# IV WORKSHOP

# BIPMed - Brazilian Initiative on Precision Medicine

**www.bipmed.org**

**SAVE THE DATE**

The São Paulo Research Foundation, FAPESP, under the scope of the program Research, Innovation and Dissemination Centers (RIDCs), invite you to the

Launching of the
# BRAZILIAN INITIATIVE ON PRECISION MEDICINE BIPMed

**November, 13, 2015**
1:30pm to 5:00pm
Venue:  FAPESP - Rua Pio XI, 1500
Alto da Lapa – São Paulo

**Partnership**

Global Alliance
for Genomics & Health

**Supported by**

affymetrix          illumina

# Mission

- To help implement precision medicine in Brazil by acting as a catalytic element to foster collaboration among different stake holders (scientist, physicians, health authorities, hospitals, society)

First product: BIPMed genomic database

# Support

# Steering Committee

- **Iscia Lopes Cendes** – Professor of the School of Medical Science, University of Campinas (FCM/UNICAMP)

- **Munir Skaf –** Professor of the Institute of Chemistry, University of Campinas (IQ/UNICAMP)

- **Wilson Araújo da Silva Jr** – Associate Professor of the School of Medicine, University of São Paulo at Ribeirão Preto (FMRP/USP)

- **Claudia Bauzer Medeiros** – Professor of the Institute of Computing, University of Campinas (IC/UNICAMP)

- **Benilton de Sá Carvalho** – Assistant Professor – Institute of Mathematics, Statistics and Computer Sciences, University of Campinas (IMECC/UNICAMP)

- **Marcelo Briones** – Associate Professor – UNIFESP/EPM

# Technical Committee

**Benilton de Sá Carvalho** – Assistant Professor – Institute of Mathematics, Statistics and Computer Sciences, University of Campinas (IMECC/UNICAMP)

**Cristiane Rocha** – Research Associate, Biostatistics and Computation Biology Laboratory (BCB), School of Medical Science, University of Campinas (FCM/UNICAMP) – **PROJECT MANAGER**
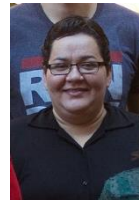
# GENOMICS

# 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.

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.

**POLICY**

# BEACON



https://beacon-network.org

# 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-Francois Deleuze
- Brazilian Society of Medical Genetics: Iscia Cendes-Lopes          ← **BIPMed**
- 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)

**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).
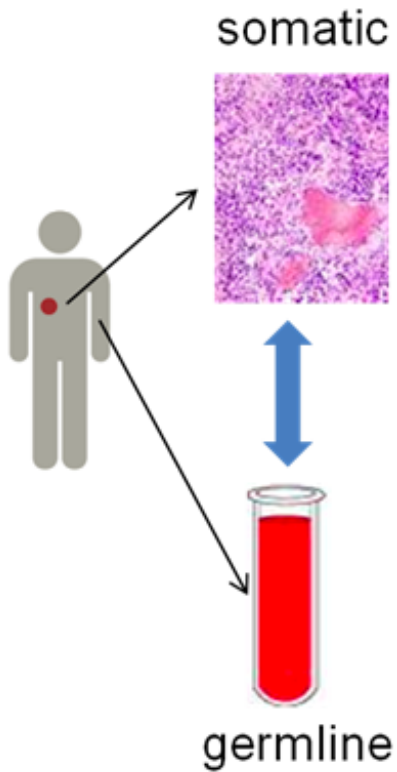
# Precision Medicine



Yamamoto et al. 2014
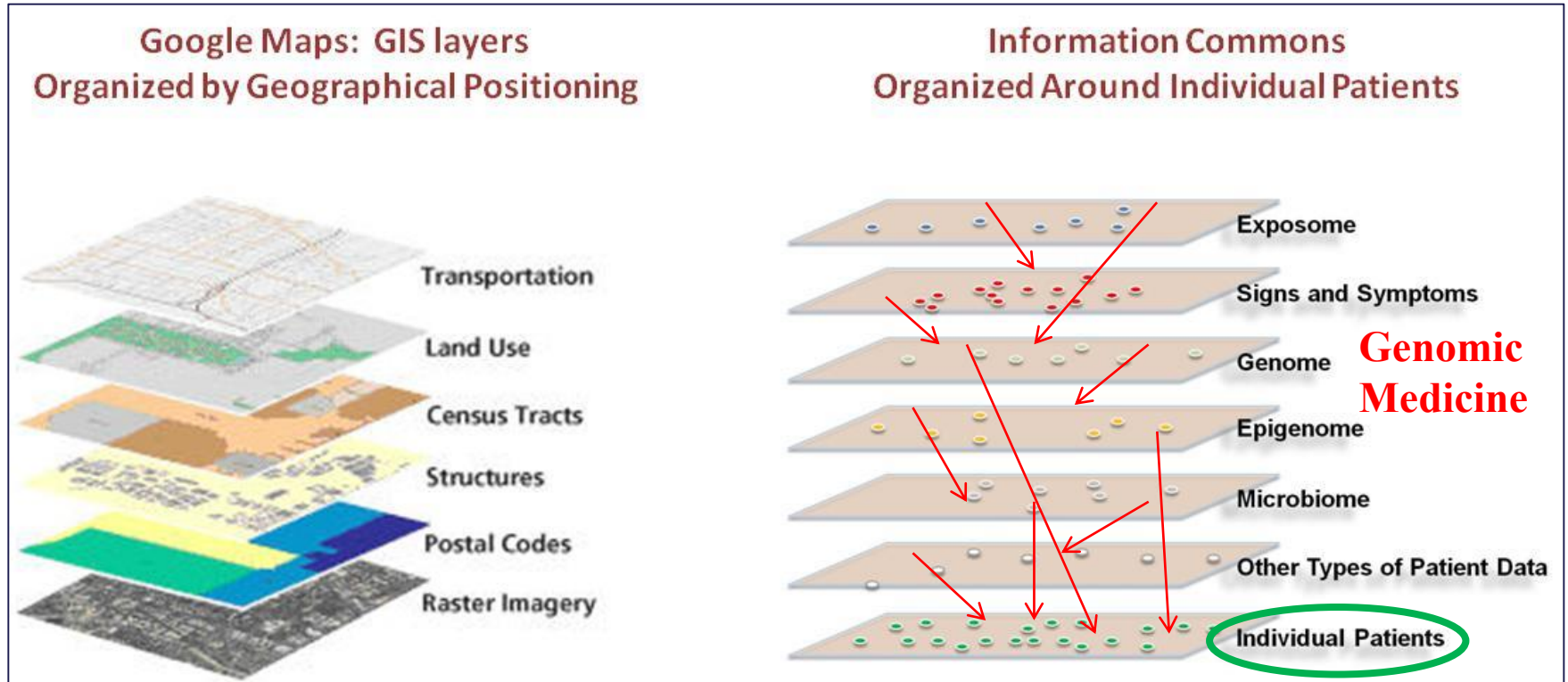
# Precision Medicine

- Diverse data types: *e.g., -omics, imaging (e.g., brain activity, longitudinal MRI), population studies, environmental effects.*

- Digital health: *wearable sensors (biosensors)*

- Data acquisition, aggregation, integration, analysis

- Data storage, security, selective access

- Data sorting and visualization

- **Data sharing**

# Populations are Important

# WGS with ancestral haplotypes
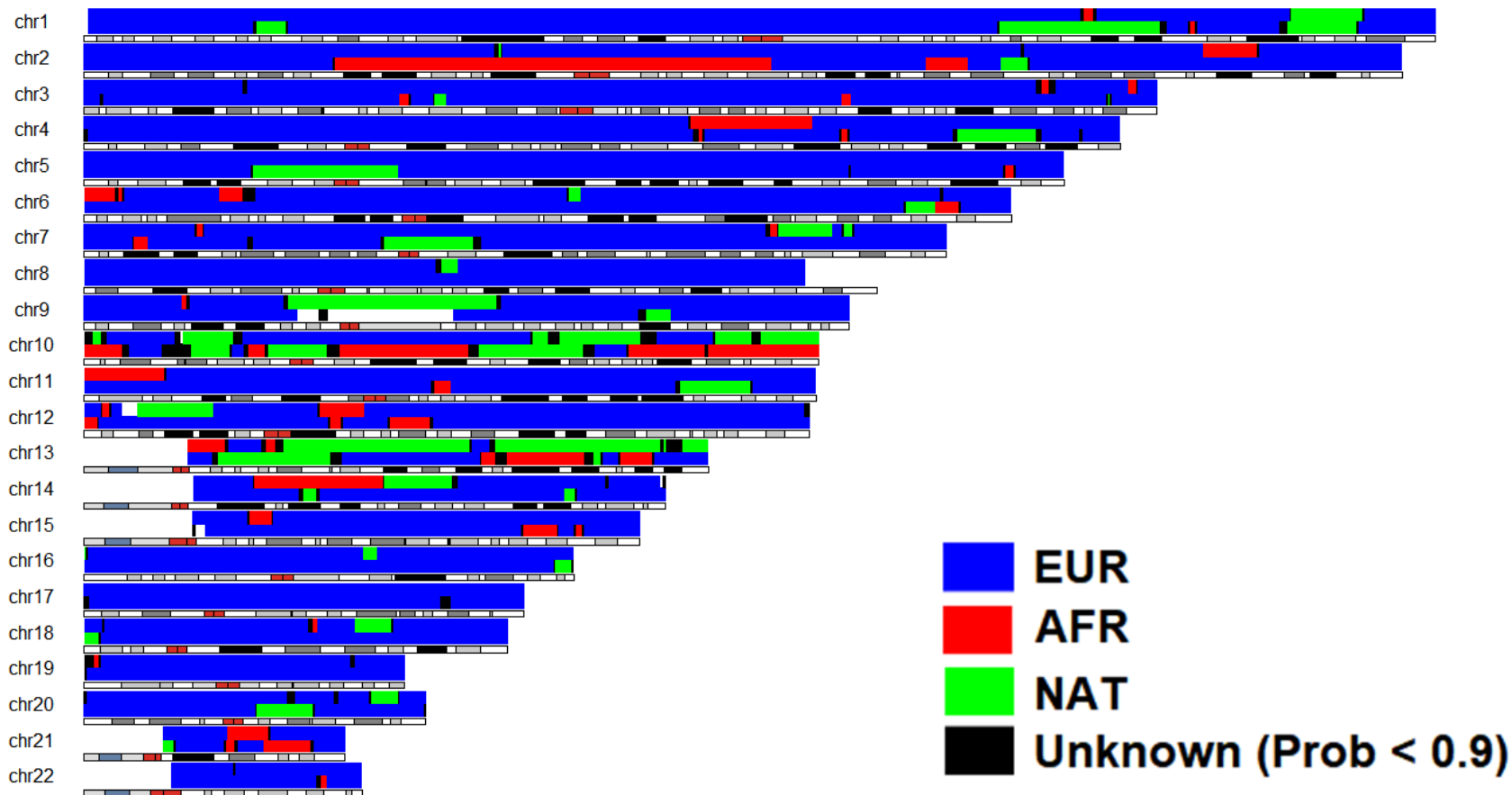
# Visit us at

# www.bipmed.org