# EBI web resources II: Ensembl and InterPro

Yanbin Yin Spring 2013

# Outline

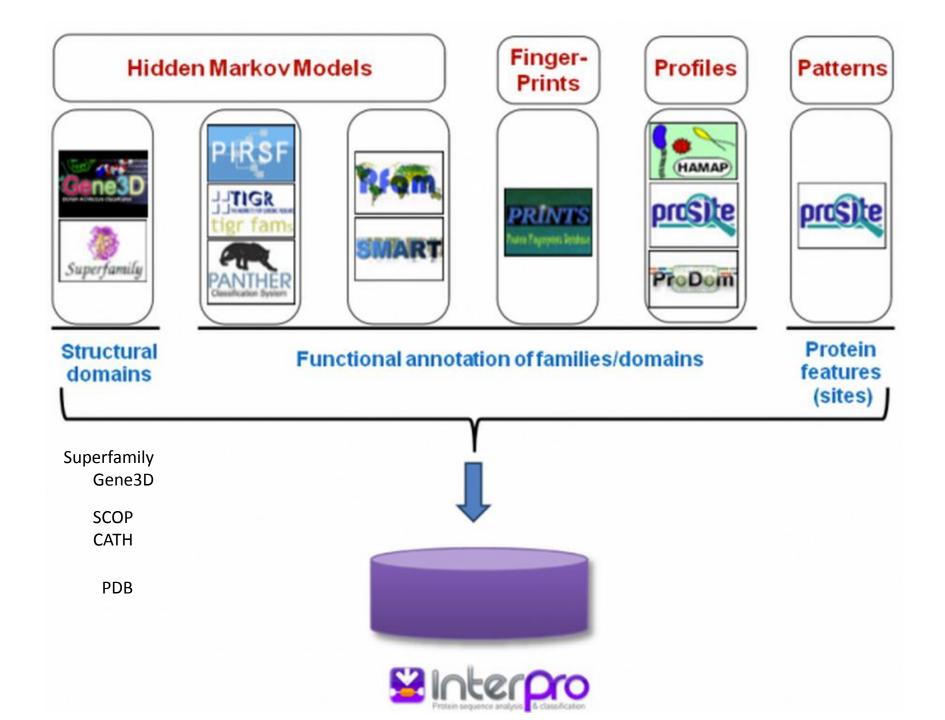
- Intro to genome annotation
- Protein family/domain databases
   InterPro, Pfam, Superfamily etc.
- Genome browser
  - Ensembl
- Hands on Practice

# Genome annotation

- Predict genes (where are the genes?)
  - protein coding
  - RNA coding

- Function annotation (What are the genes?)
  - Search against UniProt or NCBI-nr (GenPept)
  - Search against protein family/domain databases
  - Search against Pathway databases

Function vocabularies defined in Gene Ontology



# InterPro components

1. CATH/Gene3D University College, London, UK 2. PANTHER University of Southern California, CA, USA Protein Information Resource, Georgetown University, USA 3. PIRSF Pfam Wellcome Trust Sanger Institute, Hinxton, UK University of Manchester, UK 5. PRINTS 6. ProDom **PRABI Villeurbanne, France** 7. PROSITE Swiss Institute of Bioinformatics (SIB), Geneva, Switzerland SMART EMBL, Heidelberg, Germany 9. SUPERFAMILY University of Bristol, UK 10. TIGRFAMs J. Craig Venter Institute, Rockville, MD, US Swiss Institute of Bioinformatics (SIB), Geneva, Switzerland **11. HAMAP** 

# CDD components

Pfam, SMART, TIGRFAM, COG, KOG, PRK, CD, LOAD Each InterPro entry is assigned one of a number of types which tell you what you can infer when a protein matches the entry. The entry types are:



# Family

A protein family is a group of proteins that share a common evolutionary origin reflected by their related functions, similarities in sequence, or similar primary, secondary or tertiary structure. A match to an InterPro entry of this type indicates membership of a protein family.

# Domain

Domains are distinct functional, structural or sequence units that may exist in a variety of biological contexts. A match to an InterPro entry of this type indicates the presence of a domain.



# Repeat

A match to an InterPro entry of this type identifies a short sequence that is typically repeated within a protein.



# Site

A match to an InterPro entry of this type indicates a short sequence that contains one or more conserved residues. The type of sites covered by InterPro are active sites, binding sites, post-translational modification sites and conserved sites.

# **Protein Classification**

Nearly all proteins have structural similarities with other proteins and, in some of these cases, share a common evolutionary origin. Proteins are classified to reflect both structural and evolutionary relatedness. Many levels exist in the hierarchy, but the principal levels are family, superfamily and fold, described below.

# Family: Clear evolutionarily relationship

Proteins clustered together into families are clearly evolutionarily related. Generally, this means that pairwise residue identities between the proteins are 30% and greater.

## Superfamily: Probable common evolutionary origin

Proteins that have low sequence identities, but whose structural and functional features suggest that a common evolutionary origin is probable are placed together in superfamilies.

## **Fold:** Major structural similarity

Proteins are defined as having a common fold if they have the same major secondary structures in the same arrangement and with the same topological connections. Different proteins with the same fold often have peripheral elements of secondary structure and turn regions that differ in size and conformation. Proteins placed together in the same fold category may not have a common evolutionary origin: the structural similarities could arise just from the physics and chemistry of proteins favoring certain packing arrangements and chain topologies.

http://scop.mrc-lmb.cam.ac.uk/scop/intro.html

Structural Classification of Proteins



Welcome to **SCOP**: Structural Classification of Proteins. **1.75 release** (June 2009)

38221 PDB Entries. 1 Literature Reference. 110800 Domains. (excluding nucleic acids and theoretical models). Folds, superfamilies, and families <u>statistics here</u>. <u>New folds superfamilies families</u>. List of obsolete entries and their replacements.



Authors. Alexey G. Murzin, John-Marc Chandonia, Antonina Andreeva, Dave Howorth, Loredana Lo Conte, Bartlett G. Ailey, Steven E. Brenner, Tim J. P. Hubbard, and Cyrus Chothia. scop@mrc-lmb.cam.ac.uk

Reference: Murzin A. G., Brenner S. E., Hubbard T., Chothia C. (1995). SCOP: a structural classification of proteins database for the investigation of sequences and structures. J. Mol. Biol. 247, 536-540. [PDF]

Recent changes are described in: Lo Conte L., Brenner S. E., Hubbard T.J.P., Chothia C., Murzin A. (2002). SCOP database in 2002: refinements accommodate structural genomics. *Nucl. Acid Res.* 30(1), 264-267. [PDF],

Andreeva A., Howorth D., Brenner S.E., Hubbard T.J.P., Chothia C., Murzin A.G. (2004). SCOP database in 2004: refinements integrate structure and sequence family data. <u>Nucl. Acid Res. 32:D226-D229</u>. [PDF], and

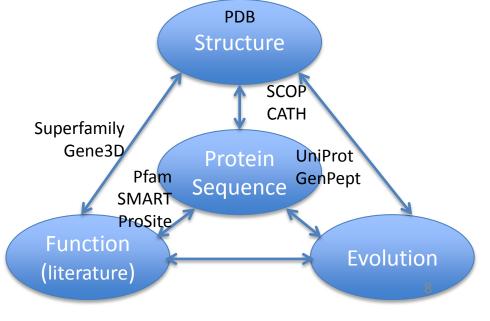
Andreeva A., Howorth D., Chandonia J.-M., Brenner S.E., Hubbard T.J.P., Chothia C., Murzin A.G. (2007). Data growth and its impact on the SCOP database: new developments. *Nucl. Acids Res.* 2008 36: D419-D425; doi:10.1093/nar/gkm993 [PDF].

#### **Postdoc Wanted**

• Want to help us design and build the next generation of SCOP and ASTRAL? Get more details and apply here.

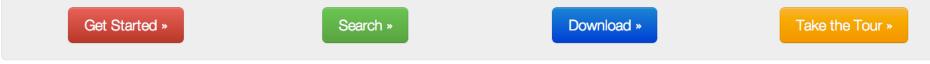
#### Access methods

- Enter scop at the top of the hierarchy
- Keyword search of SCOP entries
- <u>SCOP parseable files</u>
- All SCOP releases and reclassified entry history
- pre-SCOP preview of the next release
- SCOP domain sequences and pdb-style coordinate files (ASTRAL)
- Hidden Markov Model library for SCOP superfamilies (SUPERFAMILY)
- Structural alignments for proteins with non-trivial relationships (SISYPHUS)



# CATH / Gene3D

16 million protein domains classified into 2,626 superfamilies



# What's New?

The CATH website has recently undergone a big overhaul. We really hope you find the new pages more useful, easier to use and quicker to load. Please get in touch and let us know what you think.

# Searching CATH

- Search by ID / keyword
- Search by FASTA sequence
- Search by PDB structure

# Example pages

- PDB "1dan"
- Domain "1cukA01"
- Relatives of "1cukA01"
- Superfamily "HUPs"
- Functional Family
- FunFam Alignment
- Search for "enolase"
- Superfamily Comparison

# Latest News



"Using CATH-Gene3D to study the evolution of your protein and find its function" - Prof Orengo presents the new CATH website at ECCB

# Latest Release

CATH v3.5 bas	ed on PDB dated September 20, 2011
173,536	CATH Domains
2,626	CATH Superfamilies
51,334	PDBs

Gene3D v11 released March 18, 2012		
1,639	Cellular Genomes	
1,016	Viral Genomes	
14,963,305	Protein Sequences	
16,297,076	CATH Domain Predictions	

Depth	Letter	Name	Clustering criteria
1		Class	Secondary structure content
2		Architecture	General spatial arrangement of secondary structures
3		Topology	Spatial arrangement and connectivity of secondary structures (fold)
4		Homologous Superfamily	Manual curation of evidence of evolutionary relationship (at least two criteria
5		Sequence Family (S35)	>= 35% sequence similarity
6		Orthologous Family (S60) *	>= 60% sequence similarity
7		âLikeâ domain (S95) *	>= 95% sequence similarity
8		Identical domain (S100)	100% sequence similarity
9		Domain counter	Unique domains

fold ~ class – superfamily ~ clan – family – subfamily – domain sequence

# Hands on exercise 1: search against protein family databases

# Google "interpro"

EBI > Databases > InterPro

**Release notes** Training & tutorials About InterPro FAOs Download Contact Home

#### What is InterPro?



InterPro provides functional analysis of proteins by classifying them into families and predicting domains and important sites. We combine protein signatures from a number of member databases into a single searchable resource, capitalising on their individual strengths to produce a powerful integrated database and diagnostic tool. more



#### DOCUMENTATION

About InterPro: core concepts, update frequency, how to cite, team and consortium members.

FAOs: what are entry types and why are they important, interpreting results, downloading InterPro?

#### PROTEIN FOCUS

#### Are we really related? The Rad9/Ddc1 family



Protein family classification is often achieved using computerised multiple

protein sequence alignment and structural analysis. However, it's

#### PUBLICATIONS

InterPro in 2011: new developments in the family and domain prediction database



A recently published paper describing new developments with the InterPro database (Nucleic



New features include:

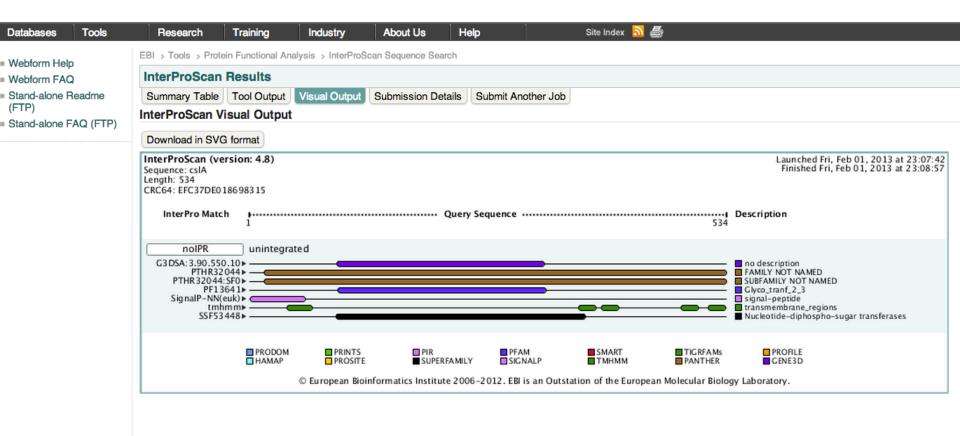
- An update to PIRSF (2.82)
- from the PANTHER, PIRSF, Pfam

announce the release of InterProScan 5RC4: the fourth release candidate of InterProScan version 5.

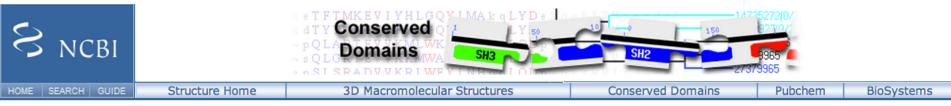
Read documentation

Feedback

We are delighted to announce that the new InterPro website is available as a



# Google "NCBI CDD search"



#### Search for <u>Conserved Domains</u> within a protein or coding nucleotide sequence

NEW! Use Batch CD-search to submit multiple query proteins at once!	OPTIONS
Enter <b>protein</b> or <b>nucleotide</b> query as accession, gi, or sequence in <u>FASTA format</u> >AT5G22740.1 AT5G22740.1 cslA MDGVSPKFVLPETFDGVRMEITGQLGMIWELVKAPVIVPLLQLAVYICLLMSVMLLCERVYMGIVIVLVKLFWKKPDKRY KFEPIHDDELGSSNFPVVLVQIPMENEREVYKLSIGAACGLSWPSDRLVIQVLDDSTDPTVKQMVEVECQRWASKGINI RYQIRENRVGYKAGALKEGLKRSYVKHCEYVVIFDADFQPEPDFLRRSIPFLMHNPNIALVQARWRFVNSDECLLTRMQE MSLDYHFTVEQEVGSSTHAFFGFNGTAGIWRIAAINEAGGWKDRTTVEDMDLAVRASLRGWKFLYLGDLQVKSELPSTFR AFRFQQHRWSCGPANLFRKMVMEIVRNKKVRFWKKVYVIYSFFFVRKIIAHWVTFCFYCVVLPLTILVPEVKVPIWGSVY IPSIITILNSVGTPRSIHLLFYWILFENVMSLHRTKATLIGLFEAGRANEWVVTAKLGSGQSAKGNTKGIKRFPRIFKLP DRLNTLELGFAAFLFVCGCYDFVHGKNNYFIYLFLQTMSFFISGLGWIGTYVPS*	Search against database ?: CDD v3.08 - 43334 PSSMs Expect Value ? threshold: 0.01 Apply low-complexity filter ? Force live search ? Maximum number of hits ? 500 Result mode • Concise ? Full ?
Submit Reset	

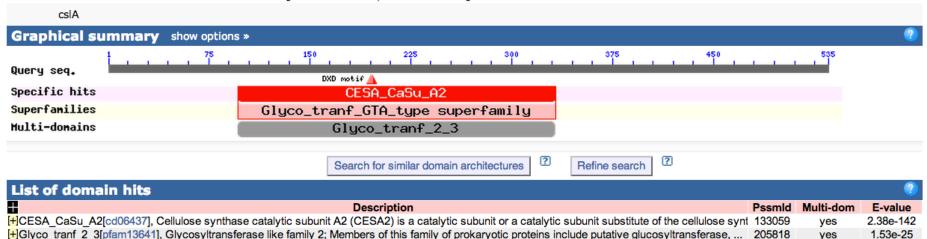
Retrieve previous CD-search	
Request ID:	Retrieve
References: Marchler-Bauer A et al. (2013), "CDD: conserved domains and protein three-dimensional structure. Marchler-Bauer A et al. (2011), "CDD: a Conserved Domain Database for the functional annotation of Marchler-Bauer A, Bryant SH (2004), "CD-Search: protein domain annotations on the fly.", Nucleic	of proteins.", Nucleic Acids Res.39(D)225-9.

Help | Disclaimer | Write to the Help Desk NCBI | NLM | NIH



## Conserved domains on [AT5G22740.1|AT5G22740.1]

View full result



#### **References:**

💯 Marchler-Bauer A et al. (2013), "CDD: conserved domains and protein three-dimensional structure.", Nucleic Acids Res.41(D1)348-52.

💯 Marchler-Bauer A et al. (2011), "CDD: a Conserved Domain Database for the functional annotation of proteins.", Nucleic Acids Res.39(D)225-9.

💯 Marchler-Bauer A, Bryant SH (2004), "CD-Search: protein domain annotations on the fly.", Nucleic Acids Res.32(W)327-331.

Help | Disclaimer | Write to the Help Desk NCBI | NLM | NIH Google "Pfam"

You will see two pfam sites: Sanger pfam and Janellia pfam



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#### Pfam 26.0 (November 2011, 13672 families)

The Pfam database is a large collection of protein families, each represented by **multiple sequence** alignments and hidden Markov models (HMMs). <u>More...</u>

<b>QUICK LINKS</b>	YOU CAN FIND DATA IN PFAM IN VARIOUS WAYS	
SEQUENCE SEARCH	Analyze your protein sequence for Pfam matches	
VIEW A PFAM FAMILY	View Pfam family annotation and alignments	
VIEW A CLAN	See groups of related families	
VIEW A SEQUENCE	Look at the domain organisation of a protein sequence	
VIEW A STRUCTURE	Find the domains on a PDB structure	
<b>KEYWORD SEARCH</b>	Query Pfam by keywords	
JUMP TO	enter any accession or ID Go Example	
	Enter any type of accession or ID to jump to the page for a Pfam family or clan, UniProt sequence, PDB structure, etc.	
	Or view the help pages for more information	



Search Pfam			0 architectures	o sequences	0 interactions	0 species	0 structures
Sequence	Sequence search						
Batch search Keyword		sequence of interest. Paste your protein sec	quence into the box belo	ow, to have it search	ed for matching Pfam	families. <u>More</u>	
Functional similarity	Sequence	1		]			
Domain architecture							
DNA sequence							
Taxonomy							
Jump to 🕸							
enter ID/acc Go	Cut-off	Gathering threshold					
		• Use E-value					
	E-value	1.0					
	Search for PfamBs	Note that we search only the 20,000 largest Pfam-	-B families				
		Submit Reset Example					

Questions or comments: pfam@janelia.hhmi.org Howard Hughes Medical Institute



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## Sequence search results

**Show** the detailed description of this results page.

We found **1** Pfam-A match to your search sequence (all significant). You did not choose to search for Pfam-B matches.



**Show** the search options and sequence that you submitted.

<u>Return</u> to the search form to look for Pfam domains on a new sequence.

#### Significant Pfam-A Matches

Show or hide all alignments.

	Family	Description	Entry	Clan	En
	ганну	Description	type	Cian	Star
	<u>Glyco_tranf_2_3</u>	Glycosyltransferase like family 2	Domain	<u>CL0110</u>	97
#HMM	vavvvptlneddvlarvl	esilaldy.aprlevivvvdgsdaetldvaeelaaayp.dvrvrvvvrprnpgptg	aralnealqaik	.sdlvlllDaDsvv	dpdtlrr1
#МАТСН	v v++p +ne +v+ ++	+++ l + ++rl + v++d + t++ e +++ + +++++ + r++ +++}	<pre>ka+al+e+l++</pre>	+++v+++DaD +	+pd+lrr
#PP	89**********	*****99777888888888555.55665444455544435567777778888888**	**********6555	9******	* * * * * * * *
#SEQ	VLVQIPMFNEREVYKLSI	GAACGLSWpSDRLVIQVLDDST-DPTVKQMVEVECQRWaSKGINIRYQIRENRVGYA	<b>KAGALKEGLKRSYv</b> k	hCEYVVIFDADFQP	EPDFLRRS

Questions or comments: pfam@janelia.hhmi.org Howard Hughes Medical Institute



## What is InterPro?



InterPro provides functional analysis of proteins by classifying them into families and predicting domains and important sites. We combine protein signatures from a number of member databases into a single searchable resource, capitalising on their individual strengths to produce a powerful integrated database and diagnostic tool. more

Text cellulose synthase	<sup>9</sup> Search
FASTA         Sequence	Search For additional options, please use InterProScan.

#### DOCUMENTATION

About InterPro: core concepts, update frequency, how to cite, team and consortium members.

FAQs: what are entry types and why are they important, interpreting results, downloading InterPro?

#### PROTEIN FOCUS

Are we really related? The Rad9/Ddc1 family



Protein family classification is often achieved using computerised multiple protein sequence alignment

and structural analysis. However, it's not always straightforward to define a

#### PUBLICATIONS

InterPro in 2011: new developments in the family and domain prediction database



A recently published paper describing new developments with the InterPro database (*Nucleic Acids Research*, 20129 Vol. EBI > Databases > InterPro



#### Cellulose synthase, subunit A (IPR003919)

... with strains lacking cellulose synthase activity. Nucleotide sequence analysis showed the cellulose synthase operon... of similarit

Overview	E Family	
Proteins matched (1195)	Cellulose synthase (IPR005150)	Contributing sig
Domain organisation (18)		Signatures from I member database
Pathways & interactions	Short name: Cellulose_synth	construct an entr
Species	Family relationships	<b>Pfam</b> ■ 团 PF03552 (Cellu
Structures		
Related resources	None.	
References (2)	Description	

Cellulose, an aggregate of unbranched polymers of beta-1,4-linked glucose residues, is the major component of wo paper, and is synthesized by plants, most algae, some bacteria and fungi, and even some animals. The genes that s cellulose in higher plants differ greatly from the well-characterised genes found in Acetobacter and Agrobacterium s correctly designated as "cellulose synthase catalytic subunits", plant cellulose synthase (CesA) proteins are integral proteins, approximately 1,000 amino acids in length. There are a number of highly conserved residues, including se shown to be necessary for processive glycosyltransferase activity [I PubMed: 8901635].

## GO terms

<b>Biological Process:</b>	GO:0030244 cellulose biosynthetic process
Molecular Function:	♂ GO:0016760 cellulose synthase (UDP-forming) activity
Cellular Component:	♂ GO:0016020 membrane

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25 architectures

G.AIE GTA.E GSMYE

1124 sequences

4

0 interactions



0 structur

Ē

128 species

# Family: Cellulose\_synt (PF03552)

Summary	Summary: Cellulose synthase				
Domain organisation					
Clan	Pfam includes annotations and additional family information from a range of different sources. These sources can be accessed via the tabs below.				
Alignments	No Wikipedia article Pfam Interpro				
HMM logo					
Trees	This tab holds the annotation information that is stored in the Pfam database. As we move to using Wikipedia as our main source of annotation, the contents of this tab will gradually replaced by the Wikipedia tab.				
Curation & model	Cellulose synthase Add annotation				
Species	centrose synthase war antotator				
Interactions	Cellulose, an aggregate of unbranched polymers of beta-1,4-linked glucose residues, is the major component of wood and thus paper, and is synthesised by plants, most a some bacteria and fungi, and even some animals. The genes that synthesise cellulose in higher plants differ greatly from the well-characterised genes found in Acetobacter				
Structures	Agrobacteria and fungi, and even some animals. The genes that synthesise cellulose in higher plants differ greatly from the well-characterised genes found in Acetobacter Agrobacterium sp. More correctly designated as 'cellulose synthase catalytic subunits', plant cellulose synthase (CesA) proteins are integral membrane proteins, approxima 1,000 amino acids in length. There are a number of highly conserved residues, including several motifs shown to be necessary for processive glycosyltransferase activity [1]				
Jump to 🌵 👘	Literature references				
enter ID/acc Go	1. Pear JR, Kawagoe Y, Schreckengost WE, Delmer DP, Stalker DM; , Proc Natl Acad Sci U S A 1996;93:12637-12642.: Higher plants contain homologs of the bacterial celA genes encoding the catalytic subunit of cellulose synthase. PUBMED:8901635 대				
	2. Richmond T; , Genome Biol 2000;1:1-6.: Higher plant cellulose synthases. PUBMED:11178255 과				
	Clan				
	This family is a member of clan <b><u>GT-A</u></b> ( <u>CL0110</u> ), which has a total of <u><b>44</b> members</u> .				
	External database links				
	PANDIT: PF03552 @				
	Pseudofam: PF03552 d <sup>2</sup>				
	SYSTERS: <u>Cellulose_synt</u> 삼				

# http://supfam.cs.bris.ac.uk/SUPERFAMILY/

Superfamily 1.75

HMM library and genome assignments server

Search SUPERFAMILY

#### Home

#### SEARCH

Keyword search

Sequence search

#### BROWSE

Organisms

---- Taxonomy

SCOP

SCOF

Hierarchy

- Ontologies
- .... <u>GO</u>
- ---- <u>EC</u>
- ----- Phenotype

#### TOOLS

Compare genomes Phylogenetic trees Web services Downloads SUPERFAMILY 2+1 +28 Recommend this on Google

Follow @SUPERFAMILY

SUPERFAMILY is a database of structural and functional annotation for all proteins and genomes.

The SUPERFAMILY annotation is based on a collection of **hidden Markov models**, which represent structural prodomains at the <u>SCOP</u> superfamily level. A superfamily groups together domains which have an evolutionary relat The annotation is produced by scanning protein sequences from over <u>2,478 completely sequenced genomes</u> ac hidden Markov models.

For each protein you can:

- Submit sequences for <u>SCOP classification</u>
- View domain organisation, sequence alignments and protein sequence details

#### For each **genome** you can:

- Examine superfamily assignments, phylogenetic trees, domain organisation lists and networks
- Check for over- and under-represented superfamilies within a genome

#### For each **superfamily** you can:

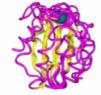
- Inspect SCOP classification, functional annotation, Gene Ontology annotation, InterPro abstract and genome assignments
- Explore taxonomic distribution of a superfamily across the tree of life

All annotation, models and the database dump are freely available for download to everyone. Description cont.

Jump to [ SUPERFAMILY description · Recent news ]

ABOUT

Superfamily 1.75



HMM library and genome assignments server

Search SUPERFAMILY

Home > Assign SCOP domains

#### SEARCH

Keyword search

# Search Sequences for SCOP domains

Sequence search Assign SCOP domains to your sequences using the SUPERFAMILY hidden Markov models.

BROWSE Organisms	Amino acid sequence Split on stop codons (nucleotide only): Yes   No O
<u>Taxonomy</u>	Sequences:
Statistics	>AT5G22740.1 AT5G22740.1 cs1A
SCOP	MDGVSPKFVLPETFDGVRMEITGQLGMIWELVKAPVIVPLLQLAVYICLLMSVMLLCERVYMGIVIVLVK
<u>Hierarchy</u>	LFWKKPDKRYKFEPIHDDEELGSSNFPVVLVQIPMFNEREVYKLSIGAACGLSWPSDRLVIQVLDDSTDP
Ontologies	TVKQMVEVECQRWASKGINIRYQIRENRVGYKAGALKEGLKRSYVKHCEYVVIFDADFQPEPDFLRRSIP
<u>GO</u>	Multiple sequence FASTA file: Choose File No file chosen
<u>EC</u>	
<u>Phenotype</u>	Notification: E-mail 💌
TOOLS	Email address: yanbin.yin@gmail.com
<u>Compare genomes</u>	
<u>Phylogenetic trees</u>	Use an <u>example sequence</u> (1plc plastocyanin) 🗖
Web services	Submit Reset
<u>Downloads</u>	(show further options)

#### SEARCH

Keyword search

#### Sequence search

#### <u>requeriee beu</u>

#### BROWSE

Nucleotide-diphospho-sugar transferases

AT5G22740.1|AT5G22740.1|cslA

1

# Organisms

.... <u>Taxonomy</u>

Scop

SCOP

Hierarchy

Ontologies

- ..... <u>GO</u>
- ---- <u>EC</u>
- Phenotype

#### TOOLS

Compare genomes Phylogenetic trees Web services Downloads

#### ABOUT

<u>Description</u> <u>Publications</u> <u>Documentation</u>

#### HELP

<u>User support</u> <u>Contact us</u>

#### Click on the picture above to see genome sequences with the same domain architecture

Sequence:	AT5G22740.1 AT5G22740.1 cslA	
Domain Number 1	Region: 94-377	
Classification Leve	Classification	E-value
Superfamily	Nucleotide-diphospho-sugar transferases	5.42e-44
Family	MGS-like	0.09
Further Details:	Family Details Alignments Genome Assignments Domain	Combinations
	·	

The results are sorted from lowest E-value to highest E-value. Strong classifications hav library classifications have an E-value greater than 0.0001. They are shown in gray. Amb domain architecture.

The family level classification is conditional on the domain being a member of the specin possibility that the selected domain is a member of a sub-family for which no structure I the family E-value will likely be > 0.01.

A machine-readable file of the assignments is available here.

Databases       Tools       Research       Training       Industry       About Us       Help       Site Index         • Tools Home       • Tools A-Z       • EBI > Tools       http://www.ebi.ac.uk/Tools/         • ID Mapping       • Literature       We provide a comprehensive range of bioinformatics tools.         • Microarray Analysis       This page shows a selection of those that are used most frequently. These include tools for the analysis and
Tools A-Z       Tools at the EBI         ID Mapping       We provide a comprehensive range of bioinformatics tools.
ID Mapping     Literature     We provide a comprehensive range of bioinformatics tools.
Literature     We provide a comprehensive range of bioinformatics tools.
• Microarray Analysis This page shows a selection of those that are used most frequently. These include tools for the analysis and
Protein Functional comparison of nucleotide and protein sequences, data from functional genomics experiments, text mining of
Analysis the scientific literature and tools for determination and visualisation of macromolecular structures.
** Proteomic Services All these tools can be accessed over the web and most provide Web Services interfaces using SOAP or RES
Sequence Analysis APIs.
Similarity & Homology
Structural Analysis     Nucleotide and Protein sequence searching
<ul> <li>Web Services</li> <li>Nucleotide sequence searches</li> <li>Protein sequence searches</li> </ul>
<ul> <li>Databases</li> <li>The sequence databases that can be searched with the tools outlined below include EMBL-Bank,</li> <li>The protein sequence databases available to searched below include UniProtKB, sequences derived from</li> </ul>
Downloads Coding Sequences, immunoglobulins and High macro molecular structures, immunoglobulins and
throughput cDNA: sequences from patents:
ENA Search     BLAST Nucleotide     BLAST Nucleotide     PSI-Search
Fasta Genomes     Fasta Proteomes
<u>Ssearch Genomes</u> <u>Ssearch Proteomes</u>
Multiple Sequence Alignment Pairwise Sequence Alignments
Alignment of three or more sequences to identify Alignment of two sequences to identify regions
regions of conservation which may indicate where the sequence is conserved and conversely functional constraints and infer evolutionary regions where the sequence is not conserved.
relationships.

Clustel Omega

- Needle

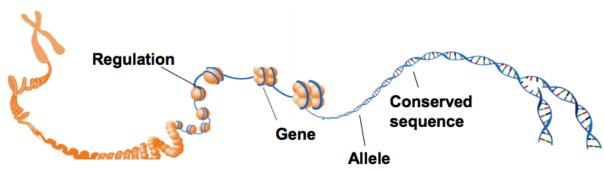
The Ensembl project aims to automatically annotate genome sequences, integrate these data with other biological information and to make the results freely available to geneticists, molecular biologists, bioinformaticians and the wider research community. Ensembl is jointly headed by Dr Stephen Searle at the Wellcome Trust Sanger Institute and Dr Paul Flicek at the European Bioinformatics Institute (EBI).

Search: All species	New to Ensembl?
e.g. BRCA2 or rat X:100000200000 or coronary heart disease	
e.g. Breez of fat A. 100000.200000 of coronary heart disease	Did you know you can: Learn how to use Ense with our video tutorials and y
Browse a Genome	e? Add custom tracks
The Ensembl project produces genome databases for vertebrates and other eukaryotic species, and	using our new Control Panel
makes this information freely available online.	Upload and analyse you and save it to your Ensemble
Popular genomes	<ul> <li>Search for a DNA or pro</li> </ul>
Human GRCh37 Mouse GRCm38	using BLAST or BLAT
GRCm37	Fetch only the data you from our public database, us
Zebrafish	Download our database
Zv9	in FASTA, MySQL and othe
★ Log in to customize this list http://www.ensembl.org/	Mine Ensembl with Bio and export sequences or tal
All genomes	Still got questions? Try our FAQs
Select a species	
View full list of all Ensembl species	What's New in Release 70 (
Other species are available in <u>Ensembl <i>Pre!</i></u> and <u>EnsemblGenomes</u>	
	Human BodyMap data reana     New assemblies for Rat (Rno

# What do we need genome browsers?

To make the bare DNA sequence, its properties, and the associated annotations more accessible through graphical interface.

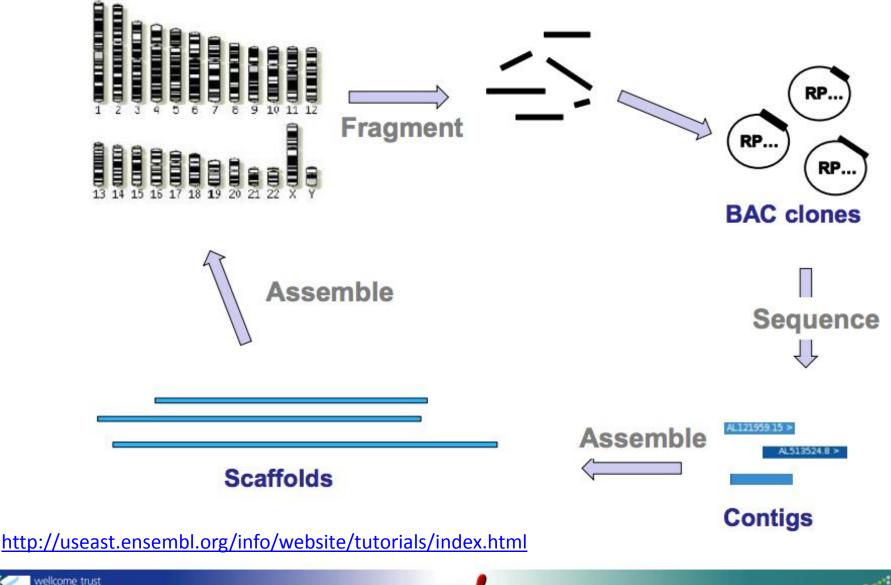
Genome browsers provide access to large amounts of sequence data via a graphical user interface. They use a visual, high-level overview of complex data in a form that can be grasped at a glance and provide the means to explore the data in increasing resolution from megabase scales down to the level of individual elements of the DNA sequence.



- Splice variants, proteins, non-coding RNA
- Small and large scale sequence variation, phenotype associations
- Whole genome alignments, protein trees
- Potential promoters and enhancers, DNA methylation
- User upload, custom data

Figure adapted from the ENCODE project www.nature.com/nature/focus/encode/

# **Genome Sequencing**



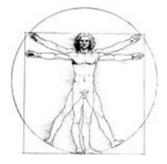
EMBL-EBI



<sup>S</sup> R <sub>C</sub>	Genor	ne Re	ferenc	e Con	sortiur	ТСАТСАААТ АСС Патсаат асс Патаста сас Статасаса стт	ATCATGAAAT ACGATCATGAAAT ACGATCATGAAAT ACGA GTTAATGAAT ACGGTTAATGAAT ACGGTTAATGAAT ACGG CTAATAATTA GACCTAATAATTA GACCTAATAATTA GACCT ACTATAGACA CTTACTATAGACA CTTACTATAGACA CTTAC
	GRC Home	Data	<u>Help</u>	<u>Report an Issue</u>	Contact Us	<u>Credits</u>	Curators Only
	Human Mou	<u>se</u> <u>Zebrafish</u>	Paper Supplem	ental Data			

# **Genome Assemblies**

The GRC has built tools to facilitate the curation of genome assemblies based on the sequence overlaps of long, high quality sequences (Clones and PCR products, not short sequence reads). The GRC currently supports production of assemblies for human, mouse or zebrafish. If your assembly data fits this model and you are interested in using these tools please contact us using the 'Contact Us' page.



## Human

The human genome assembly was produced as part of the Human Genome Project (HGP). The previous assembly (NCBI36) was the last one produced by the HGP and was described in 2004(PMID: 15496913); this was the starting point for the GRC. The assembly is based largely on assembling overlapping clone sequences.

#### Human assembly information

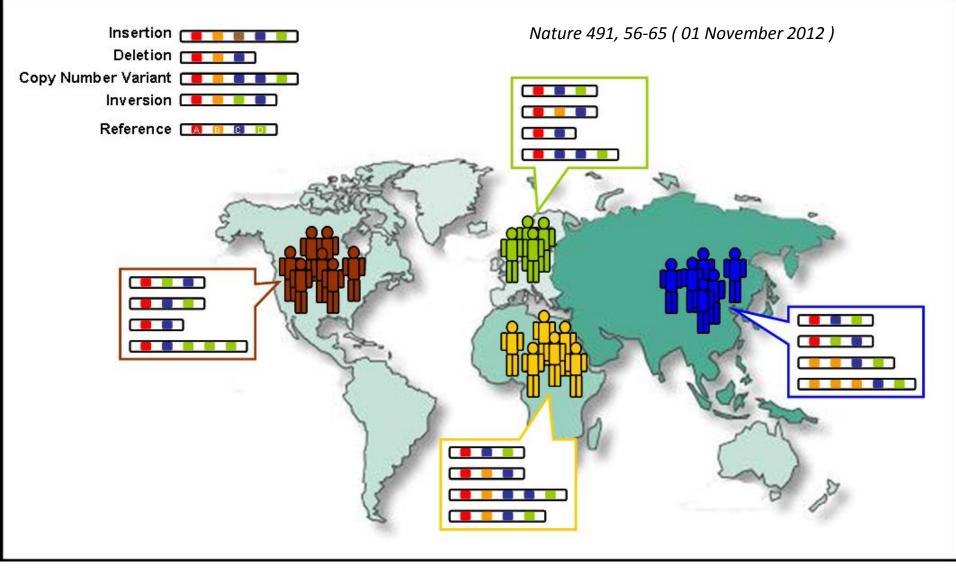
Current Major Assembly	GRCh37		
Regions with Alternate Loci	3		
Assembly N50	46,39	5,641 bp	
Remaining Gaps	357		
Remaining Gaps Patch Release Version	357 p11		

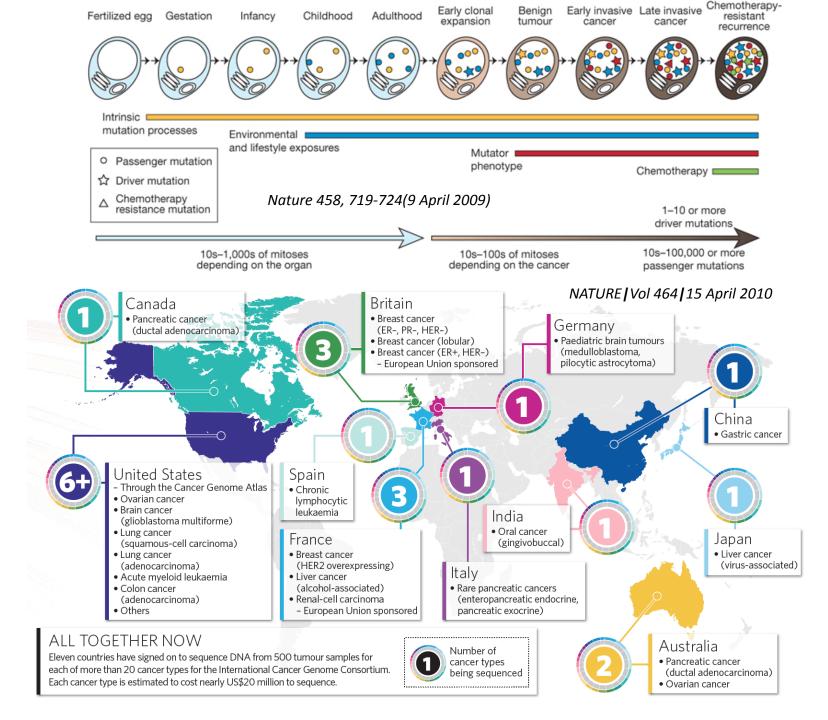
The Genome Reference Consortium consists of:



More Human assembly statistics...

1000 Genomes





While a user may start browsing for a particular gene, the user interface will display the area of the genome containing the gene, along with a broader context of other information available in the region of the chromosome occupied by the gene.

This information is shown in "tracks," with each track showing either the genomic sequence from a particular species or a particular kind of annotation on the gene. The tracks are aligned so that the information about a particular base in the sequence is lined up and can be viewed easily.

In modern browsers, the abundance of contextual information linked to a genomic region not only helps to satisfy the most directed search, but also makes available a depth of content that facilitates integration of knowledge about genes, gene expression, regulatory sequences, sequence conservation between species, and many other classes of data.

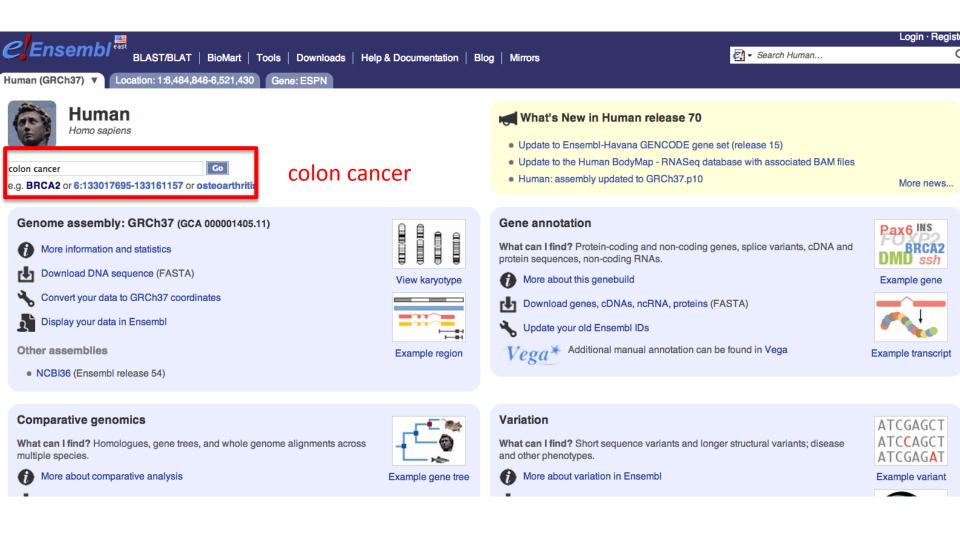
- Ensembl Genome Browsers: http://www.ensemblgenomes.org
- NCBI Map Viewer: http://www.ncbi.nlm.nih.gov/mapview/
- UCSC Genome Browser: http://genome.ucsc.edu

Each uses a centralized model, where the web site provides access to a large public database of genome data for many species and also integrates specialized tools, such as BLAST at NCBI and Ensembl and BLAT at UCSC.

The public browsers provide a valuable service to the research community by providing tools for free access to whole genome data and by supporting the complex and robust informatics infrastructure required to make the data accessible

# Hands on exercise 2: Ensembl gene search

# http://www.ensembl.org/



<b>Ensembl</b> BLAST	/BLAT   BioMart   Tools   Downloads	s   Help & Documer	ntation   Blog   Mirrors	
Search Ensembl	<b>Results Summary</b>			
<ul> <li>Configure this page</li> <li>Add your data</li> </ul>	Your search of Human with 'cold	on cancer' returne	ed the following results:	
La Export data	By Feature type		By Species	
	Total	104	Total	104
+ Bookmark this page	► Domain	1	► Human	104
Share this page	Family	5		
Cindio uno pugo	▶ Gene	19		
	Phenotype	7		
	Transcript	61		

11

Ensembl release 70 - January 2013 © WTSI / EBI

Permanent link - View in archive site

Variation



#### Human (GRCh37) 🔻

Search	Ensembl
L New	Search

### **Result in Detail**

🌣 Configure this page

#### 📌 Add your data

- Export data
- + Bookmark this page
- < Share this page

#### 19 Genes match your query ('colon cancer') in Human

Showing results 1-10

1 2 Next »

#### SDCCAG3P2

Description	serologically defined colon cancer antigen 3 pseudogene 2 [Source:HGNC Symbol;Acc:391
Gene ID	ENSG00000181101
Location	<u>1:175013762-175014784:-1</u>
Variations	Variation Table
Source	e70

#### SDCCAG8

Source

Description	serologically defined colon cancer antigen 8 [Source:HGNC Symbol;Acc:10671] [Type: prot
Gene ID	ENSG0000054282
Location	<u>1:243419320-243663394:1</u>
Variations	Variation Table
Source	e70
MACC1	
MACC1 Description	metastasis associated in colon cancer 1 [Source:HGNC Symbol;Acc:30215] [Type: protein c
	metastasis associated in colon cancer 1 [Source:HGNC Symbol;Acc:30215] [Type: protein c
Description	
Description Gene ID	ENSG0000183742

#### Gene-based displays

- Gene summary
- Splice variants (5)
- Supporting evidence Sequence
- External references
- Regulation
- Comparative Genomics
- Genomic alignments
- Gene tree (image)
- Gene tree (text)
- Gene tree (alignment)
- └ Gene gain/loss tree
- Orthologues (56)
- Paralogues (1)
- Protein families (1)

#### Phenotype

- Genetic Variation
  - Variation table
  - Variation image
     Structural variation
- External data
- Personal annotation
- ID History
- Gene history

#### 🌣 Configure this page

- 💦 Add your data
- 🛃 Export data
- H Bookmark this page

< Share this page

#### Gene: MACC1 ENSG00000183742

Description	metastasis associated in colon cancer 1 [Source:HGNC Symbol;Acc:30215]
Location	Chromosome 7: 20,174,905-20,257,027 reverse strand.
INSDC coordinates	chromosome:GRCh37:CM000669.1:20174905:20257027:1
Transcripts 🖃	This gene has 5 transcripts

Show/hide	e columns				Filter	
Name 🔅	Transcript ID 🔅	Length (bp) 🔅	Protein ID 🕴	Length (aa) 🔅	Biotype 🔅	CCDS 🔅
MACC1-001	ENST0000400331	8532	ENSP00000383185	852	Protein coding	CCDS5369
MACC1-002	ENST00000332878	2994	ENSP00000328410	852	Protein coding	CCDS5369
MACC1-005	ENST00000589011	2686	ENSP00000466864	852	Protein coding	CCDS5369
MACC1-003	ENST00000471019	304	No protein product	-	Processed transcript	
MACC1-004	ENST00000483317	608	No protein product	-	Retained intron	-

#### **Transcript and Gene level displays**

In Ensembl we provide displays at two levels:

• Transcript views which provide information specific to an individual transcript such as the cDNA and CDS sequences and protein domain annotation.

• Gene views which provide displays for data associated at the gene level such as orthologues, paralogues, regulatory regions and splice variants.

This view is a gene level view. To access the transcript level displays select a Transcript ID in the table above and then navigate to the information you want using at the left hand side of the page. To return to viewing gene level information click on the Gene tab in the menu bar at the top of the page.

### Gene summary ()

Name	MACC1 (HGNC Symbol)
Synonyms	7A5, SH3BP4L [To view all Ensembl genes linked to the name click here.]
CCDS	This gene is a member of the Human CCDS set: CCDS5369
Ensembl version	ENSG00000183742.7
Gene type	Known protein coding
Prediction Method	Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see article.
Alternative genes	This gene corresponds to the following database identifiers:
	Havana gene: OTTHUMG00000128415 (version 4)

# A consensus set of protein coding sequences



- Reaching a consensus coding sequence set for human and mouse.
- 26,473 (human)
   22,187 (mouse) (\*as of Sept 2011)
- If you see a "CCDS ID", the coding sequence is agreed upon.

Genome Res. 2009 Jul;19(7):1316-23. Epub 2009 Jun 4

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## VEGA/Havana (human, mouse, z-fish)

 Automatic annotation pipeline: Gene building all at once (whole genome)
 <u>Ensembl</u>

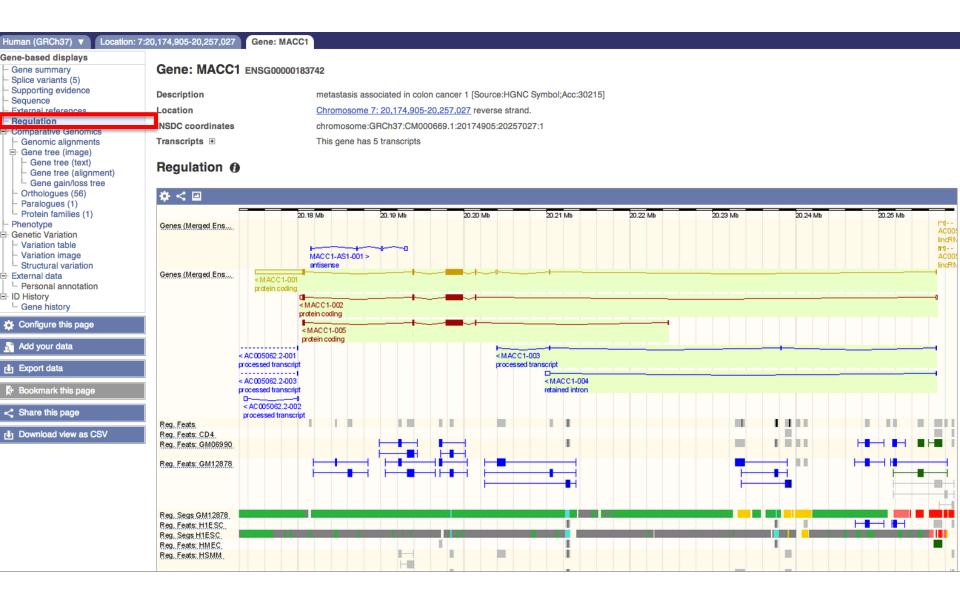
Manual curation: reviewed by experts
 <u>VEGA: Vertebrate Genome Annotation</u>
 <u>Havana</u>





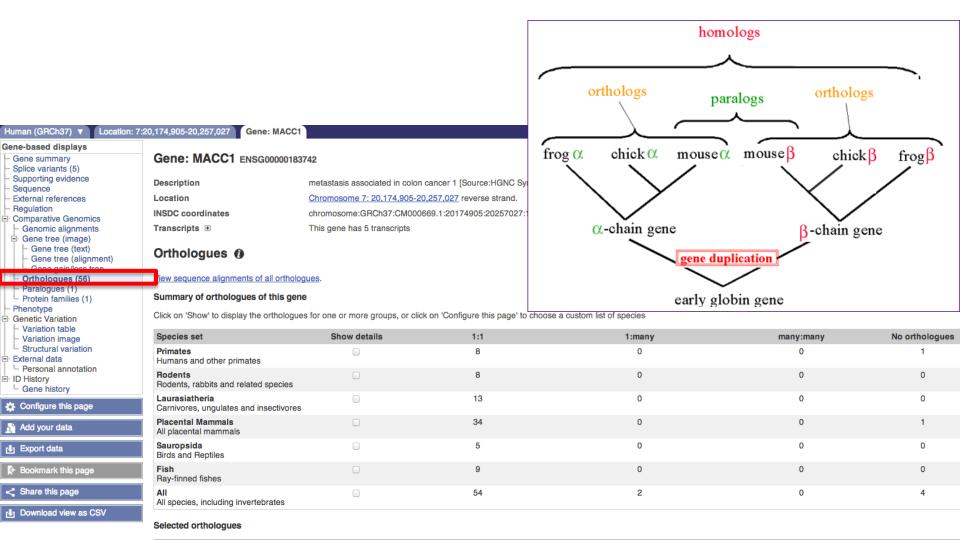
12 of 31

CEnsembl and BLAST/	/BLAT   BioMart   Tools   Downloads	Help & Documentation   Blog   Mirrors		🛃 🕶 Search Human
Human (GRCh37) ▼ Location: 7:2	20,174,905-20,257,027 Gene: MACC1			
Gene-based displays - Gene summary - Splice variants (5)	Gene: MACC1 ENSG0000018	3742		
<ul> <li>Supporting evidence</li> </ul>	Description	metastasis associated in colon cancer 1 [Source:HGN0	C Symbol;Acc:30215]	
Sequence     External references	Location	Chromosome 7: 20,174,905-20,257,027 reverse strand	I.	
- Regulation	INSDC coordinates	chromosome:GRCh37:CM000669.1:20174905:202570	027:1	
Comparative Genomics Comparative Genomics Comparative Genomic alignments Comparative Genomic alignments Comparative Genomic alignments Comparative Genomics	Transcripts	This gene has 5 transcripts		
<ul> <li>Gene tree (text)</li> <li>Gene tree (alignment)</li> </ul>	Marked-up sequence Ø			
<ul> <li>Gene gain/loss tree</li> <li>Orthologues (56)</li> <li>Paralogues (1)</li> </ul>	Кеу			
Protein families (1)	Exons All exons in this region	ACC1 exons		
Phenotype     Genetic Variation     Variation table     Variation image     Structural variation     External data	Intronic M Splice donor S	prime UTRFrameshiftissenseSplice acceptorplice regionStop gainedpstreamFrameshift		
└─ Personal annotation ⊡· ID History └─ Gene history		5:20257627:-1 CCAGA <mark>R</mark> CATTTTAGAAGATGAAATGCCAAAAGGTCTCCA TTTGAGTGACAATCACAGTGCTGATGTAGAGGGAAAGGG	60 <u>27: rs140553504</u> ; 120	
🌣 Configure this page	121 GGAACTAGTTAGACACTGTCA 181 <mark>GTGAGGCACCTTCAGCTCTGA</mark>	CTCAC <mark>CTGGGAAGGCTTTATTCACCTGTTCCACAGGGCA</mark> ATCACCGAAAGA <mark>R</mark> AATCTGGTGGGGGCAAGTTCCAGCTGC	180 240 <u>214: rs146815034</u> ;	
Add your data	301 CTCCCTGGCAAACATTAAACC	.TATTTTTTACTTATTGCTAACACTGAGGGTGCCTTCTTA CCTTTTATTTCCTTTCATGGAAATAAG <mark>M</mark> TTATATTTACA .C <mark>CTGATTTTTTTTTTAATTGCTTTTCCACCTGCTTCCCCT</mark>	300 360 <u>349: rs149806223</u> ; 420 <u>370: rs186812749</u> ;	
🛃 Export data	421 <b>TTCTTCTT</b> AGGGTGAAACTCT 481 ATTGTGGGTAT <mark>Y</mark> GTAGTTCTC	AGCCATACGCCCTCTTCTGGTTTCGGGTGARGAGMCTGA TTGCCTTTTGGGGTTTCTAGTTGGGCAGCTTTGGAGCCA	480 <u>472: rs116306795; 4</u> 540 <u>492: rs150976176</u> ;	76: rs139790502;
🗲 Bookmark this page	601 ATACGGAGCAARGCATGTTTG	CAGGTGGCTCAGGAAGCAGGGCTGCAGTTGCCTGCCTGC AAGAGTACCCGGGTTTGGTAGAGTGACTTCTATTCACTA	600 <u>545: rs142618519; 5</u> 660 <u>612: rs116031677;</u> 720 677; rs181670505; 7	
< Share this page	721 TTT <mark>W</mark> AAAATAATA <mark>K</mark> TCCTTTC	<b>GAAGCTTGGGCTCACTTCCACAAAT</b> GTAAGTG <mark>Y</mark> TGATTT ACTATTGATATGTGCTACACACTAGGAAGTTGTATAATT A <mark>R</mark> CTGCTTAACAAATCAAGCCCTGCTTAAATAAAGGGAA	720 <u>677: rs181670505; 7</u> 780 <u>724: rs186298838; 7</u> 840 803: rs3095006;	
L Download view as RTF	841 GGTAACTGGTATTCTGTGGGC	GTTTATGGGACATGTGTTAGGATAAACCTCATGACATAGAA GGTTTAGGAATAAAGTAGTAGTTTATTTTAAAAAAAA	900 960	
Q BLAST this sequence	961 ACCCAGGTCACA <mark>WK</mark> AACCATC 1021 TGTAGATGGCTGAAAKCCCCT 1081 AAAAGACTGTGCTTTGAAGAT	CAAAGGCAAAAACAAAACACTCTATGAGTTGCTTAAAATA CAAAAACTTTAAATATCTCAAAAAGCATACAGGTATGTTA CAGATAAAGTTGCAATCAGCAGCCCCYACTGCTTATTAGAT TTCACCTCRCAAAGTCTCAGTTTCCTCATCTGTAAAGTG	1020 <u>973: rs75473490; 97</u> 1080 <u>1036: rs147441166;</u> 1140 <u>1126: rs181892367;</u> 1200 1143: rs142694437;	



CEnsemble BLAST	BLAT   BioMart   Tools	Downloads   Help & Documentation   Blog   Mirrors
Human (GRCh37) V Location: 7:	20,174,905-20,257,027	iene: MACC1
Gene-based displays	Gene: MACC1 EN	ISG0000183742
<ul> <li>Splice variants (5)</li> <li>Supporting evidence</li> </ul>		
- Sequence	Description	metastasis associated in colon cancer 1 [Source:HGNC Symbol;Acc:30215]
<ul> <li>External references</li> <li>Regulation</li> </ul>	Location	Chromosome 7: 20,174,905-20,257,027 reverse strand.
Comparative Genomics	INSDC coordinates	chromosome:GRCh37:CM000669.1:20174905:20257027:1
Genomic alignments	Transcripts 🗉	This gene has 5 transcripts
Gene tree (text)     Gene tree (alignment)     Gene gain/loss tree	Genomic alignm	ents 🕖
Orthologues (56)     Paralogues (1)     Protein families (1)	Alignment: 6 primates E	PO   Co
Phenotype     Genetic Variation	Go to a graphical view o	f this alignment
<ul> <li>Variation table</li> <li>Variation image</li> </ul>	Кеу	
└── Structural variation ⊡· External data └── Personal annotation	Features All exons	
□ ID History □ Gene history	Pan troglodytes > ch	romosome:GRCh37:7:20174305:20257627:-1 romosome:CHIMP2.1.4:7:18822240:18841988:-1 romosome:CHIMP2.1.4:7518822240:18841988:-1
🌣 Configure this page		romosome:CHIMP2.1.4:7:18758798:18822239:-1 romosome:gorGor3.1:7:20332069:20397845:-1
者 Add your data		romosome:PPYG2:7:64291238:64310848:1 romosome:PPYG2:7:64310849:64375069:1
	Macaca mulatta > ch	romosome:MMUL_1:3:105974599:106040805:1
🛃 Export data		romosome:C_jacchus3.2.1:8:35737834:35755617:1 romosome:C_jacchus3.2.1:8:35755618:35817355:1
🛠 Bookmark this page	Homo sapiens Pan troglodytes	TTAAAGTGTTATCTTAAAAATCCAGAGCATTTTAGAAGATGAAATGCCAAAAGGTCTCCATTATGTCTATATGTCTATGTCTTTGAGTGACAATCACAGTGCTGATGTAGAGGGGAAAGGG TTAAAGTGTTATCTTAAAAATCCAGAGCATTTTAGAAGATGAAATGCCAAAAGGTCTCCATTATGTCTATATGTCTATGTCTTTGAGTGACAATCACAGTGCTGATGTAGGGGGAAAGGG
Share this page	Gorilla gorilla goril Pongo abelii	lla
	Pongo abelli Macaca mulatta Callithrix jacchus	TGCTCTTTTAAAAATCCAGAGCATTTTAGAAGATGAAATGCCAAAAGGTCTCCATTATGTCTATATATCTATGTCTTTGAGTGACAATCACAGTGCTGATGTAGGGGGAAAGGG
	Homo sapiens Pan troglodytes	GGAACTAGTTAGACACTGTCACTCACCTGGGAAGGCTTTATTCACCTGTTCCACAGGGCAGTGAGGCACCTTCAGCTCTGAATCACCGAAAGAGAATCTGGTGGGGCCAAGTTCCAGCTGC GGAACTAGTTAGACACTGTCACTCACCTGGGAAGGCTTTATTCACCTGTTCCACAGGGCAGTGAGGCACCTTCAGCTCTGAATCACCGAAAGAGAATCTGGTGGGGCCAAGTTCCAGCTGC
	Gorilla gorilla goril Pongo abelii	lla GGAACTAGTTAGACACTGTCACTCGGGGAAGATTTTATTCACCTGTTCCACAGGGCAGTGAGGCACCCTCAGCTCTGAATCACCGAAAGAGAATCTGGTGGGGGCAAGTTCCAGCTGC
	Macaca mulatta Callithrix jacchus	GGAACGAGTGAGACACCATCACTCACCCAGGAAGCCTTCATTCA
	Homo sapiens Pan troglodytes	ATGAGGATTTGCTTGCATAAATATTTTTTACTTATTGCTAACACTGAGGGTGCCTTCTTACTCCCTGGCAAACATTAAACCACTTTATTTCCTTTCATGGAAATAAGATTATATTTACA ATGAGGATTTGTTTGCATAAATATTTTTTACTTACTTATTGCTAACACTGAGGGTGCCTTCTTACTCCCTGGCAAACATTAAACCACTTTTATTTCCTTTCATGGAAATAAGATTATATTTACA
	Gorilla gorilla goril Pongo abelii Monco angletta	lla ATGAGCATTTGTTTGCATAAATATTTTTTAGTTATTGCCAACACTGAGAGTGCCTTCTTACTCCCTGGCAAACATTAAACCACTTTTATTTCCTTTCATAGAAATAAGATTATCTTTACA
	Homo sapiens Pan troglodytes Gorilla gorilla goril Pongo abelii	ATGAGGATTTGTTTGCATAAATATTTTTTACTTATTGCTAACACTGAGGGTGCCTTCTTACTCCCTGGCAAACATTAAACCACTTTATTTCCTTTCATGGAAATAAGATTATATTTACA

Human (GRCh37) V Location: 7:2	20,174,905-20,257,027 Gene: MACC				
Gene-based displays					
<ul> <li>Gene summary</li> <li>Splice variants (5)</li> </ul>	Gene: MACC1 ENSG0000018	3742			
<ul> <li>Supporting evidence</li> <li>Sequence</li> </ul>	Description	metastasis associated in colon cancer 1 [Source:HGNC Symbol;Acc:30215]			
- External references	Location	Chromosome 7: 20,174,905-20,257,027 reverse strand.			
Regulation	INSDC coordinates	chromosome:GRCh37:CM000669.1:20174905:20257027:1			
Comparative Genomics     Genomic alignments	Transcripts •	This gene has 5 transcripts			
⊡ Gene tree (image)					
<ul> <li>Gene tree (text)</li> <li>Gene tree (alignment)</li> <li>Gene gain/loss tree</li> </ul>	Gene tree (image) 🚯				
<ul> <li>Orthologues (56)</li> <li>Paralogues (1)</li> </ul>	GeneTree ENSGT00390000013151				
Protein families (1)	Number of genes	116			
Phenotype     Genetic Variation	Number of speciation nodes	108			
Variation table	Number of duplication	2			
<ul> <li>Variation image</li> </ul>	Number of ambiguous	5			
└── Structural variation	Number of gene split events	0			
Personal annotation	Rumber of gene spin events	•			
⊡ ID History	� < ⊡				
└ Gene history		Reptiles: 5 homologs			
Configure this page		Hominines: 2 homologs			
💦 Add your data		MACCI, Human			
		MACCI, Orangutan			
Export data		MACC1, Gibbon			
		Lo MACC1, Macaque			
F+ Bookmark this page		MACC1, Mamoset			
< Share this page		MACC1, Tarsier			
Contacto and page		MACC1, Bushbaby			
		MACC1, Tree Shrew			
	-	Rodents and Rabbits: 7 homologs			
	••••••	Laurasiatherian mammals: 13 homologs			
		Xenarthran mammals: 2 homologs			
		African mammals: 3 homologs			
		Marsupials: 3 homologs			
		MACC1, Platypus			
		🗖 macc1, Xenopus			
		MACC1, Coelacanth			
		Ray-finned fsh: 8 homologs			



Show All 🛊 entries				Show/hide columns		Filter	**
Species	_ Туре	_ dN/dS	Ensembl identifier & gene name	Compare	Location	Target %id	Query %id
Alpaca ( <i>Vicugna pacos</i> )	1-to-1	n/a	ENSVPAG0000002084 MACC1 metastasis associated in colon cancer 1 [Source:HGNC Symbol;Acc:30215]	<ul> <li>Region Comparison</li> <li>Alignment (protein)</li> <li>Alignment (cDNA)</li> <li>Gene Tree (image)</li> </ul>	<u>GeneScaffold_1891:823408-842968:-1</u>	84	84
Anole lizard	1-to-1	n/a	ENSACAG00000011775	Region Comparison	6:29083541-29105343:-1	61	61

#### Human (GRCh37) V Location: 7:20,174,905-20,257,027 Gene: MACC1

#### Gene-based displays

Gene summary Splice variants (5) Supporting evidence Sequence External references Regulation **Comparative Genomics** Genomic alignments

Gene tree (image)

- Gene tree (text) - Gene tree (alignment)
- Gene gain/loss tree
- Orthologues (56)
- Paralogues (1)
- Protein families (1)
- Phenotype
- **Genetic Variation** - Variation table
- Variation image
- Structural variation
- External data
- Personal annotation D History
- └ Gene history

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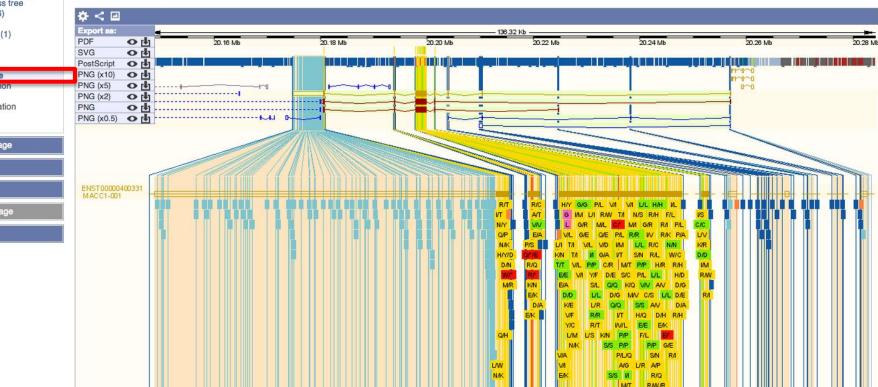
< Share this page

#### Gene: MACC1 ENSG00000183742

Description	metastasis associated in colon cancer 1 [Source:HGNC Symbol;Acc:30215]
Location	Chromosome 7: 20,174,905-20,257,027 reverse strand.
INSDC coordinates	chromosome:GRCh37:CM000669.1:20174905:20257027:1
Transcripts 🗉	This gene has 5 transcripts

#### Description

#### Variation image ()



CEnsemblPlants 
BLAST | Sequence Search | BioMart | Tools | Downloads | Help & Documentation

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C

Search: All species ; for e.g. Carboxy* or chx28 Popular genomes Arabidopsis thaliana TAIR10 Oryza sati MSU6		Featured content         This release of Ensembl Plants includes the draft genome of <u>Hordeum vulgare</u> (barley) [1]. One of the first domesticated cereal grains, originating in the Fertile Crescent over 10,000 years ago, barley played an important role in the development human civilization in southwest Eurasia [2]. At 5.3 Gbp, barley has the largest diploid genome sequenced to date. It serves as a model for adaptation, coping with a range of biotic and abiotic stresses [3]. <u>Read more</u> References         1. The International Barley Genome Sequencing Consortium (IBSC). <u>A physical, genetical and functional sequence assembly of the barley genome</u> . Nature. 2012.
Zea mays AGPv2     Brachypodium distachyon       V1.0     Physcomitrella patens       V1.0     ASM242v1		<ol> <li><u>Barley in Wikipedia</u>.</li> <li>The International Barley Genome Sequencing Consortium (IBSC). <u>At the Threshold of Efficient Access to the Barley Genome</u>. <i>Plant Physiology</i>. 2009.</li> </ol>
All genomes		Emerging resources
Select a species \$		Ensembl Plants includes an extensive set of <i>Triticum aestivum</i> (bread wheat) gene sequences and homoeologous SNPs (SNPs distinguishing genes in the component A, B, and D genomes of wheat) aligned to the <u>Brachypodium distachyon</u> genome. Currently, the size and complexity of the wheat genome precludes a chromosome-scale assembly. However, significant sequences resources have been used to produce a gene-space assembly, included here in the syntenic context of brachypodium, a model cereal and pooid relative of wheat. Sequences of diploid progenitor and ancestral species permitted homoeologous SNPs to be classified into two groups, 1) SNPs that differ between the A and D genomes (where the B genome is unknown) and, 2) SNPs that are the same between the A and D genomes, but differ in B. <u>Read more</u>
View full list of all Ensembl Plants species		
What's new in Release 16 (October 2012)		
New genomes		
<ul> <li><u>Hordeum vulgare</u> (barley)</li> </ul>	Did you know?	Ensembl Plants is developed in coordination with other plant genomics and bioinformatics groups via the EBI's role in
<ul> <li><u>Solanum tuberosum</u> (potato)</li> </ul>	For genomes where we have variation data from multiple individuals, we calculate and display linkage disequilibrium data.	the <u>transPLANT</u> consortium. The transPLANT project is funded by the <u>European Commission</u> within its <u>7th Framework Programme</u> , under the thematic area "Infrastructures", contract number <u>283496</u> .  transPLANT
<ul> <li><u>Musa acuminata</u> (banana)</li> </ul>		
<ul> <li>Updated genomes</li> </ul>		
<ul> <li>Updated gene models for <u>Glycine max</u> (soybean)</li> </ul>	For example, LD between four SNPs in	
New data	<u>Arabidopsis thaliana</u> .	
<ul> <li>Updated and improved <i>Triticum aestivum</i> (wheat) homoeologous SNPs and 'gene space' assembly alignments to <u>Brachypodium distachyon</u> (purple false brome)</li> </ul>		Ensembl Plants is produced in collaboration with Gramene
New EST alignments for:		
<ul> <li><u>Physcomitrella patens</u> (moss)</li> </ul>		
Oryza brachyantha (an ancestral rice)		Ensembl Plants is part of the Ensembl Genomes project

# Next lecture: ExPASy and DTU tools