

The new Human Genome and Stem-Cell Research Center (HUG-CELL) at the University of São Paulo raises the scope of the original Human Genome Research Center (HGRC – RIDC I) to a new level. The Center was initiated in 2000 with the aim of improving basic knowledge and diagnosis of prevalent genetic diseases in the Brazilian population. The original center concentrated initially on Mendelian disorders, mainly neuromuscular, craniofacial, and mental disabilities. These activities were expanded in 2005 by introducing stem-cell research to understand gene expression and differentiation in complex genetic disorders such as autism and amyotrophic lateral sclerosis and to evaluate stem-cell-based disease therapy. However, the unanticipated complexity of the transcriptional mechanisms that emerged from the Human Genome, and the modest advances in improving genetic health care have demanded the opening of further fields of investigation.

Accordingly, this revised project has been expanded to include research on the genetics and genomic instability associated with aging and degenerative diseases, epigenetic mechanisms involved in disease manifestation and phenotypic variability between individuals with identical Mendelian disease mutations. In addition, the “over 80 project” will compare genome variation and brain functioning (MRI) of healthy Brazilian individuals older than 80 with a group older than 60 without prior selection based on good health in old age. To address these questions, the Center will use state of the art approaches, particularly next generation sequencing and sophisticated cell sorting; incorporate a much broader base of scientific expertise; and optimize inter-group synergy, including national and international research collaborations. The plans also contribute to translational medicine, particularly to stem-cell therapies in preclinical studies using different animal models and therapeutic trials for particular genetic disorders.

The transfer technology project is based on the great number of patients with different genetic disorders that have been ascertained and



*Stem-cells culture for investigating genetic disorders and cell therapy*

registered at the Center, the largest in Latin America. The ethnic variability of the Brazilian population provides a rich foundation for the proposed studies. Molecular diagnosis and genetic counseling has been offered to the population by our genetic services division. Accordingly the plan is to develop new kits for the diagnosis of rare diseases using genomic technologies and the information on Brazilian genetic variation and to establish partnerships with start-up biotechnology enterprises. It is anticipated that the knowledge gained from these activities will have an important impact on genetic health care in Brazil.

The diffusion/education project aims to make science, and particularly genetics, more accessible to society. This includes high-school education and activities to the general public that have already reached thousands of students since 2000. In the new project, specialized courses in genetics for professionals from health and media areas will be offered. Programs to assist teachers and students inside public high schools aimed at motivating students to study science and improving their genetics knowledge will be mediated through various activities including: scientific exhibitions, implementation of short term travelling laboratory classes and maintenance of centralized repositories of instructional materials that can be loaned to schools. Additionally, the HUG-CELL center is part of a large new national project, “Adventures in Science”, involving the creation of science kits for young students aimed at motivating and teaching fundamental scientific concepts in a clear and precise way.

## Host Institution

University of São Paulo (USP, campus São Paulo)

## Associated Institutions

Federal University of São Paulo (UNIFESP)  
Albert Einstein Hospital  
Fleury S. A.  
Zerbini Foundation  
Utrecht University  
Heart Institute, University of São Paulo (INCOR-USP)

## Principal Investigator

Mayana Zatz, USP

## Education and Knowledge Diffusion Coordinator

Eliana Maria Beluzzo Dessen, USP

## Technology Transfer Coordinator

Maria Rita dos Santos e Passos Bueno, USP

## Co-Principal Investigators

Angela Maria Vianna Morgante, USP  
Carla Rosenberg, USP  
Celia Priszkulnik Koiffmann, USP  
Eliana Maria Beluzzo Dessen, USP  
Esper Abrão Cavalheiro, UNIFESP  
Maria Rita dos Santos e Passos Bueno, USP  
Mariz Vainzof, USP  
Oswaldo Keith Okamoto, USP  
Peter Lees Pearson, Utrecht University  
Regina Celia Mingroni Netto, USP

## Associated Researchers

Carlos Frederico Martins Menck, USP  
David Schlesinger, Albert Einstein Hospital  
Débora Romeo Bertola, USP  
Edson Amaro Junior, USP  
Fernando Kok, Fleury S. A.  
João Paulo F. Whitaker Kitajima  
Jorge Elias Kalil Filho, Zerbini Foundation  
Luciana Amaral Haddad, USP  
Luís Eduardo Soares Netto, USP  
Maria Lúcia Lebrão, USP  
Merari de Fátima Ramires Ferrari, USP  
Nivaldo Alonso, USP  
Rita de Cassia Pavanello  
Tatiana Teixeira Torres, USP  
Venâncio Avancini Ferreira Alves, USP  
Verônica Porto Carreiro de Vasconcellos Coelho, INCOR-USP  
Yeda Aparecida de Oliveira Duarte, USP