Presentation of bioinformatics resources for gene regulation

Max 3 minutes per resource

GREEKC Training-hackathon
Marseille (France) Apr 23-26, 2019

Ensembl

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23 April 2019

GreekC Hackathon Marseille

Data in Ensembl

What is Ensembl?

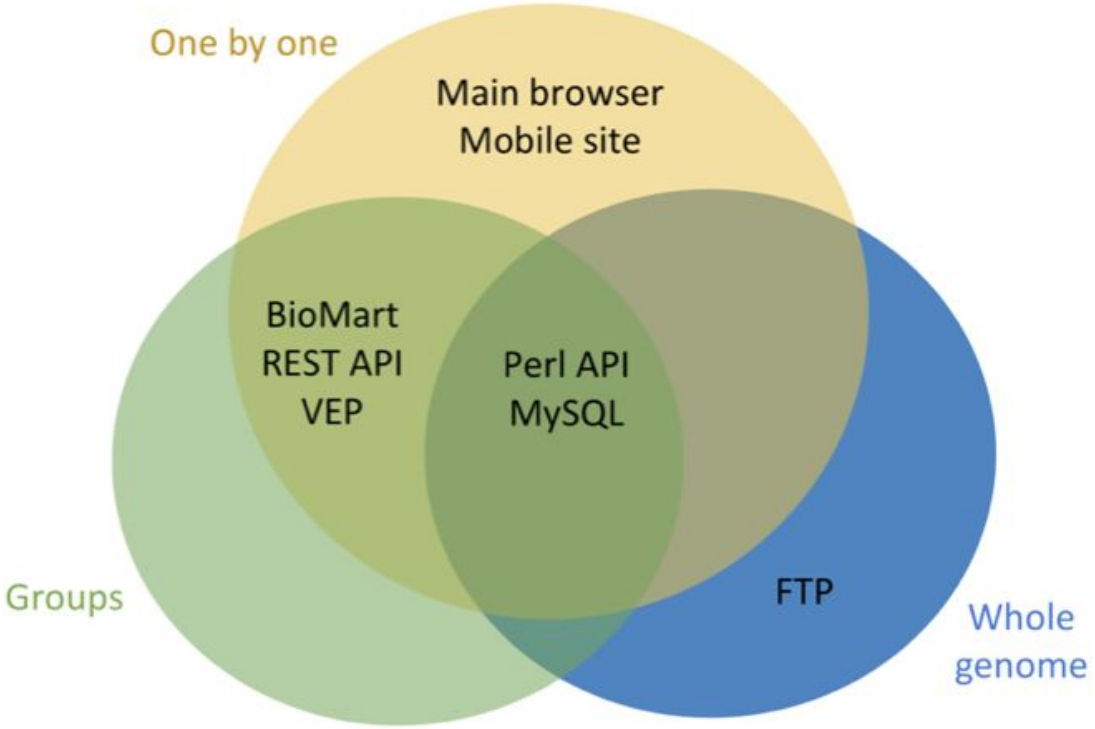
- Genome Browser, providing a single point of access to annotated genomes

- Aim of Ensembl used to be to annotate human genome with gene models and other available data
- Genome annotation
- Added in species - human/mouse/birds/reptiles/fish
- EG species – fungi, plants, bacteria, metazoa

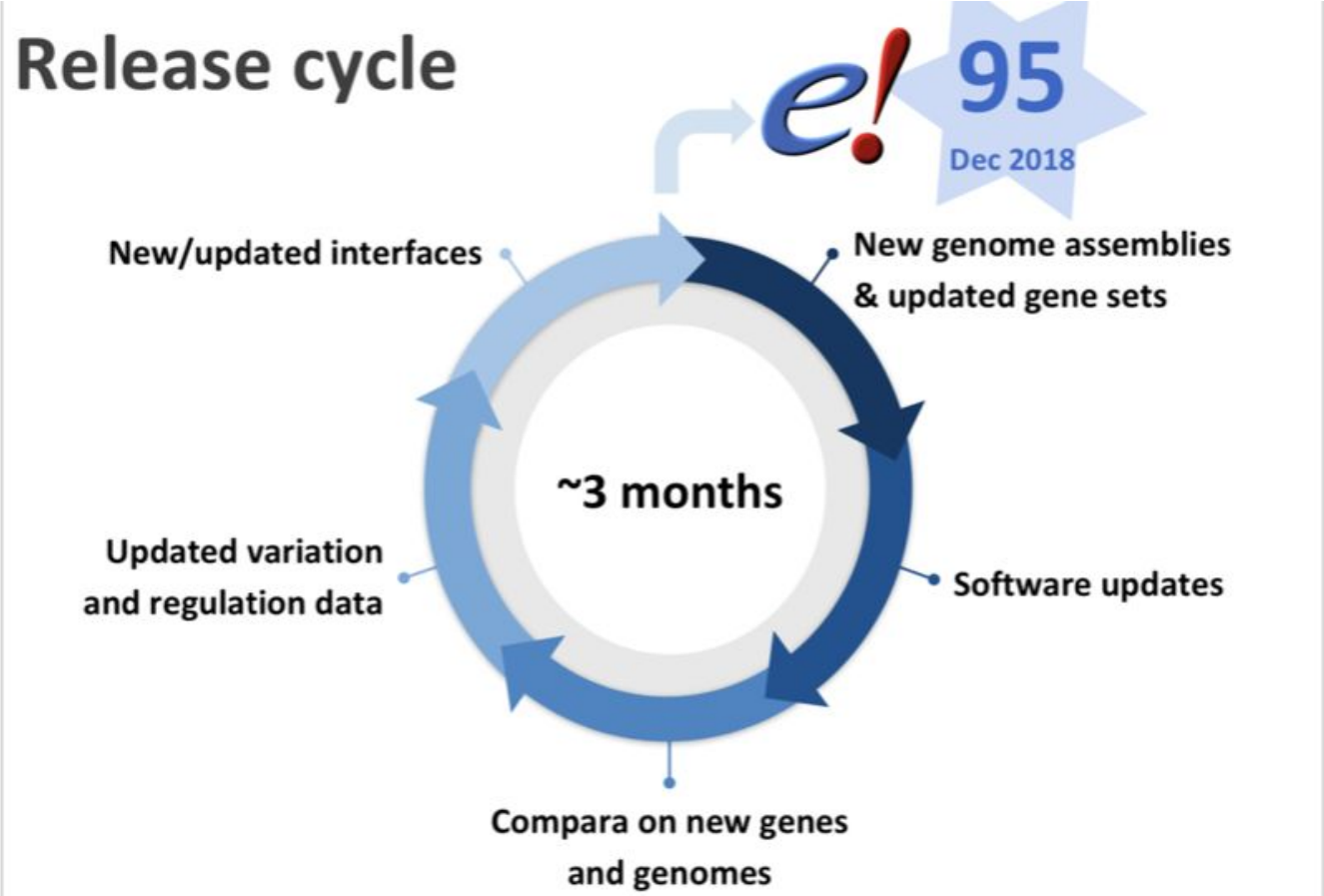
Data in Ensembl

- Genome assemblies for ~140 species
- Gene Trees
- Regulatory Build (Human and mouse)
- Variation display and VEP
- BioMart (Custom bulk data export)
- Programmatic access via the APIs
- Completely open source

Many ways to access Ensembl data



Ensembl Release Cycle



Ensembl Regulation

The goal of the Ensembl Regulation team is to annotate the genome with features that may play a role in regulation of gene expression.

- Predicted open/closed chromatin
  - DNase I sensitivity
- Transcription factor binding sites
- Epigenetic marks
  - Histone modifications
  - DNA methylation

Gene Regulation Data

- **Regulatory regions** ⇨ part of the DNA sequence
  - Promoters & flanking regions
  - Enhancers
  - CTCF binding sites
  - Transcription factor binding sites
  - Open chromatin regions
- **Epigenetic marks** ⇨ base DNA sequence unchanged
  - DNA methylation data
  - Histone modifications

Ensembl data

Where does the data come from?

Species	Data source	Assay types	Epigenomes
	ENCODE epigenomics	• ChIP-seq (histone mods) • TF binding sites • RNApol • DNase sensitivity (open chromatin)	Cultured cell lines
	ENCODE	• ChIP-seq (histone mods) • TF binding sites • RNApol • DNase sensitivity (open chromatin)	Cultured cell lines
	BLUEPRINT epigenome	• ChIP-seq (histone mods) • DNase sensitivity (open chromatin)	Primary cells from haematopoietic cell lineage (direct from human cells)

ensembl.org/info/genome/funcgen/regulation_sources.html

What does Ensembl <u>not</u> do?

- We do not link regulatory features to genes
  - We allow you see the location of features.
- We do not link regulatory features to gene expression.
  - We have cell-line specific regulation data and tissue specific expression data

You are required to make your own inferences about this data

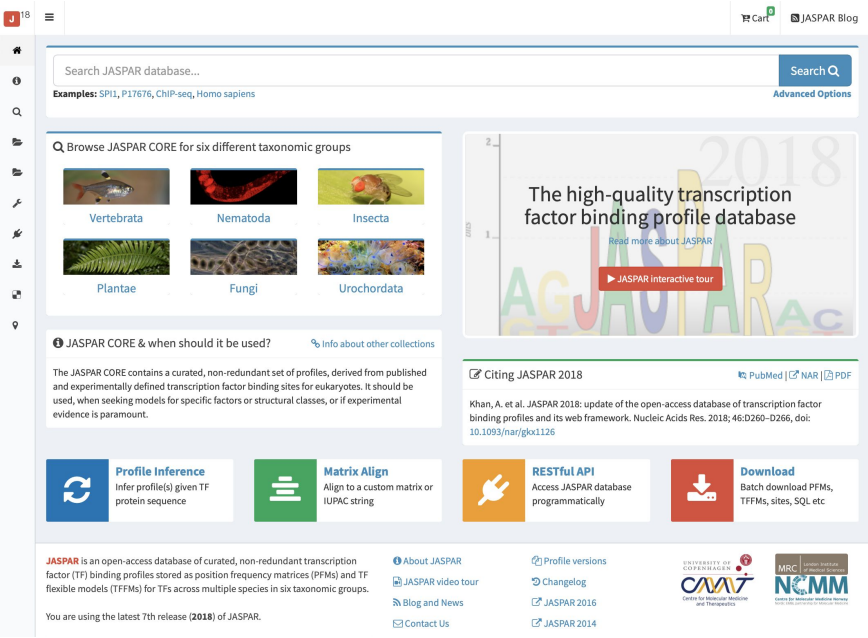
Regulatory data is incredibly complex and still in relative infancy. There is no comprehensive database of regulation data...yet!

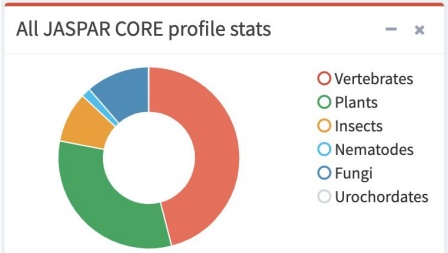
Ensembl

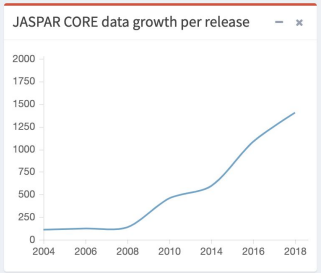
Thank you

JASPAR - database for eukaryotic TF binding profiles

- URL: jaspar.genereg.net
- TF profiles stored as position frequency matrices (PFMs) and TF flexible models (TFFMs)
- TFs across multiple species in six taxonomic groups
- 2018 release has 1404 non-redundant PFMs
  - 579 for vertebrates, 489 for plants, 176 for fungi, 133 for insects, 26 for nematodes and 1 for urochordata
  - >1.8 million pageviews by >100K users from 144 countries
- Access to data
  - REST API: **jaspar.genereg.net/api**
  - Download**:** http://jaspar.genereg.net/downloads
  - R/Bioconductor package: **JASPAR2018**

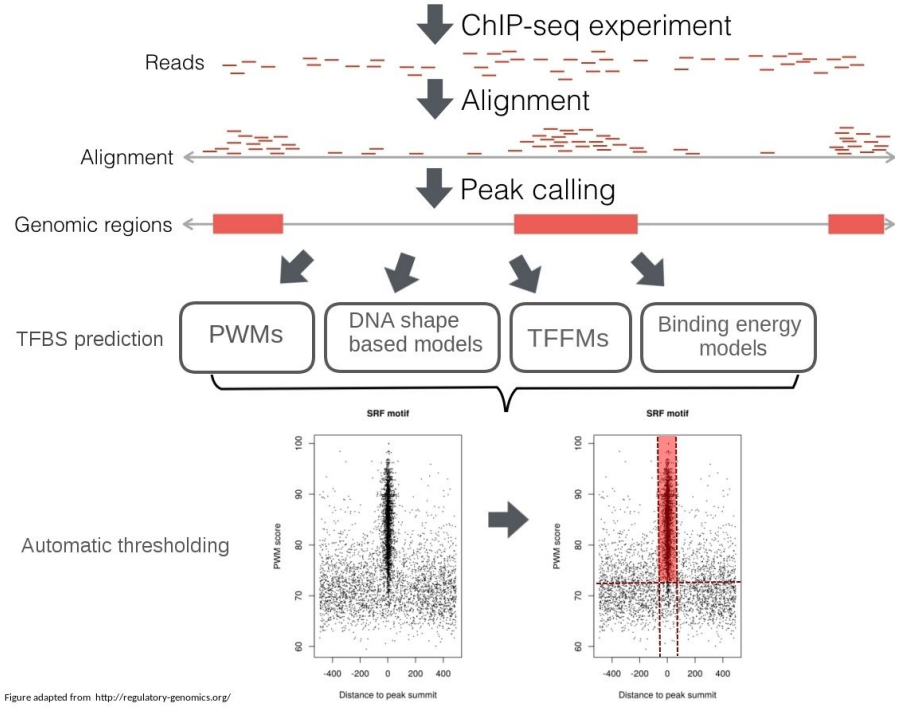


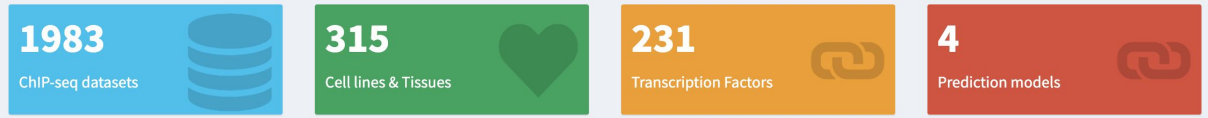




UniBind - A map of direct TF-DNA interactions in the human genome

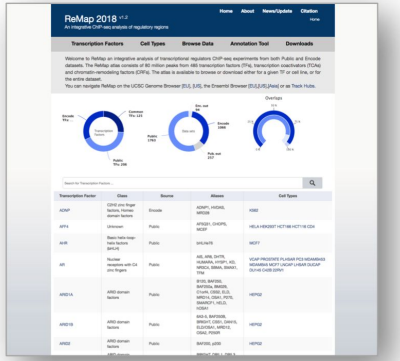
- URL: **unibind.uio.no**
- A comprehensive map of direct interactions between transcription factor (TFs) and DNA.
- High confidence TF binding site predictions were obtained from uniform processing of thousands of ChIP-seq data
- UniBind covers >4% of the human genome
- Access to data
  - Download: unibind.uio.no/downloads
- Hackathon project
  - Develop a fully functional REST API





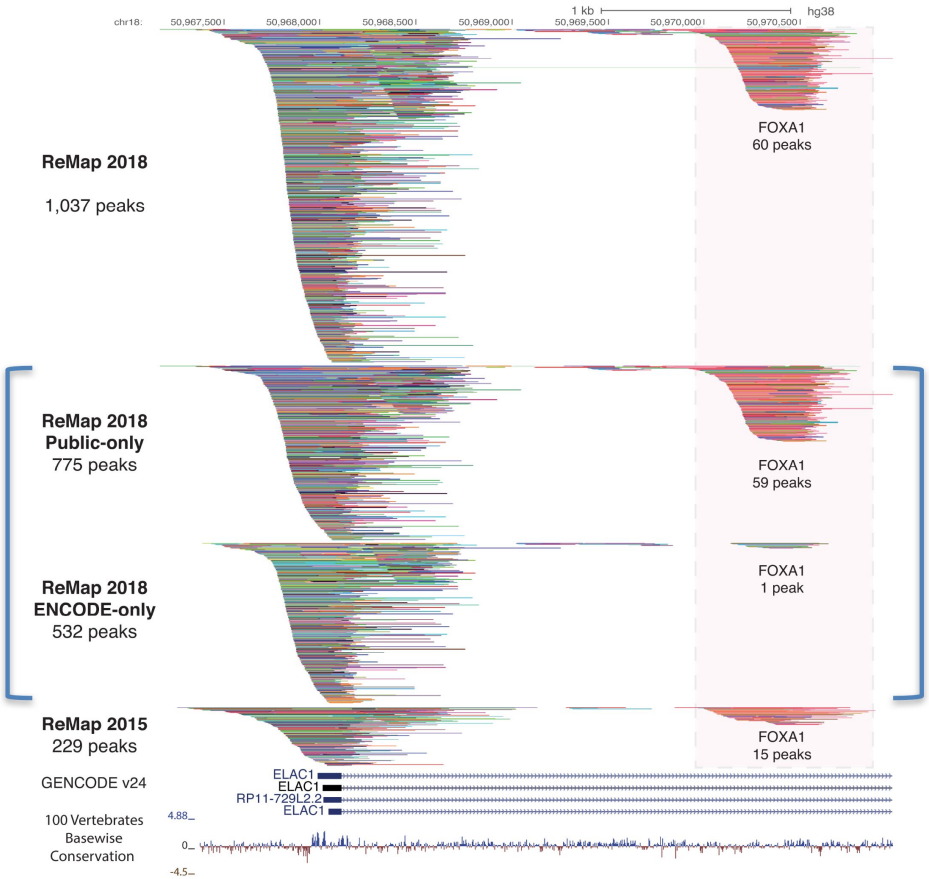
ReMap : Catalogue of ChIP-seq peaks for TR

ReMap website



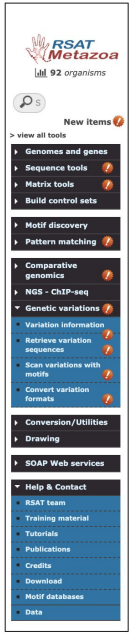
http://remap.cisreg.eu

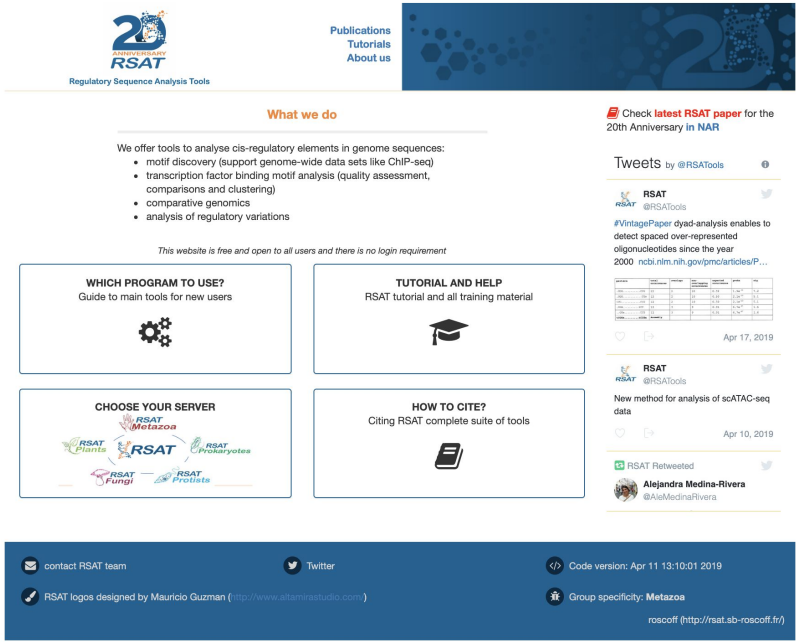
- Public data (GEO + ENCODE)
  - manual curation
- 485 TR (mainly TFs)
- 346 cell lines
- 80 millions peaks



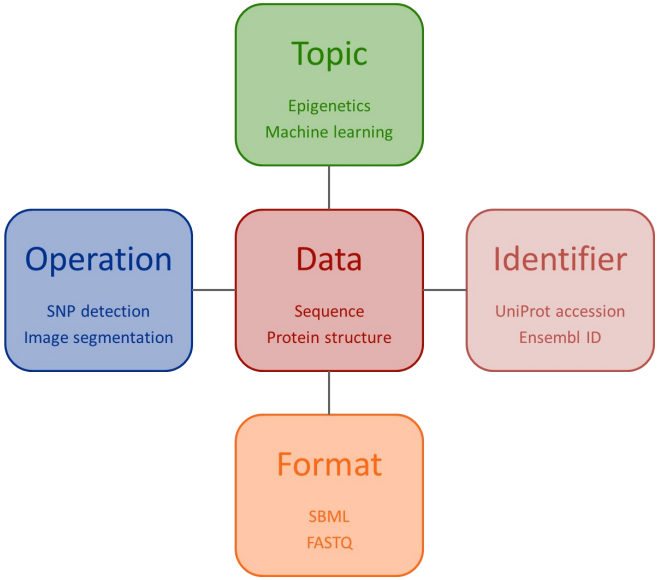
RSAT – Regulatory Sequence Analysis Tools

- URL: http://rsat.eu/
- Software suite for the detection of cis-regulatory elements in non-coding sequences.
- 6 servers worldwide
  - Bacteria, Fungi, Protists, Plants, Metazoa
  - Teaching
- Access modes
  - Web interface: 52 tools
  - Command line: many more
  - SOAP/WSDL Web services since 2008
  - REST Web services: in development *(will evolve during the hackathon)*





EDAM ontology (http://edamontology.org)



3,354 bioinformatics concepts

EDAM bioinformatics operations, types of data, data formats, identifiers, and topics

Last uploaded: July 30, 2018

Summary | Classes | Properties | Notes | Mappings | Widgets

Jump to:

- Data
- DeprecatedClass
- Format
- Operation
  - Alignment
  - Analysis
    - Enrichment analysis
    - Expression analysis
    - Genetic variation analysis
    - Image analysis
    - Pathway or network analysis
      - Expression profile pathway mapping
      - **Gene regulatory network analysis**
      - Pathway or network comparison
      - Pathway or network prediction
      - Pathway or network visualisation
      - Protein interaction network analysis
      - Weighted correlation network analysis
    - Phylogenetic tree analysis
    - Protein function analysis
    - Sequence analysis
    - Spectral analysis
    - Structure analysis
    - Text mining
    - Transmembrane protein analysis
  - Annotation
  - Calculation
  - Classification
  - Clustering

Details | Visualization | Notes (0) | Class Mappings (2)

Preferred Name	Gene regulatory network analysis
Definitions	Analyse a known network of gene regulation.
ID	http://edamontology.org/operation_1781
Created in	beta12orEarlier
hasDefinition	Analyse a known network of gene regulation.
inSubset	http://purl.obolibrary.org/obo/edam#edam http://purl.obolibrary.org/obo/edam#operations
label	Gene regulatory network analysis
prefixIRI	operation_1781
prefLabel	Gene regulatory network analysis
subClassOf	Pathway or network analysis

https://bioportal.bioontology.org/ontologies/EDAM

EDAM ontology browser (https://ifb-elixirfr.github.io/edam-browser)

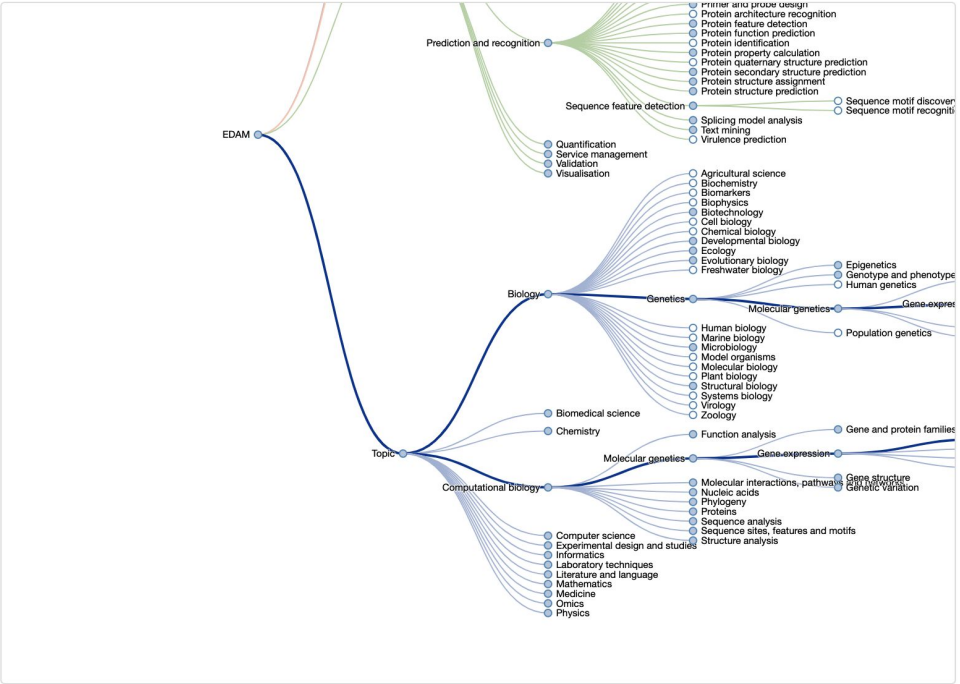
https://ifb-elixirfr.github.io/edam-browser/#topic_0204

EDAM ontology

EDAM | Custom

Gene regulation

EDAM is a simple ontology of well established, familiar concepts that are prevalent within bioinformatics [edamontology.org]



Collapse not selected elements | Reset zoom | +

1.21_dev

Details of term "Gene regulation"

Term	Gene regulation
Definition	The regulation of gene expression.
Comment	
Exact synonyms	
Narrow synonyms	Regulatory genomics
URI	http://edamontology.org/topic_0204
Parents	Gene expression Gene expression

Community usage

bio.tools	156 times
Biosphere	1 times by appliances, 1 times by tools.
BioWeb	not used
TeSS	1 times
Links	Open in AberOWL, BioPortal, OLS or WebProtégé.

Previously seen:

Details of term "Sequence motif discovery"

bio.tools

elixir bio.tools

Search bio.tools 12198 tools About Login Sign-up

JASPAR RESTful API (biotools:jaspar_api) ID Verified

http://jaspar.genereg.net/api/

Data acquisition › DNA › Protein interactions ›

OPENEBENCH 1 9

GPL-3.0

Web API Python

Widely used open-access database of curated, non-redundant transcription factor binding profiles. Currently, data from JASPAR can be retrieved as flat files or by using programming language-specific interfaces. Here, we present a programming language-independent application programming interface (API) to access JASPAR data using the Representational State Transfer (REST) architecture.

Query and retrieval ›

Credits & Support

Anthony Mathelier
Primary contact | anthony.mathelier at ncmm.uio.no

Aziz Khan
Primary contact | aziz.khan at ncmm.uio.no

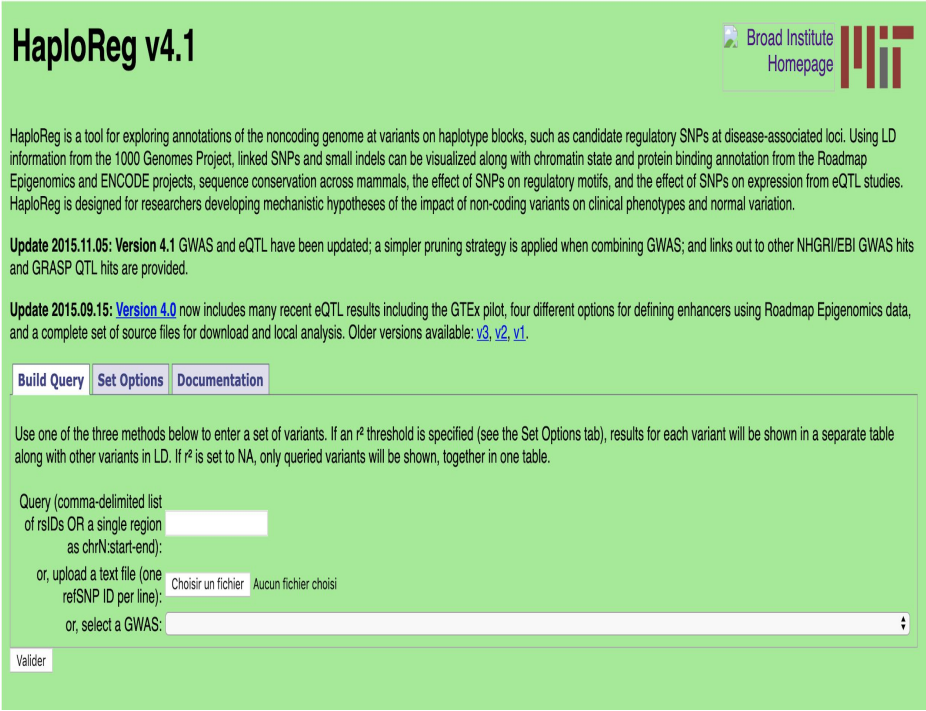
Documentation

General ›

12198 registered tools

HaploReg

- URL:https://pubs.broadinstitute.org/mammals/haploreg/haploreg.php
- Tool for exploring annotations of non-coding genome at variant on haplotype blocks, such as candidate regulatory SNPs at disease-associated loci.
- Access modes
  - Web interface
  - R package (haploR)



dbSNPs

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