PIBMed - Brazilian Initiative on Precision Medicine

BIPMed

- BIPMed is an initiative of five Research Innovation and Dissemination Centers (RIDCs) supported by FAPESP
  - The Brazilian Research Institute for Neuroscience an 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.*

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

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)

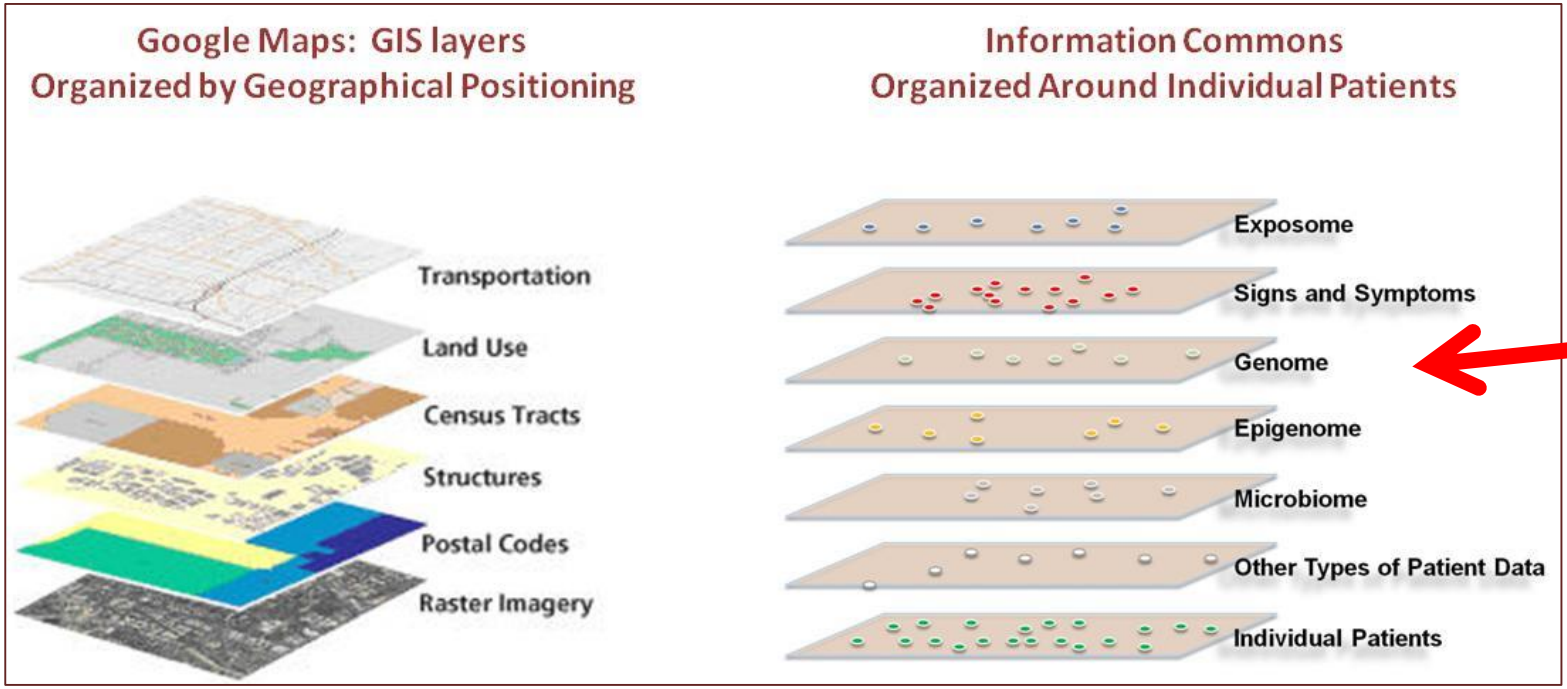
Technical Committee

- **Benilton de Sá Carvalho** – Assistant Professor – Institute of Mathematics, Statistics and Computer Sciences, University of Campinas (IMECC/UNICAMP)
- **Guilherme Telles** – Assistant Professor - Institute of Computing, University of Campinas (IC/UNICAMP)
- **Cristiane Rocha** – Research Associate, Biostatistics and Computation Biology Laboratory (BCB), School of Medical Science, University of Campinas (FCM/UNICAMP)

Precision Medicine

- ***Use massive data network that aggregates and analyzes information from large patient cohorts, healthy populations, experimental organisms and others to reach towards disease mechanisms, and precise diagnosis and treatment for each individual (Yamamoto et al. 2014).***

Precision Medicine



Yamamoto et a. 2014

Precision Medicine: informatics challenges

- *Big data*
- Diverse data types: e.g., -omics, imaging, population studies, environmental effects
- Digital health: wearable sensors (biosensors)
- Data acquisition, aggregation, integration, analysis
- Continuous learning
- Data storage, security, selective access
- Data sorting and visualization
- Data sharing

The NEW ENGLAND JOURNAL of MEDICINE

Perspective

FEBRUARY 26, 2015

A New Initiative on Precision Medicine

Francis S. Collins, M.D., Ph.D., and Harold Varmus, M.D

"Tonight, I'm launching a new Preci Initiative to bring us closer to curi cancer and diabetes — and to give all of personalized information we need to kee our families healthier."

— President Barack Obama, State of the Union Ad

President Obama has long expressed a strong conviction that science offers great potential for improving health. Now, the President has announced a research initiative that aims to accelerate progress toward a new era of precision medicine (www.whitehouse.gov/precisionmedicine). We believe that the time is right for this visionary initiative, and the National Institutes of Health (NIH) and other partners will work to achieve this vision.

The concept of precision medicine — prevention and treatment strategies that take individual variability ir new[1]; blood has been u transfusions tury. But the this concep dramatically cent develo biologic data man genom ful method patients (s metabolomi cellular ass health tech tational tool sets of data.

The White House

Office of the Press Secretary

For Immediate Release

January 30, 2015

FACT SHEET: President Obar

Key Investments to Launch the Precision Medicine Initiative:

Complementing robust investments to broadly support research, development, and innovation, the President's 2016 Budget will provide a $215 million investment for the National Institutes of Health (NIH), together with the Food and Drug Administration (FDA), and the Office of the National Coordinator for Health Information Technology (ONC) to support this effort, including:

$130 million to NIH for development of a voluntary national research cohort of a million or more volunteers to propel our understanding of health and disease and set the foundation for a new way of doing research through engaged participants and open, responsible data sharing.

$70 million to the National Cancer Institute (NCI), part of NIH, to scale up efforts to identify genomic drivers in cancer and apply that knowledge in the development of more effective approaches to cancer treatment.

$10 million to FDA to acquire additional expertise and advance the development of high quality, curated databases to support the regulatory structure needed to advance innovation in precision medicine and protect public health.

$5 million to ONC to support the development of interoperability standards and requirements that address privacy and enable secure exchange of data across systems.

ARTICLE

Privacy Risks from Genomic Data-Sharing Beacons

Suyash S. Shringarpure[1,*] and Carlos D. Bustamante[1,*]

The human genetics community needs robust protocols that enable secure sharing of genomic data from participants in genetic research. Beacons are web servers that answer allele-presence queries—such as "Do you have a genome that has a specific nucleotide (e.g., A) at a specific genomic position (e.g., position 11,272 on chromosome 1)?"—with either "yes" or "no." Here, we show that individuals in a beacon are susceptible to re-identification even if the only data shared include presence or absence information about alleles in a beacon. Specifically, we propose a likelihood-ratio test of whether a given individual is present in a given genetic beacon. Our test is not dependent on allele frequencies and is the most powerful test for a specified false-positive rate. Through simulations, we showed that in a beacon with 1,000 individuals, re-identification is possible with just 5,000 queries. Relatives can also be identified in the beacon. Re-identification is possible even in the presence of sequencing errors and variant-calling differences. In a beacon constructed with 65 European individuals from the 1000 Genomes Project, we demonstrated that it is possible to detect membership in the beacon with just 250 SNPs. With just 1,000 SNP queries, we were able to detect the presence of an individual genome from the Personal Genome Project in an existing beacon. Our results show that beacons can disclose membership and implied phenotypic information about participants and do not protect privacy a priori. We discuss risk mitigation through policies and standards such as not allowing anonymous pings of genetic beacons and requiring minimum beacon sizes.

Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

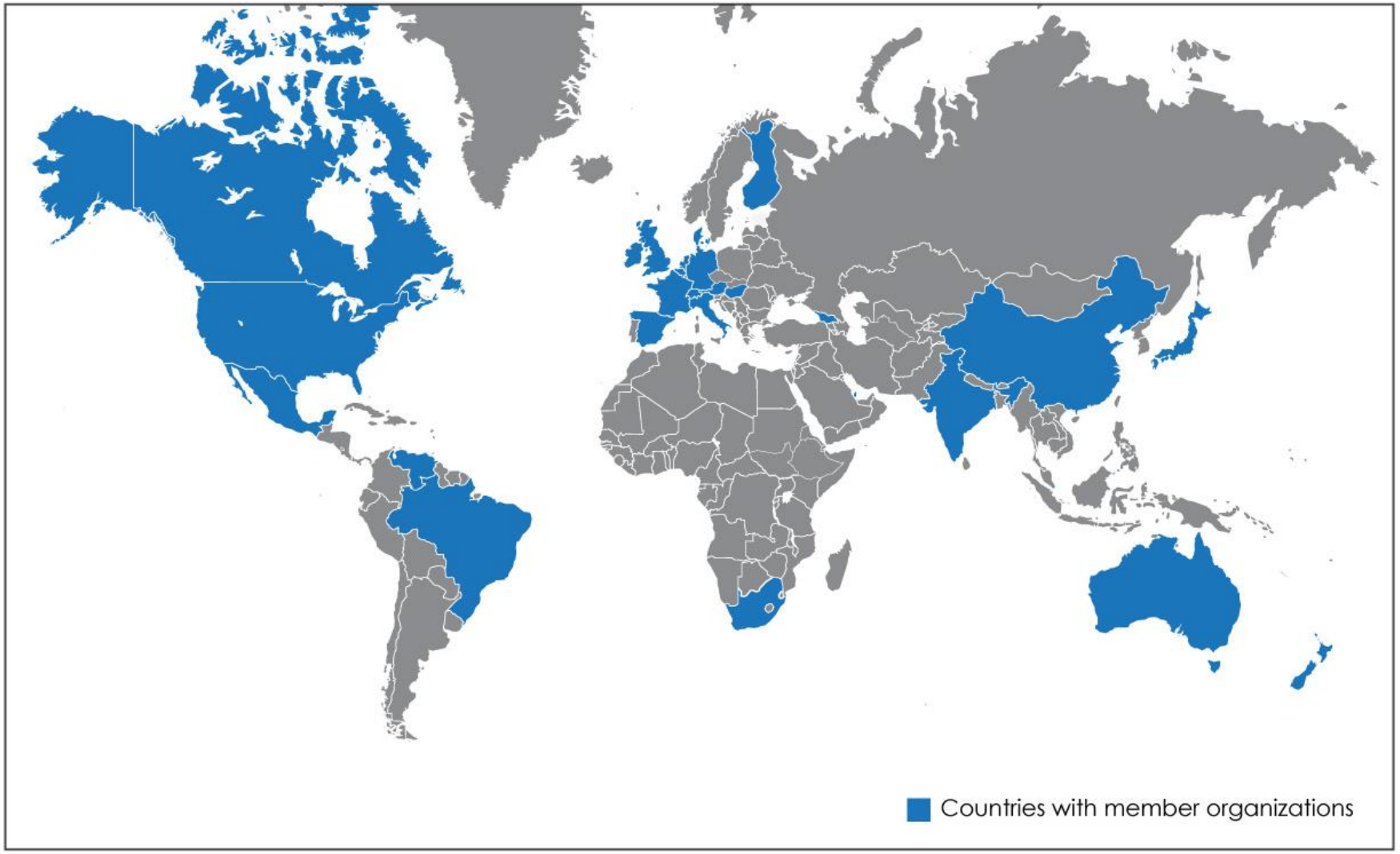
Mission

Global Alliance for Genomics & Health

To accelerate progress in human health by helping to establish a common framework of harmonized approaches to enable effective and responsible sharing of genomic and clinical data, and by catalyzing data sharing projects that drive and demonstrate the value of data sharing

Member Location

Global Alliance for Genomics & Health



Current data sharing projects

Global Alliance for Genomics & Health

Undertaken by the members, not by GA4GH as an organization. Catalyzed and supported by GA4GH coordinators and working groups.

Drive learning, identify requirements, evaluate value, coordinate activity.

- **Matchmaker Exchange**
- **BRCA Challenge**
- **Beacon Project**

BIPMed

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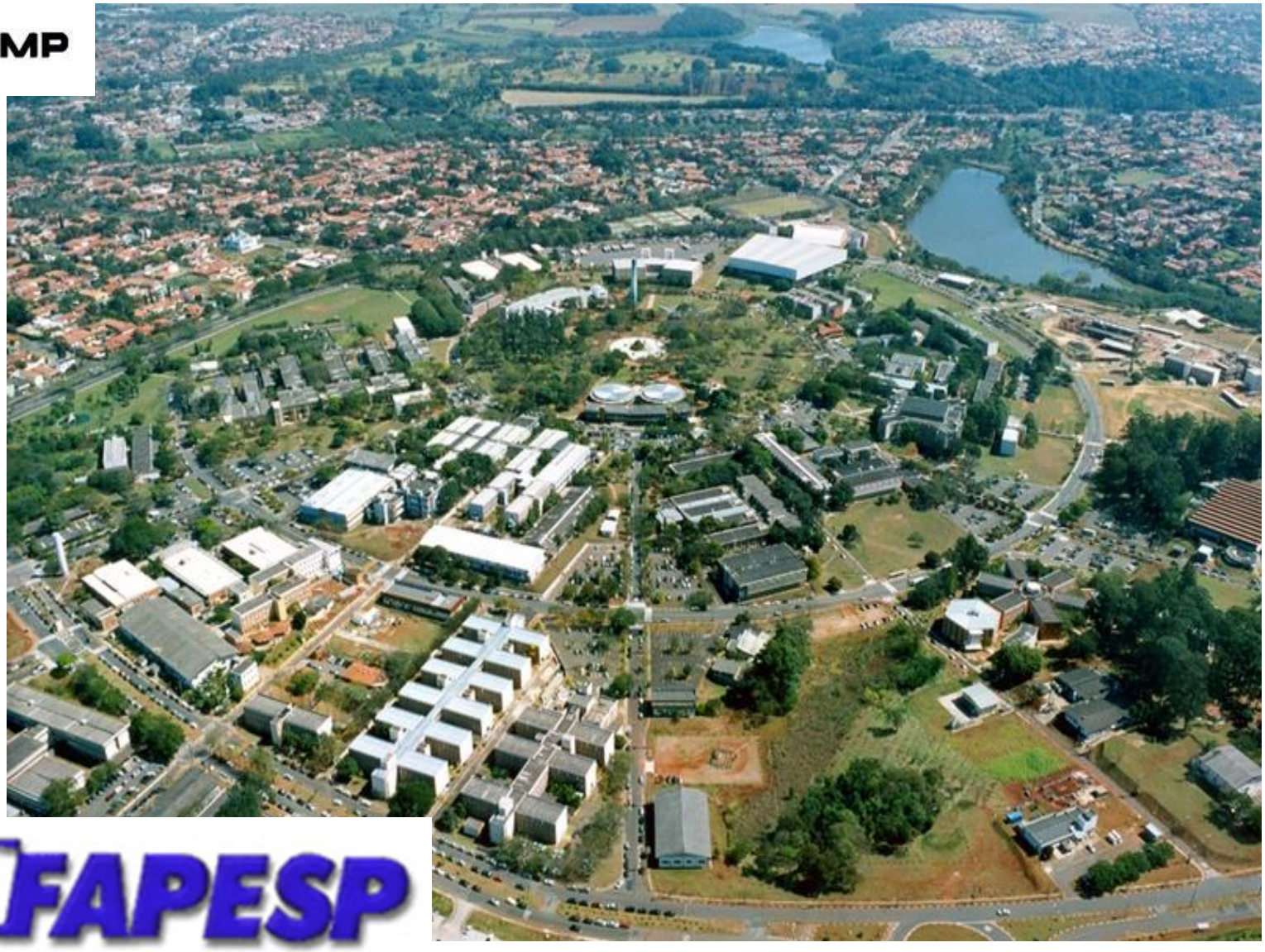

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