From 80plus (2008) to ABraOM (2016)

CEPID: CENTRO DE PESQUISA EM GENOMA E CÉLULAS-TRONCO: (CEGH-CEL)
RDIC- HUMAN GENOME AND STEM-CELL CENTER (HUG-CELL)

Oswaldo Frota-Pessoa 1917-2010

Human and Medical Genetics

It was started in the 60s in the Department of Genética e Biologia evolutiva

In 2000 the Human Genome Center was inaugurated and selected as a FAPESP/CEPID

In 2013 it was expanded to Human Genome and stem-cell center (HUG-CEL)

Oswaldo
Frota-Pessoa
1917-2010

Two-ways road

Patients

generating

New researches


Two-ways road
Patients
generating
helping
New researches

Molecular genetics

Introduced in the 80s

Research, diagnosis and carrier detection

Maria Rita Passos-Bueno

Next generation sequencing -based tests

introduced in 2012

- More than 1000 whole exomes and 2000 NGS panels

Webpage: 2015
Non-profit laboratory for genetic tests
Lowest possible costs and high quality

http://laboratorio.genoma.ib.usp.br

Genetic tests: Certification of Quality

EMQN

The European Molecular Genetics Quality Network

Certificate of Participation

2015 (Autumn)

This is to certify that

University of São Paulo

EMQN

The European Molecular Genetics Quality Network

Certificate of Participation

2016

This is to certify that

University of São Paulo

Genetic counseling

More than 100.000 families were attended so far:

Diagnosis

Carrier detection in “at risk” families

Genetic counseling including written reports

Orientation about management, available treatment, prognosis and research state of art

Multidisciplinary team: geneticists, neurologists, psychoanalists , psychologists, physiotherapists

Neuromuscular Disorders

Mariz Vainzof, Rita Pavanello, Fernando Kok, Karina Weinmann, Mayana Zatz

From genetic couseling to Psychoanalytical support neuromuscular affected patients and relatives















DEVELOPMENTAL DISORDERS:

Autism
Craniofacial
Deafness
Intellectual Disability
Skeletal Dysplasia

PIs: Ana Krepischi, Angela Vianna-Morgante, Célia Koiffmann, Debora Bertola, Fernando Kok, Regina C. Mingroni, Paulo A. Otto, M. Rita Passos-Bueno

The 80plus project: started in 2008

inct institutos nacionais de ciência e tecnologia

FAPESP

Aims

To serve as a databank of genomic variants based on our elderly population

Contribute to evaluate the pathogenicity of variants for diagnosis of different disorders

And explore some other ideas.....

The 80plus project








Some are centenarians

Cleonice Berardinelli

THE GOOD NEWS: NO SEVERE CALORIC RESTRICTION

The 80plus project was expanded

Michel Naslavsky

Guilherme Yamamoto

Collaboration project SABE → "Saúde, Bem Estar e Envelhecimento"

(Health, well-being and aging)

Follow-up cohort – Public Health School USP (FSP-USP)

Population study (representative of São Paulo census)

>=60 years old.

Maria Lucia Lebrão (in memorian)

Yeda Duarte

Brain Imaging: 580 individuals

Partnership with Instituto de Pesquisas Hospital Albert Einstein

- 3Teslas MR system (32 channel head coil)
- Neuronavigator TMS
- EEG (fMRI compatible - 64 channel)
- Research instruments developed in house

Edson Amaro Jr.

What was achieved until now?
From 80plus ☞ AbraOM

Online Archive of Brazilian mutations

Michel Naslavsky

Guilherme Yamamoto

609

Whole exome sequencing (WES)

Naslavsky et al.,
Human mutation, 2017

Whole genome sequencing

1230 individuals

Partnership with Human Longevity Institute (HLI)

Opening several new collaborations

2017-2021

Envelhecimento e doenças genéticas: genômica e metagenômica

Aging and genetic diseases: Genomics and metagenomics

MUITO OBRIGADA

To all CEPID team
Volunteers and patients

Guilherme

FAPESP

CEPID
CENTROS DE PESQUISA, INOVAÇÃO E DIFUSÃO

Instituto de Biociências

CNPq
Conselho Nacional de Desenvolvimento Científico e Tecnológico

inct
institutos nacionais de ciência e tecnologia

Centro de Pesquisa sobre o Genoma Humano e Células-Tronco