

BIPMed – New Projects and Advances

www.bipmed.org



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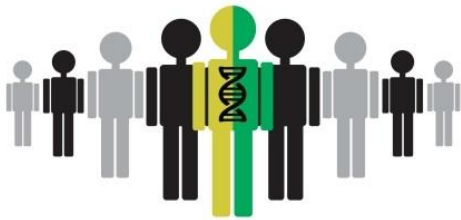
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Databases



- Leiden Open Variation Database 3.0
- Web-based gene sequence variation database
- API enables simple queries or listing of gene and variant data
- Freely available tool for Gene-centered collection and display of DNA variations.



BIPMed Brazilian Initiative on
PRECISION MEDICINE

Databases

List of public LOVD installations



If you are looking for a specific gene database, please also check our [list of LSDBs](#).

Please note that the latest available build is always installed on our [Leiden server](#). We offer free use of this LOVD installation interested, e.g. to maintain/start a LSDB. [Contact us](#) for more information.

Show only LOVDs with gene symbol:



You can also [download this list](#) to see the complete list of genes in the databases.

In total: 3,913,937 variants (2,943,353 unique) in 424,351 individuals in 85 LOVD installations.

http://databases.lovd.nl/whole_genome/

LOVD 3.0-20a

22002 genes

Whole genome datasets

A1BG,A1BG-AS1,A1CF,A2M,A2M-AS1,A2ML1,A2MP1,A4GALT,A4GNT,A...

<http://bipmed.igm.unicamp.br/>

LOVD 3.0-17

20930 genes

BIPMed WES

A1BG,A1BG-AS1,A1CF,A2M,A2M-AS1,A2ML1,A2MP1,A3GALT2,A4GALT...

<https://databases.lovd.nl/shared/>

LOVD 3.0-20b

22977 genes

Shared database

A1BG,A1BG-AS1,A1CF,A2LD1,A2M,A2M-AS1,A2ML1,A2MP1,A3GALT2,...

<http://proteomics.bio21.unimelb.edu.au/lovd/>

LOVD 3.0-07

14772 genes

LOVD - Leiden Open Variation Database

A1BG,A1BG-AS1,A1CF,A2M,A2ML1,A4GALT,A4GNT,AAAS,AACS,AACSP...

<http://bipmed.igm.unicamp.br/cfa/>

LOVD 3.0-20

41 genes

Craniofacial anomalies

APOC2,APOC4-APOC2,AXIN2,BCL3,BMP4,CLPTM1,DVL2,ERBB2,FGF22...

<https://ab-openlab.csiir.res.in/mitoilsdb/>

LOVD 2.0-35

37 genes

MitoLSDB

MTATP6,MTATP8,MTCO1,MTCO2,MTCO3,MTCYB,MTND1,MTND2,MTND3,M...



Mirrors

BIPMed has 4 installations

- Main site: IQ – Unicamp - Campinas
- Mirror 1: FCM – Unicamp - Campinas
- Mirror 2: USP – Ribeirão Preto
- Mirror 3: UNIFESP – São Paulo



BIPMed

Our Products:

- Genomic databases:
 - BIPMed-WES-db: 256 individuals **REFERENCE POPULATION**
 - BIPMed-Array-db: 264 individuals **REFERENCE POPULATION**
 - **DISEASE SPECIFIC DATABASES**
- BIPMed Beacon
- GA4GH R client
-

Disease/Phenotype specific projects

- Epilepsy (BRAINN, ILAE-ALADE) – EE
 - DOOSE SYNDROME
 - DRAVET SYNDROME
- Craniofacial Anomalies – Dr. Vera Lucia Gil Da Silva Lopes
 - Cleft lip and palate (BCFP)
- BRCA Challenge (GA4GH, HVP) – Dr. Patricia Prolla and Edenir Palmero



**BRCA
EXCHANGE**

BRCA Challenge

- Dr. Patricia Prolla, Dr. Edenir Palmero and Dr. Maria Aparecida Folgueira
- Supported by GA4GH, LOVD and the Human Variome Project
 - Understanding of the genetic basis of breast cancer, ovarian cancer and other diseases
 - Pooling data on BRCA1/2 genetic variants and corresponding clinical data from around the world.
- The BRCA Challenge on BIPMed
 - **75 variants in BRCA1**
 - **87 variants in BRCA2**



Craniofacial Anomalies

- Dr. Vera Lucia Gil Da Silva Lopes
- 253 variantes in 41 genes
- 358 Healthy individuals with no history of oral clefting in the past three generations
- 157 patients with Non-syndromic Cleft Lip and Palate



Under development

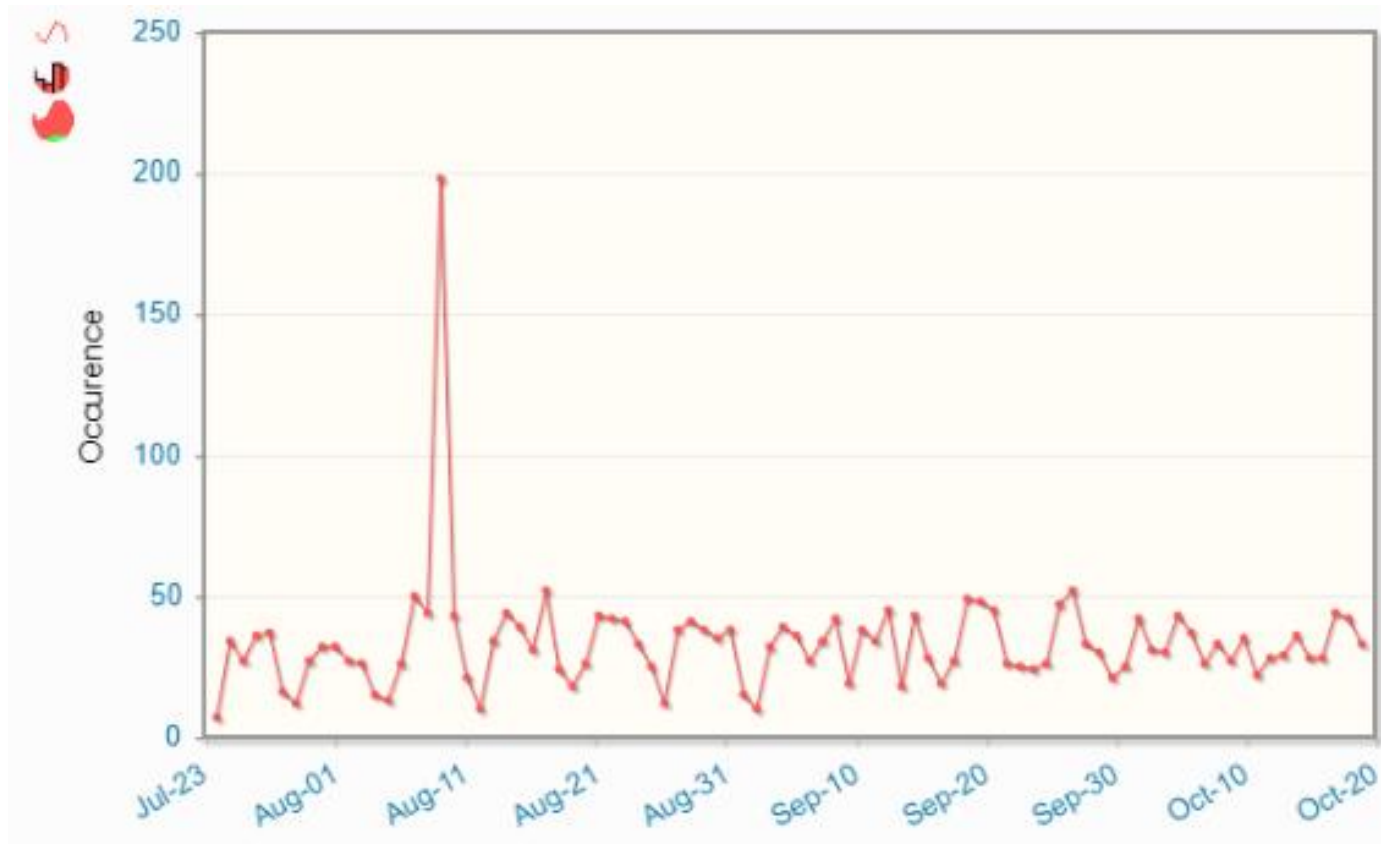
- Copy number databases;
- Craniofacial anomalies SNP Array
 - Affymetrix Genome-Wide Human SNP Array 6.0;
- Stroke (BRAINN, ISGC-Latin American Initiative);
- Pathogenic hemoglobins – Global Globin (HVP) – Dr. Monica Melo;
- ApoE Challenge (BRAINN, ABN) – Dr. Marcio Balthazar;



Under development

- 40 reference individuals born in Ribeirão Preto region with more than 65 years old – Prof. Wilson Silva Junior
 - Whole Exome Sequencing;
 - Affymetrix Genome-Wide Human SNP Array 6.0
- 40 reference individual born in Barretos region – Prof. Vinicius Vasquez
 - Whole Exome Sequencing;
 - Affymetrix Genome-Wide Human SNP Array 6.0

Access Statistics

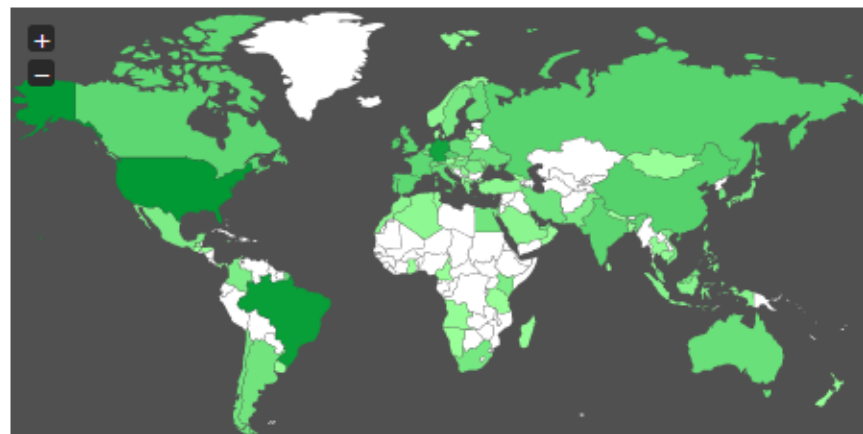
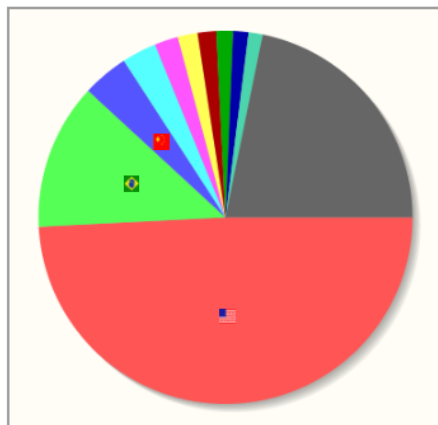




BIPMed Brazilian Initiative on **PRECISION MEDICINE**

Access Statistics

Countries	
United States -	889 49.1 %
Brazil -	229 12.6 %
China -	72 3.9 %
India -	54 2.9 %
Russian Federation -	37 2.0 %
France -	31 1.7 %
Great Britain -	29 1.6 %
Germany -	26 1.4 %
Philippines -	24 1.3 %
Canada -	22 1.2 %
Others	394 21.8 %



Requests Accesses Hosts Traffic

	Hosts	Percentage	Requests	Percentage	Accesses	Percentage	Traffic (Mb)	Percentage
Europe	305	16.9 %	16786	32.4 %	12162	47.4 %	232.7	45.2 %
South-America	252	13.9 %	14412	27.8 %	7307	28.5 %	97.9	19.0 %
North-America	918	50.8 %	18132	35.0 %	5350	20.9 %	152.1	29.5 %
Central-America	7	0.4 %	7	0.0 %	7	0.0 %	0.3	0.1 %
Africa	40	2.2 %	245	0.5 %	70	0.3 %	4.0	0.8 %
Oceania	17	0.9 %	193	0.4 %	69	0.3 %	1.9	0.4 %
Asia	267	14.8 %	2067	4.0 %	1686	2.7 %	26.0	5.0 %
Misc	1	0.1 %	1	0.0 %	1	0.0 %	0.0	0.0 %

LatinGen



- Development of the portal **latingen.org**
- Disseminate the idea of federated databases
- Act as a centralizer of genomic information in Latin America
- Provide assistance for other research groups

contact@latingen.org

LatinGen



- **Future projects:**
 - Develop integration of participants databases
 - Projects using the LOVD interface provide easy integration
 - Projects with other database structure can be challenging
 - Provide bioinformatics training in Latin America



Acknowledgment





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Visit us at

www.bipmed.org