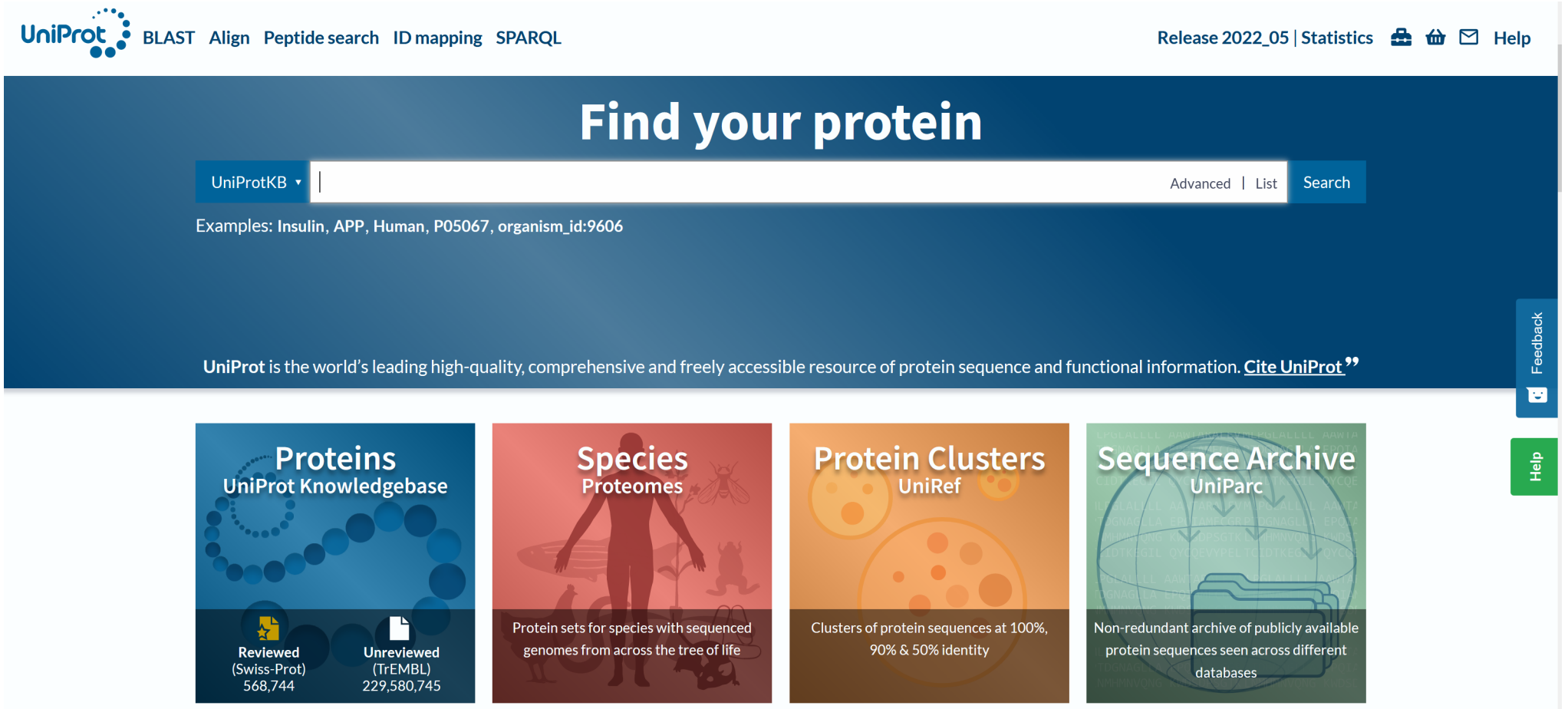
Annotation	UniProt		TrEMBL	
Evidence at protein level	90,921	16,5%	118,013	0,2%
Evidence at transcript level	57,673	10,5%	971,005	1,8%
Inferred from homology	**387,632**	**70,5%**	**11,091,443**	**21,1%**
Predicted	**11,465**	**2,1%**	**40,603,140**	**76,9%**
Uncertain	1,955	0,4%	0	0%

Evolution des bases de données protéiques

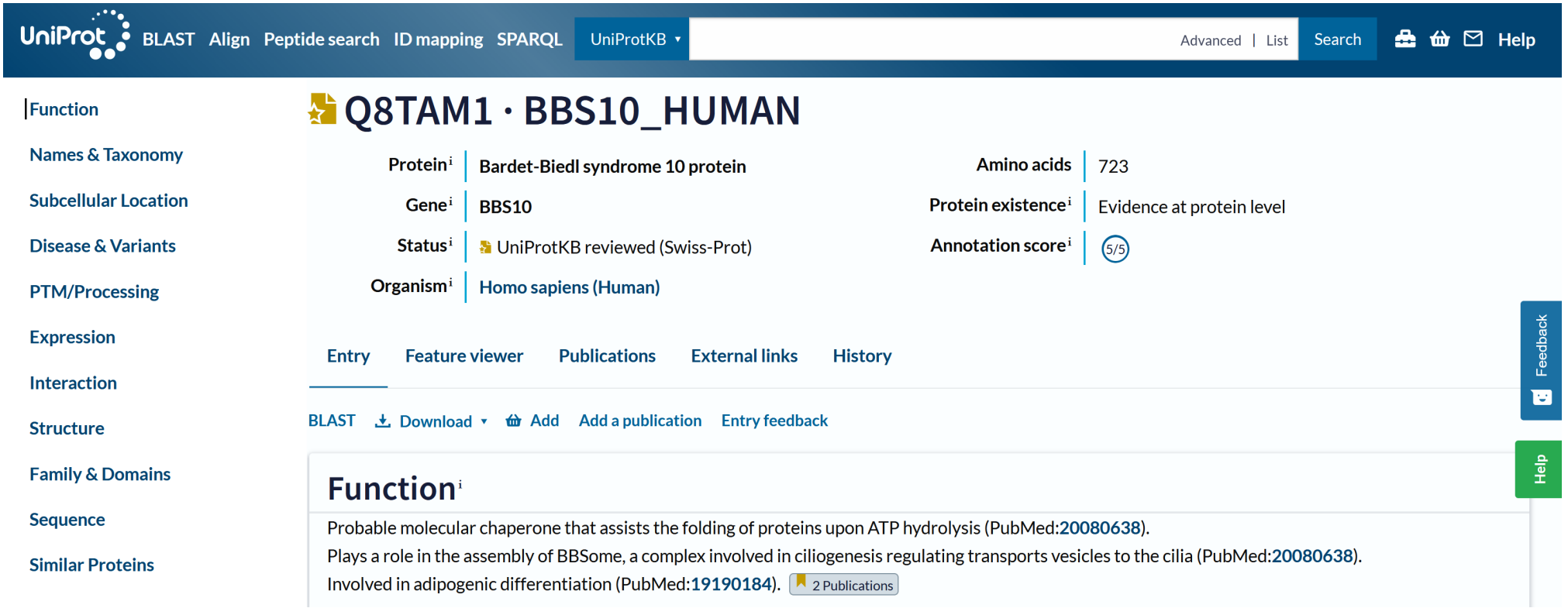




Uniprot


UniProt
BLAST
Align
Peptide search
ID mapping
SPARQL
Release 2022_05 | 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
Proteins
UniProt Knowledgebase
Reviewed
(Swiss-Prot)
568,744
Unreviewed
(TrEMBL)
229,580,745
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
Feedback
Help

Une entrée Swiss-Prot



Une entrée Swiss-Prot

Un enregistrement (entrée) :

- les informations liées à la séquence
- la séquence elle-même
- indicateur de fin d'enregistrement

Les champs :

- regrouper les informations d'un même type
- faciliter l'accès à l'information

Format général (flat file) :

- enregistrements organisés séquentiellement
- fichier texte (ASCII)
- fichiers disponibles en XML

```
ID   BBS10_HUMAN             Reviewed;         723 AA.
AC   Q8TAM1; Q96CW2; Q9H5D2;
DT   16-MAY-2006, integrated into UniProtKB/Swiss-Prot.
DT   16-MAY-2006, sequence version 2.
DT   14-OCT-2015, entry version 117.
DE   RecName: Full=Bardet-Biedl syndrome 10 protein;
DR   CCDS; CCDS9014.2; -.
DR   RefSeq; NP_078961.3; NM_024685.3.
DR   STRING; 9606.ENSP00000376946; -.
DR   Ensembl; ENST00000393262; ENSP00000376946; ENSG00000179941.
DR   GeneID; 79738; -.
DR   KEGG; hsa:79738; -.
DR   GO; GO:0005929; C:cilium; IEA:UniProtKB-SubCell.
DR   GO; GO:0005524; F:ATP binding; IEA:UniProtKB-KW.
DR   GO; GO:0001103; F:RNA polymerase II repressing transcription factor
binding; IPI:MGI.
DR   GO; GO:0051131; P:chaperone-mediated protein complex assembly;
IMP:MGI.
DR   GO; GO:0035058; P:nonmotile primary cilium assembly; IMP:BHF-UCL.
DR   GO; GO:0045494; P:photoreceptor cell maintenance; IMP:BHF-UCL.
DR   InterPro; IPR002423; Cpn60/TCP-1.
DR   InterPro; IPR027413; GROEL-like_equatorial.                    major
DR   Pfam; PF00118; Cpn60_TCP1; 2.
PE   1: Evidence at protein level;
KW   ATP-binding; Bardet-Biedl syndrome; Cell projection; Chaperone;
KW   Ciliopathy; Complete proteome; Disease mutation; Mental retardation;
KW   Nucleotide-binding; Obesity; Polymorphism; Reference proteome;
KW   Sensory transduction; Vision.
FT   VARIANT     715    715       H -> R. {ECO:0000269|PubMed:21344540}.
FT                                /FTId=VAR_066261.
SQ   SEQUENCE   723 AA;  80838 MW;  558143FFA5F191DD CRC64;
     MLSSMAAAGS VKAALQVAEV LEAIVSCCVG PEGRQVLCTK PTGEVLLSRN GGRLLEALHL
     EHPIARMIVD CVSSHLKKTG DGAKTFIIFL CHLLRGLHAI TDREKDPLMC ENIQTHGRHW
     KNCSRWKFIS QALLTFQTQI LDGIMDQYLS RHFLSIFSSA KERTLCRSSL ELLLEAYFCG
     RVGRNNHKFI SQLMCDYFFK CMTCKSGIGV FELVDDHFVE LNVGVTGLPV SDSRIIAGLV
     LQKDFSVYRP ADGDMRMVIV TETIQPLFST SGSEFILNSE AQFQTSQFWI MEKTKAIMKH
     LHSQNVKLLI SSVKQPDLVS YYAGVNGISV VECLSSEEVS LIRRIIGLSP FVPPQAFSQC
     EIPNTALVKF CKPLILRSKR YVHLGLISTC AFIPHSIVLC GPVHGLIEQH EDALHGALKM
     LRQLFKDLDL NYMTQTNDQN GTSSLFIYKN SGESYQAPDP GNGSIQRPYQ DTVAENKDAL
     EKTQTYLKVH SNLVIPDVEL ETYIPYSTPT LTPTDTFQTV ETLTCLSLER NRLTDYYEPL
     LKNNSTAYST RGNRIEISYE NLQVTNITRK GSMLPVSCKL PNMGTSQSYL SSSMPAGCVL
     PVGGNFEILL HYYLLNYAKK CHQSEETMVS MIIANALLGI PKVLYKSKTG KYSFPHTYIR
     AVHALQTNQP LVSSQTGLES VMGKYQLLTS VLQCLTKILT IDMVITVKRH PQKVHNQDSE
     DEL
//
```

Les banques de structures

- **La Protein Data Bank (PDB)**

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RCSB PDB PROTEIN DATA BANK

200,069 Structures from the PDB

1,000,357 Computed Structure Models (CSM)

3D Structures | Enter search term(s), Entry ID(s), or sequence | Include CSM

Advanced Search | Browse Annotations Help

PDB-101 wwPDB EMDataResource NUCLEIC ACID DATABASE wwPDB Foundation

NEW! Computed Structure Models (CSM) Learn more

Welcome

Deposit

Search

Visualize

Analyze

Download

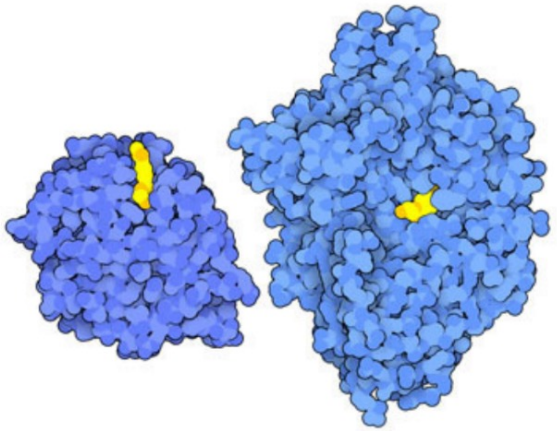
RCSB Protein Data Bank (RCSB PDB) enables breakthroughs in science and education by providing access and tools for exploration, visualization, and analysis of:

Experimentally-determined 3D structures from the **Protein Data Bank (PDB)** archive

Computed Structure Models (CSM) from AlphaFold DB and ModelArchive

These data can be explored in context of external annotations providing a structural view of biology.

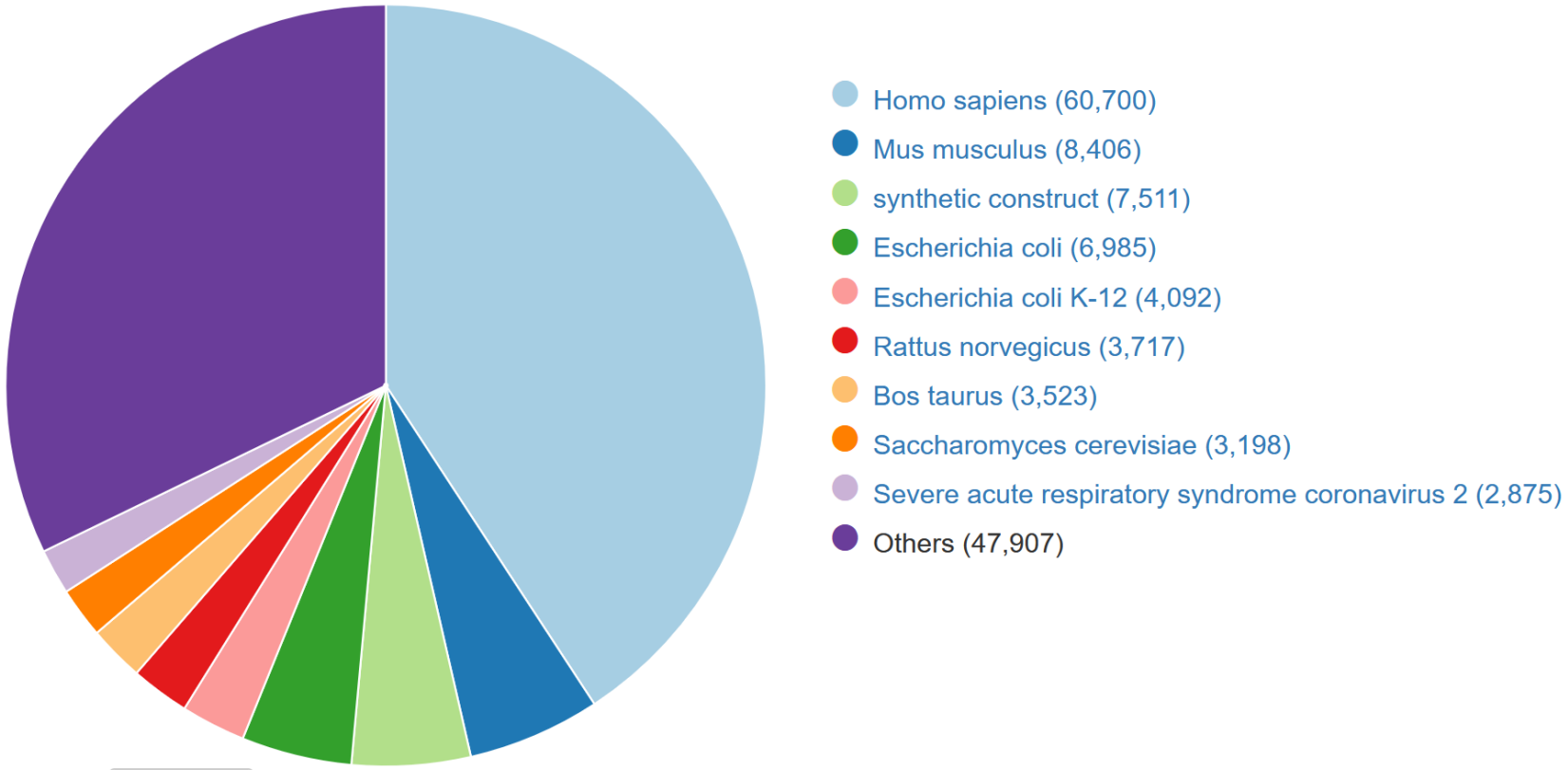
January **Molecule of the Month**



http://www.rcsb.org/pdb/

Les banques de structures

Distribution en fonction de l'organisme d'origine



Accès 01/2023 http://www.rcsb.org/pdb/

Hétérogénéité de la qualité en fonction de leur origine

La séquence des protéines est prédite!

La **qualité** des séquences de protéines dépend de la source et est dont **très hétérogène**

cDNA clonés et séquencés individuellement => protéine
(complets, séquençage multiple, vérification)


HTC (High-Throughput cDNA) => protéine
(full-length mais séquence brute, indels, multiple codons initiateur)

Structure 3D => protéine
(attention au substitutions ponctuelles/délétions)

Séquence génomique procaryote => protéine prédite
(prédiction réalisée par outils bioinformatiques, erreurs de codon initiateur de traduction fréquents, indels en Nter)

Séquence génomique eucaryote => protéine prédite
(prédiction réalisée par outils bioinformatiques, erreurs de prédictions de sites d'épissage fréquents, frameshifts, indels)

Hétérogénéité de la qualité en fonction de leur origine

1) Annotations manuelles

Réalisées par des experts, les entrées sont traitées une par une (UniProt/SwissProt)

2) Annotations automatiques

Réalisées par des outils bioinformatiques de prédiction de domaines, de fonctions…

« **by silmilarity** » , « **homologous to** », « **related to** », « **-like** », « **putative** », « **potential** »

Sont produites en haut-débit (ex: annotation de génomes)

Elles sont légions dans les banques ... et en attente d'une validation

3) Absence d'annotations

« hypothetical protein »

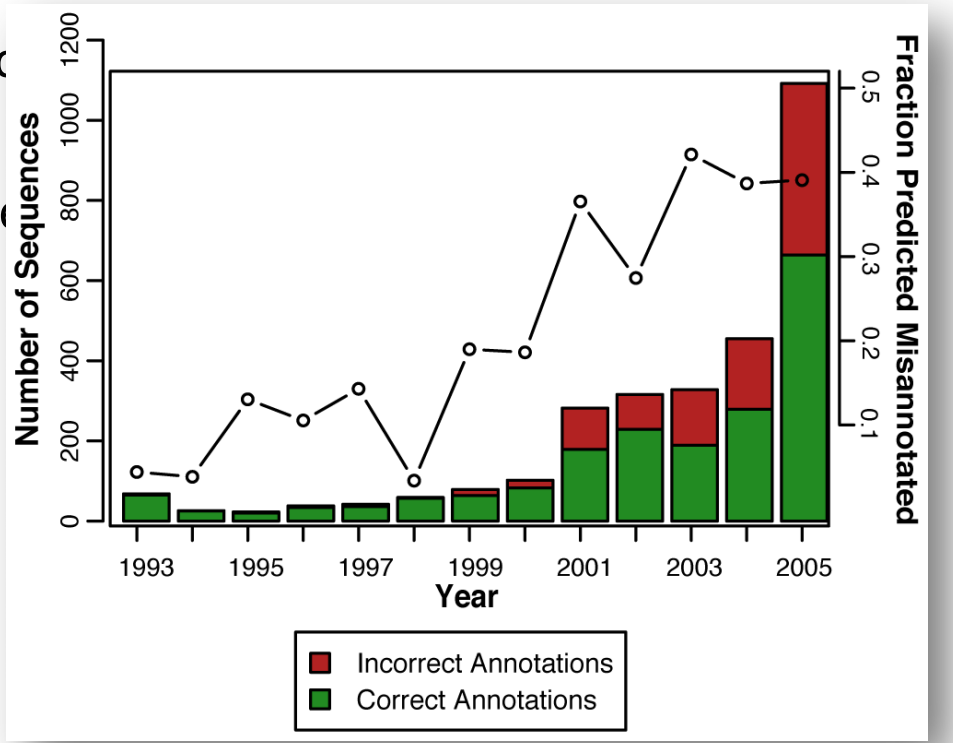
Exemple de l'importance de l'annotation

Exemple 1: DUF domain = **D**omain of **U**nknown **F**unction

Exemple 2: FAM20C = **Fam**ily with sequence similarity **20**, member **C**

Exemple 3: Analyse de 37 familles de pro

L'augmentation de la **quantité** de donné
ne signifie pas une augmentation de la
qualité de ces données.



Schnoes *et al* PLOS Genetics 2009

Evolution des bases de données protéiques

Bases de données majeures

collecte des données individuelles et collectives

Attention à la qualité de ces données

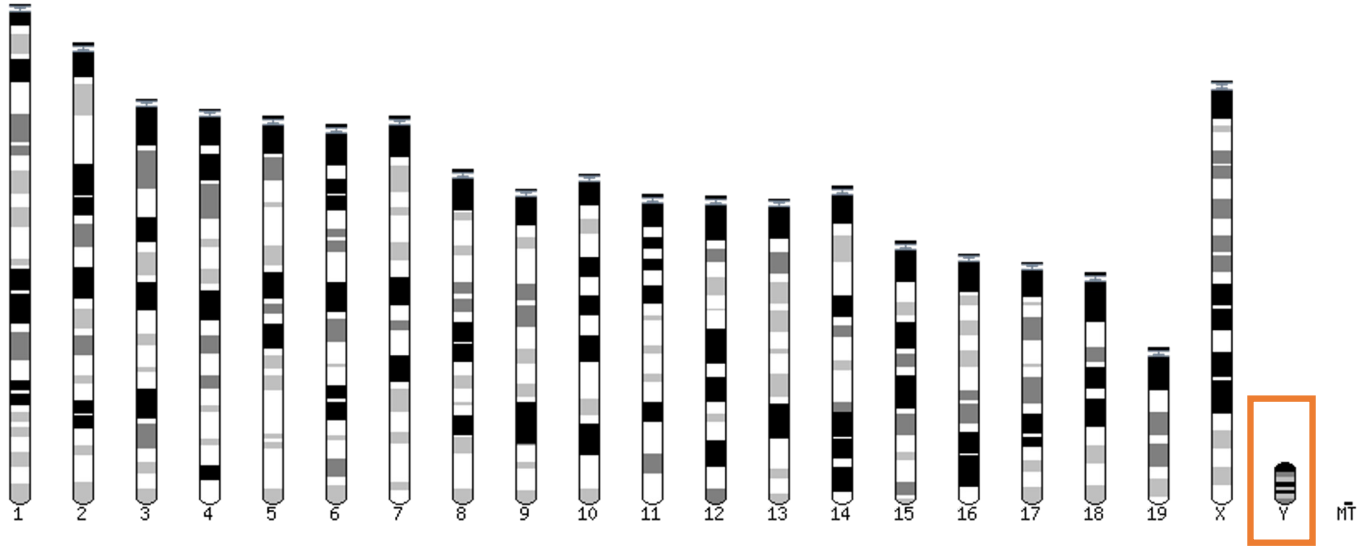
bases avec les Raw data vs Annotation

Ces données seront agrégées sur le génome humain

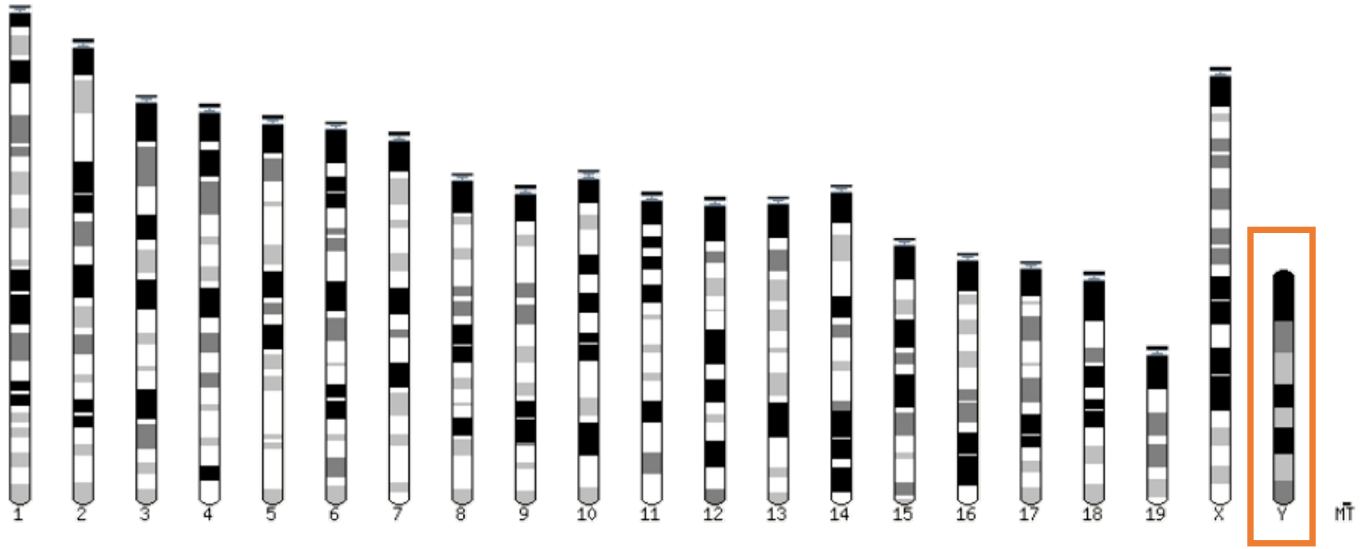
Genome browsers

Genome builds

mm9



mm10



Human Genome Builds

SPECIES	UCSC VERSION	RELEASE DATE	RELEASE NAME	STATUS
MAMMALS				
Human	hg38	Dec. 2013	Genome Reference Consortium GRCh38	Available
	hg19	Feb. 2009	Genome Reference Consortium GRCh37	Available
	hg18	Mar. 2006	NCBI Build 36.1	Available
	hg17	May 2004	NCBI Build 35	Available
	hg16	Jul. 2003	NCBI Build 34	Available
	hg15	Apr. 2003	NCBI Build 33	Archived
	hg13	Nov. 2002	NCBI Build 31	Archived
	hg12	Jun. 2002	NCBI Build 30	Archived
	hg11	Apr. 2002	NCBI Build 29	Archived (data only)
	hg10	Dec. 2001	NCBI Build 28	Archived (data only)
	hg8	Aug. 2001	UCSC-assembled	Archived (data only)
	hg7	Apr. 2001	UCSC-assembled	Archived (data only)
	hg6	Dec. 2000	UCSC-assembled	Archived (data only)
	hg5	Oct. 2000	UCSC-assembled	Archived (data only)
	hg4	Sep. 2000	UCSC-assembled	Archived (data only)
	hg3	Jul. 2000	UCSC-assembled	Archived (data only)
	hg2	Jun. 2000	UCSC-assembled	Archived (data only)
	hg1	May 2000	UCSC-assembled	Archived (data only)

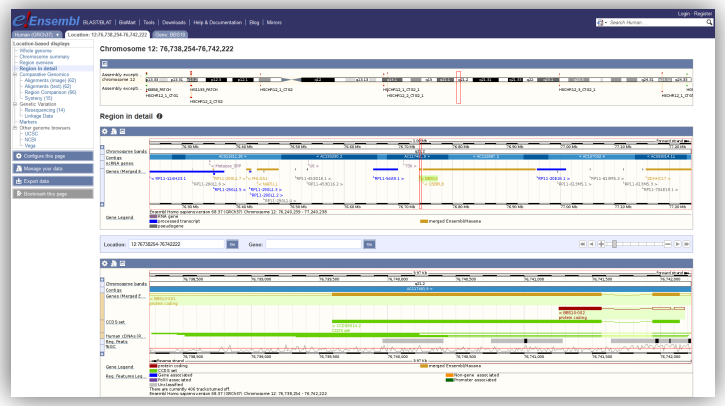
Source: https://genome.ucsc.edu/FAQ/FAQreleases.html

Genome Browsers – L'outil de référence

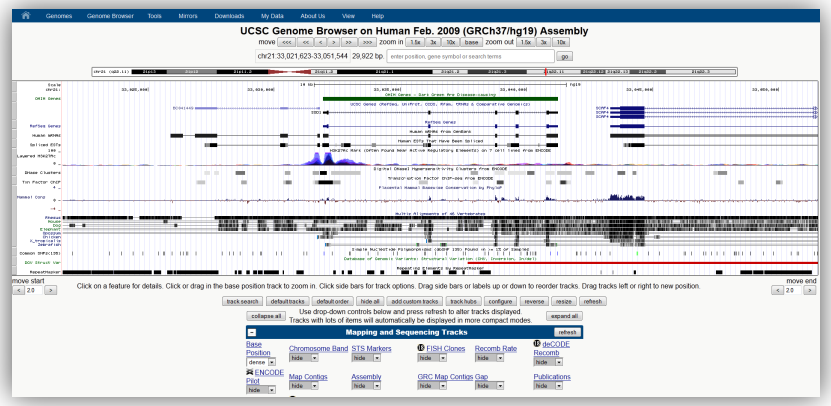
- Elément de référence absolue le **génome**
- Agrégateur et générateur d'informations/annotations
  - Prédictions de gènes
  - Protéines
  - Données d'expression
  - Variations
- Synthèse rapide et visuelle de données primordiales

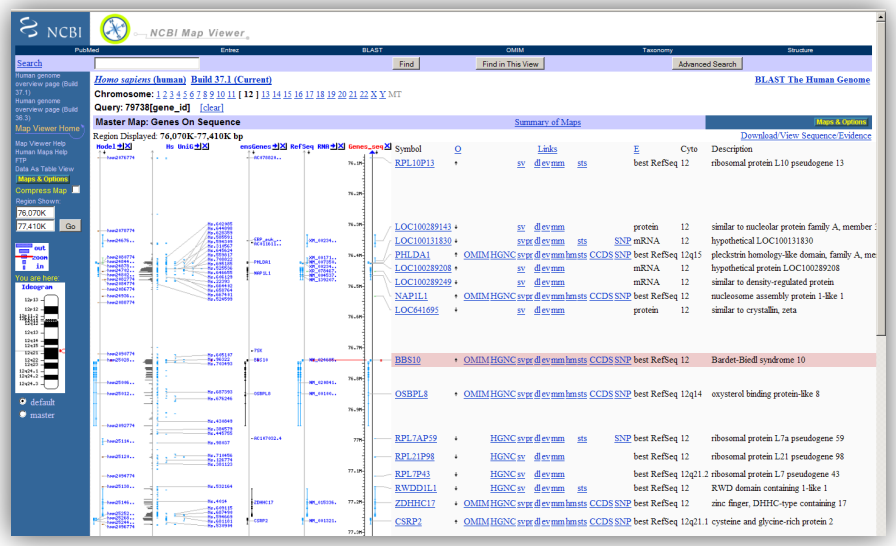
Il y a Genome Browsers…

EBI - Ensembl



UCSC – Genome Browser

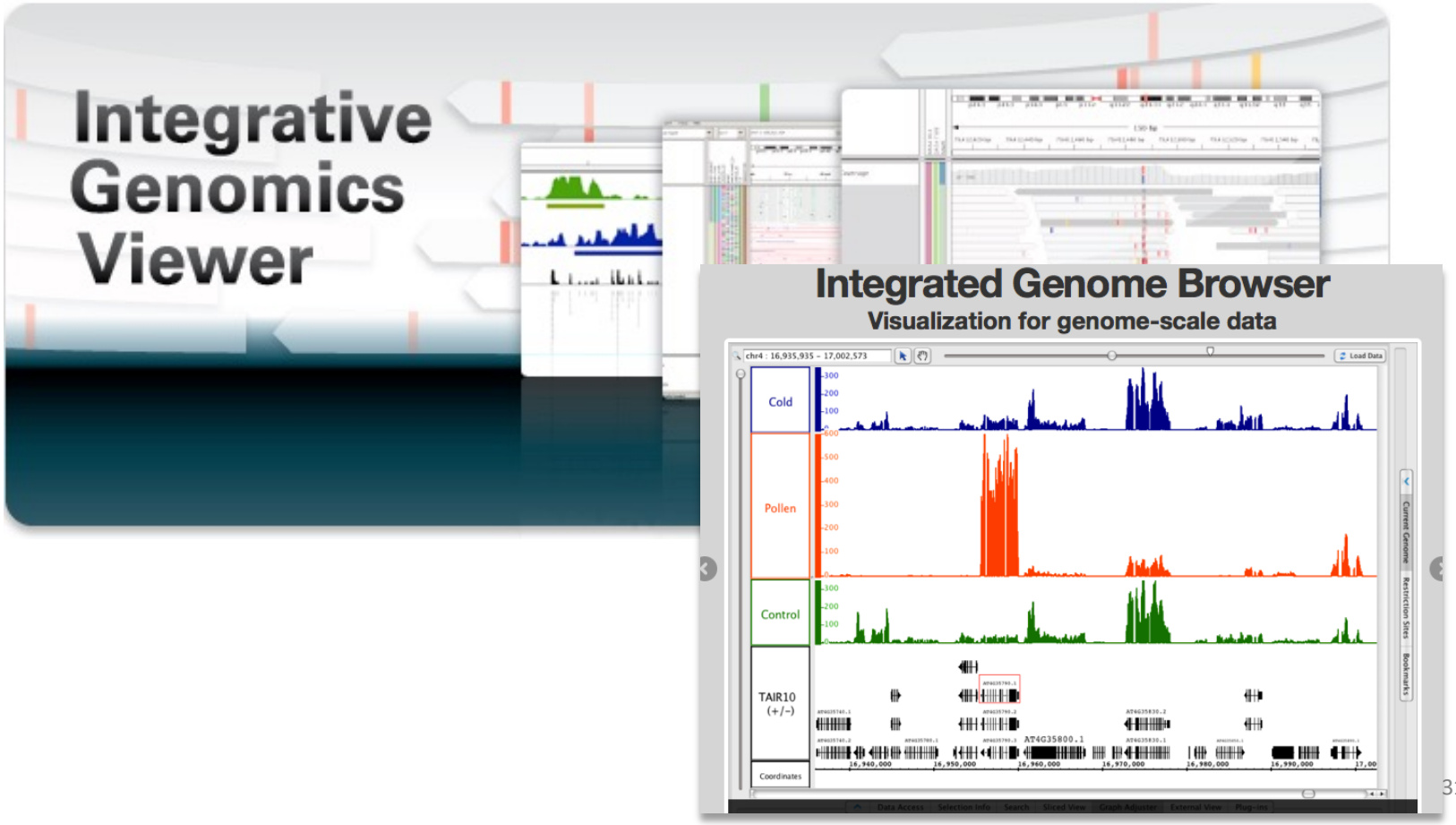




NCBI – Map Viewer

Et Genome browsers

JBrowse



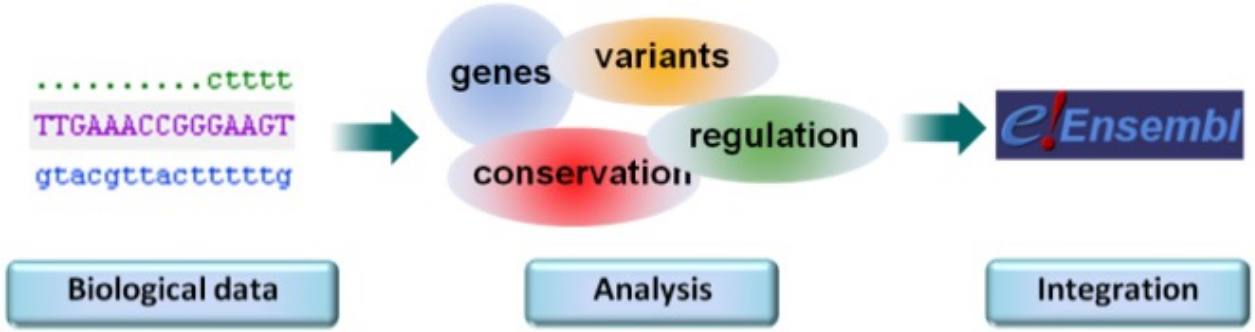
Ensembl

Le projet Ensembl

- Initié en 1999 (avant la première version du génome humain)
- Projet en collaboration entre l'European Bioinformatics Intitute (EBI) et le Wellcome Trust Sanger Institute (WTSI)
- Objectif :
  - Annoter automatiquement les génomes
  - Ajouter des données biologiques aux annotations
  - Rendre publique les annotations sur le web
- Ensembl ne produit pas ses propres données d'assemblage de génome!

Le projet Ensembl

- Données disponibles :
  - **Génomes**
  - **Données de génomique comparative**
  - **Variations**
  - **Elément régulateur des gènes**
  - Annotations externes

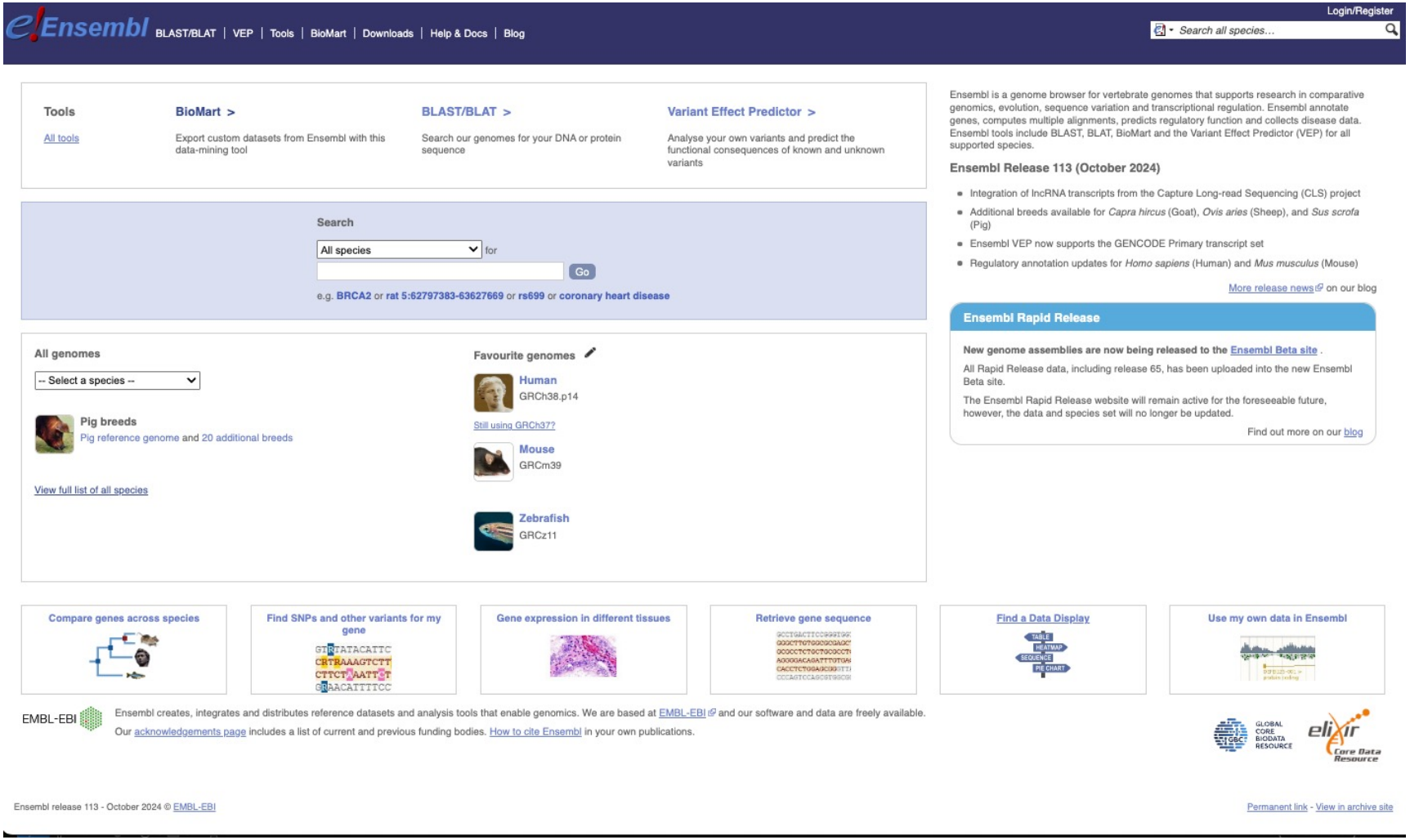


- Lancement du site web en juillet 2000 (au début il n'y avait que le génome humain)

Les génomes d'Ensembl

- Espèces de vertébrés dans http://ensembl.org
- EnsemblGenomes (avril 2009) : https://ensemblgenomes.org/
  - Métazoaires : http://metazoa.ensembl.org
  - Bactéries : http://bacteria.ensembl.org
  - Plantes : http://plants.ensembl.org
  - Fungi : http://fungi.ensembl.org
  - Protistes : http://protists.ensembl.org

L’interface web



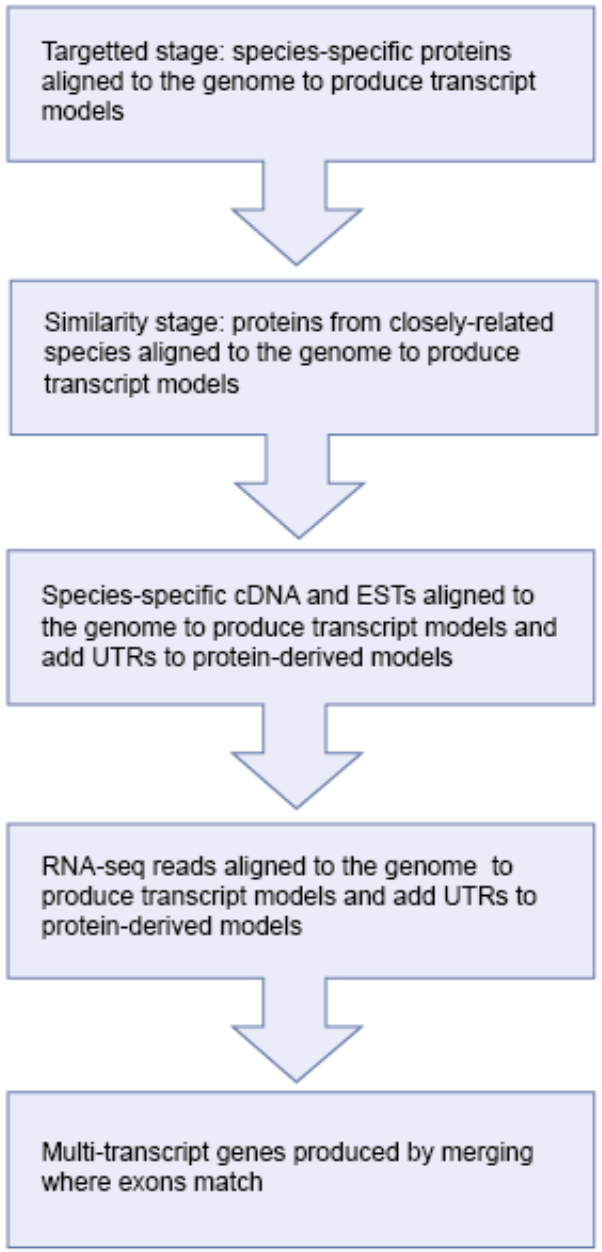
Comprendre ENSEMBL

Les annotations

- 3 à 6 mois
- Annotation par Ensembl
  - Annotation automatique (Ensembl Genebuild) :
    - Détermination des transcrits dans le génome entier
    - Basées sur des séquences d'ARNm et protéiques extraites des banques de données publiques
  - *Curation* manuelle : au cas par cas. Ex: l'humain, la souris, le rat, le zebrafish + autres vertébrés (produit par le groupe HAVANA du WTSI)
  - Fusion des annotations automatiques et manuelles (Gold)
- + Annotations importées depuis flyBase, WormBase, SGD

Les annotations

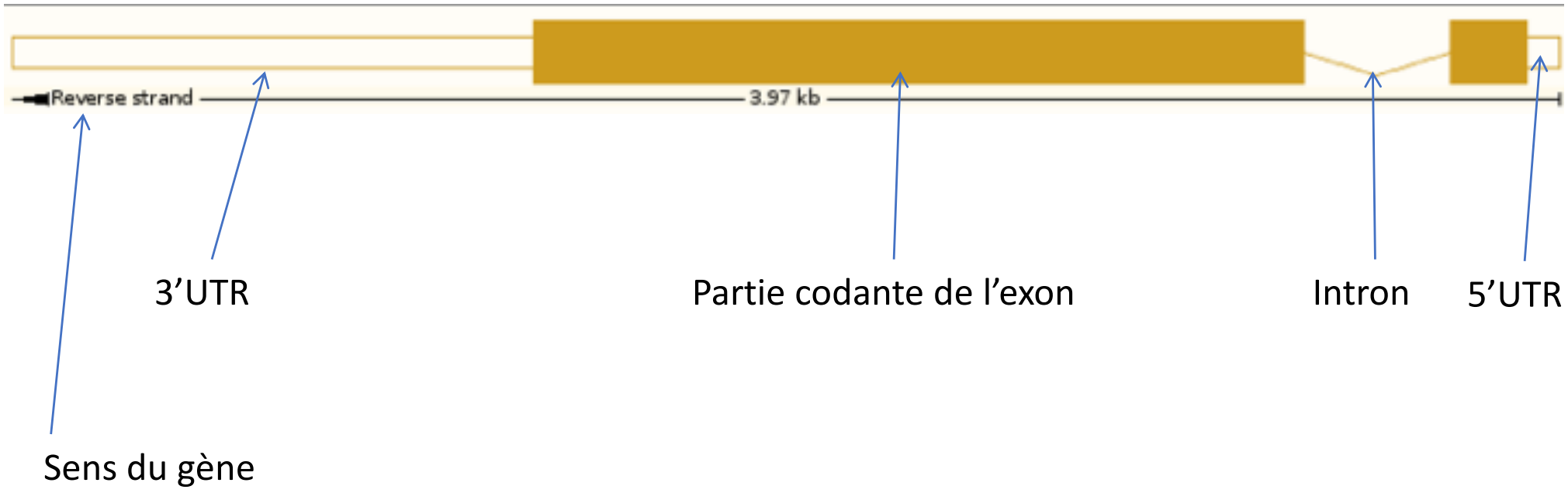
- **Les annotations automatiques des gènes codants** sont basés sur :
  - European Nucleotide Archive (ENA)
  - UniProtKB (curation manuelle)
  - NCBI refseq (curation manuelle)
- Les annotations automatiques des genes non-codants sont basés sur :
  - RFAM (base de données de familles d'ARNs)
  - miRbase
  - tRNAscan-SE (algorithm de prediction des tRNA)



Les annotations

- Les annotations des gènes peuvent varier entre les différents genome browsers (Ensembl, UCSC, NCBI)
- CCDS (Consensus CDS) est un jeu de données de gènes codants validés par tous les membres du consortium (EBI, NCBI, HGNC, MGI)
  - http://www.ncbi.nlm.nih.gov/CCDS/CcdsBrowse.cgi
  - Il faut que l'assemblage du génome soit suffisamment stable pour identifier les gènes dont les positions sont identiques entre les différentes sources (chez humain et souris)

Transcrits Ensembl



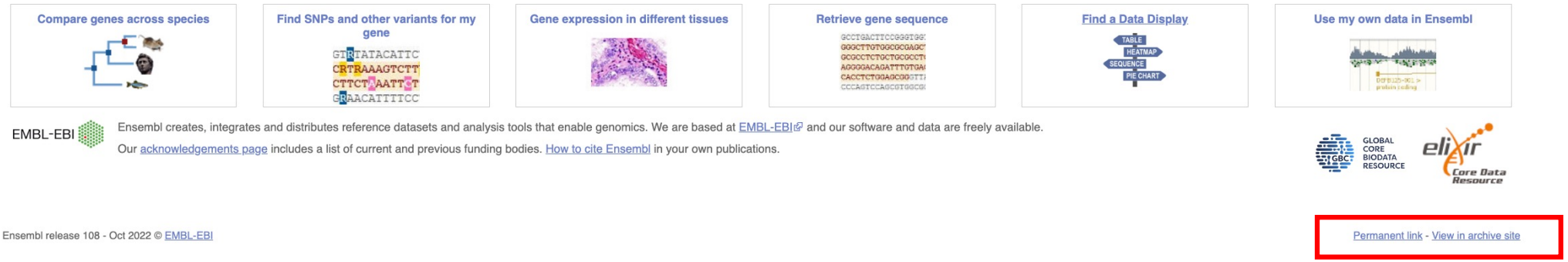
Identifiants Ensembl

- ENSG### Ensembl Gene ID
- ENST### Ensembl Transcript ID
- ENSP### Ensembl Peptide ID
- ENSE### Ensembl Exon ID

- Ajout d'un suffix pour les autres espèces
  - MUS (*Mus musculus*) pour la souris: ENSMUSG###
  - DAR (*Danio rerio*) pour le zebrafish: ENSDARG###
  - etc.

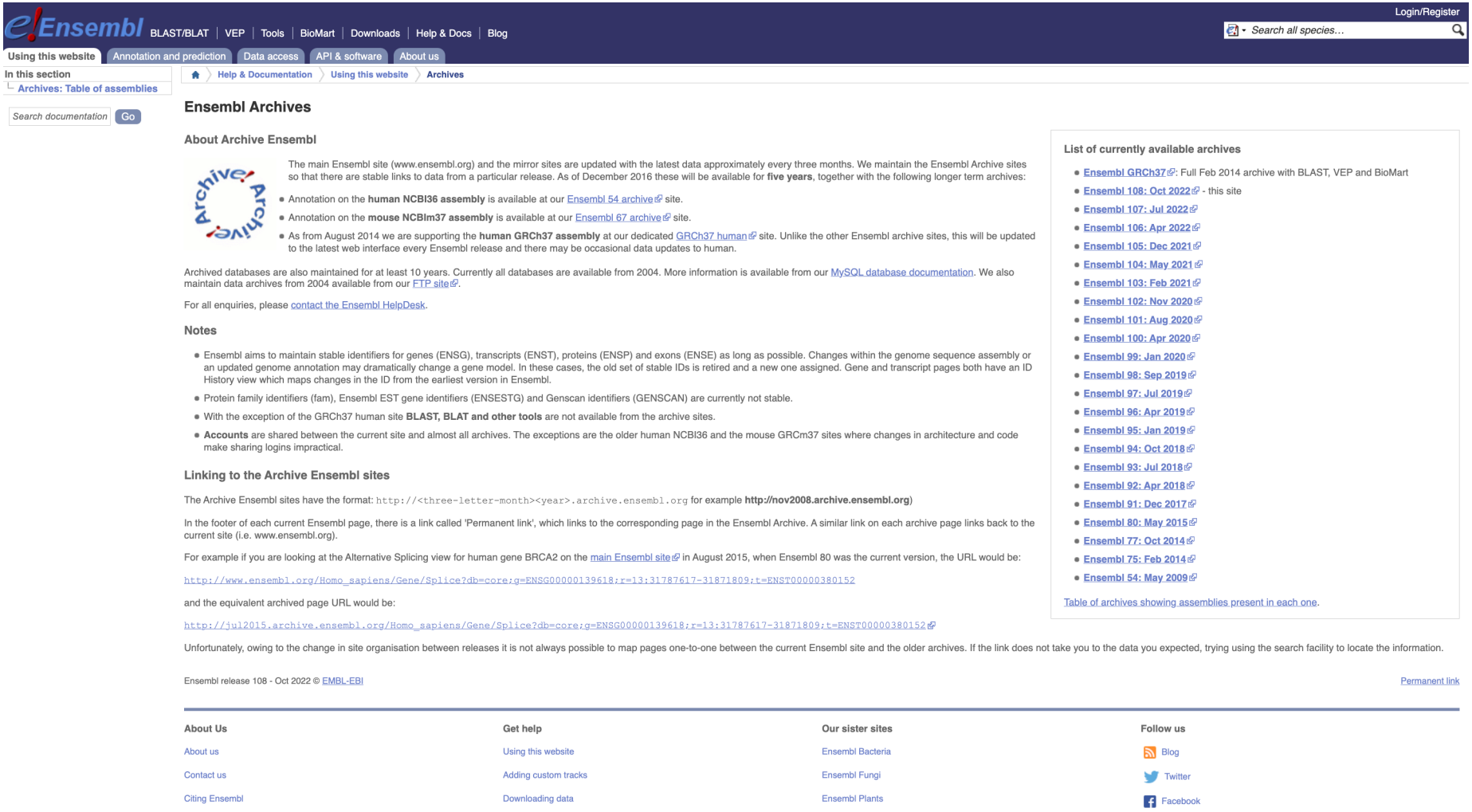
Version (Release)

- ~ tous les 3 mois
- Lien vers la dernière version d'Ensembl est toujours : http://www.ensembl.org



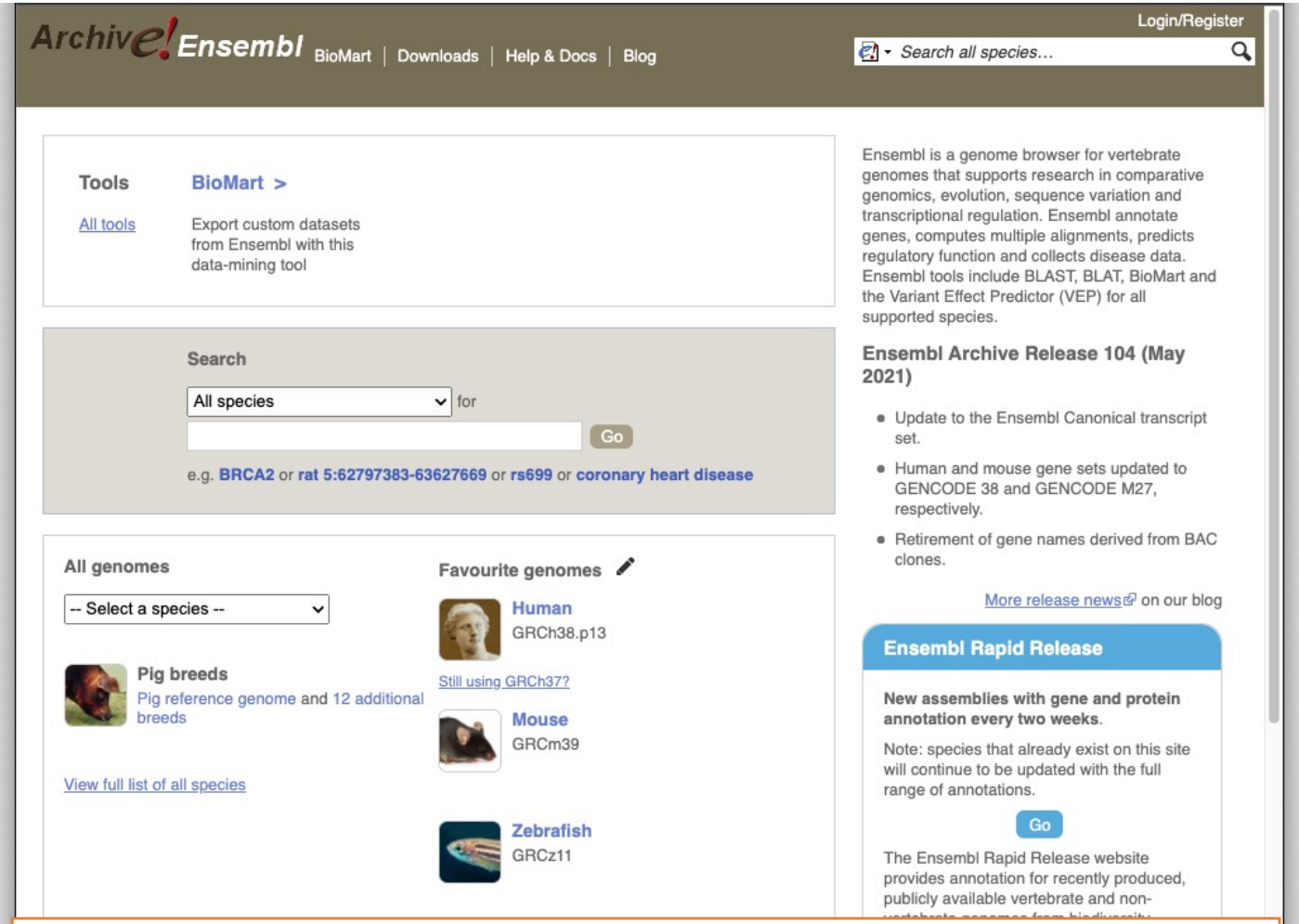
- Lien vers une version particulière d'Ensembl : http://Oct2022.archive.ensembl.org/index.html

Ensembl : Archives



http://www.ensembl.org/info/website/archives/index.html

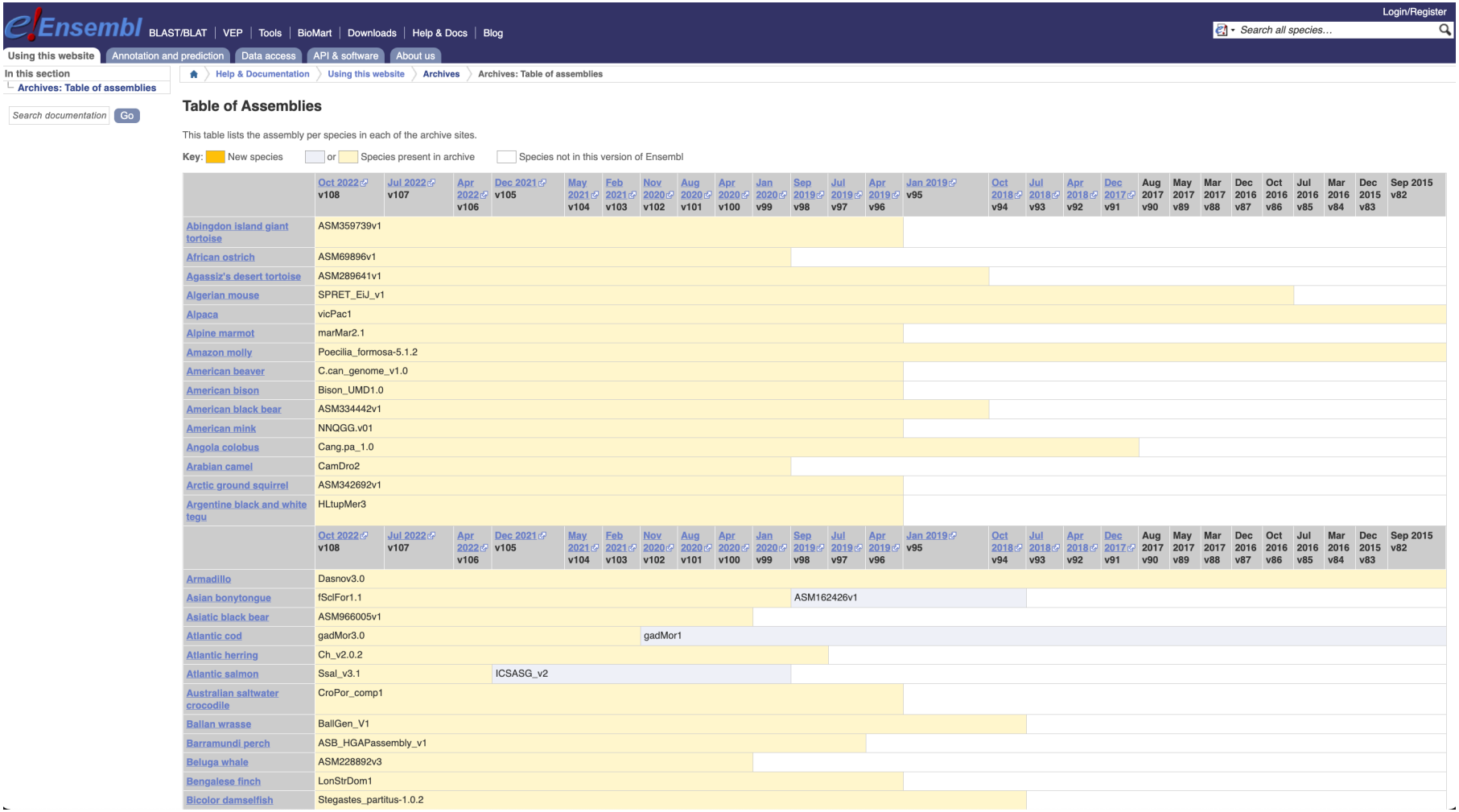
Ensembl : Archives



Les anciennes version d'Ensembl sont conservées pendant 5 ans sauf si elles contiennent la dernière version de l'annotation d'un génome.

Ensembl : Archives

- http://www.ensembl.org/info/website/archives/assembly.html



Ensembl BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog — Login/Register — Search all species…

Using this website | Annotation and prediction | Data access | API & software | About us

In this section: Archives: Table of assemblies

Search documentation — Go

Help & Documentation › Using this website › Archives › Archives: Table of assemblies

Table of Assemblies

This table lists the assembly per species in each of the archive sites.

Key: New species | or | Species present in archive | Species not in this version of Ensembl

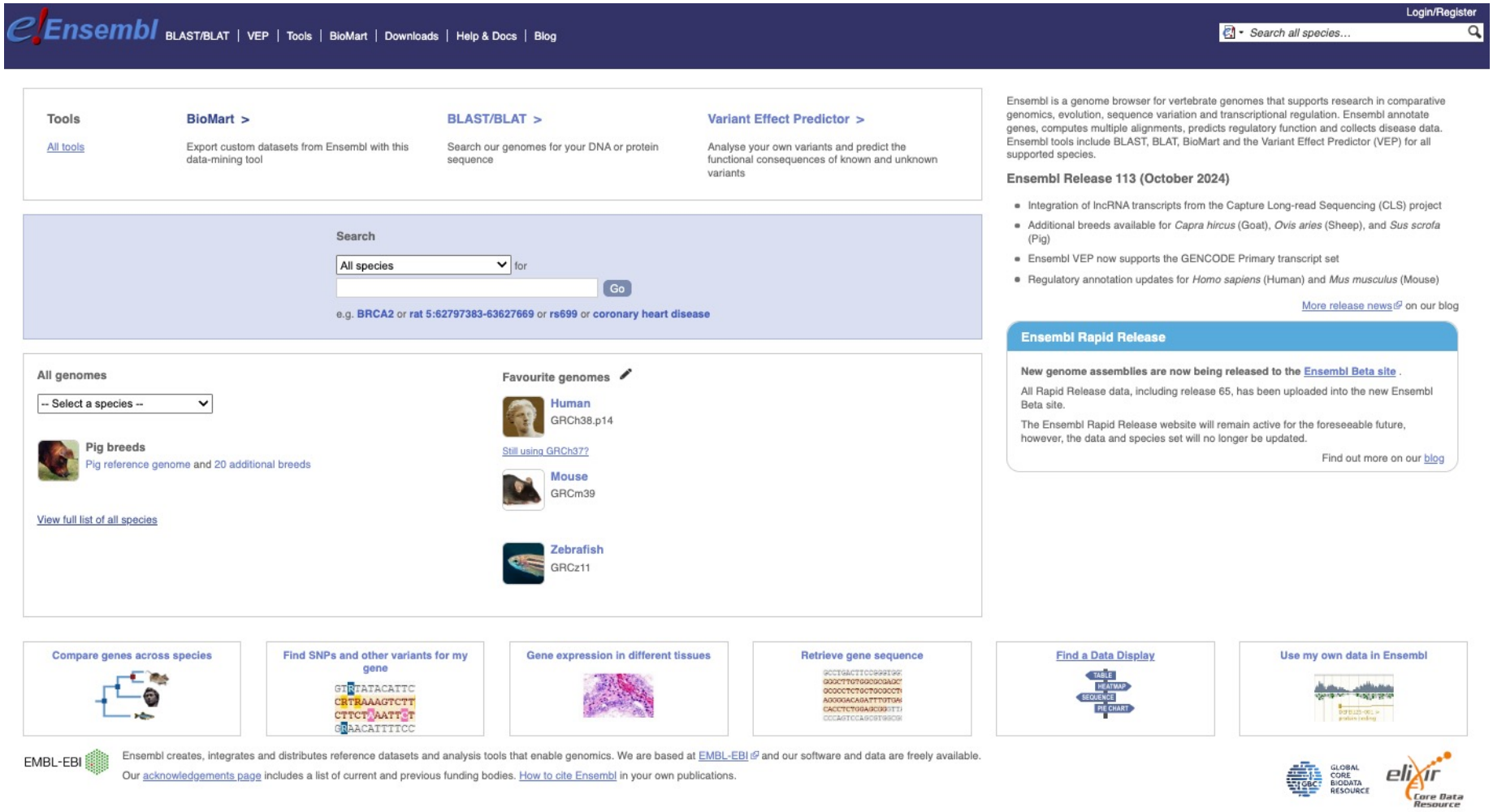
	Oct 2022 v108	Jul 2022 v107	Apr 2022 v106	Dec 2021 v105	May 2021 v104	Feb 2021 v103	Nov 2020 v102	Aug 2020 v101	Apr 2020 v100	Jan 2020 v99	Sep 2019 v98	Jul 2019 v97	Apr 2019 v96	Jan 2019 v95	Oct 2018 v94	Jul 2018 v93	Apr 2018 v92	Dec 2017 v91	Aug 2017 v90	May 2017 v89	Mar 2017 v88	Dec 2016 v87	Oct 2016 v86	Jul 2016 v85	Mar 2016 v84	Dec 2015 v83	Sep 2015 v82
Abingdon island giant tortoise	ASM359739v1																										
African ostrich	ASM69896v1																										
Agassiz's desert tortoise	ASM289641v1																										
Algerian mouse	SPRET_EiJ_v1																										
Alpaca	vicPac1																										
Alpine marmot	marMar2.1																										
Amazon molly	Poecilia_formosa-5.1.2																										
American beaver	C.can_genome_v1.0																										
American bison	Bison_UMD1.0																										
American black bear	ASM334442v1																										
American mink	NNQGG.v01																										
Angola colobus	Cang.pa_1.0																										
Arabian camel	CamDro2																										
Arctic ground squirrel	ASM342692v1																										
Argentine black and white tegu	HLtupMer3																										
	Oct 2022 v108	Jul 2022 v107	Apr 2022 v106	Dec 2021 v105	May 2021 v104	Feb 2021 v103	Nov 2020 v102	Aug 2020 v101	Apr 2020 v100	Jan 2020 v99	Sep 2019 v98	Jul 2019 v97	Apr 2019 v96	Jan 2019 v95	Oct 2018 v94	Jul 2018 v93	Apr 2018 v92	Dec 2017 v91	Aug 2017 v90	May 2017 v89	Mar 2017 v88	Dec 2016 v87	Oct 2016 v86	Jul 2016 v85	Mar 2016 v84	Dec 2015 v83	Sep 2015 v82
Armadillo	Dasnov3.0																										
Asian bonytongue	fSclFor1.1										ASM162426v1																
Asiatic black bear	ASM966005v1																										
Atlantic cod	gadMor3.0							gadMor1																			
Atlantic herring	Ch_v2.0.2																										
Atlantic salmon	Ssal_v3.1				ICSASG_v2																						
Australian saltwater crocodile	CroPor_comp1																										
Ballan wrasse	BallGen_V1																										
Barramundi perch	ASB_HGAPassembly_v1																										
Beluga whale	ASM228892v3																										
Bengalese finch	LonStrDom1																										
Bicolor damselfish	Stegastes_partitus-1.0.2																										

Aide et documentations

- Vidéo Youtube (workshop…)
- FAQ
- Exercices
- Cours en ligne
- Publications :
  - Flicek, P. et al. **Ensembl 2013**. Nucleic Acids Res. Advanced Access (Database Issue). http://www.ncbi.nlm.nih.gov/pubmed/23203987
  - Xosé M. Fernández-Suárez and Michael K. Schuster. **Using the Ensembl Genome Server to Browse Genomic Sequence Data**. UNIT 1.15 in Current Protocols in Bioinformatics, Jun 2010
  - Giulietta M Spudich and Xosé M Fernández Suárez. **Touring Ensembl: A practical guide to genome browsing**. BMC Genomics 2010, 11:295 (11 May 2010)

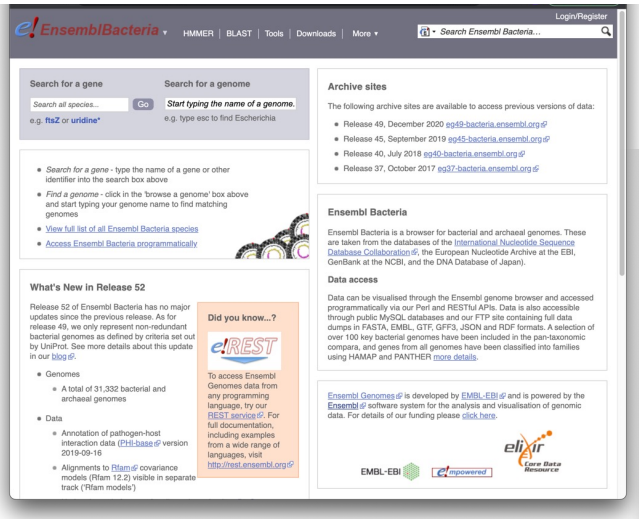
Naviguer dans ensembl

www.ensembl.org

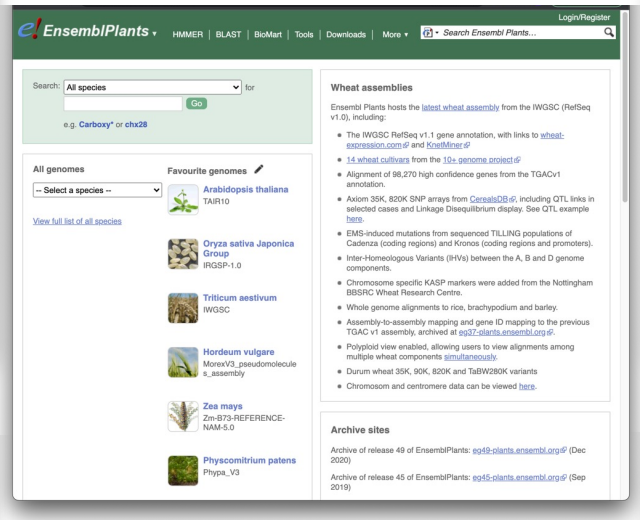


Ensembl Genomes

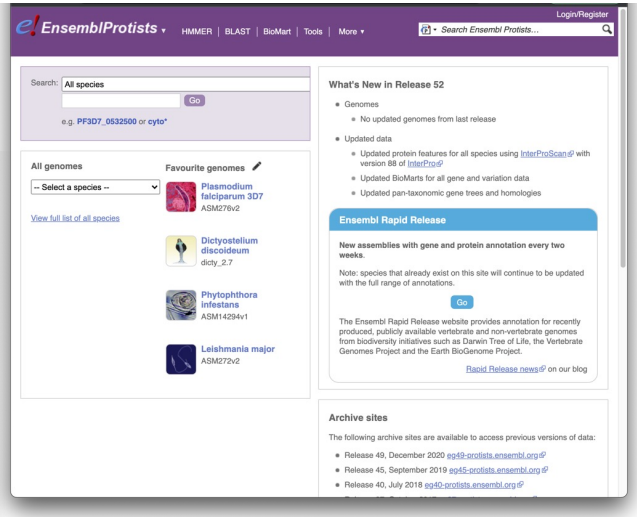
Bactéries



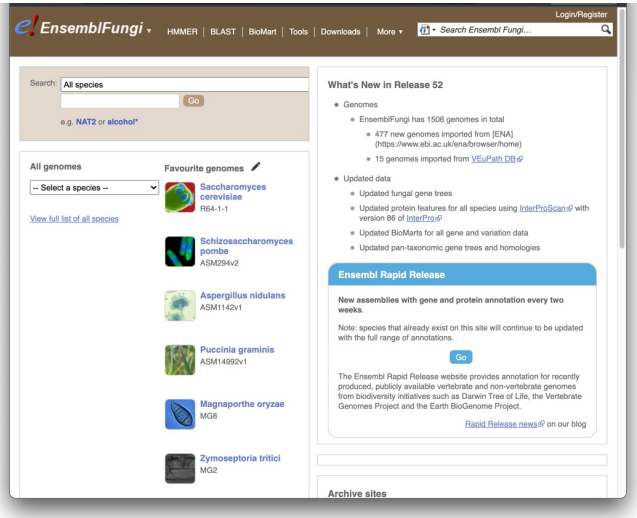
Plantes



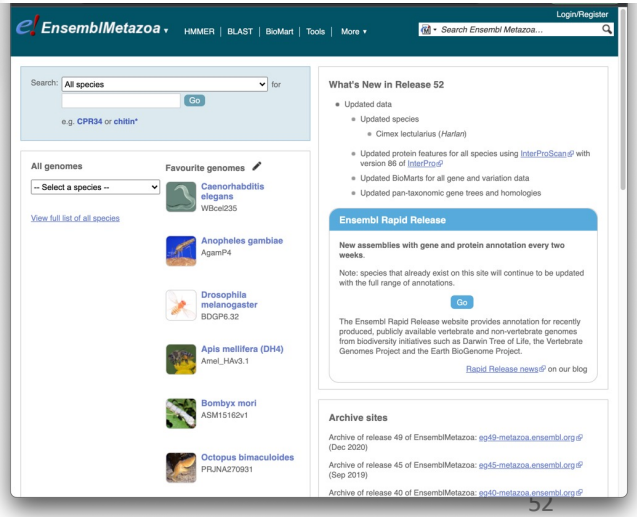
Protistes



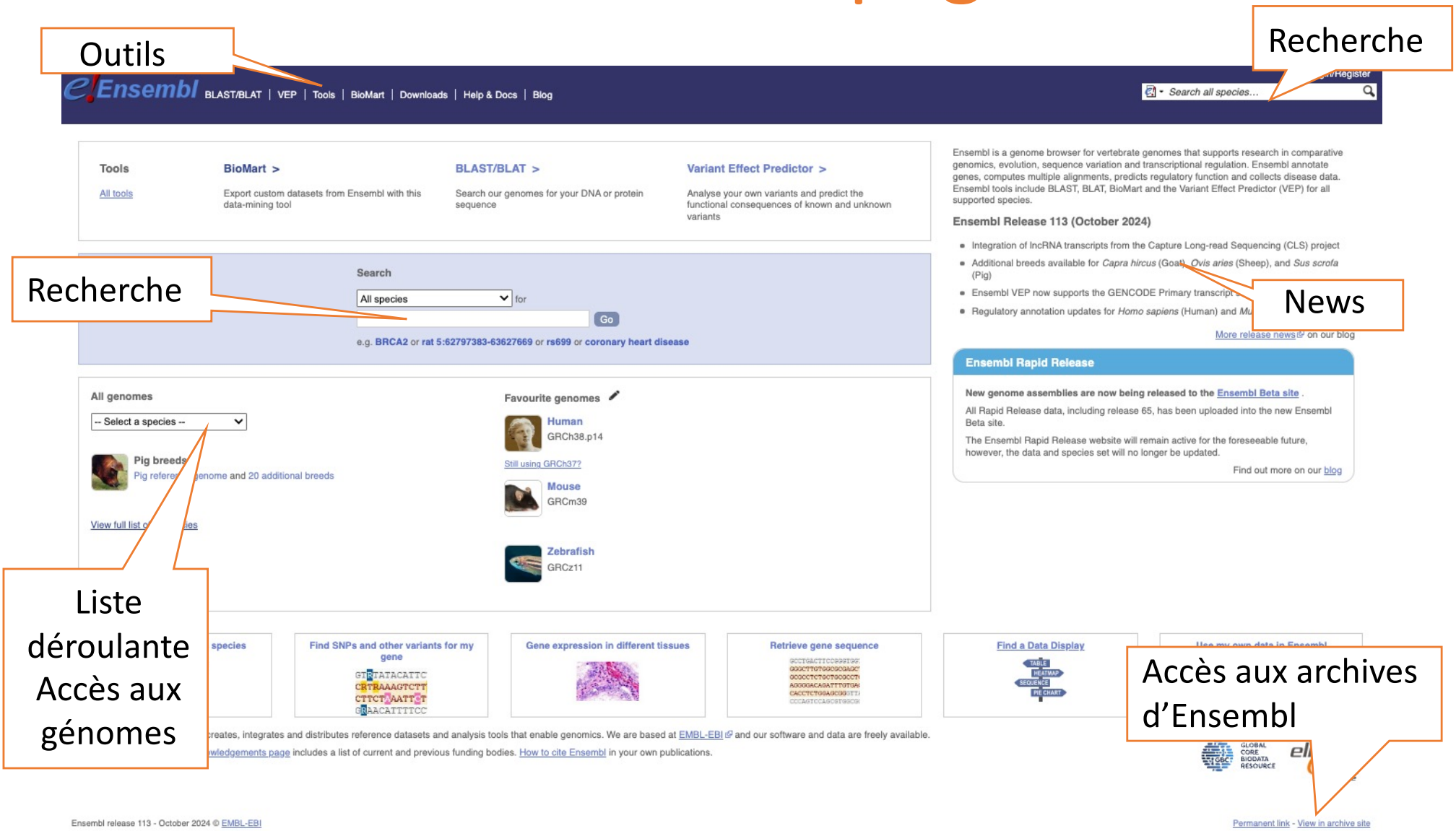
Fungi



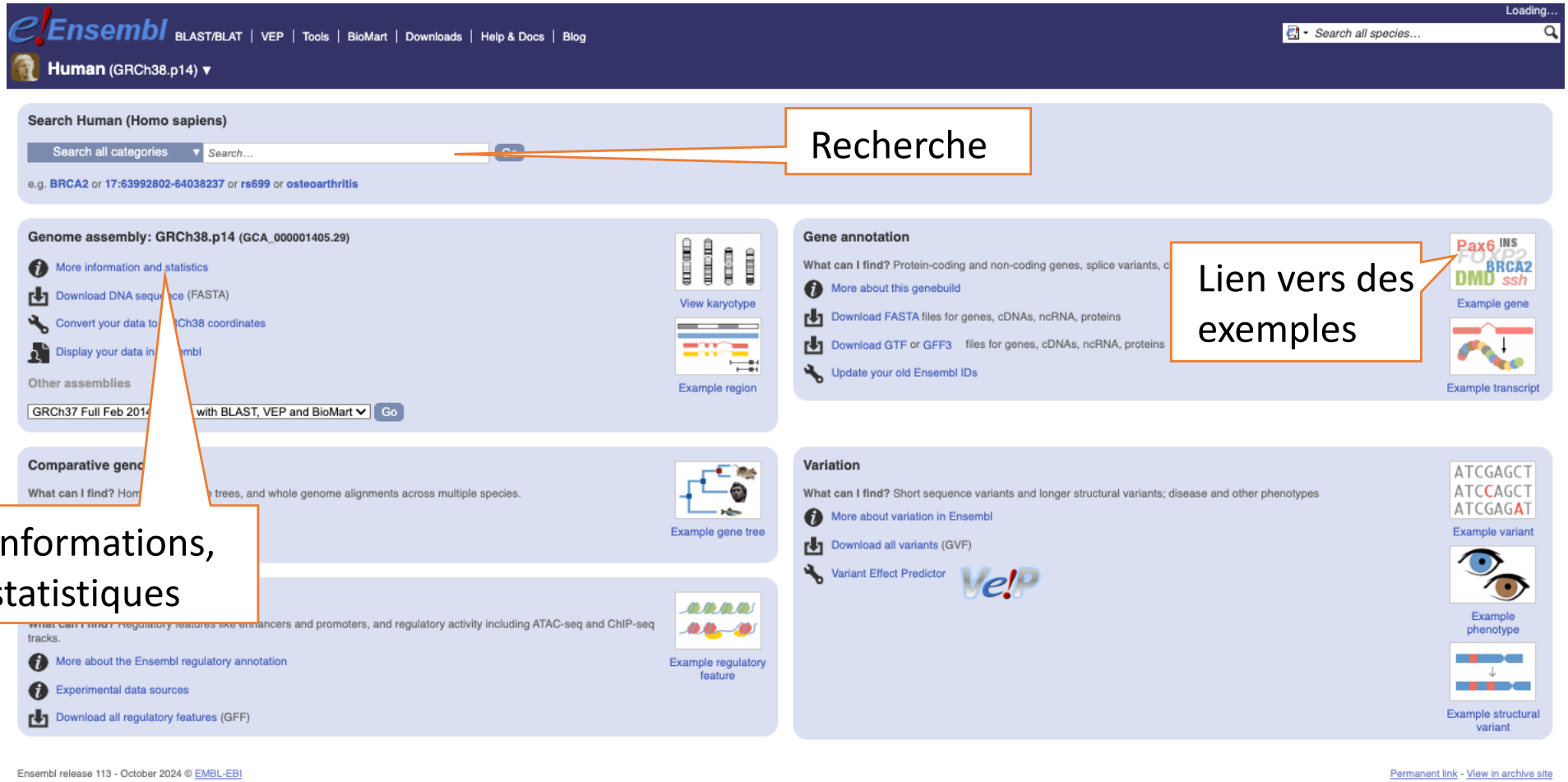
Métazoaires



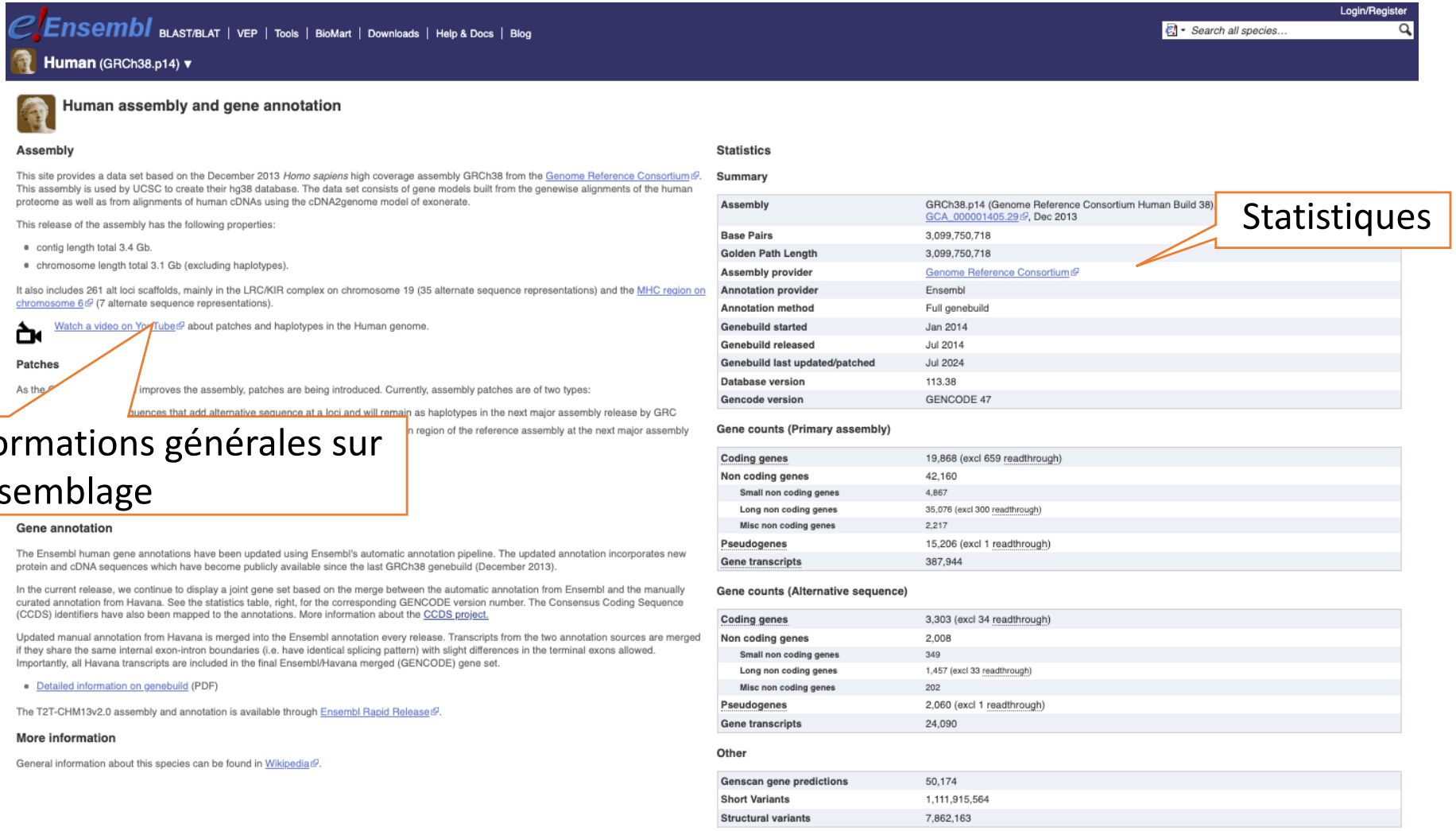
Le site web Ensembl: page d'accueil



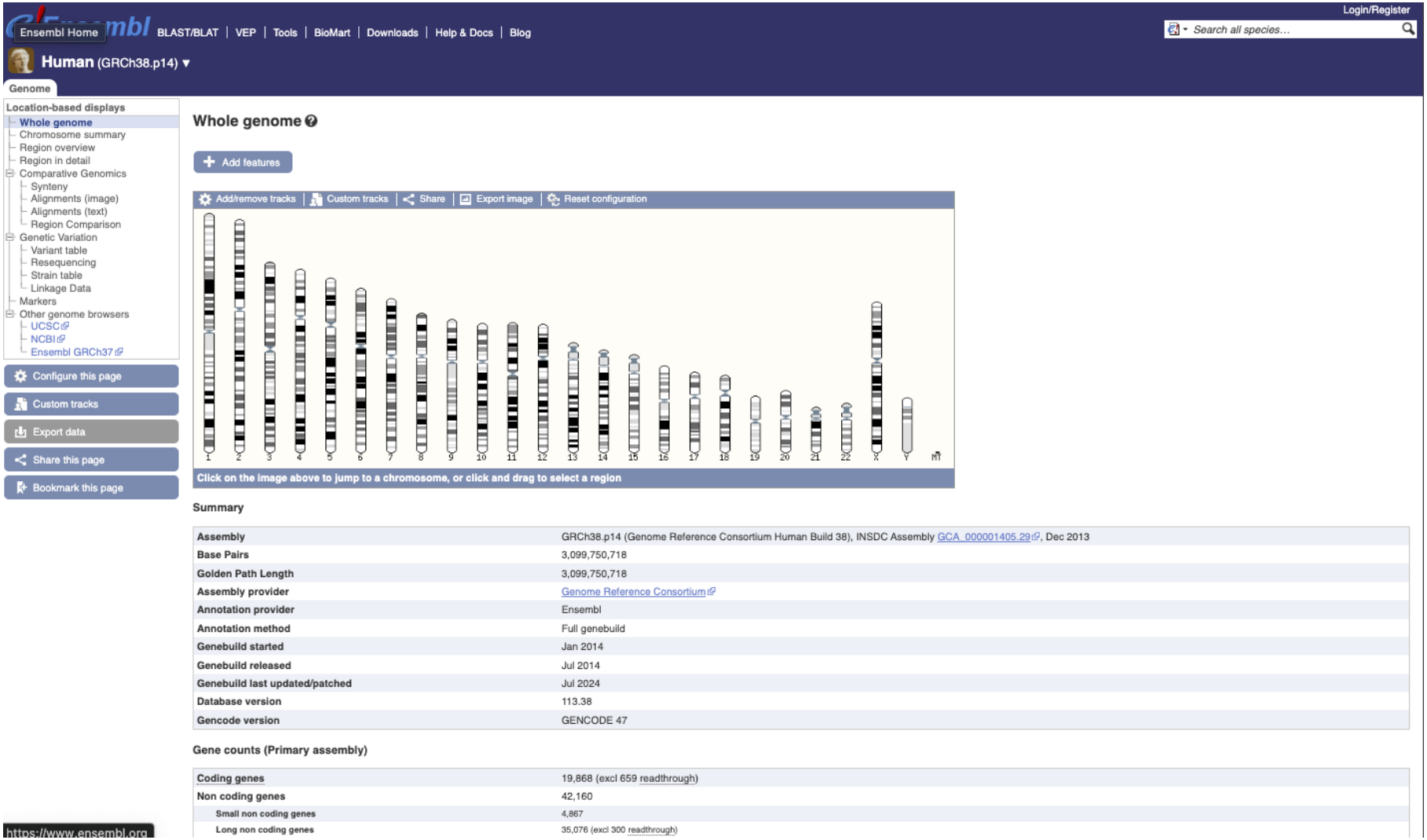
Le site web Ensembl: les génomes



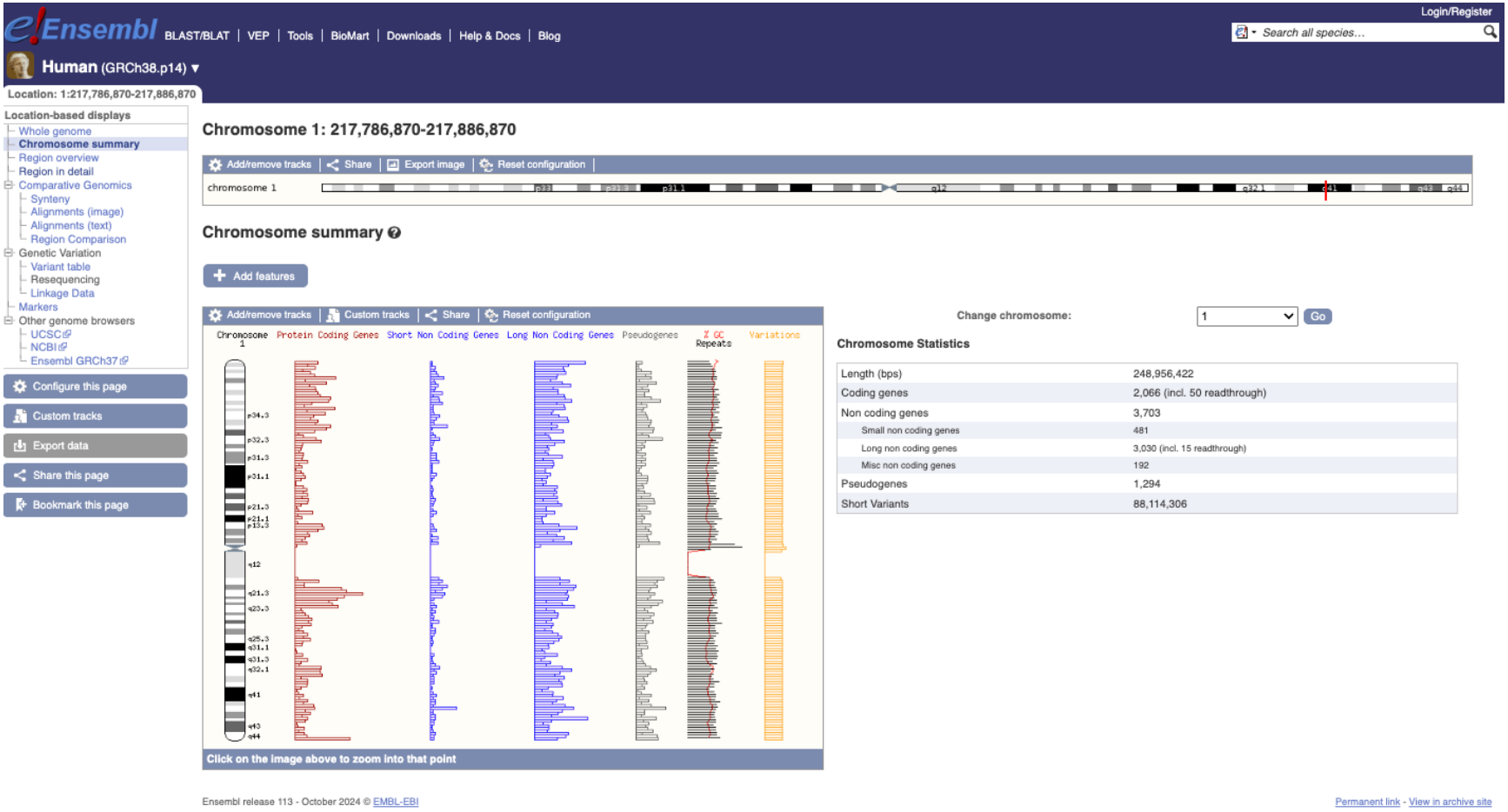
Le site web Ensembl: statistiques des génomes



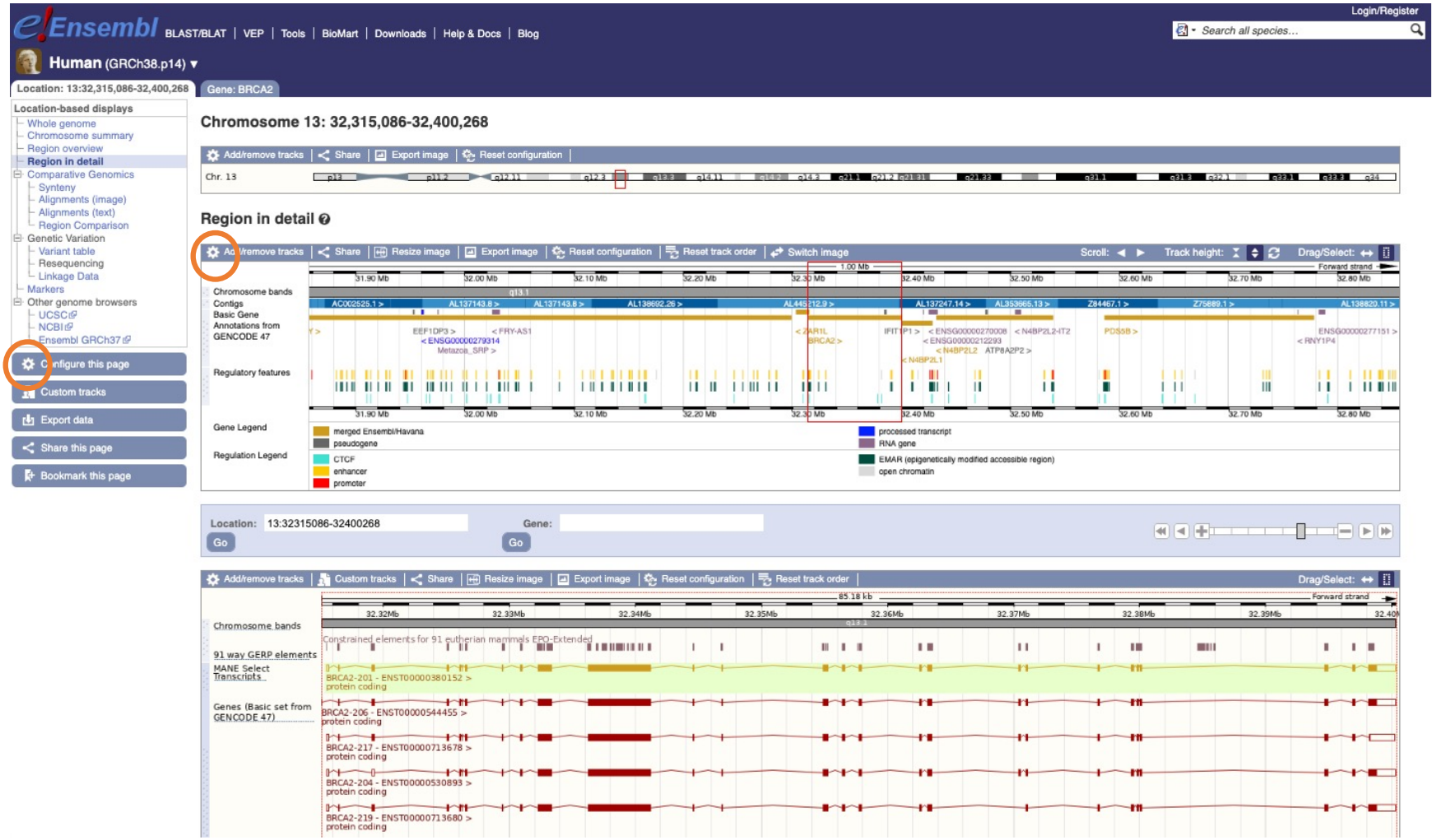
Le site web Ensembl: caryotype



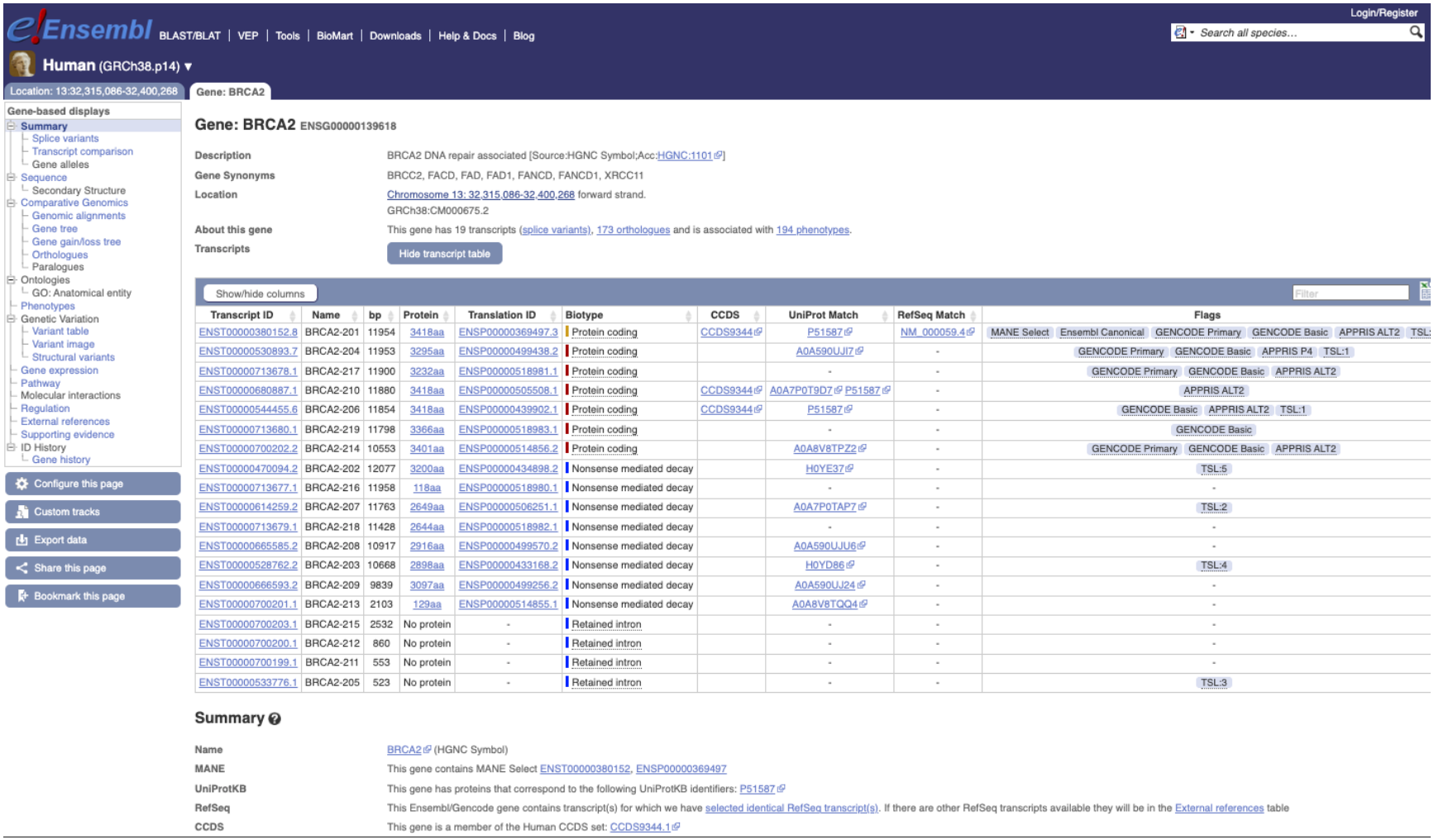
Le site web Ensembl : statistiques par chromosome



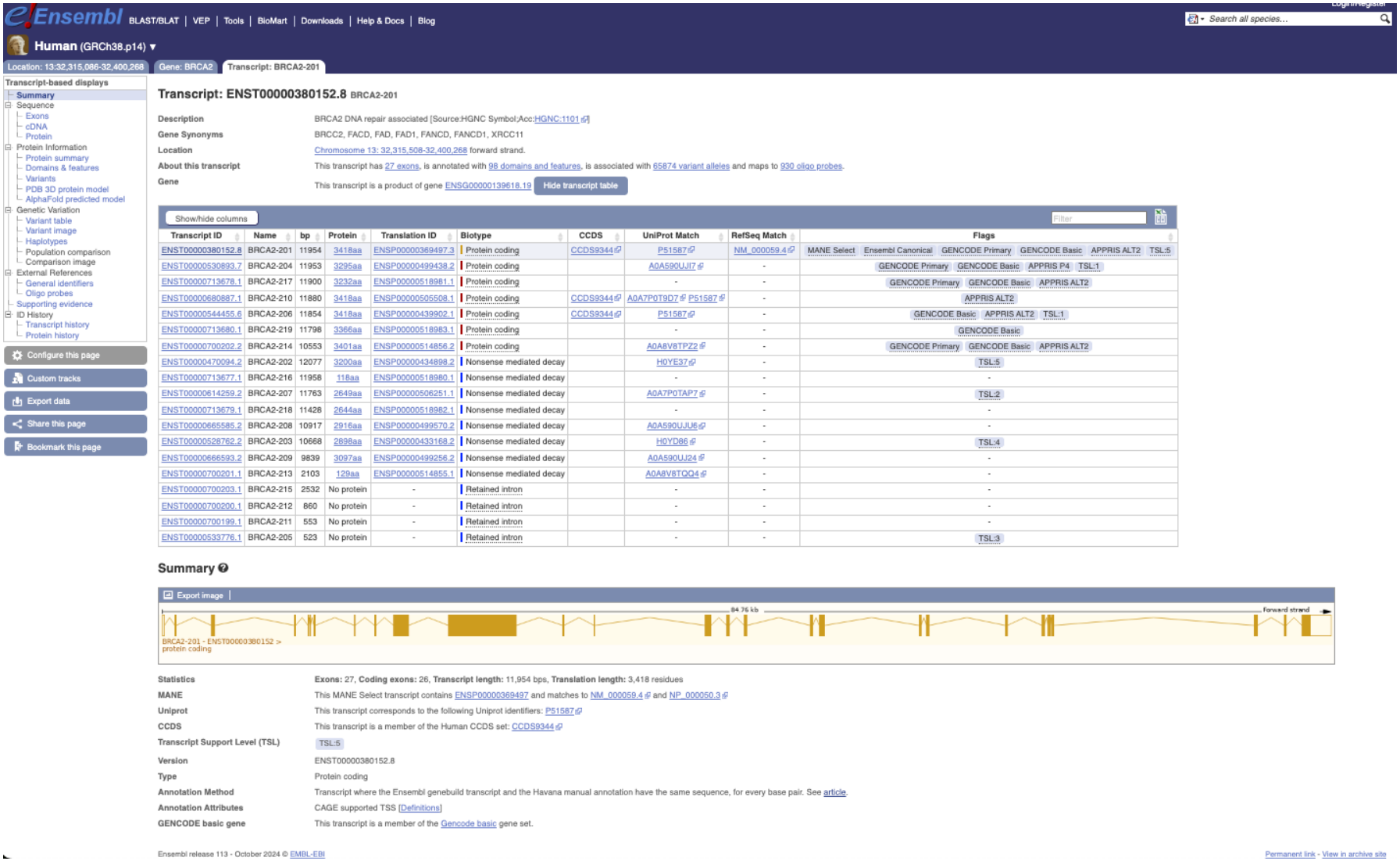
Le site web Ensembl : navigateur de génome



Le site web Ensembl : le gène

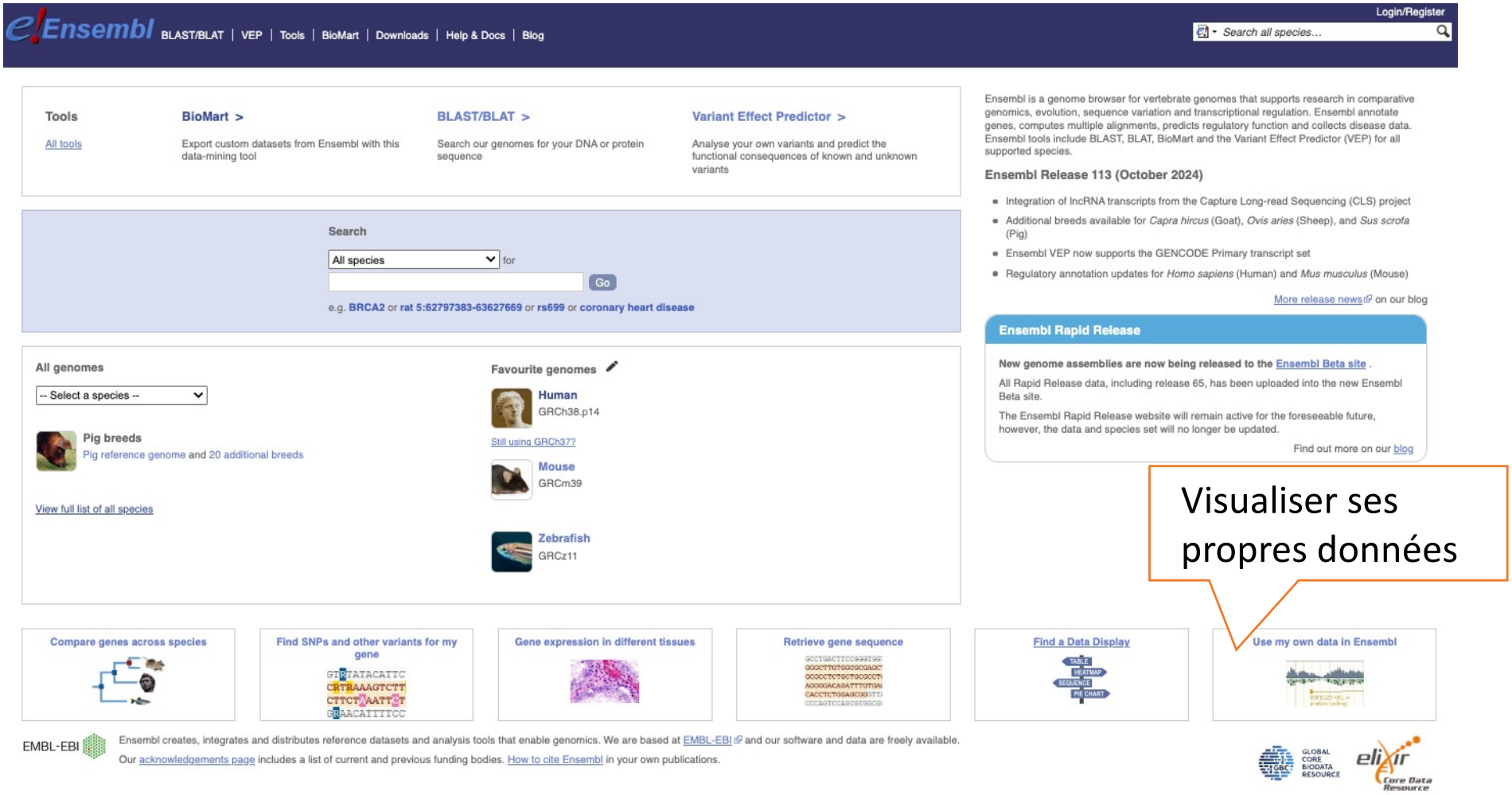


Le site web Ensembl : le transcrit



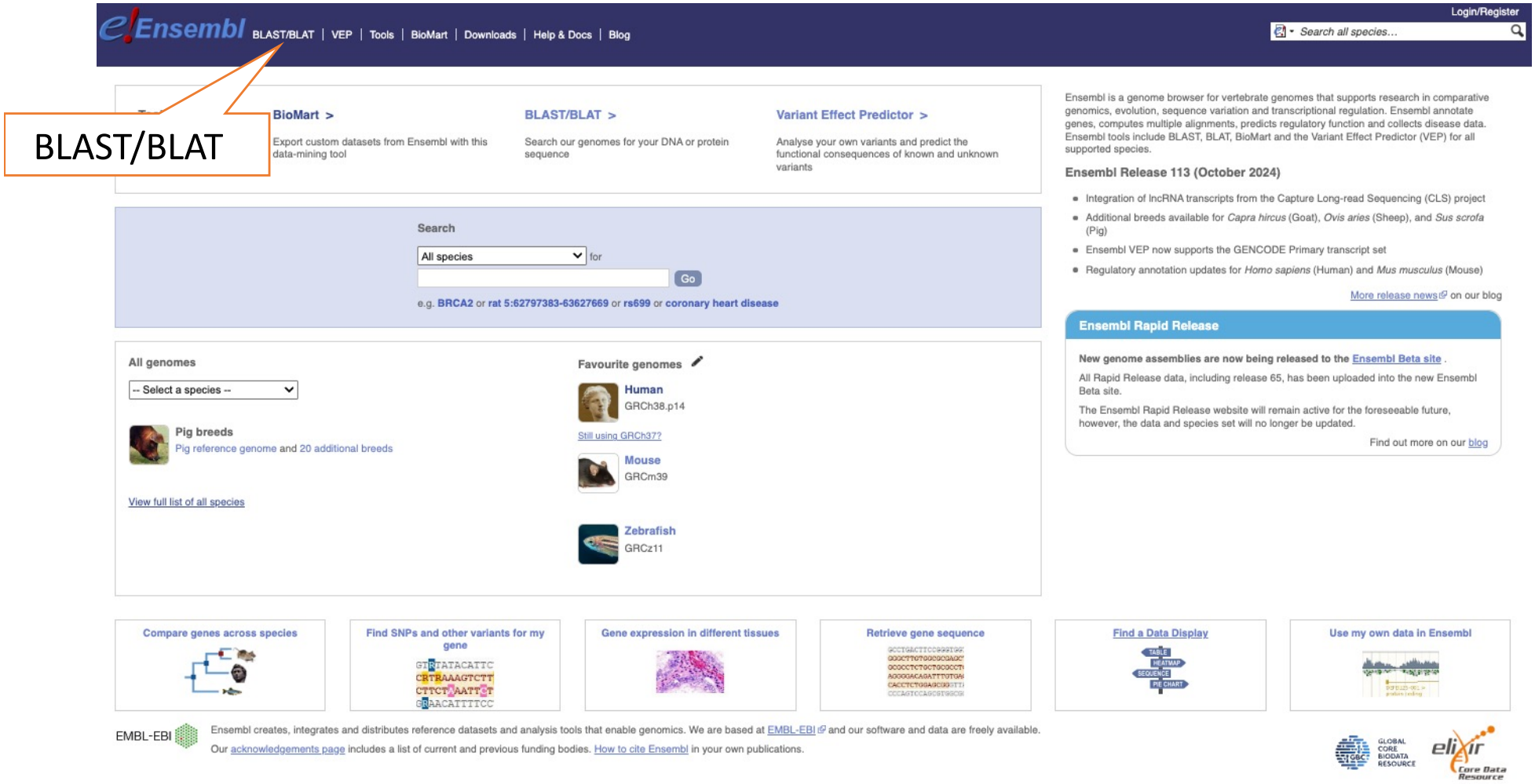
Naviguer dans Ensembl : Partie pratique

Visualiser ses propres données

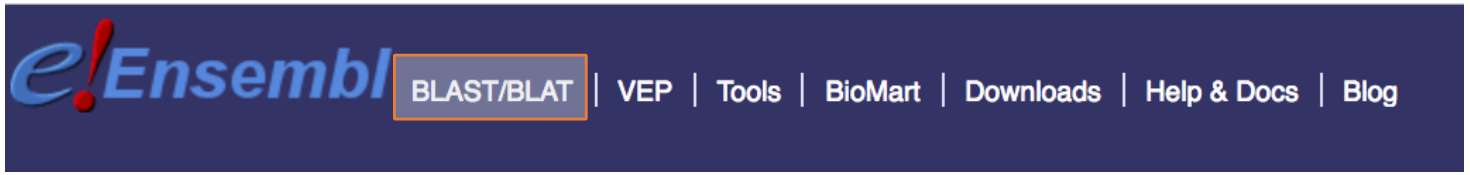


LES OUTILS

Les outils


BLAST/BLAT
Ensembl
BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog
Login/Register
Search all species…
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 --
Pig breeds
Pig reference genome and 20 additional breeds
View full list of all species
Favourite genomes
Human
GRCh38.p14
Still using GRCh37?
Mouse
GRCm39
Zebrafish
GRCz11
Ensembl is a genome browser for vertebrate genomes that supports research in comparative genomics, evolution, sequence variation and transcriptional regulation. Ensembl annotate genes, computes multiple alignments, predicts regulatory function and collects disease data. Ensembl tools include BLAST, BLAT, BioMart and the Variant Effect Predictor (VEP) for all supported species.
Ensembl Release 113 (October 2024)
Integration of lncRNA transcripts from the Capture Long-read Sequencing (CLS) project
Additional breeds available for Capra hircus (Goat), Ovis aries (Sheep), and Sus scrofa (Pig)
Ensembl VEP now supports the GENCODE Primary transcript set
Regulatory annotation updates for Homo sapiens (Human) and Mus musculus (Mouse)
More release news on our blog
Ensembl Rapid Release
New genome assemblies are now being released to the Ensembl Beta site .
All Rapid Release data, including release 65, has been uploaded into the new Ensembl Beta site.
The Ensembl Rapid Release website will remain active for the foreseeable future, however, the data and species set will no longer be updated.
Find out more on our blog
Compare genes across species
Find SNPs and other variants for my gene
Gene expression in different tissues
Retrieve gene sequence
Find a Data Display
Use my own data in Ensembl
EMBL-EBI
Ensembl creates, integrates and distributes reference datasets and analysis tools that enable genomics. We are based at EMBL-EBI and our software and data are freely available.
Our acknowledgements page includes a list of current and previous funding bodies. How to cite Ensembl in your own publications.
Ensembl release 113 - October 2024 © EMBL-EBI
Permanent link - View in archive site

Blast



- Recherche de similarité
  - 1 séquence (***Query***) comparée à des milliers ou des millions de séquences (***base de données***) par comparaison 2 à 2.
- But:
  - Détecter des séquences proches
  - Annotation simple (domaines protéiques, localisation génomique, nombre d'exons)

Les différentes comparaisons

BLAST : Basic Local Alignment Search Tool

Altschul *et al.* Basic local alignment search tool. *J. Mol. Biol. 1990*
Altschul *et al.* Gapped BLAST and PSI-BLAST: a new generation of protein database search programs. *Nucleic Acids Res 1997*

Programmes	Requête	Banque	Comparaison	Exemples d'utilisation
Blastn	ADN	ADN	nucléique	Recherche d'ARN structuraux, d'éléments régulateurs
Blastp	Protéine	protéines	protéique	Recherche de protéines homologues
Tblastn	Protéine	ADN (traduit dans les 6 cadres)	protéique	Recherche de similarités entre une protéine et une séquence génomique mal annotée
Blastx	ADN (traduit dans les 6 cadres)	protéines	protéique	Recherche des phases de lecture dans une séquence codante
Tblastx	ADN (traduit dans les 6 cadres)	ADN (traduit dans les 6 cadres)	protéique	Avantages de tblastn et blastx mais très long

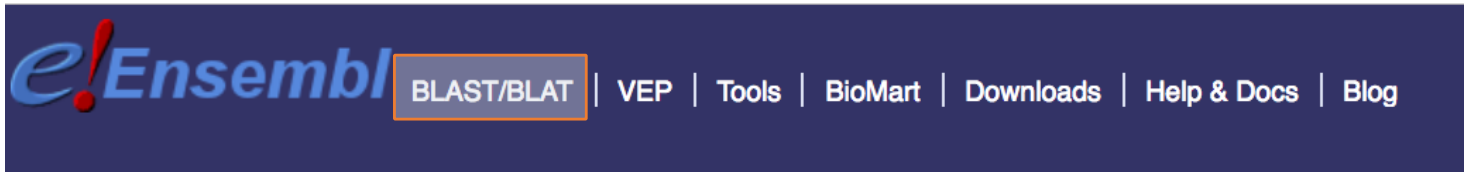
Les différentes comparaisons

BLAT (BLAST-Like Alignment Tool)

- An mRNA/DNA and cross-species protein sequence analysis tool to quickly find sequences of ≥ 95% similarity of length ≥40 bases.
- was developed by Jim Kent at the University of California Santa Cruz (UCSC) in the early 2000s to assist in the assembly and annotation of the human genome.
- The target database of BLAT is not a set of GenBank sequences, but instead an index derived from the assembly of the entire genome. **Blat works by keeping an index of an entire genome in memory.**
- By default, the index consists of all non-overlapping 11-mers for DNA and 4-mers for protein.

- Kent, W.J.. BLAT -- The BLAST-Like Alignment Tool. *Genome Research 2002*

Blast



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QTCGELMFSLEPRLRHLGLGKEGITTYFSGNCTMEDAKLAQDFLDSQNLSAYNTRLFKEVDGEGKPYYEVRL
ASVLGSEPSLDSEVTSKLKSYEFRGSPFQVTRGDYAPILQKVVEQLEKAKAYAANSHQGQMLAQYIESFTQG
SIEAHKRGSRFWIQDKGPIVESYIGFIESYRDPFGSRGEFEGFVAVVNKAMSAKFERLVASAEQLLKELPWP
PTFEKDKFLTPDFTSLDVLTFAGSGIPAGINIPNYDDLRQTEGFKNVSLGNVLAVAYATQREKLTFLEEDDK
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SYEECRAESVGLYLCLHPQVLEIFGFEGADAEDVIYVNWLNMVRAGLLALEFYTPEAFNWRQAHMQARFVIL
RVLLEAGEGLVTITPTTGSDGRPDARVRLDRSKIRSVGKPALERFLRRLQVLKSTGDVAGGRALYEGYATVT
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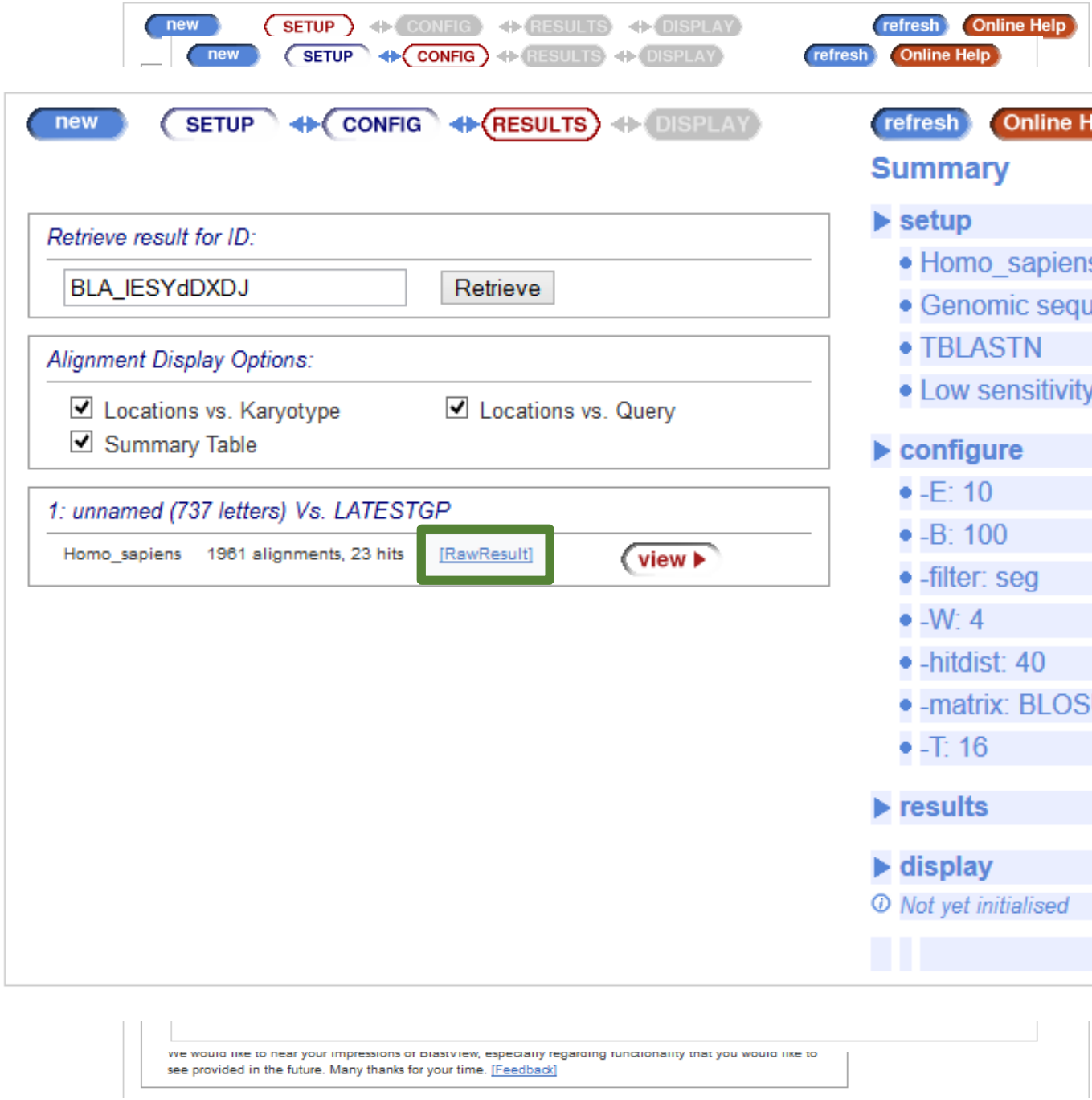
new
SETUP
CONFIG
RESULTS
DISPLAY
refresh
Online Help
Retrieve result for ID:
BLA_IESYdDXDJ
Retrieve
Alignment Display Options:
Locations vs. Karyotype
Locations vs. Query
Summary Table
1: unnamed (737 letters) Vs. LATESTGP
Homo_sapiens 1961 alignments, 23 hits
[RawResult]
view
Summary
setup
Homo_sapiens
Genomic sequence
TBLASTN
Low sensitivity
configure
-E: 10
-B: 100
-filter: seg
-W: 4
-hitdist: 40
-matrix: BLOSUM80
-T: 16
results
display
Not yet initialised
see provided in the future. Many thanks for your time. [Feedback]

```
Content-type: text/plain

TBLASTN 2.0MP-WashU [04-May-2006] [linux26-x64-I32LPF64 2006-05-10T17:22:28]

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All Rights Reserved.

Reference:  Gish, W. (1996-2006) http://blast.wustl.edu

Query=  unnamed
        (737 letters)

WARNING:  Precomputed values for Lambda, K and H are unavailable for the
          BLOSUM80 scoring matrix, when used with gap penalties +9 and +2.
          Unless overridden on the command line, the values computed for
          ungapped alignments will be used instead, but the reported E-values
          and P-values may be much too low.

Database:  Homo_sapiens.GRCh37.dna.toplevel.fa
           297 sequences; 32,036,512,383 total letters.

WARNING:  Use of the hspsepSmax parameter should be considered with long
          database sequences, to improve the biological relevance of the HSP
          groups that are assembled and to improve the statistical
          discrimination of these groups from random background.
Searching....10....20....30....40....50....60....70....80....90....100% done

WARNING:  hspmax=1000 was exceeded by 37 of the database sequences, causing
          the associated cutoff score, S2, to be transiently set as high as
          73.
```

Sequences producing High-scoring Segment Pairs:	Reading Frame	High Score	Smallest Sum Probability P(N)	N
9 dna:chromosome chromosome:GRCh37:9:1:141213431:1 REF	-3	1765	0.	6
11 dna:chromosome chromosome:GRCh37:11:1:135006516:1 REF	+3	763	3.2e-292	9
4 dna:chromosome chromosome:GRCh37:4:1:191154276:1 REF	+3	1542	5.5e-250	4
20 dna:chromosome chromosome:GRCh37:20:1:63025520:1 REF	-1	131	0.0035	9
16 dna:chromosome chromosome:GRCh37:16:1:90354753:1 REF	+1	120	0.014	10
12 dna:chromosome chromosome:GRCh37:12:1:133851895:1 REF	-2	126	0.060	11
19 dna:chromosome chromosome:GRCh37:19:1:59128983:1 REF	-1	128	0.069	9
22 dna:chromosome chromosome:GRCh37:22:1:51304566:1 REF	+1	130	0.10	10
GL000199.1 dna:supercontig supercontig:GRCh37:GL000199.1:...	+3	149	0.11	2
14 dna:chromosome chromosome:GRCh37:14:1:107349540:1 REF	+2	167	0.21	8
1 dna:chromosome chromosome:GRCh37:1:1:249250621:1 REF	-1	134	0.25	8
GL000220.1 dna:supercontig supercontig:GRCh37:GL000220.1:...	-3	124	0.26	4
5 dna:chromosome chromosome:GRCh37:5:1:180915260:1 REF	+1	127	0.33	9
GL000224.1 dna:supercontig supercontig:GRCh37:GL000224.1:...	-2	126	0.49	2
7 dna:chromosome chromosome:GRCh37:7:1:159138663:1 REF	-3	129	0.88	9
21 dna:chromosome chromosome:GRCh37:21:1:48129895:1 REF	-2	131	0.98	9
GL000237.1 dna:supercontig supercontig:GRCh37:GL000237.1:...	-2	89	0.98	5
GL000202.1 dna:supercontig supercontig:GRCh37:GL000202.1:...	+1	111	0.995	3
GL000218.1 dna:supercontig supercontig:GRCh37:GL000218.1:...	-1	145	0.996	5
15 dna:chromosome chromosome:GRCh37:15:1:102531392:1 REF	+2	134	0.999	12
6 dna:chromosome chromosome:GRCh37:6:1:171115067:1 REF	-2	118	0.9991	13
3 dna:chromosome chromosome:GRCh37:3:1:198022430:1 REF	-3	118	0.9998	11
GL000206.1 dna:supercontig supercontig:GRCh37:GL000206.1:...	-3	92	0.99992	6

```
>9 dna:chromosome chromosome:GRCh37:9:1:141213431:1 REF
       Length = 141,213,431

 Score = 1765 (578.9 bits), Expect = 0., Sum P(6) = 0.
 Identities = 220/261 (84%), Positives = 230/261 (88%), Frame = -3

Query:      477 INPETGEQIQSWYRSGETWDSKFSTIASSYEECRAESVGLYLCLHPQVLEIFGFEGADAE 536
               INPE  EQIQSWYRS +TWDSKFSTI SSYEECRAESVGLYLCLHPQVLE FGFEGADAE
Sbjct: 76090065 INPEMREQIQSWYRSMKTWDSKFSTIVSSYEECRAESVGLYLCLHPQVLETFGFEGADAE 76089886

Query:      537 DVIYVNWLNMVRAGLLALEFYTPEAFNWRQAHMQARFVILRVLLEAGEGLVTITPTTGSD 596
               +VI VNWLNMV AGLLALEFYTPEA NW+QAH++AR VILRVL EAGEGL TITPT GSD
Sbjct: 76089885 EVISVNWLNMVGAGLLALEFYTPEASNWQQAHIRARIVILRVLPEAGEGLGTITPTAGSD 76089706

Query:      597 GRPDARVRLDRSKIRSVGKPALERFLRRLQVLKSTGDVAGGRALYEGYATVTDAPPECFL 656
               GRP+A+VRLDRSKI+SVG PALERFLRR     STGDVAGG  LYE YA V DAPPE FL
Sbjct: 76089705 GRPEAQVRLDRSKIQSVGNPALERFLRRCW---STGDVAGGWTLYERYAAVADAPPEGFL 76089535

Query:      657 TLRDTVLLRKESRKLIVQPNTRLEGSDVQLLEYEASAAGLIRSFSERFPEDGPELEEILT 716
               TLRD VLLRKES KLIVQPN RLEGSDVQLLEYE SAAGLIRSFSE FPEDG ELE+ILT
Sbjct: 76089534 TLRDRVLLRKESWKLIVQPNIRLEGSDVQLLEYEVSAAGLIRSFSEHFPEDGLELEDILT 76089355

Query:      717 QLATADARFWKGPSEAPSGQA 737
               QLATADA+F KGPSEAPSGQA
Sbjct: 76089354 QLATADAQF*KGPSEAPSGQA 76089292

 Score = 1700 (557.6 bits), Expect = 0., Sum P(6) = 0.
 Identities = 212/252 (84%), Positives = 221/252 (87%), Frame = -2

Query:      224 PSLDSEVTSKLKSYEFRGSPFQVTRGDYAPILQKVVEQLEKAKAYAANSHQGQMLAQYIE 283
               P L  +  SKLKS EFRGSPFQVT G+Y PILQKVVEQLEKAK YAANSHQ QMLAQYIE
Sbjct: 76090816 PGLRGD--SKLKS*EFRGSPFQVTWGNYMPILQKVVEQLEKAKTYAANSHQEQMLAQYIE 76090643

Query:      284 SFTQGSIEAHKRGSRFWIQDKGPIVESYIGFIESYRDPFGSRGEFEGFVAVVNKAMSAKF 343
               SFTQGS EAHK+GSRFWI DKGPIVESYI FI+SYRD FGSRG  EGFVAVVNKAMSAKF
Sbjct: 76090642 SFTQGSTEAHKKGSRFWI*DKGPIVESYIEFIQSYRDSFGSRGVCEGFVAVVNKAMSAKF 76090463

Query:      344 ERLVASAEQLLKELPWPPTFEKDKFLTPDFTSLDVLTFAGSGIPAGINIPNYDDLRQTEG 403
               E LV SAEQLLKELPW P FEKDKFLTPDFTS+DVLTFAGSGI AGINI NY+DL+QTEG
Sbjct: 76090462 EWLVVSAEQLLKELPWSPAFEKDKFLTPDFTSVDVLTFAGSGIAAGINISNYNDLKQTEG 76090283

Query:      404 FKNVSLGNVLAVAYATQREKLTFLEEDDKDLYILWKGPSFDVQVGLHELLGHGSGKLFVQ 463
               FKNVSLGNVLAV  ATQ EKLT LEE DKDLYI+  GPSFDVQVGLHELLG+GSGKL  Q
Sbjct: 76090282 FKNVSLGNVLAVV*ATQWEKLTVLEESDKDLYIVLMGPSFDVQVGLHELLGYGSGKLIEQ 76090103

Query:      464 DEKGAFNFDQET 475
               DEKGAFNFDQET
Sbjct: 76090102 DEKGAFNFDQET 76090067
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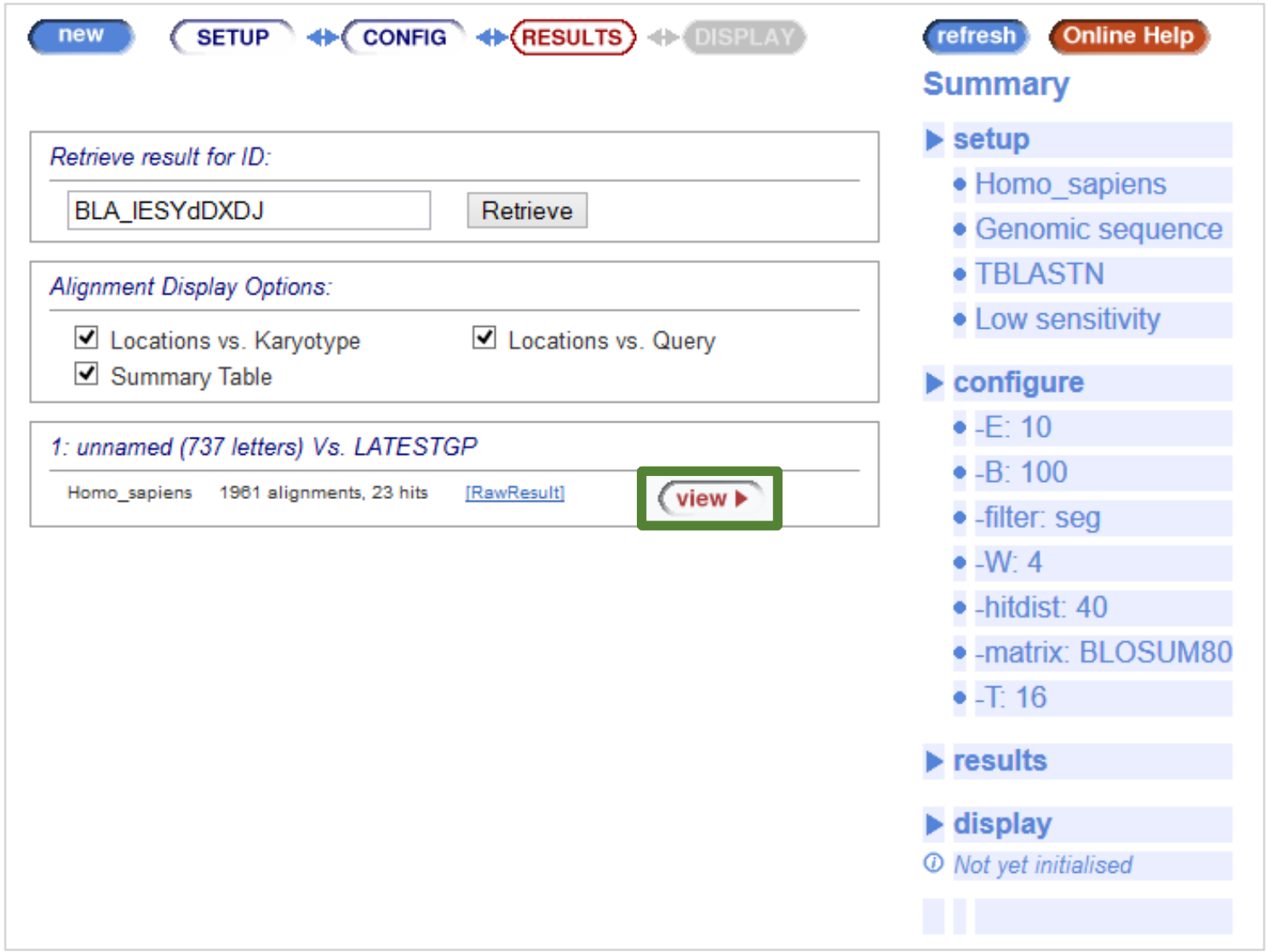

new
SETUP
CONFIG
RESULTS
DISPLAY
refresh
Online Help
Summary
Retrieve result for ID:
BLA_IESYdDXDJ
Retrieve
Alignment Display Options:
Locations vs. Karyotype
Locations vs. Query
Summary Table
1: unnamed (737 letters) Vs. LATESTGP
Homo_sapiens 1961 alignments, 23 hits
[RawResult]
view
setup
Homo_sapiens
Genomic sequence
TBLASTN
Low sensitivity
configure
-E: 10
-B: 100
-filter: seg
-W: 4
-hitdist: 40
-matrix: BLOSUM80
-T: 16
results
display
Not yet initialised

new | SETUP | CONFIG | RESULTS | DISPLAY

refresh | Online Help

Displaying unnamed sequence alignments vs Homo_sapiens LATESTGP database

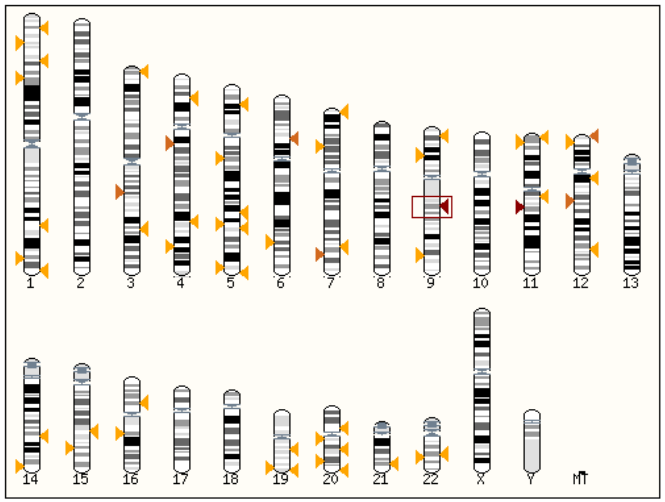
Showing top 100 **alignments of 1961, sorted by** Raw Score refresh

Summary

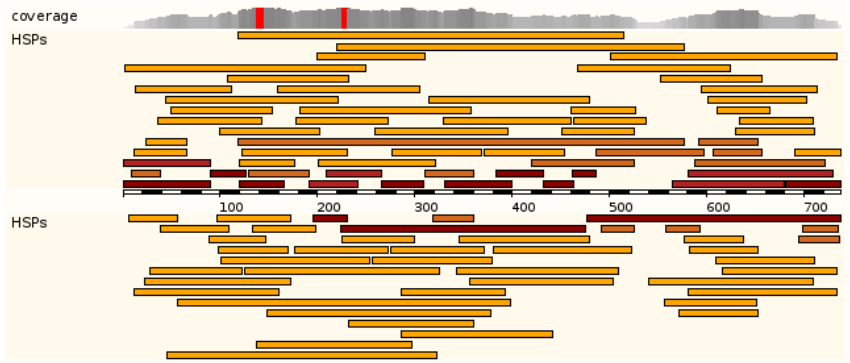
- **setup**
  - Homo_sapiens
  - Genomic sequence
  - TBLASTN
  - Low sensitivity
- **configure**
  - -E: 10
  - -B: 100
  - -filter: seg
  - -W: 4
  - -hitdist: 40
  - -matrix: BLOSUM80
  - -T: 16
- **results**
- **display**

Not yet initialised

Alignment Locations vs. Karyotype (click arrow to hide)



Alignment Locations vs. Query (click arrow to hide)



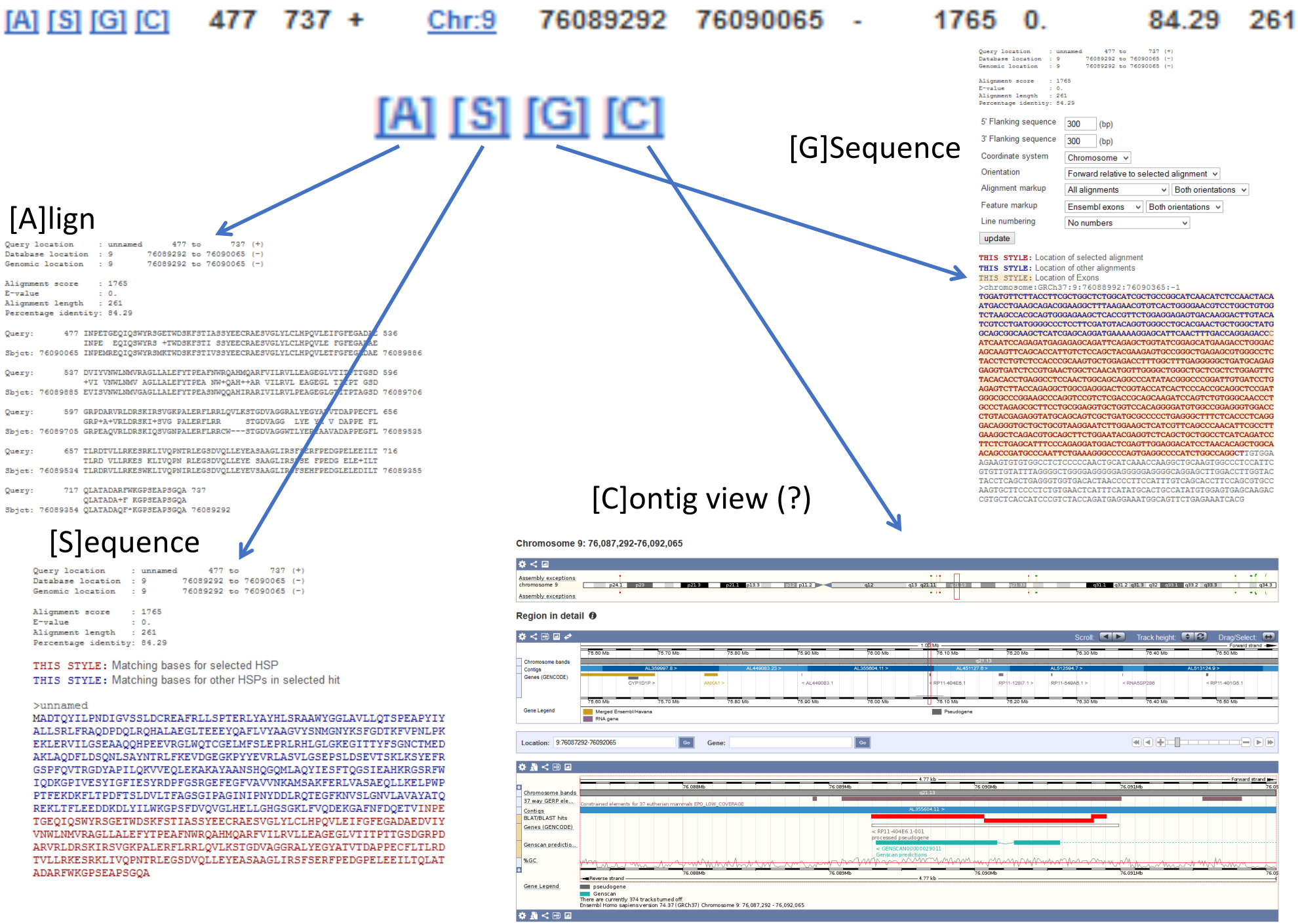
Alignment Summary (click arrow to hide)

Select rows to include in table, and type of sort
(Use the 'ctrl' key to select multiples)

refresh

Query	Subject	Chromosome	Supercontig	Clone	Contig	Lrg	Stats	Sort By
off	_off_	_off_	_off_	_off_	_off_	_off_	_off_	>Lrg
Name	Name	Name	Name	Name	Name	Name	Score	<Score
Start	Start	Start	Start	Start	Start	Start	E-val	>Score

Links	Query Start	Query End	Ori	Chromosome Name	Chromosome Start	Chromosome End	Ori	Stats Score	E-val	%ID	Length
[A] [S] [G] [C]	477	737	+	Chr:9	76089292	76090065	-	1765	0.	84.29	261
[A] [S] [G] [C]	224	475	+	Chr:9	76090067	76090816	-	1700	0.	84.13	252
[A] [S] [G] [C]	119	577	+	Chr:4	65296878	65298248	+	1542	5.5e-250	49.70	497
[A] [S] [G] [C]	581	729	+	Chr:4	65298493	65298930	+	854	5.5e-250	74.83	151
[A] [S] [G] [C]	1	90	+	Chr:11	66249672	66249941	+	763	3.2e-292	100.00	90
[A] [S] [G] [C]	330	399	+	Chr:11	66260186	66260395	+	552	3.2e-292	95.71	70
[A] [S] [G] [C]	565	679	+	Chr:11	66264763	66265104	+	531	3.2e-292	63.71	124
[A] [S] [G] [C]	1	90	+	Chr:4	65296627	65296899	+	529	5.5e-250	76.09	92
[A] [S] [G] [C]	588	721	+	Chr:11	66271972	66272364	+	487	1.7e-276	55.63	142
[A] [S] [G] [C]	681	737	+	Chr:11	66276549	66276719	+	477	3.2e-292	100.00	57
[A] [S] [G] [C]	120	166	+	Chr:11	66254008	66254148	+	391	1.8e-273	97.87	47
[A] [S] [G] [C]	420	526	+	Chr:11	66262674	66262961	+	384	3.2e-292	53.57	112
[A] [S] [G] [C]	486	597	+	Chr:11	66263006	66263296	+	377	1.7e-276	51.72	116
[A] [S] [G] [C]	266	309	+	Chr:11	66258962	66259093	+	375	3.2e-292	97.73	44
[A] [S] [G] [C]	209	266	+	Chr:11	66258657	66258854	+	370	3.2e-292	75.76	66
[A] [S] [G] [C]	384	432	+	Chr:11	66260513	66260650	+	310	5.1e-263	83.67	49
[A] [S] [G] [C]	90	126	+	Chr:11	66252641	66252751	+	272	3.2e-292	89.19	37
[A] [S] [G] [C]	432	463	+	Chr:11	66261009	66261104	+	270	1.7e-276	96.88	32
[A] [S] [G] [C]	192	242	+	Chr:11	66255385	66255576	+	268	1.3e-266	64.06	64
[A] [S] [G] [C]	196	230	+	Chr:9	76090801	76090905	-	257	0.	88.57	35
[A] [S] [G] [C]	129	191	+	Chr:11	66254628	66254813	+	248	3.2e-292	56.06	66


[A] [S] [G] [C] 477 737 + Chr:9 76089292 76090065 - 1765 0. 84.29 261
[A] [S] [G] [C]
[A]lign
Query location : unnamed 477 to 737 (+)
Database location : 9 76089292 to 76090065 (-)
Genomic location : 9 76089292 to 76090065 (-)
Alignment score : 1765
E-value : 0.
Alignment length : 261
Percentage identity: 84.29
[S]equence
Query location : unnamed 477 to 737 (+)
Database location : 9 76089292 to 76090065 (-)
Genomic location : 9 76089292 to 76090065 (-)
Alignment score : 1765
E-value : 0.
Alignment length : 261
Percentage identity: 84.29
THIS STYLE: Matching bases for selected HSP
THIS STYLE: Matching bases for other HSPs in selected hit
>unnamed
MADTQYILPNDIGVSSLDCREAFRLLSPTERLYAYHLSRAAWYGGLAVLLQTSPEAPYIY
ALLSRLFRAQDPDQLRQHALAEGLTEEEYQAFLVYAAGVYSNMGNYKSFGDTKFVPNLPK
EKLERVILGSEAAQQHPEEVRGLWQTCGELMFSLEPRLRHLGLGKEGITTYFSGNCTMED
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VNWLNMVRAGLLALEFYTPEAFNWRQAHMQARFVILRVLLEAGEGLVTITPTTGSDGRPD
ARVRLDRSKIRSVGKPALERFLRRLQVLKSTGDVAGGRALYEGYATVTDAPPECFLTLRD
TVLLRKESRKLIVQPNTRLEGSDVQLLEYEASAAGLIRSFSERFPEDGPELEEILTQLAT
ADARFWKGPSEAPSGQA
[G]Sequence
Query location : unnamed 477 to 737 (+)
Database location : 9 76089292 to 76090065 (-)
Genomic location : 9 76089292 to 76090065 (-)
Alignment score : 1765
E-value : 0.
Alignment length : 261
Percentage identity: 84.29
5' Flanking sequence 300 (bp)
3' Flanking sequence 300 (bp)
Coordinate system Chromosome
Orientation Forward relative to selected alignment
Alignment markup All alignments Both orientations
Feature markup Ensembl exons Both orientations
Line numbering No numbers
update
THIS STYLE: Location of selected alignment
THIS STYLE: Location of other alignments
THIS STYLE: Location of Exons
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TCTAAGCCACGCAGTGGGAGAAGCTCACCGTTCTGGAGGAGAGTGACAAGGACTTGTACA
TCGTCCTGATGGGGCCCTCCTTCGATGTACAGGTGGGCCTGCACGAACTGCTGGGCTATG
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AGCAAGTTCAGCACCATTGTCTCCAGCTACGAAGAGTGCCGGGCTGAGAGCGTGGGCCTC
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GCCCTAGAGCGCTTCCTGCGGAGGTGCTGGTCCACAGGGGATGTGGCCGGAGGGTGGACC
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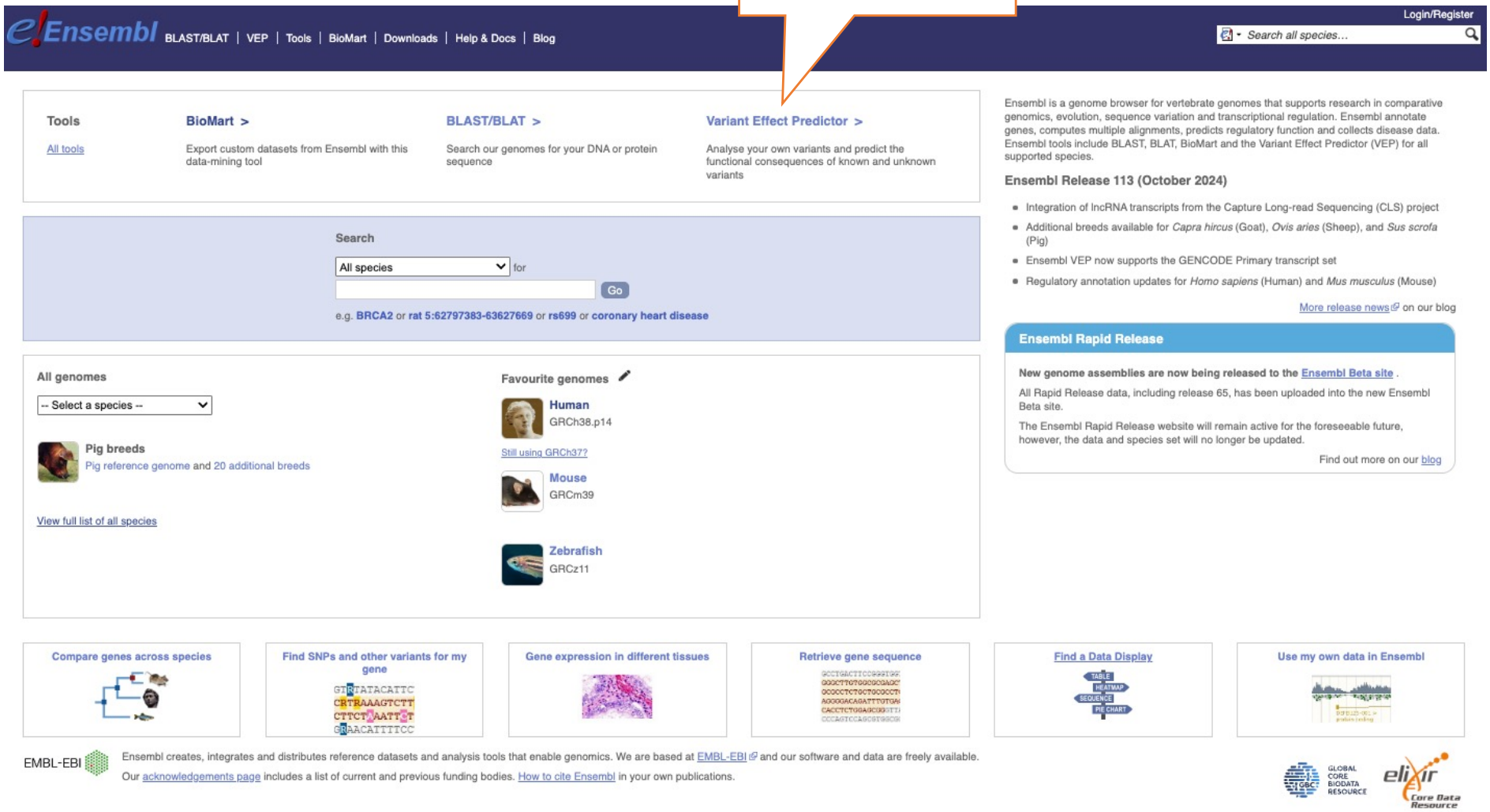
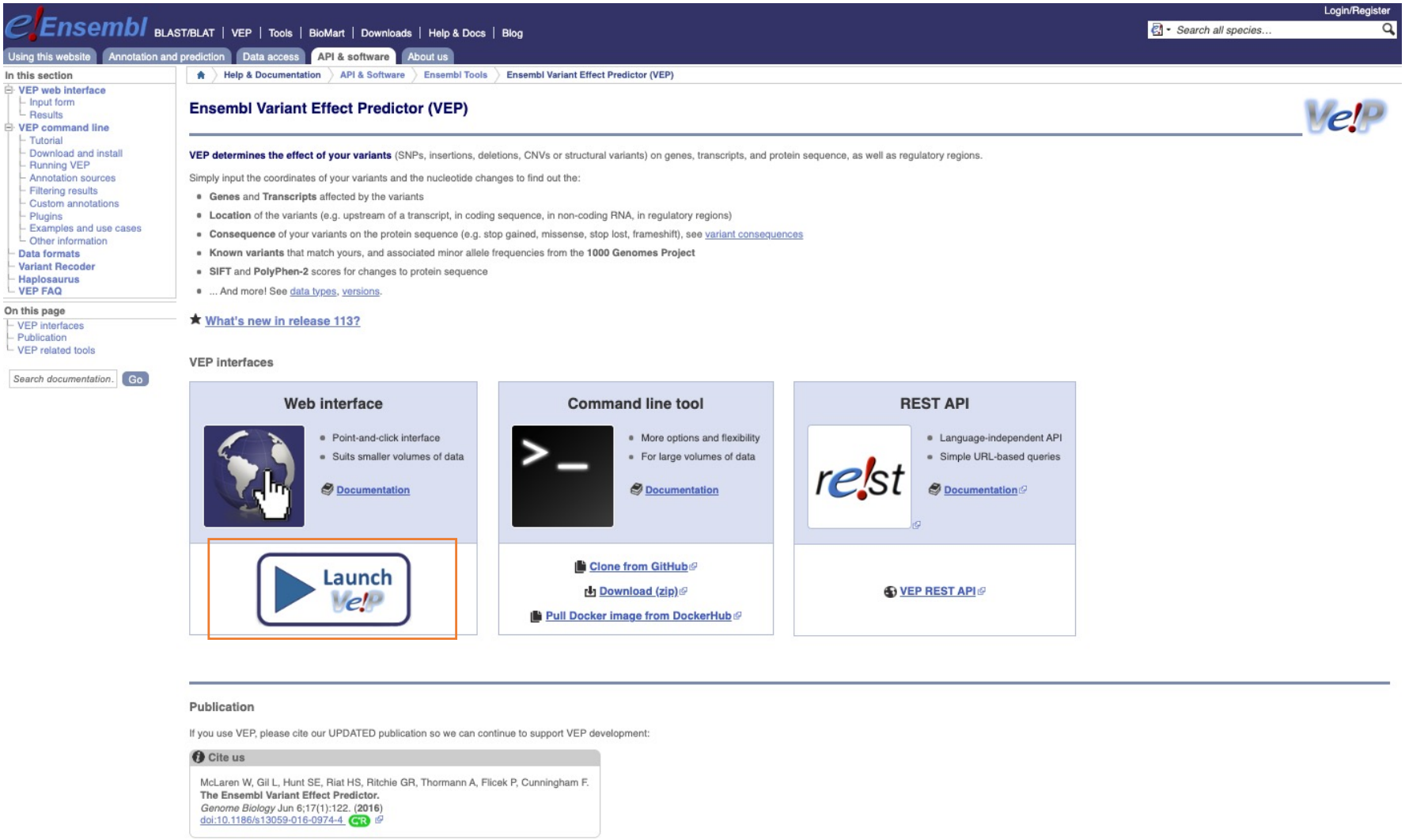
GTGTTGTATTTAGGGGCTGGGGAGGGGGAGGGGGAGGGGCAGGAGCTTGGACCTTGGTAC
TACCTCAGCTGAGGGTGGTGACACTAACCCCTTCCATTTGTCAGCACCTTCCAGCGTGCC
AAGTGCTTCCCCTCTGTGAACTCATTTCATATGCACTGCCATATGTGGAGTGAGCAAGAC
CGTGCTCACCATCCCGTCTACCAGATGAGGAAATGGCAGTTCTGAGAAATCACG
[C]ontig view (?)
Chromosome 9: 76,087,292-76,092,065
Region in detail
Location: 9:76087292-76092065
Gene:
Go
Gene Legend
Merged Ensembl/Havana
RNA gene
Pseudogene
Reverse strand
Forward strand
There are currently 374 tracks turned off.
Ensembl Homo sapiens version 74.37 (GRCh37) Chromosome 9: 76,087,292 - 76,092,065

Les outils

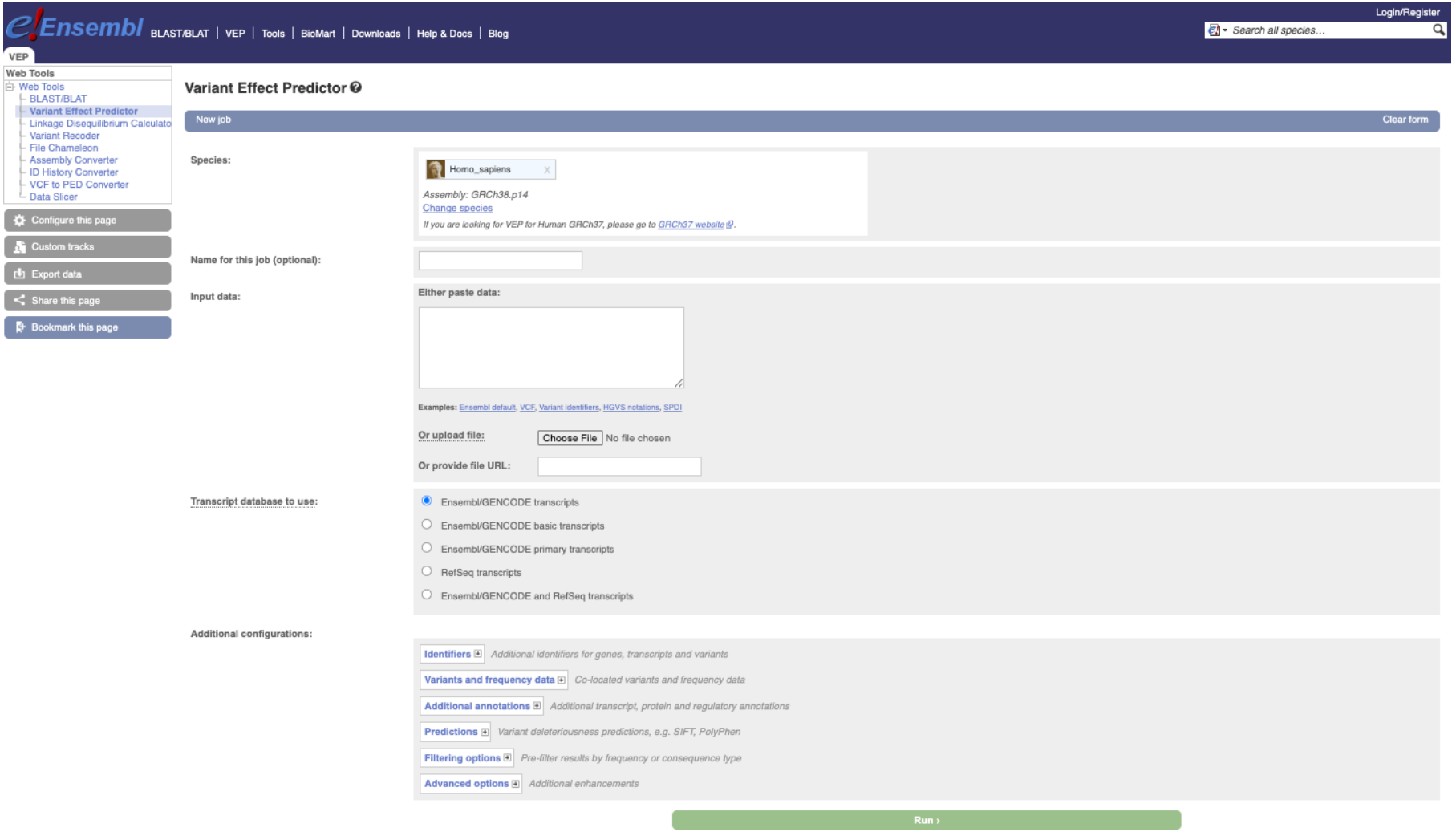
Annotation de variants



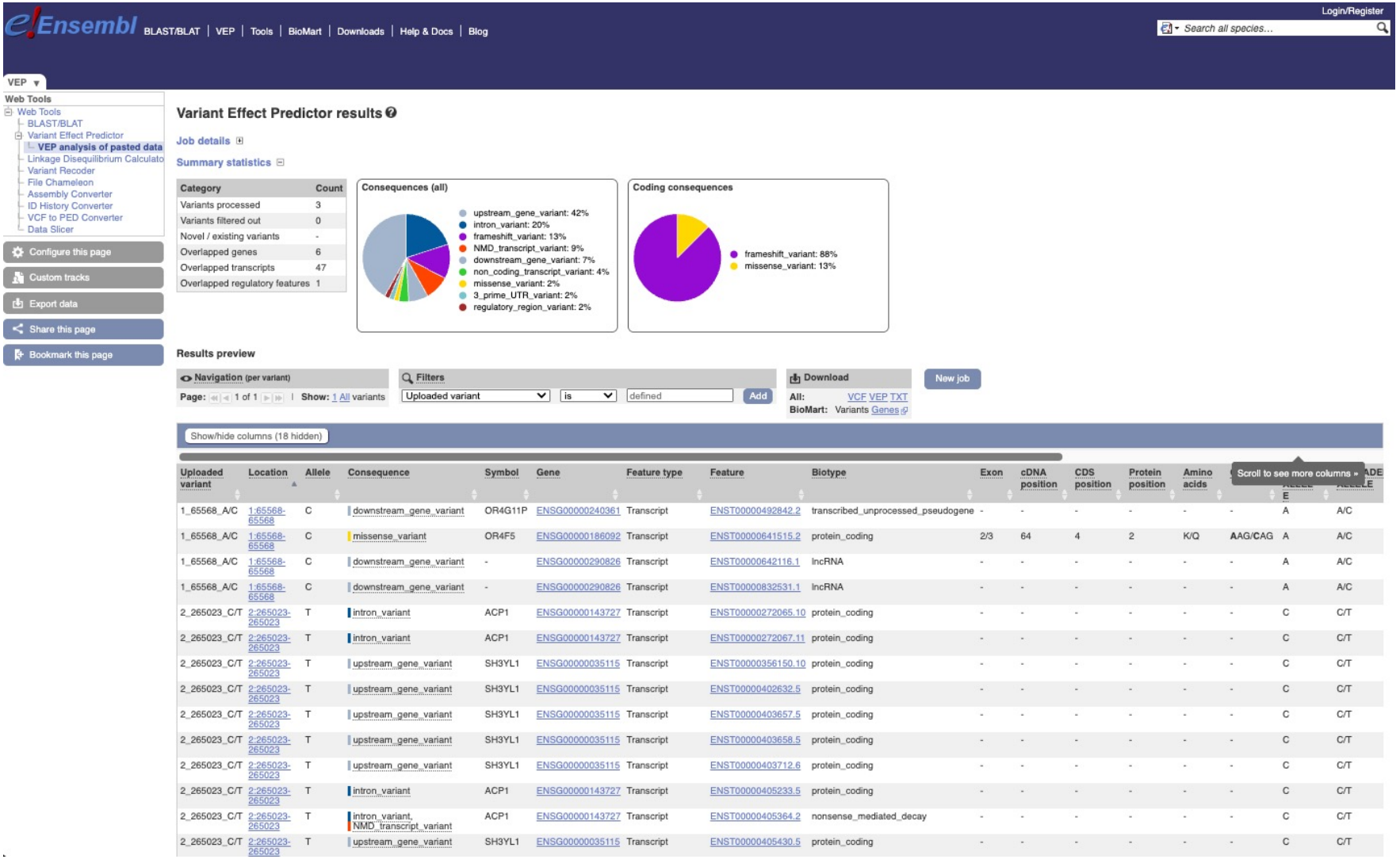
Variant Effect Predictor



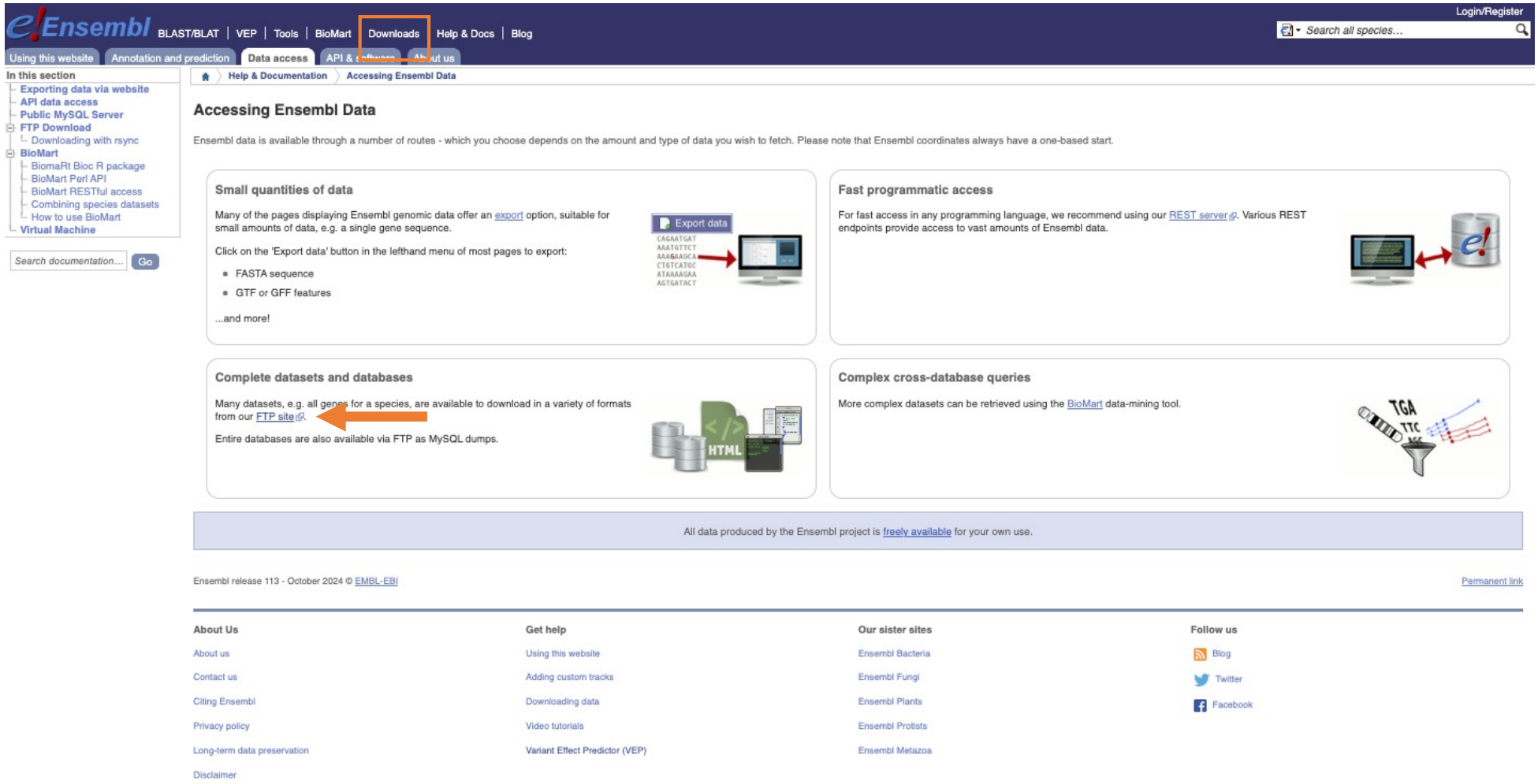
Variant Effect Predictor



Variant Effect Predictor



Outils de récupération de données



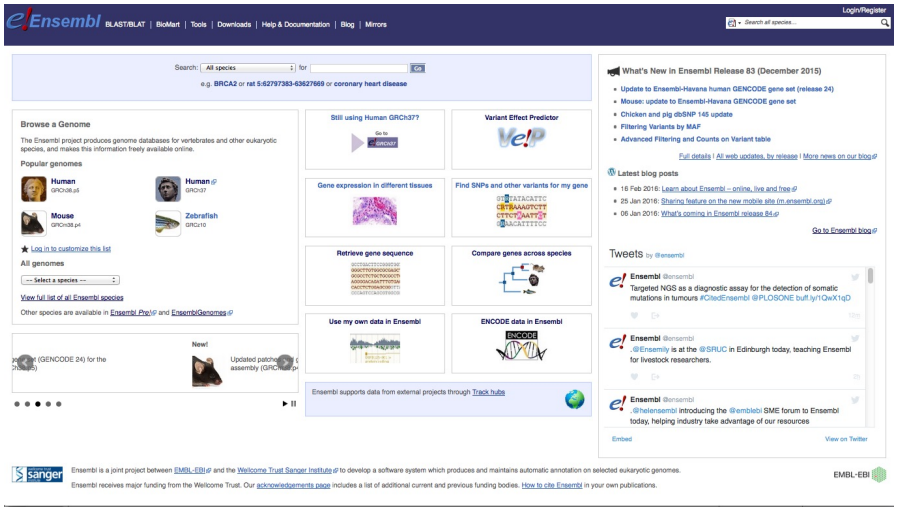
BioMart

Le projet BioMart

- Développé conjointement par :
  - EBI
  - Cold Spring Harbor Laboratory (CSHL)
- Arek Kasprzyk : « BioMart can access diverse databases from a single interface »
- Créer un système générique de stockage et de gestion de données
- « Data-agnostic » : manipulation de n'importe quel type de donnée avec le même software
- Applicable à
  - Tout type de données descriptives (y compris des données biologiques)
  - de grands volumes de données

Accéder aux données d'Ensembl

Site web

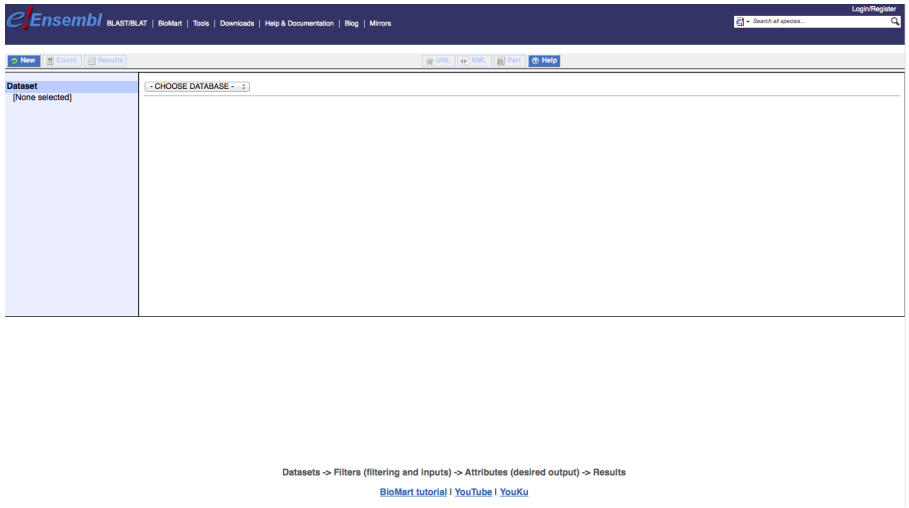



Simple d'utilisation


Facile à comprendre


Une seule requête à la fois

Outil de fouille: BioMart

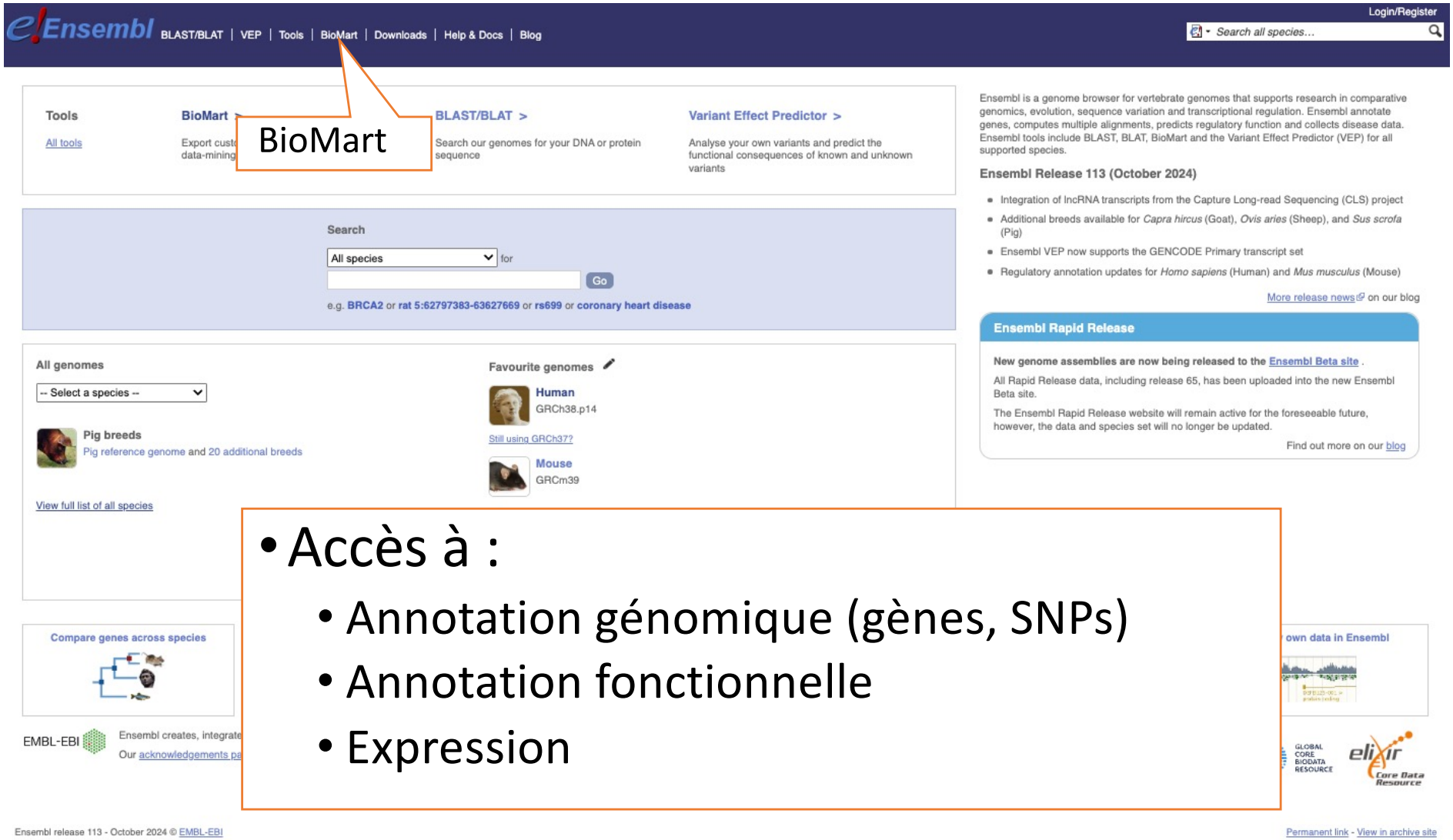



Requête complexe


Rapide

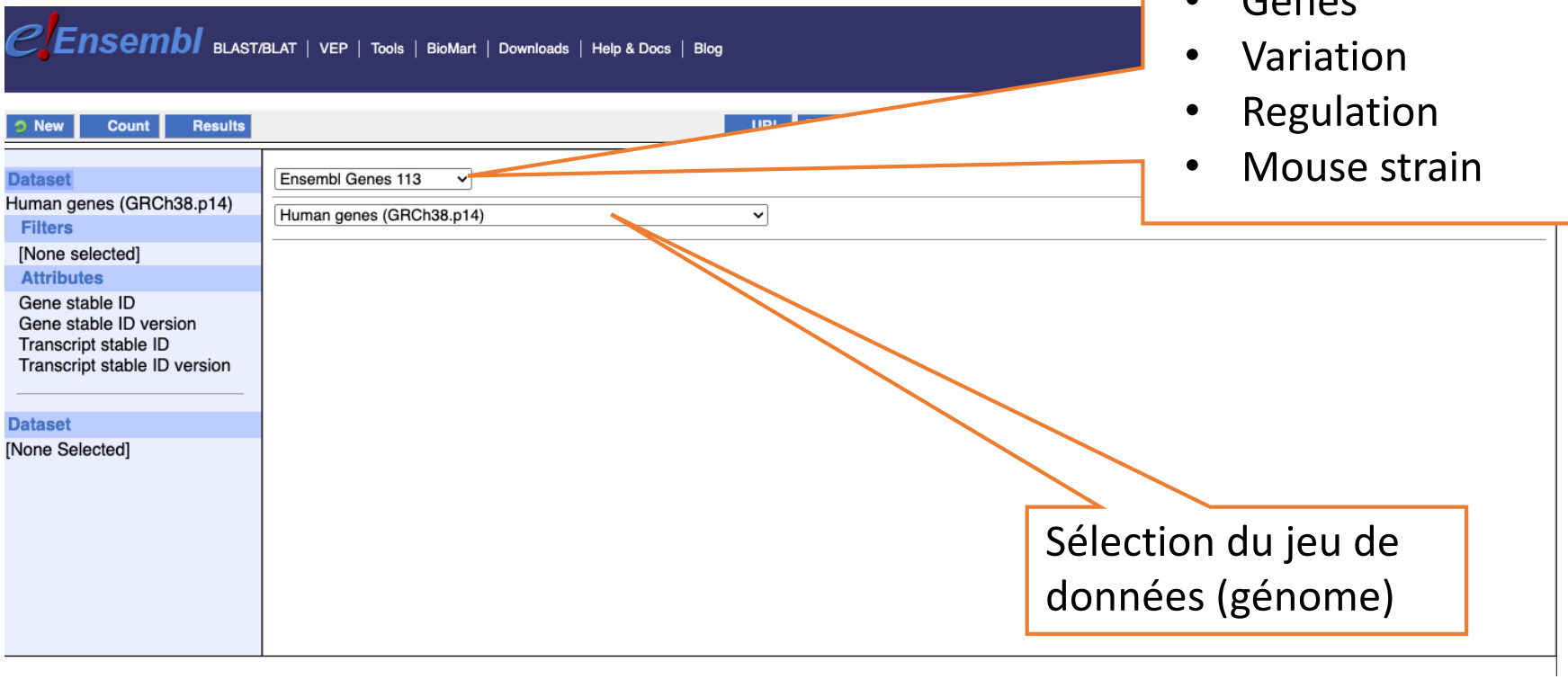

Requiert une formation

BioMart/Ensembl

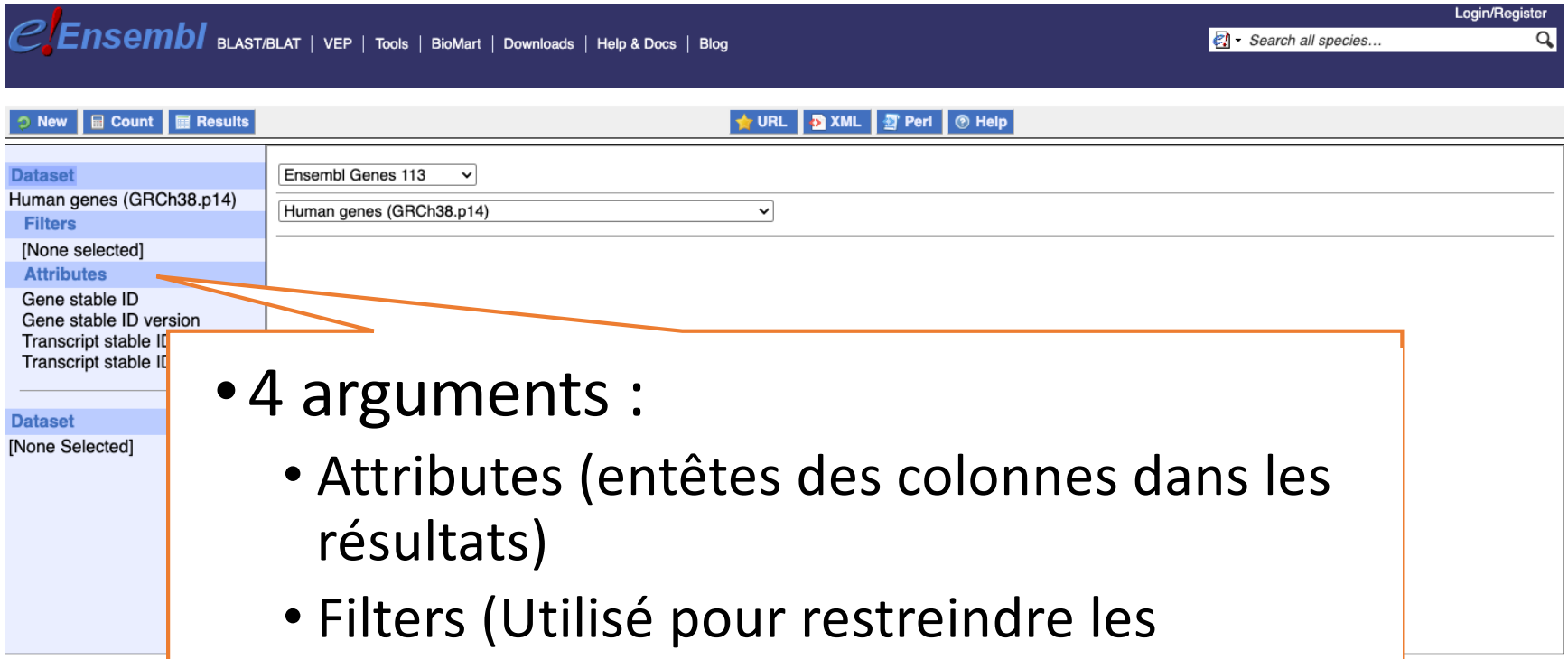


- Accès à :
  - Annotation génomique (gènes, SNPs)
  - Annotation fonctionnelle
  - Expression

BioMart/Ensembl



BioMart/Ensembl



- 4 arguments :
  - Attributes (entêtes des colonnes dans les résultats)
  - Filters (Utilisé pour restreindre les résultats)
  - Values (identifiants utilisés pour filtrer)
  - Dataset (selection des jeux de données)

Biomart :
Partie pratique

Comparaison des browsers

- Différences majeures entre Ensembl vs UCSC/NCBI
  - NCBI vs ensembl (UCSC?) – à l'origine de l'assemblage
  - Utilisation d'un pipeline automatique pour la création des jeux de données
  - Utilisation:
    - Visuel: ensembl/UCSC vs NCBI
    - Web: ensembl vs UCSC/NCBI
    - Rapidité/confort: UCSC vs ensembl/NBI
    - Organisation: ensembl/UCSC? Vs NCBI