BIOGRAPHICAL SKETCH

Provide the following information for the Senior/key personnel and other significant contributors in the order listed on Form Page 2. Follow this format for each person. **DO NOT EXCEED FOUR PAGES.**

NAME	POSITION TITLE
Mendonca, Berenice Bilharinho de	Full Professor of Endocrinology
eRA COMMONS USER NAME (credential, e.g., agency login)	

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	MM/YY	FIELD OF STUDY
Faculdade de Medicina do Triangulo Mineiro	MD	1973	Medicine
University of Sao Paulo School of Medicine, São Paulo, Brazil	Residency	1976	Endocrinology
University of Sao Paulo School of Medicine, Sao Paulo, Brazil	MSc		
University of Sao Paulo School of Medicine, Sao Paulo, Brazil	PhD	1984	Endocrinology

A. Personal Statement

I am a physician-scientist focused on basic and translational investigation of developmental endocrinology. I have established the disorders of sex development as my clinical and research area of expertise. I have held a leadership role as Chief, Division of Endocrinology, at University of Sao Paulo since 2005.

B. Positions and Honors

Appointments

1971-1972 Research Fellow, Cardiology Department, School of Medicine, Triangulo Mineiro, MG, Brazil
1974-1975 Resident, First Department of Internal Medicine, University of Sao Paulo, SP, Brazil
1975-1977 Resident, Division of Endocrinology, University of Sao Paulo, SP, Brazil
1977-1978 Chief Resident, Division of Endocrinology, University of Sao Paulo, SP, Brazil
1981-present Clinical Associate Professor of Endocrinology, University of Sao Paulo, SP, Brazil
1995-present Chief of the Hormonal and Molecular Endocrinology Laboratory of the Hospital das Clinicas, University of Sao Paulo School of Medicine
2005-present Professor of Endocrinology, University of Sao Paulo, Brazil
2005-present Director of Endocrinology and Metabolism Service, Clinical Hospital, Medical School at Sao Paulo University.
2007-present Member of the Director Consultive Committee of the Clinical Hospital, Medical School at Sao Paulo University
2008-present Diretor Técnico da Divisão de Clinica Médica I e II.
2008-2011 Head of the Internal Medicine Department of Medical School, Sao Paulo University

Other Experiences and Selected Professional Memberships

Member: Brazilian Endocrine Society, 1976-present; Brazilian Medical Association, 1977-present; Sao Paulo Medical Association, 1980-present; Latin American Pediatric Endocrinology Society, 1987-present; Endocrine Society, 1992-present. Membership In Committees: Medical Residence Committee, Division of Endocrinology, SP, Brazil; Annual Steering Committee of Endocrine Society 2006-2009; Annual Scientific & Educational Programs Core Committee (SCIED) 2011-2014. Editorial Board: Brazilian Archives of Endocrinology and Metabolism; Editorial Board, Clinics 2009-present; Clinical Endocrinology 2004-2007; JOE and JME 2012-2014; JCEM 2014; Journal of the Endocrine Society (JES) 2016-2018 Postgraduate Student Formation: Doctor's Degree,1988-2016; (24 PhD theses as adviser; 10 PhD theses as co-adviser); 5 *Master's Degree* dissertations (1996-2012).

Awards and Honors (selected listing)

Brazilian Society of Endocrinology Award for the recognition of the scientific career, 2009; Scopus Award given by Elsevier Publisher with CAPES (Coordenação de Aperfeiçoamento de Pessoal de Nível Superior, the section of the Ministry of Education for University Education) to recognize Brazilian scientists with elevated scientific production of great relevance and excellence based on the Scopus database, 2009; Clinical Endocrinology Trust Visiting Professor in the UK, 2012; SLEP Maestra de La Endocrinologia Pediatrica. Award for the recognition of the scientific career, 2012; Endocrine Society International Excellence In Endocrinology Award, 2013.

C. Selected peer-reviewed publications

1. Teles MG, Bianco SD, Brito VN, Trarbach EB, Kuohung W, Xu S, Seminara SB, Mendonca BB, Kaiser UB, Latronico AC. A GPR54-activating mutation in a patient with central precocious puberty. **N Engl J Med** 358: 709-715, 2008. [PMID: 18272894] [PMCID: PMC2859966]
2. Mendonca BB, Domenice S, Arnhold IJ, Costa EM. 46,XY disorders of sex development (DSD). **Clin Endocrinol (Oxf)** 70: 173-187, 2009. [PMID: 18811725]
3. Mendonca BB, Costa EM, Belgorosky A, Rivarola MA, Domenice S. 46,XY DSD due to impaired androgen production. **Best Pract Res Clin Endocrinol Metab** 24: 243-262, 2010. [PubMed PMID: 20541150].
4. Sircili MH, e Silva FA, Costa EM, Brito VN, Arnhold IJ, Dénes FT, Inacio M, de Mendonca BB. Long-term surgical outcome of masculinizing genitoplasty in large cohort of patients with disorders of sex development. **J Urol** 184: 1122-1127,2010. [PMID: 20650476]
5. Nishi MY, Costa EM, Oliveira SB, Mendonca BB, Domenice S. The role of SRY mutations in the etiology of gonadal dysgenesis in patients with 45,X/46,XY disorder of sex development and variants. **Horm Res Paediatr** 75: 26-31, 2011. [PMID: 20699606]
6. da Silva TE, Nishi MY, Costa EM, Martin RM, Carvalho FM, Mendonca BB, Domenice S. A novel WT1 heterozygous nonsense mutation (p.K248X) causing a mild and slightly progressive nephropathy in a 46,XY patient with Denys-Drash syndrome. **Pediatr Nephrol** 26: 1311-1315, 2011. [PMID: 21559934]
7. Wilson JD, Rivarola MA, Mendonca BB, Warne GL, Josso N, Drop SL, Grumbach MM. Advice on the management of ambiguous genitalia to a young endocrinologist from experienced clinicians. **Semin Reprod Med** 30: 339-350, 2012. [PMID: 23044870]
8. Costa EM, Domenice S, Sircili MH, Inacio M, Mendonca BB. DSD due to 5α-reductase 2 deficiency - from diagnosis to long term outcome. **Semin Reprod Med** 30: 427-431, 2012. [PMID: 23044880]
9. Montenegro LR, Silveira LF, Tusset C, de Castro M, Versiani BR, Latronico AC, Mendonca BB, Trarbach EB. Combined use of multiplex ligation-dependent probe amplification and automatic sequencing for identification of KAL1 defects in patients with Kallmann syndrome. **Fertil Steril** 100: 854-859, 2013. [PMID: 23721716]
10. Gomes LG, Madureira G, Mendonca BB, Bachega TA. Mineralocorticoid replacement during infancy for salt wasting congenital adrenal hyperplasia due to 21-hydroxylase deficiency. **Clinics (Sao Paulo)** 68: 147-152, 2013. [PMID: 23525308] [PMCID: PMC3584273]
11. Moura-Massari VO, Bugano DD, Marcondes JA, Gomes LG, Mendonca BB, Bachega TA. CYP21A2 genotypes do not predict the severity of hyperandrogenic manifestations in the nonclassical form of congenital adrenal hyperplasia. **Horm Metab Res** 45: 301-307, 2013. [PMID: 23322511]
12. Abreu AP, Dauber A, Macedo DB, Noel SD, Brito VN, Gill JC, Cukier P, Thompson IR, Navarro VM, Gagliardi PC, Rodrigues T, Kochi C, Longui CA, Beckers D, de Zegher F, Montenegro LR, Mendonca BB, Carroll RS, Hirschhorn JN, Latronico AC, Kaiser UB. Central precocious puberty caused by mutations in the imprinted gene MKRN3. **N Engl J Med** 368: 2467-2475, 2013. [PMID: 23738509] [PMCID: PMC3808195]
13. Carvalho LC, Brito VN, Martin RM, Zamboni AM, Gomes LG, Inácio M, Mermejo LM, Coeli-Lacchini F, Teixeira VR, Gonçalves FT, Carrilho AJ, Del Toro Camargo KY, Finkielstain GP, Taboada GF, Frade Costa EM, Domenice S, Mendonca BB.Clinical, hormonal, ovarian, and genetic aspects of 46,XX patients with congenital adrenal hyperplasia due to CYP17A1 defects. **Fertil Steril. 2016** Jun;105(6):1612-9.

14. Achermann JC, Domenice S, Bachega TA, Nishi MY, Mendonca BB.
Disorders of sex development: effect of molecular diagnostics. **Nat Rev Endocrinol.** 2015 Aug;11(8):478-88. doi: 10.1038/nrendo.2015.69.. Review.

15. Bianchi PH, Gouveia GR, Costa EM, Domenice S, Martin RM, de Carvalho LC, Pelaes T, Inacio M, Codarin RR, Sator de Faria MB, Francisco RP, Baracat EC, Serafini PC, Mendonca BB.
Successful Live Birth in a Woman With 17α-Hydroxylase Deficiency Through IVF Frozen-Thawed Embryo Transfer. **J Clin Endocrinol Metab. 2016** Feb;101(2):345-8. doi: 10.1210/jc.2015-3201.

D. Research Support

Current Support

Sponsor: FAPESP, (2013/02162-8) 2014-2019
Title: Molecular pathogenesis and characterization of monogenic developmental diseases: a route to translational medicine

Completed Research Support:

Sponsor: Personal Grant (CNPq, 305743/2011-2)
Title: TRH-Stimulated TSH and Prolactin Levels in Patients with PROP1 Mutations: Model for Exclusive Congenital Pituitary Deficiency.

Sponsor: National consortium FAPESP (1997/1196-1) 1997-2001
Title: Molecular diagnosis of sexual differentiation disorders.

Sponsor: National consortium FAPESP (1999/06468-5) 2000-2003
Title: Analysis of mutations in CYP21B in patients with congenital adrenal hyperplasia due to 21-hydroxilase deficiency.

Sponsor: National consortium FAPESP (2000/06677-2-5) 2000-2003
Title: Analysis of LHX3 and GHRHR genes in children with growth hormone deficiency.

Sponsor: National consortium FAPESP (2000/14339-0) 2000-2003
Title: DAX1 and SF1 molecular analysis in patients with abnormal gonadal determination.

Sponsor: National consortium FAPESP (2000/14092-4) 2000-2005
Title: Molecular diagnosis of abnormalities in GH–GHR–IGF-I axis.

Sponsor: National consortium FAPESP (2003/07966-6) 2003-2005
Title: Identificação do novo *locus* autossômico responsável pela resistência familial ovariana e testicular ao hormônio luteinizante.

Sponsor: National consortium FAPESP (2004/05379-9) 2004-2006
Title: Gonadotropin isoforms in hypogonadotropic hypogonadism.

Sponsor: National consortium FAPESP (2004/15046-7) 2005-2007
Title: Expression of illicit receptors in adrenal tissue of patients with adrenocortical diseases.

Sponsor: National consortium FAPESP (2005/04726-0) 2006-2011
PI: Berenice BB
Title: Molecular defects characterization in congenital developmental disorders.

Sponsor: Edital MCT/CNPq (14/2009 2009-483416/2009-6) 2009-2011
Co-I: Berenice BB
Title: Universal Faixa B - Absence of inactivating mutations in *FGF9* and *FGFR2* genes in 46,XY patients with disorders of sex development (DSD) due to gonadal dysgenesis.

Sponsor: National consortium FAPESP, 2011-2013
Title: Determinação dos Hormônios Esteróides por Cromatografia Líquida Associada à Espectrometria de Massas em Tadem (LC-MSMS): impacto na pesquisa em endocrinologia.

Sponsor: International consortium (1454RR19484-01) 2004-2009
Co-I: Berenice BB (PI: Maria New)
Title: The Natural History of Rare Genetic Steroid Disorders
An international consortium to study several rare genetic steroid disorders from conception to old age with data stored at a central data technology center; one of eight consortia funded through NIH Rare Disease Network.