# ABraOM

## Arquivo Brasileiro Online de Mutações
## Online Archive of Brazilian Mutations

http://abraom.ib.usp.br


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Arquivo **Bra**sileiro **O**nline de **M**utações

Online Archive of Brazilian Mutations

Centro de Pesquisa sobre o Genoma Humano e Células-Tronco

CEPID Centros de Pesquisa, Inovação e Difusão

FAPESP

instituto de biociências

USP

inct institutos nacionais de ciência e tecnologia

CNPq

# HUG-CELL

**1,000+** Brazilian whole exome sequences

400+ rare Mendelian disorders

**609** elderly from São Paulo (SABE)

Allele frequencies publicly available online at **ABraOM**

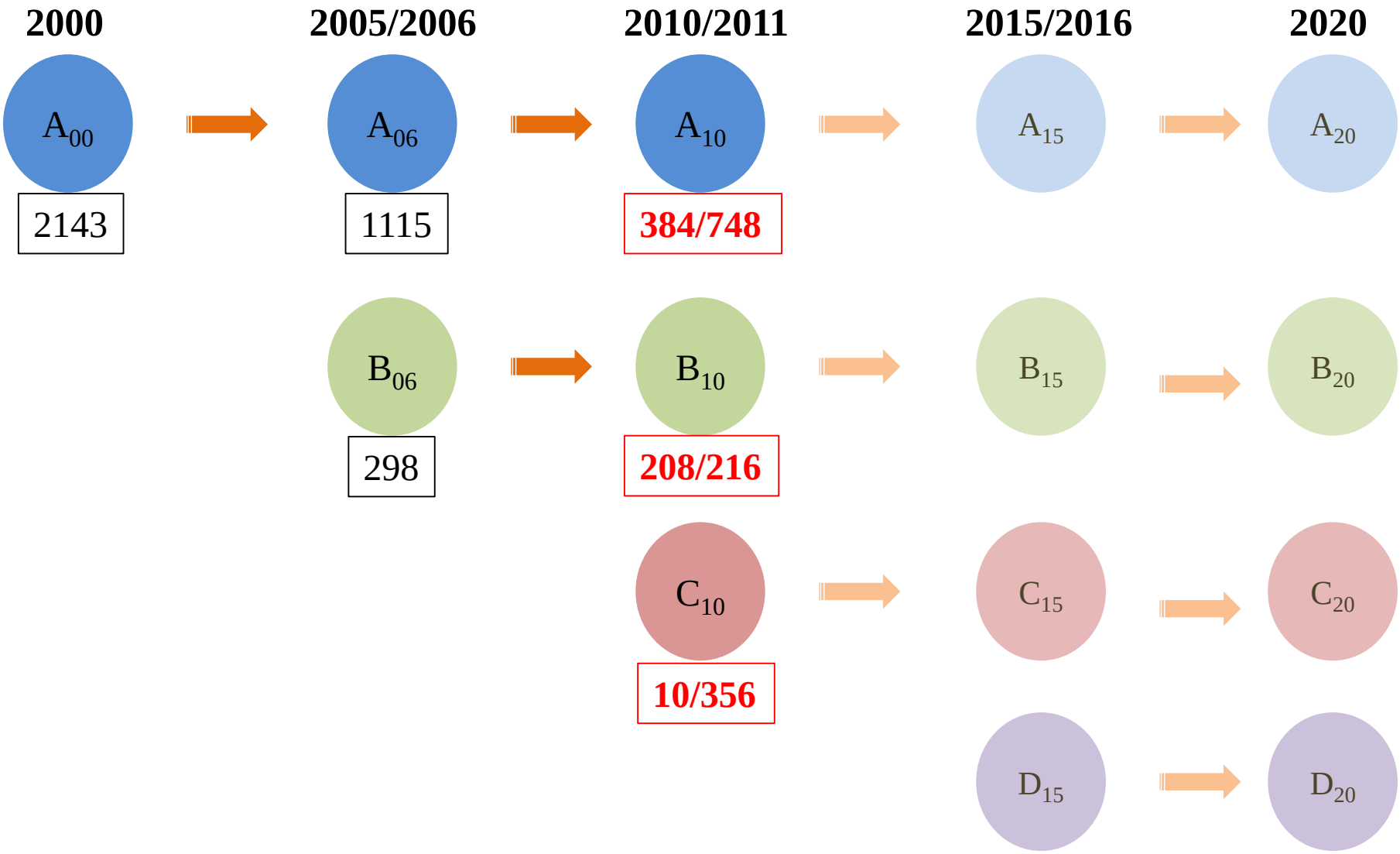
# Our aims

Database of reference controls with ancestry matching

Tool for interpretation of variant pathogenicity

Collaboration for studies in healthy aging

# SABE - Whole Exome Sequencing




Whole exome sequencing: **609/1320**

Dr. Mayana Zatz
USP

Survey
(150 pages)

Dr. Maria Lucia Lebrão & Dr. Yeda Duarte
USP

Genomics

Blood analyses

# SABE

Dr. Edson Amaro
USP/HIAE

Brain MRI

Biometry & Functional tests

# Results – Database application

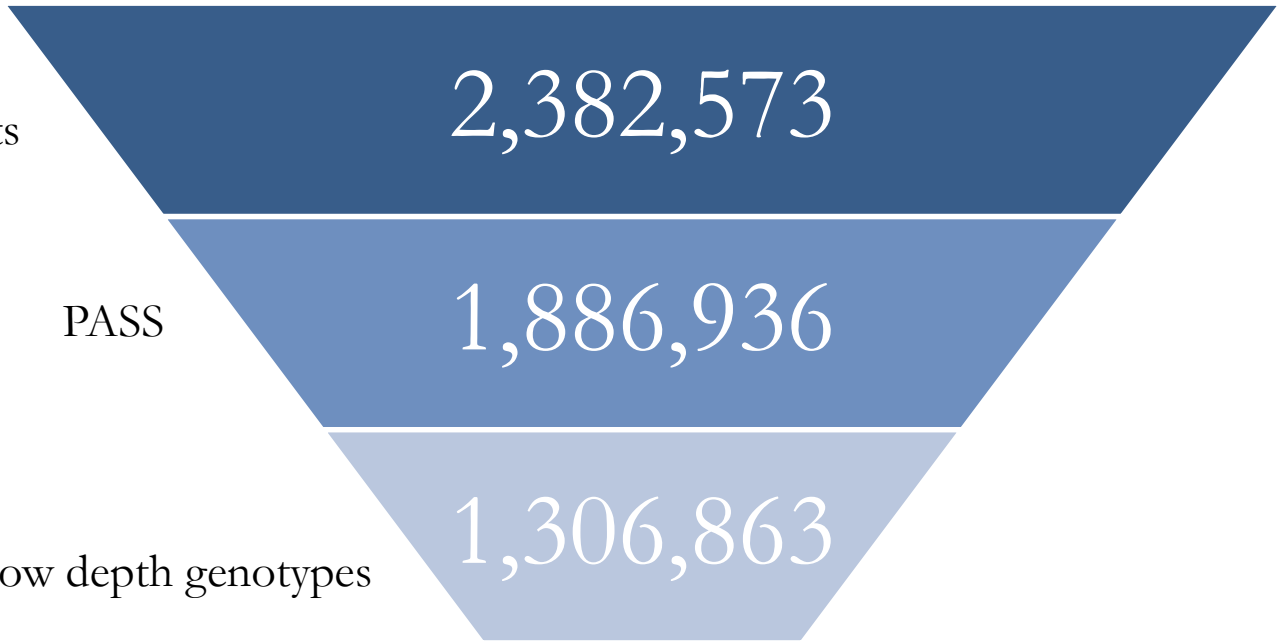
**Integration of allele frequencies to the clinical genomics diagnostic service and research projects of HUG-CELLL:**

Filtering over 1700 panels of disease-related genes and 400 whole exomes

**Collaboration in several studies:**

8 articles published between 2014 and 2016 cited over 80 times

# ABraOM variants




**PASS also excluding low confidence allele balance genotypes** 1,264,224

→ (3,6%)

**Absent from public databases**

# Results – Brazilian variants

| Frequency bins | Synonymous SNVs | Nonframeshift | | Frameshift | | Stop codon | | Splicing | Nonsynonymous SNVs | **Total (%)** |
|---|---|---|---|---|---|---|---|---|---|---|
| | | Deletion | Insertion | Deletion | Insertion | Gain | Loss | | | |
| 0.01% to 0.1% | 12331 | 384 | 129 | 812 | 368 | 787 | 27 | 495 | 25885 | 41218 (90.89) |
| 0.1% to 1% | 1249 | 86 | 65 | 65 | 80 | 53 | 1 | 58 | 2391 | 4048 (8.93) |
| 1% to 10% | 0 | 5 | 24 | 5 | 8 | 0 | 0 | 3 | 0 | 45 (0.1) |
| Above 10% | 0 | 2 | 21 | 4 | 4 | 1 | 0 | 4 | 0 | 36 (0.08) |
| **Total (%)** | 13580 (29.95) | 477 (1.05) | 239 (0.53) | 886 (1.95) | 460 (1.01) | 841 (1.85) | 28 (0.06) | 560 (1.23) | 28276 (62.35) | 45347 (100) |
| Singletons (%) | 12699 (93.51) | 410 (85.95) | 154 (64.44) | 827 (93.34) | 395 (85.87) | 801 (95.24) | 28 (100) | 546 (97.5) | 26543 (93.87) | 42403 (93.51) |

**Variants <1%: 2.744 with potential loss of function!**

**3-5 Loss of function (LoF) variants per individual on average**

# LoF in ACMG genes

**1.15%** (7/609) have pathogenic LoF variants in ACMG-56 genes, all in cancer-related genes.

ESP6500 found **1%** (African-ancestry) and **1.67%** (European-ancestry) [Amendola, 2015]

2/7 individuals have reported cancer diagnosis

The other 5 aged 69 to 87 years old are asymptomatic.

# ABraOM database


http://abraom.ib.usp.br

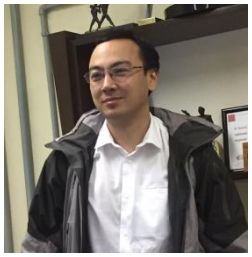
# Next steps

**Integrate genomic database (ABraOM) with clinical database (Zen)**

**Inclusion of other cohort datasets into ABraOM**

**Make public datasets available for download**

# Acknowledgments



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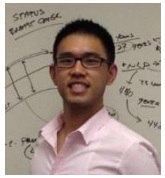

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