

PIBMed - Brazilian Initiative on Precision Medicine

Summary of results for the first year



Dra. Cristiane Rocha



BIPMed

Our Products:

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



Genomic Databases

BIPMed-WES-db

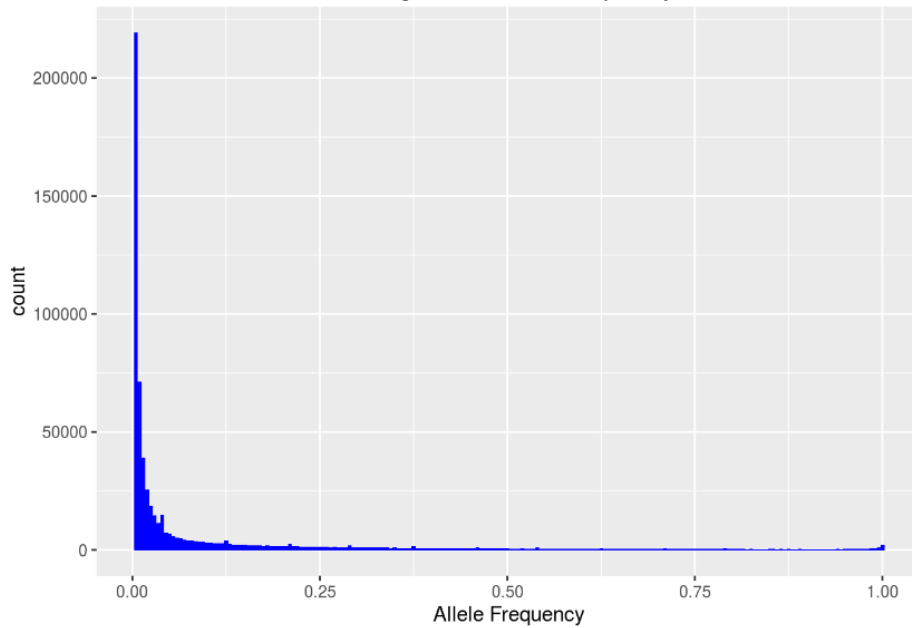
- Whole Exome Sequencing experiments from 106 Brazilian Reference individuals
- Use Leiden Open Variation Database (LOVD) interface
- Open database at <http://bipmed.iqm.unicamp.br>
- Over 600.000 different variants



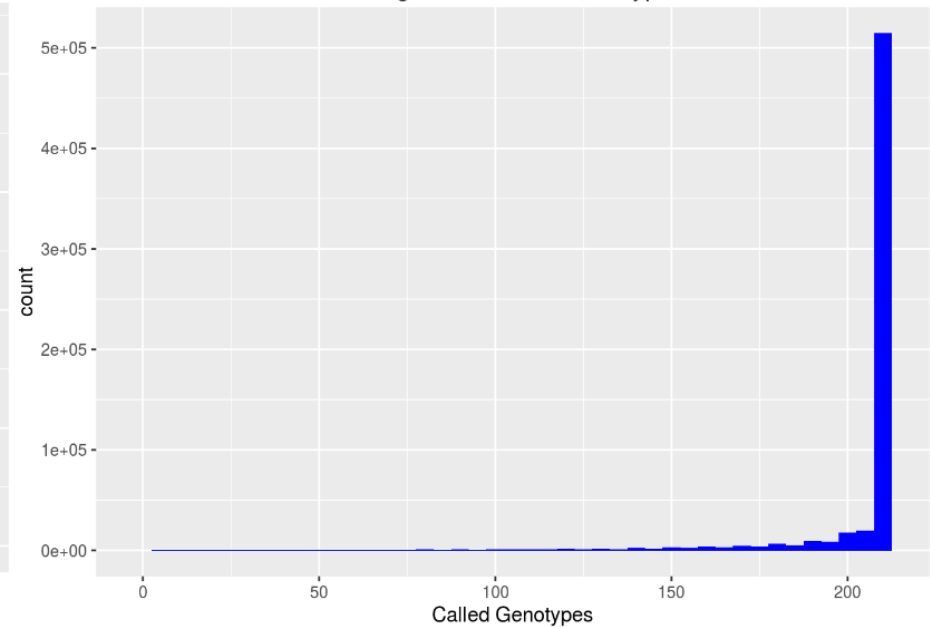
Genomic Databases

BIPMed-WES-db

Histogram of Allele Frequency



Histogram of Called Genotypes

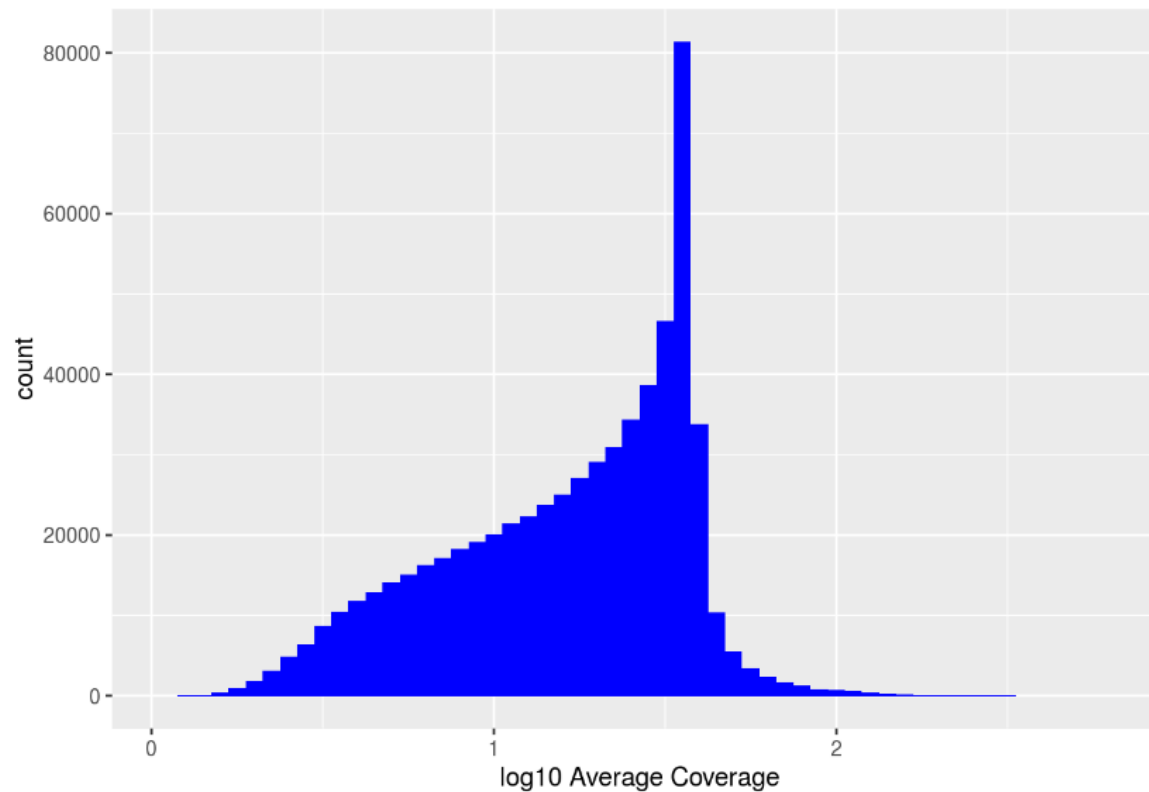




Genomic Databases

BIPMed-WES-db

Average Coverage





Genomic Databases

BIPMed-WES-db

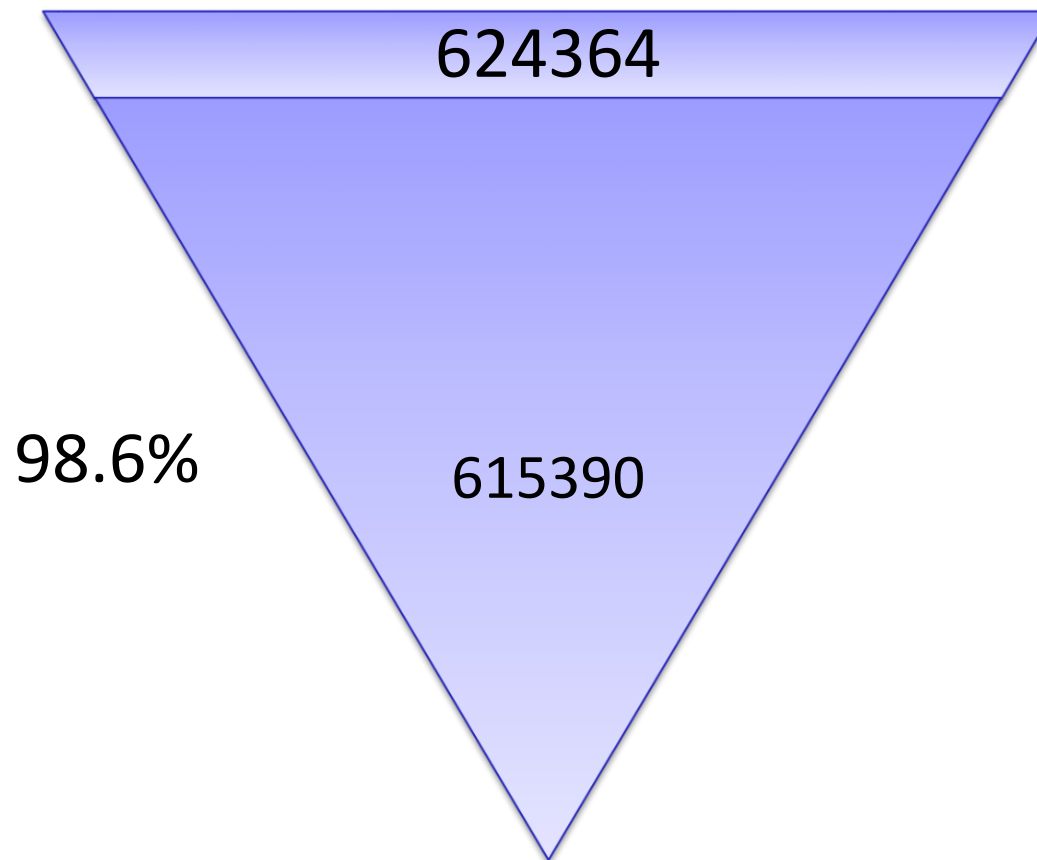
Total variants

624364

Genomic Databases

BIPMed-WES-db

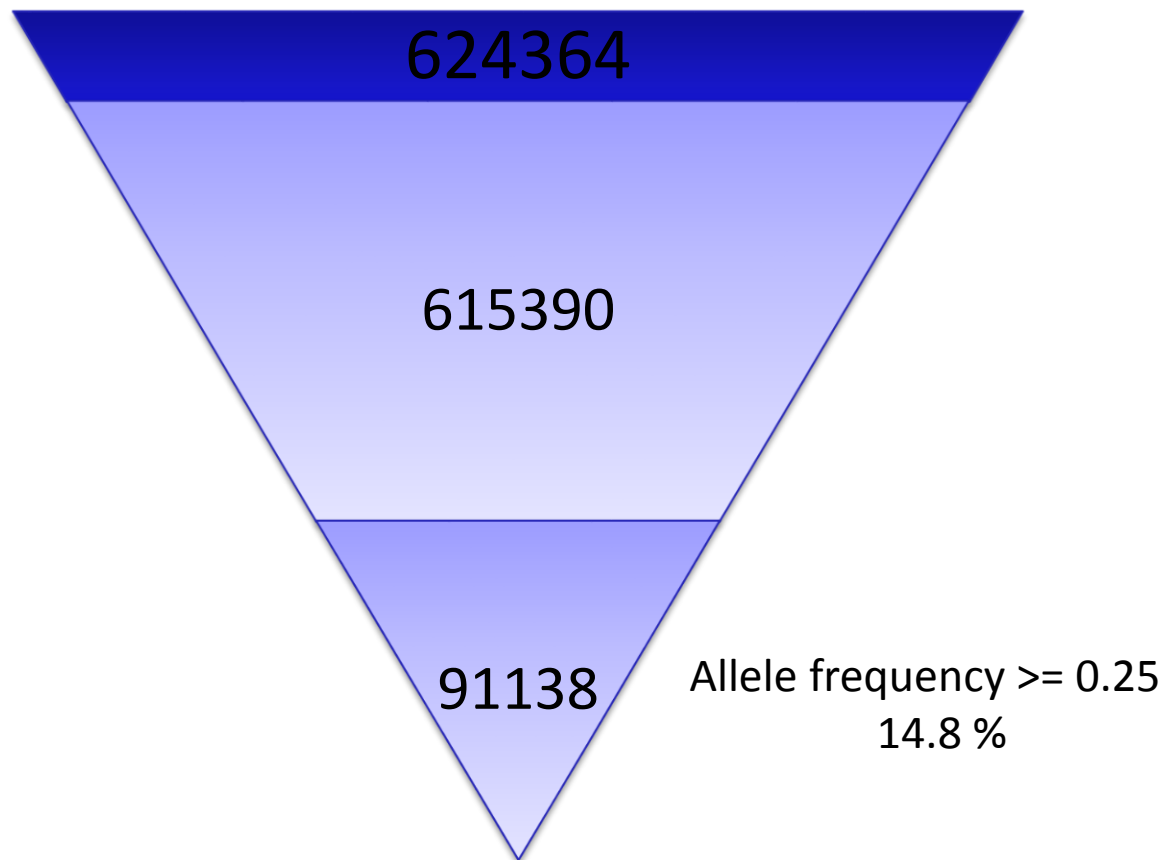
Filter variants with coverage in more than 50% samples



Genomic Databases

BIPMed-WES-db

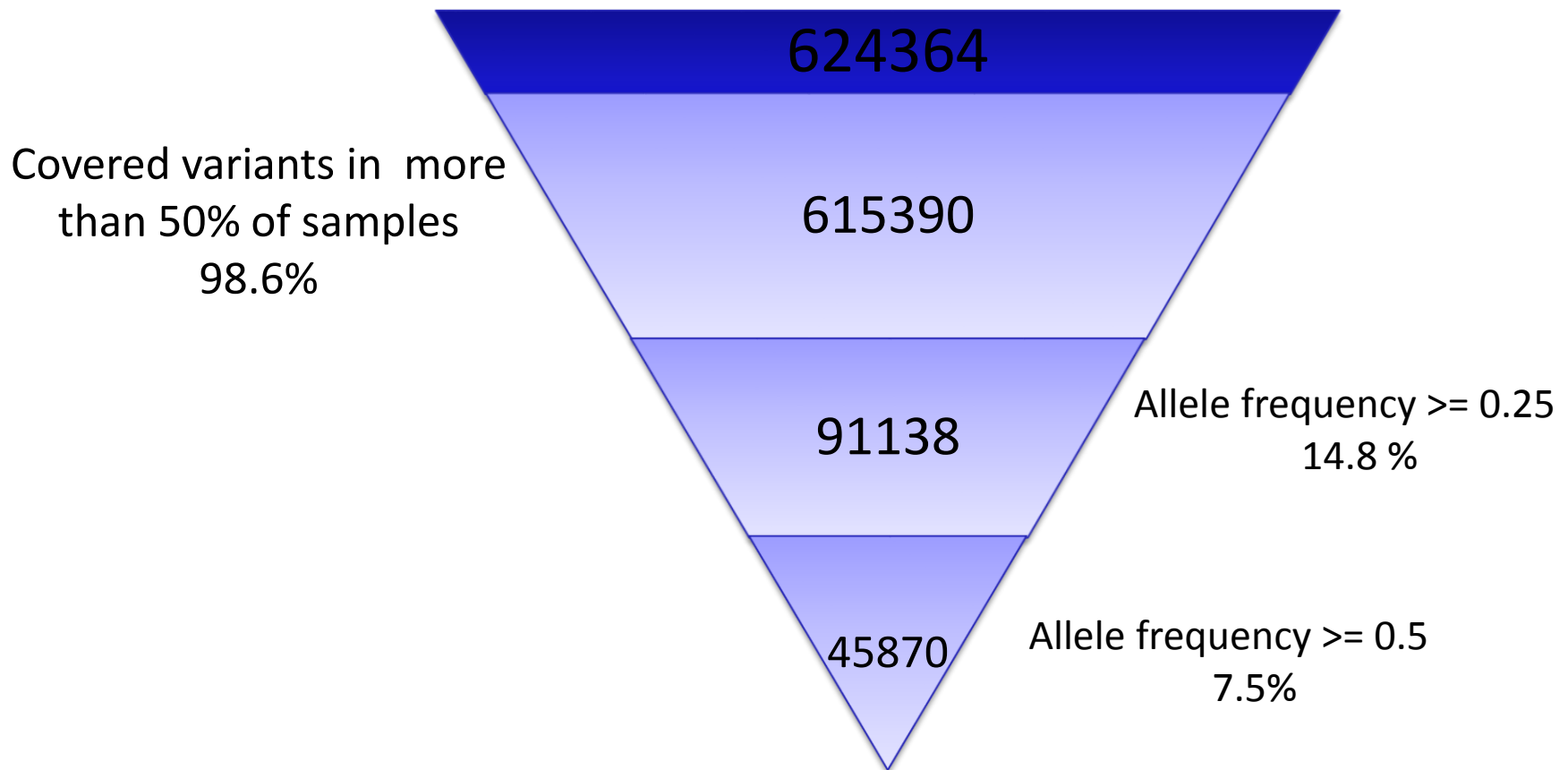
Filter variants according to Allele frequency



Genomic Databases

BIPMed-WES-db

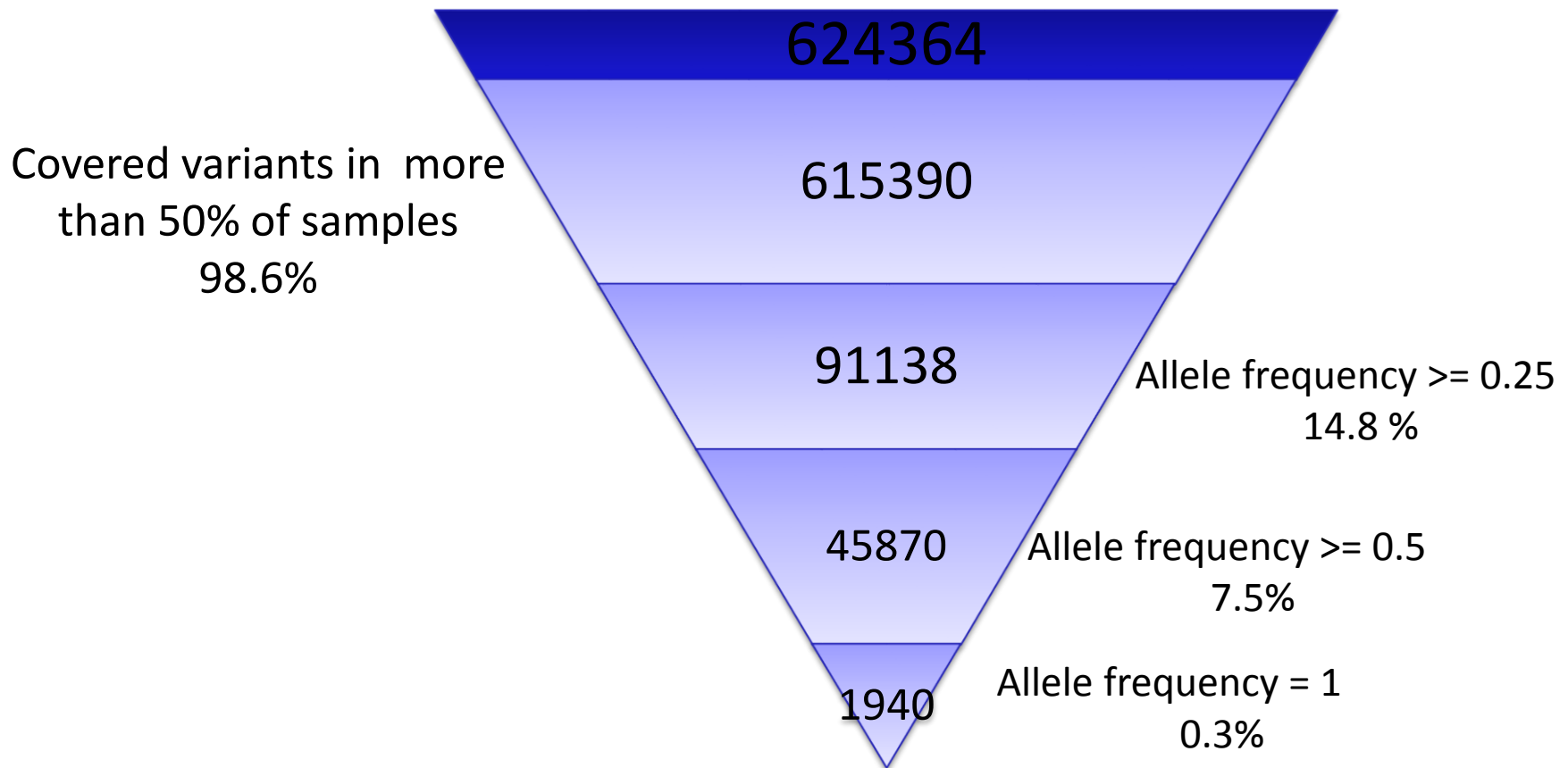
Filter variants according to Allele frequency



Genomic Databases

BIPMed-WES-db

Filter variants according to Allele frequency

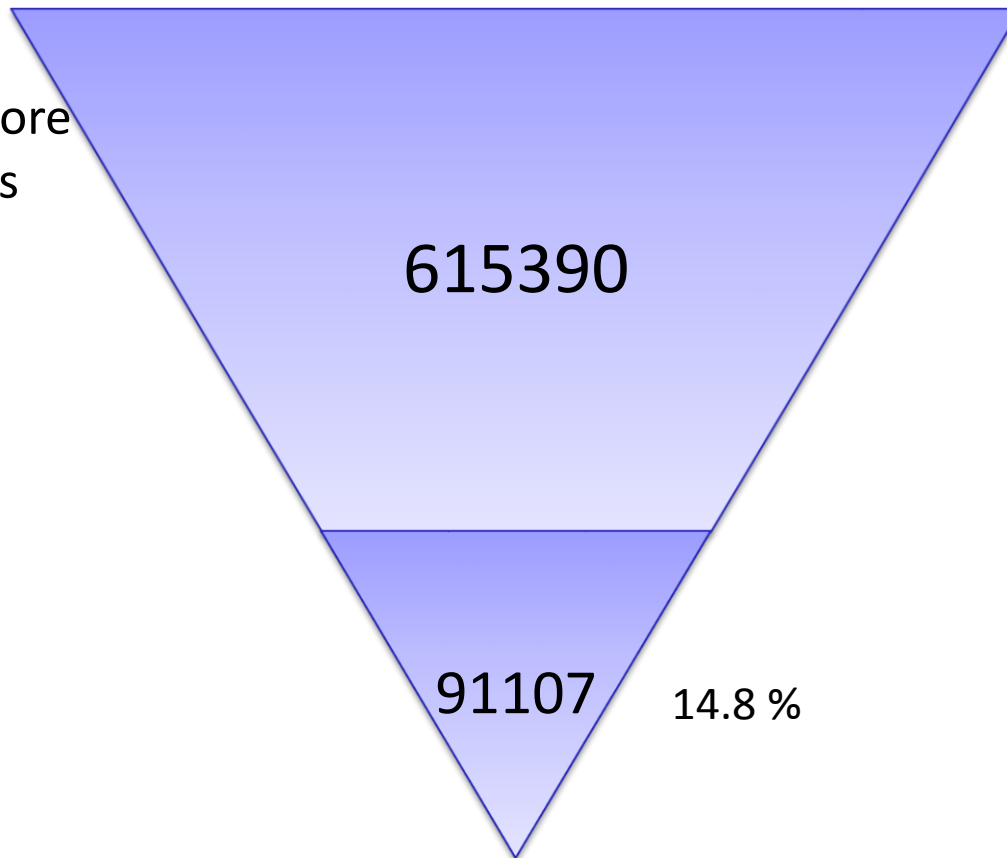




Genomic Databases

BIPMed-WES-db

Filter variants without dbSNP ID

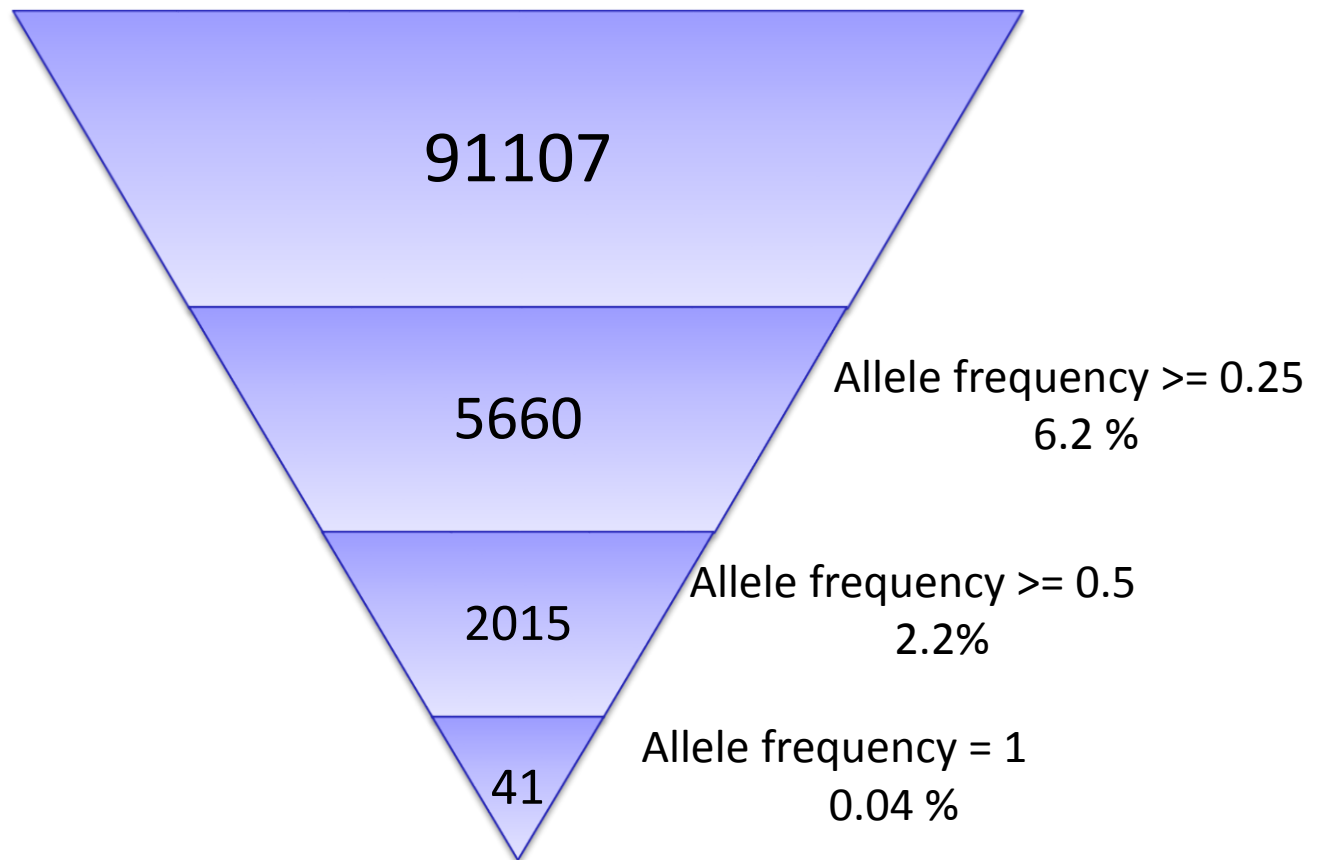


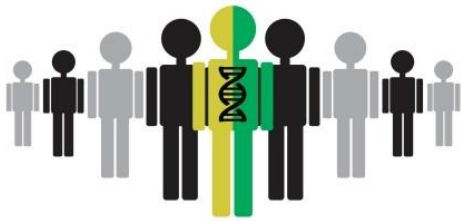
Covered variants in more than 50% of samples

Genomic Databases

BIPMed-WES-db

Filter “no dbSNP” variants according to Allele frequency

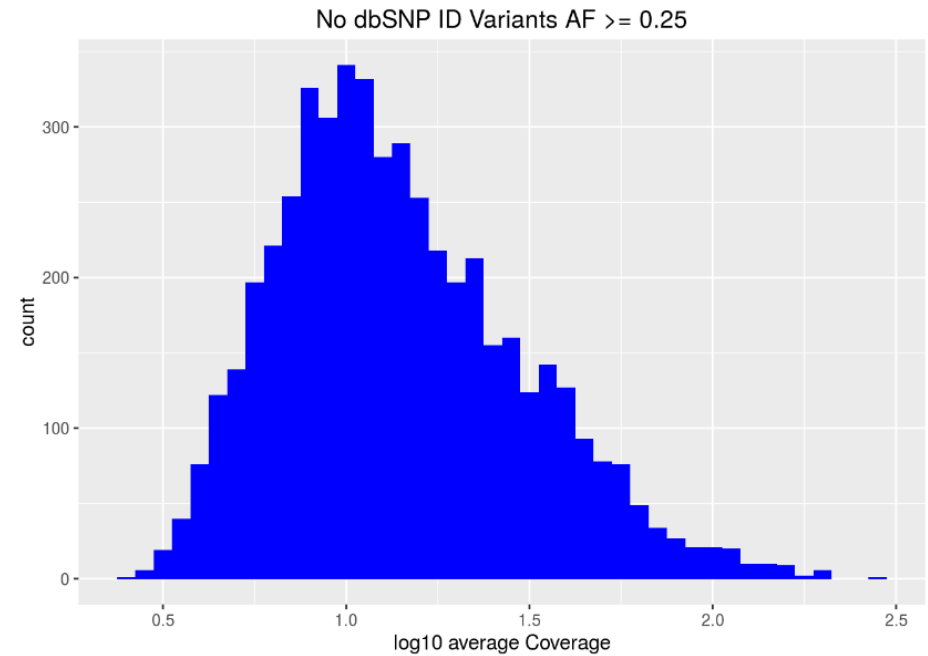
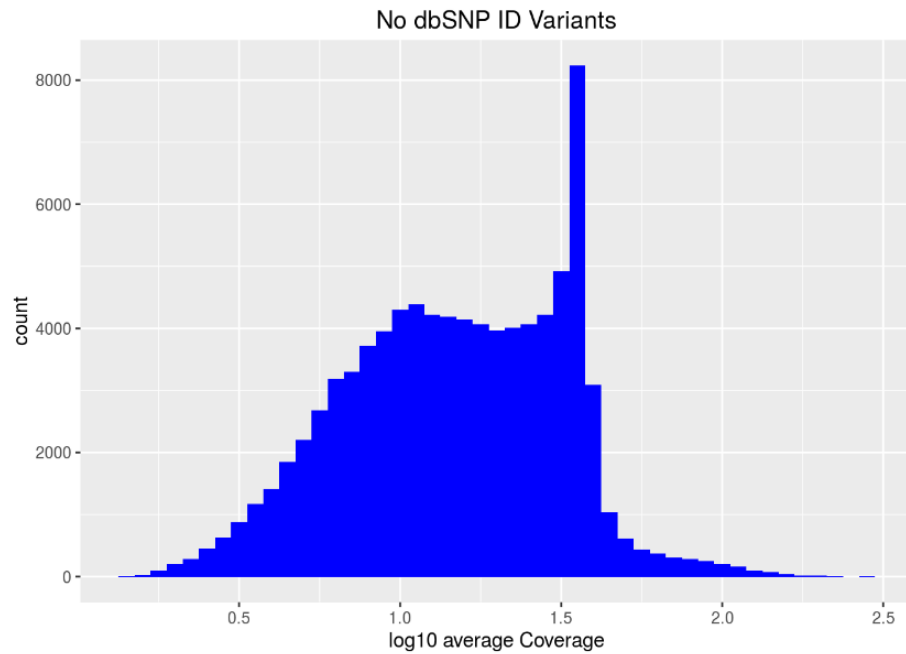


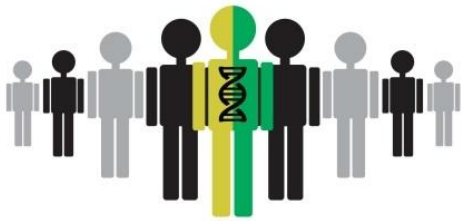


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Genomic Databases

BIPMed-WES-db

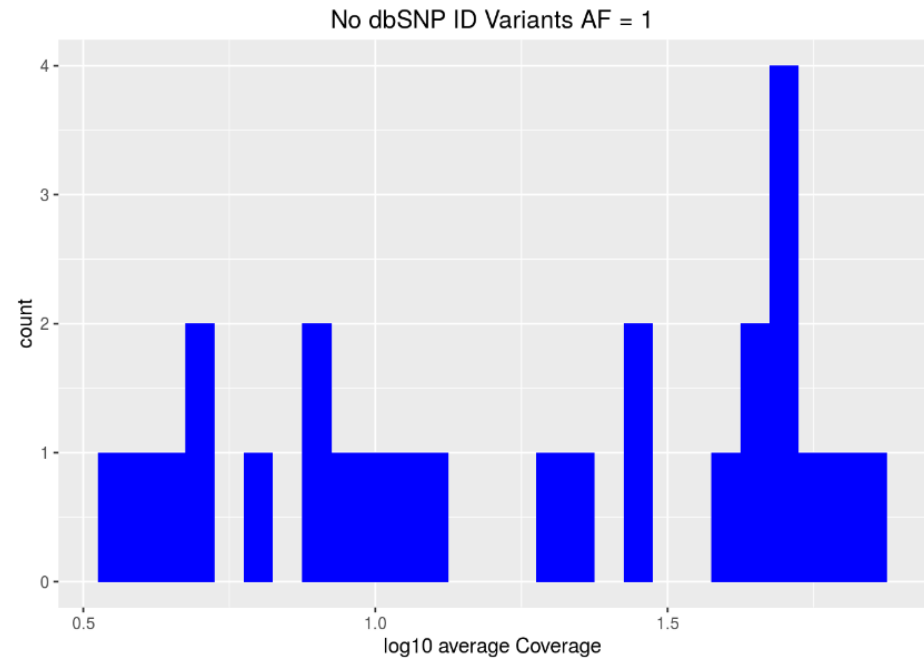
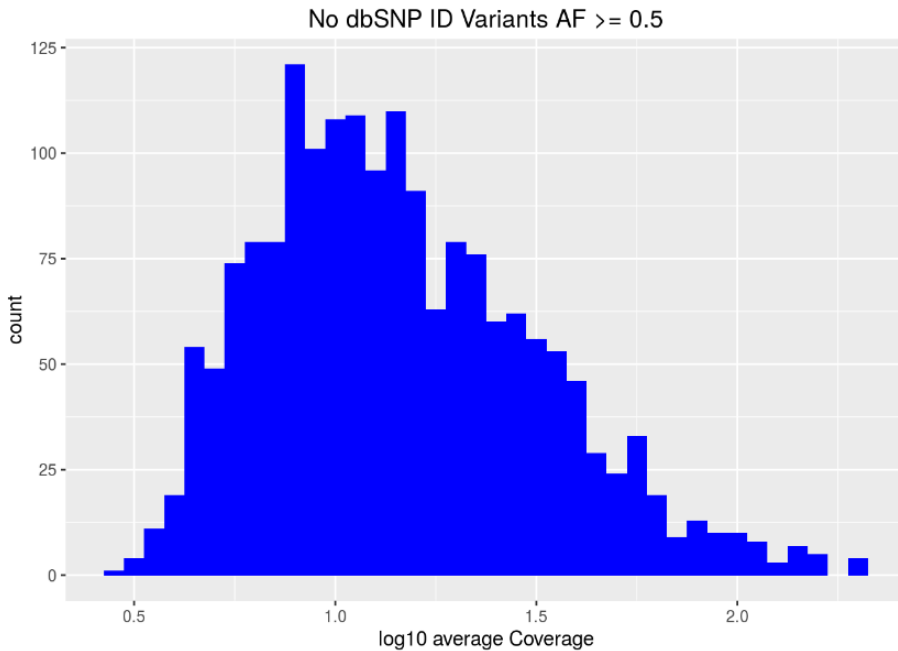




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Genomic Databases

BIPMed-WES-db





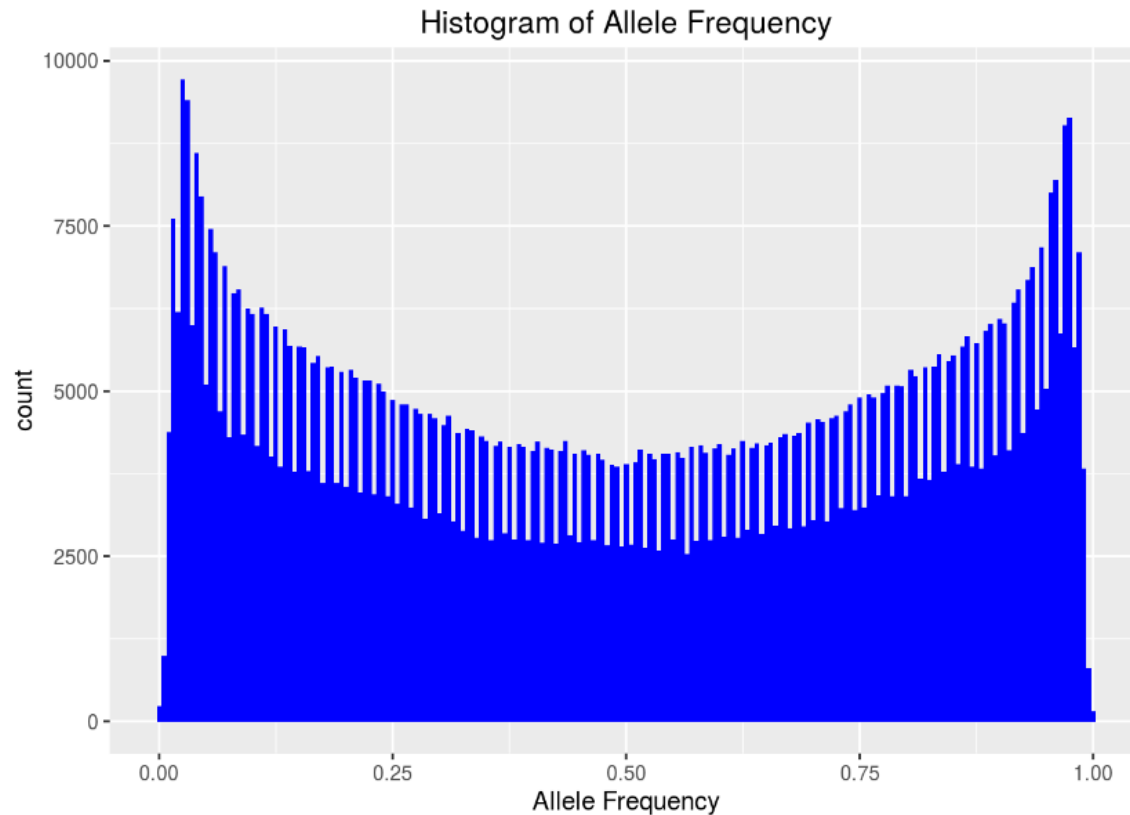
Genomic Databases

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

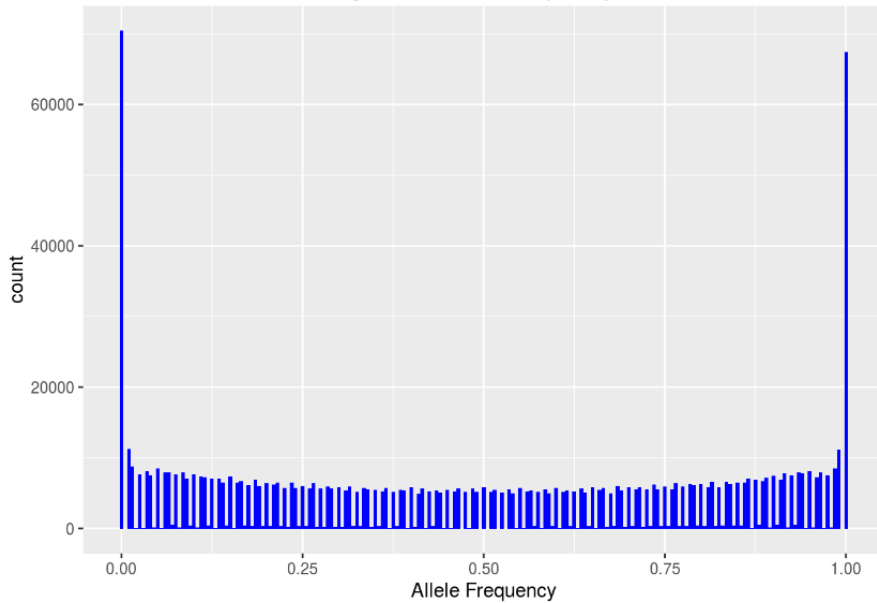




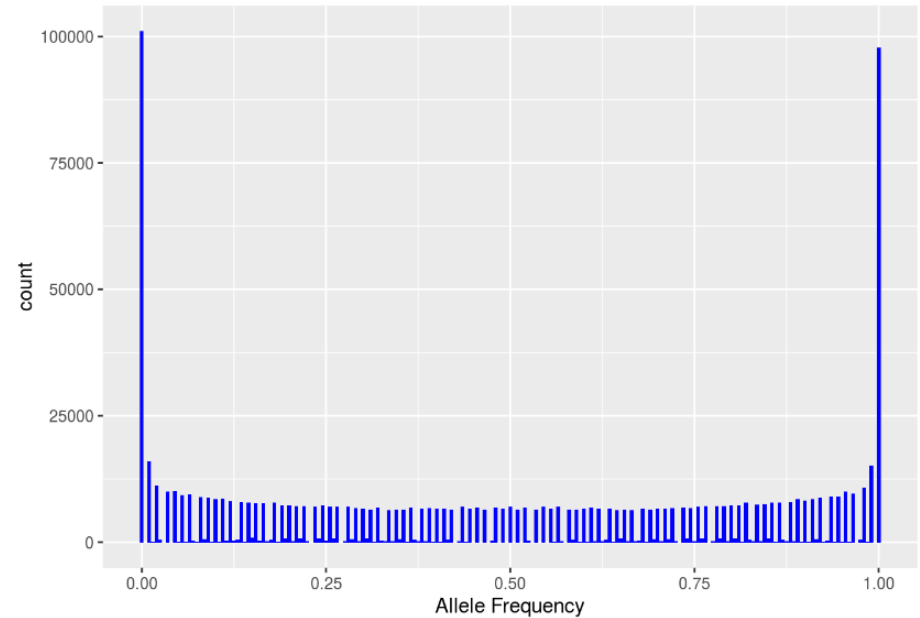
Genomic Databases

BIPMed-Array-db

Histogram of Allele Frequency CEU



Histogram of Allele Frequency CHB

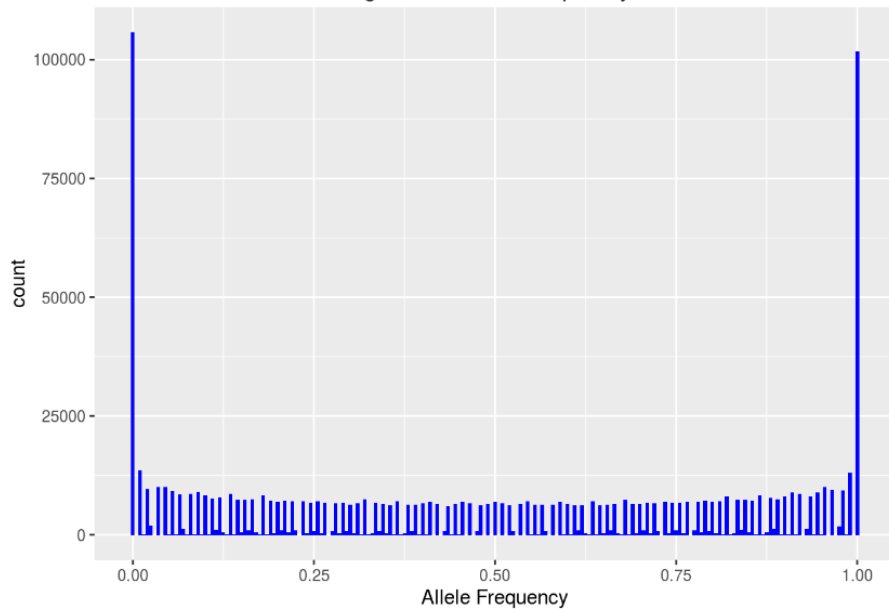




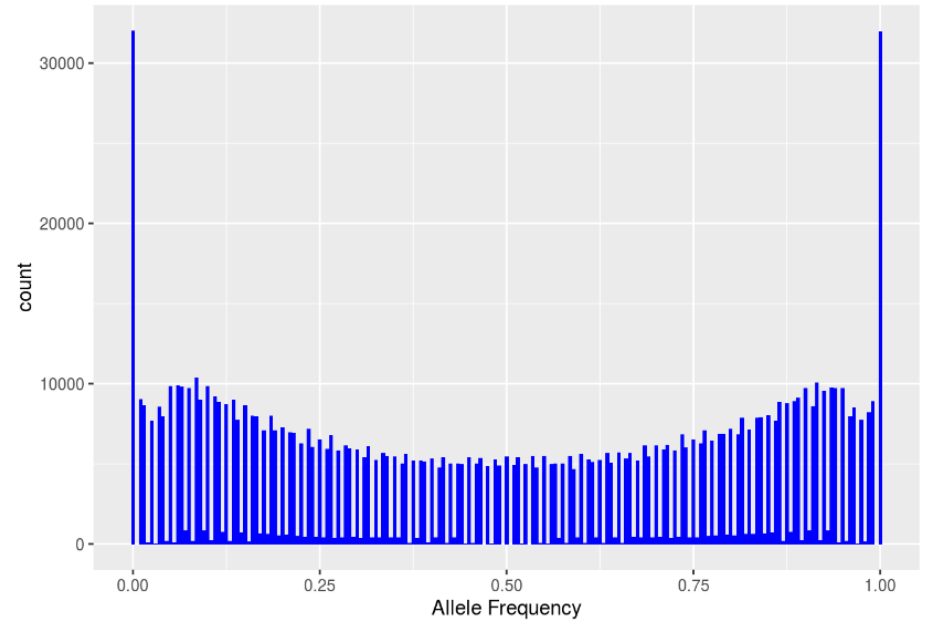
Genomic Databases

BIPMed-Array-db

Histogram of Allele Frequency JPT



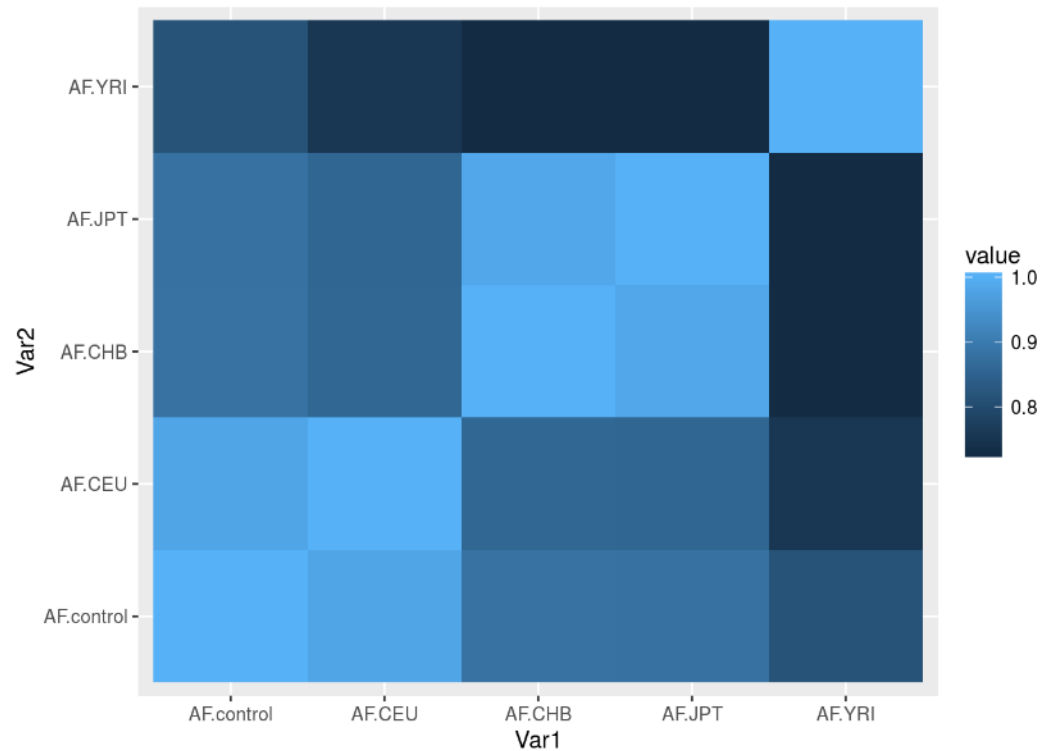
Histogram of Allele Frequency YRI



Genomic Databases

BIPMed-Array-db

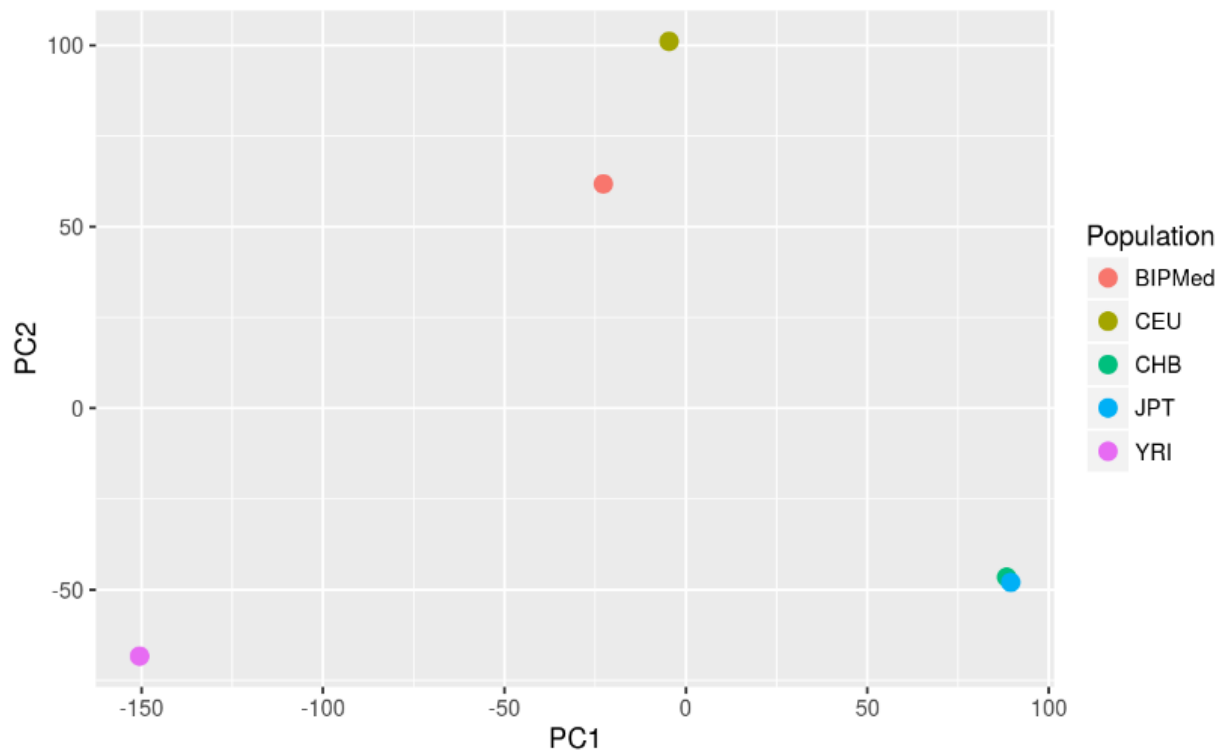
Correlation



Genomic Databases

BIPMed-Array-db

PCA





Global Alliance
for Genomics & Health

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.



GA4GHclient

- Bioconductor package for accessing GA4GH API data servers.

GA4GHclient 0.99.0 - <http://bioinf1.fcm.unicamp.br/ga4gh/>

Dataset:

Variant Set:

Gene Symbol:

Genomic Feature:
 Genes
 Transcripts
 Exons
 CDS
 Promoters

Reference Name:

Start:

End:

Variants

Show entries

Search:

	DNA change	dbSNP ID	Reference Name	Start	End	Reference Bases	Alternate Bases	ExcessHet	AC	BaseQRankSum	MI
1	g.165991411A>G	-	chr2	165991411	165991411	A	G	3.03090000153	2	2.78999996185	0.00
2	g.165991493G>C	-	chr2	165991493	165991493	G	C	3.01029992104	1	1.48000001907	0.00
3	g.165991857C>T	-	chr2	165991857	165991857	C	T	3.03090000153	2	-0.29699999094	0.00
4	g.165992330G>A	-	chr2	165992330	165992330	G	A	3.01029992104	1	-1.49800002575	0.00
5	g.165992388G>A	-	chr2	165992388	165992388	G	A	3.03090000153	2	0.165999993682	0.00
6	g.165992564T>A	-	chr2	165992564	165992564	T	A	0.105499997735	4	1.64999997616	0.02
7	g.165995836_165995840delinsT	-	chr2	165995836	165995840	TTTAA	T	3.03150010109	2	0.921000003815	0.00
8	g.165995889G>C	-	chr2	165995889	165995889	G	C	3.07249999046	3	-2.90499997139	0.01
9	g.165998005C>T	-	chr2	165998005	165998005	C	T	3.07369995117	3	-1.14999997616	0.01
10	g.165999649G>A	-	chr2	165999649	165999649	G	A	3.21959996223	5	2.25999999046	0.02

Showing 1 to 10 of 66 entries

Previous 2 3 4 5 6 7 Next



BEACON

<https://beacon-network.org>



[Search](#) [Directory](#) [Developers](#)

Advanced Options

GRCh38 ▾

19 : 58353113 T > C

Search

Response All None

- Found 3
- Not Found 13
- Error 46

Organization All None

- AMPLab, UC Berkeley
- BGI
- BioReference Laboratori...
- Brazilian Initiative on Pre...
- BRCA Exchange



AMPLab - 1000 Genomes Pr...

AMPLab, UC Berkeley

Found



BIPMed

Brazilian Initiative on Precision Medicine

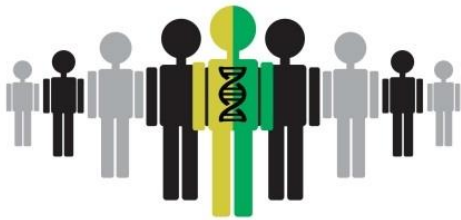
Found



OpenSNP - Personal Genom...

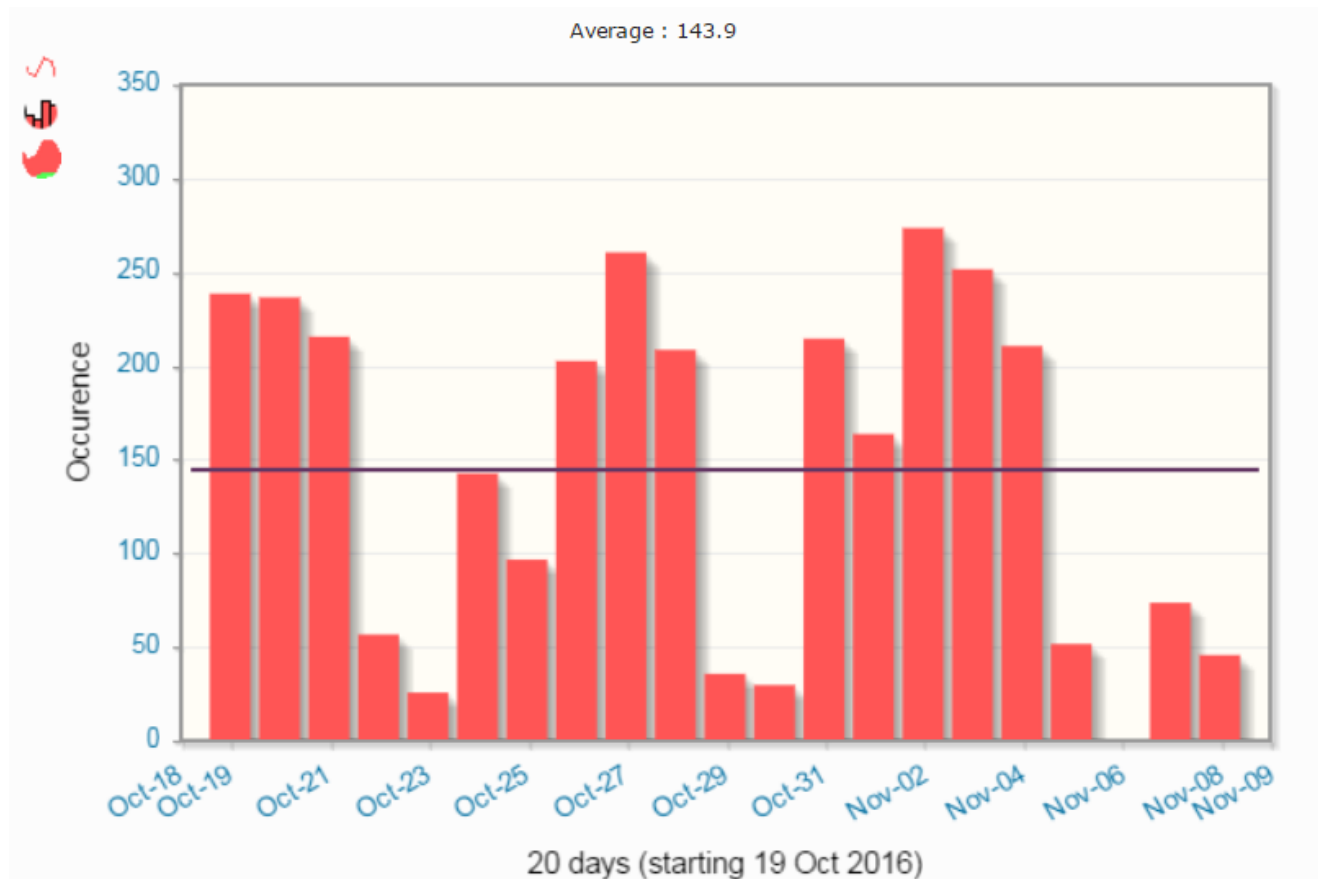
OpenSNP

Found



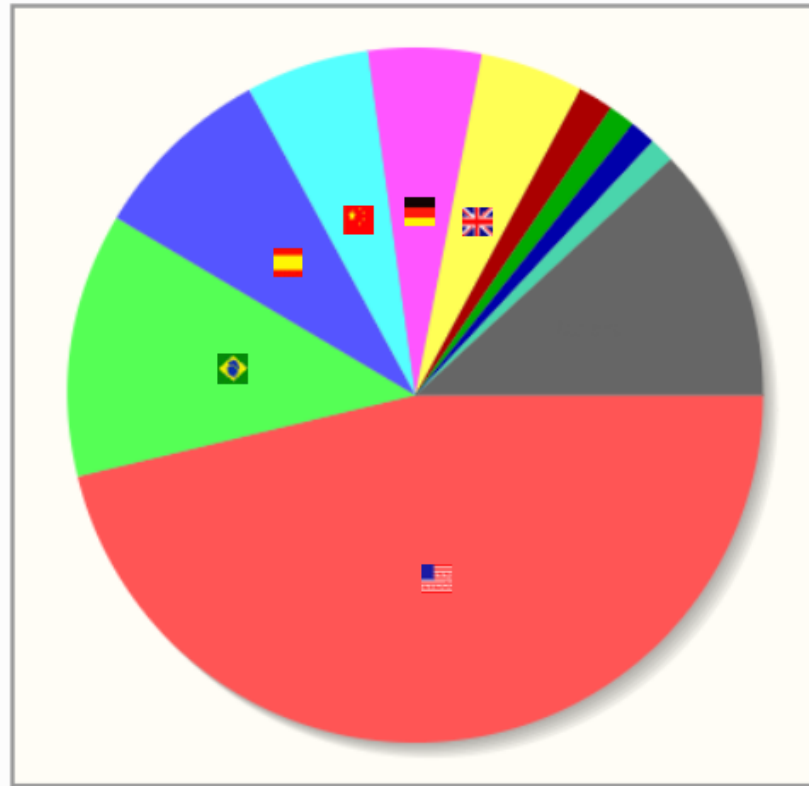
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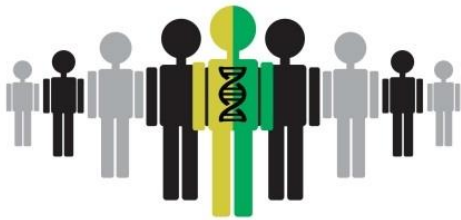
Access Statistics



Access Statistics

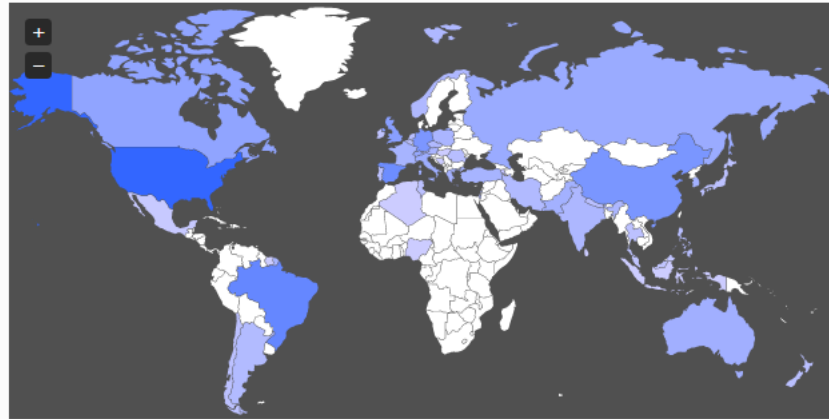
Countries		
United States - 	9414	46.2 %
Brazil - 	2501	12.2 %
Spain - 	1741	8.5 %
China - 	1168	5.7 %
Germany - 	1072	5.2 %
Great Britain - 	979	4.8 %
Italy - 	332	1.6 %
Australia - 	252	1.2 %
Russian Federation - 	248	1.2 %
Canada - 	238	1.1 %
Others	2415	11.8 %





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Access Statistics




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	<i>Total</i>	<i>external</i>	<i>local</i>
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

