

# Bioinformatics Tools for HLA

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Antalya, 2022



# Outline

**HLA and its unique features**

**Genome-wide bioinformatics tools**

**HLA-specific bioinformatics tools**

# HLA: Unique Features

Genome (3.2Gb)	xHLA Region (25.7 to 33.4Mb)	Comparison
<b>Total No of Genes</b> 60155	<b>Total No of Genes</b> 674	...
<b>Protein-coding genes</b> 19881	<b>Protein-coding genes</b> 453	32.7 vs 67.2% $P < 0.0001$
<b>Non-coding RNA Genes</b> 25411	<b>Non-coding RNA Genes</b> 54	42.6 vs 8.0% $P < 0.0001$
<b>Long non-coding RNA genes</b> 15877	<b>Long non-coding RNA genes</b> 13	26.4 vs 1.9% $P < 0.0001$
<b>Small non-coding RNA genes</b> 9534	<b>Small non-coding RNA genes</b> 7	15.9 vs 1.0% $P < 0.0001$
<b>Pseudogenes</b> 14467	<b>Pseudogenes</b> 172	24.0 vs 25.5% $P = 0.37$

**xHLA makes up 0.24% of the genome, but contains 0.40% of all SNPs in the human genome**

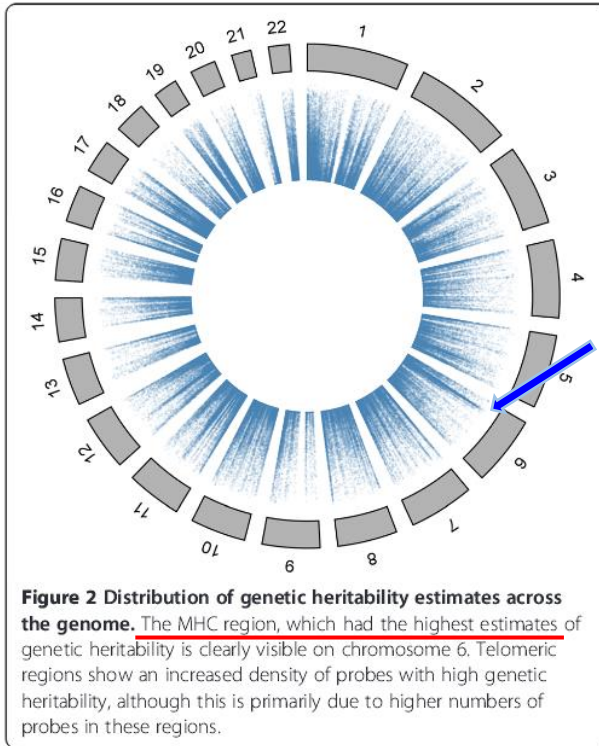
**A systematic analysis of the gene and variation content of the extended HLA region**

Ertan KANBUR, Mustafa DOGAN, Mehmet Tefvik DORAK

*Presented at EFI 2017*

# HLA: Unique Features

## Heritability of DNA methylation



McRae et al. *Genome Biology* 2014, 15:R73  
http://genomebiology.com/2014/15/5/R73



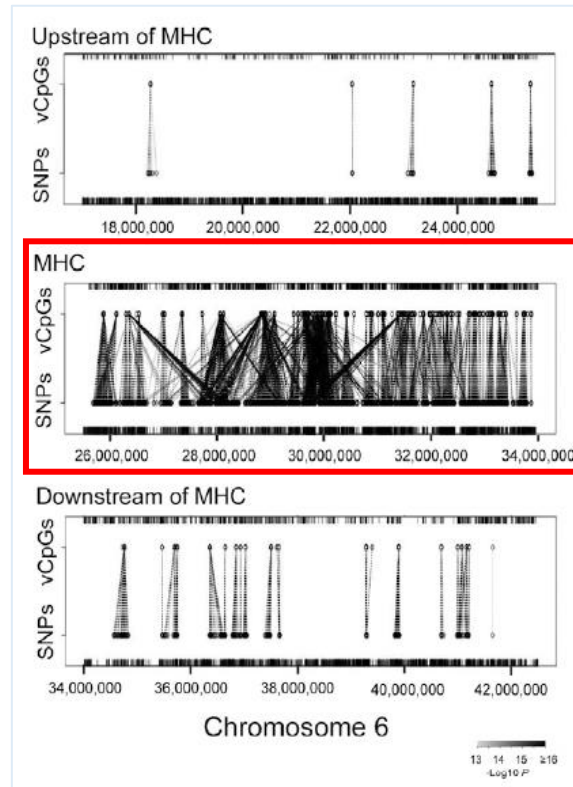
### RESEARCH

### Open Access

## Contribution of genetic variation to transgenerational inheritance of DNA methylation

Allan F McRae<sup>1,2\*</sup>, Joseph E Powell<sup>1,2</sup>, Anjali K Henders<sup>3</sup>, Lisa Bowdler<sup>3</sup>, Gibran Hemani<sup>1,2</sup>, Sonia Shah<sup>1,2</sup>, Jodie N Painter<sup>3</sup>, Nicholas G Martin<sup>3</sup>, Peter M Visscher<sup>1,2\*</sup> and Grant W Montgomery<sup>1\*</sup>

## meQTL density

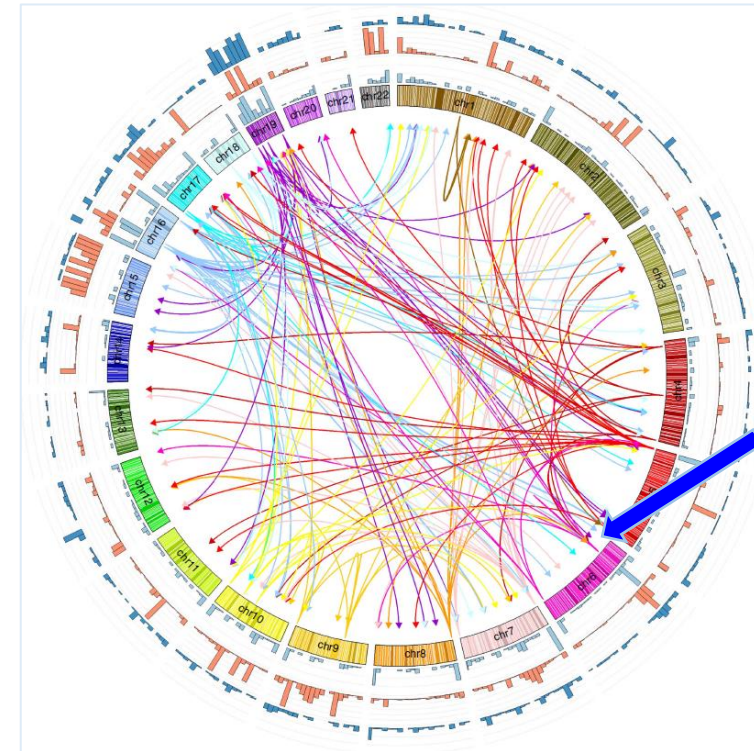


(E) Associations between CpG sites and SNPs upstream of (top panel), within (middle panel), or downstream of (bottom panel) the major histocompatibility complex (MHC) region. Each dashed line represents a significant association, and the shades of black indicate significance of the associations.

## GeMes, Clusters of DNA Methylation under Genetic Control, Can Inform Genetic and Epigenetic Analysis of Disease

Yun Liu,<sup>1,2,9</sup> Xin Li,<sup>1,2,9</sup> Martin J. Aryee,<sup>1,3</sup> Tomas J. Ekström,<sup>4,5</sup> Leonid Padyukov,<sup>4,6</sup> Lars Klareskog,<sup>4,6</sup> Amy Vandiver,<sup>1,2</sup> Ann Zenobia Moore,<sup>7</sup> Toshiko Tanaka,<sup>7</sup> Luigi Ferrucci,<sup>7</sup> M. Daniele Fallin,<sup>1,8,\*</sup> and Andrew P. Feingberg<sup>1,2,\*</sup>

## trans-eQTL/meQTL density

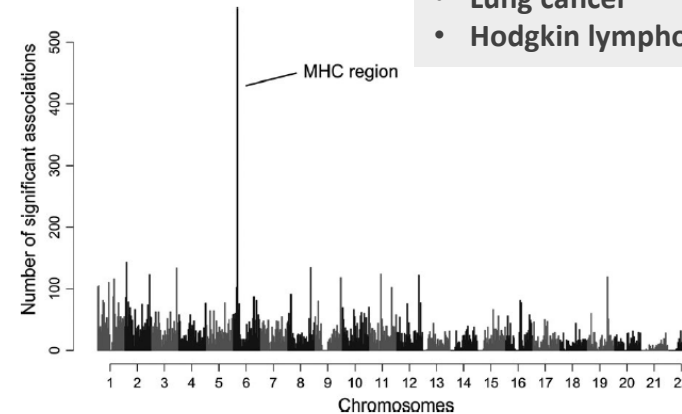
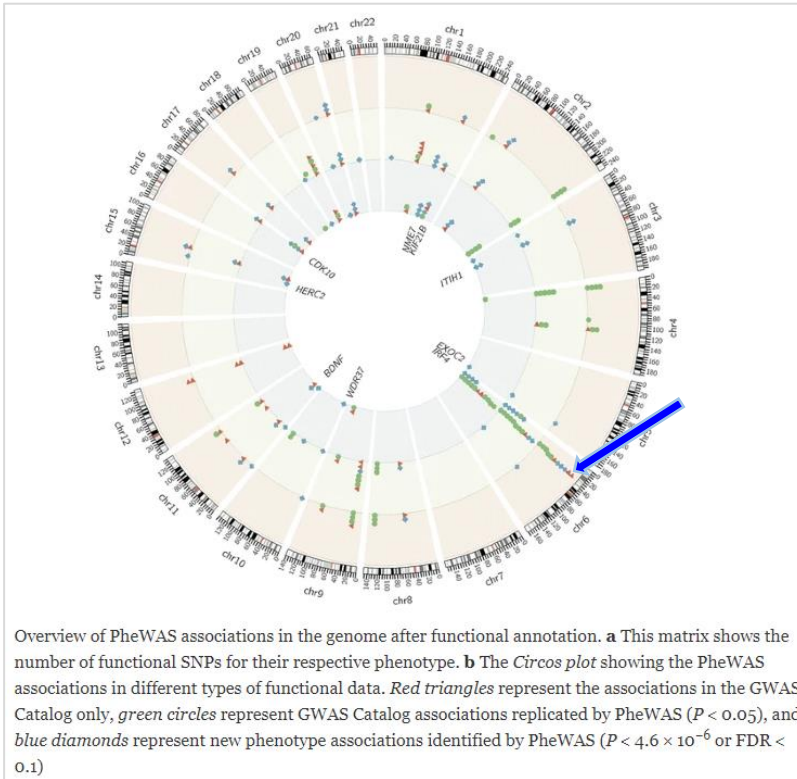


**Figure 1** | Enrichment of features in regions harbouring SNPs involved in distal SNP-CpG associations. Outer histograms: number of SNPs involved in distal SNP-CpG associations (light blue), calculated in 7.5 Mb bins; number of piRNA sequences (orange); number of transcription factors (dark blue). Inner links: SNP regions associated with four or more CpG sites. Arrows are pointing from SNPs to the CpG sites they are associated with, and are coloured according to the chromosomes where the SNPs reside.

## Long-range epigenetic regulation is conferred by genetic variation located at thousands of independent loci

Mathieu Lemire<sup>1</sup>, Syed H.E. Zaidi<sup>1</sup>, Maria Ban<sup>2</sup>, Bing Ge<sup>3</sup>, Dylan Aissi<sup>4,5,6</sup>, Marine Germain<sup>4,5,6</sup>, Irfahan Kassam<sup>7</sup>, Mike Wang<sup>1</sup>, Brent W. Zanke<sup>8</sup>, France Gagnon<sup>7</sup>, Pierre-Emmanuel Morange<sup>9,10,11</sup>, David-Alexandre Tréguët<sup>4,5,6</sup>, Philip S. Wells<sup>8</sup>, Stephen Sawcer<sup>2</sup>, Steven Gallinger<sup>12,13</sup>, Tomi Pastinen<sup>3</sup> & Thomas J. Hudson<sup>1,14,15</sup>

# HLA: Unique Features



including:

- Schizophrenia
- Alzheimer disease
- Parkinson disease
- Lung cancer
- Hodgkin lymphoma

**FIGURE 1** Number of significant GWAS associations along the genome. The chromosomal location of significant trait associations from GWAS ( $N = 18,682$ ) is shown for all autosomes. Data from NHGRI GWAS catalog. Reproduced from "Lenz TL, Spirin V, Jordan DM, Sunyaev SR. Excess of Deleterious Mutations around HLA Genes Reveals Evolutionary Cost of Balancing Selection. Mol Biol Evol 2016;33(10):2555-64. <https://doi.org/10.1093/molbev/msw127>" by permission of Oxford University Press on behalf of the Society for Molecular Biology and Evolution

.... despite already showing the highest number of disease associations, the true extent of the involvement of the MHC region in disease genetics may not have been uncovered.

Zhao et al. Genome Medicine (2018) 10:7  
DOI: 10.1186/s13073-018-0513-x

Genome Medicine

RESEARCH

Open Access



An integrative functional genomics framework for effective identification of novel regulatory variants in genome-phenome studies

Junfei Zhao<sup>1†</sup>, Feixiong Cheng<sup>2,3†</sup>, Peilin Jia<sup>1</sup>, Nancy Cox<sup>4,5</sup>, Joshua C. Denry<sup>5,6</sup> and Zhongming Zhao<sup>1,7\*</sup>

Received: 9 March 2017 | Revised: 16 June 2017 | Accepted: 20 July 2017

DOI: 10.1111/jgi.12332

REVIEW

WILEY INTERNATIONAL JOURNAL OF IMMUNOGENETICS

What has GWAS done for HLA and disease associations?

A. E. Kennedy<sup>1</sup> | U. Ozbek<sup>2,3</sup> | M. T. Dorak<sup>4</sup>



# Bioinformatics Tools: Genome-wide



Human Mouse How to access data FAQ



## More about GENCODE Human

[Current human data](#)

[Release history](#)

[Statistics](#)

[Data format](#)

[FTP site](#)

## General stats

Total No of Genes	61544
Protein-coding genes	19988
Long non-coding RNA genes	18805
Small non-coding RNA genes	7567
Pseudogenes	14774

HGNC

Search symbols, keywords or IDs



[Home](#) [Gene data](#) [Tools](#) [Downloads](#) [VGNC](#) [Contact us](#) [More](#)

[Request symbol](#)

## Statistics & download files ?

Filter statistics and download files by chromosome

All chromosomes

### Statistics

Locus Group	Total by Locus Group	Locus Type	Total by Locus Type
protein-coding gene	19220	gene with protein product	19220
non-coding RNA	8883	RNA, Y	4
		RNA, cluster	119
		RNA, long non-coding	5545
		RNA, micro	1912



National Library of Medicine  
National Center for Biotechnology Information

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### Eukaryotic genomes annotated at NCBI

Hundreds of eukaryotic genomes have been annotated by the NCBI Eukaryotic Genome Annotation Pipeline (see [graphs](#)). The latest annotation release available for each genome is shown in the tables below. The tables are organized by taxonomic group and provide links to the annotation report, FTP site, genome BLAST page, and Genome Data Viewer page.

Only completed annotations are shown here. Please browse the [annotation runs currently in progress](#) to see what will become available in a few days.

[Show/Hide All](#)

Featured (6)

[FTP](#) - FTP Download [B](#) - Organism-specific BLAST [AR](#) - Annotation Report [GDV](#) - Genome Data Viewer

Species	RefSeq assembly(ies)	Annotation Release	Freeze Date	Release Date	Links
<a href="#">Homo sapiens (human)</a>	<a href="#">GRCh38.p13</a> (GCF_000001405.39)	109.20211119	2021-11-19	2021-11-22	<a href="#">FTP</a> <a href="#">B</a> <a href="#">AR</a> <a href="#">GDV</a>
<a href="#">Mus musculus (house mouse)</a>	<a href="#">GRCm39</a> (GCF_000001635.27)	109	2020-09-09	2020-09-22	<a href="#">FTP</a> <a href="#">B</a> <a href="#">AR</a> <a href="#">GDV</a>
<a href="#">Rattus norvegicus (Norway rat)</a>	<a href="#">mRatBN7.2</a> (GCF_015227675.2)	108	2021-01-07	2021-01-21	<a href="#">FTP</a> <a href="#">B</a> <a href="#">AR</a> <a href="#">GDV</a>
<a href="#">Danio rerio (zebrafish)</a>	<a href="#">GRCz11</a> (GCF_000002035.6)	106	2017-06-02	2017-06-26	<a href="#">FTP</a> <a href="#">B</a> <a href="#">AR</a> <a href="#">GDV</a>
<a href="#">Apis mellifera (honey bee)</a>	<a href="#">Amel_HAv3.1</a> (GCF_003254395.2)	104	2018-09-13	2018-09-19	<a href="#">FTP</a> <a href="#">B</a> <a href="#">AR</a> <a href="#">GDV</a>
<a href="#">Zea mays (maize)</a>	<a href="#">Zm-B73-REFERENCE-NAM-5.0</a> (GCF_902167145.1)	103	2020-08-09	2020-09-01	<a href="#">FTP</a> <a href="#">B</a> <a href="#">AR</a> <a href="#">GDV</a>

# Bioinformatics Tools: Genome-wide

**Ensembl** BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog

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

In this section












- Ensembl Variant Effect Predictor
  - VEP web interface
  - VEP command line
  - Data formats
  - Variant Recoder
  - Haplosaurus
  - VEP FAQ
  - Variant Simulator
  - VCF to PED Converter

Search documentation... **Go**

## Ensembl Tools

We provide a number of ready-made tools for processing both our data and yours. We routinely delete results from our servers after 10 days, but if you have an [ensembl account](#) you will be able to save the results indefinitely.

### Processing your data

Name	Description	Online tool	Upload limit
<a href="#">Variant Effect Predictor</a> 	Analyse your own variants and predict the functional consequences of known and unknown variants via our Variant Effect Predictor (VEP) tool.		50MB*
<a href="#">Variant Recoder</a>	Translate a variant identifier, HGVS notation or genomic SPDI notation to all possible variant IDs, HGVS, VCF format and genomic SPDI.		Maximum 1000 variants recommended
<a href="#">BLAST/BLAT</a>	Search our genomes for your DNA or protein sequence.		50MB
<a href="#">File Chameleon</a>	Convert Ensembl files for use with other analysis tools		
<a href="#">Assembly Converter</a>	Map (liftover) your data's coordinates to the current assembly.		50MB
<a href="#">ID History Converter</a>	Convert a set of Ensembl IDs from a previous release into their current equivalents.		50MB
<a href="#">Linkage Disequilibrium Calculator</a>	Calculate LD between variants using genotypes from a selected population.		
<a href="#">VCF to PED converter</a>	Parse a vcf file to create a linkage pedigree file (ped) and a marker information file, which together may be loaded into Id visualization tools like Haploview.		
<a href="#">Data Slicer</a>	Get a subset of data from a BAM or VCF file.		
<a href="#">Post-GWAS</a>	Upload GWAS summary statistics and highlight likely causal gene candidates.		

## IGSR: The International Genome Sample Resource

Supporting open human variation data

[Home](#) [About](#) [Data](#) [Help](#)

## 1KG Project!

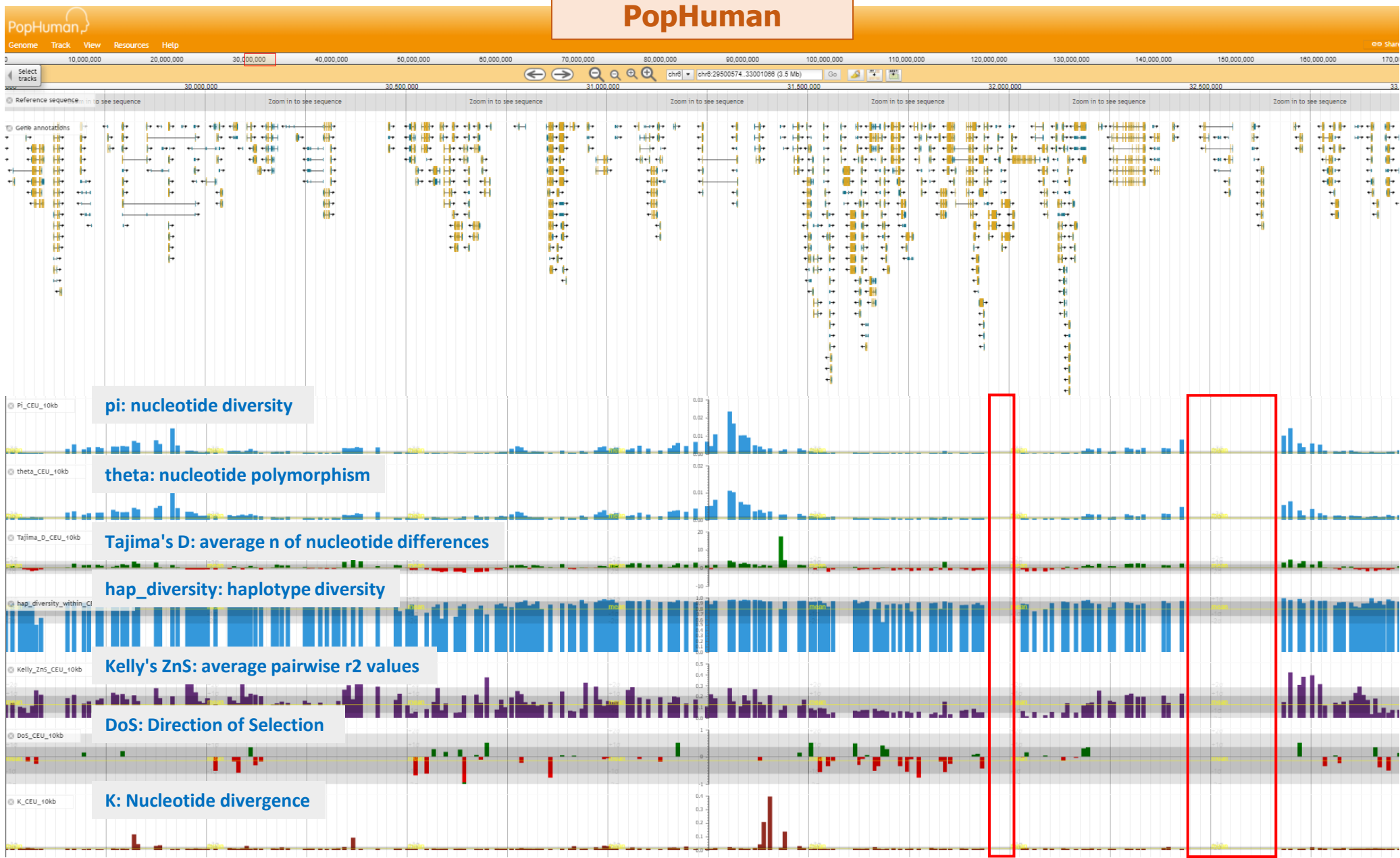
### The International Genome Sample Resource

The 1000 Genomes Project created a catalogue of common human genetic variation, using openly consented samples from people who declared themselves to be healthy.

The International Genome Sample Resource (IGSR) maintains and shares the human genetic variation resources built by the 1000 Genomes Project.

# Bioinformatics Tools: Genome-wide

## PopHuman





# Bioinformatics Tools: Genome-wide

## SNPs

The VarNote web application interface features a navigation bar with links to HOME, APPLICATIONS, UTILS, DOCUMENTATION, JOBS, and UPDATES. Below the navigation bar, there are three tabs: 1. VarNote-REG (selected), 2. VarNote-PAT, and 3. VarNote-CAN. The main content area is titled "Recent Updates:" and contains a list of updates. The first update mentions the introduction of various annotation and prioritization modules. The second update mentions the development of a database called VannoPortal. The third update mentions the implementation of useful online/local toolkits. Below the updates, there is a section titled "VarNote-REG. By integrating 127 Roadmap tissue/cell type-specific epigenomic profiles and 5 tissue/cell type-specific prediction tools (including cchip, GenoSkylinePlus, FUN-LDA, FitCons2, GenoNet), VarNote-REG can efficiently prioritize causal regulatory variants in the LD of each GWAS signal and provide combined scores." Below this section, there is a "Demo result for 161 fine-mapped loci for coronary artery disease (van der Harst and Verweij 2018)" section. The main query area is titled "Query information, required." and contains a "Query" input field with a "Paste query variants (tab-delimited or comma-delimited)" placeholder. Below the input field, there is a "Query Format" section with radio buttons for rsID, VCF, VCF-Like, and Coord-Allele. Below the "Query Format" section, there is a "Data Delimiter" section with radio buttons for TAB and Comma. Below the "Data Delimiter" section, there is a "Reference genome" section with radio buttons for GRCh37/hg19 and GRCh38/hg38. Below the "Reference genome" section, there is an "Upload File" button.

The VannoPortal web application interface features a navigation bar with links to HOME, DOCUMENTATION, FAQ, UPDATES, and CONTACT US. Below the navigation bar, there is a section titled "VannoPortal is a variant annotation database that comprehensively collects and integrates genome-wide variant annotations and prediction scores from various biological domains, including allele frequency, linkage disequilibrium, evolutionary signature, disease/trait association, pathogenesis, allele imbalance, base-wise functional prediction and tissue/cell type-specific functional profile. It greatly expands context-dependent variant annotation to incorporate large-scale epigenomic maps across human tissues/cell types, and compiles many genome-scale base-wise prediction scores for pathogenic/regulatory variant classification beyond protein-coding region. VannoPortal focuses more on interpretability of variant annotations using many intuitive visualizations and interactive web components." Below this section, there is a section titled "GENOME-SCALE HUMAN GENETIC VARIATION ANNOTATION AND INTERPRETATION". Below this section, there is a search bar with the text "rs2395185" and a search button. Below the search bar, there are two radio buttons: GRCh37/hg19 and GRCh38/hg38.

The Open Targets Genetics web application interface features a logo and the text "Open Targets Genetics". Below the logo, there is a search bar with the text "Search for a gene, variant, study, or trait...". Below the search bar, there are four results: PCSK9, 1\_154453788\_C\_T, rs4129267, and LDL cholesterol (Willer CJ et al. 2013). Below the results, there is a note: "Note: genomic coordinates are based on GRCh38". Below the note, there is a section titled "Last updated:" with the text "February 2022 (22.02)".



[https://genetics.opentargets.org/variant/6\\_32465390\\_G\\_T](https://genetics.opentargets.org/variant/6_32465390_G_T)



<http://www.mulinlab.org/vportal/apir.html?q=rs2395185&g=hg38>

<http://www.mulinlab.org/varnote/application.html#REG>



# Bioinformatics Tools: Genome-wide

## SNPs



 **Basic Information** 

Variant Summary

 **Evolution** 

Conservation **1**

Positive Selection **1**

 **Phenotype** 



LD Information **94**

Trait Association


Causal Variant **14**


eQTL **239**


sQTL **207**

 **Regulatory Potential** 

LD Prioritization **94**

Roadmap Epigenomics 



Epimap Epigenomics 

3D Genomes 

Motif Altered **10**

TF Binding **2**

Allele Imbalance

 **Pathogenicity** 

Pathogenicity Score **1**

Missense Pathogenicity


Splicing Alteration

ClinVar

Somatic Recurrence


Oncogenicity Score **1**

Mutation Actionability

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VannoPortal is a variant annotation database that comprehensively collects and integrates [genome-wide variant annotations and prediction scores](#) from various biological domains, including allele frequency, linkage disequilibrium, evolutionary signature, disease/trait association, pathogenesis, allele imbalance, base-wise functional prediction and tissue/cell type-specific functional profile. It greatly expands [context-dependent variant annotation](#) to incorporate large-scale epigenomic maps across human tissues/cell types, and compiles many [genome-scale base-wise prediction scores](#) for pathogenic/regulatory variant classification beyond protein-coding region. VannoPortal focuses more on interpretability of variant annotations using many intuitive visualizations and interactive web components.

### GENOME-SCALE HUMAN GENETIC VARIATION ANNOTATION AND INTERPRETATION



☐ GRCh37/hg19 ☒ GRCh38/hg38

# Bioinformatics Tools: Genome-wide

## eQTLs

### FIVEx: Visualization for Genotypes, Expressions, and Tissues

Currently hosting eQTL and sQTL data from 16 different studies from the [EBI eQTL Catalogue](#)

Search for a variant, region, or gene: chr19:488506, rs10424907, or SHC2



☒ eQTL ☐ sQTL

Search for: **Variant by position:** chr1:109274968 • rsID: rs12740374

**Region:** chr1:108774968-109774968 • **Gene:** SORT1

First time? [View the tutorial here](#) to see what FIVEx can do



## eQTL Catalogue

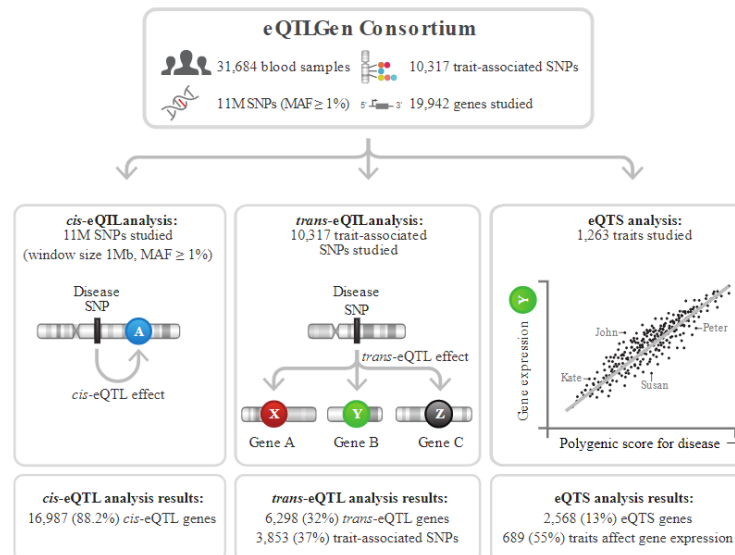
Expression and splicing QTLs recomputed from public datasets

### eQTLGen

[Cis-eQTLs](#) [Trans-eQTLs](#) [eQTS](#) [Replications](#) [Publications](#) [sc-eQTLGen](#)

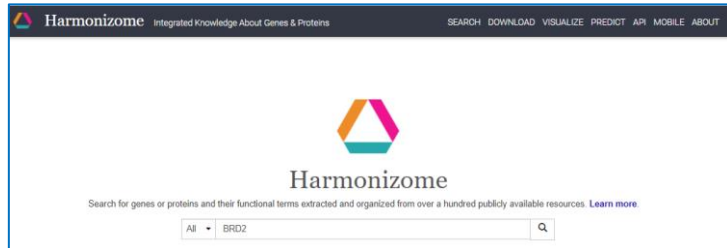
### Welcome to the eQTLGen Consortium

The eQTLGen Consortium has been set up to identify the downstream consequences of trait-related genetic variants. The consortium incorporates 37 datasets, with a total of 31,684 individuals. You can find the [cis-eQTL](#), [trans-eQTL](#), [eQTS](#) and replication results from our forthcoming paper on this website.



# Bioinformatics Tools: Genome-wide

## Genes



### Functional Associations

BRD2 has 4,992 functional associations with biological entities spanning 8 categories (molecular profile, organism, chemical, functional term, phrase or reference, disease, phenotype or trait, structural feature, cell line, cell type or tissue, gene, protein or microRNA) extracted from 80 datasets.

Click the + buttons to view associations for BRD2 from the datasets below.

If available, associations are ranked by **standardized value** ⓘ

	Dataset	Summary
+	<a href="#">Achilles Cell Line Gene Essentiality Profiles</a>	Cell lines with fitness changed by BRD2 gene knockdown relative to other cell lines from the Achilles Cell Line Gene Essentiality Profiles dataset.
+	<a href="#">Allen Brain Atlas Adult Human Brain Tissue Gene Expression Profiles</a>	Tissues with high or low expression of BRD2 gene relative to other tissues from the Allen Brain Atlas Adult Human Brain Tissue Gene Expression Profiles dataset.
+	<a href="#">Allen Brain Atlas Adult Mouse Brain Tissue Gene Expression Profiles</a>	Tissues with high or low expression of BRD2 gene relative to other tissues from the Allen Brain Atlas Adult Mouse Brain Tissue Gene Expression Profiles dataset.
+	<a href="#">Allen Brain Atlas Developing Human Brain Tissue Gene Expression Profiles by Microarray</a>	Tissue samples with high or low expression of BRD2 gene relative to other tissue samples from the Allen Brain Atlas Developing Human Brain Tissue Gene Expression Profiles by Microarray dataset.
+	<a href="#">Allen Brain Atlas Developing Human Brain Tissue Gene Expression Profiles by RNA-seq</a>	Tissue samples with high or low expression of BRD2 gene relative to other tissue samples from the Allen Brain Atlas Developing Human Brain Tissue Gene Expression Profiles by RNA-seq dataset.
+	<a href="#">Allen Brain Atlas Prenatal Human Brain Tissue Gene Expression Profiles</a>	Tissues with high or low expression of BRD2 gene relative to other tissues from the Allen Brain Atlas Prenatal Human Brain Tissue Gene Expression Profiles dataset.
+	<a href="#">BioGPS Cell Line Gene Expression Profiles</a>	Cell lines with high or low expression of BRD2 gene relative to other cell lines from the BioGPS Cell Line Gene Expression Profiles dataset.
+	<a href="#">BioGPS Human Cell Type and Tissue Gene Expression Profiles</a>	Cell types and tissues with high or low expression of BRD2 gene relative to other cell types and tissues from the BioGPS Human Cell Type and Tissue Gene Expression Profiles dataset.
+	<a href="#">BioGPS Mouse Cell Type and Tissue Gene Expression Profiles</a>	Cell types and tissues with high or low expression of BRD2 gene relative to other cell types and tissues from the BioGPS Mouse Cell Type and Tissue Gene Expression Profiles dataset.
+	<a href="#">CCLE Cell Line Gene CNV Profiles</a>	Cell lines with high or low copy number of BRD2 gene relative to other cell lines from the CCLE Cell Line Gene CNV Profiles dataset.
+	<a href="#">CCLE Cell Line Gene Expression Profiles</a>	Cell lines with high or low expression of BRD2 gene relative to other cell lines from the CCLE Cell Line Gene Expression Profiles dataset.
+	<a href="#">CCLE Cell Line Gene Mutation Profiles</a>	Cell lines with BRD2 gene mutations from the CCLE Cell Line Gene Mutation Profiles dataset.
+	<a href="#">CHEA Transcription Factor Binding Site Profiles</a>	Transcription factor binding site profiles with transcription factor binding evidence at the promoter of BRD2 gene from the CHEA Transcription Factor Binding Site Profiles dataset.
+	<a href="#">CHEA Transcription Factor Targets</a>	Transcription factors binding the promoter of BRD2 gene in low- or high-throughput transcription factor functional studies from the CHEA Transcription Factor Targets dataset.
+	<a href="#">CMAP Signatures of Differentially Expressed Genes for Small Molecules</a>	Small molecule perturbations changing expression of BRD2 gene from the CMAP Signatures of Differentially Expressed Genes for Small Molecules dataset.
+	<a href="#">COMPARTMENTS Curated Protein Localization Evidence Scores</a>	Cellular components containing BRD2 protein from the COMPARTMENTS Curated Protein Localization Evidence Scores dataset.
+	<a href="#">COMPARTMENTS Text-mining Protein Localization Evidence Scores</a>	Cellular components co-occurring with BRD2 protein in abstracts of biomedical publications from the COMPARTMENTS Text-mining Protein Localization Evidence Scores dataset.
+	<a href="#">COSMIC Cell Line Gene Mutation Profiles</a>	Cell lines with BRD2 gene mutations from the COSMIC Cell Line Gene Mutation Profiles dataset.

# Bioinformatics Tools: Genome-wide

## Genes

The screenshot shows the GeneCards website homepage. The header includes the GeneCards logo and navigation links for GeneCardsSuite, GeneCards, GeneCaRNA, MalaCards, PathCards, VarElect, GeneAnalytics, GeneALaCart, and GenesLikeMe. A search bar is prominently displayed. The main content area features the text 'GeneCards®: The Human Gene Database' and a description of the database. Below this, there is a section 'Explore a Gene' with a search bar containing 'BRD2' and a 'GO' button. To the right, there are links to 'NGS Analysis' and 'Affiliated Databases' including MalaCards, PathCards, and GeneCaRNA.

The screenshot shows the GeneCards website page for the BRD2 gene. The header is identical to the previous screenshot. The main content area is titled 'BRD2 Gene - Bromodomain Containing 2' and includes a 'Protein Coding (GC06P076900)' label. Below the title, there is a table with various gene-related information. The table has columns for 'Jump to section', 'Aliases', 'Disorders', 'Domains', 'Drugs', 'Expression', 'Function', 'Genomics', 'Localization', and 'Orthologs'. The 'Aliases' column lists 'Paralogs', 'Antibodies', and 'Cell Lines'. The 'Disorders' column lists 'Pathways', 'Assays', and 'Clones'. The 'Domains' column lists 'Products', 'Proteins', and 'Primers'. The 'Drugs' column lists 'Proteins', 'Antibodies', 'Assays', 'Genes', 'shRNA', 'Primers', 'CRISPR', and 'Lentiviral Particles'. The 'Expression' column lists 'Publications', 'CRISPR', and 'Free Bioinformatics Tools'. The 'Function' column lists 'Sources', 'Exp. Assays', and 'miRNA'. The 'Genomics' column lists 'Summaries' and 'Drugs'. The 'Localization' column lists 'Transcripts' and 'Animal Models'. The 'Orthologs' column lists 'Variants' and 'Animal Models'. Below the table, there is a section 'Aliases for BRD2 Gene' with a list of aliases and their corresponding GeneCards symbols. The aliases include 'Bromodomain-Containing Protein 2', 'BRD2 Intronic Transcript 1', 'BRD2-IT1', 'O27.1.1', 'Female Sterile Homeotic-Related Gene 1', 'Bromodomain-Containing 2', 'RNF3', and 'FSH'.

Jump to section	Aliases	Disorders	Domains	Drugs	Expression	Function	Genomics	Localization	Orthologs
Research Products	Antibodies Cell Lines	Assays Clones	Proteins Primers	Inhib. RNA Genotyping	CRISPR	Exp. Assays	miRNA	Drugs	Animal Models

**Aliases for BRD2 Gene**

GeneCards Symbol: **BRD2** <sup>2 3 5</sup>

**Bromodomain Containing 2** <sup>2 3 5</sup>

RING3 <sup>2 3 4 5</sup>

KIAA9001 <sup>2 4 5</sup>

D6S113E <sup>2 3 5</sup>

FSRG1 <sup>2 3 5</sup>

NAT <sup>2 3 5</sup>

Really Interesting New Gene 3 Protein <sup>3 4</sup>

Bromodomain-Containing Protein 2 <sup>3 4</sup>

BRD2 Intronic Transcript 1 <sup>2 3</sup>

BRD2-IT1 <sup>3 5</sup>

O27.1.1 <sup>3 4</sup>

Female Sterile Homeotic-Related Gene 1 <sup>3</sup>

Bromodomain-Containing 2 <sup>2</sup>

RNF3 <sup>3</sup>

FSH <sup>3</sup>

# Bioinformatics Tools: HLA-specific

TISSUE ANTIGENS  
IMMUNE RESPONSE GENETICS

Tissue Antigens ISSN 0001-2815

REVIEW ARTICLE

## Bioinformatic Databases and Resources in the public domain to aid HLA research

W. Helmberg

Department for Blood Group Serology and Transfusion Medicine, Medical University Graz, Graz, Austria

Clinicians Network Payer **Bioinformatics** Search Bioinformatics

NATIONAL MARRIAGE PROGRAM BE THE MATCH! About Us

HLA Resources Search Strategies Policies Contact Us

**Bioinformatics and HLA Expertise**

Find resources and access expertise for immunogenetic-focused research and operational bioinformatics. Access frequently used HLA tools and more.

HaploStats Lookup Tool Allele Code Update  
GTAAGTTGATGAGAGAGAAATGTGT

### Key Areas

Access Popular HLA Resources:

- Allele Code Update to WHO Nomenclature
- Histoimmunogenetics Markup Language (hIML)
- List of Common and Well-Documented (CWD) Alleles v2.0
- MAG Service UI (NMDP allele code creation)

### Haplotype Frequencies

Access helpful resources:

- HaploStats: A Lookup Tool for Haplotype & Haplotype Pair Frequencies
- Haplotype Frequency Estimates
- Be The Match Registry Frequencies

### Learn More

- Contact Us for research support or to request HLA resources
- View our videos: Genetic Ancestry for a Better Match, HLA: Making the Match
- NMCP Network Members can visit the Network section for HLA education, research information, HaploLogic search algorithm, Search Strategy, Advice and more

IPD-IMGT/HLA

Overview IMGT/HLA KIR MHC NHKIR HPA ESTDAB

IPD / IMGT/HLA

## Welcome to IPD-IMGT/HLA

### Release 3.48.0 (2022-04) Version Report - Build 207cdce

The IPD-IMGT/HLA Database provides a specialist database for sequences of the human major histocompatibility complex (MHC) and includes the official sequences named by the [WHO Nomenclature Committee For Factors of the HLA System](#). The IPD-IMGT/HLA Database was originally part of the international ImMunoGeneTics project (IMGT). For more information about the database and what data and tools are available please see our [about](#) page.

**Alignment**

The alignment tool provides access to pre-compiled alignments for individual HLA genes and sequence features

**Alleles**

Query the IPD-IMGT/HLA database for officially named alleles with the allele query tool

**Statistics**

Latest IPD-IMGT/HLA Statistics and Release reports

**Download**

Access to the IPD-IMGT/HLA ftp for downloading sequence files

**Matching**

DPB T-Cell Epitope + HLA-B Leader matching tools

**Cells**

Perform complex queries on the IPD cell database

**Tools**

A complete list of tools for querying the IPD-IMGT/HLA Database

**Submit**

Submit a sequence to the IPD-IMGT/HLA Database

ANTHONY NOLAN saving the lives of people with blood cancer

What we do Help save a life

HLA Nomenclature

ImMunoGeneTics Information system

<http://www.imgt.org>

Allele Frequency Net Database

About us Links Publications Automated Access FAQs Contact

Home Populations HLA KIR Other polymorphisms HLA-ADR KDDB EUROSTAM



# Bioinformatics Tools: HLA-specific

## HLA Data Handling

GENE[RATE] TOOLS

HLA-net > Gene[rate] Tools

May 2021 : Check-out our section with tools for binding and population diversity

### TOOLS FOR HANDLING AND ANALYSING DATA WITH AMBIGUITIES

These programs expect **plain text files in UNIFORMAT v3** as input. A download link to the results file is provided once the execution is completed (on screen and/or by email). Read more about these tools (and see examples of input files) in the [usage overview](#).

Please note that the results files are deleted after 48 hours, therefore if you have not saved your results file you have to run the programs again.

#### File conversions

Converts data files from different formats (tabulated, columnar, pdf files of Luminex reports, Arlequin projects) into UNIFORMAT, and vice versa.

LAUNCH

#### Uniformate

Checks UNIFORMAT files for formatting errors and performs automatic translations of allele and haplotype names (new nomenclature, expansion of valid abbreviations,...).

LAUNCH

#### Basic statistics

Provides all the basic statistics for one and two-locus, including allele and haplotype frequencies, tests for Hardy-Weinberg, neutrality and linkage disequilibrium, and graphs.

LAUNCH

#### Haplotype frequency estimation

Calculates multi-locus haplotype frequencies (two or more loci) or allele frequencies under two alternative models (either Hardy-Weinberg equilibrium or inbreeding-like).

LAUNCH

#### Family phaser

Establishes haplotype phase for family data. Input is UNIFORMAT file with a modified identifier. There are no limits on the number of families included in each file.

LAUNCH

#### Phenotype

Calculates phenotypes from probe lists and reactivity data and returns a UNIFORMAT file.

LAUNCH

TISSUE ANTIGENS  
IMMUNE RESPONSE GENETICS



Tissue Antigens ISSN 0001-2815

#### REVIEW ARTICLE

**The *HLA-net GENE[RATE]* pipeline for effective HLA data analysis and its application to 145 population samples from Europe and neighbouring areas**

J. M. Nunes, S. Buhler, D. Roessli, A. Sanchez-Mazas & the HLA-net 2013 collaboration<sup>†</sup>

# Bioinformatics Tools: HLA-specific

## HLA SPREAD

HLA Web Resource for **S**NPs, **P**opulations, **R**esources, **A**DRs, **D**iseases

[Home](#)[Visualization](#)[Search](#)[Team](#)[Workflow](#)

2910

Diseases

689

HLA alleles

163

Adverse Drug Reactions

286

Biomarker reports

### HLA-SPREAD: A PLATFORM FOR HLA RESOURCES (HLA- SNPs Populations REsources ADRs Diseases)



**DATA RETRIEVAL**  
Parsing of MEDLINE Abstracts  
Data Compilation



**KEYWORDS DICTIONARY**  
IPD-IMGIT/HLA Database for Alleles  
UMLS (MeSH ID) for Diseases



**NAMED ENTITY RECOGNITION**  
Keyword Matching in Sentences  
Tags: Populations, Drugs & SNPs



**SEMANTIC ASSESSMENT**  
Relation between Entities  
Hybrid Approach: N-GRAM & Bio-Verbs



**DATA ANALYSIS**  
HLA Alleles in Disease  
Population Frequencies  
SNPs & ADR Associations



**GUI & DATABASE**  
Integration of Relational Database  
Advanced HLA Search

Dholakia et al. BMC Genomics (2022) 23:10  
<https://doi.org/10.1186/s12864-021-08239-0>

BMC Genomics

DATABASE

Open Access

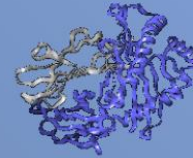
HLA-SPREAD: a natural language processing based resource for curating HLA association from PubMed abstracts

Dhwani Dholakia<sup>1,2\*</sup>, Ankit Kalra<sup>3†</sup>, Bishnu Raman Misir<sup>4</sup>, Uma Kanga<sup>5</sup> and Mittali Mukerji<sup>1,4\*</sup>

# Bioinformatics Tools: HLA-specific

## HLA3D

Home Statistics Analysis Submission Help Contact



Please choose a pipeline for prediction

Risk Alignment for Transplant

Antigenic Peptides Prediction



### Key Points

- To provide comprehensive analysis of HLA for different populations, we developed HLA3D, a comprehensive toolkit that collected 1296 sequences, 256 PDB structures, 212 modelled structures, 120 000 frequency data, 73 000 associated literature, 39 000 disease-associated SNP of HLA and 1604 oncogenic mutations.
- Based on common HLA molecules in HLA3D, we qualified the HLA structure differences and obtained 100 000 RMSD records. In addition, we predicted 370 000 antigenic peptides with high affinity in common tumours, which helps to narrow the range of candidate neoantigens and promote subsequent experimental verification.
- By integrating the knowledge of differential immunogenicity in HLA sequences and structures, HLA3D established a risk alignment pipeline, providing users with the functions of structure alignment, sequence alignment, 3D-View and risk report, to help users predict the severity of aGVHD of mismatch HLA donors before transplantation.
- In view of the key characteristics that affect the immunogenicity of mutated peptides, HLA3D established an antigenic peptide prediction pipeline to provide users with a series of applications, such as mutation prediction, peptide prediction, immunogenicity assessment and docking simulation, to help users complete the prediction and analysis of immunogenic peptides.



"in press"

Briefings in Bioinformatics, 2022, 1–13  
<https://doi.org/10.1093/bib/bbac076>  
 Problem Solving Protocol

HLA3D: an integrated structure-based computational toolkit for immunotherapy

Xingyu Li<sup>1</sup>, Xue Lin<sup>1</sup>, Xueyin Mei, Pin Chen, Anna Liu, Weicheng Liang<sup>2</sup>, Shan Chang<sup>2</sup> and Jian Li<sup>1</sup>

# Bioinformatics Tools: HLA-specific

[Easy-HLA](#)[Language](#)[Register](#)[Login](#)

# Easy-HLA

[EasyMatch-R](#)[HLA-Upgrade](#)[HLA-2-Haplo](#)[HLA-Epi](#)

EasyMatch-R

HLA-Upgrade

HLA-2-Haplo

Epitopes

For research use only

EasyMatch-R is a tool designed to find unrelated donors in silico. We aim to improve efficiency by saving time and resources in HLA labs. From a patient's HLA typing in input, Easymatch-R displays probability of finding a potential unrelated bone marrow donor according to the required matching level. In addition to the number of expected donors, Easymatch-R provides complementary typings recommendations.

HLA-Upgrade uses the power of haplotypes to predict full high resolution HLA-A, B, C, DRB1, DQB1 genotypes.

HLA-2-Haplo predicts pairs of haplotypes from HLA genotypes.

Compare donors and recipient in terms of epitopic HLA compatibilities

*Bioinformatics*, 36(7), 2020, 2157–2164  
doi: 10.1093/bioinformatics/btaz675  
Advance Access Publication Date: 21 November 2019  
Original Paper

Genetics and population analysis  
**Easy-HLA: a validated web application suite to reveal the full details of HLA typing**  
Estelle Geffard<sup>1,\*</sup>, Sophie Limou<sup>1</sup>, Alexandre Walencik<sup>1,2</sup>, Michelle Daya<sup>3</sup>, Harold Watson<sup>4</sup>, Dara Torgerson<sup>5</sup>, Kathleen C. Barnes on behalf of CAAPA<sup>3</sup>, Anne Cesbron Gautier<sup>2</sup>, Pierre-Antoine Gourraud<sup>1,\*</sup> and Nicolas Vince<sup>1</sup>

Received: 6 January 2020 | Reviewed: 19 June 2020 | Accepted: 3 July 2020  
DOI: 10.1002/gp.1.22134

RESEARCH ARTICLE

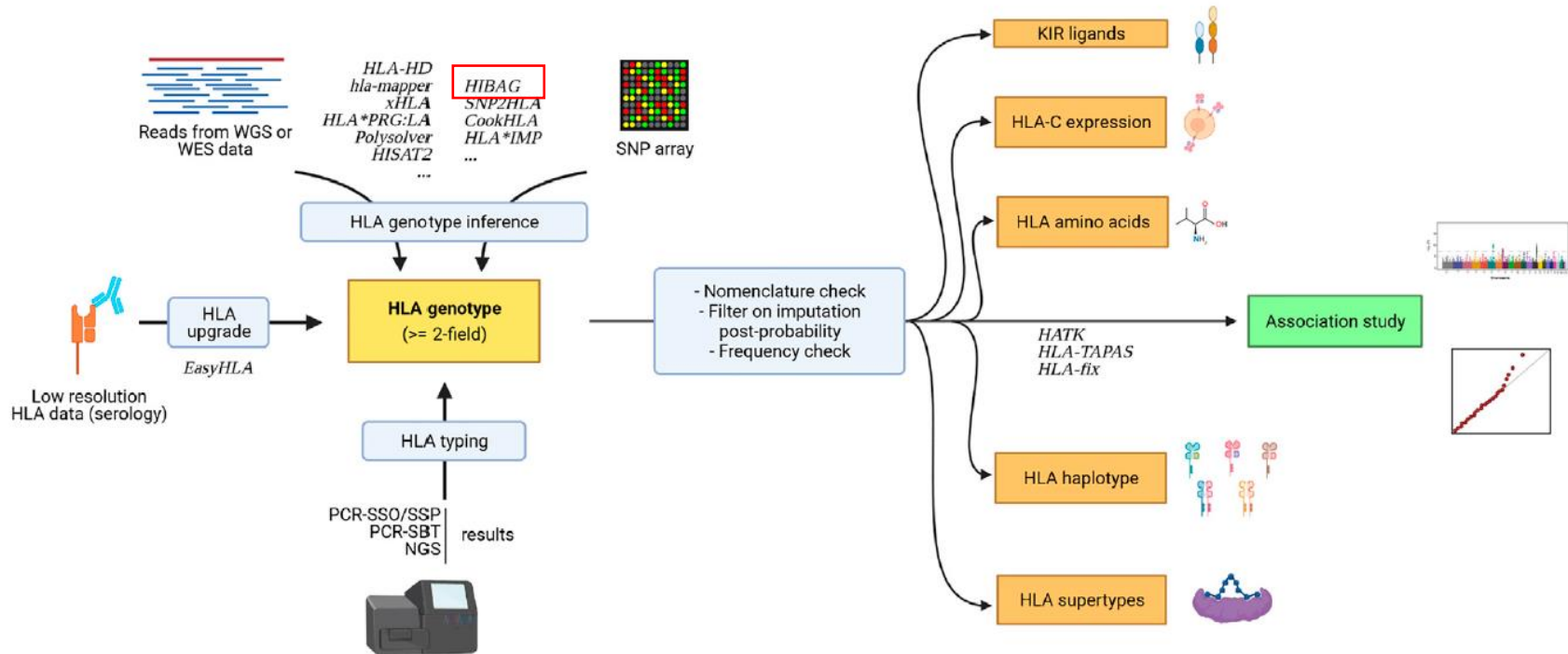
Genetic Epidemiology

OFFICIAL JOURNAL  
INTERNATIONAL GENETIC  
EPIDEMIOLOGY SOCIETY  
wileyonlinelibrary.com/genetic

**SNP-HLA Reference Consortium (SHLARC): HLA and SNP data sharing for promoting MHC-centric analyses in genomics**

Nicolas Vince<sup>1</sup> | Venceslas Douillard<sup>1</sup> | Estelle Geffard<sup>1</sup> | Diogo Meyer<sup>2</sup> | Erick C. Castelli<sup>3</sup> | Steven J. Mack<sup>4</sup> | Sophie Limou<sup>1,5</sup> | Pierre-Antoine Gourraud<sup>1</sup>

# Bioinformatics Tools: HLA-specific



**FIGURE 3 |** Association study pipeline for HLA data and surrounding immunogenetic factors. Created with biorender.com.

## Approaching Genetics Through the MHC Lens: Tools and Methods for HLA Research

Venceslas Douillard<sup>1</sup>, Erick C. Castelli<sup>2</sup>, Steven J. Mack<sup>3</sup>, Jill A. Hollenbach<sup>4,5</sup>, Pierre-Antoine Gourraud<sup>1</sup>, Nicolas Vince<sup>1,6</sup> and Sophie Limou<sup>1,6,7</sup> on behalf of the Covid-19/HLA & Immunogenetics Consortium and the SNP-HLA Reference Consortium

# Bioinformatics Tools: HLA-specific

## HLA-IMPUTER: HLA Allele Imputation via population specific HIBAG models

ImputeHLA Impute Association

### Follow 4 Simple Steps:

1. Upload plink genotypes files in hg19

#### Choose bed File

Browse... No file selected

#### Choose fam file

Browse... No file selected

#### Choose bim file

Browse... No file selected

### 2. Choose Reference Panel to Use

Han Chinese

Han Chinese

Pan Asian

European

Multiethnic

### 4. Impute Alleles

Input Email to receive results

5. Load Data

### Preview of Genotypes Loaded

### Predicted Alleles

Best Predicted HLA Alleles

[Posterior Probabilities Of All HLA Alleles](#)

Download Best Predicted

Download All Predicted

*Bioinformatics*, 35(7), 2019, 1244–1246  
doi: 10.1093/bioinformatics/bty730  
Advance Access Publication Date: 31 August 2018  
Applications Note

OXFORD

Genetics and population analysis

### HLA-IMPUTER: an easy to use web application for HLA imputation and association analysis using population-specific reference panels

Jiangshan J. Shen<sup>1,†</sup>, Chao Yang<sup>2,†</sup>, Yong-Fei Wang<sup>1</sup>, Ting-You Wang<sup>1</sup>, Mengbiao Guo<sup>1</sup>, Yu Lung Lau<sup>1</sup>, Xuejun Zhang<sup>2</sup>, Yujun Sheng<sup>2,\*</sup> and Wanling Yang<sup>1,\*</sup>



# Bioinformatics Tools: HLA-specific

TABLE 1 | Tools for HLA analyses.

HLA application name	Description	URL
Alphard-nt (Hayashi et al., 2019)	Identification of somatic mutations in HLA molecules from whole-genome and exome data using Bayesian algorithms	—
BIGDAWG (Pappas et al., 2016)	Open-source R package for the case-control analysis of highly polymorphic data at the allele, haplotype and amino-acid level	<a href="https://CRAN.R-project.org/package=BIGDAWG">https://CRAN.R-project.org/package=BIGDAWG</a>
Easy-HLA (Geffard et al., 2020)	Website with HLA alleles haplotyping, upgrading and inference from HLA genotypes, prediction of HLA-C expression	<a href="http://hla.univ-nantes.fr/">http://hla.univ-nantes.fr/</a>
HATK (Choi et al., 2021)	Open-source <i>Python</i> pipeline for HLA association studies, including tools for HLA data formatting	<a href="https://github.com/WansonChoi/HATK">https://github.com/WansonChoi/HATK</a>
HLA-check (Jeanmougin et al., 2017)	Perl tool evaluating the probability of accurate HLA genotype imputation by comparing it to SNP imputation in the exonic region of HLA.	<a href="https://github.com/mcleggrand/HLA-check/">https://github.com/mcleggrand/HLA-check/</a>
HLA-EMMA (Kramer et al., 2020)	Donor/recipient compatibility assessment based on solvent-accessible amino acids, based on intralocus comparisons	<a href="http://www.HLA-EMMA.com">http://www.HLA-EMMA.com</a>
HLAfix	Open-source R pipeline for HLA association studies. Performing SNP quality control steps, stratification, HLA imputation and representation of the results	<a href="https://univ-nantes.io/Nico_V/hlafix">https://univ-nantes.io/Nico_V/hlafix</a>
HLAHapV (Osoegawa et al., 2016)	A Java-based HLA Haplotype Validator for quality assessments of HLA typing	<a href="https://github.com/nmdp-bioinformatics/ImmunogeneticDataTools">https://github.com/nmdp-bioinformatics/ImmunogeneticDataTools</a>
HLA-NET (Nunes et al., 2014)	Set of tools to manipulate HLA data, infer haplotypes, convert files format, and information about typing	<a href="https://hla-net.eu/">https://hla-net.eu/</a>
HLApers (Aguiar et al., 2020)	Genotyping and quantification of HLA expression from RNA-seq data	<a href="https://github.com/genevol-usp/HLApers">https://github.com/genevol-usp/HLApers</a>
HLA-TAPAS (Luo et al., 2020)	Open-source <i>Python</i> pipeline for creation of reference panels and HLA association studies	<a href="https://github.com/immunogenomics/HLA-TAPAS">https://github.com/immunogenomics/HLA-TAPAS</a>
MergeReference (Cook and Han, 2017)	SNP2HLA compatible tool to concatenate multiple reference panels in order to gain accuracy during HLA imputation	<a href="http://software.buhmhan.com/MergeReference">http://software.buhmhan.com/MergeReference</a>
pyHLA (Fan and Song, 2017)	Association analysis for HLA alleles in <i>Python</i> language	<a href="https://github.com/felixfan/PyHLA">https://github.com/felixfan/PyHLA</a>



## Approaching Genetics Through the MHC Lens: Tools and Methods for HLA Research

Venceslas Douillard<sup>1</sup>, Erick C. Castelli<sup>2</sup>, Steven J. Mack<sup>3</sup>, Jill A. Hollenbach<sup>4,5</sup>, Pierre-Antoine Gourraud<sup>1</sup>, Nicolas Vince<sup>1,6</sup> and Sophie Limou<sup>1,6,7</sup> on behalf of the Covid-19/HLA & Immunogenetics Consortium and the SNP-HLA Reference Consortium


# Bioinformatics Tools: HLA-specific

## R Packages

### MiDAS—Meaningful Immunogenetic Data at Scale

Maciej Migdal, Dan Fu Ruan, William F. Forrest, Amir Horowitz, Christian Hammer 

### HATK: HLA analysis toolkit

Wanson Choi, Yang Luo, Soumya Raychaudhuri, Buham Han 

*Bioinformatics*, Volume 37, Issue 3, 1 February 2021, Pages 416–418,  
<https://doi.org/10.1093/bioinformatics/btaa684>

[Home](#) » [Bioconductor 3.14](#) » [Software Packages](#) » immunotation

## immunotation

platforms **all** rank **1967 / 2083** support **0 / 0** in Bioc **1 year**  
build **ok** updated **before release** dependencies **68**

DOI: [10.18129/B9.bioc.immunotation](https://doi.org/10.18129/B9.bioc.immunotation)  

### Tools for working with diverse immune genes

Bioconductor version: Release (3.14)

MHC (major histocompatibility complex) molecules are cell surface complexes that present antigens to T cells. The repertoire of antigens presented in a given genetic background largely depends on the sequence of the encoded MHC molecules, and thus, in humans, on the highly variable HLA (human leukocyte antigen) genes of the hyperpolymorphic HLA locus. More than 28,000 different HLA alleles have been reported, with significant differences in allele frequencies between human populations worldwide. Reproducible and consistent annotation of HLA alleles in large-scale bioinformatics workflows remains challenging, because the available reference databases and software tools often use different HLA naming schemes. The package immunotation provides tools for consistent annotation of HLA genes in typical immunoinformatics workflows such as for example the prediction of MHC-presented peptides in different human donors. Converter functions that provide mappings between different HLA naming schemes are based on the MHC restriction ontology (MRO). The package also provides automated access to HLA allele frequencies in worldwide human reference populations stored in the Allele Frequency Net Database.

Author: Katharina Imkeller [cre, aut]



# Bioinformatics Tools

[Genome Biology](#) [Genetics](#) [Biostatistics](#) [R](#) [Population Genetics](#) [Genetic Epidemiology](#) [HLA](#) [MHC](#) [Homepage](#)

## BIOINFORMATICS TOOLS

*(for Genetic Epidemiologists)*

Mehmet Tevfik DORAK, MD, PhD

### *Open Access Reviews / Journals*

ENCODE Resources ([Pazin MJ. CSHP 2015](#)); Database Tools ([Bianco, Genomics 2013](#); [Zou, 2015](#)); UCSC Browser ([Mangan, 2014](#))

*Journals:* [Nucleic Acids Research \(NAR\)](#); [NAR Genomics & Bioinformatics](#); [Database; Bioinformatics](#)

[Nucleic Acids Research Databases Catalog](#)

[Scientific Data](#)

### *Article Collections*

[ENCODE \(Web Focus\)](#) [FANTOM5 \(RIKEN\)](#) [IHEC](#) [Epigenome RoadMap](#) [TCGA](#) [GTEx](#)

[F1000Research: Bioconductor](#)

### *Societies*

[ISCB](#) [ISMB](#) [AMIA](#) [ECCB](#)

### *Online Suites*

[Galaxy \(101 - Support\)](#) [ExPASy \(ref\)](#) [Taverna 2.5.0/Archive \(ref\)](#) [Apache-Taverna](#) [BioMart \(ref\)](#) [Artemis \(manual\)](#) [Babelomics \(ref / tutorial\)](#)

[Bioconductor \(manual\)](#) [BioPython \(tutorial & cookbook / PDF\)](#)

[EDGE: Empowering the Development of Genomics Expertise \(ref\)](#)

### *Biomarts*

[BioTools: Biomart](#)

[Ensembl BioMart \(YouTube tutorial\)](#) [UCSC Table Browser](#) [GWAS Central GWAS Mart \(tutorial\)](#) [SPS Mart \(allele freqs\)](#) [Fantom5](#)

### *Genome Browsers*

[BioTools: Genome Visualization](#)

[UCSC Human Genome Bioinformatics](#) [GENCODE](#) [FANTOM5 \(ref / tutorial / wiki / user guide \(pp.12-19\)\)](#) - [ZENBU - SLIDEBASE \(tutorial\)](#)

[Ensembl Genome Browser \(Human\)](#) [Vega Genome Browser \(Human\)](#)

[NIH RoadMap Epigenomic Browser \(tutorial\)](#) [WashU EpiGenome](#) [Human Epigenome Atlas](#) [NCBI Epigenomics \(help\)](#) [NGSmethDB \(ref\)](#)

[1KG \(Browser - Quick Start Guide\)-\(NCBI\)](#) [PopHuman \(ref\)](#)

[UK10K \(ref\)](#) [GENCODE Browser](#) [SNIIPA](#) [\(gnomAD\) genome Aggregation Browser \(ref\)](#)

[CNV Browser](#) [DECIPHER](#) [eQTL Browser](#) [seeQTL](#) [GTEx eQTL Browser](#)

[IncRBase](#) [NonCode](#) [FANTOM CAT \(lncRNA Atlas\) Browser \(ref\)](#) [Integrative Genomics \(ref\)](#)

[VISTA Enhancer Browser](#) [Swiss Regulon](#) [Reactome Pathway Browser](#) [ImmunoBase Browser](#) [ImmunPop Browser \(ref\)](#)

[NCBI Genome Data Viewer \(GDV\) \(ref / tutorial\)](#)

### *General Resources*

[BioTools](#) [OmicX](#)

[NCBI Global Search](#) [GoPubMed](#) [HuGE](#) [LabMeeting](#) [NCBI Tutorials / Handbook / Help Manual](#) [NIH Helix](#) [NCBI Tools for Data Mining](#) [NCI Data Analysis Tools](#)

[Sanger: Software / Databases & Tools](#) [EMBL-EBI](#) [Swiss Institute of Bioinformatics: ExPASy](#) [JJ Wang Lab](#)

[NCBI Gene .. GeneCards .. GeneMap .. GenAtlas .. iHOP .. BioGPS Gene Annotation .. Harmonizome \(ref\)](#)

[NCBI \(tutorials on YouTube\) .. BLAST .. MimicMe \(ref\) .. e-PCR .. In Silico PCR .. GeneNote .. GeneLoc .. CGAP-GAL .. Aceview](#)

[ENCODE \(ref / tutorial1 / tutorial2 / cell types\)](#) [GENCODE](#) [RegulomeDB](#) [FANTOM Gateway \(papers / software\)](#) [Broad Institute Portals](#)

[Database of Genomic Variants \(CNV\)](#) [CNV inspector](#) [Copy Number Variation Resources](#) [DECIPHER](#)

[OMIM .. OMIM Map .. NCBI Genetic Association Database \(GAD\) \[Now Retired\] - HuGE Navigator .. Variant N](#)

[Gene Exp Omnibus -GEO \(NCBI\) .. NCBI dbGaP .. ArrayExpress \(EBI\) .. SOURCE \(genomics tool\)](#)

<http://www.dorak.info/mtd/bioinf.html>

# **Bioinformatics Tools for HLA**

**The amount of biological data related to the HLA genes and HLA region is ever increasing**

**Cell/tissue-specific, validated, high quality data should be exploited at every opportunity to find answers to research questions**

**To do that, user friendly tools and apps are available**

**There is, however, no substitute for expert advice from a trained bioinformatician**

**Experimental verification is also still needed**

***Thank You***

