INTRODUCTION TO BIOINFORMATICS

Please take the initial BIOINF525 questionnaire:

< http://tinyurl.com/bioinf525-questions >

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COURSE LOGISTICS

Lectures: Tuesdays 2:30-4:00 PM
Rm. 2062 Palmer Commons

Labs: Thursdays 2:30-4:00 PM
Rm. 2036 Palmer Commons

Website: http://tinyurl.com/bioinf525-w17
Lecture, lab and background reading material plus homework and course announcements

MODULE OVERVIEW

Objective: Provide an introduction to the practice of bioinformatics as well as a practical guide to using common bioinformatics databases and algorithms

1.1. ▸ *Introduction to Bioinformatics*

1.2. ▸ *Sequence Alignment and Database Searching*

1.3 ▸ *Structural Bioinformatics*

1.4 ▸ *Genome Informatics: High Throughput Sequencing Applications and Analytical Methods*

TODAYS MENU

Overview of bioinformatics

- The *what*, *why* and *how* of bioinformatics?
- Major bioinformatics research areas.
- Skepticism and common problems with bioinformatics.

Bioinformatics databases and associated tools

- Primary, secondary and composite databases.
  - Nucleotide sequence databases (GenBank & RefSeq).
  - Protein sequence database (UniProt).
  - Composite databases (PFAM & OMIM).

Database usage vignette

- Searching with ENTREZ and BLAST.
- Reference slides and handout on major databases.

HOMEWORK

- ☑ Complete the **initial course questionnaire**:
  http://tinyurl.com/bioinf525-questions

- ☑ Check out the "**Background Reading**" material online:
  PDF1 (bioinformatics review),
  PDF 2 (bioinformatics challenges).

- ☑ Complete the **lecture 1.1 homework questions**:
  http://tinyurl.com/bioinf525-quiz1

Q. What is Bioinformatics?

"Bioinformatics is the application of computers to the collection, archiving, organization, and analysis of biological data."

… Bioinformatics is a hybrid of biology and computer science

… **Bioinformatics is computer aided biology!**

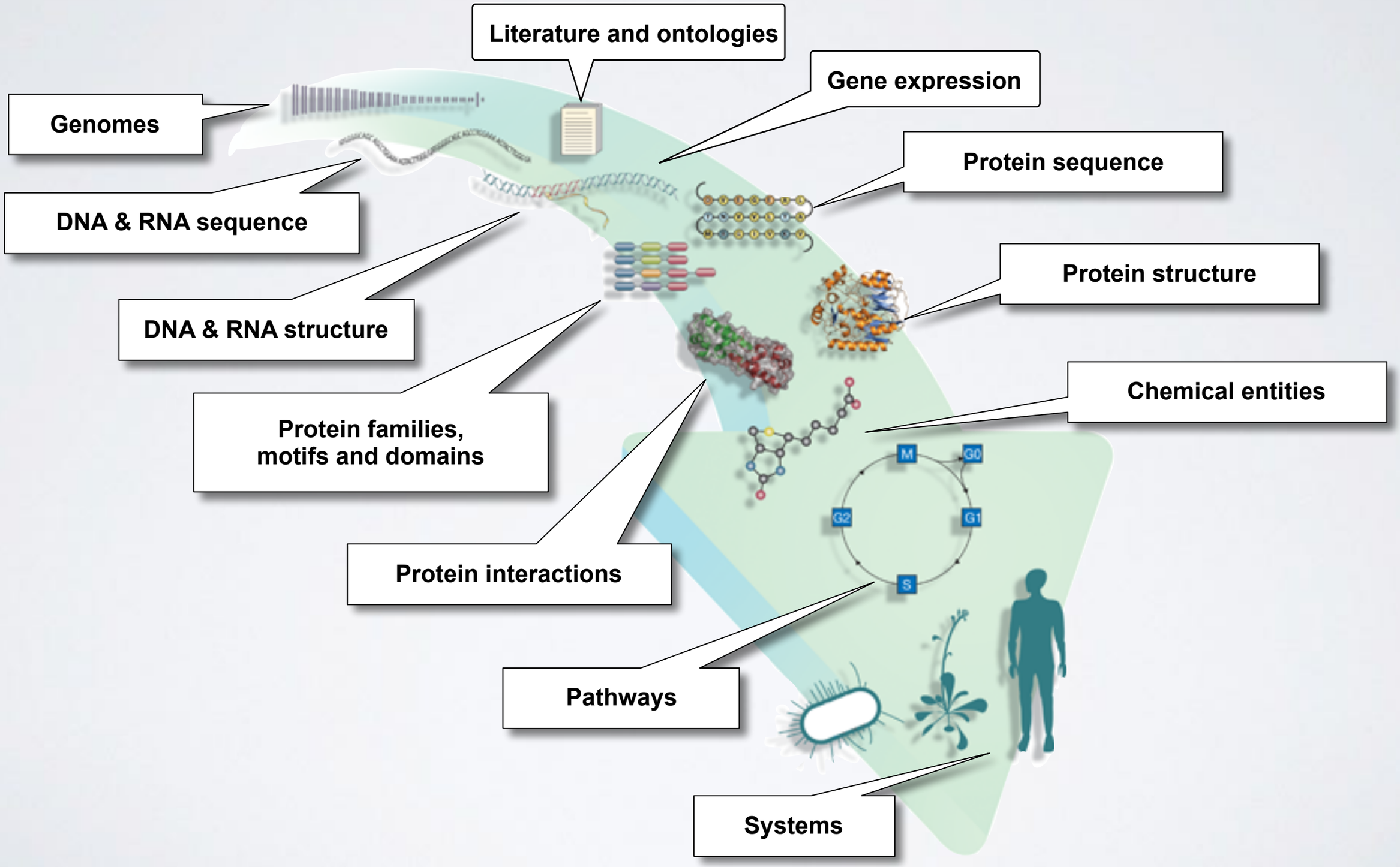
Computer based management and analysis of biological and biomedical data with useful applications in many disciplines, particularly genomics, proteomics, metabolomics, etc...

MORE DEFINITIONS

- “Bioinformatics is conceptualizing biology in terms of **macromolecules** and then applying **"informatics" techniques** (derived from disciplines such as applied maths, computer science, and statistics) to **understand** and **organize** the information associated with these molecules, on a **large-scale**.
  Luscombe NM, *et al*. Methods Inf Med. 2001;40:346.

- “Bioinformatics is research, development, or application of **computational approaches** for expanding the use of **biological**, **medical**, **behavioral** or **health data**, including those to **acquire**, **store**, **organize** and **analyze** such data.”
  National Institutes of Health (NIH) (http://tinyurl.com/l3gxr6b)

Major types of Bioinformatics Data



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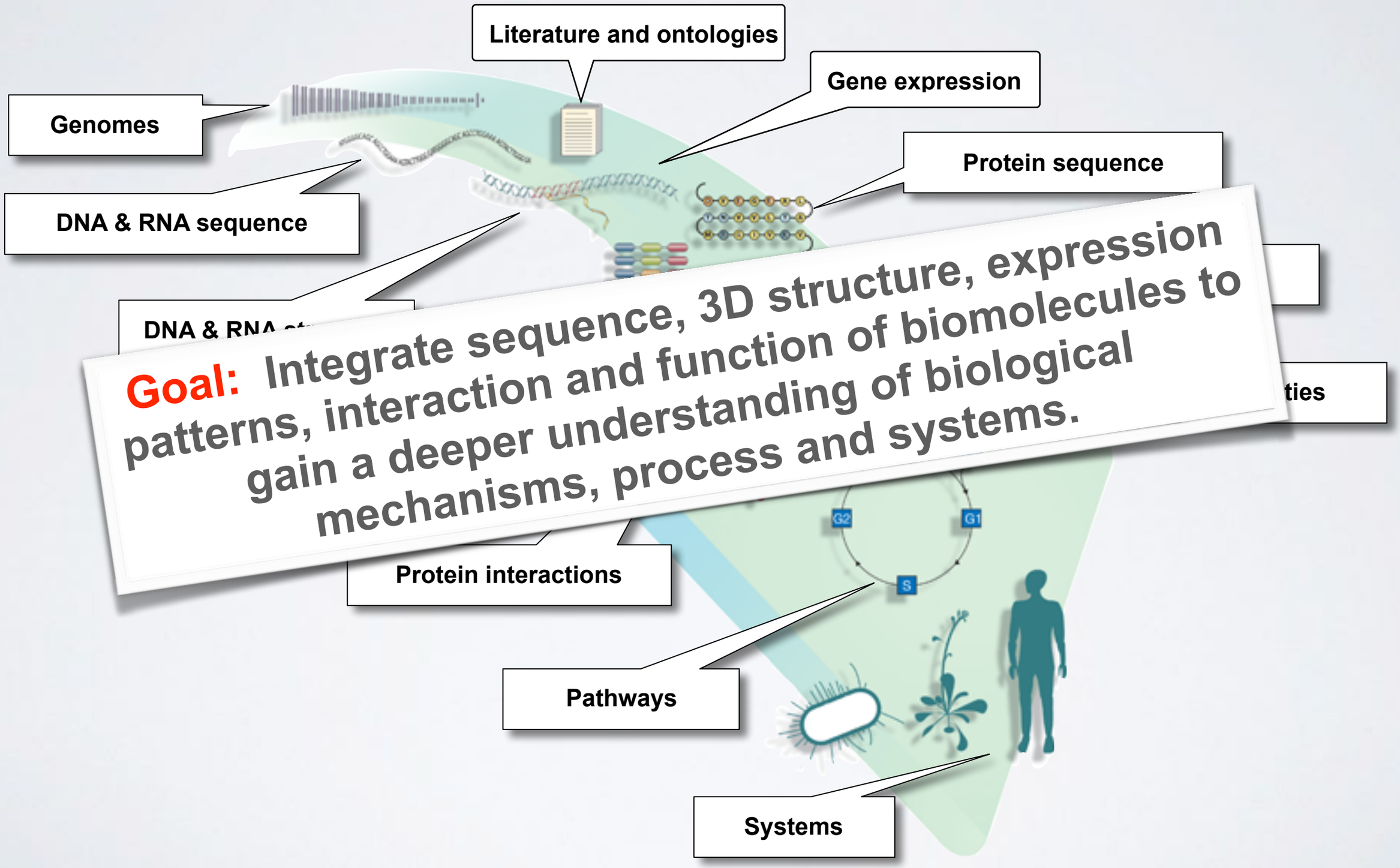
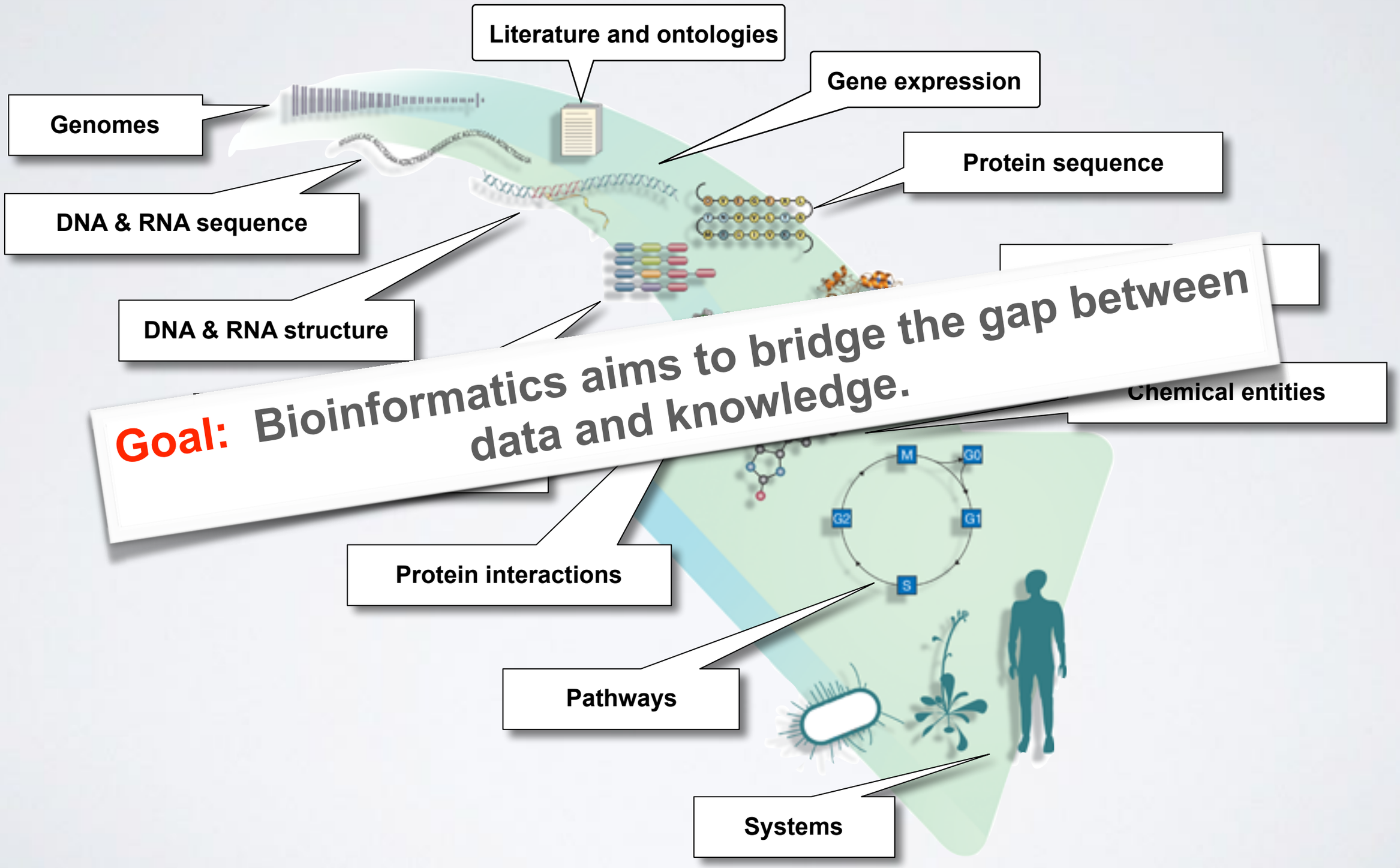


Goal: Integrate sequence, 3D structure, expression patterns, interaction and function of biomolecules to gain a deeper understanding of biological mechanisms, process and systems.

Major types of Bioinformatics Data



Goal: Bioinformatics aims to bridge the gap between data and knowledge.

BIOINFORMATICS RESEARCH AREAS

Include but are not limited to:

- Organization, classification, dissemination and analysis of biological and biomedical data (particularly '-omics' data).
- Biological sequence analysis and phylogenetics.
- Genome organization and evolution.
- Regulation of gene expression and epigenetics.
- Biological pathways and networks in healthy & disease states.
- Protein structure prediction from sequence.
- Modeling and prediction of the biophysical properties of biomolecules for binding prediction and drug design.
- Design of biomolecular structure and function.

With applications to Biology, Medicine, Agriculture and Industry

Where did bioinformatics come from?

Bioinformatics arose as molecular biology began to be transformed by the emergence of molecular sequence and structural data

Recap: The key dogmas of molecular biology

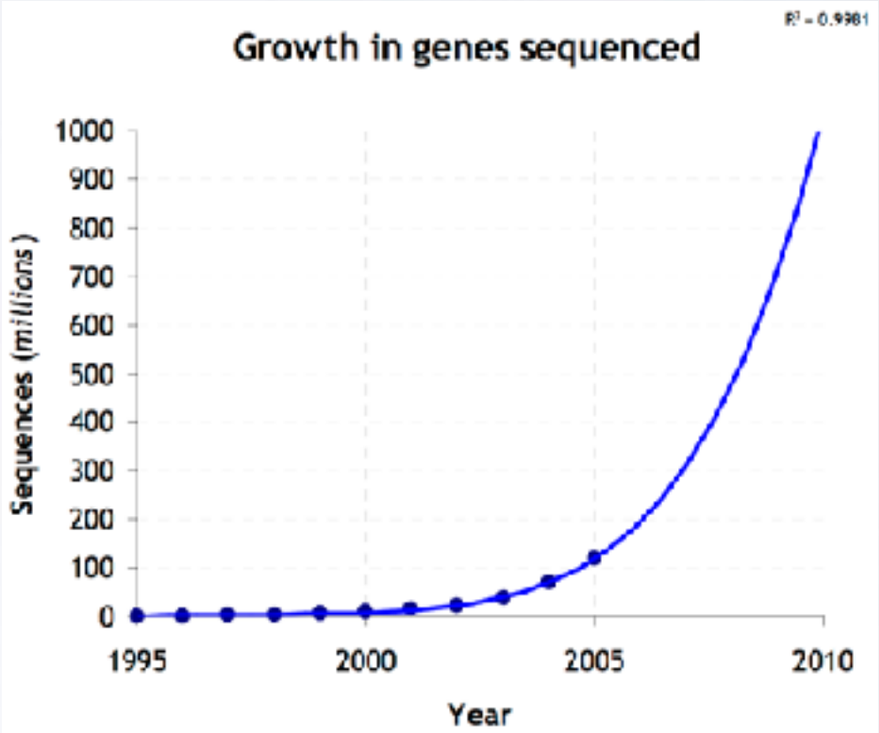
- *DNA sequence* determines *protein sequence.*
- *Protein sequence* determines *protein structure.*
- *Protein structure* determines *protein function.*
- *Regulatory mechanisms* (e.g. gene expression) determine the amount of a particular *function in space and time.*

Bioinformatics is *now* essential for the archiving, organization and analysis of data related to all these processes.

Why do we need Bioinformatics?

Bioinformatics is necessitated by the rapidly expanding quantities and complexity of biomolecular data

- Bioinformatics provides methods for the efficient:
  - **storage**
  - **annotation**
  - **search** and **retrieval**
  - data **integration**
  - data **mining** and **analysis**



E.G. data from sequencing, structural genomics, microarrays, proteomics, new high throughput assays, *etc…*

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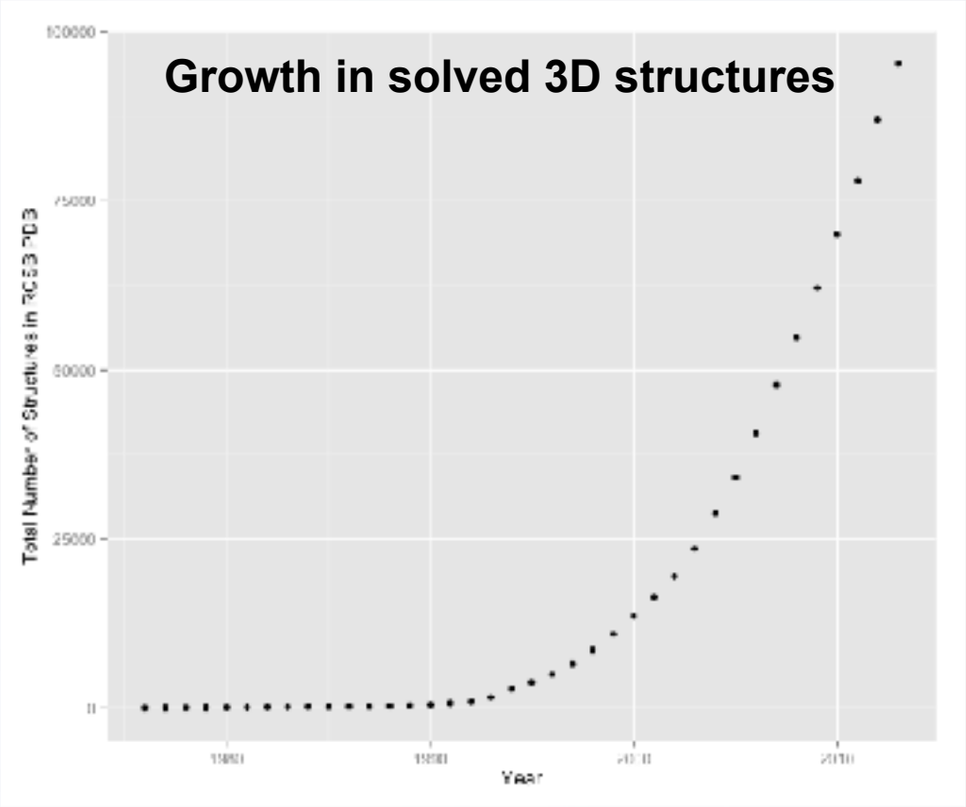


E.G. data from sequencing, structural genomics, microarrays, proteomics, new high throughput assays, *etc…*

How do we do Bioinformatics?

- A "*bioinformatics approach*" involves the application of **computer algorithms**, **computer models** and **computer databases** with the broad goal of understanding the action of both individual genes, transcripts, proteins and large collections of these entities.

DNA	→	RNA	→	Protein
Genome	→	**Transcriptome**	→	**Proteome**

How do we actually do Bioinformatics?

Pre-packaged tools and databases

- Many online
- New tools and time consuming methods frequently require downloading
- Most are free to use

Tool development

- Mostly on a UNIX environment
- Knowledge of programing languages frequently required (Python, **R**, Perl, C Java, Fortran)
- May require specialized or high performance computing resources…

Skepticism & Bioinformatics

We have to approach computational results the same way we do wet-lab results:

- Do they make sense?
- Is it what we expected?
- Do we have adequate controls, and how did they come out?
- Modeling is modeling, but biology is different...
  What does this model actually contribute?
- Avoid the miss-use of ‘black boxes’

Common problems with Bioinformatics

Confusing multitude of tools available

- Each with many options and settable parameters

Most tools and databases are written by and for nerds

- Same is true of documentation - if any exists!

Most are developed independently

Notable exceptions are found at the:

- **EBI** (European Bioinformatics Institute) and
- **NCBI** (National Center for Biotechnology Information)

Protein BLAST: search protein databases using a protein query

blast.ncbi.nlm.nih.gov/Blast.cgi?PROGRAM=blastp&BLAST_PROGRAMS=blastp&PAGE_TYPE=BlastSearch&SHOW_DEFAULTS=on&LINK_LOC=blasthome

General Parameters

Max target sequences 500

Select the maximum number of aligned sequences to display

Short queries ☑ Automatically adjust parameters for short input sequences

Expect threshold 10

Word size 3

Max matches in a query range 0

Scoring Parameters

Matrix BLOSUM62

Gap Costs Existence: 11 Extension: 1

Compositional adjustments Conditional compositional sco

Filters and Masking

Filter ☐ Low complexity regions

Mask ☐ Mask for lookup table only ☐ Mask lower case letters

PSI/PHI/DELTA BLAST

Upload PSSM Optional — Choose File no file selected

PSI-BLAST Threshold 0.005

Pseudocount 0

Even Blast has many settable parameters

STEP 3 - Set you

Related tools with different terminology

PROGRAM					
FASTA					
MATRIX	GAP OPEN	GAP EXTEND	KTUP	EXPECTATION UPPER VALUE	EXPECTATION LOWER VALUE
BLOSUM50	-10	-2	2	10	0 (default)
DNA STRAND	HISTOGRAM	FILTER		STATISTICAL ESTIMATES	
N/A	no	none		Regress	
SCORES	ALIGNMENTS	SEQUENCE RANGE	DATABASE RANGE	MULTI HSPs	
50	50	START-END	START-END	no	
SCORE FORMAT					
Default					

Key Online Bioinformatics Resources: NCBI & EBI

The NCBI and EBI are invaluable, publicly available resources for biomedical research

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

https://www.ebi.ac.uk

National Center for Biotechnology Information (NCBI)

- Created in 1988 as a part of the National Library of Medicine (NLM) at the National Institutes of Health
- NCBI's mission includes:
  - Establish **public databases**
  - Develop **software tools**
  - **Education** on and dissemination of biomedical information

- We will cover a number of core NCBI databases and software tools in the lecture

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

National Center for Biotechnology Information

www.ncbi.nlm.nih.gov

NCBI Resources How To Sign in to NCBI

NCBI
National Center for Biotechnology Information

All Databases Search

- NCBI Home
- Resource List (A-Z)
- All Resources
- Chemicals & Bioassays
- Data & Software
- DNA & RNA
- Domains & Structures
- Genes & Expression
- Genetics & Medicine
- Genomes & Maps
- Homology
- Literature
- Proteins
- Sequence Analysis
- Taxonomy
- Training & Tutorials
- Variation

Welcome to NCBI

The National Center for Biotechnology Information advances science and health by providing access to biomedical and genomic information.

About the NCBI | Mission | Organization | Research | RSS Feeds

Get Started

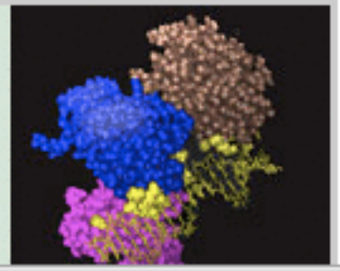
- Tools: Analyze data using NCBI software
- Downloads: Get NCBI data or software
- How-To's: Learn how to accomplish specific tasks at NCBI
- Submissions: Submit data to GenBank or other NCBI databases

Popular Resources

PubMed
Bookshelf
PubMed Central
PubMed Health
BLAST
Nucleotide
Genome
SNP
Gene
Protein
PubChem

3D Structures

Explore three-dimensional structures of proteins, DNA, and RNA molecules. Examine sequence-structure relationships, active sites, molecular interactions, biological activities of bound chemicals, and associated biosystems



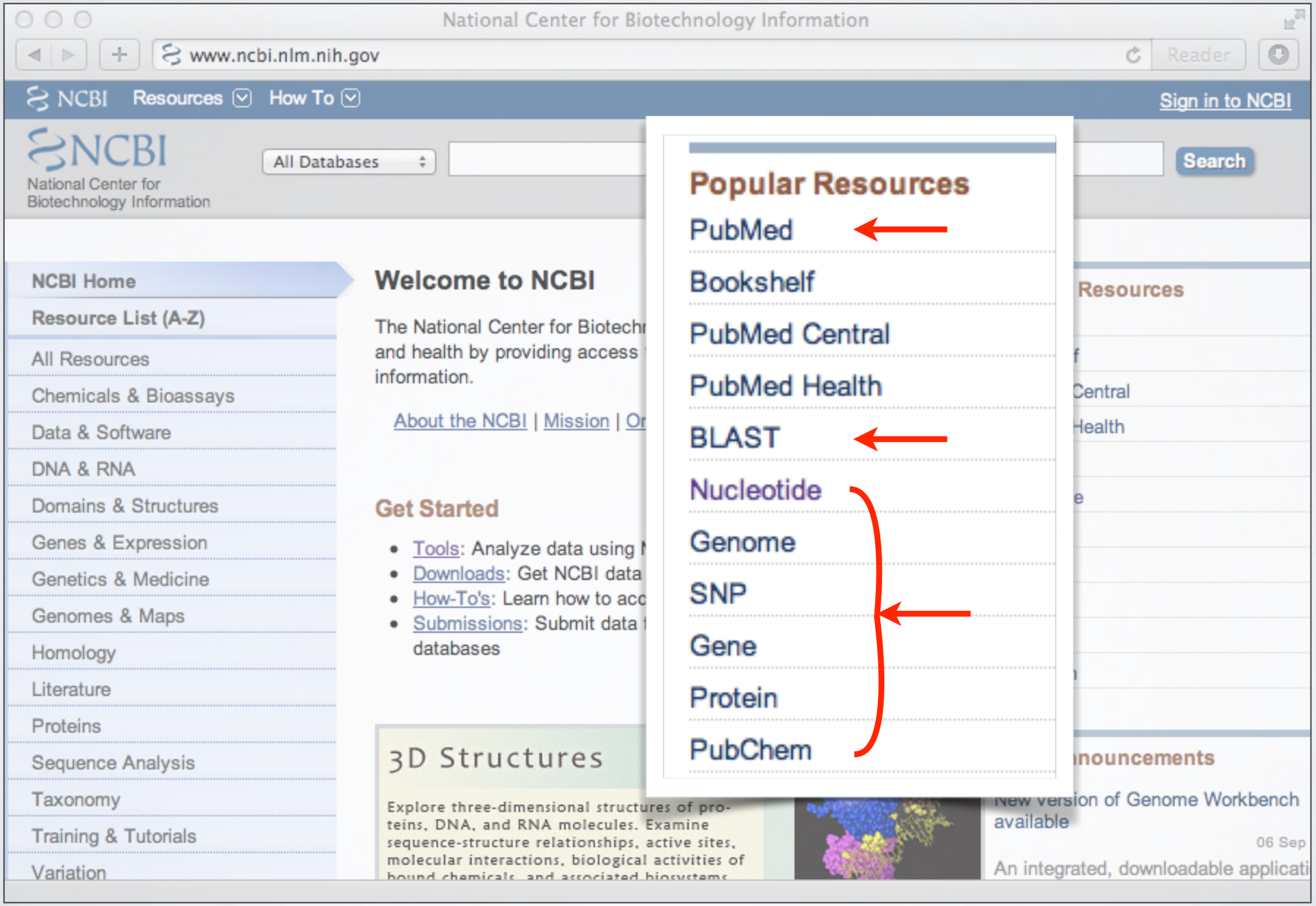
NCBI Announcements

New version of Genome Workbench available

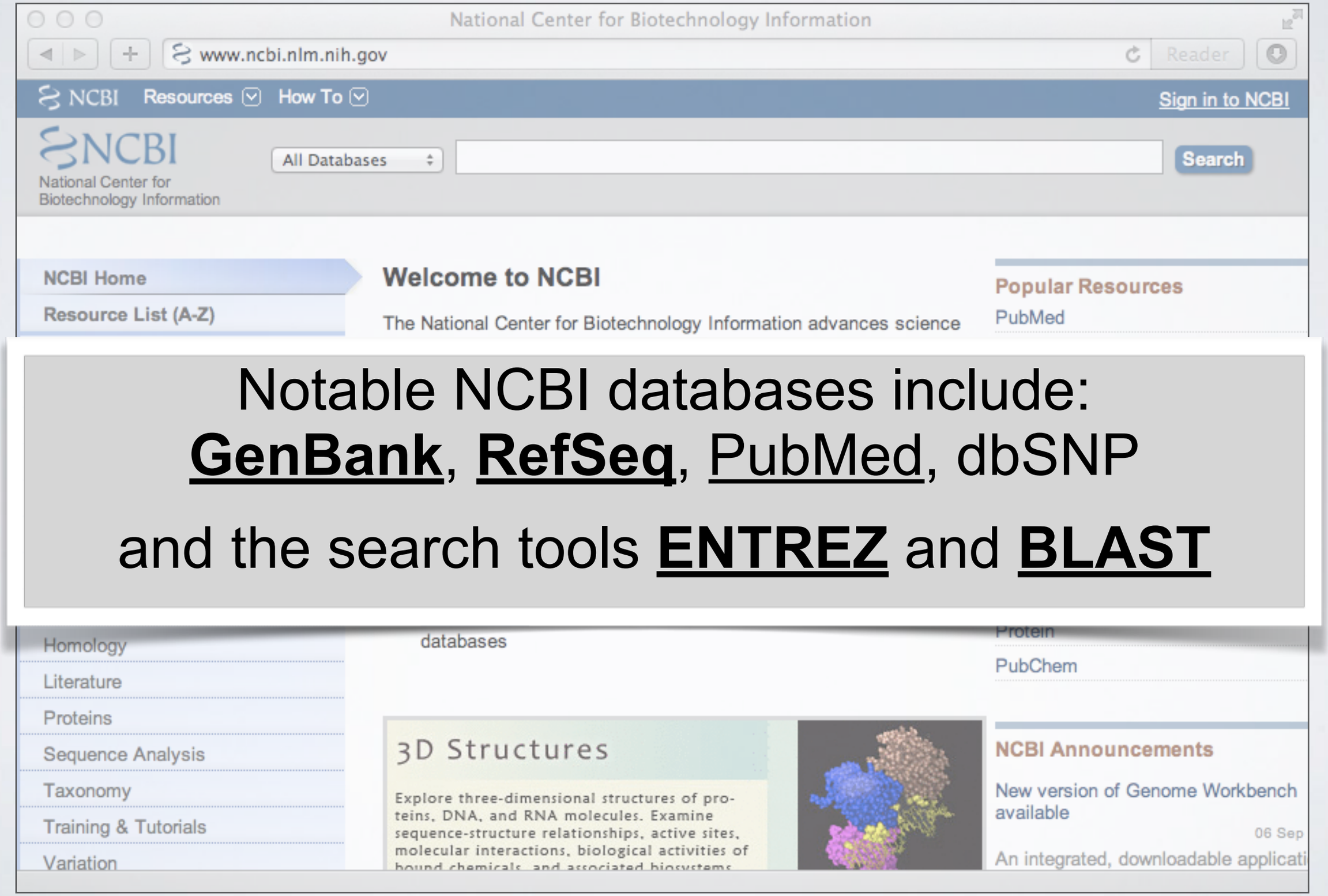
06 Sep

An integrated, downloadable applicati

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



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



Notable NCBI databases include:
GenBank, **RefSeq**, PubMed, dbSNP

and the search tools **ENTREZ** and **BLAST**

Key Online Bioinformatics Resources: NCBI & EBI

The NCBI and EBI are invaluable, publicly available resources for biomedical research

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

https://www.ebi.ac.uk

European Bioinformatics Institute (EBI)

- Created in 1997 as a part of the European Molecular Biology Laboratory (EMBL)
- EBI's mission includes:
  - providing freely available **data** and **bioinformatics services**
  - and providing advanced **bioinformatics training**

- We will briefly cover several EBI databases and tools that have advantages over those offered at NCBI

The EBI maintains a number of high quality curated **secondary databases** and associated tools

EMBL European Bioinforma...

www.ebi.ac.uk

EMBL-EBI

Services | Research | Training | About us

The European Bioinformatics Institute

Part of the European Molecular Biology Laboratory

EMBL-EBI provides freely available data from life science experiments, performs basic research in computational biology and offers an extensive user training programme, supporting researchers in academia and industry.

Find a gene, protein or chemical:

Search

Examples: blast, keratin, bfl1...

Services | Research | Training | Industry | European Coordination | EMBL ALUMNI

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- Research
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- Jobs
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EMBL 40

Upcoming events

Plant and Animal Genome conference (PAG XXIV)

Sunday 10 - Tuesday 12 January 2016

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Services < EMBL-EBI

www.ebi.ac.uk/services

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Services

Overview | A to Z | Data submission | Support

Bioinformatics services

We maintain the world's most comprehensive range of **freely available** and up-to-date molecular databases. Developed in collaboration with our colleagues worldwide, our services let you share data, perform complex queries and analyse the results in different ways. You can work locally by downloading our data and software, or use our web services to access our resources programmatically. You can read more about our services in the journal *Nucleic Acids Research*.

DNA & RNA
genes, genomes & variation

Gene expression
RNA, protein & metabolite expression

Proteins
sequences, families & motifs

Structures
Molecular & cellular structures

Systems
reactions, interactions & pathways

Chemical biology
chemogenomics & metabolomics

Ontologies
taxonomies & controlled vocabularies

Literature
Scientific publications & patents

Cross domain
cross-domain tools & resources

Popular

- Ensembl
- BLAST
- UniProt
- Europe PMC
- PDBe
- Reactome
- ArrayExpress
- Train online
- ChEMBL
- Support

Service news

Programmatic access

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Bioinformatics services

We maintain the world's most comprehensive range of **freely available** and up-to-date molecular databases. Developed in collaboration with our colleagues worldwide, our services let you share data, perform complex queries and analyse the results in different ways. You can work locally by downloading our data and software, or use our web services to access our resources programmatically. You can read more about our services in the journal *Nucleic Acids Research*.

DNA & RNA
genes, genomes & variation

Gene expression
RNA, protein & metabolite expression

Proteins
sequences, families & motifs

Structures
Molecular & cellular structures

Systems
reactions, interactions & pathways

Chemical biology
chemogenomics & metabolomics

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Literature
Scientific publications & patents

Cross domain
cross-domain tools & resources

Popular

- Ensembl
- UniProt
- PDBe
- ArrayExpress
- ChEMBL

Programmatic access

https://www.ebi.ac.uk

The EBI makes available a wider variety of **online tools** than NCBI

Proteins

Popular services

UniProt: The Universal Protein Resource

The gold-standard, comprehensive resource for protein sequence and functional annotation data.

InterPro

A database for the classification of proteins into families, domains and conserved sites.

PRIDE: The Proteomics Identifications Database

An archive of protein expression data determined by mass spectrometry.

Pfam

A database of hidden Markov models and alignments to describe conserved protein families and domains.

Clustal Omega

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

HMMER - protein homology search

Fast sensitive protein homology searches using profile hidden Markov models (HMMs). Variety of different search methods for querying against both sequence and HMM target databases.

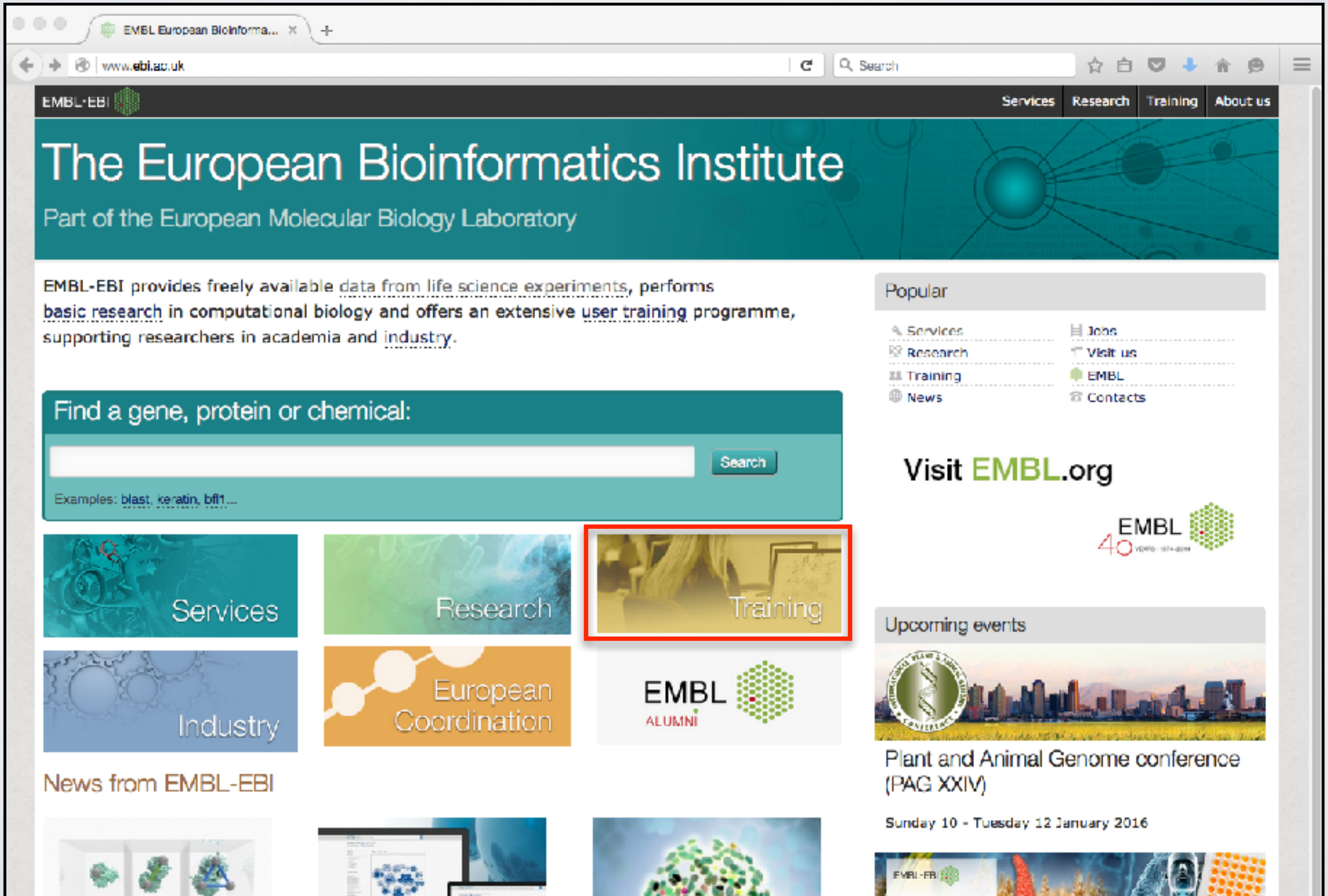
InterProScan 5

InterProScan 5 searches sequences against InterPro's predictive protein signatures. Please note that InterProScan 4.8 has been retired.

Quick links

- Popular services in this category
- All services in this category
- Project websites in this category

The EBI also provides a growing selection of **online tutorials** on EBI databases and tools



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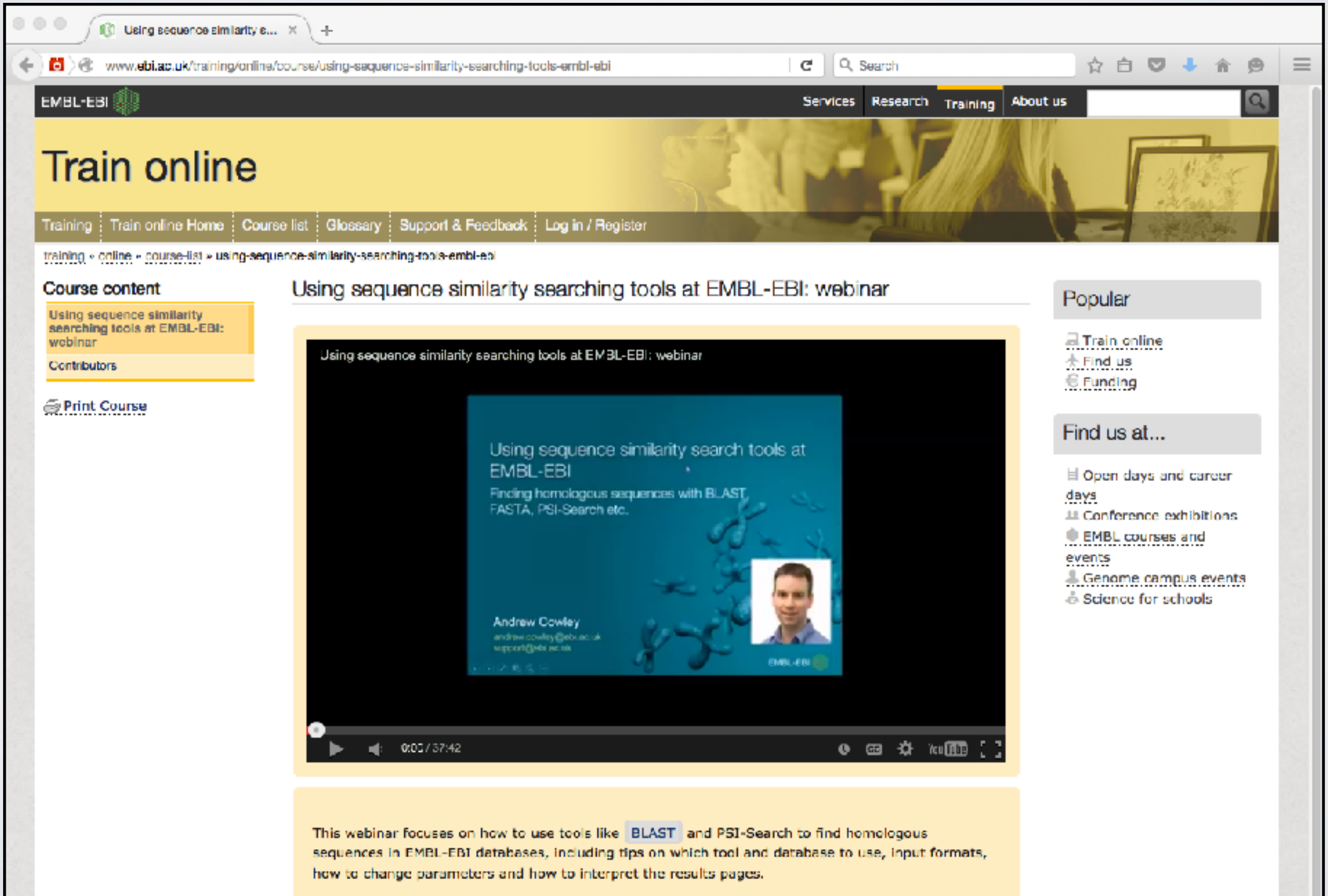

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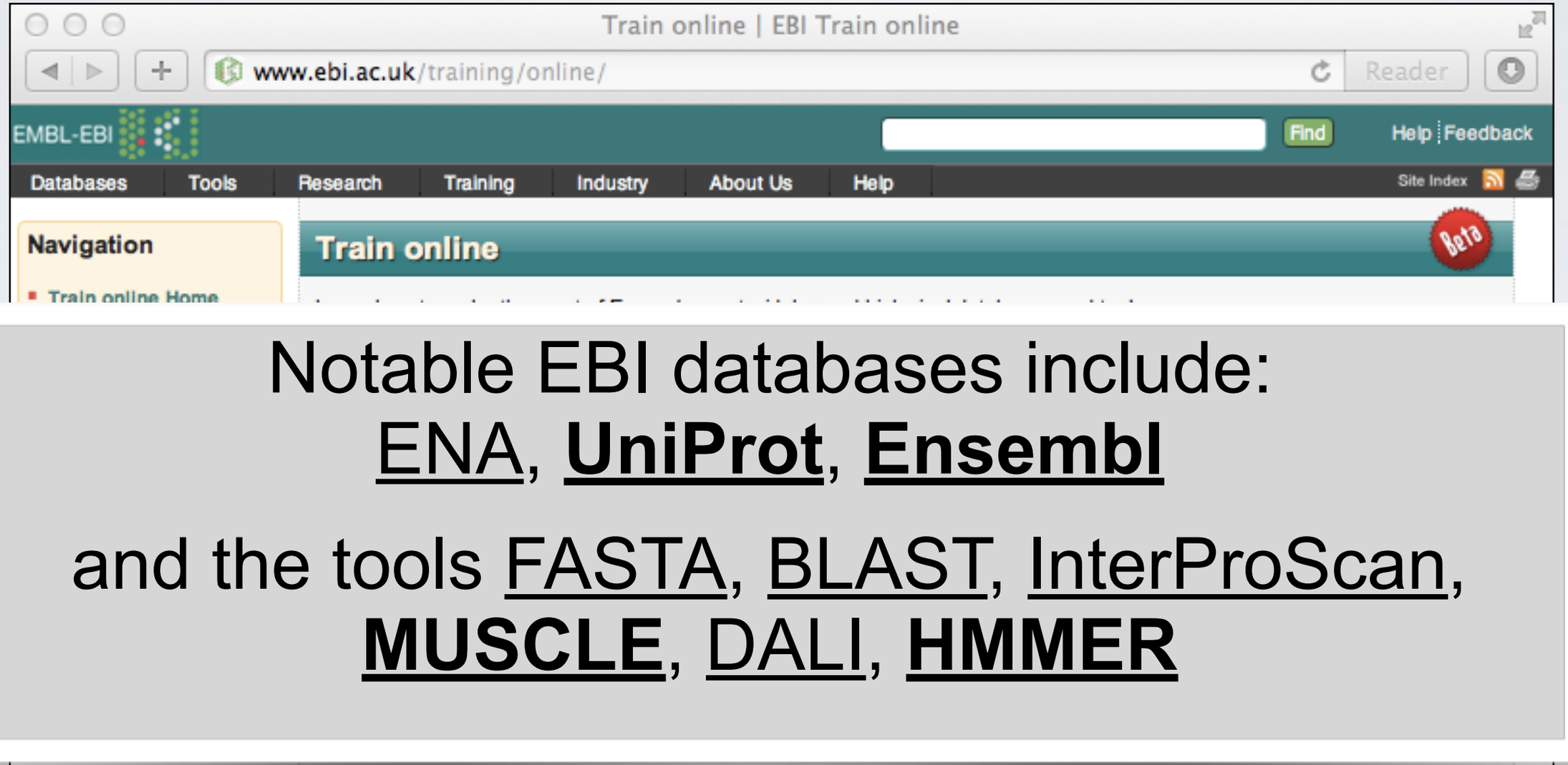

Notable EBI databases include:
ENA, **UniProt**, **Ensembl**

and the tools FASTA, BLAST, InterProScan, **MUSCLE**, DALI, **HMMER**

BIOINFORMATICS DATABASES AND ASSOCIATED TOOLS

What is a database?

Computerized store of data that is organized to provide efficient retrieval.

- Uses standardized data (record) formats to enable computer handling

Key database features allow for:

- Adding, changing, removing and merging of records
- User-defined queries and extraction of specified records

Desirable features include:

- Contains the data you are interested in
- Allows fast data access
- Provides annotation and curation of entries
- Provides links to additional information (possibly in other databases)
- Allows you to make discoveries

Bioinformatics Databases

AATDB, AceDb, ACUTS, ADB, AFDB, AGIS, AMSdb, ARR, AsDb,BBDB, BCGD, Beanref, BioImage, BioMagResBank, BIOMDB, BLOCKS, BovGBASE, BOVMAP, BSORF, BTKbase, CANSITE, CarbBank, CARBHYD, CATH, CAZY, CCDC, CD4OLbase, CGAP, ChickGBASE, Colibri, COPE, CottonDB, CSNDB, CUTG, CyanoBase, dbCFC, dbEST, dbSTS, DDBJ, DGP, DictyDb, Picty_cDB, DIP, DOGS, DOMO, DPD, DPInteract, ECDC, ECGC, EC02DBASE, EcoCyc, EcoGene, EMBL, EMD db, ENZYME, EPD, EpoDB, ESTHER, FlyBase, FlyView, GCRDB, GDB, GENATLAS, Genbank, GeneCards, Genlilesne, GenLink, GENOTK, GenProtEC, GIFTS, GPCRDB, GRAP, GRBase, gRNAsdb, GRR, GSDB, HAEMB, HAMSTERS, HEART-2DPAGE, HEXAdb, HGMD, HIDB, HIDC, HIVdb, HotMolecBase, HOVERGEN, HPDB, HSC-2DPAGE, ICN, ICTVDB, IL2RGbase, IMGT, Kabat, KDNA, KEGG, Klotho, LGIC, MAD, MaizeDb, MDB, Medline, Mendel, MEROPS, MGDB, MGI, MHCPEP5 Micado, MitoDat, MITOMAP, MJDB, MmtDB, Mol-R-Us, MPDB, MRR, MutBase, MycDB, NDB, NRSub, 0-lycBase, OMIA, OMIM, OPD, ORDB, OWL, PAHdb, PatBase, PDB, PDD, Pfam, PhosphoBase, PigBASE, PIR, PKR, PMD, PPDB, PRESAGE, PRINTS, ProDom, Prolysis, PROSITE, PROTOMAP, RatMAP, RDP, REBASE, RGP, SBASE, SCOP, SeqAnaiRef, SGD, SGP, SheepMap, Soybase, SPAD, SRNA db, SRPDB, STACK, StyGene,Sub2D, SubtiList, SWISS-2DPAGE, SWISS-3DIMAGE, SWISS- MODEL Repository, SWISS-PROT, TeIDB, TGN, tmRDB, TOPS, TRANSFAC, TRR, UniGene, URNADB, V BASE, VDRR, VectorDB, WDCM, WIT, WormPep, etc ..... !!!!

Bioinformatics Databases

AATDB, AceDb, ACUTS, ADB, AFDB, AGIS, AMSdb, ARR, AsDb,BBDB, BCGD, Beanref, BioImage, BioMagResBank, BIOMDB, BLOCKS, BovGBASE, BOVMAP, CATKbase, CANSITE, CarbBank, CARBHYD, CATH, CAZY, CMAP, ChickGBASE, Colibri, COPE, CottonDB, CSDB, dbEST, dbSTS, DDBJ, DGP, DictyDb, Bi[illegible] CDC, ECGC, EC02DBASE, E[illegible] THER, FlyBase, Fl[illegible] Link, G[illegible] DB, HAEMB, H[illegible] HIVdb, HotMolecBase, H[illegible] IEZRGbase, IMGT, Kabat, KDNA, K[illegible] DB, Medline, Mendel, MEROPS, MGDB, MGI, MHC[illegible] TOMAP, MJDB, MmtDB, Mol-R-Us, MPDB, MRR, MutBase, Myc[illegible] db, 0-lycBase, OMIA, OMIM, OPD, ORDB, OWL, PAHdb, PatBase, PD[illegible] DD, Pfam, PhosphoBase, PigBASE, PIR, PKR, PMD, PPDB, PRESAGE, PRINTS, ProDom, Prolysis, PROSITE, PROTOMAP, RatMAP, RDP, REBASE, RGP, SBASE, SCOP, SeqAnaiRef, SGD, SGP, SheepMap, Soybase, SPAD, SRNA db, SRPDB, STACK, StyGene,Sub2D, SubtiList, SWISS-2DPAGE, SWISS-3DIMAGE, SWISS- MODEL Repository, SWISS-PROT, TelDB, TGN, tmRDB, TOPS, TRANSFAC, TRR, UniGene, URNADB, V BASE, VDRR, VectorDB, WDCM, WIT, WormPep, etc ..... !!!!

There are lots of Bioinformatics Databases

For a annotated listing of major bioinformatics databases please see the online handout

< Handout_Major_Databases.pdf >

Side-note: Databases come in all shapes and sizes





Databases can be of variable quality and often there are multiple databases with overlapping content.

Finding Bioinformatics Databases

Oxford Journals | Life Sciences | Nucleic Acids Research | Database Summary Paper Categories

www.oxfordjournals.org/nar/database/cat/8

Oxford Journals | Life Sciences | Nucleic Acids Research | Database Summary Pa... | The 2014 Nucleic Acids Research Database Issue and an updated NAR online M...

OXFORD JOURNALS — CONTACT US — MY BASKET — MY ACCOUNT

Nucleic Acids Research

ABOUT THIS JOURNAL — CONTACT THIS JOURNAL — SUBSCRIPTIONS — CURRENT ISSUE — ARCHIVE — SEARCH

Oxford Journals > Life Sciences > Nucleic Acids Research > Database Summary Paper Categories

2014 NAR Database Summary Paper Category List

- Compilation Paper
- Category List
- Alphabetical List
- Category/Paper List
- Search Summary Papers

Nucleotide Sequence Databases
RNA sequence databases
Protein sequence databases
Structure Databases
Genomics Databases (non-vertebrate)
Metabolic and Signaling Pathways
Human and other Vertebrate Genomes
Human Genes and Diseases
CancerResource
Protein Mutant Database
General human genetics databases
General polymorphism databases
Cancer gene databases
Gene-, system- or disease-specific databases
Microarray Data and other Gene Expression Databases
Proteomics Resources
Other Molecular Biology Databases
Organelle databases
Plant databases
Immunological databases
Cell biology

http://www.oxfordjournals.org/nar/database/c/

- Compilation Paper
- Category List
- Alphabetical List
- Category/Paper List
- Search Summary Papers

Oxford University Press is not responsible for the content of external internet sites

Major Molecular Databases

The most popular bioinformatics databases focus on:

- Biomolecular sequence (e.g. GenBank, UniProt)
- Biomolecular structure (e.g. PDB)
- Vertebrate genomes (e.g. Ensemble)
- Small molecules (e.g. PubChem)
- Biomedical literature (e.g. PubMed)

The are also many popular "*boutique*" databases for:

- Classifying protein families, domains and motifs (e.g. PFAM, PROSITE)
- Specific organisms (e.g. WormBase, FlyBase)
- Specific proteins of biomedical importance (e.g. KinaseDB, GPCRDB)
- Specific diseases, mutations (e.g. OMIM, HGMD)
- Specific fields or methods of study (e.g. GOA, IEDB)

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See Online: 'Handout_Major_Databases.pdf'

Primary, secondary & composite databases

Bioinformatics databases can be usefully classified into *primary*, *secondary* and *composite* according to their data source.

- **Primary databases** (or *archival databases*) consist of data derived experimentally.
  - **GenBank**: NCBI's primary nucleotide sequence database.
  - **PDB:** Protein X-ray crystal and NMR structures.
- **Secondary databases** (or *derived databases*) contain information derived from a primary database.
  - **RefSeq**: non redundant set of curated reference sequences primarily from GenBank
  - **PFAM**: protein sequence families primarily from UniProt and PDB
- **Composite databases** (or *metadatabases*) join a variety of different primary and secondary database sources.
  - **OMIM**: catalog of human genes, genetic disorders and related literature
  - **GENE**: molecular data and literature related to genes with extensive links to other databases.

GENBANK & **REFSEQ**:

NCBI'S NUCLEOTIDE SEQUENCE DATABASES

What is GenBank?

- GenBank is NCBI's primary **nucleotide only** sequence database
  - Archival in nature - reflects the state of knowledge at time of submission
  - Subjective - reflects the submitter point of view
  - Redundant - can have many copies of the same nucleotide sequence
- GenBank is actually three collaborating international databases from the US, Japan and Europe
  - GenBank (US)
  - DNA Database of Japan (DDBJ)
  - European Nucleotide Archive (ENA)

GenBank sequence record

Homo sapiens kinesin family member 5A (KIF5A), mRNA – Nucleotide – NCBI

www.ncbi.nlm.nih.gov/nuccore/NM_004984.2

NCBI Resources How To Sign in to NCBI

Nucleotide (KIF5A) AND "Homo sapiens" Search

Limits Advanced Help

Display Settings: GenBank Send:

Homo sapiens kinesin family member 5A (KIF5A), mRNA

NCBI Reference Sequence: NM_004984.2

FASTA Graphics

Go to:

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LOCUS       NM_004984               3897 bp    mRNA    linear   PRI 10-JAN-2014
DEFINITION  Homo sapiens kinesin family member 5A (KIF5A), mRNA.
ACCESSION   NM_004984
VERSION     NM_004984.2  GI:45446748
KEYWORDS    RefSeq.
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE   1  (bases 1 to 3897)
  AUTHORS   Kawaguchi,K.
  TITLE     Role of kinesin-1 in the pathogenesis of SPG10, a rare form of
            her
  JOURNAL   Neu
   PUBMED   227
  REMARK    Gen
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            Rev
REFERENCE   2
  AUTHORS   Pro
            Boh
  TITLE     alpha-Synuclein oligomers inpair neuronal microtubule-kinesin
            interplay
  JOURNAL   J. Biol. Chem. 288 (30), 21742-21754 (2013)
   PUBMED   23744071
```

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Highlight Sequence Features

Find in this Sequence

Articles about the KIF5A gene

α-Synuclein oligomers impair neuronal microtubule-kinesin interp [J Biol Chem. 2013]

Peptide hormone metabolism

MHC class II antigen presentation

GenBank flat file format has defined fields including unique identifiers such as the **ACCESSION** number.

This same general format is used for other sequence database records too.

Side node: Database accession numbers

Database **accession numbers** are strings of letters and numbers used as **identifying labels** for sequences and other data within databases

- Examples (all for retinol-binding protein, RBP4):

X02775	GenBank genomic DNA sequence	DNA
NT_030059	Genomic contig	
N91759.1	An expressed sequence tag (1 of 170)	RNA
NM_006744	RefSeq DNA sequence (from a transcript)	
NP_007635	RefSeq protein	Protein
AAC02945	GenBank protein	
Q28369	UniProtKB/SwissProt protein	
1KT7	Protein Data Bank structure record	
PMID: 12205585	PubMed IDs identify articles at NCBI/NIH	Literature

GenBank sequence record

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www.ncbi.nlm.nih.gov/nuccore/NM_004984.2

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Nucleotide | Nucleotide | (KIF5A) AND "Homo sapiens" | Search

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NCBI Reference Sequence: NM_004984.2

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  AUTHORS   Kawaguchi,K.
  TITLE     Role of kinesin-1 in the pathogenesis of SPG10, a rare form of
            hereditary spastic paraplegia
  JOURNAL   Neuroscientist 19 (4), 336-344 (2013)
   PUBMED   22785106
  REMARK    GeneRIF: A review of the mechanism of pathogenesis involved in
            spastic paraplegia type 10 when KIF5A is inactivated by mutations.
            Review article
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  AUTHORS   Prots,I., Veber,V., Brey,S., Campioni,S., Buder,K., Riek,R.,
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  TITLE     alpha-Synuclein oligomers impair neuronal microtubule-kinesin
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See all...

Pathways for the KIF5A gene

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www.ncbi.nlm.nih.gov/nuccore/NM_004984.2

NCBI Resources How To Sign in to NCBI

Nucleotide | Nucleotide | (KIF5A) AND "Homo sapiens" | Search

Limits Advanced Help

Display Settings: GenBank Send: Change region shown

Homo sapiens kinesin family member 5A (KIF5A), mRNA

NCBI Reference Sequence: NM_004984.2

FASTA Graphics

Can set different display formats here

Go to:

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LOCUS       NM_004984                3897 bp    mRNA    linear   PRI 10-JAN-2014
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  ORGANISM  Homo sapiens
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REFERENCE   2  (bases 1 to 3897)
  AUTHORS   Prots,I., Veber,V., Brey,S., Campioni,S., Buder,K., Riek,R.,
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  TITLE     alpha-Synuclein oligomers impair neuronal microtubule-kinesin
            interplay
  JOURNAL   J. Biol. Chem. 288 (30), 21742-21754 (2013)
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- Pick Primers
- Highlight Sequence Features
- Find in this Sequence

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See all...

Pathways for the KIF5A gene

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- MHC class II antigen presentation

FASTA sequence record

Homo sapiens kinesin family member 5A (KIF5A), mRNA – Nucleotide – NCBI

www.ncbi.nlm.nih.gov/nuccore/45446748?report=fasta

NCBI Resources How To Sign in to NCBI

Nucleotide Nucleotide Search

Limits Advanced Help

Display Settings: FASTA Send:

Homo sapiens kinesin family member 5A (KIF5A), mRNA

NCBI Reference Sequence: NM_004984.2

GenBank Graphics

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FASTA sequence files consist of records where each record begins with a “>” and header information on that same line. Each subsequent line of the record is sequence information.

This format is commonly used by sequence analysis programs.

Change region shown

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Pathways for the KIF5A gene

Peptide hormone metabolism

MHC class II antigen presentation

GenBank ‘graphics’ sequence record

NM_004984.2: Homo sapiens kinesin family member 5A (KIF5A), mRNA

www.ncbi.nlm.nih.gov/nuccore/45446748?report=graph

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Homo sapiens kinesin family member 5A (KIF5A), mRNA

NCBI Reference Sequence: NM_004984.2

GenBank FASTA

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NM_004984.2: 1..3.9K (3.9Kbp) Tools Configure

Genes - Exon

Genes

KIF5A

NP_004979.2

KISc

Globular

KISc_KHC_KIF5

ATP binding site (c...

acetylo... Microtubule-binding

microtubule interact...

STS Markers

D12S1839

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See all...

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Dopaminergic synapse

See all...

Reference sequence information

RefSeq alternative splicing

See the other reference mRNA sequence splice variant for the KIF5A gene

GenBank sequence record, cont.

Homo sapiens kinesin family member 5A (KIF5A), mRNA - Nucleotide - NCBI

www.ncbi.nlm.nih.gov/nuccore/NM_004984.2

NCBI Resources How To Sign in to NCBI

Nucleotide | Nucleotide | (KIF5A) AND "Homo sapiens" | Search

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Display Settings: GenBank Send:

Homo sapiens kinesin family member 5A (KIF5A), mRNA

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ACCESSION   NM_004984
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KEYWORDS    RefSeq.
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
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REFERENCE   1  (bases 1 to 3897)
  AUTHORS   Kawaguchi,K.
  TITLE     Role of kinesin-1 in the pathogenesis of SPG10, a rare form of
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  JOURNAL   Neuroscientist 19 (4), 336-344 (2013)
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  REMARK    GeneRIF: A review of the mechanism of pathogenesis involved in
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REFERENCE   2  (bases 1 to 3897)
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  JOURNAL   J. Biol. Chem. 288 (30), 21742-21754 (2013)
   PUBMED   23744071
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GenBank sequence record, cont.

Homo sapiens kinesin family member 5A (KIF5A), mRNA - Nucleotide - NCBI

www.ncbi.nlm.nih.gov/nuccore/45446748?report=genbank&to=3897#feature_45446748

Reader

Homo sapiens kinesin family member 5A (KIF5A), mRNA - Nucleotide - NCBI

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OMIM
Probe
Protein
PubMed
PubMed (RefSeq)

The **FEATURES** section contains annotations including a conceptual translation of the nucleotide sequence.

Recent activity

Turn Off Clear

Homo sapiens kinesin family member 5A (KIF5A), mRNA Nucleotide

(kinesin) AND "Homo sapiens"[porgn] (1351) Nucleotide

kinesin (37064) Nucleotide

See more...

GenBank sequence record, cont.

Homo sapiens kinesin family member 5A (KIF5A), mRNA - Nucleotide - NCBI

www.ncbi.nlm.nih.gov/nuccore/45446748?report=genbank&to=3897#sequence_45446748

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The actual sequence entry starts after the word **ORIGIN**

RefSeq: NCBI's Derivative Sequence Database

- RefSeq entries are hand curated best representation of a transcript or protein (in their judgement)
- Non-redundant for a given species although alternate transcript forms will be included if there is good evidence

- Experimentally verified transcripts and proteins
  accession numbers begin with "NM_" or "NP_"
- Model transcripts and proteins based on bioinformatics predictions with little experimental support
  accession numbers begin with "XM_" or "XP_"
- RefSeq also contains contigs and chromosome records

UNIPROT:

THE PREMIER PROTEIN SEQUENCE DATABASE

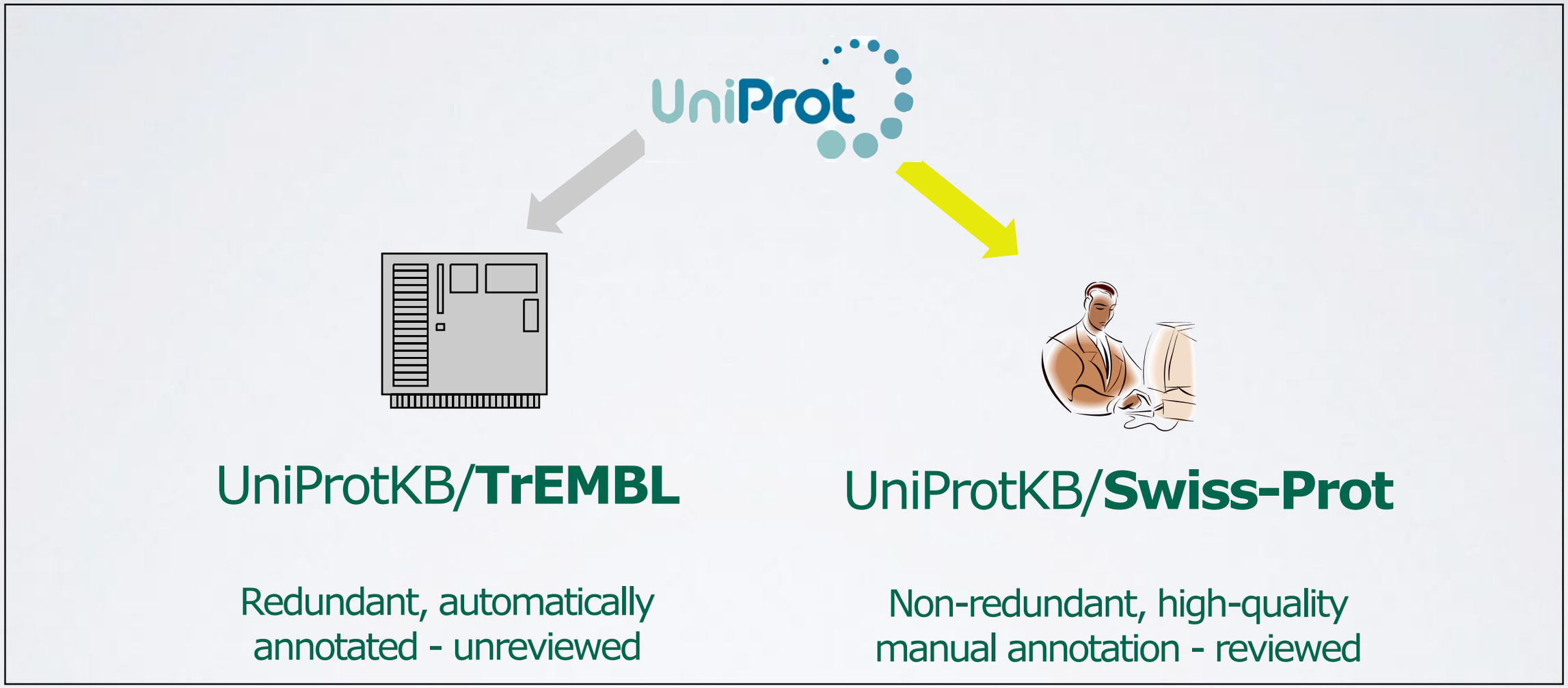
UniProt: Protein sequence database

UniProt is a comprehensive, high-quality resource of protein sequence and functional information

- UniProt comprises four databases:

1. **UniProtKB** (Knowledgebase)
   Containing **Swiss-Prot** and **TrEMBL** components
   (these correspond to hand curated and automatically annotated entries respectively)
2. **UniRef** (Reference Clusters)
   Filtered version of UniProtKB at various levels of sequence identity
   e.g. UniRef90 contains sequences with a maximum of 90% sequence identity to each other
3. **UniParc** (Archive) with database cross-references to source.
4. **UniMES** (Metagenomic and Environmental Sequences)

The two sides of UniProtKB



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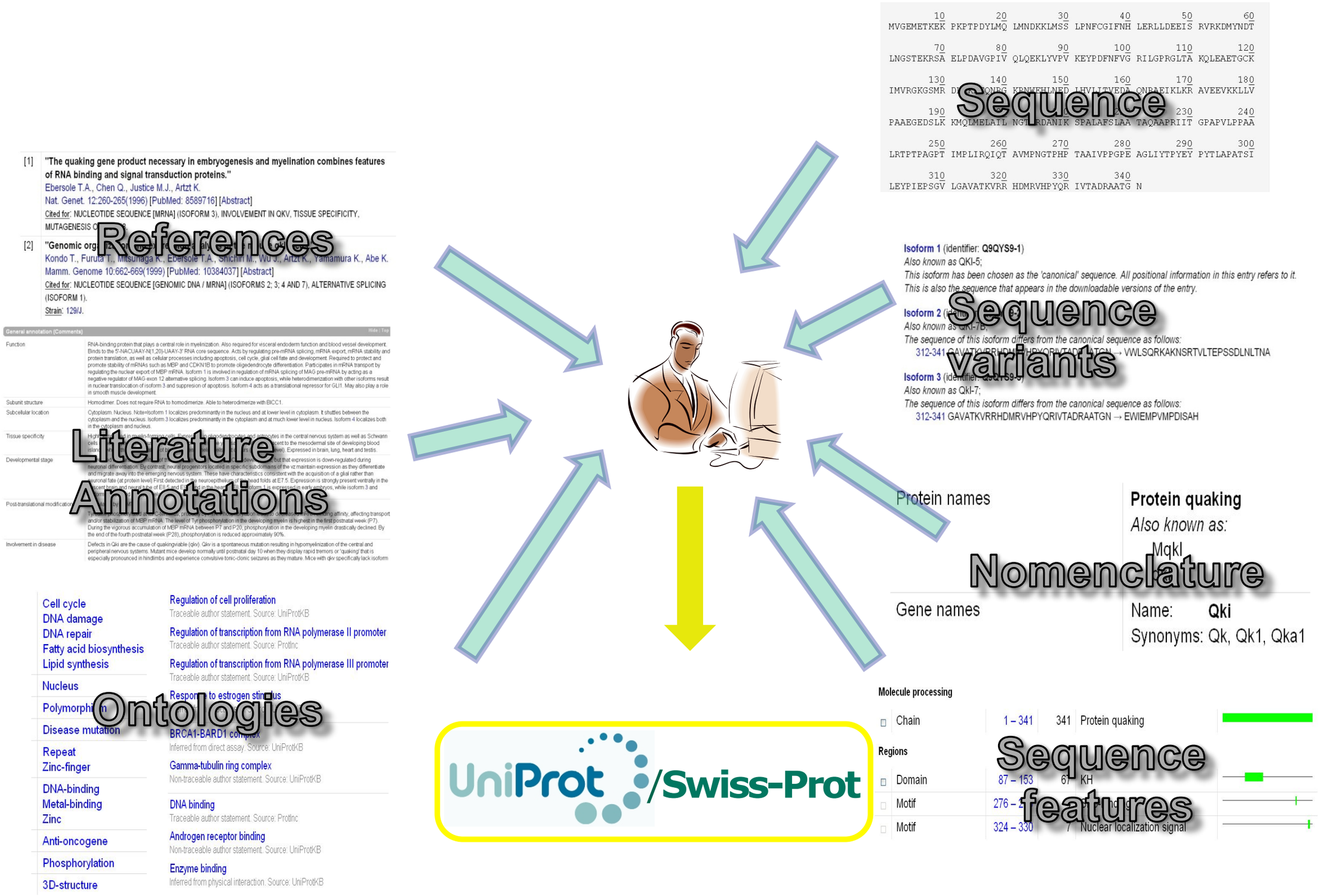
Reviewed, UniProtKB/Swiss-Prot **P38398** (BRCA1_HUMAN)

Indicators of which part of UniProt an entry belongs to include the color of the stars and the ID

The main information added to a UniProt/Swiss-Prot entry



Organism-specific databases

AGD
BuruList
CGD
CYGD
dictyBase
EchoBASE
EcoGene
euHCVdb
FlyBase
GeneDB_Spombe
GeneFarm
GeneLynx
Gramene
H-InvDB
HGNC
HPA
LegioList
Leproma
ListiList
MaizeGDB
MGI
MIM
MypuList
Orphanet
PharmGKB
PseudoCAP
PhotoList
RGD
SagaList
SGD
StyGene
SubtiList
TAIR
TubercuList
WormBase
WormPep
ZFIN

Sequence databases

EMBL
PIR
RefSeq
UniGene

Enzyme and pathway databases

BioCyc
Reactome

Family and domain databases

Gene3D
HAMAP
InterPro
PANTHER
PIRSF
Pfam
PRINTS
ProDom
PROSITE
SMART
TIGRFAMs

2D-gel databases

2DBase-Ecoli
ANU-2DPAGE
Aarhus/Ghent-2DPAGE
COMPLUYEAST-2DPAGE
Cornea-2DPAGE
DOSAC-COBS-2DPAGE
ECO2DBASE
HSC-2DPAGE
OGP
PHCI-2DPAGE
PMMA-2DPAGE
Rat-heart-2DPAGE
REPRODUCTION-2DPAGE
Siena-2DPAGE
SWISS-2DPAGE
World-2DPAGE

UniProt

UniProt provides cross-references to a large number other resources and can serve as a useful “portal” when you first begin to investigate a particular protein

Genome annotation databases

Ensembl
GenomeReviews
GeneID
KEGG
NMPDR
TIGR
Vectorbase

PTM databases

GlycoSuiteDB
PhosSite
PhosphoSite

3D structure databases

DisProt
HSSP
PDB
PDBsum
SMR

Protein family/group databases

GermOnline
MEROPS
PeroxiBase
PptaseDB
REBASE

Miscellaneous

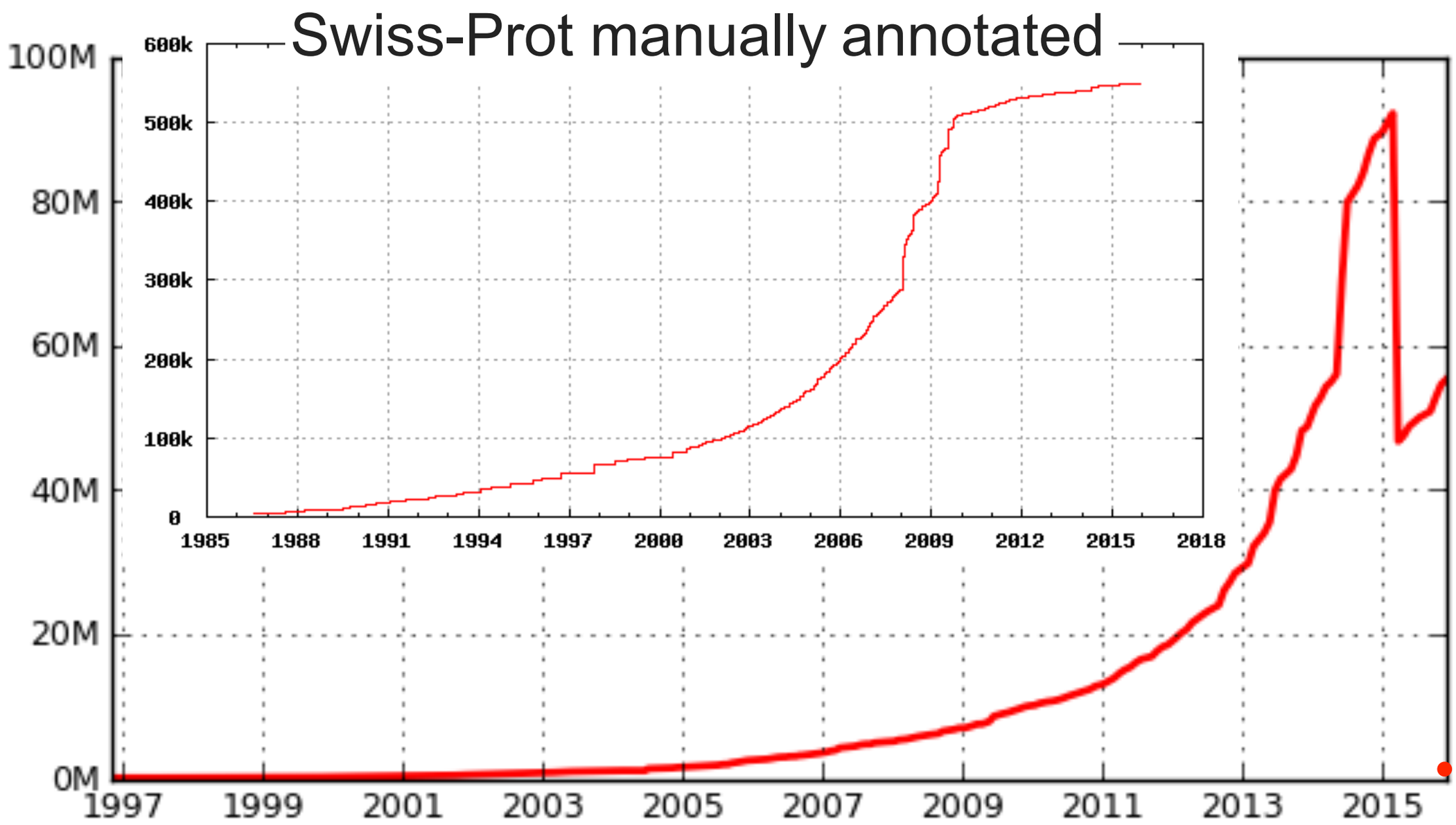
ArrayExpress
BindingDB
CleanEx
dbSNP
DIP
DrugBank
GO
HOGENOM
HOVERGEN
IntAct
LinkHub
PeptideAtlas
ProMEX

UniProt/Swiss-Prot *vs* UniProt/TrEMBL

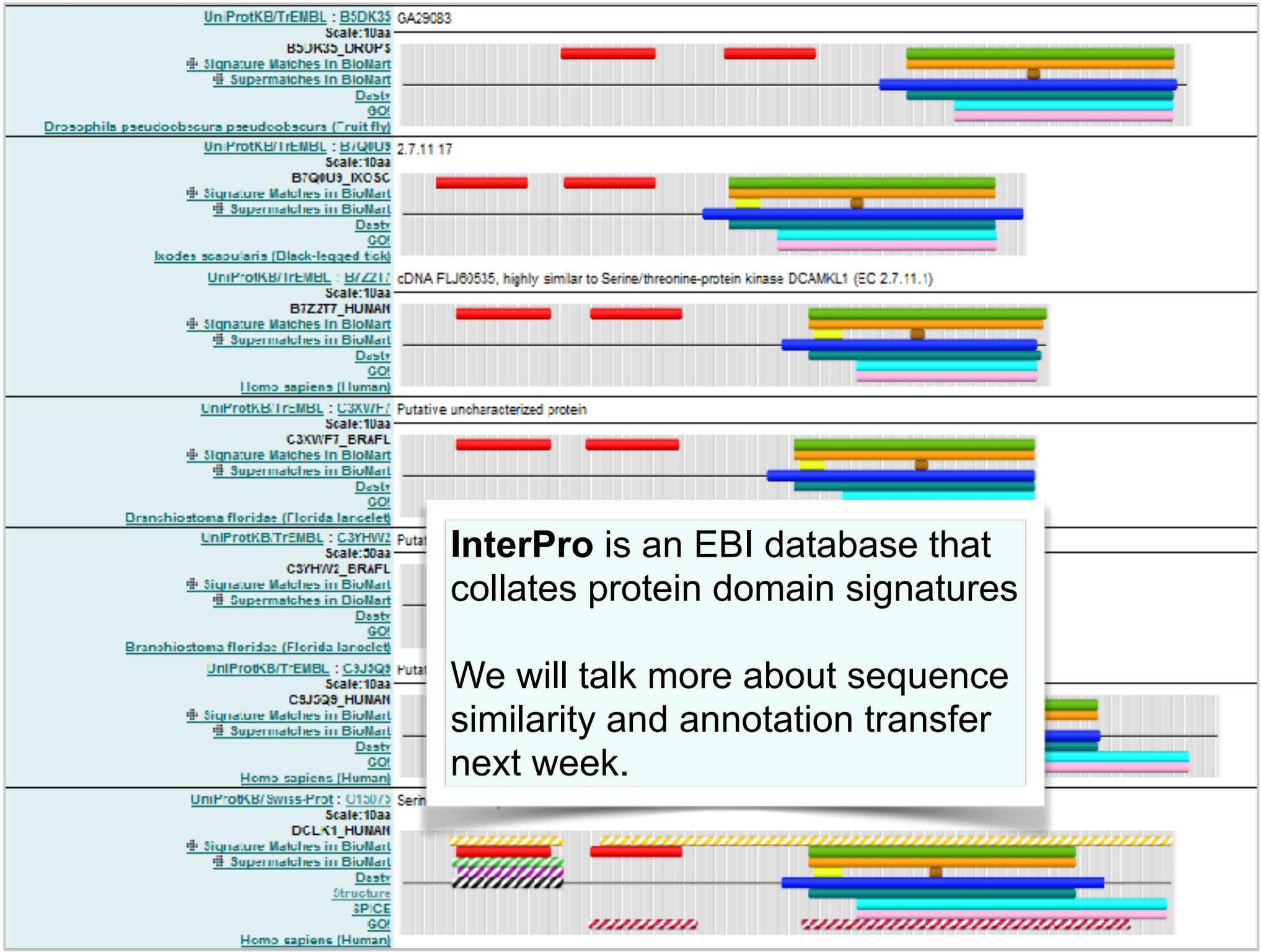
- *UniProtKB/Swiss-Prot* is a **non-redundant** database with one entry per protein

- *UniProtKB/TrEMBL* is a **redundant** database with one entry per translated ENA entry (ENA is the EBI's equivalent of GenBank)
  - Therefore TrEMBL can contain multiple entries for the same protein
  - Multiple UniProtKB/TrEMBL entries for the same protein can arise due to:
    - Erroneous gene model predictions
    - Sequence errors (Frame shifts)
    - Polymorphisms
    - Alternative start sites
    - Isoforms
    - OR because the same sequence was submitted by different people

Side note: Automatic Annotation (sharing the wealth)



Same domain composition = same function = annotation transfer



InterPro is an EBI database that collates protein domain signatures

We will talk more about sequence similarity and annotation transfer next week.

DATABASE VIGNETTE

You have just come out a seminar about gastric cancer and one of your co-workers asks:

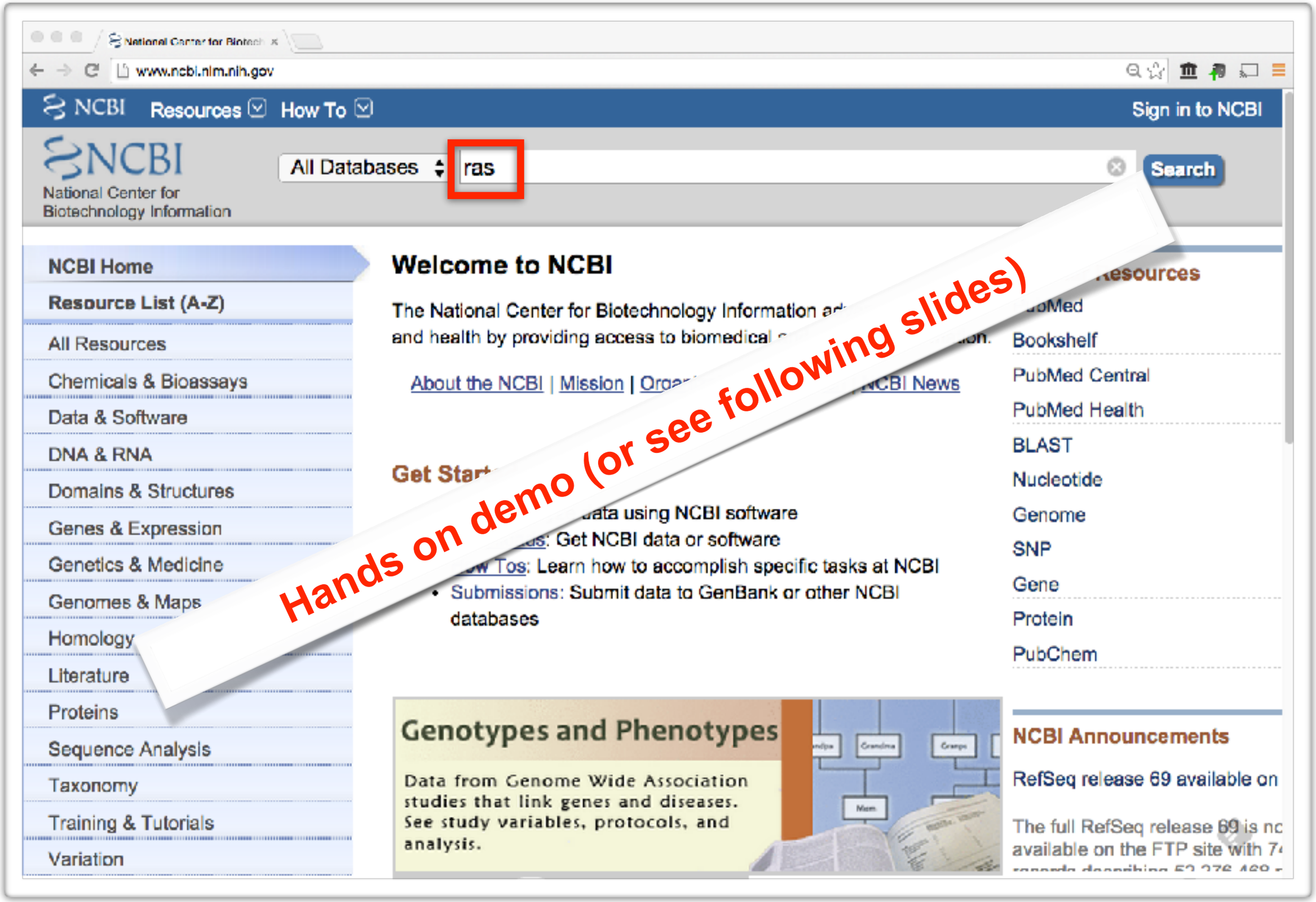
> *"What do you know about that 'Kras' gene the speaker kept taking about?"*

You have some recollection about hearing of 'Ras' before. How would you find out more?

- Google?
- Library?
- **Bioinformatics databases at NCBI and EBI!**

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

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



www.ncbi.nlm.nih.gov/gquery/?term=ras

NCBI Resources How To Sign in to NCBI

Search NCBI databases

Help

ras Search

About 2,978,774 search results for "ras"

Literature

Books	1,677	books and reports
MeSH	402	ontology used for PubMed indexing
NLM Catalog	223	books, journals and more in the NLM Collections
PubMed	54,672	scientific & medical abstracts/citations
PubMed Central	96,114	full-text journal articles

Health

ClinVar	759	human variations of clinical significance
dbGaP	120	genotype/phenotype interaction studies
GTR	1,879	genetic testing registry

Genes

EST	3,985	expressed sequence tag sequences
Gene	87,165	collected information about gene loci
GEO DataSets	3,732	functional genomics studies
GEO Profiles	1,622,789	gene expression and molecular abundance profiles
HomoloGene	696	homologous gene sets for selected organisms
PopSet	2,254	sequence sets from phylogenetic and population studies
UniGene	4,770	clusters of expressed transcripts

Proteins

www.ncbi.nlm.nih.gov/gene/?term=ras

NCBI Resources How To Sign in to NCBI

Gene | Gene | ras | Search

Save search Advanced Help

Show additional filters

Clear all

Gene sources
Genomic
Mitochondria
Organelles
Plasmids
Plastids

Categories
Alternatively spliced
Annotated genes
Non-coding
Protein-coding
Pseudogene

Sequence content
CCDS
Ensembl
RefSeq

Display Settings: Tabular, 20 per page, Sorted by Relevance **Send to:**

Hide sidebar >>

Did you mean ras as a gene symbol?
Search Gene for ras as a symbol.

<< First < Prev Page 1 of 4282 Next > Last >>

Results: 1 to 20 of 85633

Filters activated: Current only. Clear all to show 87165 items.

Name/Gene ID	Description	Location	Aliases
ras ID: 19412	resistance to audiogenic seizures [*Mus musculus* (house mouse)]		asr
ras ID: 43873	raspberry [*Drosophila melanogaster* (fruit fly)]	Chromosome X, NC_004354.4 (10744502..10749097)	Dmel_CG1799, CG11485, CG1799, Dmel\CG1799, EP(X)1093,

Filters: Manage Filters

Top Organisms [Tree]
Homo sapiens *(1126)*
Mus musculus *(823)*
Rattus norvegicus *(625)*
Oreochromis niloticus *(533)*
Neolamprologus brichardi *(507)*
All other taxa *(82019)*
More...

Find related data
Database:
Select
Find items

Search details
ras[All Fields] AND alive[property]

www.ncbi.nlm.nih.gov/gene

NCBI Resources How To Sign in to NCBI

Gene | Gene | (ras) AND "Homo sapiens"[porgn:__txid9606] | Search

Save search Advanced Help

Show additional filters

Clear all

Gene sources
Genomic

Categories
Alternatively spliced
Annotated genes
Non-coding
Protein-coding
Pseudogene

Sequence content
CCDS
Ensembl
RefSeq

Status clear
✓ Current only

Chromosome locations
Select

Display Settings: Tabular, 20 per page, Sorted by Relevance Send to:

Hide sidebar >>

Results: 1 to 20 of 1126 << First < Prev Page 1 of 57 Next > Last >>

Filters activated: Current only. Clear all to show 1499 items.

Name/Gene ID	Description	Location	Aliases
NRAS ID: 4893	neuroblastoma RAS viral (v-ras) oncogene homolog [*Homo sapiens* (human)]	Chromosome 1, NC_000001.11 (114704464..114716894, complement)	RP5-1000E10.2, ALPS4, CMNS, N-ras, NCMS1, NS6, NRAS
KRAS ID: 3845	Kirsten rat sarcoma viral oncogene homolog [*Homo sapiens* (human)]	Chromosome 12, NC_000012.12 (25205246..25250923, complement)	C-K-RAS, CFC2, K-RAS2A, K-RAS2B, K-RAS4A, K-RAS4B, KI-RAS1, KRAS2, NS, NS3, RASK2

Filters: Manage Filters

Find related data

Database:
Select

Find items

Search details

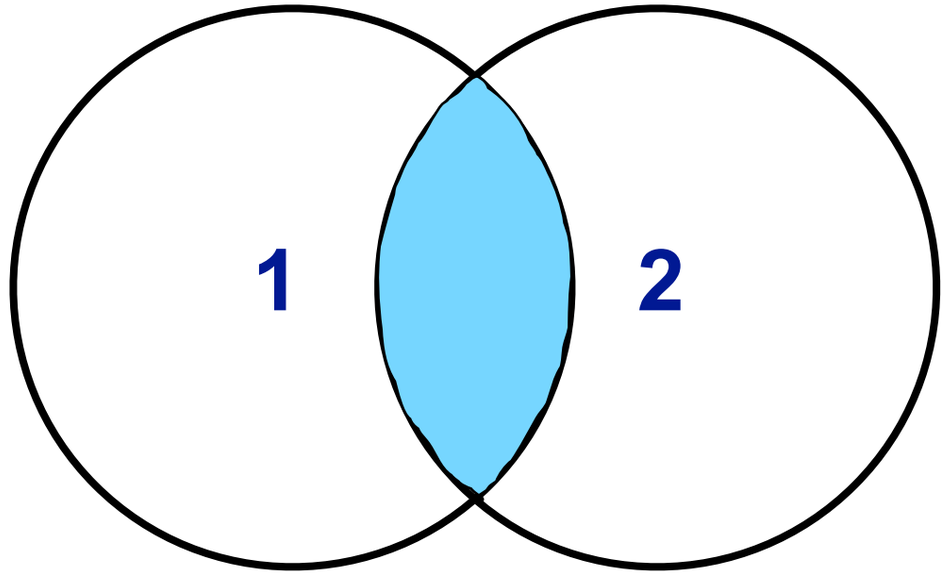
```
ras[All Fields] AND "Homo
sapiens"[porgn] AND
alive[property]
```

Search See more...

Recent activity

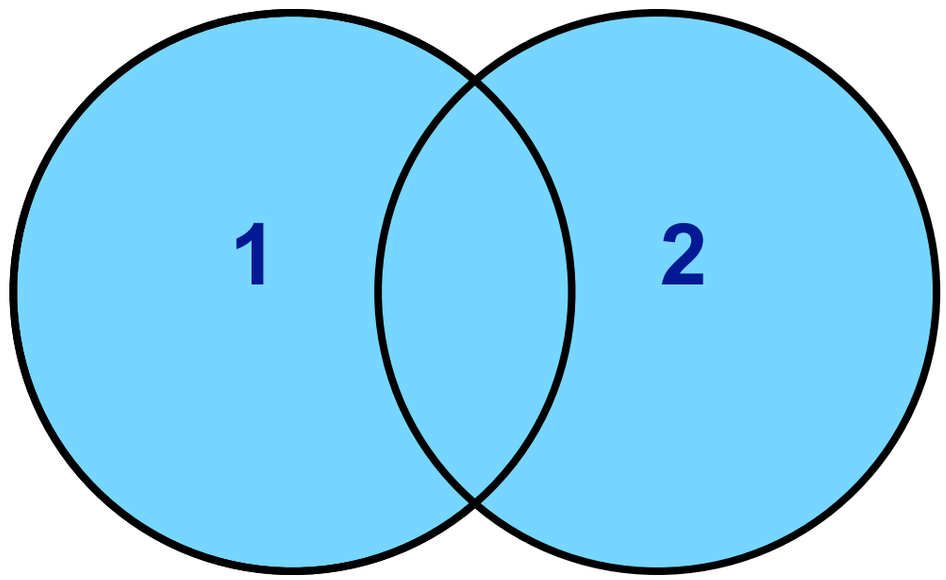
Turn Off Clear

1 AND 2



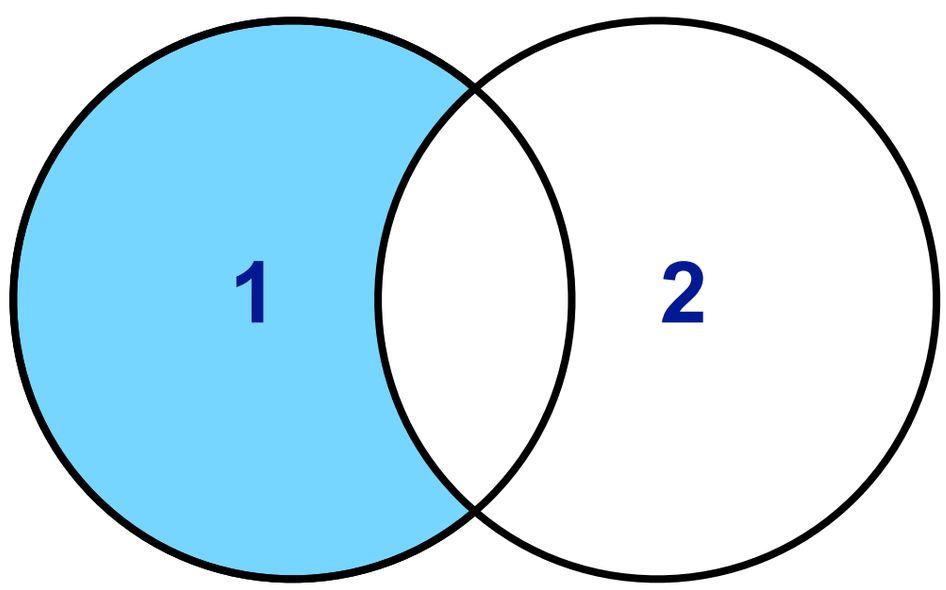
ras AND disease
(1185 results)

1 OR 2



ras OR disease
(134,872 results)

1 NOT 2



ras NOT disease
(84,448 results)

www.ncbi.nlm.nih.gov/gene

NCBI Resources How To Sign in to NCBI

Gene

Gene (ras) AND "Homo sapiens"[porgn:__txid9606] Search

Save search Advanced Help

Show additional filters

Clear all

Gene sources
Genomic

Categories
Alternatively spliced
Annotated genes
Non-coding
Protein-coding
Pseudogene

Sequence content
CCDS
Ensembl
RefSeq

Status clear
✓ Current only

Chromosome locations

Display Settings: Tabular, 20 per page, Sorted by Relevance Send to:

Results: 1 to 20 of 1126 << First < Prev Page 1 of 57 Next > Last >>

Filters activated: Current only. Clear all to show 1499 items.

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Hide sidebar >>

Filters: Manage Filters

Find related data

Database:
Select

Find items

Search details

```
ras[All Fields] AND "Homo
sapiens"[porgn] AND
alive[property]
```

Search See more...

Recent activity

Turn Off Clear

www.ncbi.nlm.nih.gov/gene/3845

NCBI Resources How To Sign in to NCBI

Gene Gene Search

Advanced Help

Display Settings: Full Report Send to: Hide sidebar >>

KRAS Kirsten rat sarcoma viral oncogene homolog [*Homo sapiens* (human)]

Gene ID: 3845, updated on 4-Jan-2015

Summary

Official Symbol	KRAS provided by HGNC
Official Full Name	Kirsten rat sarcoma viral oncogene homolog provided by HGNC
Primary source	HGNC:HGNC:6407
See related	Ensembl:ENSG00000133703; HPRD:01817; MIM:190070; Vega:OTTHUMG00000171193
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	NS; NS3; CFC2; KRAS1; KRAS2; RASK2; KI-RAS; C-K-RAS; K-RAS2A; K-

Table of contents

Example Questions:

What chromosome location and what genes are in the vicinity?

www.ncbi.nlm.nih.gov/gene/3845

NCBI Resources How To Sign in to NCBI

Gene Search Help

Display S

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KRAS

(human

Gene ID: 3845, updated on 4-Jan-2015

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Table of contents

- Summary
- Genomic context
- Genomic regions, transcripts, and products
- Bibliography
- Phenotypes
- Variation
- HIV-1 interactions
- Pathways from BioSystems
- Interactions
- General gene information
  - Markers, Related pseudogene(s), Homology, Gene Ontology
- General protein information
- NCBI Reference Sequences (RefSeq)

www.ncbi.nlm.nih.gov/gene/3845#genomic-context

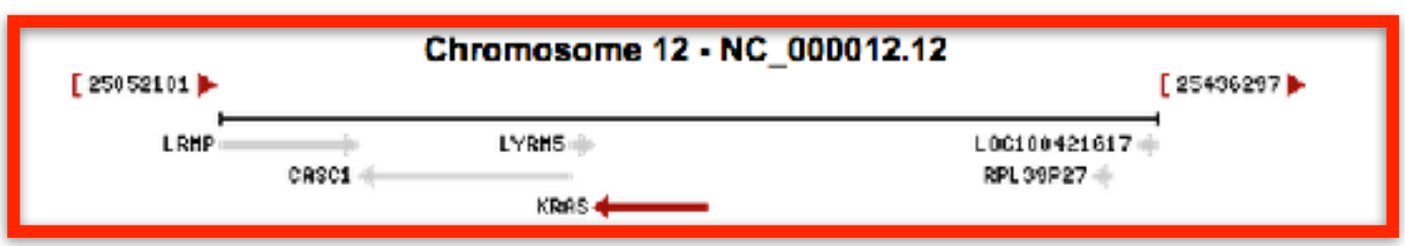
Genomic context

Location: 12p12.1

Exon count: 6

See KRAS in Epigenomics, MapViewer

Annotation release	Status	Assembly	Chr	Location
106	current	GRCh38 (GCF_000001405.26)	12	NC_000012.12 (25205246..25250923, complement)
105	previous assembly	GRCh37.p13 (GCF_000001405.25)	12	NC_000012.11 (25358180..25403870, complement)



Genomic regions, transcripts, and products

Go to reference sequence details

Genomic Sequence: NC_000012.12 chromosome 12 reference GRCh38 Primary Assembly

Go to nucleotide: Graphics FASTA GenBank

BioAssay by Target (Summary)
BioAssay, by Gene target
BioAssays, RNAi Target, Active
BioAssays, RNAi Target, Tested
BioProjects
BioSystems
Books
CCDS
ClinVar
Conserved Domains
dbVar
EST
Full text in PMC
Full text in PMC_nucleotide
Gene neighbors
Genome
GEO Profiles
GTR
HomoloGene
Map Viewer
MedGen
Nucleotide

Example Questions:

What 'molecular functions', 'biological processes', and 'cellular component' information is available?

www.ncbi.nlm.nih.gov/gene/3845

NCBI Resources How To Sign in to NCBI

Gene Search Help

Display Settin

Hide sidebar >>

KRAS Ki
(human)]

Gene ID: 3845

Summa

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  - Markers, Related pseudogene(s), Homology, Gene Ontology
- General protein information
- NCBI Reference Sequences (RefSeq)

www.ncbi.nlm.nih.gov/gene/3845#gene-ontology

Gene Ontology Provided by GOA

Function	Evidence Code	Pubs
GDP binding	IEA	
GMP binding	IEA	
GTP binding	IEA	
LRR domain binding	IEA	
protein binding	IPI	PubMed
protein complex binding	IDA	PubMed

Items 1 - 25 of 33 < Prev Page 1 of 2 Next >

Process	Evidence Code	Pubs
Fc-epsilon receptor signaling pathway	TAS	
GTP catabolic process	IEA	
MAPK cascade	TAS	
Ras protein signal transduction	TAS	
actin cytoskeleton organization	IEA	
activation of MAPKK activity	TAS	
axon guidance	TAS	
blood coagulation	TAS	

GO: Gene Ontology

GO provides a controlled vocabulary of terms for describing gene product characteristics and gene product annotation data

www.ebi.ac.uk/GOA

EMBL-EBI

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UniProt-GOA

Search

Examples: GO:0006915, tropomyosin, P06727

Overview | New to UniProt-GOA | FAQ | Contact Us

Gene Ontology Annotation (UniProt-GOA) Database

The UniProt GO annotation program aims to provide high-quality Gene Ontology (GO) annotations to proteins in the UniProt Knowledgebase (UniProtKB). The assignment of GO terms to UniProt records is an integral part of UniProt biocuration . UniProt manual and electronic GO annotations are supplemented with manual annotations supplied by external collaborating GO Consortium groups, to ensure a comprehensive GO annotation dataset is supplied to users .

UniProt is a member of the GO Consortium .

Menu

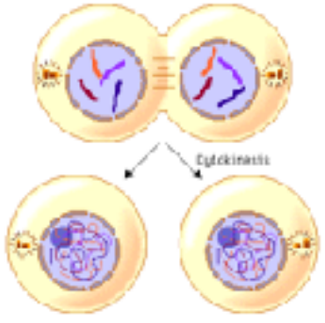
- Downloads
- Searching UniProt-GOA
- Annotation Methods
  - Annotation Tutorial
  - Manual Annotation Efforts
    - Reference Genome Annotation Initiative
    - Cardiovascular Gene Ontology Annotation Initiative
    - Renal Gene Ontology Annotation Initiative
    - Exosome Gene

Why do we need Ontologies?

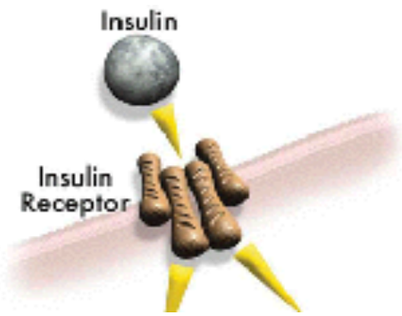
- Annotation is essential for capturing the understanding and knowledge associated with a sequence or other molecular entity
- Annotation is traditionally recorded as “free text”, which is easy to read by humans, but has a number of disadvantages, including:
  - Difficult for computers to parse
  - Quality varies from database to database
  - Terminology used varies from annotator to annotator
- Ontologies are annotations using standard vocabularies that try to address these issues
- GO is integrated with UniProt and many other databases including a number at NCBI

GO Ontologies

- There are three ontologies in GO:
  - **Biological Process**
    A commonly recognized series of events
    e.g. cell division, mitosis,



  - **Molecular Function**
    An elemental activity, task or job
    e.g. kinase activity, insulin binding



  - **Cellular Component**
    Where a gene product is located
    e.g. mitochondrion, mitochondrial membrane

www.ncbi.nlm.nih.gov/gene/3845#gene-ontology

Gene Ontology Provided by GOA

Function	Evidence Code	Pubs
GDP binding		
GMP binding		
GTP binding		
LRR domain binding		
protein binding		
protein complex binding		

The 'Gene Ontology' or GO is actually maintained by the EBI so lets switch or link over to UniProt also from the EBI.

Process	Code	Pubs
Fc-epsilon receptor signaling pathway	TAS	
GTP catabolic process	IEA	
MAPK cascade	TAS	
Ras protein signal transduction	TAS	
actin cytoskeleton organization	IEA	
activation of MAPKK activity	TAS	
axon guidance	TAS	
blood coagulation	TAS	

Scroll down to **UniProt** link

UniProt will detail much more information for protein coding genes such as this one

www.ncbi.nlm.nih.gov/gene/3845#gene-ontology

genomic	X01669.1	CAA25828.1

Items 1 - 25 of 43 < Prev Page 1 of 2 Next >

Protein Accession	Links	
	GenPept Link	UniProtKB Link
P01116.1	GenPept	UniProtKB/Swiss-Prot:P01116

Additional links

Scroll down to **UniProt** link

You are here: NCBI > Genes & Expression > Gene

Write to the Help Desk

GETTING STARTED
NCBI Education
NCBI Help Manual
NCBI Handbook
Training & Tutorials

RESOURCES
Chemicals & Bioassays
Data & Software
DNA & RNA
Domains & Structures
Genes & Expression
Genetics & Medicine
Genomes & Maps
Homology
Literature
Proteins
Sequence Analysis
Taxonomy

POPULAR
PubMed
Bookshelf
PubMed Central
PubMed Health
BLAST
Nucleotide
Genome
SNP
Gene
Protein
PubChem

FEATURED
Genetic Testing Registry
PubMed Health
GenBank
Reference Sequences
Gene Expression Omnibus
Map Viewer
Human Genome
Mouse Genome
Influenza Virus
Primer-BLAST
Sequence Read Archive

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Research at NCBI
NCBI News
NCBI FTP Site
NCBI on Facebook
NCBI on Twitter
NCBI on YouTube

UniProt will detail much more information for protein coding genes

www.uniprot.org/uniprot/P01116

UniProt

UniProtKB | Advanced

BLAST Align Retrieve/ID Mapping Help Contact

Basket

P01116 - RASK_HUMAN

Protein	**GTPase KRas**
Gene	**KRAS**
Organism	*Homo sapiens (Human)*
Status	Reviewed - ◉◉◉◉◉ - Experimental evidence at protein level[i]

BLAST Align Format Add to basket History Feedback Help video

Display None

- FUNCTION
- NAMES & TAXONOMY
- SUBCELL. LOCATION
- PATHOL./BIOTECH
- PTM / PROCESSING
- EXPRESSION
- INTERACTION
- STRUCTURE
- FAMILY & DOMAINS
- SEQUENCES (2)
- CROSS-REFERENCES

Function

Ras proteins bind GDP/GTP and possess intrinsic GTPase activity. Plays an important role in the regulation of cell proliferation (PubMed:23698361, PubMed:22711838). 2 Publications Curated

Enzyme regulation

Alternates between an inactive form bound to GDP and an active form bound to GTP. Activated by a guanine nucleotide-exchange factor (GEF) and inactivated by a GTPase-activating protein (GAP). Interaction with SOS1 promotes exchange of bound GDP by GTP. 3 Publications

Regions

Feature key	Position(s)	Length	Description	Graphical view	Feature identifier	Actions
Nucleotide binding[i]	10 – 18	9	GTP 2 Publications			
Nucleotide binding[i]	29 – 35	7	GTP 2 Publications			
Nucleotide binding[i]	59 – 60	2	GTP 2 Publications			

www.uniprot.org/uniprot/P01116

UniProtKB | Advanced

BLAST Align Retrieve/ID Mapping Help Contact

Basket

P01116 - RASK_HUMAN

Protein	GTPase KRas
Gene	KRAS
Organism	*Homo sapiens (Human)*
Status	Reviewed -

Example Questions:

What positions in the protein are responsible for GTP binding?

Display None

- FUNCTION
- NAMES & TAXONOMY
- SUBCELL. LOCATION
- PATHOL./BIOTECH
- PTM / PROCESSING
- EXPRESSION
- INTERACTION
- STRUCTURE
- FAMILY & DOMAINS
- SEQUENCES (2)
- CROSS-REFERENCES

BLAST Align

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Regions

Feature key	Position(s)	Length	Description	Graphical view	Feature identifier	Actions
Nucleotide binding	10 – 18	9	GTP 2 Publications			
Nucleotide binding	29 – 35	7	GTP 2 Publications			
Nucleotide binding	59 – 60	2	GTP 2 Publications			

Example Questions:

What variants of this enzyme are involved in gastric cancer and other human diseases?

www.uniprot.org/uniprot/P01116

Display None

- FUNCTION
- NAMES & TAXONOMY
- SUBCELL. LOCATION
- PATHOL./BIOTECH
- PTM / PROCESSING
- EXPRESSION
- INTERACTION
- STRUCTURE
- FAMILY & DOMAINS
- SEQUENCES (2)
- CROSS-REFERENCES
- PUBLICATIONS
- ENTRY INFORMATION
- MISCELLANEOUS
- SIMILAR PROTEINS

Top

Pathology & Biotech

Involvement in disease

LEUKEMIA, ACUTE MYELOGENOUS (AML)

[MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. 1 Publication

Note: The disease is caused by mutations affecting the gene represented in this entry.

Feature key	Position(s)	Length	Description	Graphical view	Feature identifier	Actions
Natural variant	10 – 10	1	G → GG in one individual with AML; expression in 3T3 cell causes cellular transformation; expression in COS cells activates the Ras-MAPK signaling pathway; lower GTPase activity; faster GDP dissociation rate. 1 Publication		VAR_034601	

LEUKEMIA, JUVENILE MYELOMONOCYTIC (JMML)

[MIM:607785]: An aggressive pediatric myelodysplastic syndrome/myeloproliferative disorder characterized by malignant transformation in the hematopoietic stem cell compartment with proliferation of differentiated progeny. Patients have splenomegaly, enlarged lymph nodes, rashes, and hemorrhages.

Note: The disease is caused by mutations affecting the gene represented in this entry.

NOONAN SYNDROME 3 (NS3)

[MIM:609942]: A form of Noonan syndrome, a disease characterized by short stature, facial dysmorphic features such as hypertelorism, a downward eyeslant and low-set posteriorly rotated ears, and a high incidence of congenital heart

Example Questions:

Are high resolution protein structures available to examine the details of these mutations?

www.uniprot.org/uniprot/P01116

Display None

- FUNCTION
- NAMES & TAXONOMY
- SUBCELL. LOCATION
- PATHOL./BIOTECH
- PTM / PROCESSING
- EXPRESSION
- INTERACTION
- STRUCTURE
- FAMILY & DOMAINS
- SEQUENCES (2)
- CROSS-REFERENCES
- PUBLICATIONS
- ENTRY INFORMATION
- MISCELLANEOUS
- SIMILAR PROTEINS

Top

Structure

Secondary structure

1 … 189

Legend: Helix Turn Beta strand

Show more details

3D structure databases

Select the link destinations:

- PDBe
- RCSB PDB
- PDBj

Entry	Method	Resolution (Å)	Chain	Positions	PDBsum
1D8D	X-ray	2.00	P	178-188	[»]
1D8E	X-ray	3.00	P	178-188	[»]
1KZO	X-ray	2.20	C	169-173	[»]
1KZP	X-ray	2.10	C	169-173	[»]
3GFT	X-ray	2.27	A/B/C/D/E/F	1-164	[»]
4DSN	X-ray	2.03	A	2-164	[»]
4DSO	X-ray	1.85	A	2-164	[»]
4EPR	X-ray	2.00	A	1-164	[»]
4EPT	X-ray	2.00	A	1-164	[»]
4EPV	X-ray	1.35	A	1-164	[»]
4EPW	X-ray	1.70	A	1-164	[»]
4EPX	X-ray	1.76	A	1-164	[»]
4EPY	X-ray	1.80	A	1-164	[»]
4L8G	X-ray	1.52	A	1-169	[»]
4LDJ	X-ray	1.15	A	1-164	[»]
4LPK	X-ray	1.50	A/B	1-169	[»]

Example Questions:

What is known about the protein family, its species distribution, number in humans and residue-wise conservation, etc… ?

www.uniprot.org/uniprot/P01116

Display None

- FUNCTION
- NAMES & TAXONOMY
- SUBCELL. LOCATION
- PATHOL./BIOTECH
- PTM / PROCESSING
- EXPRESSION
- INTERACTION
- STRUCTURE
- FAMILY & DOMAINS
- SEQUENCES (2)
- CROSS-REFERENCES
- PUBLICATIONS
- ENTRY INFORMATION
- MISCELLANEOUS
- SIMILAR PROTEINS

▲Top

PhylomeDB[i] P01

TreeFam[i] TF3

Family and domain databases

Gene3D[i]	3.40.50.300. 1 hit.
InterPro[i]	IPR027417. P-loop_NTPase. IPR005225. Small_GTP-bd_dom. IPR001806. Small_GTPase. IPR020849. Small_GTPase_Ras. [Graphical view]
PANTHER[i]	PTHR24070. PTHR24070. 1 hit.
Pfam[i]	PF00071. Ras. 1 hit. [Graphical view]
PRINTS[i]	PR00449. RASTRNSFRMNG.
SMART[i]	SM00173. RAS. 1 hit. [Graphical view]
SUPFAM[i]	SSF52540. SSF52540. 1 hit.
TIGRFAMs[i]	TIGR00231. small_GTP. 1 hit.
PROSITE[i]	PS51421. RAS. 1 hit. [Graphical view]

PFAM is one of the best protein family databases

Sequences (2)[i]

Sequence status[i]: Complete.

Sequence processing[i]: The displayed sequence is further processed into a mature form.

This entry describes **2** isoforms[i] produced by **alternative splicing**. Align

Example Questions:

What is known about the protein family, its **species distribution**, number in humans and residue-wise conservation, etc… ?

pfam.xfam.org/family/PF00071

EMBL-EBI

HOME | SEARC

Family: *Ras* (PF00071)

332 architectures | 21243 sequences | 30 interactions | 1006 species | 663 structures

- Summary
- Domain organisation
- Clan
- Alignments
- HMM logo
- Trees
- Curation & model
- Species
- Interactions
- Structures

Jump to… enter ID/acc Go

Summary: Ras family

Pfam includes annotations and additional family information from a range of different sources. These sources can be accessed via the tabs below.

Wikipedia: Ras subfamily | Wikipedia: Ras superfamily | Pfam | InterPro

This is the Wikipedia entry entitled "Ras subfamily". More...

Ras subfamily

Edit Wikipedia article

This article is about p21/Ras protein. For the p21/waf1 protein, see p21.

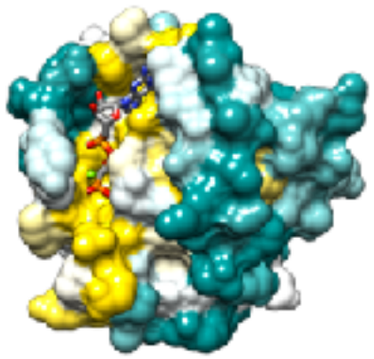
Ras is the name given to a family of related proteins which is ubiquitously expressed in all cell lineages and organs. All Ras protein family members belong to a class of protein called small GTPase, and are involved in transmitting signals within cells (cellular signal transduction). Ras is the prototypical member of the Ras superfamily of proteins, which are all related in 3D structure and regulate diverse cell behaviours.

The name 'Ras' is an abbreviation of 'Rat sarcoma', reflecting the way the first members of the protein family were discovered. The name ras is also used to refer to the family of genes encoding those proteins.

When Ras is 'switched on' by incoming signals, it subsequently switches on other proteins, which ultimately turn on genes involved in cell growth, differentiation and survival. As a result, mutations in ras genes can lead to the production of permanently activated Ras proteins. This can cause unintended and overactive signalling inside the cell, even in the absence of incoming signals.

Because these signals result in cell growth and division, overactive Ras signaling can ultimately lead to cancer.[1] The 3 Ras genes in humans (HRAS, KRAS, and NRAS) are the most common oncogenes in human cancer; mutations that permanently activate Ras are found in 20% to 25% of all human tumors and up to 90% in certain types of cancer (e.g., pancreatic cancer).[2] For this reason, Ras inhibitors are being studied as a treatment for cancer, and other diseases with Ras overexpression.

Contents [hide]

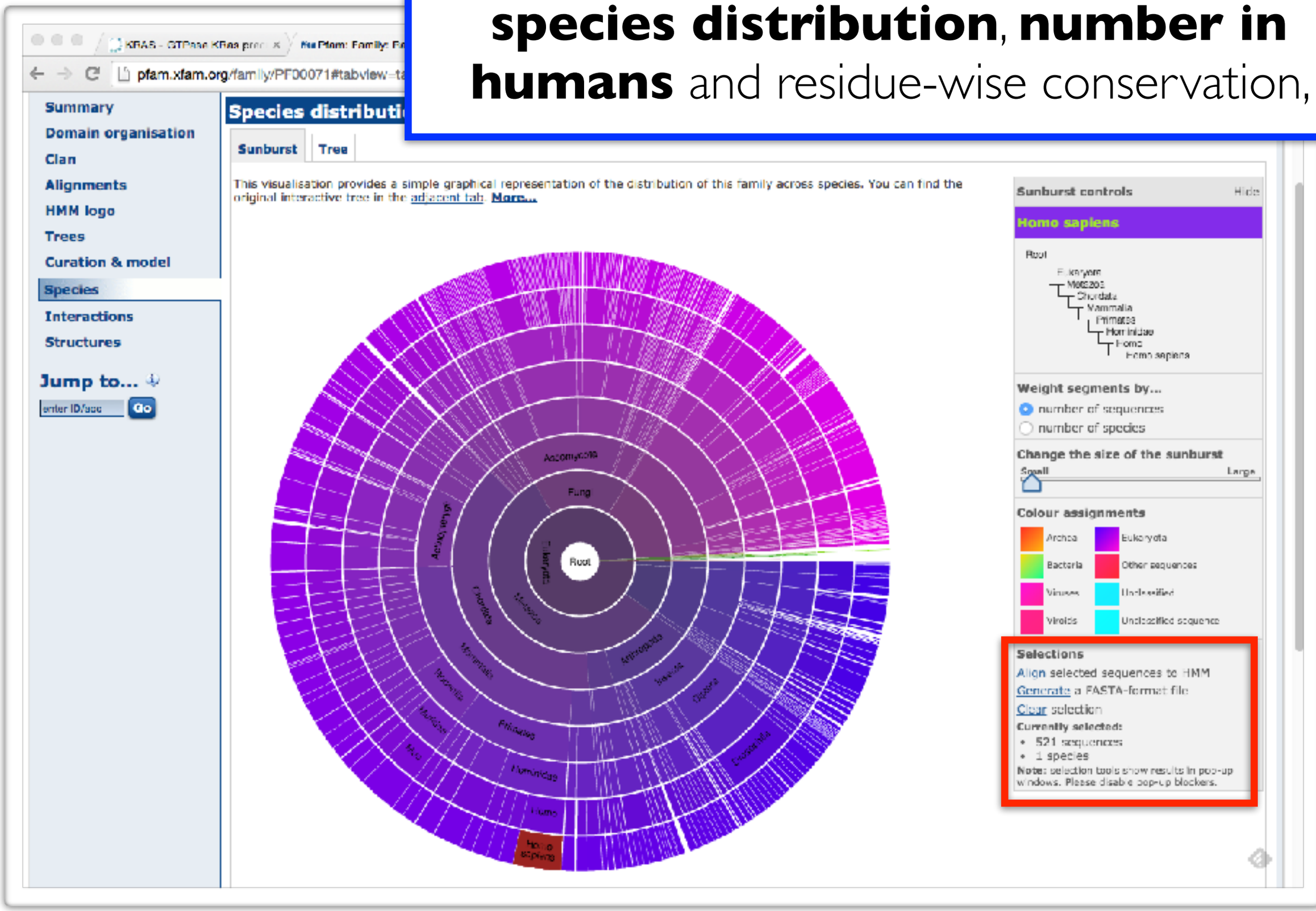


H-Ras structure PDB 121p, surface colored by conservation in Pfam seed alignment: gold, most conserved; dark cyan, least conserved.

Identifiers	
Symbol	Ras
Pfam	PF00071
InterPro	IPR013753
PROSITE	PDOC00017
SCOP	5p21
SUPERFAMILY	5p21

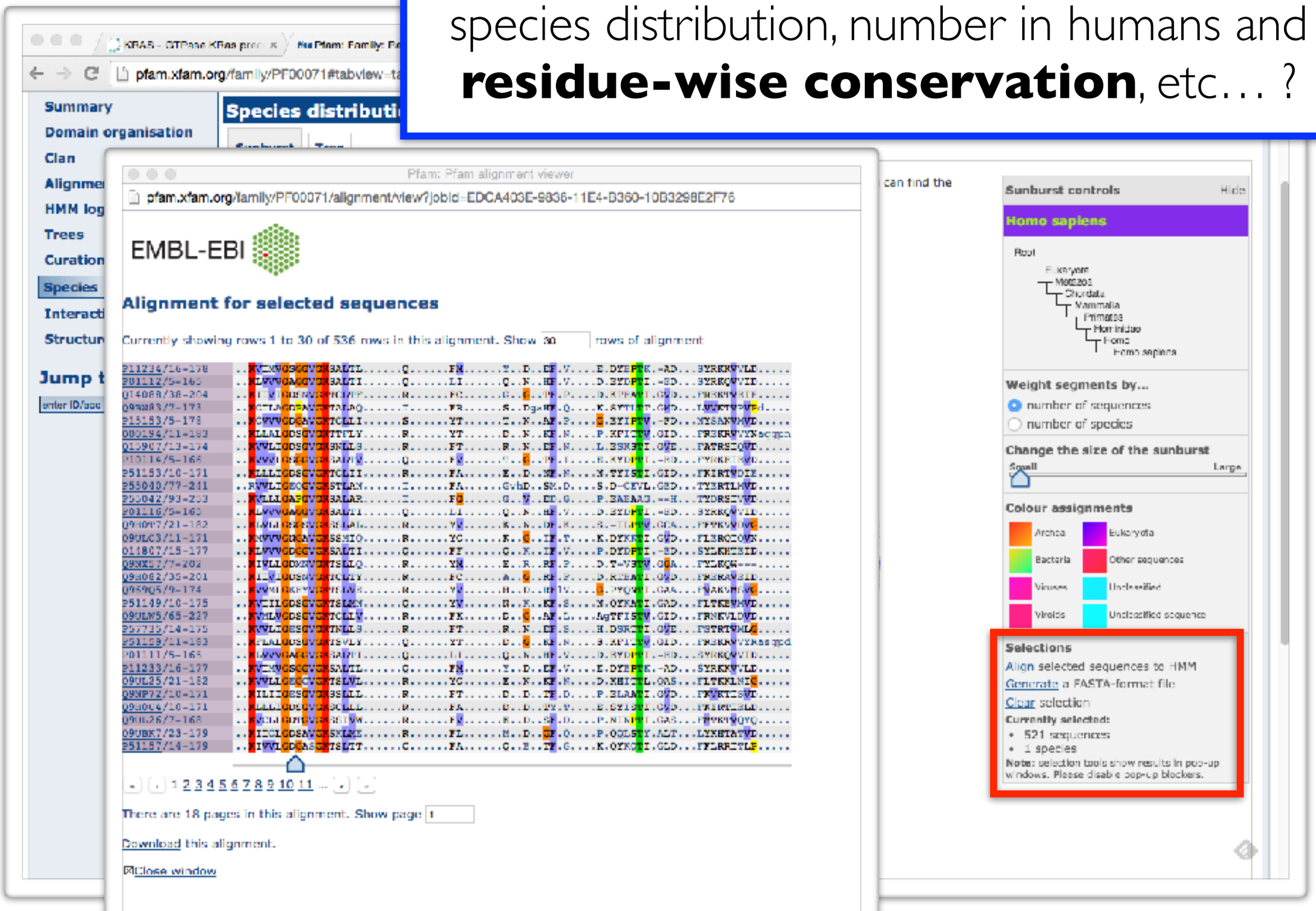
Example Questions:

What is known about the protein family, its **species distribution**, **number in humans** and residue-wise conservation,



Example Questions:

What is known about the protein family, its species distribution, number in humans and **residue-wise conservation**, etc… ?



Example Questions:

What is known about the protein family, its species distribution, number in humans and **residue-wise conservation**, etc… ?

pfam.xfam.org/family/PF00071#tabview=tab4

EMBL-EBI

HOME | SEARCH | BROWSE | FTP | HELP | ABOUT

Pfam

keyword search Go

Family: *Ras* (PF00071)

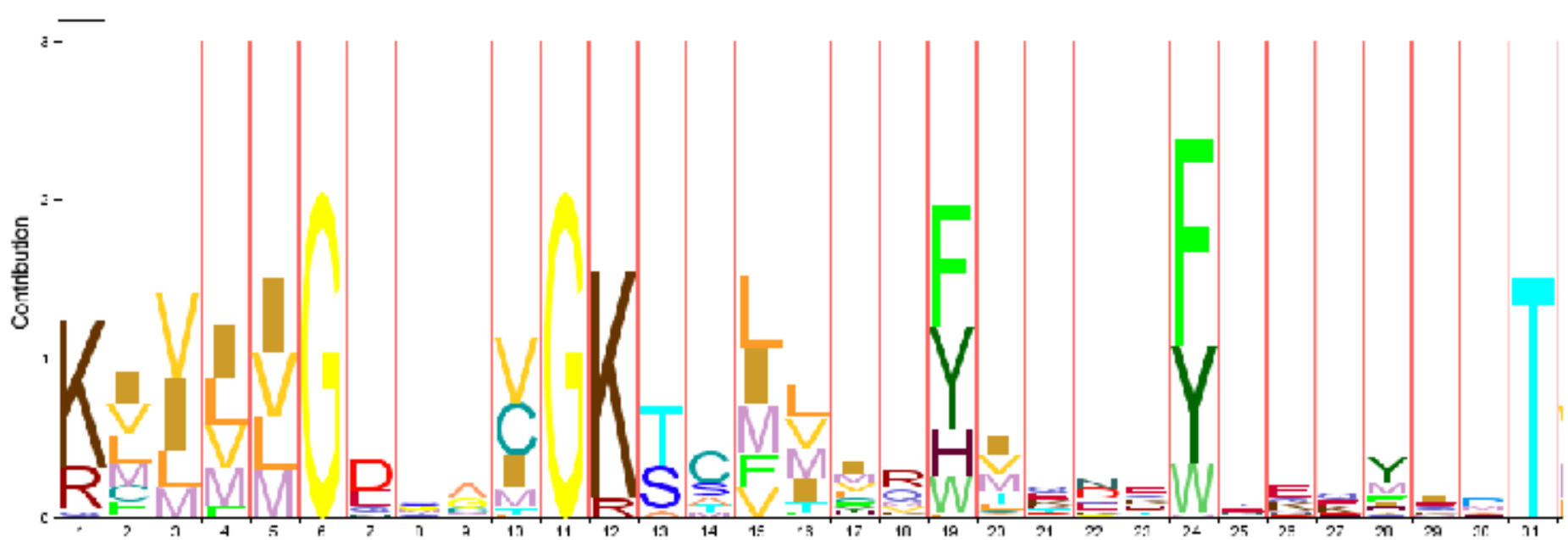
332 architectures | 21243 sequences | 30 interactions | 1006 species | 663 structures

- Summary
- Domain organisation
- Clan
- Alignments
- HMM logo
- Trees
- Curation & model
- Species
- Interactions
- Structures

Jump to... enter ID/acc Go

HMM logo

HMM logos is one way of visualising profile HMMs. Logos provide a quick overview of the properties of an HMM in a graphical form. You can see a more detailed description of HMM logos and find out how you can interpret them here. More...



Comments or questions on the site? Send a mail to pfam-help@ebi.ac.uk.

European Molecular Biology Laboratory

Family: *Kinesin* (PF00225)

Loading page components (1 remaining)...

126 architectures 4150 sequences 6 interactions 248 species 114 structures

Summary
Domain organisation
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Trees
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Species
Interactions
Structures

Jump to...
enter ID/acc Go

Interactions

There are **6** interactions for this family. More...

Tubulin Tubulin_C	Tubulin_C	Kinesin	Tubulin	Kinesin

Family: *Kinesin* (PF00225)

126 architectures 4150 sequences 6 interactions 248 species 114 structures

Summary
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Interactions
Structures

Jump to...
enter ID/acc Go

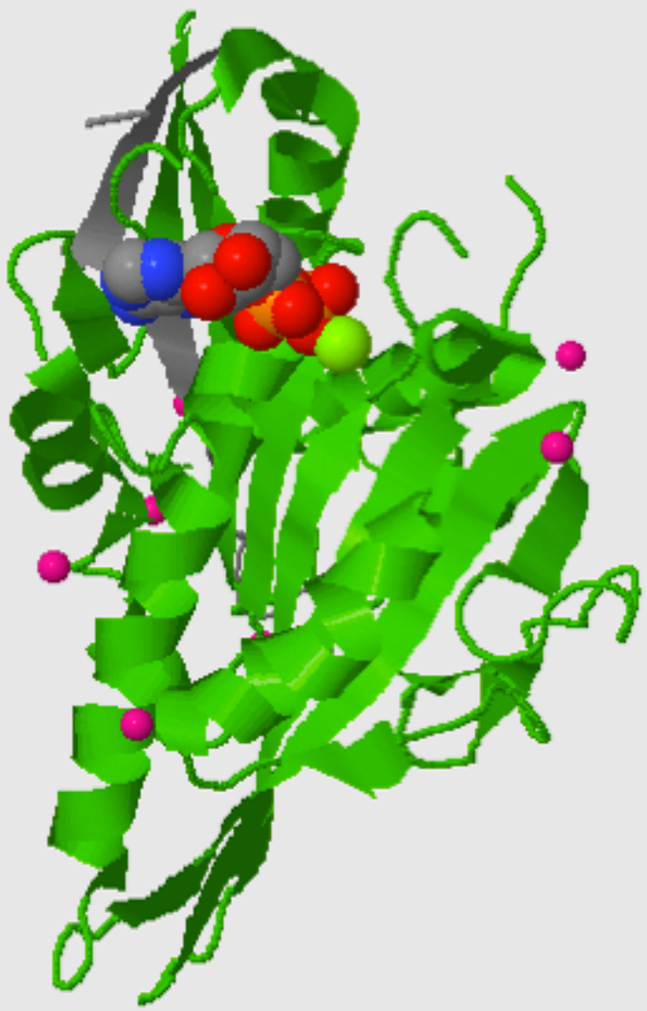
Structures

For those sequences which have a structure in the Protein DataBank, we use the mapping between UniProt, PDB and Pfam coordinate systems from the PDBe group, to allow us to map Pfam domains onto UniProt sequences and three-dimensional protein structures. The table below shows the structures on which the **Kinesin** domain has been found.

UniProt entry	UniProt residues	PDB ID	PDB chain ID	PDB residues	View
A8BKD1_GIALA	11 - 335	2vvq	A	11 - 335	Jmol AstexViewer SPICE
			B	11 - 335	Jmol AstexViewer SPICE
CENPE_HUMAN	12 - 329	1t5c	A	12 - 329	Jmol AstexViewer SPICE
			B	12 - 329	Jmol AstexViewer SPICE
KAR3_YEAST	392 - 723	1f9t	A	392 - 723	Jmol AstexViewer SPICE
		1f9u	A	392 - 723	Jmol AstexViewer SPICE
		1f9v	A	392 - 723	Jmol AstexViewer SPICE
		1f9w	A	392 - 723	Jmol AstexViewer SPICE
			B	392 - 723	Jmol AstexViewer SPICE
		3kar	A	392 - 723	Jmol AstexViewer SPICE
KI13B_HUMAN	11 - 352	3qbj	A	11 - 352	Jmol AstexViewer SPICE
			B	11 - 352	Jmol AstexViewer SPICE
			C	11 - 352	Jmol AstexViewer SPICE
		1ii6	A	24 - 359	Jmol AstexViewer SPICE
			B	24 - 359	Jmol AstexViewer SPICE
		1q0b	A	24 - 359	Jmol AstexViewer SPICE
			B	24 - 359	Jmol AstexViewer SPICE
		1x88	A	24 - 359	Jmol AstexViewer SPICE
			B	24 - 359	Jmol AstexViewer SPICE
			A	24 - 359	Jmol AstexViewer SPICE

wellcome trust sanger institute

PDB entry 3bfn



PDB			UniProt			Pfam family	Colour
Chain	Start	End	ID	Start	End		
A	49	368	KIF22_HUMAN	49	368	Kinesin (PF00225)	

Close window

SUMMARY

- Bioinformatics is computer aided biology.
- Bioinformatics deals with the collection, archiving, organization, and interpretation of a wide range of biological data.
- There are a large number of primary, secondary and tertiary bioinformatics databases.
- The NCBI and EBI are major online bioinformatics service providers.
- Introduced GenBank, RefSeq, UniProt, PDB databases as well as a number of ‘boutique’ databases including PFAM and OMIM.
- Introduced the notion of *controlled vocabularies* and *ontologies.*
- Described the use of ENTREZ and BLAST for searching databases.

HOMEWORK

- ☑ Complete the **initial course questionnaire**:
  http://tinyurl.com/bioinf525-questions

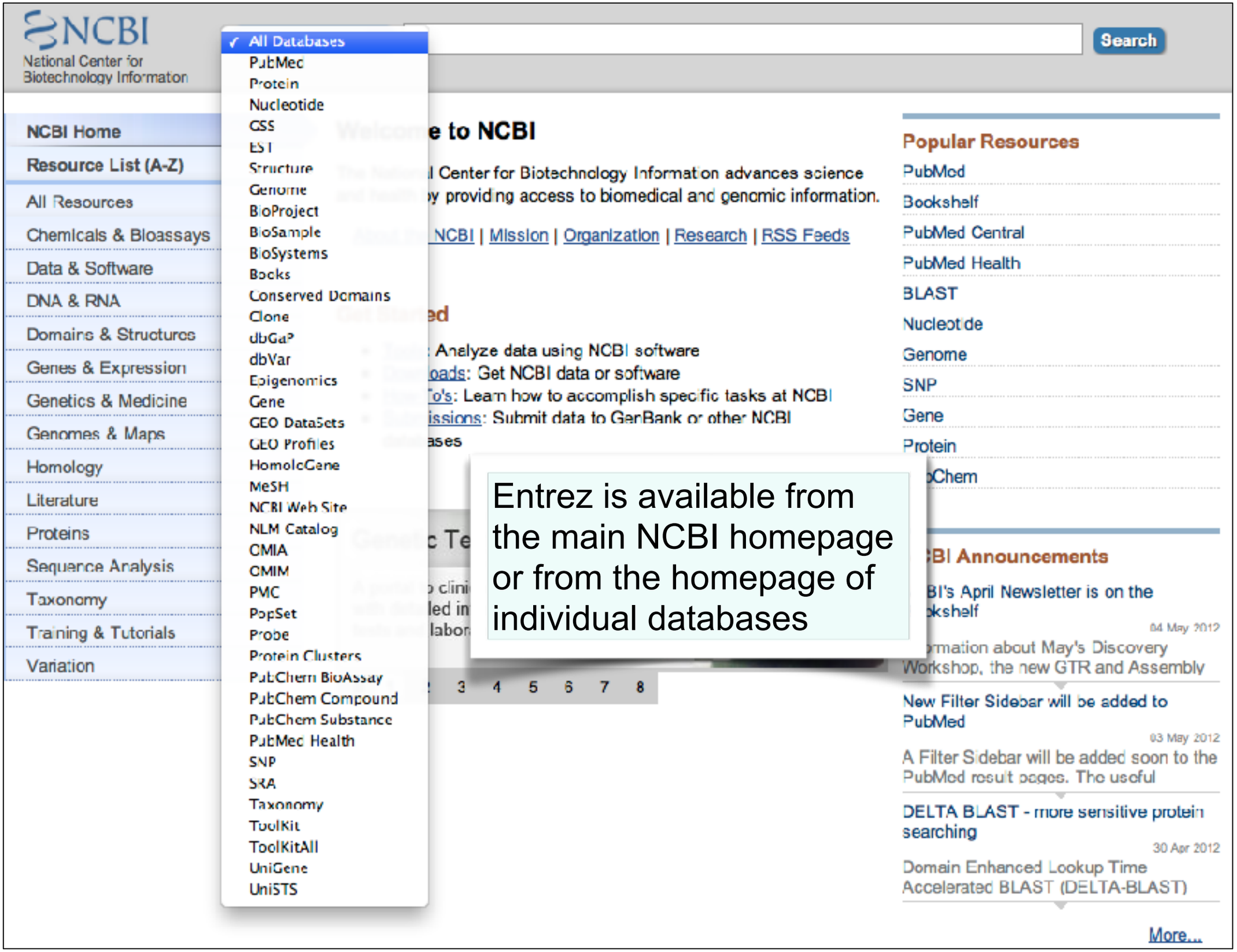
- ☑ Check out the "**Background Reading**" material online:
  PDF1 (bioinformatics review),
  PDF 2 (bioinformatics challenges).

- ☑ Complete the **lecture 1.1 homework questions**:
  http://tinyurl.com/bioinf525-quiz1

ADDITIONAL DATABASES OF NOTE (SLIDES FOR YOUR REFERENCE)

ENTREZ & **BLAST**:
TOOLS FOR SEARCHING AND ACCESSING MOLECULAR DATA AT NCBI

Entrez: Integrated search of NCBI databases



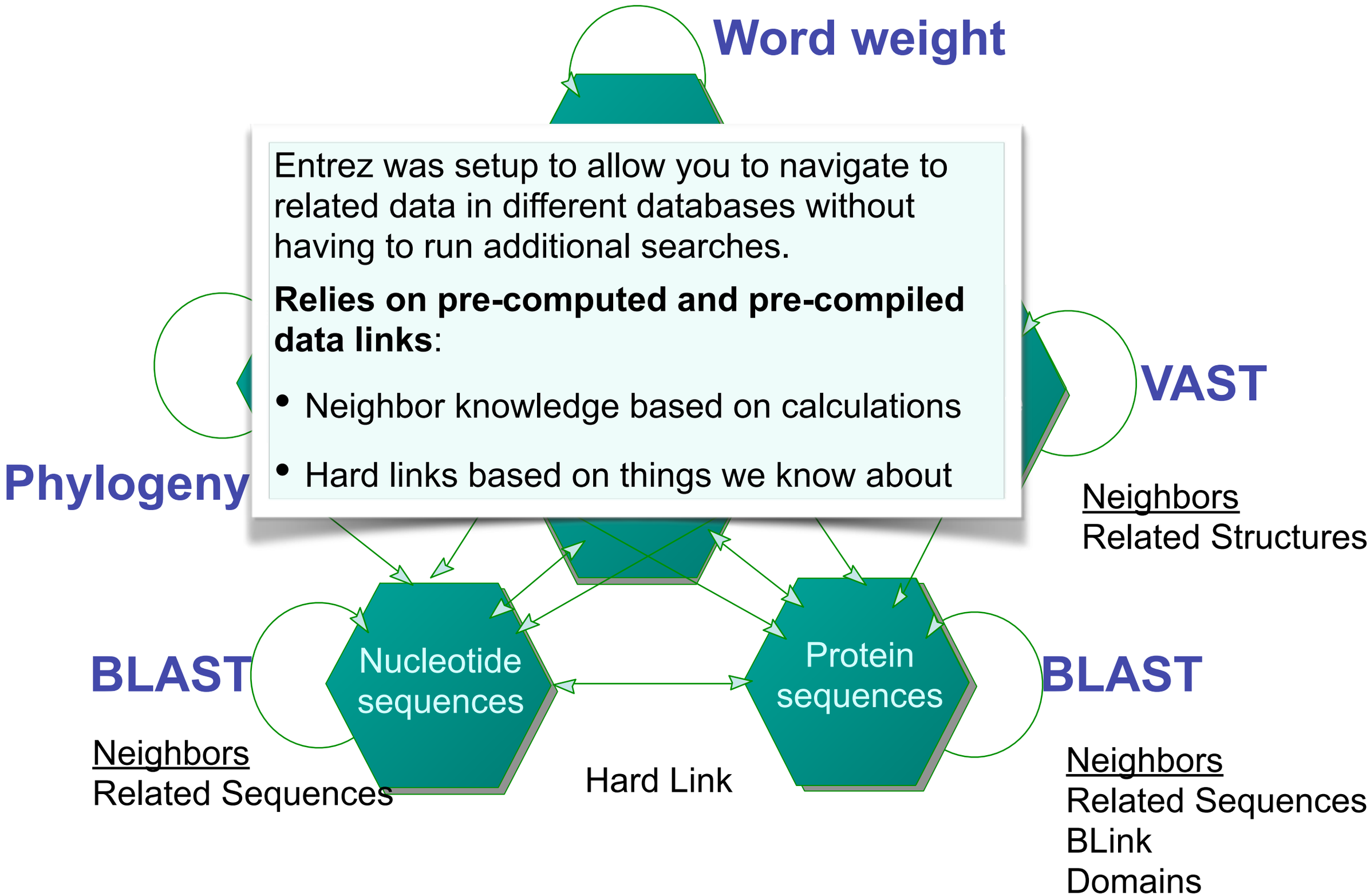
Entrez is available from the main NCBI homepage or from the homepage of individual databases

Entrez: navigating across databases

Entrez was setup to allow you to navigate to related data in different databases without having to run additional searches.

Relies on pre-computed and pre-compiled data links:

- Neighbor knowledge based on calculations
- Hard links based on things we know about



Global Entrez Query: All NCBI Databases

www.ncbi.nlm.nih.gov/gquery/?term=ras

NCBI Resources How To Sign in to NCBI

Search NCBI databases Help

ras Search

About 2,978,774 search results for "ras"

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

The Entrez system: 38 (and counting) integrated databases

Literature

Books		
MeSH	402	ontology used for PubMed indexing
NLM Catalog	223	
PubMed	54,672	
PubMed Central	96,114	full-text journal articles

Health

ClinVar	759	human variations of clinical significance
dbGaP	120	genotype/phenotype interaction studies
GTR	1,879	genetic testing registry

EST	3,985	tag sequences
GEO Profiles	1,622,789	and molecular abundance profiles
HomoloGene	696	homologous gene sets for selected organisms
PopSet	2,254	sequence sets from phylogenetic and population studies
UniGene	4,770	clusters of expressed transcripts

Proteins

Search Results

Nucleotide

Nucleotide | zebrafish creatine kinase | Search

Save search Limits Advanced Help

Display Settings: Summary, 20 per page, Sorted by Default order

Send to:

Found 2324 nucleotide sequences. Nucleotide (35) EST (2289)

Results: 1 to 20 of 35

<< First < Prev Page 1 of 2 Next > Last >>

1. **Danio rerio creatine kinase**, muscle b (ckmb), mRNA
   1,463 bp linear mRNA
   Accession: NM_001105683.1 GI: 157787180
   GenBank FASTA Graphics Related Sequences

2. **Danio rerio** zgc:63663 (zgc:63663), mRNA
   2,478 bp linear mRNA
   Accession: NM_200614.1 GI: 41055386
   GenBank FASTA Graphics Related

3. **Danio rerio creatine kinase**, muscle
   1,552 bp linear mRNA
   Accession: NM_130932.1 GI: 18858426
   GenBank FASTA Graphics Related Sequences

4. **Danio rerio creatine kinase**, mitochondrial 2 (sarcomeric), mRNA (cDNA clone MGC:198091 IMAGE:9039080), complete cds
   1,296 bp linear mRNA
   Accession: BC171364.1 GI: 213624628
   GenBank FASTA Graphics Related Sequences

5. **Danio rerio creatine kinase**, mitochondrial 2 (sarcomeric), mRNA (cDNA clone MGC:172259 IMAGE:8798676), complete cds
   1,400 bp linear mRNA
   Accession: BC154617.1 GI: 159155933
   GenBank FASTA Graphics Related Sequences

Discovery Column
(sort, filter, link)

Filter your results:

- All (35)
- Bacteria (0)
- INSDC (GenBank) (27)
- mRNA (32)
- RefSeq (8)

Manage Filters

Top Organisms [Tree]

- Danio rerio (29)
- Ictalurus furcatus (6)

Find related data

Database: Select

Find items

Search details

```
("Danio rerio"[Organism]
OR zebrafish[All Fields])
AND creatine kinase[All
Fields]
```

Search

See more...

Recent activity

Advanced: Search Builder

Nucleotide Advanced Search Builder

zebrafish[Organism] AND "creatine kinase"[Title]

Clear

Helps build complex fielded queries

Organism | zebrafish | Show index list

AND | Title | "creatine kinase"[Title] | Hide index list

creatine kinase (749)
creatine kinase 1 (6)
creatine kinase 2 (2)
creatine kinase b (30)
creatine kinase b gene (3)
creatine kinase b mrna (3)
creatine kinase b pseudogene 1 (1)
creatine kinase b subunit (1)
creatine kinase brain (43)
creatine kinase chain b (1)

Previous 200
Next 200

Refresh index

AND | All Fields | Show index list

Search or Add to history

Items from search history can be included / combined / modified

History

Clear history

Search	Add to builder	Query	Items found	Time
#7	Add	Search **zebrafish[organism] AND actin[title]**	71	12:41:16
#4	Add	Search **zebrafish actin**	1288	12:40:07
#1	Add	Search **zebrafish creatine kinase**	34	12:39:02

Complex Query Results

Display Settings: Summary, 20 per page, Sorted by Default order

Send to:

Results: 6

1. **Danio rerio creatine kinase**, brain a (ckba), mRNA
   1,481 bp linear mRNA
   Accession: NM_001077163.1 GI: 116004536
   GenBank FASTA Graphics Related Sequences

2. **Danio rerio creatine kinase**, mitochondrial 1 (ckmt1), nuclear gene encoding mitochondrial protein, mRNA

("Danio rerio"[Organism] AND "creatine kinase"[Title]) AND "refseq"[Filter] AND mrna[Filter]

3. 1,552 bp linear mRNA
   Accession: NM_130932.1 GI: 18856426
   GenBank FASTA Graphics Related Sequences

4. **Danio rerio creatine kinase**, mitochondrial 2 (sarcomeric) (ckmt2), nuclear gene encoding mitochondrial protein, mRNA
   1,401 bp linear mRNA
   Accession: NM_200697.1 GI: 41152341
   GenBank FASTA Graphics Related Sequences

5. **Danio rerio creatine kinase**, muscle b (ckmb), mRNA
   1,463 bp linear mRNA
   Accession: NM_001105683.1 GI: 157787180
   GenBank FASTA Graphics Related Sequences

6. **Danio rerio creatine kinase**, brain b (ckbb), mRNA
   1,459 bp linear mRNA
   Accession: NM_173222.1 GI: 27546192
   GenBank FASTA Graphics Related Sequences

Filter your results:

- All (6)
- Bacteria (0)
- INSDC (GenBank) (0)
- mRNA (6)
- RefSeq (6)

Manage Filters

Analyze these sequences

Run BLAST

Find related data

Database: Select

Find items

Search details

```
("Danio rerio"[Organism]
AND "creatine kinase"
[Title]) AND "refseq"
[Filter]
```

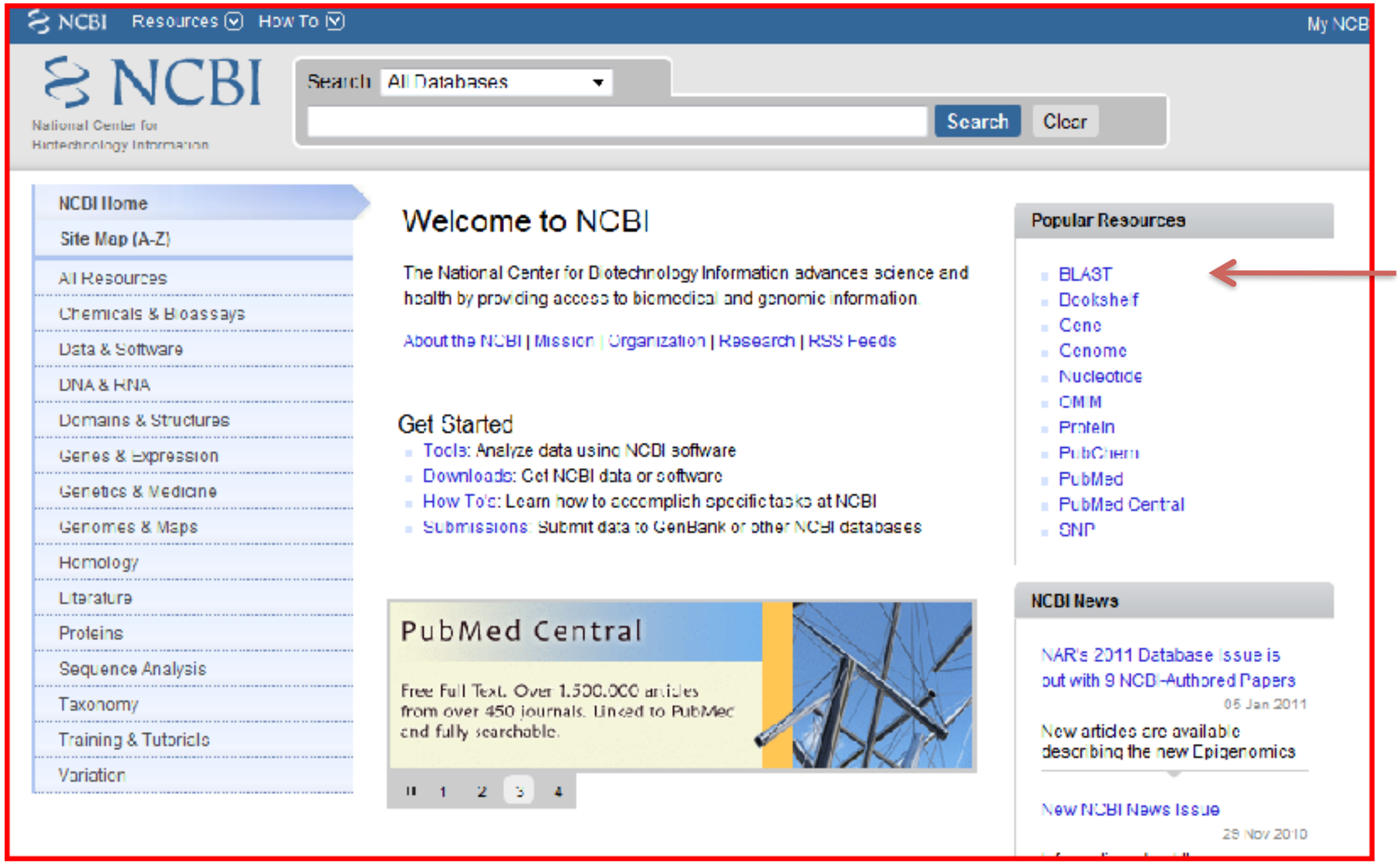
Search

See more...

Recent activity

BLAST is a very important tool available from the NCBI Homepage

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



BLAST – Basic Local Alignment Search Tool

http://blast.ncbi.nlm.nih.gov/Blast.cgi

BLAST
Basic Local Alignment Search Tool
Home | Recent Results | Saved Strategies | Help
My NCBI
[Sign In] [Registe

NCBI BLAST Home

BLAST finds regions of similarity between biological sequences. more...

New Aligning Multiple Protein Sequences? Try the COBALT Multiple Alignment Tool. Go

BLAST Assembled RefSeq Genomes

Choose a species genome to search, or list all genomic BLAST databases

- Human
- Mouse
- Rat
- *Arabidopsis thaliana*
- *Oryza sativa*
- *Bos taurus*
- *Danio rerio*
- *Drosophila melanogaster*
- *Gallus gallus*
- *Pan troglodytes*
- *Microbes*
- *Apis mellifera*

Basic BLAST

Choose a BLAST program to run

nucleotide blast	Search a **nucleotide** database using a **nucleotide** query *Algorithms:* blastn, megablast, discontiguous megablast
protein blast	Search **protein** database using a **protein** query *Algorithms:* blastp, psi-blast, phi-blast
blastx	Search **protein** database using a **translated** nucleotide query
tblastn	Search **translated nucleotide** database using a **protein** query
tblastx	Search **translated nucleotide** database using a **translated nucleotide** query

Specialized BLAST

News

New WGS BLAST page

A new WGS BLAST page allows selection of search sets by organism
Mon, 22 Nov 2010 09:00:00 EST

More BLAST news...

Tip of the Day

How to do Batch BLAST jobs.

BLAST makes it easy to examine a large group of potential gene candidates

More tips...

BLAST performs sequence similarity searches of query sequences *vs* sequence databases. We will cover this in detail in the next lecture.

NCBI Metadatabases

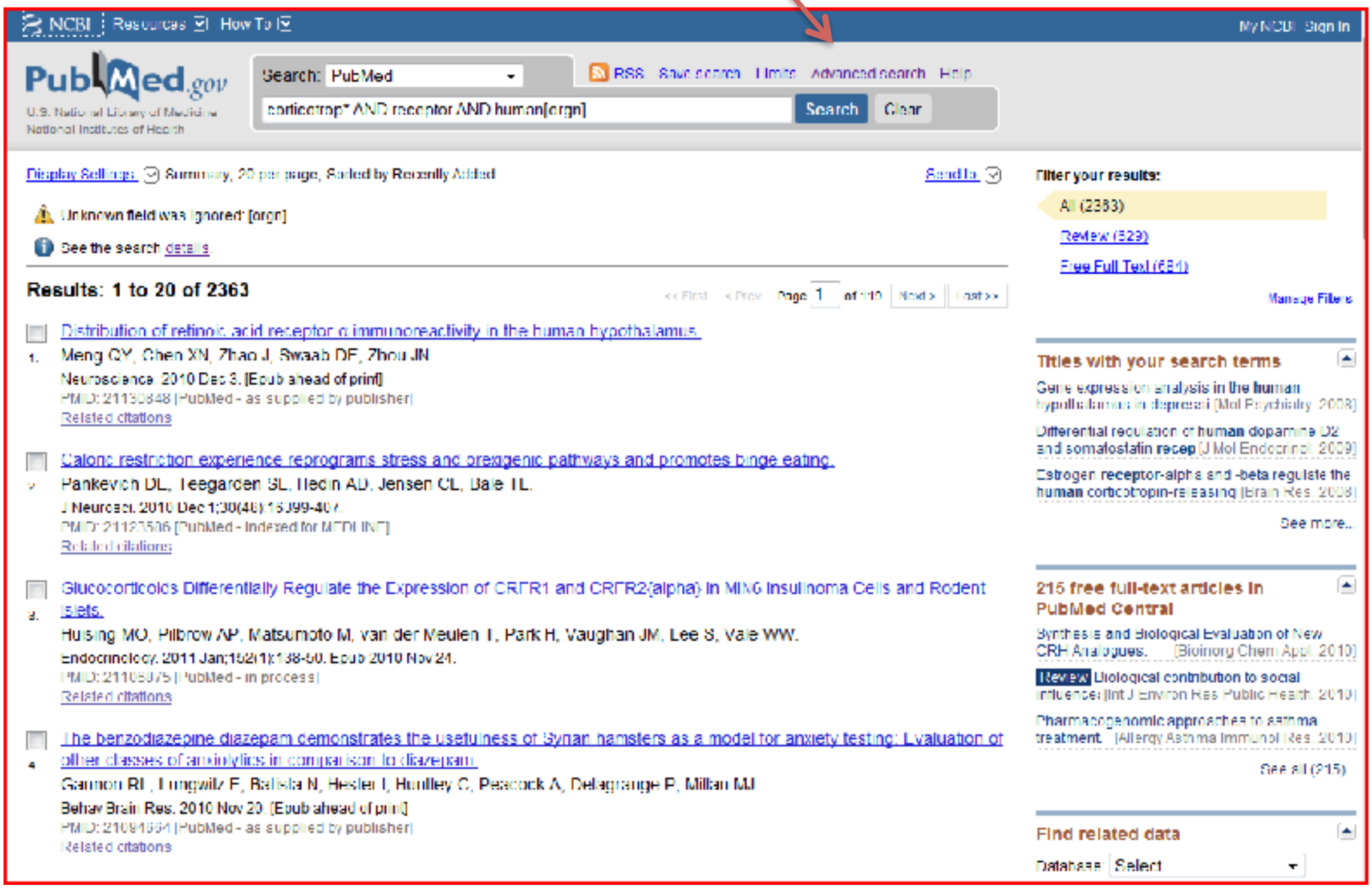
- **Gene**
  - molecular data and literature related to genes
- **HomoloGene**
  - automated collection of homologous genes from selected eukaryotes
- **Taxonomy**
  - access to NCBI data through source organism taxonomic classification
- **PubChem**
  - small organic molecules and their biological activities
- **BioSystems**
  - biochemical pathways and processes linked to NCBI genes, gene products, small molecules, and structures

PubMed

- Curated database of biomedical journal articles
- Data records are annotated with MeSH terms (Medical Subject Headings)
- Contract workers actually read all of the articles and classify them with the MeSH terms
- PubMed entries contain article abstracts
- PubMed Central contains full journal articles, but the majority are not freely re-distributable

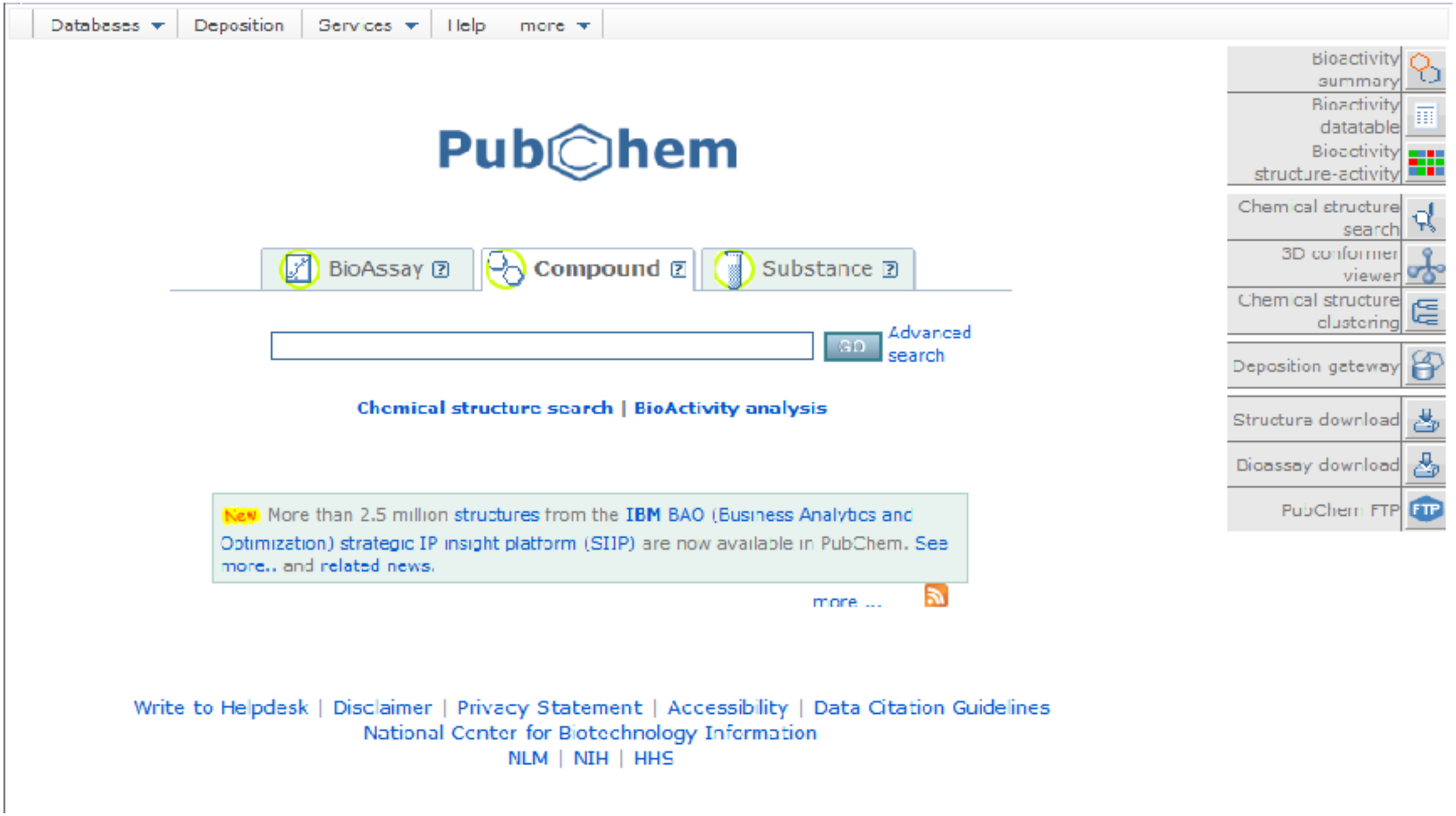
PubMed results

Limits and Advanced search can be used to refine searches



Small molecule databases have been added at NCBI

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



HomoloGene - Homologous genes from different organisms http://www.ncbi.nlm.nih.gov/homologene

NCBI

HomoloGene
Discover Homologs

Help

My NCBI
[Sign In] [Regis

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | OMIM | PMC | Journals | Books

Search HomoloGene for Go Clear

Limits | Preview/Index | History | Clipboard | Details

HomoloGene
Homepage
Query Tips
Build Procedure
FTP site

Genome Resources
Homo sapiens
Mus musculus
Rattus norvegicus
Danio rerio

HomoloGene is a system for automated detection of homologs among the annotated genes of several completely sequenced eukaryotic genomes

HomoloGene Release 65 Statistics

Initial numbers of genes from complete genomes, numbers of genes placed in a homology group, and the numbers of groups for each species.

Species	Number of Genes Input	Number of Genes Grouped	HomoloGene groups
Homo sapiens	19,943*	18,981	18,431
Pan troglodytes	25,096	16,050	15,980
Canis familiaris	19,766	16,708	15,951
Bos taurus	22,049	18,180	16,224
Mus musculus	25,388	21,766	19,005
Rattus norvegicus	21,991	19,229	17,473
Gallus gallus	17,959	13,142	11,905
Danio rerio	26,690*	21,084	14,067
Drosophila melanogaster	13,027*	9,282	7,749
Anopheles gambiae	12,460	8,867	7,541
Caenorhabditis elegans	20,132*	8,678	4,810
Schizosaccharomyces pombe	5,043	3,225	2,935
Saccharomyces cerevisiae	5,880	4,851	4,370
Kluyveromyces lactis	5,335	4,459	4,382
Eremothecium gossypii	4,722	3,928	3,884
Magnaporthe grisea	12,832	7,330	6,399
Neurospora crassa	9,821*	6,287	6,144
Arabidopsis thaliana	[illegible]*	19,961	11,343

What's New

HomoloGene release 65 includes updated annotations for the following species: Homo sapiens (NCBI release 37.2), Danio rerio (NCBI release 4.1), Drosophila melanogaster (NCBI release 9.3) Caenorhabditis elegans (NCBI release 9.1), Arabidopsis thaliana (NCBI release 9.1).

Related Resources

Entrez Genomes

A collection of complete genome sequences that includes more than 1000 viruses and over hundred microbes

- Archaea
- Bacteria
- Eukaryota

Online Mendelian Inheritance in Man – OMIM

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

NCBI

OMIM
Online Mendelian Inheritance in Man

Johns Hopkins University

My NCBI
[Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | OMIM

Search OMIM for Go Clear

Limits | Preview/Index | History | Clipboard | Details

Entrez

OMIM
Search OMIM
Search Gene Map
Search Morbid Map

Help
OMIM Help
How to Link

FAQ
Numbering System
Symbols
How to Print
Citing OMIM
Download

OMIM Facts
Statistics
Update Log

- Enter one or more search terms.
- Use **Limits** to restrict your search by search field, chromosome, and other criteria
- Use **Index** to browse terms found in OMIM records
- Use **History** to retrieve records from previous searches, or to combine searches.

OMIM ® - Online Mendelian Inheritance in Man

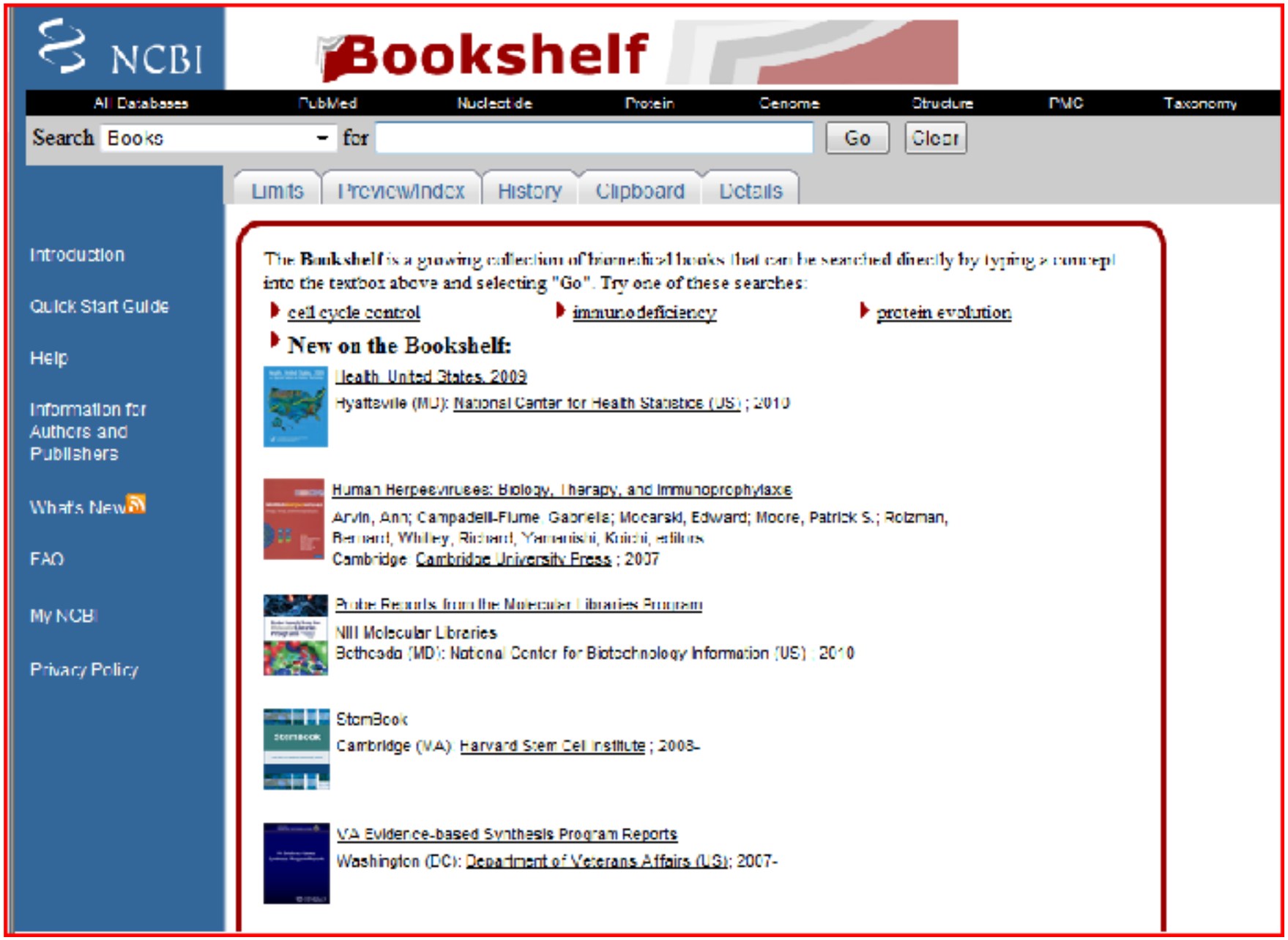
Welcome to OMIM ®, Online Mendelian Inheritance in Man ®. OMIM is a comprehensive, authoritative, and timely compendium of human genes and genetic phenotypes. The full-text, referenced overviews in OMIM contain information on all known mendelian disorders and over 12,000 genes. OMIM focuses on the relationship between phenotype and genotype. It is updated daily, and the entries contain copious links to other genetics resources.

This database was initiated in the early 1960s by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders, entitled Mendelian Inheritance in Man (MIM). Twelve book editions of MIM were published between 1966 and 1998. The online version, OMIM, was created in 1985 by a collaboration between the National Library of Medicine and the William H. Welch Medical Library at Johns Hopkins. It was made generally available on the

OMIM is essentially a set of reviews of human genes, gene function and phenotypes. Includes causative mutations where known.

The NCBI Bookshelf includes many well known molecular biology texts.

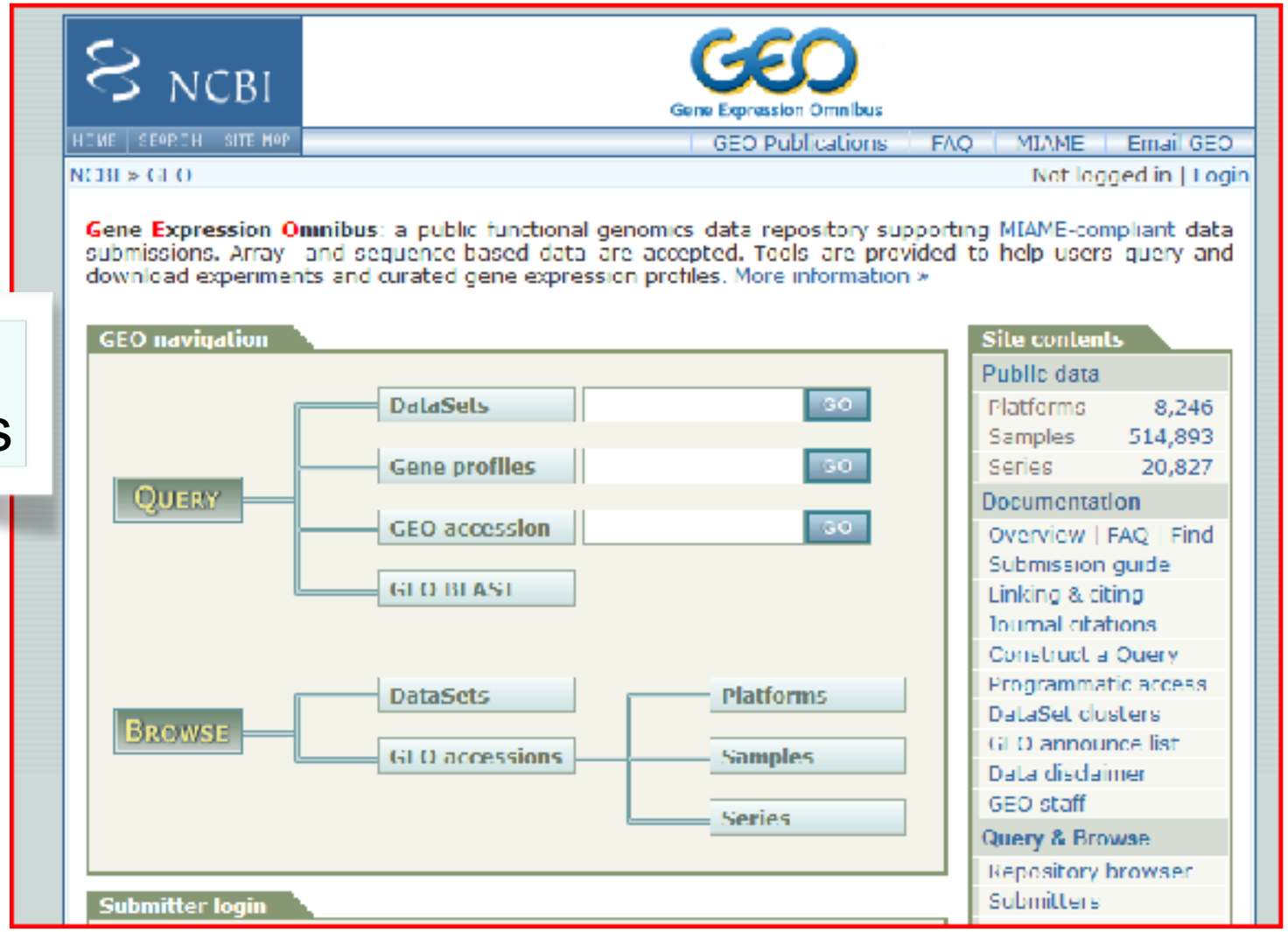
http://www.ncbi.nlm.nih.gov/books/



GEO: Gene Expression Omnibus

- Gene expression data (mostly from microarrays but also RNA-seq data, 2 methods for measuring RNA levels)

Query browse and download data sets



- **Series** - (GSExxx) is an original submitter-supplied record that summarizes a study. May contain multiple individual **Samples** (GSMxxx).

GEO Accession viewer

www.ncbi.nlm.nih.gov/geo/query/acc.cgi?acc=GSE3541

NCBI

HOME | SEARCH | SITE MAP

NCBI > GEO > Accession Displ

Scope: Self Format:

Series GSE3541

Status	Public on
Title	DNA micr type II ep
Organism	Rattus no
Experiment type	Expressio
Summary	Mechanica cellular a unknown. to investi epithelial utero. Significance Analysis of Microarrays (SAM) identified 92 genes differentially expressed by strain. Interestingly, several members of the solute carrier family of amino acid transporters, genes involved in amino acid synthesis and development, and amiloride-sensitive epithelial sodium channel gene were induced by strain. These results were confirmed by quantitative real-time polymerase chain reaction (qRT-PCR). Thus, this study identifies genes induced by strain that may be important for amino acid signaling pathways, protein

Platforms (1) GPL4091 Agilent-014693 Human Genome CGH Microarray 244A (Feature number version)

Samples (4) GSM495808 Aspc1 Cell Line
More... GSM495809 JH39 Xenograft
GSM495810 JH21 Xenograft

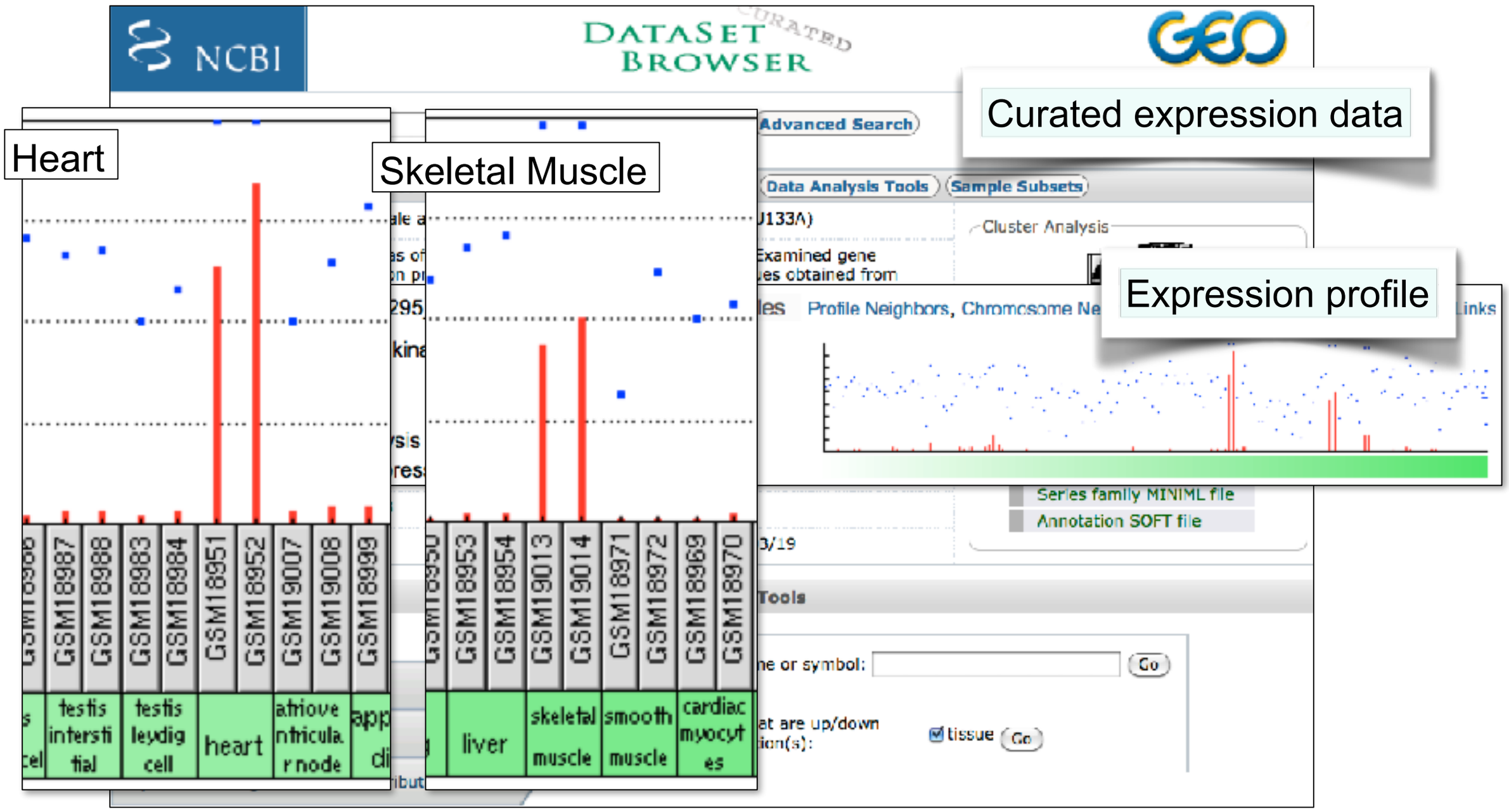
Download family	Format
SOFT formatted family file(s)	SOFT
MINiML formatted family file(s)	MINiML
Series Matrix File(s)	TXT

Supplementary file	Size	Download	File type/resource
GSE19852_RAW.tar	170.7 Mb	(ftp)(http)	TAR (of TXT)

Raw data provided as supplementary file
Processed data included within Sample table

NLM | NIH | GEO Help | Disclaimer | Section 508

- **DataSets** - (GDSxxx) are curated collections of selected Samples that are biologically and statistically comparable



QuickGO is a fast web-based browser of the Gene Ontology and Gene Ontology annotation data

QuickGO

www.ebi.ac.uk/QuickGO/

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  - geneontology.org
  - UniProt-GOA project
  - Web Services

EBI › Databases › QuickGO

QuickGO

QuickGO is a fast web-based browser for Gene Ontology terms and annotations, which is provided by the UniProt-GOA project at the EBI.

Click for example search | Search! | Web Services | Dataset | Term Basket: 0

Search and Filter GO annotation sets

Extensive filters are available from this page to allow the generation of specific subsets of GO annotations, mapped to sequence identifiers of your choice.

Investigate GO slims

GO slims are lists of GO terms that have been selected from the full set of terms available from the Gene Ontology project.

GO slims can be used to generate a focused view of part of the GO, or with annotation data they can be used to see how a set of proteins/genes can be broadly categorized (using annotation data and the relationships that exist between terms in the ontologies).

Further information on GO slims can be found at the GO Consortium web site.

View the history of changes to GO

This page allows you to view the changes to GO, optionally filtered by date, term identifier, or type of change.

QuickGO News

- 19 August 2011 - Changes to the Term Basket
- 14 June 2011 - New term history displays
- 20 April 2011 - Display improvements

QuickGO News Archive

QuickGO Tips

Tutorial

GO annotation in UniProt

An example UniProt entry for hemoglobin beta (HBB_human, P68871) with GO annotation displayed.

Hemoglobin subunit beta - Homo sapiens (Human)

www.uniprot.org/uniprot/P68871

Hemoglobin subunit beta - Homo sapiens (Human)

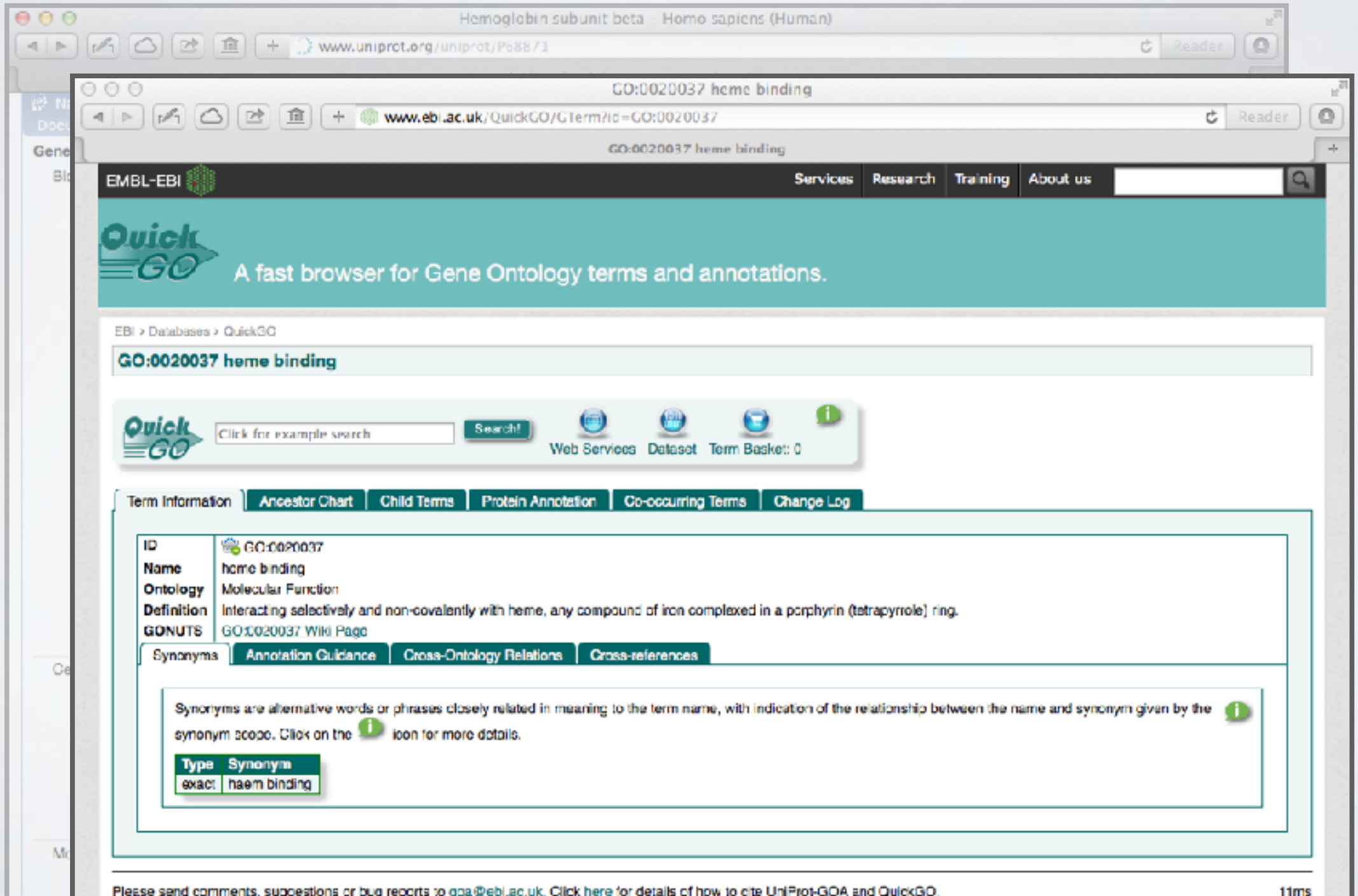
Names · Attributes · General annotation · Ontologies · Interactions · Sequence annotation · Sequences · References · Web links · Cross-refs · Entry info · Documents Customize order

Gene Ontology (GO)

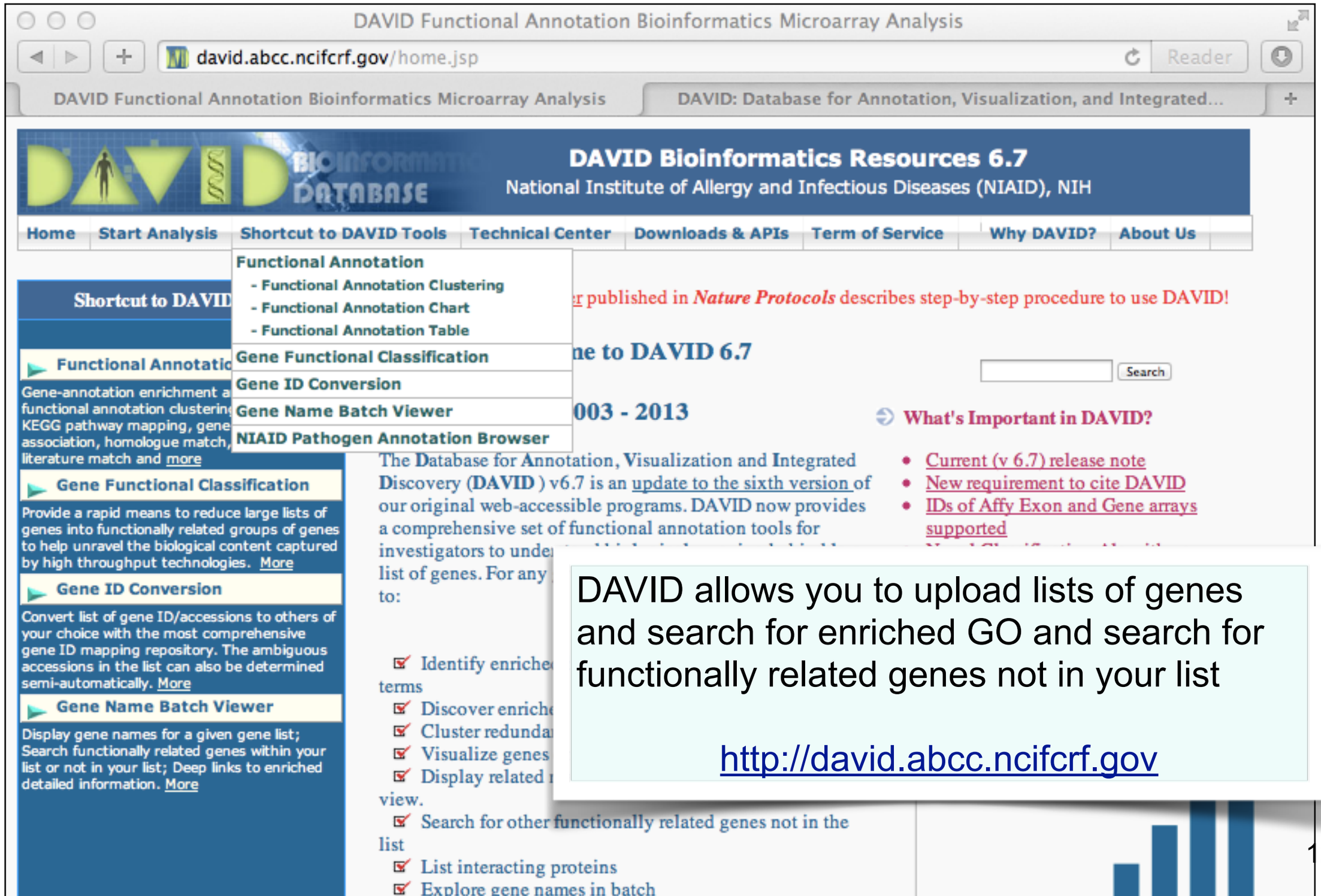
Biological_process	bicarbonate transport Traceable author statement. Source: Reactome blood coagulation Traceable author statement. Source: Reactome hydrogen peroxide catabolic process Inferred from direct assay (PubMed 13740759). Source: BHF-UCL nitric oxide transport Non-traceable author statement (PubMed 8292032). Source: UniProtKB positive regulation of cell death Inferred from direct assay (PubMed 13740759). Source: BHF-UCL positive regulation of nitric oxide biosynthetic process Non-traceable author statement (PubMed 7865123). Source: UniProtKB protein heterooligomerization Inferred from direct assay (PubMed 13740759). Source: BHF-UCL regulation of blood pressure Inferred from electronic annotation. Source: UniProtKB-KW regulation of blood vessel size Inferred from electronic annotation. Source: UniProtKB-KW renal absorption Inferred from mutant phenotype (PubMed 15465030) (PubMed 10974565). Source: UniProtKB small molecule metabolic process Traceable author statement. Source: Reactome
Cellular_component	endocytic vesicle lumen Traceable author statement. Source: Reactome extracellular region Traceable author statement. Source: Reactome haptoglobin-hemoglobin complex Inferred from direct assay (PubMed 13740759). Source: BHF-UCL hemoglobin complex Non-traceable author statement (Ref.33) (Ref.72). Source: UniProtKB
Molecular_function	heme binding Inferred from electronic annotation. Source: InterPro hemoglobin binding

GO annotation in UniProt

An example UniProt entry for hemoglobin beta (HBB_human, P68871) with GO annotation displayed.



DAVID: a online tool for assessing GO term enrichment in gene lists



DAVID allows you to upload lists of genes and search for enriched GO and search for functionally related genes not in your list

http://david.abcc.ncifcrf.gov

Example output: enriched functions from GO

DAVID: Database for Annotation, Visualization, and Integrat...ID); Science Applications International Corporation (SAIC)

david.abcc.ncifcrf.gov/chartReport.jsp?annot=25

DAVID: Functional Annotation Result Summary | Database for Annotation, Visualization, and Integrated Discov...

DAVID Bioinformatics Resources 6.7
National Institute of Allergy and Infectious Diseases (NIAID), NIH

Functional Annotation Chart

Help and Manual

Current Gene List: List_1
Current Background: Homo sapiens
14 DAVID IDs
⊞ **Options**

Rerun Using Options | Create Sublist

10 chart records — Download File

Sublist	Category	Term	RT	Genes	Count	%	P-Value	Benjamini
☐	GOTERM_BP_FAT	regulation of osteoclast differentiation	RT		2	14.3	2.1E-2	9.9E-1
☐	GOTERM_BP_FAT	response to organic substance	RT		4	28.6	2.9E-2	9.6E-1
☐	GOTERM_BP_FAT	regulation of myeloid leukocyte differentiation	RT		2	14.3	3.9E-2	9.5E-1
☐	GOTERM_BP_FAT	positive regulation of transcription from RNA polymerase II promoter	RT		3	21.4	4.8E-2	9.4E-1
☐	GOTERM_BP_FAT	regulation of myeloid cell differentiation	RT		2	14.3	6.5E-2	9.5E-1
☐	GOTERM_BP_FAT	cartilage development	RT		2	14.3	6.9E-2	9.3E-1
☐	GOTERM_BP_FAT	positive regulation of transcription, DNA-dependent	RT		3	21.4	7.5E-2	9.2E-1
☐	GOTERM_BP_FAT	positive regulation of RNA metabolic process	RT		3	21.4	7.6E-2	8.9E-1
☐	GOTERM_BP_FAT	response to protein stimulus	RT		2	14.3	9.8E-2	9.3E-1
☐	GOTERM_BP_FAT	positive regulation of transcription	RT		3	21.4	1.0E-1	9.1E-1

8 gene(s) from your list are not in the output.

Please cite *Nature Protocols 2009; 4(1):44* & *Genome Biology 2003; 4(5):P3* within any publication that makes use of any methods inspired by **DAVID**.