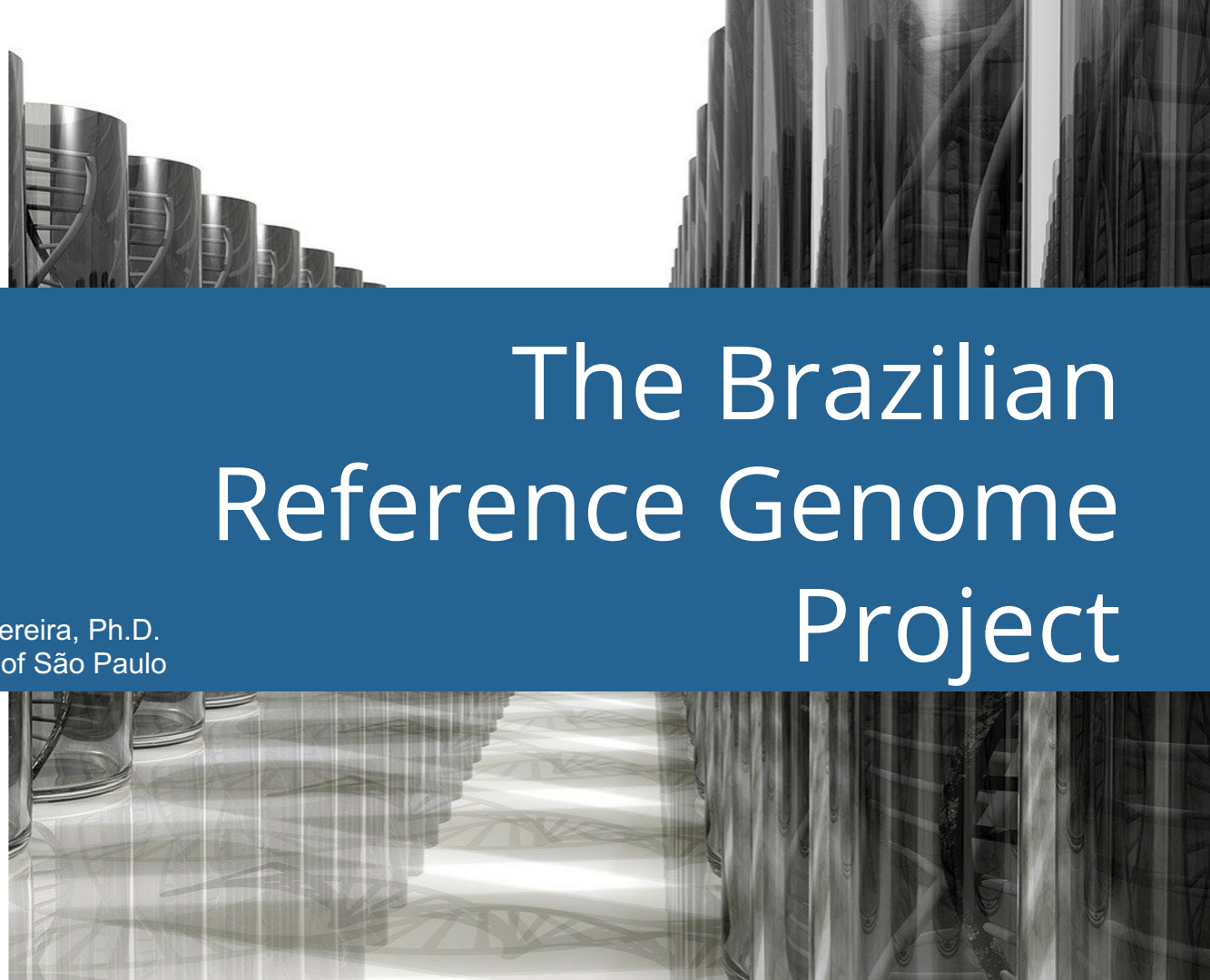
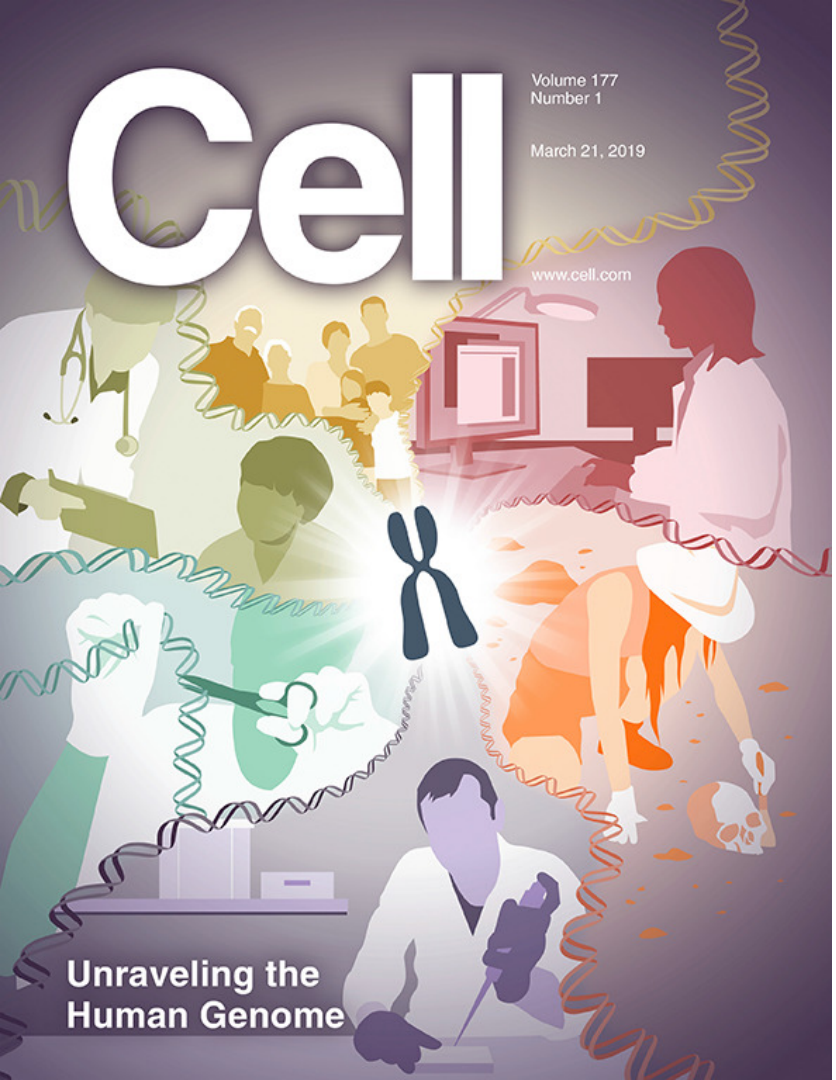
May 7th, 2020



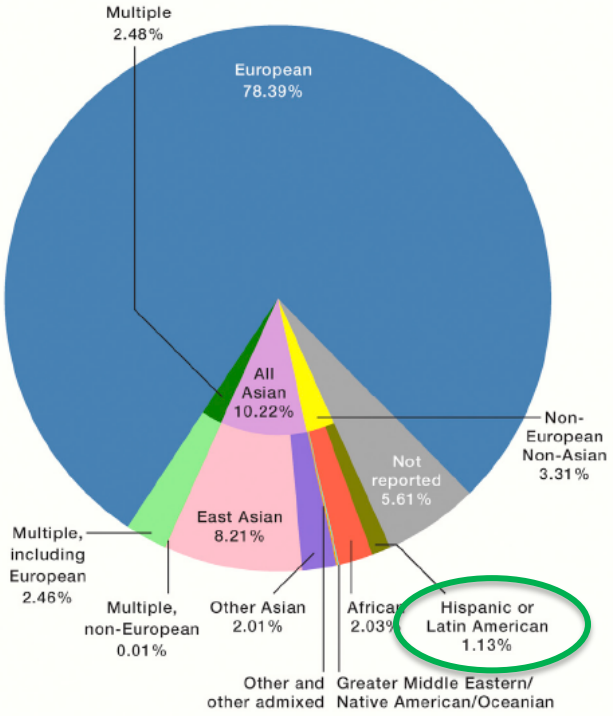
The Brazilian Reference Genome Project

Lygia V. Pereira, Ph.D.
University of São Paulo

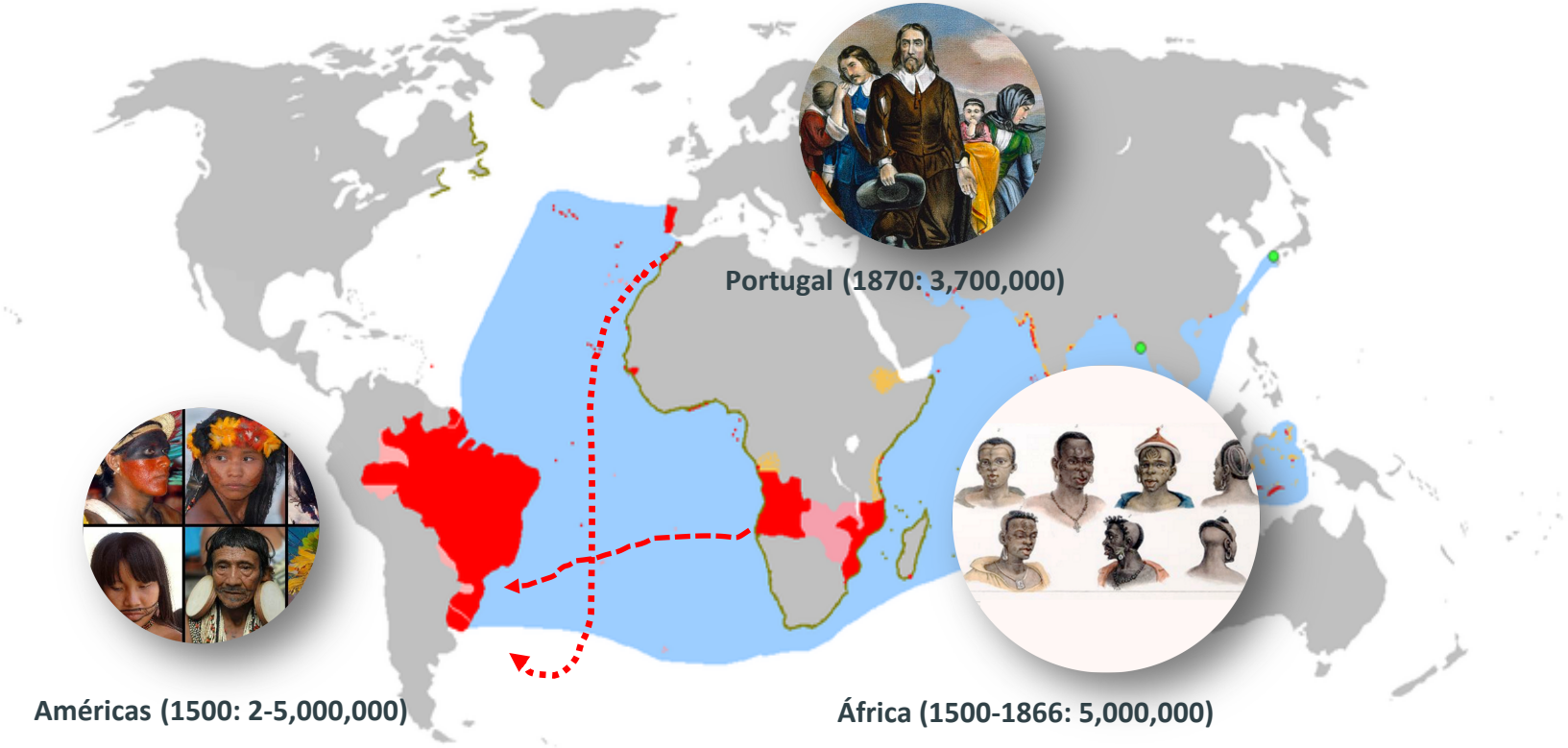
Global Genomic Medicine Collaborative



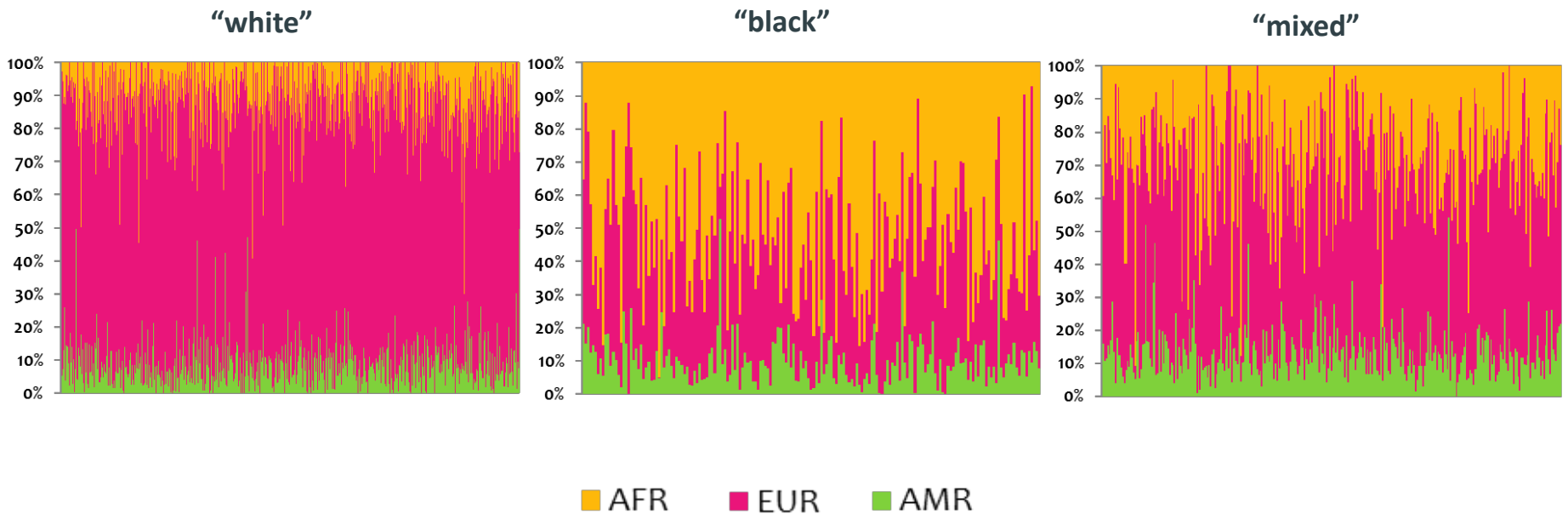
The Missing Diversity in Human Genetic Studies

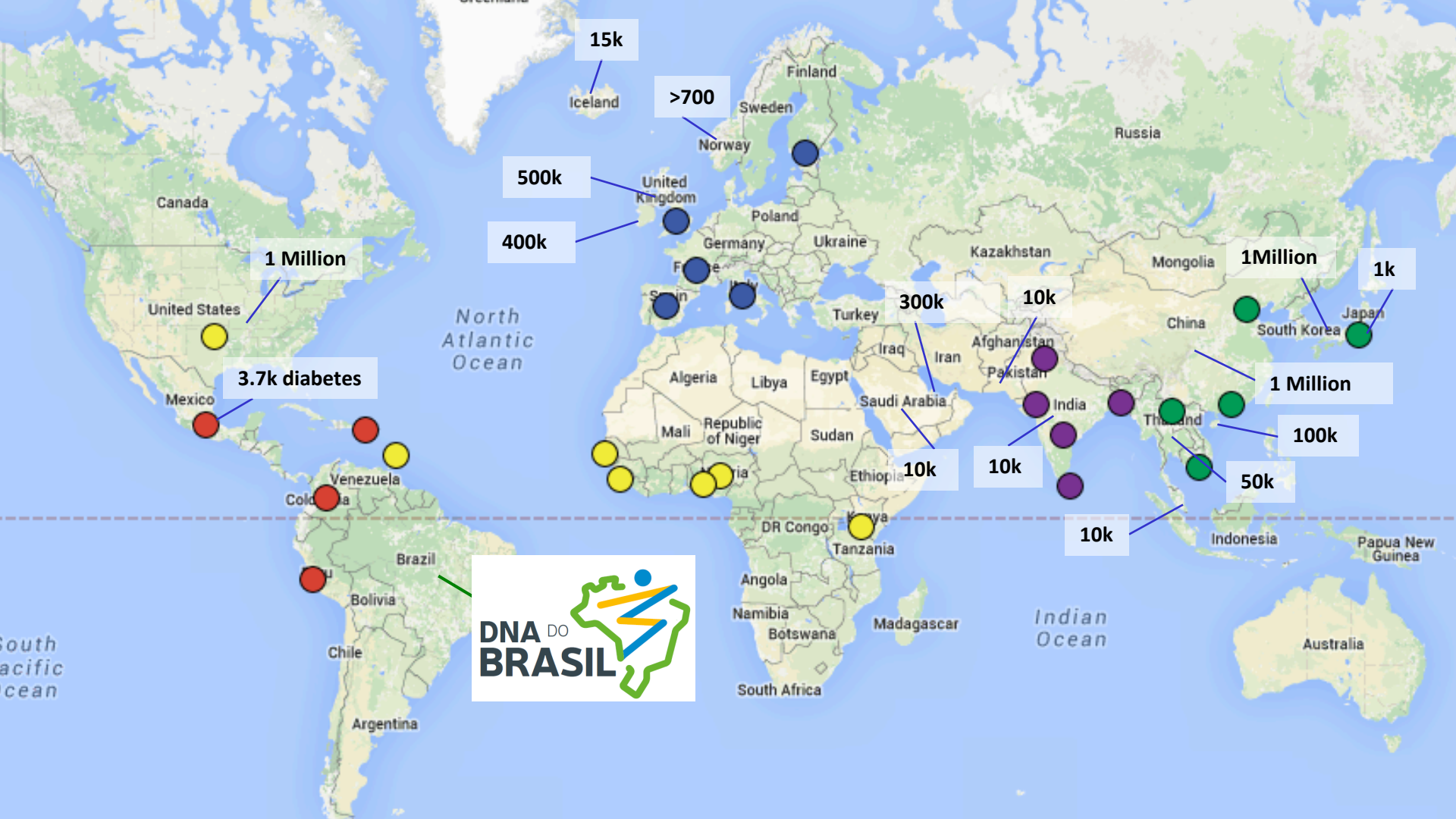


Genetic Background of the Brazilian Population



Brazilian Admixture



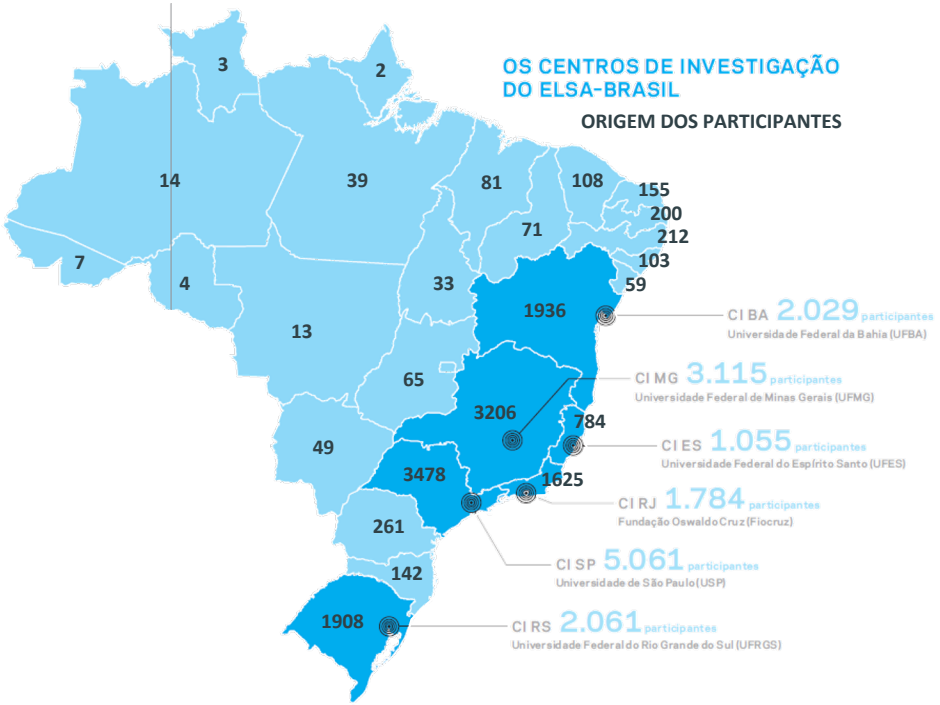

15k
Iceland
>700
Norway
Finland
Sweden
Russia
500k
United Kingdom
400k
Poland
Germany
Ukraine
Canada
1 Million
United States
North Atlantic Ocean
Kazakhstan
Mongolia
1Million
1k
Japan
South Korea
China
300k
10k
Turkey
Iraq
Iran
Afghanistan
Pakistan
3.7k diabetes
Mexico
Algeria
Libya
Egypt
Saudi Arabia
India
1 Million
Mali
Republic of Niger
Sudan
Ethiopia
100k
Thailand
10k
10k
50k
Venezuela
DR Congo
Tanzania
10k
Indonesia
Papua New Guinea
Brazil
Angola
Namibia
Botswana
Madagascar
Indian Ocean
Bolivia
Chile
Australia
South Africa
Argentina
DNA DO BRASIL

Estudo Longitudinal de Saúde do Adulto ELSA BRASIL

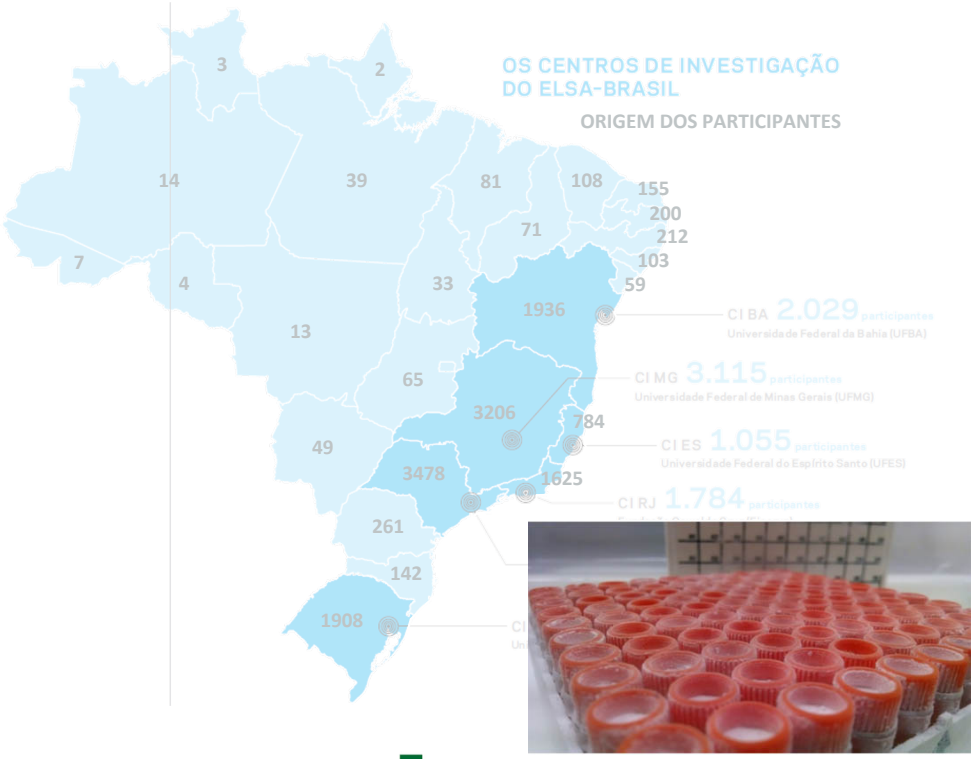
MINISTÉRIO DA SAÚDE

MINISTÉRIO DA CIÊNCIA, TECNOLOGIA, INOVAÇÕES E COMUNICAÇÕES

- **Largest epidemiologic study in Brazil**
- 15.105 public servers (35-74 yrs);
- Association btwn chronic diseases (cardiovascular/diabetes) and environmental/behaviour factors;
- Possible regional variations;



ELSA Brasil – Largest epidemiologic longitudinal study in Brazil



Estudo Longitudinal de Saúde do Adulto ELSA BRASIL

MINISTÉRIO DA SAÚDE

MINISTÉRIO DA CIÊNCIA, TECNOLOGIA, INOVAÇÕES E COMUNICAÇÕES

FUNDING ??

dasa

illumina

3.000 WGS

Google Cloud

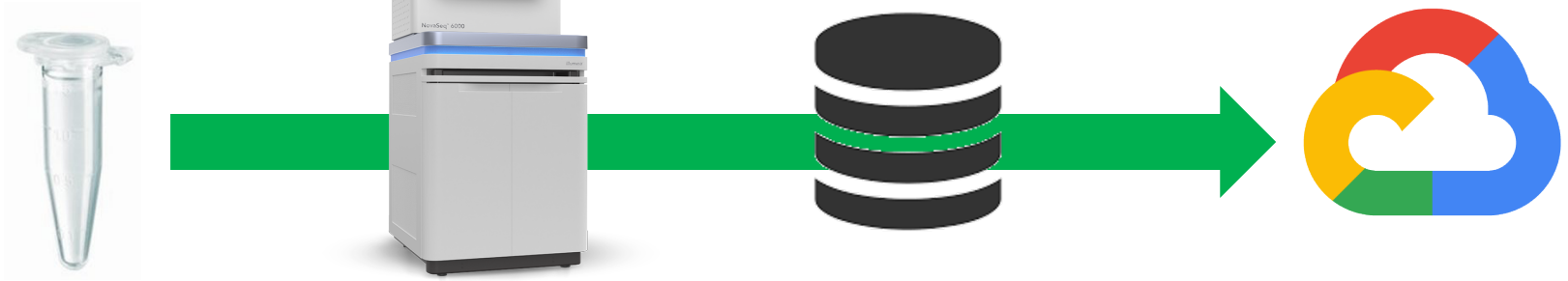
Processing/Storage

PINHEIRONETO
ADVOGADOS

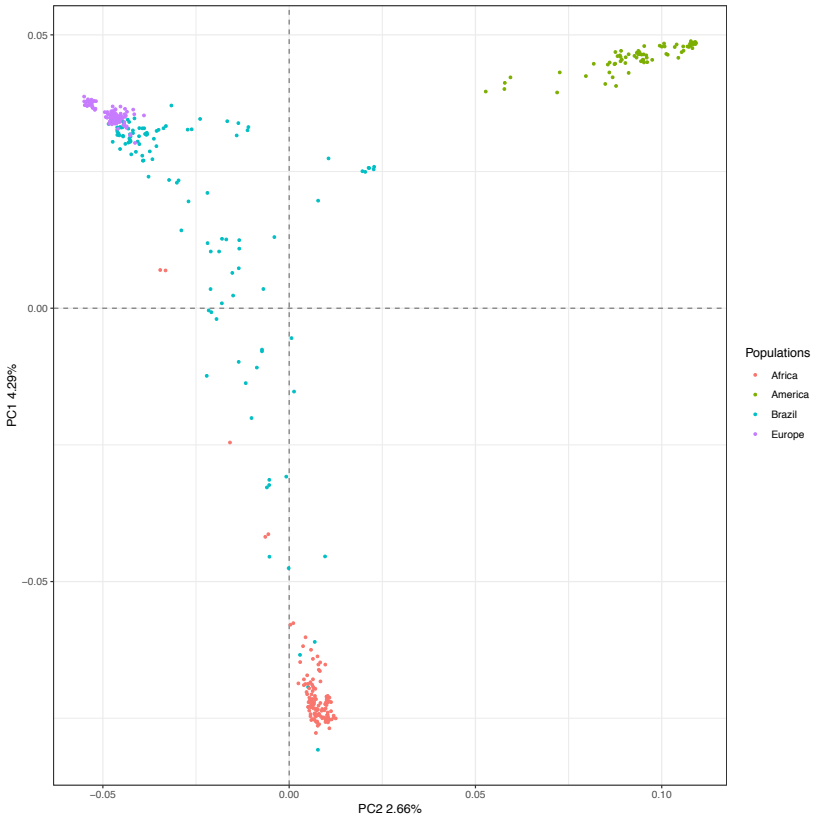
SantoDigital
Cloud Consulting Services

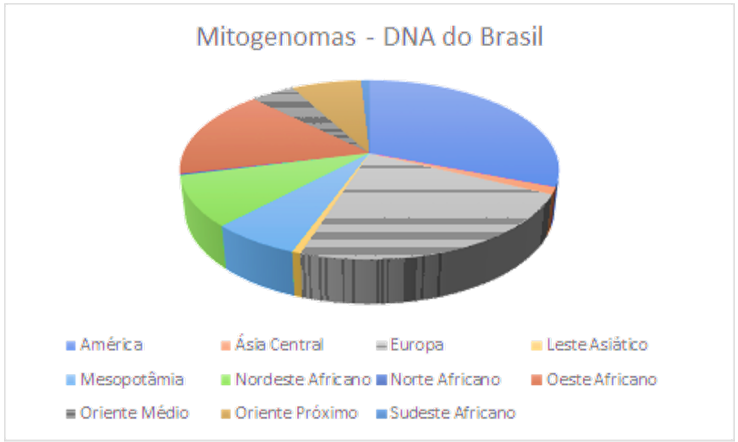
plbrasil
MAKES IT EASY

DNA DO BRASIL – the first 4 months



2.219 347 340 365


0.05
0.00
−0.05
PC1 4.29%
−0.05
0.00
0.05
0.10
PC2 2.66%
Populations
Africa
America
Brazil
Europe


Mitogenomas - DNA do Brasil
América
Ásia Central
Europa
Leste Asiático
Mesopotâmia
Nordeste Africano
Norte Africano
Oeste Africano
Oriente Médio
Oriente Próximo
Sudeste Africano

MINISTÉRIO DA SAÚDE

National Policy on Precision Medicine

GENOMICS

identification target gene

ADVANCED THERAPIES

POPULATIONAL

RARE DISEASES

GENE

CELL

BIOLOGICS

Brazilian Reference Genome

Diagnostic

Brazilian Reference Genome- Strategy

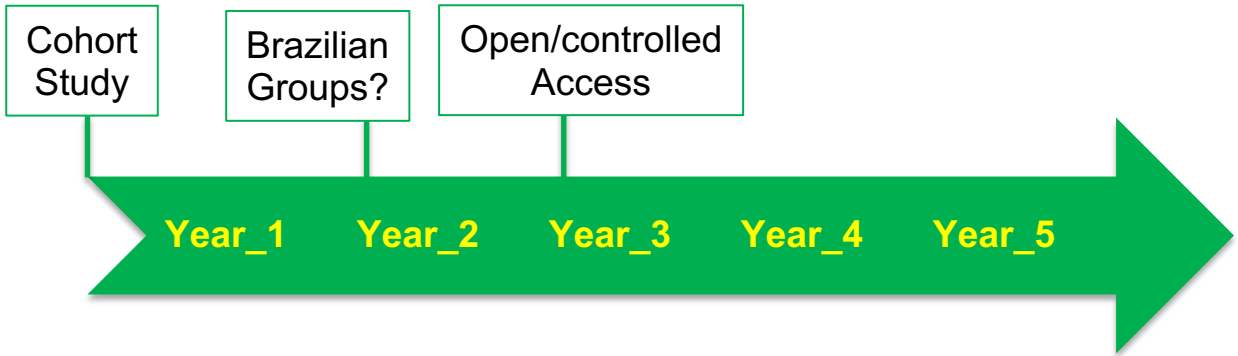
1. **Leverage investment made in cohort studies (40.000 genomes):**
    1. Cohort Identification;
    2. Harmonization of clinical data;
    3. Sequencing;
    4. Data access.

Estudo Longitudinal de Saúde do Adulto ELSA BRASIL

ELSI BRASIL

Corações de Baependi

EpiGen-Brasil



GENOMA REFERÊNCIA BRASIL - Strategy

1. Leverage investment made in cohort studies (40.000 genomes):
2. **Use Unified Public Health Service (SUS) to scale to 200k genomes:**
    1. Eletronic Health Records;
    2. Cost of sequencing;

Team

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