CURRICULAR SUMMARY

NAME: ISCIA LOPES-CENDES

ORCID: https://orcid.org/0000-0002-6221-6822

Lattes CV: https://lattes.cnpq.br/3944020856408245

Web of Science: https://www.webofscience.com/wos/author/record/B-9416-2013

MyCitations (Google Scholar): http://scholar.google.com.br/citations?user=H4g6TDcA

1. EDUCATION AND TRAINING

Year	Duration (months)	Degree or activity	Institution	Title of thesis/dissertation
1994-1999	56	Ph.D. in Neuroscience	McGill University, CANADA	Molecular studies in spinocerebellar ataxias
1991-1993	36	M.Sc. in Neuroscience	McGill University, CANADA	Genetic heterogeneity in spinocerebellar ataxias
1988-1990	24	Medical Residency in Pediatrics (Medical Specialty)	UNICAMP, BRAZIL	NOT APPLICABLE
1982-1987	72	Medical Degree (M.D.)	UNICAMP, BRAZIL	NOT APPLICABLE

1.1. Education and Training - Additional Information: NOTHING TO DECLARE.

2. ACADEMIC/PROFESSIONAL HISTORY

2.1. List of up to three current positions:

1. 04/2009-present - Full Professor of Medical Genetics and Genomic Medicine, University of Campinas (UNICAMP).

2. 09/2023-present - Chair (2023-2024) and member of the World Health Organization (WHO) Technical Advisory Group on Genomics (TAG-G).

3. 11/2025-present - Member of *The Lancet* Commission on the Global Implementation of Precision Medicine.

3. CONTRIBUTIONS TO SCIENCE (LIST UP TO FIVE):

1. Galvão IC, Lemoine M, Kandratavicius L, Yasuda CL, Alvim MKM, Ghizoni E, Blümcke I, Cendes F, Rogerio F, Lopes-Cendes I, Veiga DFT. *Cell type mapping of mild malformations of cortical development with oligodendroglial hyperplasia in epilepsy using single-nucleus multiomics.* Epilepsia. 2025;66(8):3064-3080. doi: 10.1111/epi.18413. **Impact statement:** This study used single-nucleus multiomics to characterize the cell types and states involved in mild malformations of cortical development with oligodendroglial hyperplasia, a poorly understood cause of epilepsy. As senior author, I contributed to the entire study conception, provision of biological material, and discussion of the manuscript. I was co-supervisor of the first author during her Ph.D. This work is directly aligned with the research line investigating mechanisms underlying human epileptogenesis.

2. Galvão IC, Lemoine M, Messias LA, Araújo PAOR, Geraldis JC, Yasuda CL, Alvim MKM, Ghizoni E, Tedeschi E, Cendes F, Rogerio F, Lopes-Cendes I, Veiga DFT. *Multimodal single-cell profiling reveals neuronal vulnerability and pathological cell states in focal cortical dysplasia.* iScience. 2024 Nov 6;27(12):111337. doi: 10.1016/j.isci.2024.111337. **Impact statement:** The first single-cell multiomic study of focal cortical dysplasia revealed cell type-specific regulatory alterations and pathways associated with mTOR signaling and neuronal hyperexcitability. As senior author, I contributed from study conception through manuscript preparation, consolidating the use of single-cell multiomic approaches in our group. I was co-supervisor of the first author during her Ph.D. This work is directly aligned with the research line investigating mechanisms underlying human epileptogenesis.

3. International League Against Epilepsy Consortium on Complex Epilepsies. *GWAS meta-analysis of over 29,000 people with epilepsy identifies 26 risk loci and subtype-specific genetic architecture.* Nat Genet. 2023 Sep;55(9):1471-1482. doi: 10.1038/s41588-023-01485-w. **Impact statement:** The largest genome-wide association study of epilepsy conducted at the time, it identified 26 risk loci and differences across subtypes. I led the Brazilian participation, which included the only large admixed Latin American cohort and the largest single-center series of mesial temporal lobe epilepsy. I actively participated in the analyses and manuscript preparation. This work is part of current projects on genetic diversity and susceptibility to epilepsy.

4. Avansini SH, Puppo F, Adams JW, Vieira AS, Coan AC, Rogerio F, Torres FR, Araújo PAOR, Martin M, Montenegro MA, Yasuda CL, Tedeschi H, Ghizoni E, França AFEC, Alvim MKM, et al. *Junctional instability in neuroepithelium and network hyperexcitability in a focal cortical dysplasia human model.* Brain. 2022 Jun 30;145(6):1962-1977. doi: 10.1093/brain/awab479. **Impact statement:** The first human model of focal cortical dysplasia based on patient-derived organoids revealed neuroepithelial instability, altered neuronal maturation, and hyperexcitability. As senior author, I helped integrate clinical and genetic investigation with cellular modeling. The platform is important for expanding research into disease mechanisms and the evaluation of more specific therapies. I supervised the first author during her postdoctoral fellowship. The platform supports current projects on disease mechanisms and therapeutic evaluation in patient-derived models.

5. Rocha CS, Secolin R, Rodrigues MR, Carvalho BS, Iscia Lopes-Cendes. *The Brazilian Initiative on Precision Medicine (BIPMed): fostering genomic data-sharing of underrepresented populations.* NPJ Genomic Medicine. 2020;5(1):42. doi:

10.1038/s41525-020-00149-6. **Impact statement:** BIPMed established the first public genomic reference database for a highly admixed Latin American population, improving variant interpretation. As its originator and one of those responsible for its implementation, I consolidated a line of work in genomic diversity and precision medicine. I supervised the first author during her Ph.D. The initiative supports current projects in data sharing and the clinical implementation of genomics in global health.

4. RESEARCH FUNDING (UP TO FIVE)

1. National Council for Scientific and Technological Development (CNPq), Ministry of Science and Technology, Brazil. Research Productivity Fellowship - Level A. 08/01/2026 to 07/31/2031.

2. Chan Zuckerberg Initiative on Single Cell Genomics. Single-cell analysis of the pediatric brain. Principal Investigator. 11/30/2022 to 11/30/2026.

3. National Council for Scientific and Technological Development (CNPq), Ministry of Science and Technology, Brazil. Call: National Institutes of Science and Technology (INCT), grant no. 408760/2024-9. National Institute of Science and Technology in Multiomics Applied to Precision Health (IMASP). Principal Investigator. 11/30/2025 to 10/01/2030.

4. National Council for Scientific and Technological Development (CNPq), Ministry of Science and Technology, Brazil. Universal Call, grant no. 402893/2025-5. Unraveling the molecular mechanisms of mesial temporal lobe epilepsy: integration of single-cell omics technologies to investigate gene regulation in hippocampal sclerosis. Coordinator. 11/18/2025 to 11/30/2028.

5. São Paulo Research Foundation (FAPESP). Research, Innovation and Dissemination Center - Brazilian Institute of Neuroscience and Neurotechnology (BRAINN). Principal Investigator (grant no. 2013/07559-3). 07/01/2013 to 06/30/2026. It is important to note that although FAPESP funding has ended, the institute remains active and has been incorporated into UNICAMP's official organizational structure as an Interdisciplinary Center of Excellence.

5. QUANTITATIVE INDICATORS

1. Books: 1

2. Publications in journals with selective editorial policies: 299

3. Book chapters: 46

4. Master's dissertations
   4.a) supervised and completed: 31 (+ co-supervisions: 3) = 34
   4.b) in progress: 3

5. Doctoral theses
   5.a) supervised and completed: 35 (+ co-supervisions: 5) = 40
   5.b) in progress: 2

6. Postdoctoral supervisions:
   6.a) completed: 36
   6.b) in progress: 5

7. Citations received in the international scientific literature - Google Scholar: 19,510

8. Patents filed, granted, and licensed: NOTHING TO DECLARE

9. Products developed and launched on the market: NOTHING TO DECLARE

10. Optimized processes implemented in companies or social organizations: NOTHING TO DECLARE

11. Companies created or supported: NOTHING TO DECLARE

12. Relevant technical and scientific consultancies: NOTHING TO DECLARE

6. OTHER RELEVANT BIOGRAPHICAL INFORMATION

I work in neurogenetics, focusing on the genetic and phenotypic determinants of neurological disorders, particularly the epilepsies. My research uses multiomic approaches, next-generation sequencing, bioinformatics, and single-cell studies to investigate molecular mechanisms and identify potential therapeutic targets. This trajectory combines thematic continuity in epilepsy research with methodological renewal, progressively incorporating genomics, single-cell multiomics, spatial analysis, and human cellular models. I contributed to the creation of the Brazilian Initiative on Precision Medicine (www.bipmed.org), the first public genomic reference database for a highly admixed Latin American population, and to the development of LatinGen (www.latingen.org), a platform for sharing Latin American genomic data. In 1997, I established Brazil's first outpatient clinic for presymptomatic testing of late-onset neurodegenerative diseases. I am currently a principal investigator at the Brazilian Institute of Neuroscience and Neurotechnology (BRAINN). My academic activities include experience in management, scientific evaluation, and editorial work. I served as Head of the Department of Medical Genetics and Coordinator of UNICAMP's Graduate Program in Medical Pathophysiology, as well as on committees of the International League Against Epilepsy. I served on CNPq's Advisory Committee for Medicine and currently sit on CAPES's evaluation committee for the Medicine I area. I am Section Editor of the *Brazilian Journal of Medical and Biological Research* and *Arquivos de Neuro-Psiquiatria*, Associate Editor of *Human Genomics*, and a member of the Editorial Board of *Epilepsia Open*. I have supervised 41 undergraduate research students and participate in patient care activities at HC-UNICAMP. I also remain active nationally and internationally in governance and policies for the implementation of genomic medicine.

6.a. Registered processes and software:

I participated in the development of computational tools for RNA interference, small RNA design, genetic data analysis, and sequencing data quality control. These products reflect the continued integration of technological development and bioinformatics into the group's research lines.

• DEBORA: platform for identifying targets for RNA interference.

• STRAND ANALYSIS: software for designing small interfering RNAs, registered with the Brazilian National Institute of Industrial Property (INPI).

• JINGLEFIX: software for analyzing microsatellites and single-nucleotide variants, registered with INPI.

• THERMODYNAMIC DOMAIN ASSEMBLER: software for thermodynamic analysis of DNA and RNA sequences.

• Rqc: Bioconductor package for assessing the quality of next-generation sequencing data. Available at http://www.bioconductor.org/packages/Rqc.

• GA4GHclient: R package for accessing data servers compatible with Global Alliance for Genomics and Health standards. Available at https://github.com/labbcb/GA4GHclient.

6.b. International experience:

After completing my Ph.D. in Neuroscience at McGill University in 1999, I maintained continuous involvement in international research networks in genomics, neuroscience, and epilepsy. I participate in genetics initiatives of the International League Against Epilepsy (ILAE) and in the ENIGMA consortium, contributing to published multicenter studies on the genetic architecture and structural alterations of the epilepsies, including data from underrepresented Latin American populations. Notable among these results is the largest genome-wide association study of epilepsy conducted at the time, which identified 26 risk loci and characterized genetic differences across disease subtypes.

I also participated in the EpimiRNA consortium (2015-2019), funded by the European Union's Seventh Framework Programme (FP7), which generated publications on the role of microRNAs in epilepsy and the development of RNA-based therapeutic approaches. My research also received funding from the Chan Zuckerberg Initiative for an international project on single-cell analysis of the pediatric brain.

My international work encompasses data-sharing and genomics implementation initiatives such as the Global Alliance for Genomics and Health (GA4GH), the International Common Disease Alliance, LatinGen, and LatinGenomes. I currently serve on the Executive Board of the Human Genome Organization (HUGO) and on *The Lancet* Commission on the Global Implementation of Precision Medicine. From 2023 to 2024, I chaired the World Health Organization Technical Advisory Group on Genomics (TAG-G), and I continue to serve on the same group as a member. This work has contributed to strategies for the responsible and equitable implementation of genomics in global health.

6.c. Awards, distinctions, and honors:

• 2006 - “Zeferino Vaz” Award for Academic Recognition, UNICAMP.

• 2015 - Elected member of the Brazilian Academy of Sciences.

• 2016 - Scientist and Entrepreneur of the Year Award, Instituto Nanocell.

• 2017 - Elected member of the São Paulo State Academy of Sciences.

• 2020 - “Zeferino Vaz” Award for Academic Recognition, UNICAMP.

• 2024 - Elected member of The World Academy of Sciences (TWAS).