UNICAMP BRAIN
BRAZILIAN INSTITUTE OF NEUROSCIENCE AND NEUROTECHNOLOGY

FAPESP

PIBMed - Brazilian Initiative on Precision Medicine
Summary of results for the first year

BIPMed Brazilian Initiative on PRECISION MEDICINE

Dra. Cristiane Rocha

BIPMed

Our Products:

- Genomic databases:
  - BIPMed-WES-db
  - BIPMed-Array-db
- BIPMed Beacon
- GA4GH R client

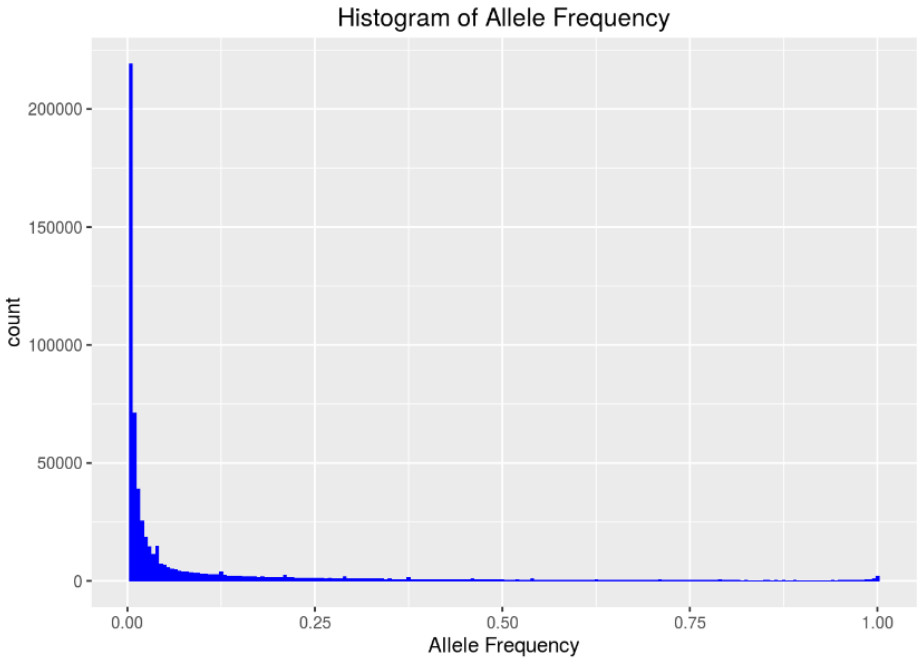
Genomic Databases

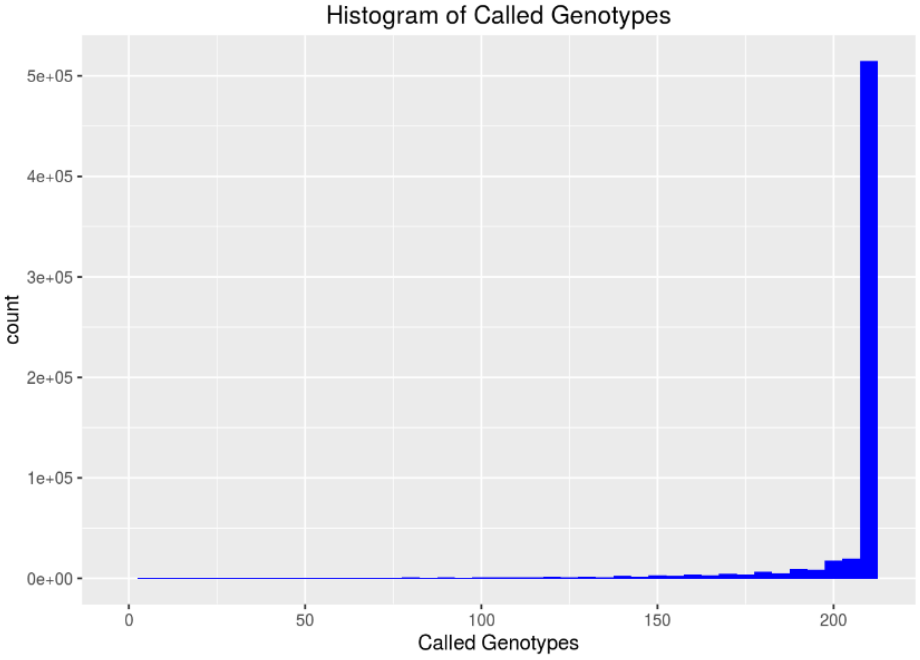
BIPMed-WES-db

- Whole Exome Sequencing experiments from 106 Brazilian Reference individuals
- Use Leiden Open Variation Database (LOVD) interface
- Open database at http://bipmed.iqm.unicamp.br
- Over 600.000 different variants

Genomic Databases

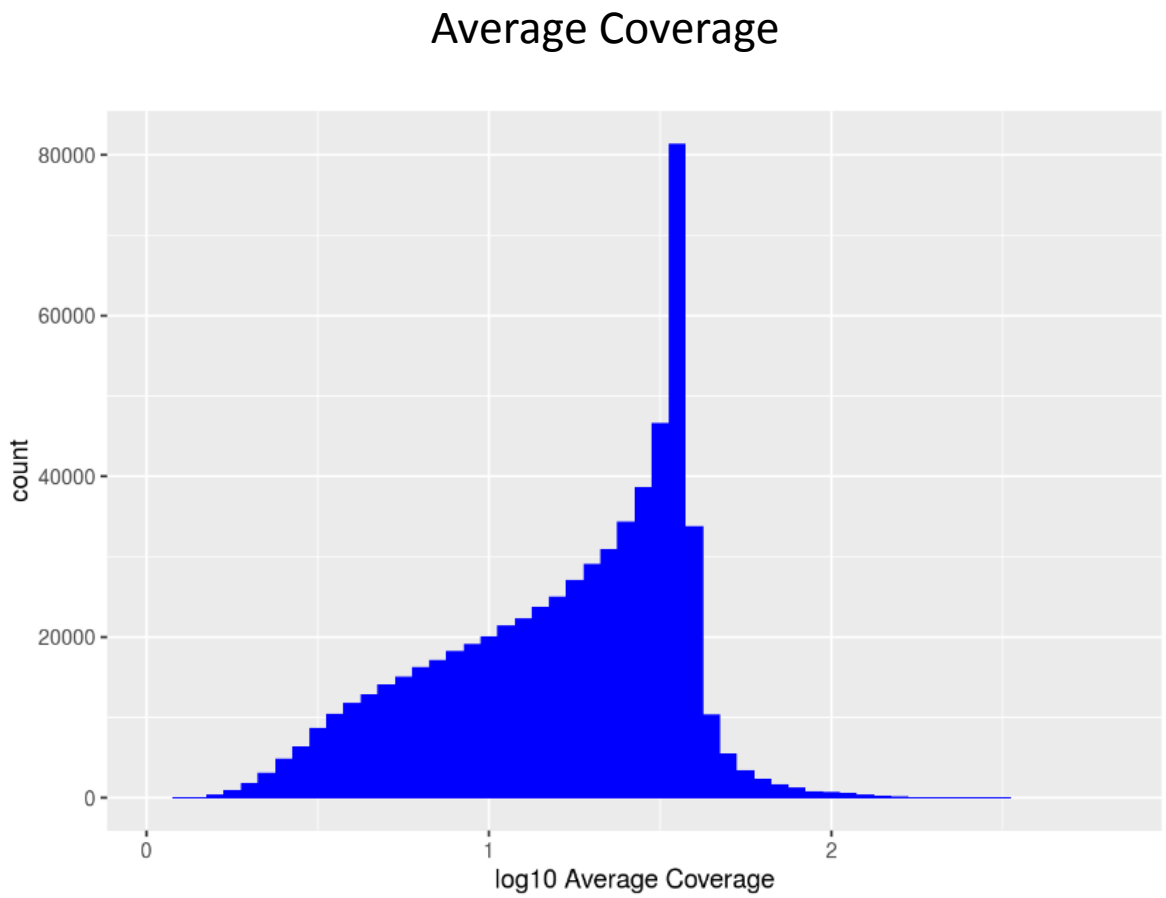
BIPMed-WES-db





Genomic Databases

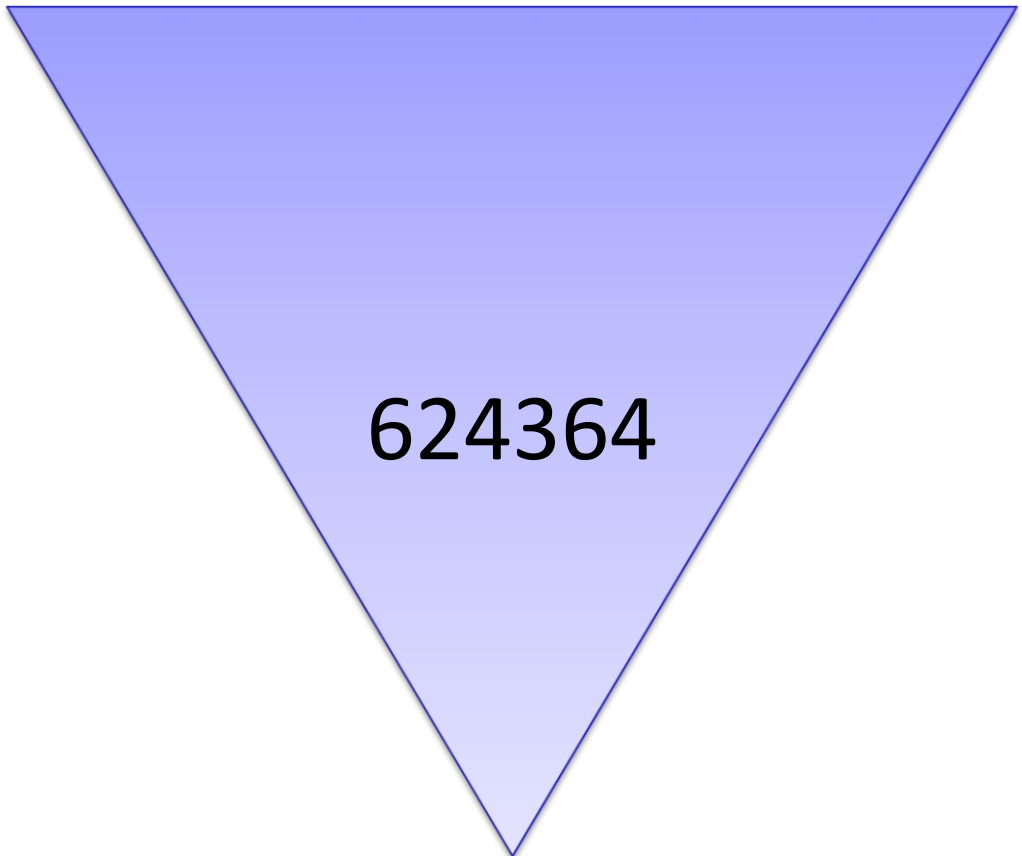
BIPMed-WES-db



Genomic Databases

BIPMed-WES-db

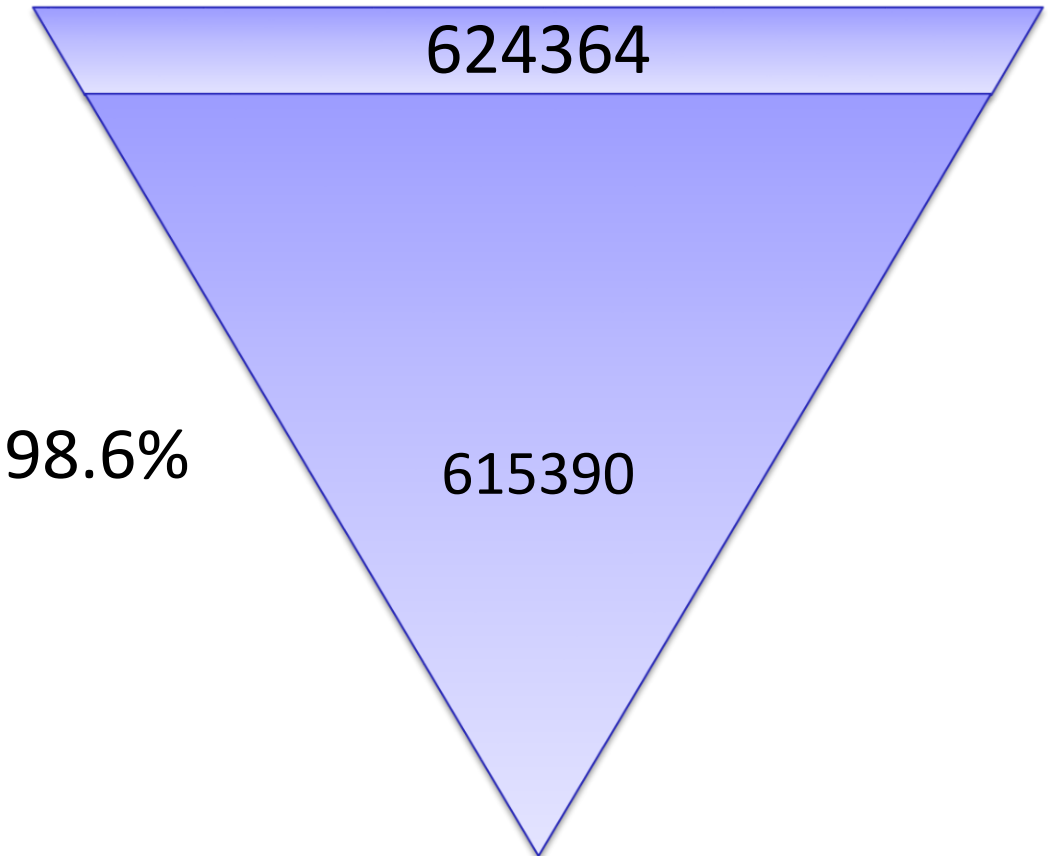
Total variants



Genomic Databases

BIPMed-WES-db

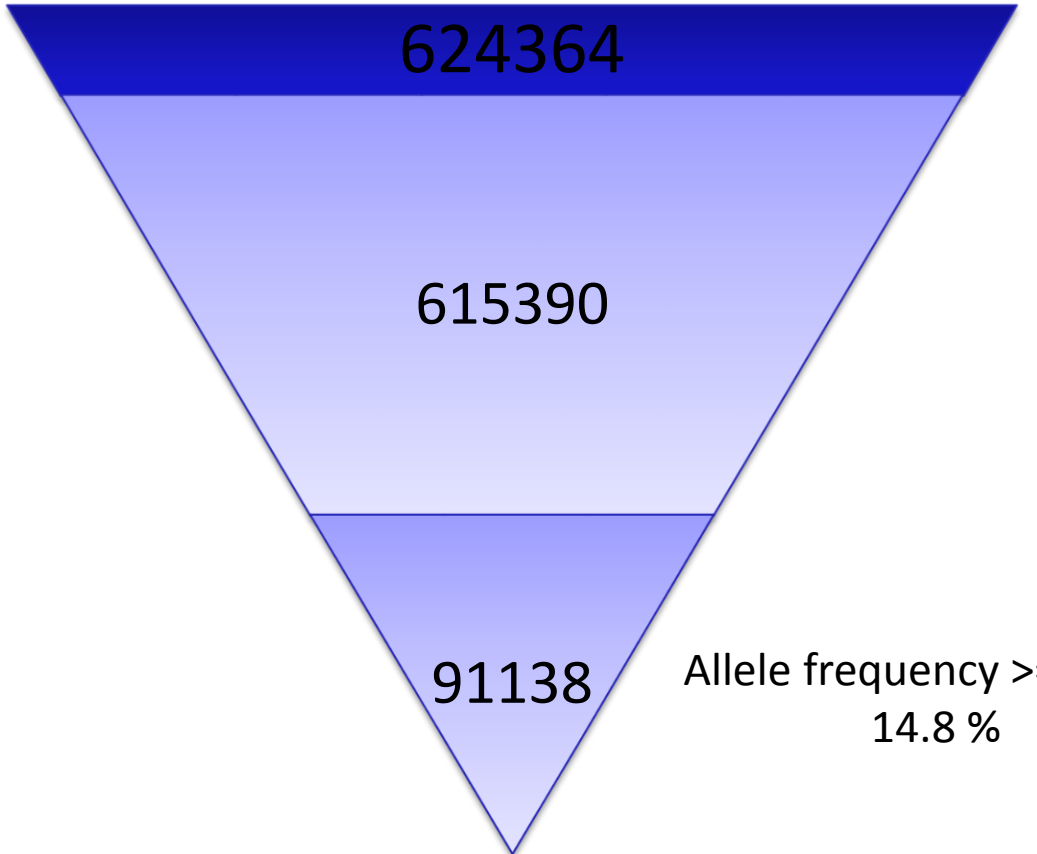
Filter variants with coverage in more than 50% samples



Genomic Databases

BIPMed-WES-db

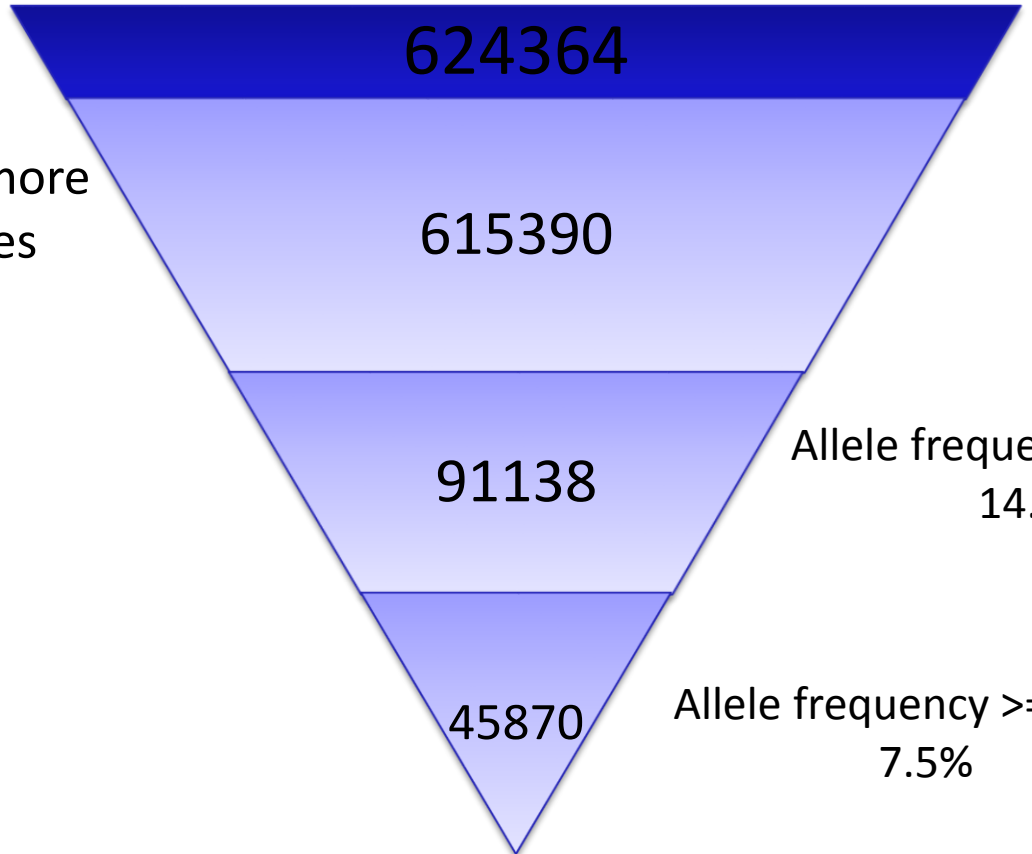
Filter variants according to Allele frequency



Genomic Databases

BIPMed-WES-db

Filter variants according to Allele frequency



Covered variants in more than 50% of samples
98.6%

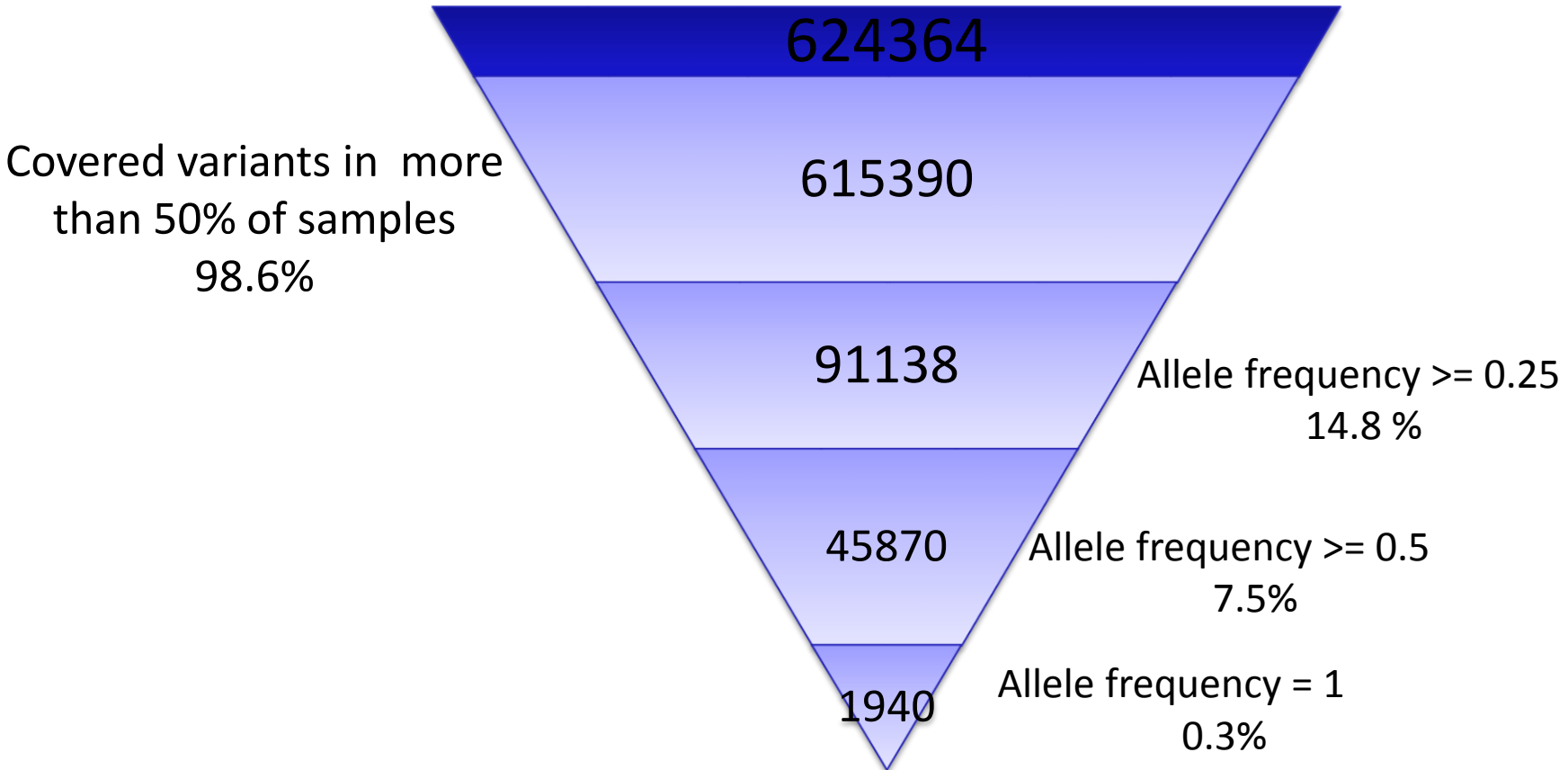
Allele frequency >= 0.25
14.8 %

Allele frequency >= 0.5
7.5%

Genomic Databases

BIPMed-WES-db

Filter variants according to Allele frequency

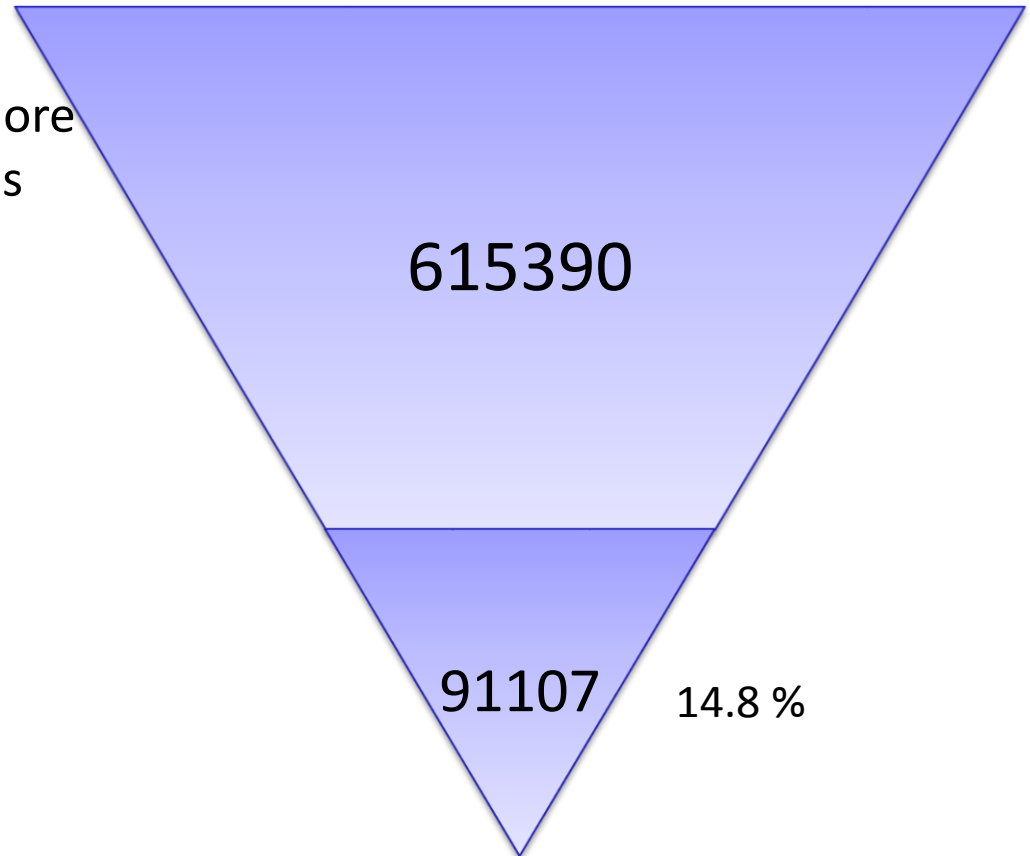


Genomic Databases

BIPMed-WES-db

Filter variants without dbSNP ID

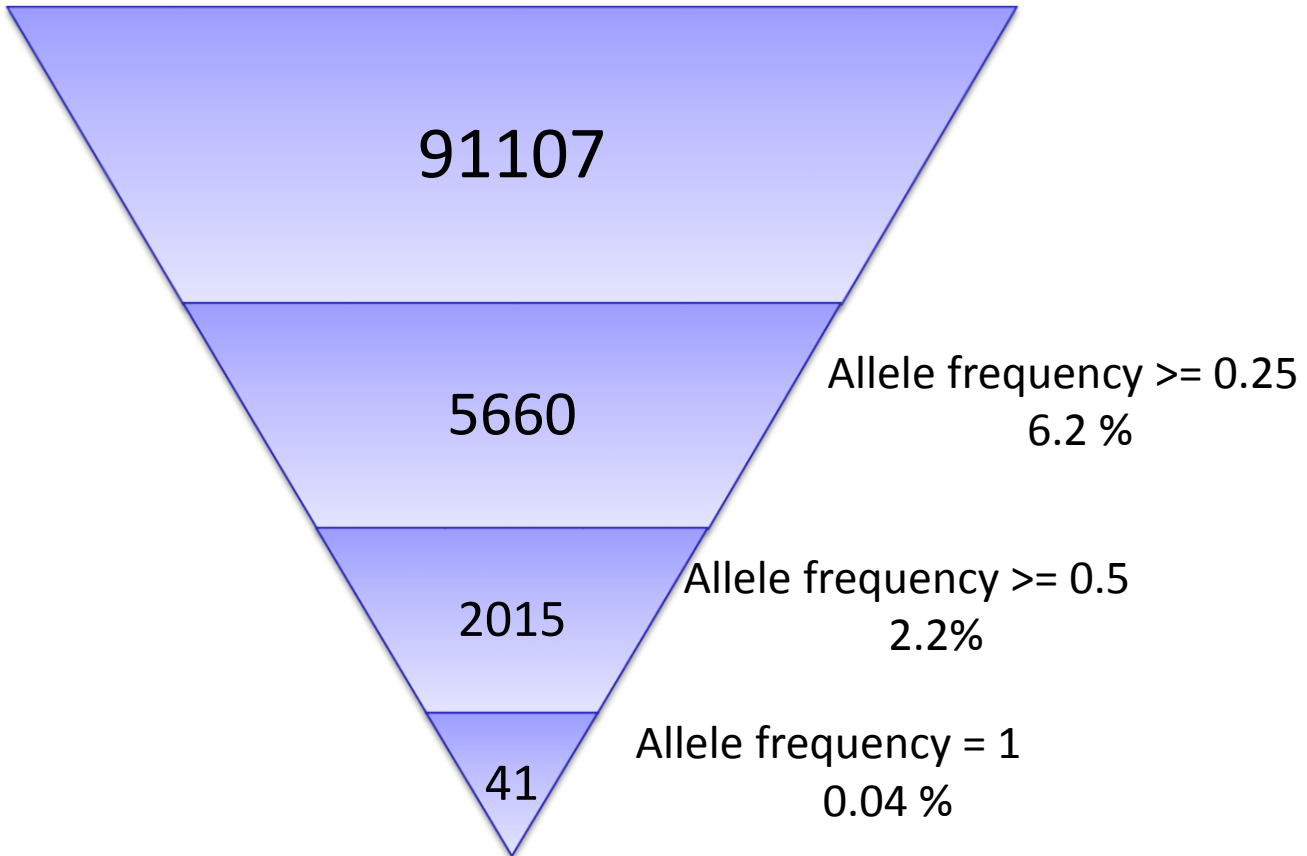
Covered variants in more than 50% of samples



Genomic Databases

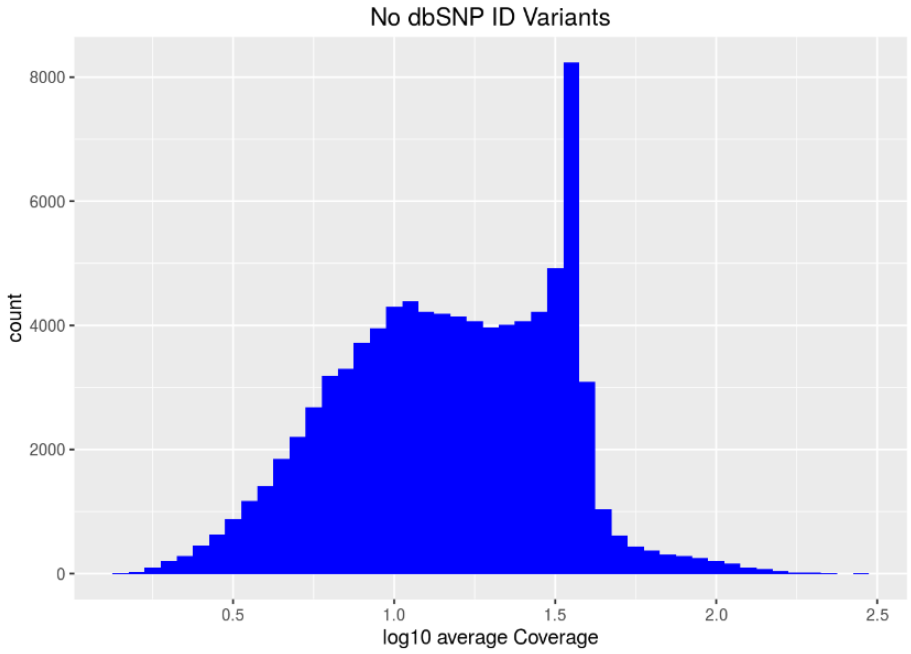
BIPMed-WES-db

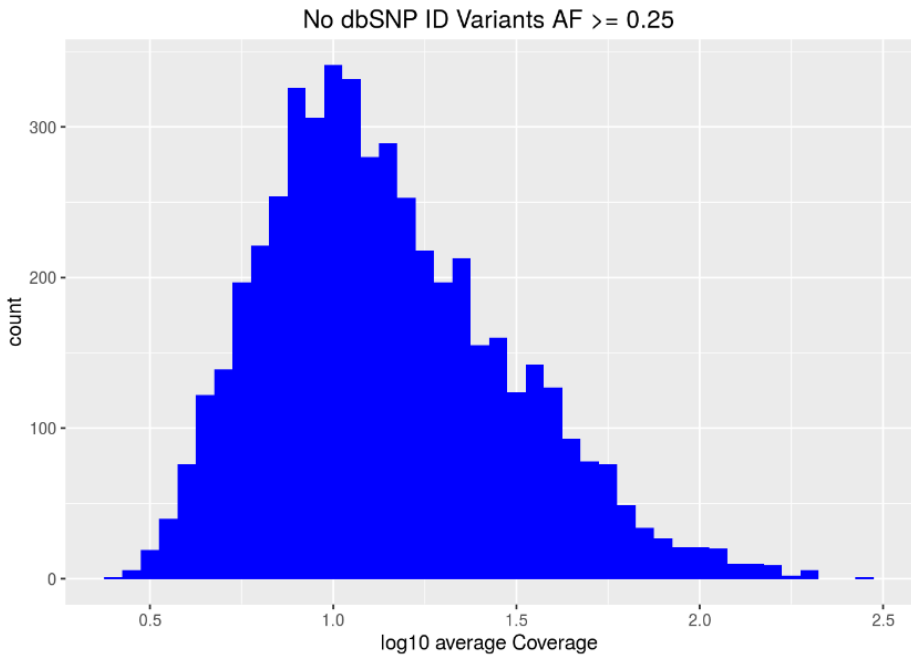
Filter “no dbSNP” variants according to Allele frequency



Genomic Databases

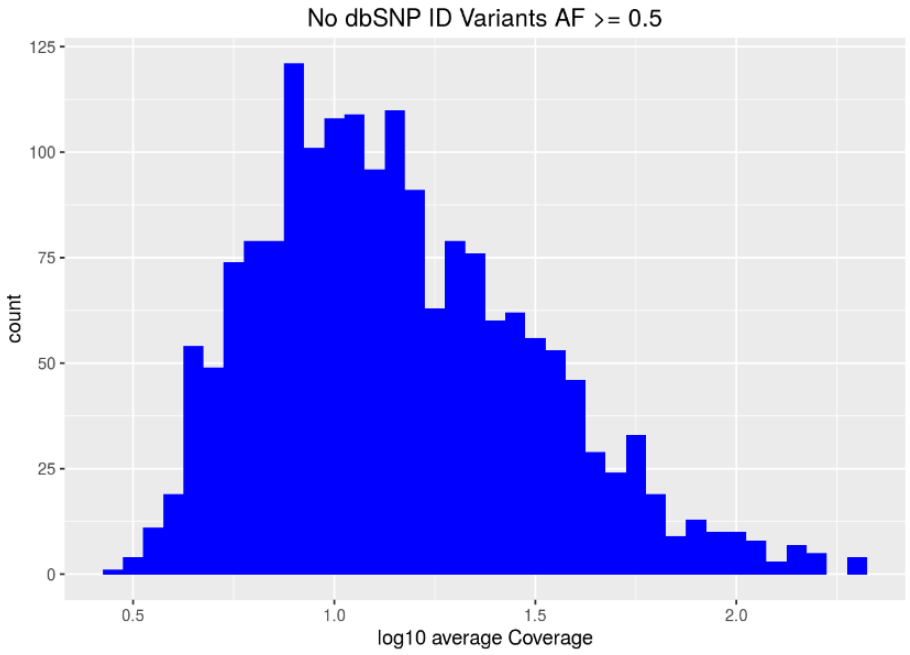
BIPMed-WES-db

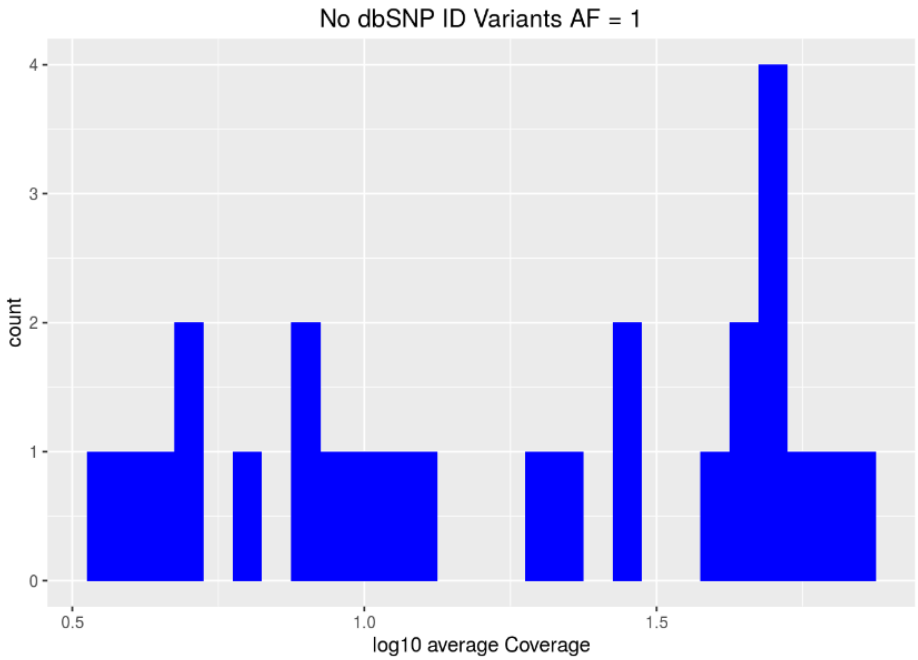




Genomic Databases

BIPMed-WES-db





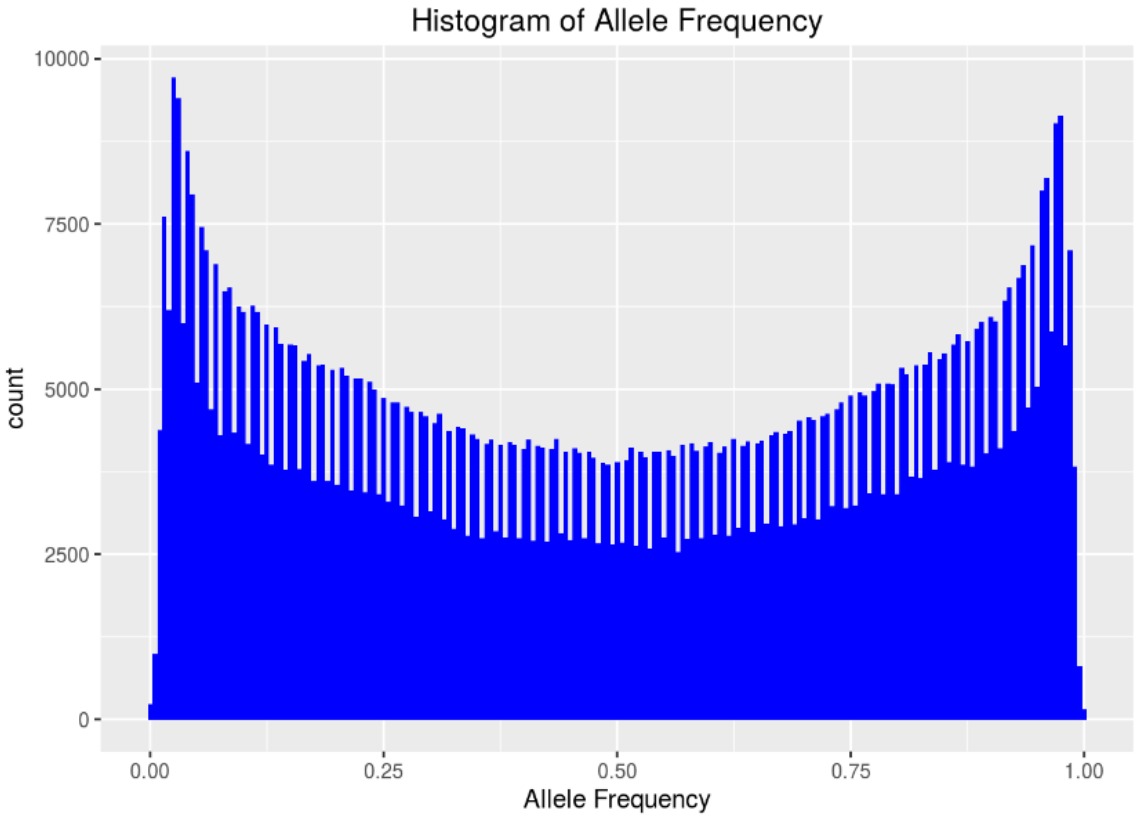
Genomic Databases

BIPMed-Array-db

- Affymetrix Genome-Wide Human SNP Array 6.0 experiments from 246 Brazilian Reference individuals
- Use Leiden Open Variation Database (LOVD) interface
- Open database at http://bipmed.iqm.unicamp.br/snparray
- Over 900.000 different variants

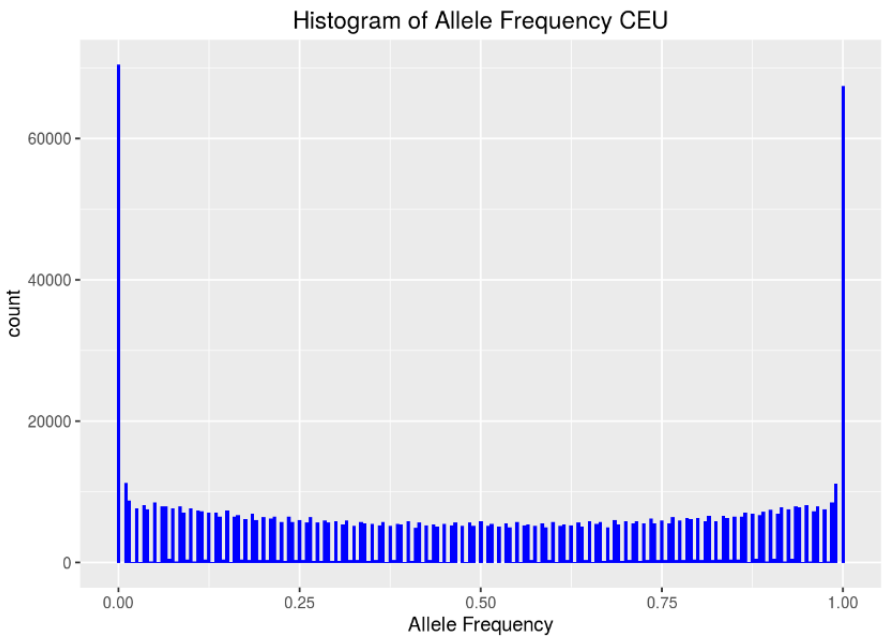
Genomic Databases

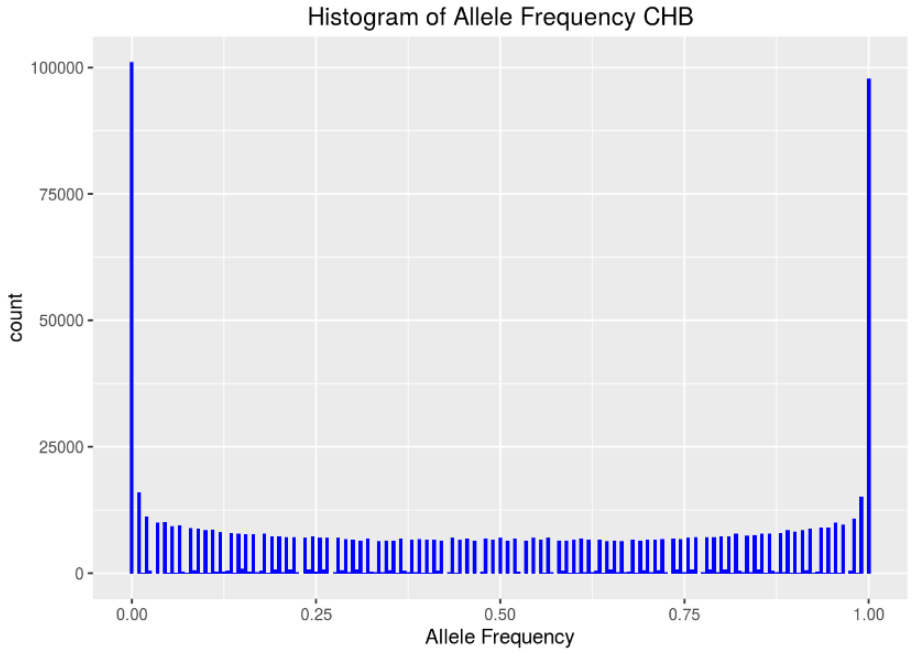
BIPMed-Array-db



Genomic Databases

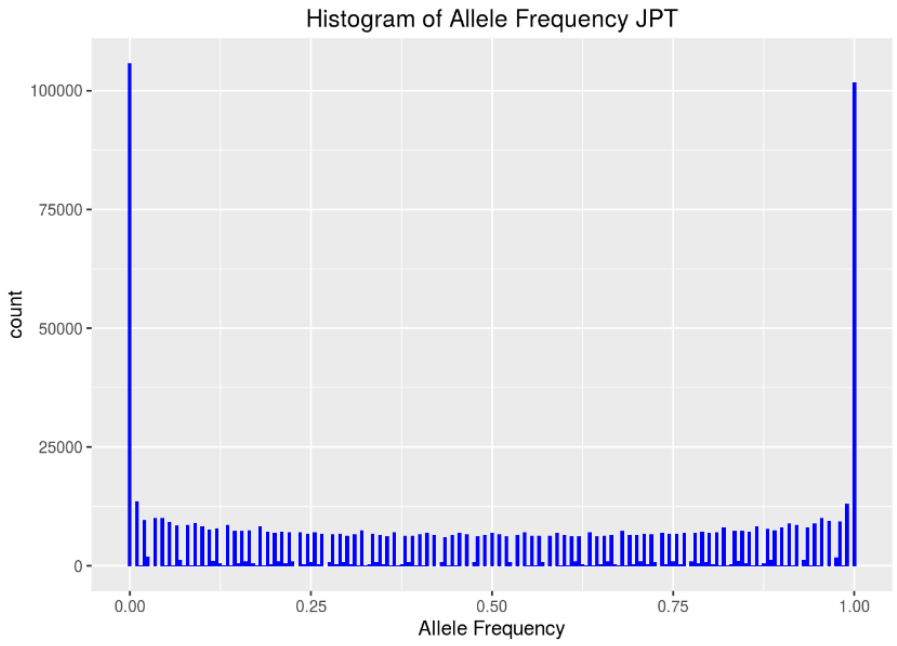
BIPMed-Array-db

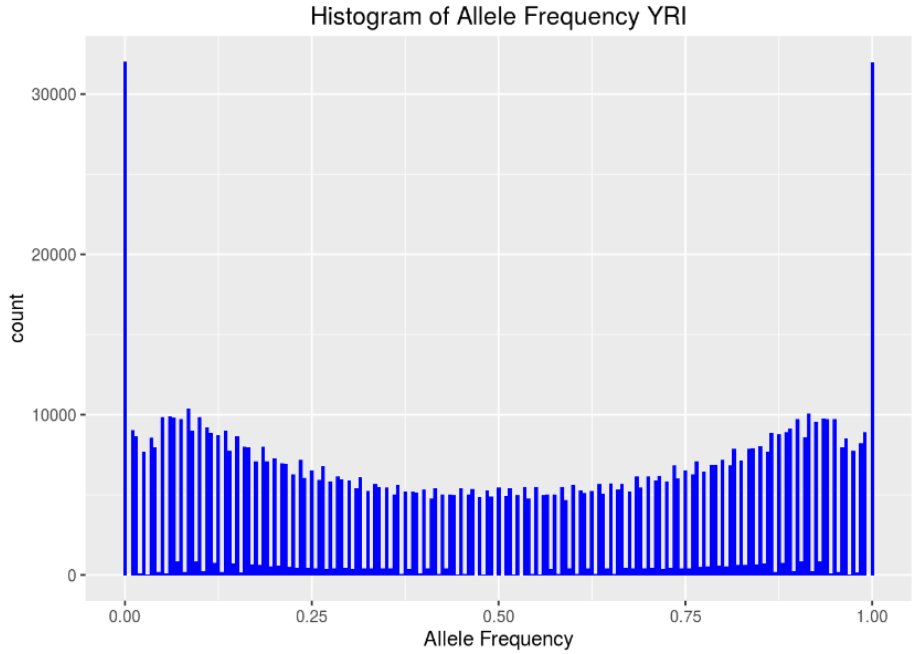




Genomic Databases

BIPMed-Array-db

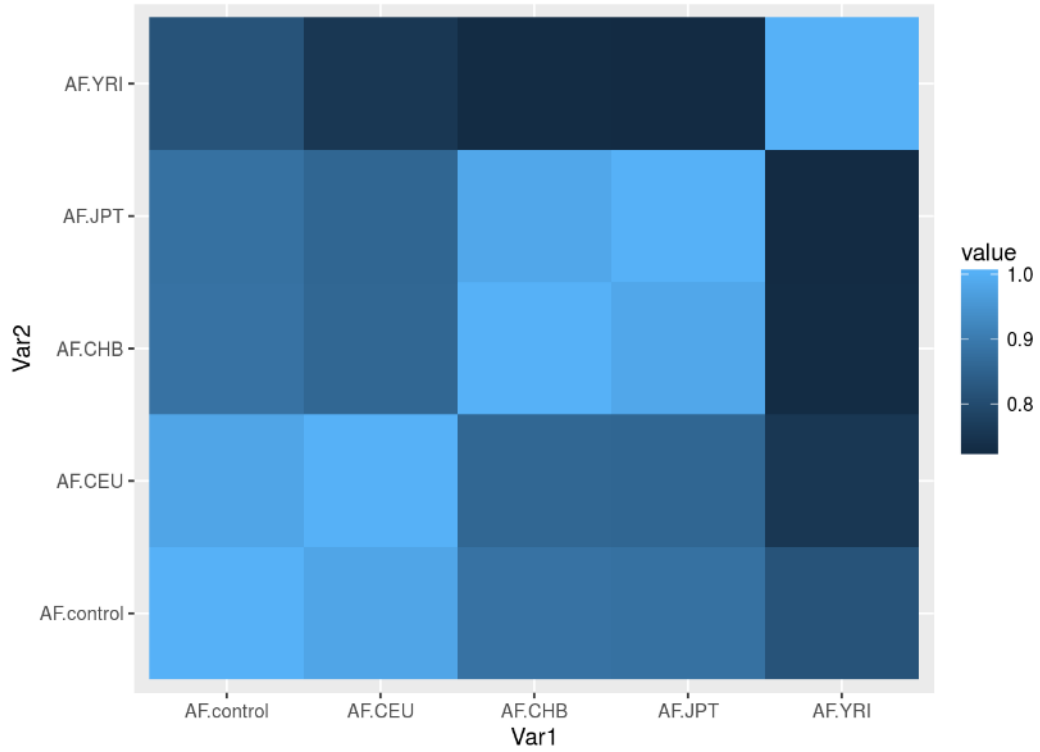




Genomic Databases

BIPMed-Array-db

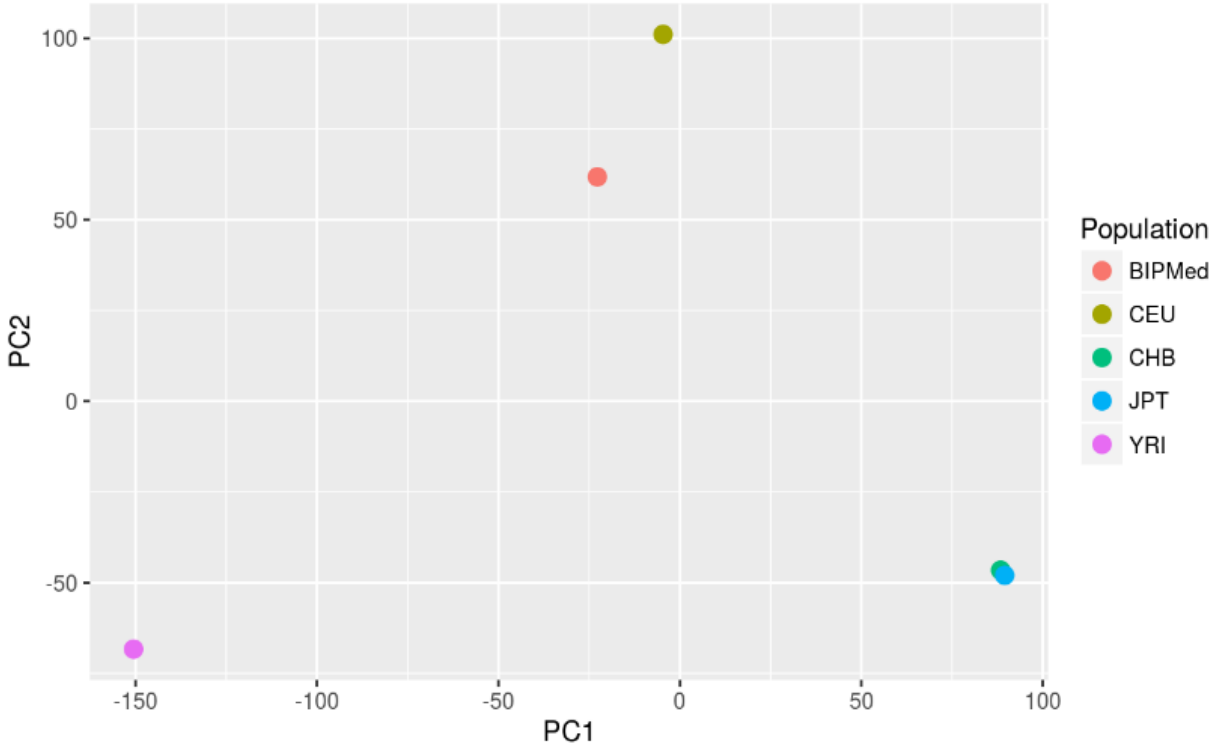
Correlation



Genomic Databases

BIPMed-Array-db

PCA



GA4GH

Global Alliance for Genomics and Health

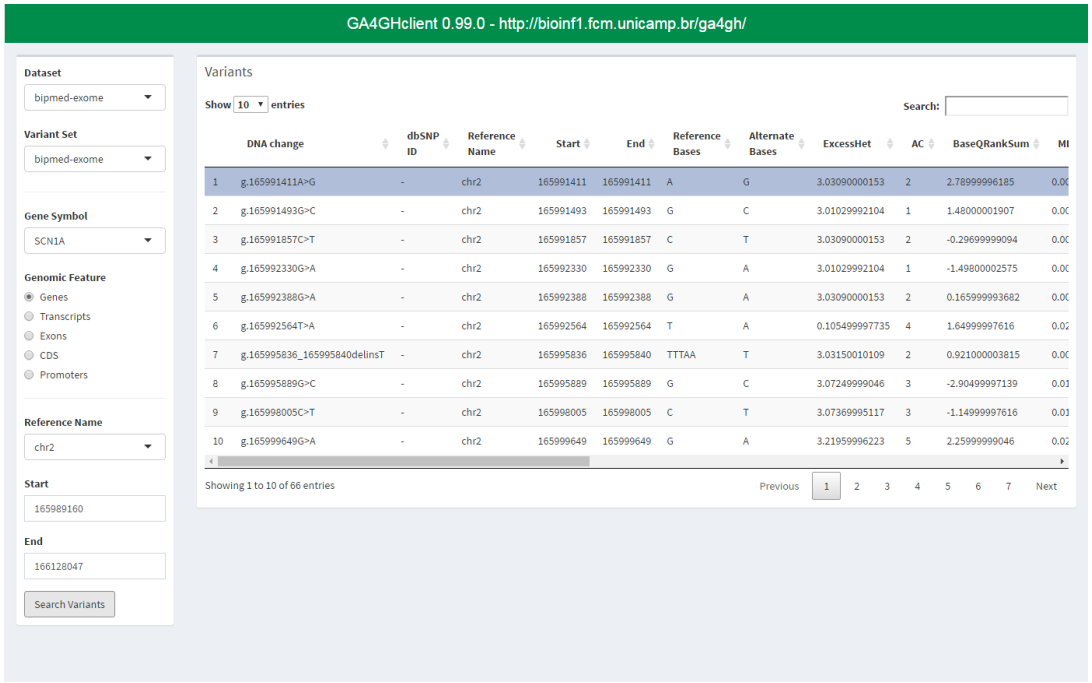
- Aim to help accelerate the potential of genomic medicine to advance human health.
- The partners in the Global Alliance are working together to create a common framework of harmonized approaches to enable the responsible, voluntary, and secure sharing of genomic and clinical data.
- The ga4gh-server software implements data schema for representing genomic variation data.
- It provides an application program interface (API) through the internet.

Global Alliance for Genomics & Health

GA4GH

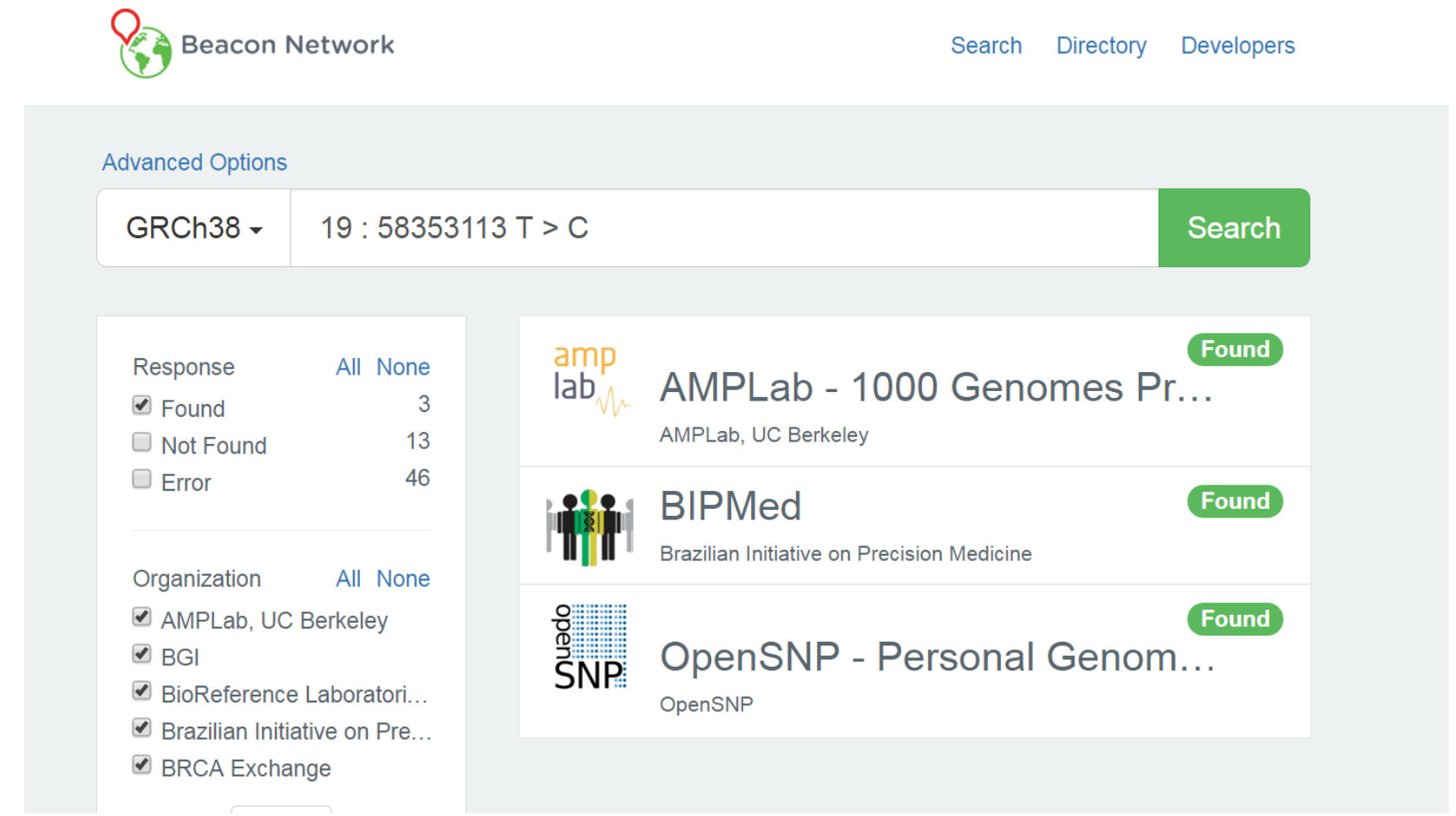
GA4GHclient

- Bioconductor package for accessing GA4GH API data servers.



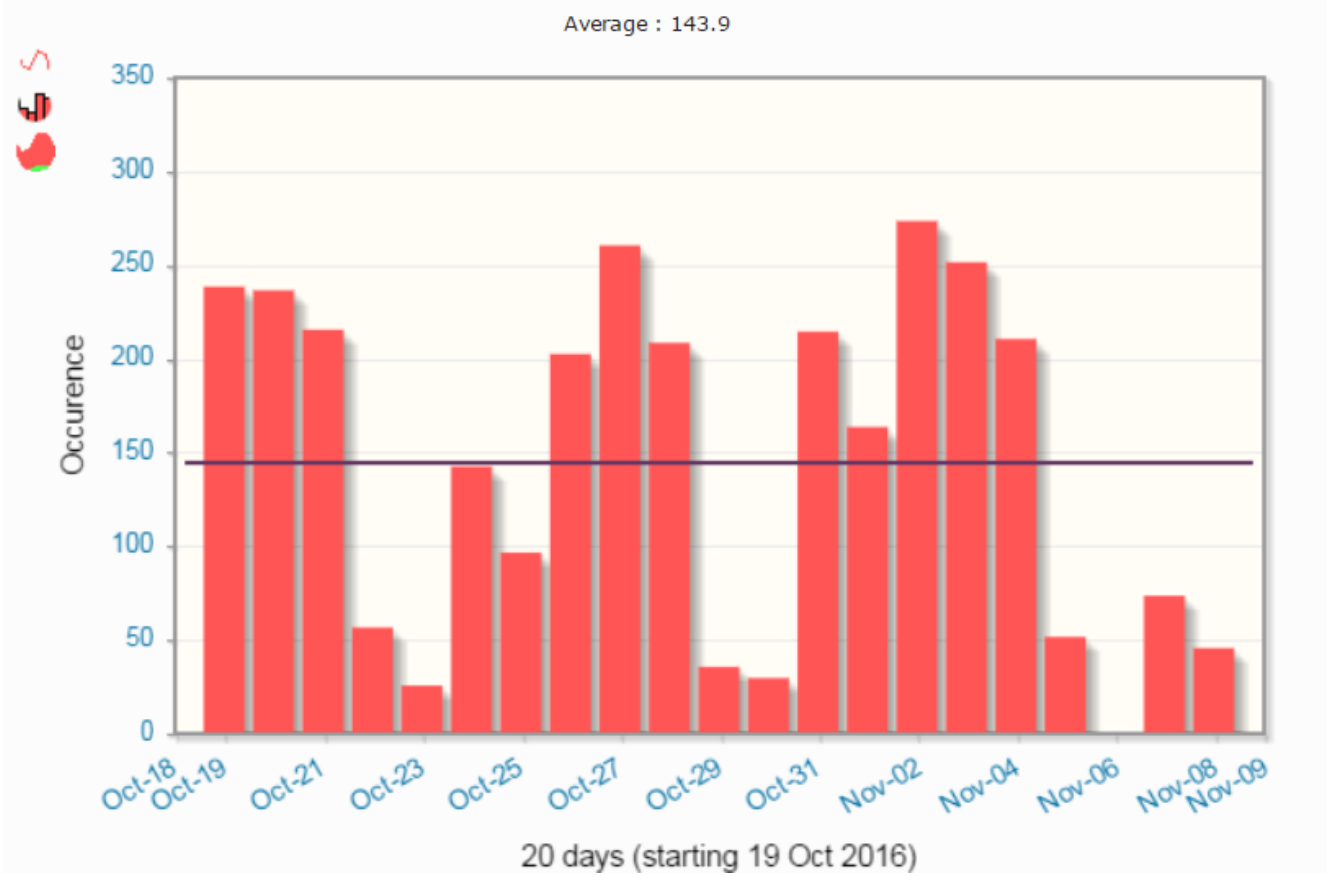
BEACON

https://beacon-network.org



BIPMed Brazilian Initiative on PRECISION MEDICINE

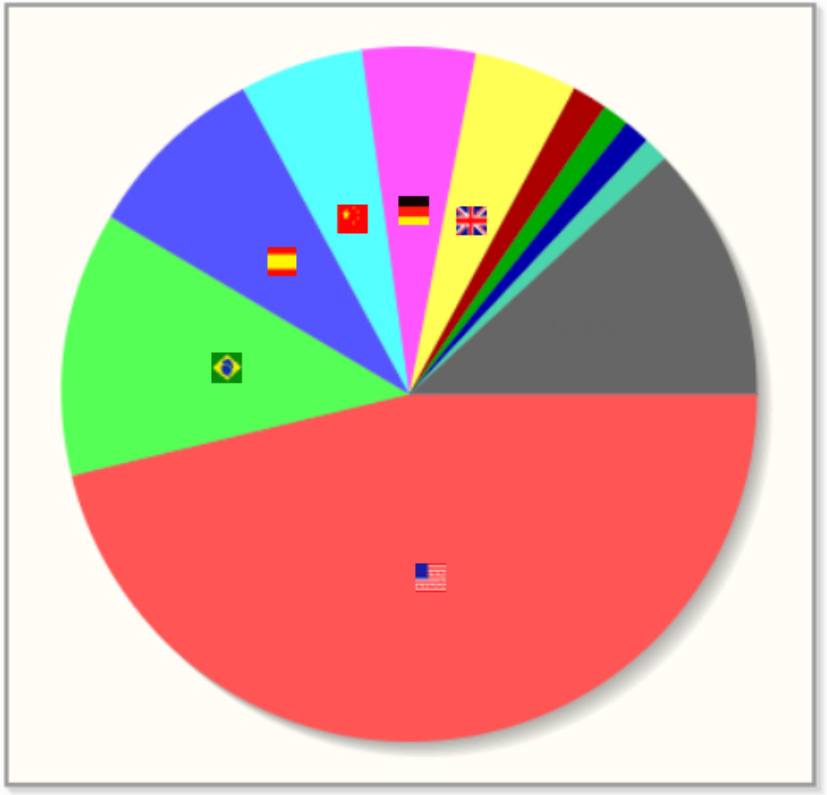
Access Statistics



BIPMed Brazilian Initiative on PRECISION MEDICINE

Access Statistics

Countries		
United States -	9414	46.2 %
Brazil -	2501	12.2 %
Spain -	1741	8.5 %
China -	1168	5.7 %
Germany -	1072	5.2 %
Great Britain -	979	4.8 %
Italy -	332	1.6 %
Australia -	252	1.2 %
Russian Federation -	248	1.2 %
Canada -	238	1.1 %
Others	2415	11.8 %



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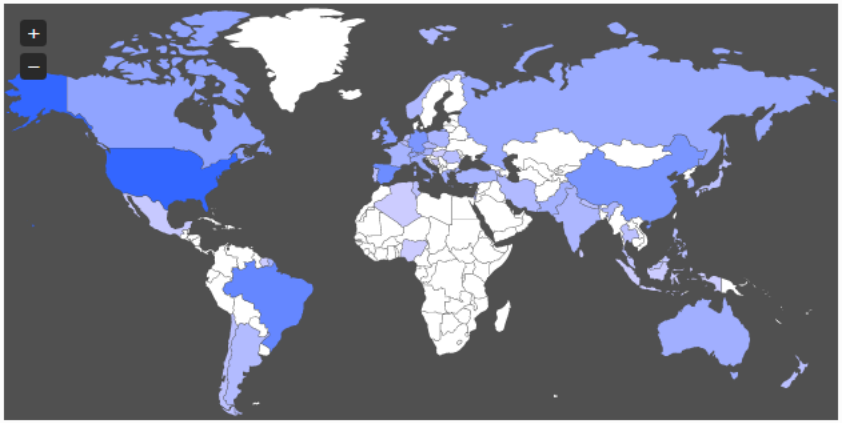

Requests Accesses Hosts Traffic

	Hosts	Percentage	Requests	Percentage	Accesses	Percentage	Traffic (Mb)	Percentage
Europe	125	23.8 %	5774	28.4 %	1961	22.2 %	281.2	28.1 %
South-America	104	19.8 %	2618	12.9 %	1074	12.1 %	85.7	8.6 %
North-America	178	33.9 %	9654	47.4 %	5079	57.4 %	541.0	54.1 %
Africa	3	0.6 %	38	0.2 %	9	0.1 %	1.6	0.2 %
Oceania	9	1.7 %	291	1.4 %	67	0.8 %	12.1	1.2 %
Asia	106	20.2 %	1985	9.7 %	657	7.4 %	78.2	7.8 %

Access Statistics

- Site summary

Global access	Total	external	local
Requests	258210	258182	28
Accesses	181381	181353	28
Documents	360	360	0
Traffic (Gb)	18.83	18.83	0
Number of different sites	2542	2541	1
Number of different pages used	37130		
Number of different resolved countries	73		

Acknowledgment