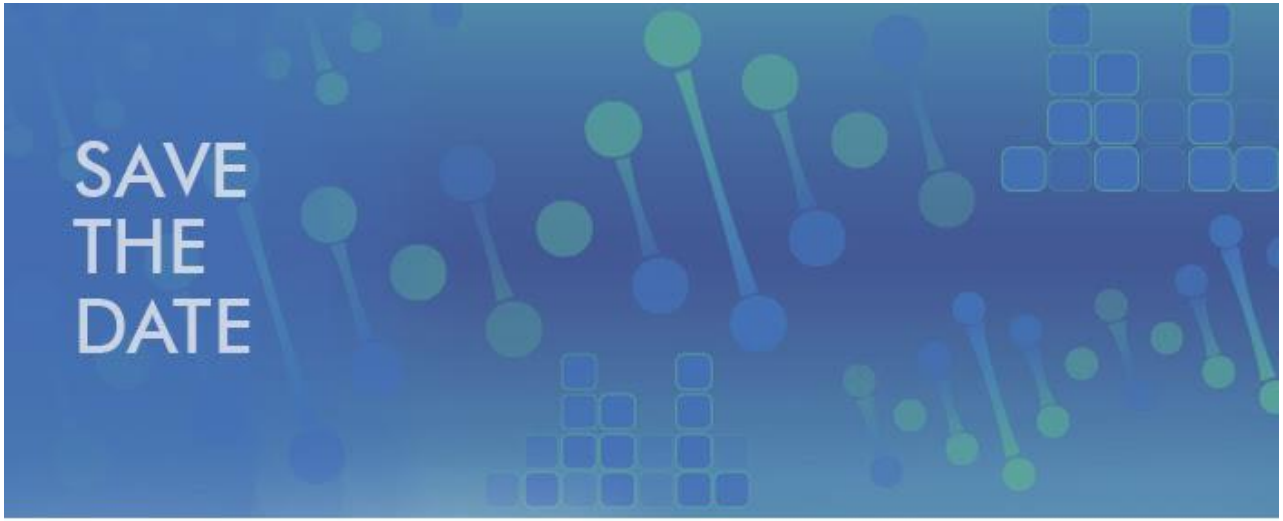
BIPMed - Brazilian Initiative on Precision Medicine

BIPMed Brazilian Initiative on PRECISION MEDICINE

www.bipmed.org

Global Alliance
for Genomics & Health



The São Paulo Research Foundation, FAPESP, under the scope of the program Research, Innovation and Dissemination Centers (RIDCs), and the Brazilian Initiative on Precision Medicine, BIPMed, invite you to the

BIPMED: ONE YEAR AND FORWARD

November 10th, 2016

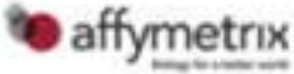
8:45am to 12:30pm

A meeting to celebrate the one-year launching of the Brazilian Initiative on Precision Medicine (BIPMed)

Partnership

Global Alliance
for Genomics & Health

Supported by



BIPMed

- BIPMed is an initiative of five Research Innovation and Dissemination Centers (RIDCs) supported by FAPESP
  - The Brazilian Research Institute for Neuroscience and Neurotechnology (BRAINN) – *Iscia Lopes-Cendes*
  - Center for Computational Science and Engineering (CECC) – *Claudia Bauzer Medeiros*
  - Center for Research in Cell Therapy (CTC) – *Wilson Silva Jr.*
  - Obesity and Comorbidities Research Center (OCRC) – *Joseane Morari*
  - Center for Research on Inflammatory Diseases (CRID) – *Wilson Silva Jr.*

BIPMed

Brazilian Initiative on Precision Medicine

FAPESP

BIPMed@bipmed.iqm.unicamp.br

QUESTIONS / SUGGESTIONS: Send us an email!

alpha-1-B glycoprotein (A1BG)

Curator: Admin

Genes | Transcripts | Variants | Diseases | Documentation

View all genes

20857 entries on 209 pages. Showing entries 1 - 100.

100 per page ▼ « First ‹ Prev 1 2 3 4 5 6 7 8 9 10 11 ... Next › Last »

Symbol	Gene	Chr	Band	Transcripts	Variants	Unique variants	Last updated	Associated with diseases
A1BG	alpha-1-B glycoprotein	19	q13.43	1	148	0	2015-12-17	-
A1BG-AS1	A1BG antisense RNA 1	19	q13.43	1	192	1	2015-12-17	-
A1CF	APOBEC1 complementation factor	10	q21.1	10	109	2	2015-12-26	-
A2M	alpha-2-macroglobulin	12	p13.31	2	304	0	2015-12-26	-
A2M-AS1	A2M antisense RNA 1 (head to head)	12	p13.31	1	22	0	2015-12-15	-
A2ML1	alpha-2-macroglobulin-like 1	12	p13	4	455	1	2015-12-26	-
A2MP1	alpha-2-macroglobulin pseudogene 1	12	p13.31	1	2	0	2015-12-15	-
A3GALT2	alpha 1,3-galactosyltransferase 2	1	p35.1	1	9	6	2015-12-13	-
A4GALT	alpha 1,4-galactosyltransferase	22	q13.2	10	147	3	2015-12-26	-
A4GNT	alpha-1,4-N-acetylglucosaminyltransferase	3	p14.3	1	107	6	2015-12-19	-
AAAS	achalasia, adrenocortical insufficiency, alacrimia	12	q13	9	200	3	2015-12-15	-
AACS	acetoacetyl-CoA synthetase	12	q24.31	5	127	6	2015-12-14	-
AACSP1	acetoacetyl-CoA synthetase pseudogene 1	5	q35	1	163	2	2015-12-26	-
AADAC	arylacetamide deacetylase	3	q25.1	2	249	16	2015-12-26	-
AADACL2	arylacetamide deacetylase-like 2	3	q25.1	2	64	9	2015-12-19	-
AADACL2-AS1	AADACL2 antisense RNA 1	3	q25.1	0	0	0	2016-03-16	-
AADACL3	arylacetamide deacetylase-like 3	1	p36.21	2	190	13	2015-12-07	-
AADACL4	arylacetamide deacetylase-like 4	1	p36.21	5	13	0	2015-12-12	-
AADAT	aminoadipate aminotransferase	4	q33	6	69	2	2015-12-26	-
AAED1	AhpC/TSA antioxidant enzyme domain containing 1	9	q22.32	3	1	0	2015-12-03	-
AAGAB	alpha- and gamma-adaptin binding protein	15	q23	6	138	3	2015-12-16	-
AAK1	AP2 associated kinase 1	2	p13.3	1	346	0	2015-12-26	-
AAMDC	adipogenesis associated, Mth938 domain containing	11	q14.1	4	46	2	2015-12-14	-
AAMP	angio-associated migratory cell protein	2	q	2	30	1	2015-12-07	-
AANAT	aralkylamine N-acetyltransferase	17	q25.1	9	37	5	2015-12-26	-
AAR2	AAR2 splicing factor homolog	20	q11.23	5	9	0	2015-12-18	-
AARD	alanine and arginine rich domain containing protein	8	q24.11	2	15	0	2015-12-24	-

bipmed.iqm.unicamp.br

Beacon

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OpenSNP - Personal Genom…
OpenSNP

GENOMICS

A federated ecosystem for sharing genomic, clinical data

Silos of genome data collection are being transformed into seamlessly connected, independent systems

The Global Alliance for Genomics and Health*

Early data-sharing efforts have led to improved variant interpretation and development of treatments for rare diseases and some cancer types (*1*–*3*). However, such benefits will only be available to the general population if researchers and clinicians can access and make comparisons across data from millions of individuals.

POLICY

Despite much progress, genomic and clinical data are still generally collected and studied in silos: by disease, by institution, and by country. Regulatory data-privacy requirements do not seamlessly lend themselves to the secure sharing of data within and across institutions and countries (*4*). Current practices in research and medicine hinder the sharing of data in ways that tangibly recognize an individual's contributions. Tools and analytical methods are nonstandardized and incompatible, and the data are often stored in incompatible file formats.

Science
AAAS

A federated ecosystem for sharing genomic, clinical data

The Global Alliance for Genomics and Health (June 9, 2016)
Science **352** (6291), 1278-1280. [doi: 10.1126/science.aaf6162]

REMAINING CHALLENGES. Shringarpure and Bustamante (*11*) used simulations to show that, in some scenarios, querying a public beacon for as few as 250 variants already known to be present in an individual's genome could reveal information distinctive to that individual. GA4GH members have been developing solutions to this potential security breach since the project's inception, including aggregating data among multiple beacons, tracking usage to restrict systematic searches and introducing tiers of secured access that require users to be authorized for data access—but these necessarily limit the scope of information that can be shared widely. Innovative policy and regulatory measures, as well as technological solutions, are needed to securely handle individual genomic and clinical data.

Finally, ensuring engagement among the entire global community is necessary from a social justice and medical perspective, although this will likely require distinct legal, cultural, and business models. In some countries, health care and research organizations are interested in GA4GH as a means to link nascent national efforts in precision medicine with other international groups, such as the Brazilian Initiative on Precision Medicine (www.fcm.unicamp.br/gtc/evento/1/trabalho/8).

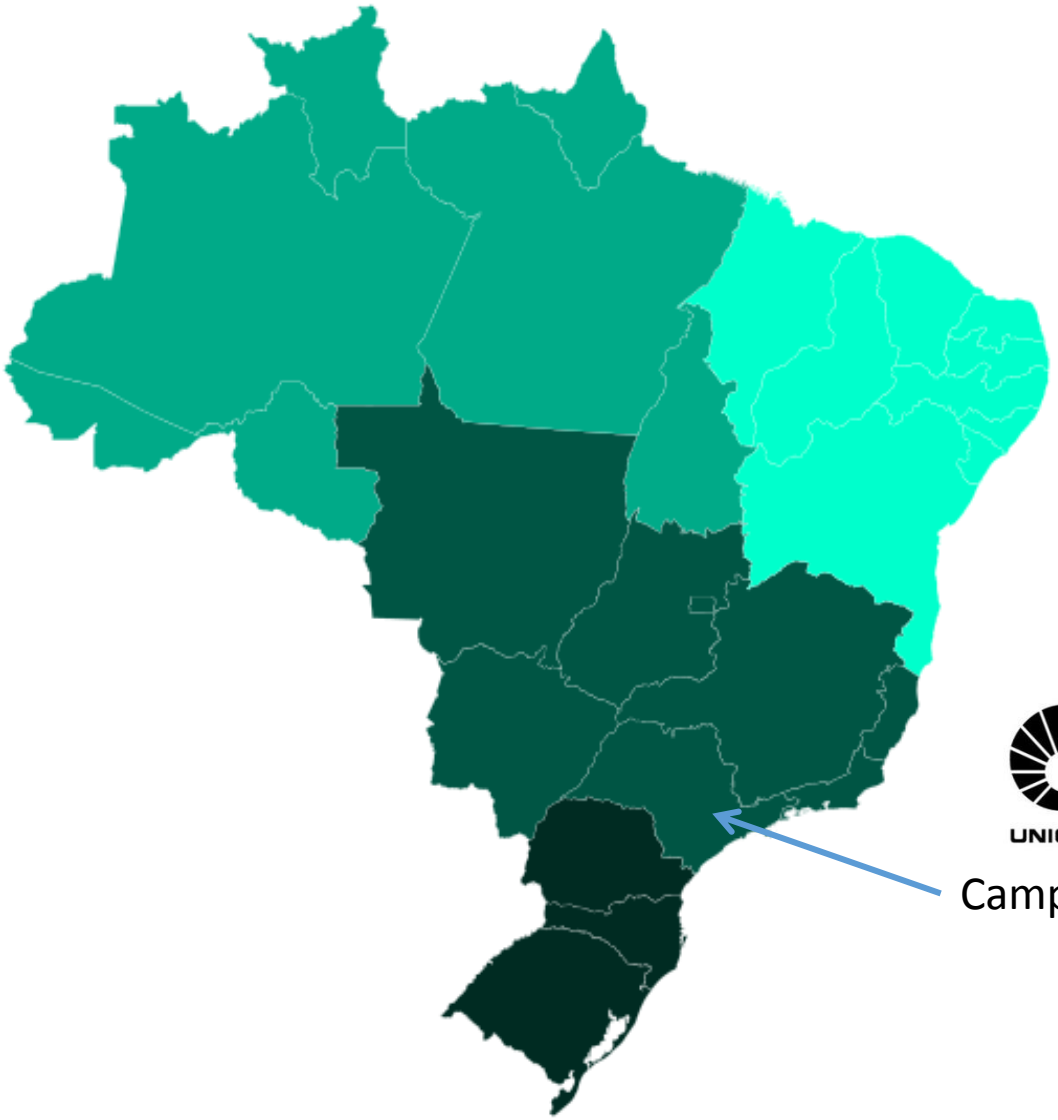
Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

National Initiatives 'Pre-Meeting' at GA4GH 4th Plenary - Agenda

Date: October 17, 2016
Time: 1-3pm PDT | 8-10pm UTC
Location: 'Ambleside 2' room at Vancouver Marriott Pinnacle Downtown Hotel
1128 W Hastings St., Vancouver, Canada
Contact: Lena Dolman (lena.dolman@genomicsandhealth.org)

Attendees:

- GA4GH: Kathryn North, Peter Goodhand, Julia Wilson, Lena Dolman
- Australia and AGHA: Sean Grimmond, John Christodoulou, Andrew Sinclair, Marcel Dinger, Clara Gaff, Sylvia Metcalfe, Oliver Hofmann
- Genomics England: Augusto Rendon, Mark Caulfield
- Genome Canada: Cindy Bell, Kate Swan
- French National Genotyping Centre: Jean-François Deleuze
- Brazilian Society of Medical Genetics: Iscia Cendes-Lopes
- H3Africa: Nicola Mulder
- Cancer Moonshot blue ribbon panel: Angel Pizarro
- Precision Medicine Initiative: David Glazer
- National Cancer Centre of Singapore: Bin Tean Teh (via Zoom)



BIPMed Brazilian Initiative on PRECISION MEDICINE

UNICAMP

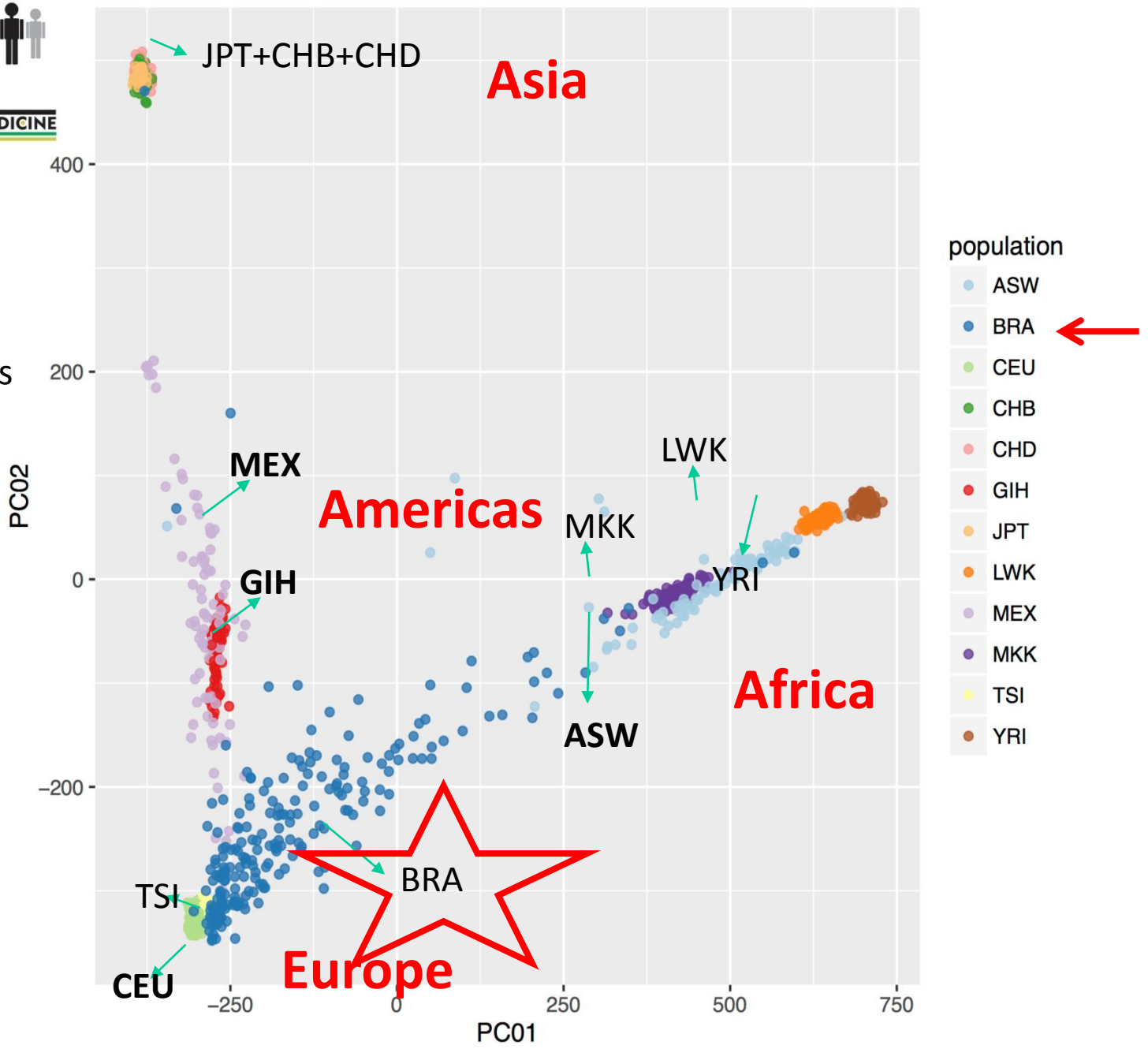
Campinas, SP

Reference Individuals

BIPMed
SNP-arrays
196 individuals

x

HapMap



Ananina G et al. 2016
submitted

Fine scale genetic structure of the populations from three Brazilian regions

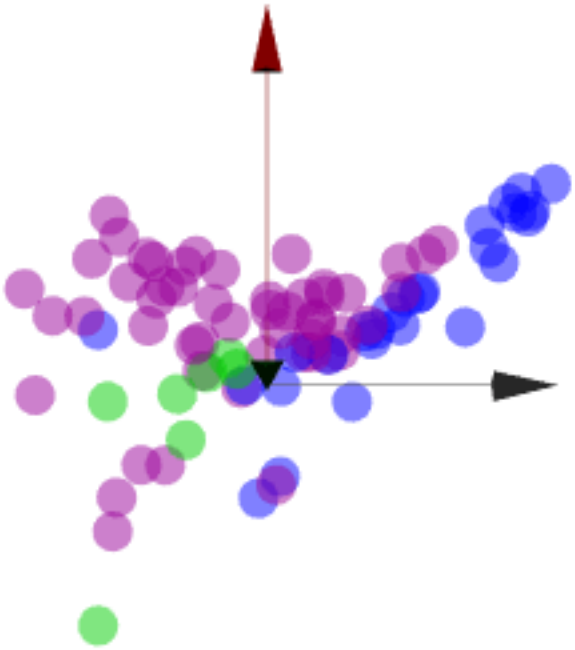
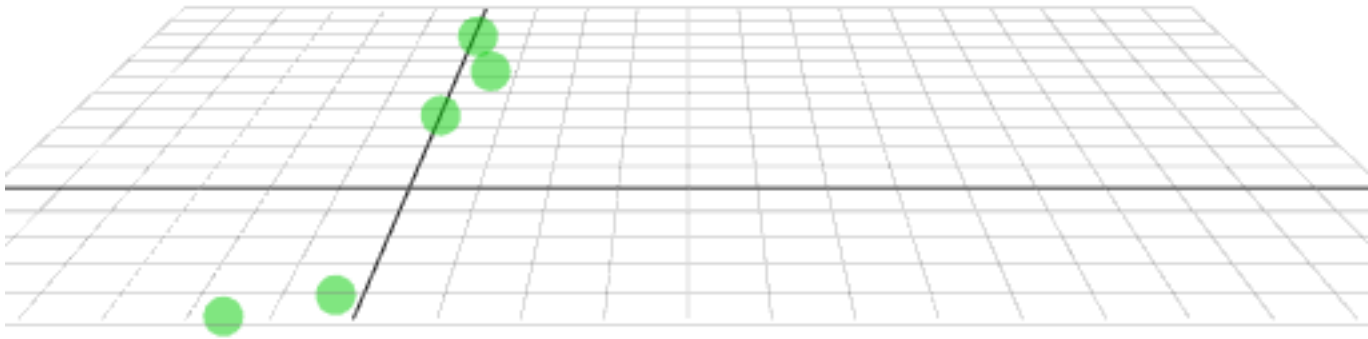
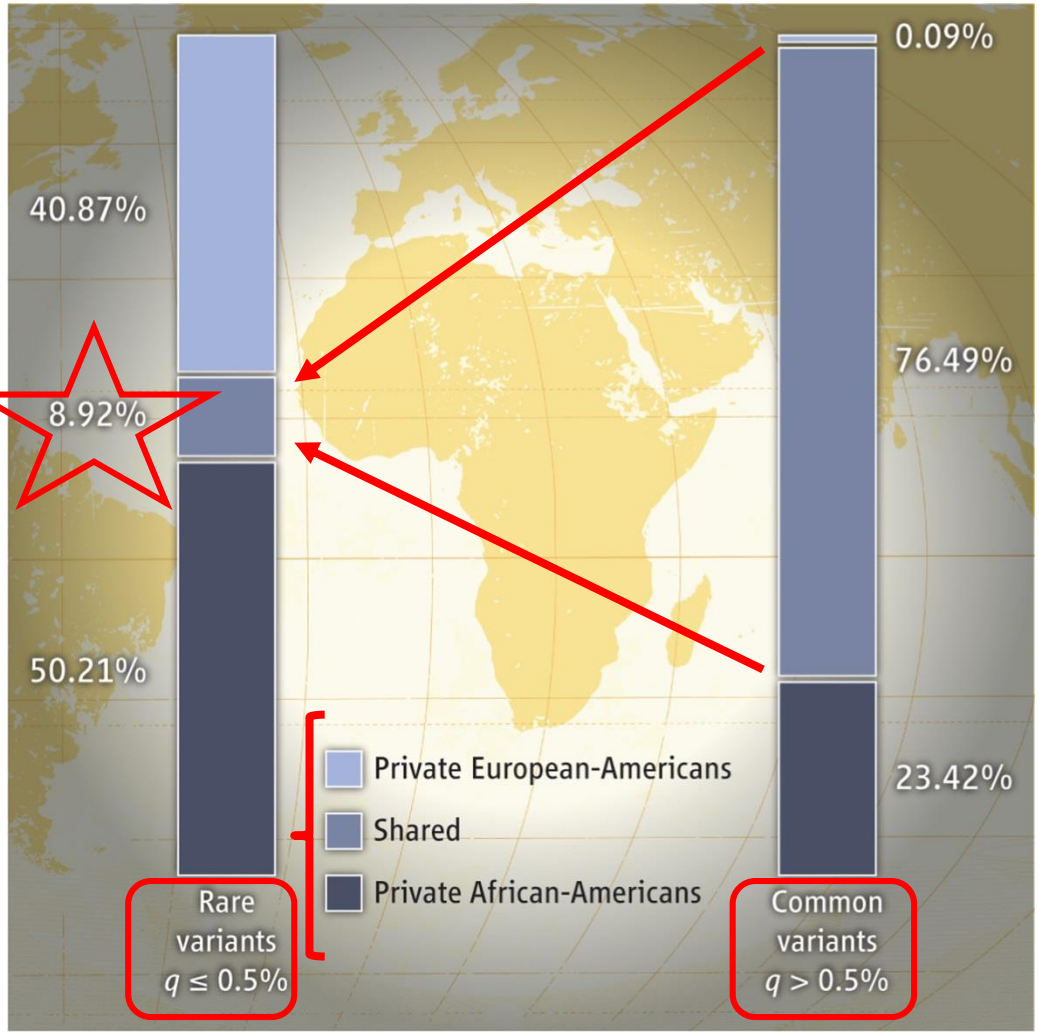


Gráfico de Componentes Principais: PC1 x PC2 x PC3

Recife, PE
Campinas, SP
Joinville,SP



Human genetic variation.Proportion of shared and unshared (private) variants between the African-American and the European-American populations [data from (1)].



Ferran Casals, and Jaume Bertranpetit Science 2012;337:39-40

Published by AAAS

Conclusions

- The first product of BIPMed has been successfully implemented
- There is worldwide interest in BIPMed
- Planed expansions in the genomic database
- PIBMed additional products:
  - Computer Interface for the identification of variants of clinical relevance in WES and WGS.
  - Work in conjunction with the hospital informatics team to improve and **integrate** electronic medical records