A. Biographical Information

Name	Gustavo Henrique Boff Maegawa
Address/contacts	University of Florida (UF), Department of Pediatrics/Genetics and Metabolism UF Department of Peds Chair's Office, PO Box 100296 1600 SW Archer Road Room R1-118D Gainesville, FL 32610-0296 (352) 294 5559 – office (352) 627 4565 – fax gmaegawa@ufl.edu
Webpage(s)	Research Lab http://maegawa.research.pediatrics.med.ufl.edu LSD Clinical Program http://maegawa.research.pediatrics.med.ufl.edu/lsd-program/

Degrees

M.D.	Universidade Federal do Parana, Medical School, Curitiba, Brazil	2000
Ph.D.	University of Toronto, Faculty of Medicine, Toronto, Ontario, Canada	2008

Current appointment

Position	Associate Professor University of Florida (UF), College of Medicine, Department of Pediatrics, Div. of Genetics and Metabolism (primary). Departments of Neuroscience, Immunology and Microbiology (affiliated)
Period	July 2015 – present

Past appointments

Position	Assistant Professor The Johns Hopkins University School of Medicine McKusick-Nathans Institute of Genetic Medicine & Department of Pediatrics, Director, Lysosomal Storage Disease (LSD) Program, Johns Hopkins Hospital
Period	2009 – June 2015

Other Positions and Public Service

Position	Special Government Employee (SGE) for FDA *Food and Drug Administration (FDA)*
Period	2013 – present
Address	Food and Drug Administration (FDA) Rockville, MD 20857
Position	Member of Grant Application Review Panel *Office of Orphan Products Development* *Food and Drug Administration (FDA)*
Period	2013 – present
Address	Food and Drug Administration (FDA) Rockville, MD 20857
Position	NIH Study Section Reviewer *NSB-B Study Section for IGNITE and NINDS CREATE Applications*
Period	2018-2026 (permanent member)
Address	Center for Scientific Review (CSR), NIH Bethesda, MD 20892

Position	Member of Scientific Advisory Council *National Tay-Sachs Disease Association and Allied Disorders (NTSAD)*
Period	2014 – present
Address	NTSAD, Boston, MA 02135
Position	NIH Study Section Reviewer *Drug Discovery for the Nervous System Standing Study Section*
Period	2014
Address	Center for Scientific Review (CSR), NIH Bethesda, MD 20892
Position	Scientific Advisory Council Member *Cure Sanfilippo Foundation*
Period	2015 – present
Address	P.O. Box 6901, Columbia, SC 29260
Position	NIH Study Section Reviewer *Therapeutic Approaches to Genetic Diseases Study Section*
Period	2016-2017
Address	Center for Scientific Review (CSR), NIH Bethesda, MD 20892
Position	Site Visit Intramural Program Reviewer *Eunice Kennedy Shriver - National Institute of Child Health and Human Development*
Period	June 1-2, 2016
Address	Eunice Kennedy Shriver NICHD Bethesda, MD 20892
Position	Research Grant Section Reviewer for NSERC *Natural Sciences and Engineering Research Council of Canada (NSERC)* *Conseil de recherches en sciences naturelles et en génie du Canada (CRSNG)*
Period	2017
Address	50 Albert Street, Ottawa ON K1A 1H5 Ottawa, ON Canada
Position	MRC Grant Reviewer Medical Research Council, United Kingdom
Period	2018
Address	2nd Floor, David Phillips Building, North Star Avenue, Swindon Wiltshire SN2 1FL, United Kingdom
Position	Grant Reviewer Parkinson's UK Research
Period	2015-present
Address	Parkinson's UK 215 Vauxhall Bridge Road London, SW1V 1EJ United Kingdom
Position	Grant Reviewer ELA – European Leukodystrophies Association
Period	2014-present
Address	2, rue Mi-les-Vignes BP 61024 54521 Laxou Cedex FRANCE

Editorial Boards	Molecular Genetics and Metabolism (MGM) Reports - ISSN: 2214-4269, Elselvier Editorial Board Member
Journal Reviewer	Molecular Therapies, Journal of Clinical Investigation, Lancet, Nature Medicine, Molecular Genetics and Metabolism, Pediatrics, Journal of Neuroscience Research, Neurology, Clinical Genetics, Journal of Inherited Metabolic Diseases

Honours and Awards

2006 Life for Luke Foundation – scholarship for research fellowship: Development of Novel Therapies with Small Molecules for Lysosomal Storage Diseases. The Hospital for Sick Children, Toronto, ON, Canada

2007 Exceptional Trainee Award – Genetics & Genomic Biology Program – Research Institute, The Hospital for Sick Children, Toronto, ON, Canada

2010 National Tay-Sachs Allied Disease Association, Inc. Research Initiative Award. May 2010

2016 The Mathew Forbes Romer Foundation, Honoree for Contributions to LSDs. Seagate Country Club, Delray Beach, FL.

2017 Shanghai Summit Forum on Pediatric Endocrine & Genetic Metabolic Diseases Appreciation Award, Shanghai, China

Affiliations and Activities

2002-present	American Society of Human Genetics (ASHG)
2003-2008	The College of Physicians and Surgeons of Ontario (CPSO)
2005-present	American Board of Medical Genetics (ABMG)
2006-2009	Canadian College of Medical Geneticists (CCMG)
2007-present	American College of Medical Genetics (ACMG)
2007-2015	Society of Inborn Error of Metabolism (SSIEM)
2016-present	Society of Inherited Metabolic Diseases (SIMD)
2009-present	Maryland Board of Physicians (MBP)
2012-present	American Society of Biochemistry and Molecular Biology (ASBMB)
2014-present	American Association for the Advancement of Science (AAAS)
2014-present	American Society for Mass Spectrometry (ASMS)

Current and Past Medical Licenses

Florida Medical Board - Physician and Surgeon Unrestricted License – M125350 (exp. 01/31/2020)

Drug Enforcement and Administration (DEA) - Controlled Substances Regulated Chemical – FM1743562 (exp.01/31/2022)

Maryland Board of Physicians – Physician and Surgeon Unrestricted License – D69890 (exp. 09/30/2019)

College of Physicians and Surgeons of Ontario (CPSO) – Canada – registration 79783 – issued 09/03/2003 – expired 06/30/2008

Medical Board Certifications

American Board of Medical Genetics and Genomics – Clinical Genetics Subspecilaty
Issued 09/01/2005. Re-certified 01/01/2016 – MOC status – meeting requirements

American Board of Medical Genetics and Genomics – Biochemical Genetics Subspecilaty

Issued 09/01/2007. Re-certified 01/01/2018 – MOC status – meeting requirements

Canadian College of Medical Geneticists – Clinical Genetics
Issued 09/30/2006

B. Academic History

Medical education

Location Universidade Federal do Paraná Medical School, Curitiba, Brazil
Coordenacao do Curso de Medicina
Rua Padre Camargo, 280 – 4th floor
Curitiba – PR, 80.060-240 Brazil
Period 1994 – 1999
Degree M.D.
Date 01/2000

Postgraduate medical education

Residency

Specialty Pediatrics and Medical Genetics
Location Hospital de Clinicas de Porto Alegre, Universidade Federal do Rio Grande do Sul Medical School
Serviço de Genética Medica
Rua Ramiro Barcelos, 2350
Porto Alegre, RS; 90035-903 Brazil.
Period 01/2000-06/2003

Fellowships

Specialty Clinical and Metabolic Genetics
Location The Hospital for Sick Children
Division of Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto
555 University Ave.
Toronto, ON, M5G 1X8, Canada
Period 07/2003 – 06/2005
Specialty Clinical & Biochemical Research fellowship
Location The Hospital for Sick Children
Lysosomal Storage Disease Research Center, Div. of Clinical and Metabolic Genetics
Genetics & Biology Program, Research Institute
555 University Ave.
Toronto, ON, M5G 1X8 Canada
Period 07/2005 – 06/2008

Graduate Education

Location University of Toronto
Faculty of Medicine
Institute of Medical Sciences
Medical Sciences Bldg. Ste. 7213
1 King's Circle, Toronto, ON, M5S 1A8 Canada
Period 01/2006-08/2008
Degree Ph.D.
Date 11/2008
Doctoral thesis Juvenile GM2 gangliosidosis: a model for investigation of small-molecule therapies for lysosomal storage diseases
Supervisor Joe T.R. Clarke M.D., Ph.D., FRCP(C)
Co-supervisor Don Mahuran, Ph.D.

Grants and Projects

Active

R21NS110406 Maegawa (PI) 07/01/19 - 01/30/21 2 calendar
NIH/NINDS $137,500/yr
Exosomal CNS-Delivery of Therapies for a Lysosomal Disorder
After the development and implementation of high throughput screening assay for globoid cell leukodystrophy (GLD), or Krabbe disease, we will characterize and validate novel therapeutic agents to treat this devastating neurodegenerative genetic disease. The availability of GLD patient neural cells and murine models will allow us to achieve a lead compound for early clinical studies.

R000 Maegawa (Co-PI) 09/01/19 - 03/31/20 0.4 calendar
The Legacy of Angels Foundation $21,000
Development of high throughput screening assay for acid ceramidase and UGT8 inhibitor
In conjuction with University of Washington, Seattle, WA and collaboration of Dr. Micahel Gelb PhD, we will develop new high throughput assays of human acid ceramidase and human UGT8. We will also carry out a pilot screening of ~1,200 commercially available compound library. This latter step is needed to optimize the assays to ensure that they are sufficiently robust to take forward to the NIH high throughput screen.

R000 Maegawa (PI) 06/01/18 - 05/31/19 1.2 calendar
Hunter's Hope Foundation $20,000
Leukodystrophy Care Network at UF Health Shands Hospital
Response to Invited Leukodystrophy Care Network Center (LCN) Certification Request for Proposals. The purpose of this RFP is to gain knowledge of services currently available at your medical institution. Currently, we are offering funding to support a portion of the LCN Clinical Coordinator position for Candidate Centers who meet the baseline requirements for LCN certification.

R000 Maegawa (PI) 05/1/15 – 04/30/19 0.24 calendar
Protalix Inc. $96,082.08/patient
A Multi Center Extension Study of PRX-102 Administered by Intravenous Infusions Every 2 Weeks for 24 Months to Adult Fabry Patients
This is an extension of previous phase I/II clinical trial to continue to assess the safety, tolerability and exploratory efficacy parameters of PRX-102 in adult patients with Fabry disease
Role: Principal Investigator – University of Florida

NV1205-008 Maegawa (PI) 08/01/18 - 02/28/21 0.6 calendar
Neuro Via Inc. $10,582
A Non-Interventional Study to Prospectively Assess Baseline Status and Disease Progression in Male Children with X-Linked Adrenoleukodystrophy.
This is a non-interventional study that follows general principles of periodic assessment of XALD patients in routine practice. No therapeutic intervention is required and no changes to the patient treatment are necessary.

RFP_2018LSD1 Maegawa (PI) 09/01/18-08/30/20 1.2 calendar
Pfizer Grants $150,000
Lysosomal Storage Disorders Fellowship for Nurse Practitioners
Pfizer educational grant program to assist training of RN or RNP for providing care of patients affected with lysosomal storage diseases.

R000 Maegawa (PI) 12/22/15 – 10/21/18 1 calendar
Dept. of Pediatrics, University of Florida
Small molecule therapy development and clinical studies in lysosomal storage diseases
The major goal is to study develop small molecules that can be used as therapeutic agents for lysosomal storage diseases. Secondly, the study aims to delineate the natural history of lysosomal storage disorders as well as determine potential biomarkers to evaluate these genetic disorders.

R000 Maegawa (PI) 10/02/15 – 10/05/18 0.5 calendar
Dept. of Pediatrics, University of Florida
Lysosomal Disease Bank
This is a specimen bank which aims to store cells, blood, urine and clinical data collected from patients diagnosed with lysosomal storage diseases.
Role: Principal Investigator

Past Awards

NV1205-008 Maegawa (PI) 08/01/18 - 02/28/21 0.6 calendar
Neuro Via Inc. $10,582
A Non-Interventional Study to Prospectively Assess Baseline Status and Disease Progression in Male Children with X-Linked Adrenoleukodystrophy.
This is a non-interventional study that follows general principles of periodic assessment of XALD patients in routine practice. No therapeutic intervention is required and no changes to the patient treatment are necessary.
Role: Principal Investigator

5R01NS079655-01A1 Maegawa (PI) 03/01/13 - 02/28/18 6 calendar
NIH/NINDS $384,989/yr
Development of a HTS Assay for a Neurological Lysosomal Disease
In this proposal, we plan to develop a neural cell-based high-throughput screening assay to identify potential small molecule that can reduce the levels of psychosine in the brain of patient affected with Krabbe disease.
Role: Principal Investigator – Johns Hopkins University/University of Florida

1R03MH098689-01 Maegawa (PI) 09/18/12 – 07/31/16 0.6 calendar
NIH/NINDS $40,500/yr
A Novel Cell-Based Assay to Identify Small Molecules for B-Galactocerebrosidase
The objective of this project is to develop a patient cell-based high throughput screening (HTS) assay for the identification of molecular probes that enhance the residual enzyme activity of β-galactocerebrosidase (GALC; deficient in Krabbe disease).
Role: Principal Investigator – Johns Hopkins University

1R21NS071535-01 Maegawa (PI) 6/1/10 – 5/31/11 4.2 calendar
NIH/NINDS $164,000
A HTS assay to identify molecular probes for arylsulfatase A
Development of a high-thorughput screen (HTS) assay to identify small molecules to assist the misfolded mutations observed in late-onset forms of metachromatic leukodystrophy (MLD)
Role: Principal Investigator – Johns Hopkins University

3R21NS071535-01S1 Maegawa (PI) 06/1/11 – 05/31/12 4.2 calendar
NIH/NINDS $41,000
A HTS assay to identify molecular probes for arylsulfatase A
Fast Track Entry of Assay Development Projects into the Roadmap Molecular Libraries Screening Center Network for HTS assay developed for arylsulfatase A (ASA).
Role: Principal Investigator – Johns Hopkins University

R000 Maegawa (PI) 06/1/10 - 05/31/12 3 calendar
National Tay-Sachs Disease $83,500
and Allied Disorders Association (NTSAD)
Characterization of small molecules identified in a HTS cell-based assay for ASA
Development of assays to validate small molecule candidates identified in the HTS for MLD
Role: Principal Investigator – Johns Hopkins University

R000 Maegawa (PI) 07/1/10 - 12/31/12 0.24 calendar
Shire HGT $100,144
An open label treatment protocol to evaluate the safety of Replagal treatment in patients with Fabry disease
This is an open-label extension of Study TKT028 designed to evaluate the long-term safety and clinical outcomes of treatment with Replagal administered to patients with Fabry disease who completed TKT028 at a dose of 0.2 mg/kg every other week for 12 months.
Role: Principal Investigator – Johns Hopkins University

R000 Maegawa (PI) 11/27/12 – 10/02/13 0.12 calendar
Genzyme-Sanofi Corp. $24,745.65
Evaluation of Glycosphingolipid Clearance in Patients Treated With Agalsidase Alfa who Switch to Agalsidase Beta (The Inform Study)
Multi-center exploratory study to evaluate changes in glycosphingolipid levels and other Fabry disease parameters in male Fabry disease patients who were previously treated with agalsidase alfa (Replagal) 0.2 mg/kg every 2 weeks and who are being switched to agalsidase beta (Fabrazyme) 1.0 mg/kg q2w.
Role: Principal Investigator – Johns Hopkins University

R000 Maegawa (PI) 01/17/13 – 12/31/13 0.6 calendar
Genzyme-Sanofi Corp. $80,000
Lysosomal Storage Disease Educational Program
This is a one-year educational grant for training a PA/RN to build clinical expertise in assisting in the care of patients with lysosomal storage diseases.
Role: Principal Investigator – Johns Hopkins University

R000 Maegawa (PI) 05/01/13 – 09/30/15 0.24 calendar
Protalix Ltd. $156,810
A Phase 1/2, Open Label, Dose Ranging Study to Evaluate the Safety, Tolerability, Pharmacokinetics and Exploratory Efficacy Parameters of PRX-102 Administered by Intravenous Infusion Every 2 Weeks for 12 Weeks to Adult Fabry Patients
This is a phase I/II clinical trial to evaluate the safety, tolerability, pharmacokinetics and exploratory efficacy parameters of PRX-102 in adult patients with Fabry disease
Role: Principal Investigator – Johns Hopkins University

R000 Maegawa (PI) 05/01/13 – 09/30/16 0.24 calendar
Protalix Ltd. $859,757
An Extension of Phase 1/2, Open Label, Dose Ranging Study to Evaluate the Safety, Tolerability, Pharmacokinetics and Exploratory Efficacy Parameters of PRX-102 Administered by Intravenous Infusion Every 2 Weeks for 38 Weeks (9 Months) to Adult Fabry Patients
This is an extension of phase I/II clinical trial to continue to assess the safety, tolerability, pharmacokinetics and but now also including further exploratory efficacy parameters of PRX-102 in adult patients with Fabry disease
Role: Co-Principal Investigator – Johns Hopkins University

R000 Maegawa (PI) 07/01/12 – 01/30/16 1.2 calendar
National Mucopolysaccharidosis (MPS) Society $45,000/yr
Induced-neuronal (iN) cells as tools to study the pathogenesis of neurological manifestations in MPS-II
Using iN cells, the characterized “neuro-pathogenic” cascades can then be used as targets to develop specific therapies for the neurodegeneration commonly observed in neuronopathic MPS-II and others lysosomal storage diseases.
Role: Principal Investigator – Johns Hopkins University/University of Florida

Teaching Activities

Position Predoctoral Teaching Faculty, Human Genetics Graduate Program, McKusick-Nathans Inst. Of
2009-2015 Genetic Medicine, Johns Hopkins School of Medicine, Baltimore, MD
Co-director Molecular Mechanisms in Genetic Diseases – course in Human Genetics Graduate Program,
2010-2015 McKusick-Nathans Inst. Of Genetic Medicine, Johns Hopkins School of Medicine, Baltimore, MD
Position Predoctoral Teaching Faculty, Interdisciplinary Graduate Program in Biomedical Sciences,
2015-present College of Medicine, Departments of Neurosciences, Immunology and Microbiology, University of Florida, Gainesville, FL

Mentored students and postdoctoral

Melanie Hare	Undergraduate student	IFAS/University of Florida	2018- present
Jairo Hernandez	Undergraduate student	IFAS/University of Florida	2018- present
Isabella Fabian	Undergraduate student	IFAS/University of Florida	2018- present
Seif Hanbali	Undergraduate student	IFAS/University of Florida	2018- 2019
Kuber Bhardwaj	Undergraduate student	IFAS/University of Florida	2018- 2019
Madison Pedreira	Undergraduate student	IFAS/University of Florida	2018- 2019
Priyanka L Devaguptapu	Undergraduate student	Health Science/University of Florida	2018- present
Dae Song Jang Ph.D	Lab Postdoctoral fellow – research	–University of Florida	2015-2017
Melani Solomon Ph.D.	Lab Postdoctoral fellow – research	– Johns Hopkins University	2013 - 2015
Alvaro Reategui	Undergraduate Molecular Biology	Johns Hopkins University	2014 - 2015
Meeka Garcia BSc.	Post-baccalaureate - Medicine candidate	-– Johns Hopkins University	2014 – 2015
Samantha Wax	Undergraduate - Medicine candidate	-– Johns Hopkins University	2014 – 2015
Maria Reategui	Undergraduate Molecular Biology	Johns Hopkins University	2014 – 2015
Shin-ichi Kano, M.D., Ph.D.	Postdoctoral fellow – research	Johns Hopkins University	2012 – 2014
Shilpa Patil Ph.D.	Lab Postdoctoral fellow – research	– Johns Hopkins University	2013 – 2014
Kristen Pusateri R.N.	Research RN trainee in LSD clinical research	– Johns Hopkins University	2013
Haifeng Geng Ph.D.	Lab Postdoctoral fellow – research	– Johns Hopkins University	2011 – 2013
Dan-Ju Tso Ph.D.	Lab Postdoctoral fellow – research	– Johns Hopkins University	2010 – 2011

Thesis commitees

Kranthi Vysyaraju B.Sc.	Master of Science Candidate, Thesis Committee Member	2012- 2013

Janet Hsu BSc. Ph.D. Candidate Graduate Student 2012 - 2014
Thesis Committee Member

Position Clinical Teaching Faculty

Program Residency Programs - Medical Genetics (3-year), Combined Pediatrics/Medical Genetics (4-year) and Combined Internal Medicine/Medical Genetics (5-year), Combined Maternal Fetal Medicine (MFM)/Medical Genetics (4-year), Pediatrics (3-year) McKusick-Nathans Inst. of Genetic Medicine, The Johns Hopkins University School of Medicine

Activities Mentor both clinical and elective research trainees (residents and fellows) in medical genetics and genomics to acquire competencies in medical genetics to care for patients with the full spectrum of genetic disorders and predispositions, as well as to obtain the necessary teaching skills to contribute to the integration of genetics to medicine. To train residents to be competent in cutting-edge clinical and/or basic research in genetics to play a leadership role in the future of genetics and genomics medicine.
Trainees are also available to compete for research funding and for academic career in genetic medicine.

Mentored Pediatric/Genetics Residents and Clinical and Biochemical Genetics Residents - Johns Hopkins

Hans Bjornsson MD, PhD, Jill Fahrner MD, PhD, Jessica Duis MD, Carlos Ferreira MD, Karin Weiss MD, Nara Sobreira, MD, PhD, Hind Al Saif, MD, Pedro Argoti, MD, Mellisa Russo MD, An Dang Do MD, PhD, Jasmin Roohi, MD, PhD, Mauricio DeCastro MD, Teresa Martino MD, Hilary Vernon MD, PhD, Vinayak Kottor MD, Regina Zambrano MD, Abe Ellis MD, Michael Walsh MD.

Mentored Pediatric Residents - UF

Seon Lee MD, Parvathi Nataraj MD, Ahsan Rizvi MD, John Copolla MD, Chelsea Zimmerman MD, Elizabeth Donner MD.

Mentored Pediatric Residents - UF

Kathryn Patrick MD

Mentoring Young Faculty UF

Anatalia Labiloy MD

Publications

Book chapters

1. Maegawa GHB, Steiner R. Treatment of Genetic Disorders. Chapter 16 in **Medical Genetics in Pediatric Practice**. American Academy of Pediatrics. Editor: Robert Saul MD, FAAP, FAMCG. 2012
2. Maegawa GHB. Chapter: Lysosomal Storage Diseases. **Clinical Decision Support: Pediatrics, LLC** – online content Management System Online. Editors: Julia McMillan MD. 2013.
3. Maegawa GHB, Pastores G. GM2 gangliosidosis. **Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease**. **5th Ed.** Elsevier. Editors: Roger N. Rosenberg MD, Salvatore DiMauro MD, Henry L. Paulson MD, Louis Ptácek MD, Eric J. Nestler MD. *In press. 2014*
4. Maegawa GHB. Chapter: Lysosomal Disorders of The Nervous System. **Neurobiology of Disease, 2nd Ed.** Editor: Sid Gilman MD. 2014

Articles (refereed)

https://www.ncbi.nlm.nih.gov/myncbi/gustavo.maegawa.1/bibliography/public/

1. Jiang W, Yi M, Maegawa GHB, Zhang H. Ambroxol improves skeletal and hematological manifestations on a child with Gaucher disease. J Hum Genet. 2019 Dec 11;. doi: 10.1038/s10038-019-0704-3. PMID: 31822786.
2. Microphthalmia and linear skin defects syndrome: Precise diagnosis guides prognosis.Satcher KG, Maegawa GHB, Schoch JJ*. Pediatr Dermatol*. 2019 Aug 2.PMID: 31373408
3. O'Donnell-Luria AH, Pais LS, Faundes V, Wood JC, Sveden A, Luria V, Abou Jamra R, Accogli A, Amburgey K, Anderlid BM, Azzarello-Burri S, Basinger AA, Bianchini C, Bird LM, Buchert R, Carre W, Ceulemans S, Charles P, Cox H, Culliton L, Currò A, Deciphering Developmental Disorders Study, Demurger F, Dowling JJ, Duban-Bedu B, Dubourg C, Eiset SE, Escobar LF, Ferrarini A, Haack TB, Hashim M, Heide S, Helbig KL, Helbig I, Heredia R(29), Héron D, Isidor B, Jonasson AR, Joset P, Keren B, Kok F, Kroes HY, Lavillaureix A, Lu X, Maas SM, Maegawa GHB, Marcelis CLM, Mark PR(37), Masruha MR(38), McLaughlin HM, McWalter K, Melchinger EU, Mercimek-Andrews S, Nava C, Pendziwiat M, Person R, Ramelli GP, Ramos LLP, Rauch A, Reavey C, Renieri A, Rieß A, Sanchez-Valle A, Sattar S, Saunders C, Schwarz N, Smol T, Srour M, Steindl K, Syrbe S, Taylor JC, Telegrafi A, Thiffault I, Trauner DA, van der Linden H Jr, van Koningsbruggen S, Villard L, Vogel I, Vogt J, Weber YG, Wentzensen IM, Widjaja E, Zak J, Baxter S, Banka S, Rodan LH. Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *Am J Hum Genet.* 2019 May 9. PMID: 31079897
4. Schiffmann R, Goker-Alpan O, Holida M, Giraldo P, Barisoni L, Colvin RB, Jennette JC, Maegawa G, Boyadjiev SA, Gonzalez D, Nicholls K, Tuffaha A, Atta MG, Rup B, Charney MR, Paz A, Szlaifer M, Alon S, Brill-Almon E, Chertkoff R, Hughes D. Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: a 1-year Phase 1/2 clinical trial. *J Inherit Metab Dis.* 2019 May;42(3):534-544. doi: 10.1002/jimd.12080. PMID: 30834538
5. Maegawa G.H.B. Leukodystrophies in Lysosomal Storage Diseases. *J Child Neurol.* 2019 May;34(6):339-358. PMID: 30757954
6. Katabuchi, A. U., Godoy, V., Shil, P., Moser, A., & Maegawa, GHB. Serendipitous effects of β-cyclodextrin on murine model of Krabbe disease. *Mol Genet Metab Rep.* 2018 Mar 26;15:98-99 PMID: 30023296.
7. Metz KA, Teng X, Coppens I, Wang TS, Lamb HM, Chen X, Zhang Y, Haberlandt E, Esther Leshinsky-Silver E, Rosenfeld JA, Bi W, Yang Y, Ong MT, Mordekar SR, Parker MJ, Crooks D, Wagner B, McKnight D, Markello T, Peariso K, Burrow T, Meng H, Pratt M, Makhseed N, Garnica A, Danylchuk NR, Agadi S, Gbedawo H, Stanley C, Jayakar P, Agrawal PJ, Berry GT, Loddenkemper T, Alber M, Isabelle Prehl27, Maegawa GHB, Hartman AL, Hardwick JM. KCTD7 mutations define a distinct neurodegenerative disorder with defective autophagy. *Ann Neurol.* 2018 Nov;84(5):766-780. PMID: 30295347
8. Maegawa G, Fatemi A, Furuya H, Al-Jasmin, F, Raiman J, Pastores G. Neurological Manifestations in Lysosomal Storage Diseases. Lancet Neurology. Under review. 2018.
9. Nashabat M, Maegawa G, Nissen PH, Nexo E, Al-Shamrani H, Al-Owain M , Alfadhel M. Long Term Outcome of Four Patients with Transcobalamin Deficiency Caused by Two Novel TCN2 Mutations. J Ped Hematology/Onc 2017 Nov;39(8):e430-e436. PMID: 28538514
10. Ramesh A, Diaz J, Nogee L, Duis J, Jang DS, Lawson C, Maegawa G. Premature Identical Twin Neonates with Sleep *Apnea. Clin Pediatr (Phila)*. 2017 Oct;56(11):1075-1078. doi: 10.1177/0009922817698810. Epub 2017 Mar 20. PMID: 28871878
11. Jang DS, Katabuchi A, Solomon M, Moser A, Maegawa G. Identification of psychosine-reducing small molecule agents for Krabbe disease. *Mol Genetics and Metabolism* 120 (2016) S17–

S145.

12. Jang D, Ye W, Tian G, Solomon M, Southall N, Hu X, Marugan J, Ferrer M, Maegawa G. Cell-based High-throughput Screen identifies GALC Enhancers as Potential Small Molecules Therapies for Krabbe Disease. J Neurosci Res. 2016 Nov;94(11):1231-45. PMID: 27638606
13. Klionsky DJ, Abdelmohsen K, Abe A, Abedin MJ, Abeliovich H, Acevedo Arozena A *et al.* Maegawa G, *at al.* Zughaier SM. Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). *Autophagy***.** 2016 Jan 2;12(1):1-222. PMID: 26799652
14. Lalani S, Liu P, Rosenfeld JA, Watkin LB, Chiang T, Leduc M, Zhu W, Ding Y, Pan S, Miyake C, Shinawi M, Gambin T, Eldomery MK, Akdemir ZHC, Emrick L, Wilnai Y, Schelley S, Koenig MK, Memon N, Farach LS, Coe B, Azamian M, Hernandez P, Zapata G, Jhangiani SN, Muzny DM, Lotze T, Clark G, Wilfong A, Northrup H, Adesina A, Bacino C, Scaglia F, Bonnen PE, Duis J, Crosson J, Maegawa GHB, McGill J, Boerwinkle E, Graham B Beaudet A, Eng CM, Hanchard N, Xia F, Orange JS, Gibbs RA, Lupski JR, Yang Y. Recurrent muscle weakness with rhabdomyolysis and cardiac arrhythmia due to bi-allelic TANGO2 mutations. *A J Hum Gent, 2016* 4;98(2):347-57. PMID: 26805781
15. Goker-Alpan O, Gambello MJ, Maegawa GHB, Nedd KJ, Gruskin DJ, Blankstein L, Weinreb NJ. Reduction of plasma globotriaosylsphingosine levels after switching from agalsidase alfa to agalsidase beta as enzyme replacement therapy for Fabry disease. *JIMD Rep.* 2016;25:95-106. PMID: 26303609
16. Kano SI, Yuan M, Cardarelli RA, Maegawa G, Higurashi N, Gaval-Cruz M, Wilson AM, Tristan C, Kondo MA, Chen Y, Koga M, Obie C, Ishizuka K, Seshadri S, Srivastava R, Kato TA, Horiuchi Y, Sedlak TW, Lee Y, Rapoport JL, Hirose S, Okano H, Valle D, O'Donnell P, Sawa A, Kai M. Clinical utility of neuronal cells directly converted from fibroblasts of patients for neuropsychiatric disorders: studies of lysosomal storage diseases and channelopathy**.** *Current Molecular Medicine* 2015;15(2):138-45. PMID: 25732146
17. Teixeira CA, Miranda CO, Sousa MM, Santos TE, Malheiro AR, Solomon M, Maegawa GH, Brites P, Sousa MM. Early axonal loss accompanied by impaired endocytosis, abnormal axonal transport, and decreased microtubule stability occur in the model of Krabbe's disease. *Neurobiol Dis.* 2014 Jun;66:92-103. PMID: 24607884
18. Ribbens J, Moser AB, Hubbard W, Bongarzone E, Maegawa GHB. A novel disease-cell model for a neurodegenerative lysosomal disease with pathogenic and therapeutic implications. *Mol Genet Metab.* 2014 Feb;111(2):172-83. PMID: 24094551
19. Alqahtani E, Huisman TA, Boltshauser E, Scheer I, Güngör T, Tekes A, Maegawa GH, Poretti A. Mucopolysaccharidoses type I and II: New neuroimaging findings in the cerebellum. *Eur J Paediatr Neurol.* 2013 Dec 31. pii: S1090-3798(13)00209-2. PMID: 24423630
20. Pastores GM, Maegawa GH. Clinical neurogenetics: neuronopathic lysosomal storage disorders. *Neurol Clin.* 2013 Nov;31(4):1051-71. PMID: 24176423
21. Travaglini L, Brancati F, Silhavy J, Iannicelli M, Nickerson E, Elkhartoufi N, Scott E, Spencer E, Gabriel S, Thomas S, Ben-Zeev B, Bertini E, Boltshauser E, Chaouch M, Cilio MR, de Jong MM, Kayserili H, Ogur G, Poretti A, Signorini S, Uziel G, Zaki MS; International JSRD Study Group, Johnson C, Attié-Bitach T, Gleeson JG, Valente EM. Phenotypic spectrum and prevalence of INPP5E mutations in Joubert syndrome and related disorders. MaegawaGHB *is a member of the International JSRD Study Group. Eur J Hum Genet.* 2013 Oct;21(10):1074-8.
22. Patil SA, Maegawa GH. Developing therapeutic approaches for metachromatic leukodystrophy. *Drug Des Devel Ther.* 2013 Aug 8;7:729-45. PMID: 23966770
23. Ribbens J, Whiteley G, Furuya H, Southall N, Hu X, Marugan J, Ferrer M, Maegawa GH. A high-throughput screening assay using Krabbe disease patient cells. *Anal Biochem.* 2013 Mar 1;434(1):15-25. PMID: 23138179

24. Jones SA, Parini R, Harmatz P, Giugliani R, Fang J, Mendelsohn NJ; HOS Natural History Working Group on behalf of HOS Investigators. The effect of idursulfase on growth in patients with Hunter syndrome: data from the Hunter Outcome Survey (HOS). Maegawa GHB *is one of the HOS Investigators. Mol Genet Metab.* 2013 May;109(1):41-8. PMID: 23537841
25. Zulfiqar M, Lin DM, Graaf MvD, Barker, PD, Fahrner JA, Marie S, Morava E, Boer LD, Willemsen MAA, Vining E, Horska E, Engelke U, Wevers R, Maegawa GHB. Novel Proton MR Spectroscopy Findings in Adenylosuccinate Lyase Deficiency. *J Magn Reson Imaging.* 2013 Apr;37(4):974-80. PMID: 23055421
26. Maegawa GHB. GM2 gangliosodisis: the prototype of lysosomal storage disorders. *Dev Med Child Neurol.* 2012; 54(2):104-5. PMID: 22224667
27. Geng H, Whiteley G, Ribbens J, Zheng W, Southall N, Hu X, Marugan JJ, Ferrer M, Maegawa GHB. Novel Patient Cell-Based HTS Assay for Identification of Small Molecules for a Lysosomal Storage Disease. *PLoS One.* 2011;6(12):e29504.
28. Iannicelli M, Brancati F, Mougou-Zerelli S, Mazzotta A, Thomas S, Elkhartoufi N, Travaglini L, Gomes C, Ardissino GL, Bertini E, Boltshauser E, Castorina P, D'Arrigo S, Fischetto R, Leroy B, Loget P, Bonnière M, Starck L, Tantau J, Gentilin B, Majore S, Swistun D, Flori E, Lalatta F, Pantaleoni C, Penzien J, Grammatico P; International JSRD Study Group, Dallapiccola B, Gleeson JG, Attie-Bitach T, Valente EM. *Maegawa GHB is a member of the International JSRD Study Group*. Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies. *Hum Mutat.* 2010 May;31(5):E1319-31.
29. Travaglini L, Brancati F, Attie-Bitach T, Audollent S, Bertini E, Kaplan J, Perrault I, Iannicelli M, Mancuso B, Rigoli L, Rozet JM, Swistun D, Tolentino J, Dallapiccola B, Gleeson JG, Valente EM; International JSRD Study Group, Zankl A, Leventer R, Grattan-Smith P, Janecke A, D'Hooghe M, Sznajer Y, Van Coster R, Demerleir L, Dias K, Moco C, Moreira A, Kim CA, Maegawa G, *et al*.. Expanding CEP290 mutational spectrum in ciliopathies. *Am J Med Genet A*. 2009;149A(10):2173-80.
30. Maegawa GHB, Tropak M, Buttner JD, Rigat B, Fuller M, Pandit D, Tang L, Kornhaber GJ, Hamuro Y, Clarke JTR, Mahuran DJ. Identification and characterization of ambroxol as an enzyme-enhancement agent for Gaucher disease. *J Biol Chem* 2009;284(35):23502-16.
31. Maegawa GHB, Banwell B, Blaser S, Hawkins C, Arckerley C, Hayes J, Tlopak M, Sorge G, Clarke JTR. Substrate reduction therapy in juvenile GM2 gangliosidosis. *Mol Genet Metab.* 2009;98(1-2):215-24.
32. Maegawa GHB, Giersbergen PV, Yang S, Banwell B, Morgan CP, Tifft C, Clarke JTR. Pharmacokinetics, tolerability and safety of miglustat as treatment of GM2 gangliosidosis. *Mol Genet Metab* 2009;97(4):284-91.
33. Rohrbach M, Chitayat D, Maegawa G, Shanske S, Davidzon G, Chong K, Clarke JT, Toi A, Tarnopolsky M, Robinson B, Blaser S. Intracerebral periventricular pseudocysts in a fetus with mitochondrial depletion syndrome: an association or coincidence. *Fetal Diagn Ther* 2009 Mar 25;25(2):177-182.
34. Tropak, MB, Kornhaber, GJ, Rigat,BA, Maegawa GHB, Buttner J, Blanchard JE, Murphy C, Tuske SJ, Coales SJ, Hamuro Y, Brown E, Mahuran DJ. Identification of pharmacological chaperones for Gaucher disease nd characterization of their effects on b-glucocerebrosidase by hydrogen/deuterium exchange mass spectrometry. *ChemBioChem* 2008 (9), 2650-2662.
35. Kornhaber GJ, Tropak MB, Maegawa GHB, Tuske SJ, Coales SJ, Mahuran DJ, Hamuro Y. Isofagomine Induced Stabilization of Glucocerebrosidase" *ChemBioChem* 2008 (9), 2643-2649.
36. Maegawa GHB, Poplawski N, Andresen BS, Olpin SE, Nie G, Clarke JTR, Teshima I. Interstitial Deletion of 1p22.2p31.1 and Medium-Chain Acyl-CoA Dehydrogenase Deficiency in a Patient with Developmental Delay. *Am J Med Genet (Part A)* 2008 Jun 15;146A(12):1581-6.

37. Maegawa GHB, Stockley T, Tropak M, Kok F, Giugliani R, Mahuran D, Clarke JTR. The natural history of juvenile or subacute GM2 gangliosidosis: 21 unreported cases and literature review of 134 cases. *Pediatrics* 2006, Oct 2; e1550-1562.
38. Maegawa GHB, Chitayat D, Blaser S, Whyte H, Thomas M, Kim P, Kim J, Taylor G, McNamara P. Bilateral microtia with absence of external auditory meati, fused ossicles and Mondini dysplasia, duodenal atresia, anal anomalies, thyroid hemiaplasia and biliary atresia. A new mandibulofacial dysostosis syndrome? *Clin Dysmorph* 2006, 15: 191-196.
39. Seifert W, Holder-Espinasse M, Spranger S, Hoeltzenbein M, Rossier E, Dollfus H, Lacombe D, Verloes A, Chrzanowska KH, Maegawa GHB, Chitayat D, Kotzot D, Huhle D, Meinecke P, Albrecht B, Mathijssen I, Leheup B, Raile C, Hennies HC, Horn D. Mutational spectrum of COH1 and clinical heterogeneity in Cohen syndrome. *J Med Genet* 2006; 43(5):e22.
40. Loguercio JCL, Faermann R, Stein NR, Zandona D, Maegawa G. Focal dermal hypoplasia associated with split sternum – Goltz syndrome. *Clin Dysmorphol*. 2005, 14(1):37-9.
41. Bertoli-Avella AM, Giroud-Benitez JL, Ulucan, H, Barbosa E, Schaap O, van der Linde HC, Martignoni E, Lopiano L, Lamberti P, Fincati E, Antonini A, Stocchi F, Montagna P, Squitieri F, Marini P, Abbruzzese G, Fabbrini G, Marconi R, Dalla Libera A, Trianni G, Guidi M, De Gaetano A, Boff Maegawa G, De Leo N, Gallai V, de Rosa G, Vanacore N, van Duijn CM, Meco G, Oostra BA, Heutink P, Bonifati V and The Italian Parkinson Genetics Network. The contribution of *Parkin* to Early-Onset Parkinson's Disease A clinical and genetic study. *Mov Disord* 2005, 20(4):424-31.
42. Maegawa GH, Leite JC, Felix TM, da Silveira HL, da Silveira HE.. Clinical variability in KBG syndrome: Report of three unrelated families. *Am J Med Genet* A. 2004, 131A(2):150-4.
43. Ghisolfi ES, Maegawa GHB, Becker J, Zanardo AP, Strimitzer IM, Prokopiuk AS, Pereira ML, Carvalho T, Jardim LB, Lara DR. Impaired P50 sensory gating in Machado-Joseph Disease. *Clinical Neurophysiology* 2004, 115: 2231-2235.
44. Vargas RA, Maegawa GH, Taucher SC, Leite JC, Sanz P, Cifuentes J, Parra M, Munoz H, Maranduba CM, Passos-Bueno MR. Beare-Stevenson syndrome: Two South American patients with FGFR2 analysis. *Am J Med Genet*. 2003, 121A(1):41-6.
45. Werneck LC, Scola RH, Maegawa GHB, Werneck MC. Comparative analysis of PCR-deletion detection and immunohistochemistry in Brazilian Duchenne and Becker muscular dystrophy patients. *Am J Med Genet*. 2001, 103(2):115-20.
46. Scola RH, Werneck LC, Iwamoto FM, Maegawa GHB, Faoro LN, Caldeira FH. [True neurogenic outlet syndrome: report of 2 cases]. *Arq Neuropsiquiatr* 1999; 57:659-65.
47. Scola RH, Werneck LC, Iwamoto FM, Cabral NL, Maegawa GHB, Rogacheski E. [Ossification of the posterior longitudinal ligament in the cervical spine: case report]. *Arq Neuropsiquiatr* 1998; 56:480-485.

Articles (non-refereed)

1. Maegawa GHB, Schwartz IVD, de Souza, CFM, Giugliani R. Cardiovascular presentation of inborn errors of metabolism. *Rev Soc Cardio Rio G Sul* X (3) Jul/Aug/Sept, 2001.
2. Maegawa GHB, Centa LJR. Aspects of male factor in infertility. *Fam Saud Desenv* 2000, 2(1): 7-12.

Abstract Presented in Meetings and Symposia

1. Edelmann, MJ; Oue, M; Hu, W; Hernandez, J; Mirabel, R; Maegawa, GHB. Exosomes with Unique CNS-targeting Properties Brings Novel Therapeutic Strategy to Neuronopathic LSDs. 16th WORLD Symposium, Orlando, FL, Feb. 10-14, 2020.
2. Katabuchi A; Jang DS; Hernandez J; Mirabel R; Moser A; Maegawa GHB. Psychosine-reducing molecules as potential therapies for Globoid-cell Leukodystrophy or Krabbe disease. 16th WORLD Symposium, Orlando, FL, Feb. 10-14, 2020.
3. Katabuchi A; Jang DS; Hernandez J; Mirabel R; Ye Wenjuan; Hu, Xin; Kim D; Henderson M; Southall, N; Ferrer M; Marugan J; Maegawa GHB. GALC-folding assistant molecules as

potential therapies for Krabbe disease. 16th WORLD Symposium, Orlando, FL, Feb. 10-14, 2020.

4. Salazar, D; Hernandez, J; Fabian, I; Bhardwaj K; Hanbali S; Rodrigues, JN; Sousa, MM; Gelb, M; Maegawa, GHB. Cyclodextrin Analogs reduce psychosine cytotoxicity. 16th WORLD Symposium, Orlando, FL, Feb. 10-14, 2020.
5. Bhardwaj, Hanbali, Fabian I, Salazar D, Katabuchi A, Maegawa GHB. Investigating Small Molecule Therapies for Krabbe Disease, an inherited lysosomal leukodystrophy. Rare Disease Day 2019. University of Florida, Gainesville, FL. February 28, 2019.
6. Fabian I, Salazar D, Katabuchi A, Maegawa G. Cyclodextrin effects on the neurobehavioral phenotype of the Krabbe disease murine model. Research celebration Event. College of Medicine, University of Florida, Gainesville, FL. February 19, 2019.
7. Bhardwaj K, Hanbali S, Fabian I, Salazar D, Katabuchi A, Maegawa GHB. Investigating Small Molecule Therapies for Krabbe Disease, an inherited lysosomal leukodystrophy. Research celebration Event. College of Medicine, University of Florida, Gainesville, FL. February 19, 2019.
8. Katabuchi A, Shil A, Bhardwaj K, Godoy V, Maegawa GHB. Beneficial effects of cyclodextrin in the Twitcher mouse. Lysosomal Disease Gordon Research Conference, Galveston, TX. March 3-8, 2019.
9. Katabuchi A, Shil A, Bhardwaj K, Godoy V, Maegawa GHB. The effect of beta-cyclodextrin on the murine model of Krabbe disease. 15th WORLD Symposium, Orlando, FL, Feb. 4-8, 2019.
10. Cabrera-Marcello J, Katabuchi AU, Solomon, M, Moser A, Ye J, Southall N, Hu X, Marugan J, Ferrer M, Maegawa GHB. Cell-based High-throughput Screening Assays to identify small molecule therapies for a Neurological Lysosomal Disease Pediatric Research Day, University of Florida, Gainesville, FL May 21, 2018.
11. Cabrera-Marcello J, Katabuchi AU, Santostefanos K, Terada N, Maegawa GHB. Generation of Neurologically Relevant Disease-Cell Models to Study Lysosomal Storage Diseases. Pediatric Research Day, University of Florida, Gainesville, FL May 21, 2018.
12. Godoy V, Maegawa GHB. Validating small molecule therapies in a Krabbe disease cell model. UF COM Celebration of Research, University of Florida, Gainesville, FL. February 19-20, 2018.
13. Katabuchi A, Godoy V, Shil P, Maegawa GHB. Effects of psychosine-reducing agents in the Twitcher murine model for Krabbe disease. UF COM Celebration of Research, University of Florida, Gainesville, FL. February 19-20, 2018.
14. Katabuchi A, Santostefanos K, Terada N, Maegawa GHB. Generation of neurologically relevant disease-cell models for lysosomal diseases. UF COM Celebration of Research, University of Florida, Gainesville, FL. February 19-20, 2018.
15. Katabuchi A, Godoy V, Shil P, Maegawa GHB. Effects of psychosine-reducing agents in the Twitcher murine model for Krabbe disease. Gordon Research Conferences (GRC) - Glycolipid and Sphingolipid Biology. Galveston, TX. Feb. 11-16, 2018.
16. Katabuchi A, Godoy V, Shil P, Maegawa GHB. Effects of psychosine-reducing agents in the Twitcher murine model for Krabbe disease. 13th WORLD Symposium 2018, Lysosomal Disease Network - NINDS. San Diego, CA. Feb. 5-9, 2018.
17. Katabuchi A, Santostefanos K, Terada N, Maegawa GHB. Generation of neurologically relevant disease-cell models for lysosomal diseases. 13th WORLD Symposium 2018, Lysosomal Disease Network - NINDS. San Diego, CA. Feb. 5-9, 2018.
18. Jang DS, Bayona NG, Katabuchi AU, Zelmanovich R, Maegawa GHB. Generation of Neurologically Relevant Disease-Cell Models to Study Lysosomal Storage Diseases. 3rd Annual CTSI iPSC Retreat. March 2017. Gainesville, FL. Award best poster.
19. Jang DS, Bayona NG, Katabuchi AU, Maegawa GHB. Identification of psychosine-reducing small molecule agents for Krabbe disease. 14th WORLD Symposium 2017, Lysosomal Disease Network - NINDS. San Diego, CA. Feb. 13-17, 2017.
20. Jang DS, Tian G, Solomon M, Bongarzone E, Moser A, Maegawa G. Screen and identification of small molecules therapies to reduce elevated psychosine levels in globoid-cell

leukodystrophy. 11th WORLD Symposium 2016, Lysosomal Disease Network - NINDS. San Diego, CA. Feb. 29-March 4, 2016.

21. Jang DJ, Solomon M, Moser A, Shapiro T, Bongarzone E, Ye X, Southall N, Hu X, Marugan J, Ferrer M, Maegawa G. Cell-based quantitative high-throughput assays to identify small molecule therapies a neurological lysosomal storage disease. February 22, 2016. UF COM Celebration of Research, University of Florida, Gainesville, FL.
22. Tian G, Ye W, Southal N, Xin H, Marugan J, Ferrer M, Maegawa G. February 22, 2016. UF COM Celebration of Research, University of Florida, Gainesville, FL. Quantitative high-throughput screen to identify small molecule enhancers of mutant arylsulfatase A (ASA) in metachromatic leukodystrophy patient cell lines.
23. Maegawa GHB. Development of cell-based high-throughput screening assays for lysosomal diseases. Gordon Research Conferences – Lysosomal Diseases. March 15-20, 2015. Galveston, TX, US.
24. Patil S, Kano, S-I, Sawa A, Maegawa GHB. Using small molecules to increase the direct conversion of culture fibroblasts from MPS-II patients into induced-neuronal (iN) cells. 10th WORLD Symposium 2014, Lysosomal Disease Network - NINDS. San Diego, CA. Feb. 11-13, 2014. Accepted as poster presentation.
25. Ribbens J, Whiteley G, Furuya H, Southall N, Hu X, Marugan J, Ferrer M, Maegawa GHB. A High-throughput Screening Assay using Krabbe Disease Patient Cells. 9th WORLD Symposium 2013, Lysosomal Disease Network - NINDS. Orlando, FL. Feb. 8-10, 2013. Accepted as poster presentation.
26. Kano, S-I, Tristan C, Kondo MA, Koga M, Wilson A, Ishizuka K, Yuan M, Obie C, Sedlak T, Kai M, Valle D, Sawa A, Maegawa GHB. Glycosphingolipid storage in a lysosomal disease using patient-derived induced neuronal cells. 5th Annual Symposium Maryland Stem Cell Research Symposium. Annapolis, MD. October 4-5, 2012. Accepted as poster presentation.
27. Moser A, Hubbard W, Bongarzone E, Ribbens J, Maegawa GHB. Development of a Psychosine Assay to Identify Therapeutic Small Molecules for Krabbe Disease. 8th WORLD Symposium 2012, Lysosomal Disease Network - NINDS. San Diego, CA. Feb. 8-10, 2012. Accepted as platform presentation.
28. Ribbens J, Whiteley G; Furuya H; Sakai N, Southall N, Hu X, Marugan J, Ferrer M, Maegawa GHB. A High-throughput Screening Assay using Krabbe Disease Patient Cells. 7th WORLD Symposium 2011, Las Vegas, NV Lysosomal Disease Network - NINDS. Accepted as poster.
29. Obie C, Ribbens J, Furuya H; Sakai N, Maegawa GHB. A proposed cell-based high-throughput screening assay to identify potential small molecules agents for treatment of Krabbe disease. 7th WORLD Symposium 2011 Lysosomal Disease Network - NINDS. Las Vegas, NV. Accepted as poster presentation.
30. Walsh MF, Raymond KM, Freeze HH, He P, Maegawa GHB. An unusual transferrin glycoform in a patient with developmental delay, failure to thrive, anemia, and dysmorphic features suggests a new type of congenital disorder of glycosylation. American Society of Human Genetics 60th Annual Meeting, Washigton, DC. November 6-10, 2010. Accepted as poster presentation.
31. Maegawa GHB, Banwell B, Blaser S, van Giersbergen P, Sorge G, Tlopak M, Ackerley C, Hawkins C, Hayes J, Clarke JTR. Substrate reduction therapy with miglustat in juvenile GM2 gangliosidosis. 5th Annual WORLD Symposium. Lysosomal Disease Network - NINDS. February 18-20, 2009. San Diego, CA, US. Accepted as platform presentation.
32. Maegawa GHB, Tropak M, Buttner J, Kornhaber G, Rigat B, Clarke JTR, Don J. Mahuran. Ambroxol as a potential enzyme-enhancement agent for Gaucher disease. American Society of Human Genettics Annual Meeting. November 11-15, 2008. Philadelphia, PA, US. Accepted as poster presentation.
33. Maegawa GHB, Tropak M, Buttner J, Kornhaber G, Rigat B, Clarke JTR, Don J. Mahuran. Identification of ambroxol as a potential enzyme-enhancement agent for Gaucher disease. Society of Inborn Error of Metabolism Annual Symposium. September 2-5, 2008. Lisbon, Portugal.

34. Maegawa GHB, Banwell B, Blaser S, van Giersbergen P, Sorge G, Tlopak M, Ackerley C, Hawkins C, Hayes J, Clarke JTR. Substrate reduction therapy with miglustat in juvenile GM2 gangliosidosis. Society of Inborn Error of Metabolism Annual Symposium. September 2-5, 2008. Lisbon, Portugal.
35. Maegawa GHB, Tropak M, Buttner J, Kornhaber G, Rigat B, Clarke JTR, Don J. Mahuran. Identification of ambroxol as a potential enzyme-enhancement agent for Gaucher disease. Garrod Symposium 2008. May 2-3, 2008. Toronto, ON, Canada. Plataform presentation.
36. Maegawa GHB, Banwell B, Tropak M, Buttner J, Rigat B, Hawkins C, Ackerley C, Hayes J, Tifft C, Tlopak M, Sorge G, Blaser S, Giersbergen PV, Mahuran DJ, Clarke JTR. Juvenile GM2 gangliosidosis: a model for investigation of small-molecule therapies for lysosomal storage diseases. Paediatric Research Day. May 28, 2008, Toronto, ON, Canada. Accepted.
37. Mahuran DJ, Tropak M, Maegawa GHB, Buttner J, Rigat B, Clarke JTR. Lending a helping hand: chaperones and lysosomal storage disease. Society for Inherited Metabolic Disorders (SIMD), 2008 Annual Meeting. March 2-5, 2008. Pacific Grove, California, USA.
38. Maegawa GHB, Tropak M, Buttner J, Kornhaber G, Rigat B, Clarke JTR, Mahuran D. Identification of Ambroxol as a Potential Enzyme Enhancement-Agent for Gaucher Disease. 4th WORLD Symposium 2008, Lysosomal Disease Network - NINDS. February 13-15, 2008, Las Vegas, NV, USA.
39. Maegawa GHB, Tropak M, Buttner J, Kornhaber G, Rigat B, Clarke JTR, Mahuran D. Ambroxol as a Potential Pharmacological Chaperone for Gaucher Disease. Research Institute Retreat, The Hospital for Sick Children. November 2007, Toronto, ON, Canada.
40. Maegawa GHB, Buttner J, Tropak M, Kok F, Stockley T, Clarke JTR, Mahuran DJ. Pyrimethamine as a novel pharmacological chaperone for late-onset forms of GM2 gangliosidosis. Society for the Study of Inborn Errors of Metabolism, Annual Symposium. September 4th-7th 2007, Hamburg, Germany. Oral presentation.
41. Teshima I, Maegawa GHB, Poplawski N, Andresen BS, Nie G, Clarke JTR. Interstitial deletion of 1p22.2->1p31.1 causing MCAD deficiency and review of interstitial deletions and duplications of this region. Sixth European Cytogenetic Conference, Istanbul , Turkey, July 7-10, 2007.
42. Maegawa GHB, Poplawski N, Andresen BS, Nie G, Clarke JTR, Teshima I. Novel case of *ACADM* missense mutation with 1p22.2p31.1 interstitial deletion causing MCAD deficiency. Genomic Disorders Workshop, Sanger Institute Wellcome Trust Conference, Cambridge, UK. March 21-24, 2007.
43. Maegawa GHB, Banwell B, Blaser S, Hawkins C, Hayes J, Clarke JTR. Pharmacokinetics and tolerability of miglustat in juvenile GM2 gangliosidosis. Presented as poster at 56th Annual Meeting of The American Society of Human Genetics. October 10-13, 2006. New Orleans, LO, USA.
44. Tropak M, Maegawa GHB, Kok F, Clarke JTR, Mahuran DJ. Potential Potential pharmacological chaperones for late-onset GM2 gangliosidosis. Presented as poster at 56th Annual Meeting of The American Society of Human Genetics. October 10-13, 2006. New Orleans, LO, USA.
45. Rohrbach M, Chitayat D, Maegawa G, Davidzon G, Shanske S, Chong K, Blaser S. Are periventricular intracranial cysts on prenatal MRI an early diagnostic feature for mitochondrial depletion syndrome? Presented as poster at 56th Annual Meeting of The American Society of Human Genetics. October 10-13, 2006. New Orleans, LO, USA.
46. Clarke JTR , Maegawa GHB, Banwell B, Blaser S, Hawkins C, Hayes J. Pharmacokinetics of miglustat in juvenile GM2 gangliosidosis. Presented as poster at 10th International Congress of Inborn Errors of Metabolism ICIEM, Makuhari Messe. September, 12-16 2006. Chiba, Japan.
47. Clarke, JTR, Maegawa GHB, Tropak M, Kok F, Mahuran DJ. Potential Pharmacological chaperones for juvenile and late-onset GM2 gangliosidosis. Presented as poster at 10th International Congress of Inborn Errors of Metabolism ICIEM, Makuhari Messe. September, 12-16 2006. Chiba, Japan.
48. Maegawa GHB, Tropak M, Kok F, Clarke JTR, Mahuran DJ. Novel pharmacological chaperones for juvenile and late-onset GM2 gangliosidosis. Presented as poster at 9th International Symposium on Mucopolysaccharide and Related Diseases, June 29-July 2, 2006, Venice, Italy.

49. Maegawa GHB, Tropak M, Kok F, Clarke JTR, Mahuran DJ. Novel pharmacological chaperones for juvenile and late-onset GM2 gangliosidosis. NIH/NINDS Workshop: Glycosphingolipids in Health and Disease. April 5-6, 2006. Alexandria, Virginia, USA.
50. Maegawa GHB, Tropak M, Mahuran D, Stockley T, Callahan JW, Giugliani R,Kok F, Clarke JTR. Study of natural history and genotype analysis of juvenile GM2 gangliosidosis. Submitted to the American Society of Human Genetics Meeting. October 25 – 29, 2005. Salt Lake City, USA.
51. Chitayat D, Maegawa GHB, Sroka H, Boon L, Revencu N, Clapuyt P, Vikkula M, Blaser S**.** A Novel RASA1 Mutation Associated with Vein of Galen Malformation. Submitted to the American Society of Human Genetics Meeting. October 25 – 29, 2005. Salt Lake City, USA.
52. Feigenbaum A, Maegawa G, Poplawski N, Callahan JW, Olpin S, Strauss A. Case report of mitochondrial trifunctional protein deficiency (TFP) presenting with metabolic acidosis and ketosis. American Society of Human Genetics Meeting. October 26 – 30, 2004. Toronto, Canada.
53. Fishman L, Maegawa GHB, Babul-Hirji R, Teebi A, Chitayat D. Two cases of chromosome 8p23 deletion with different clinical presentation. American Society of Human Genetics Meeting. October 26 – 30, 2004. Toronto, Canada.
54. Rossi R, Sanseverino MT, Maegawa G, Ribas C, Jann M, Costa S, Golbert M, Shuler-Faccini L. National Information Center about Teratogens (SIAT): analysis about its impact over the pregnant women health during its 14 years of service. L. American Society of Human Genetics Meeting. October 26 – 30, 2004. Toronto, Canada.
55. McNamara PJ, Maegawa GHB, Whyte H,Thomas M, Kim P, Kim J, Chitayat D. Bilateral Microtia with Absence of External Auditory Meati, Mondini type Malformation, Duodenal Atresia, Thyroid Hemiplasia and Biliary Atresia. A New Syndrome? American Society of Human Genetics Meeting. October 26 – 30, 2004. Toronto, Canada.
56. Lo B, Maegawa GHB, Satodia P, Whyte H, Sermer M, Hutchinson J, Wilson JG, Gross G, Matyas G, Hinek A, Unger S, Chitayat D. Neonatal Marfan Syndrome: two cases with some unusual clinical findings. American Society of Human Genetics Meeting. October 26 – 30, 2004. Toronto, Canada
57. Seifert W, Holder M, Spranger S, Hoeltzenbein M, Rossier E, Maegawa G, Dollfus H, Chrzanowa KH, Horn D, Hennies HC. Cohen syndrome: mutational and transcriptional analysis of *COH1*. European Human Genetics Conference May 7 – 10, 2005. Prague, Czech Republic.
58. Maegawa GHB, Tropak M, Mahuran D, Stockley T, Callahan JW, Giugliani R,Kok F, Clarke JTR. Study of natural history and genotype analysis of juvenile GM2 gangliosidosis. Research Day, The Hospital for Sick Children. May 25th, 2005. Toronto, Canada.
59. Maegawa GHB, Whyte H, Thomas M, Kim P, Kim J, McNamara PJ, Chitayat D. Bilateral Microtia with Absence of External Auditory Meati, Mondini type Malformation, Duodenal Atresia, Thyroid Hemiplasia and Biliary Atresia. A New Syndrome? Course of Mammalian and Experimental Medical Genetics, Bar Harbour, Maine, USA, June 27 – July 9, 2004.
60. Maegawa GHB, Tropak M, Mahuran DJ, Hewson SJ, Callahan JW, Clarke JTR. Five year-old girl with neurodegeneration and decreased Hex B activity. SSIEM 41st Annual Symposium, Amsterdam, the Netherlands, August 31 – September 3, 2004.
61. Maegawa GHB, Tropak M, Mahuran DJ, Callahan JW, Giugliani R, Barros C, Kok F, Clarke JTR. Study of Natural History of Juvenile GM2 gangliosidosis. SSIEM 41st Annual Symposium, Amsterdam, the Netherlands, August 31 – September 3, 2004.
62. Kelly RI, Maegawa GHB, Leite JC, Kratz L. The Male with Conradi-Hünermann Syndrome (CDPX2): A Distinct Phenotype. 25th Annual DAVID W. SMITH Workshop on Malformations and Morphogenesis. Wasatch Mountains, Utah, USA. August 18-21, 2004.
63. Maegawa GHB, B. Lo, P. Satodia, H. Whyte, M. Sermer, J. Hutchinson, G. Wilson, G. Gross, G. Matyas, A. Hinek, D. Chitayat, S. Unger. Neonatal Marfan Syndrome: two cases with some unusual clinical findings. European Human Genetics Conference, Munich, Germany. June 12-14, 2004.
64. Maegawa GHB, Chitayat D, Whyte H, Thomas M, Kim P, Kim J, McNamara PJ. Bilateral microtia with absence of external auditory meati, Mondini type malformation, duodenal atresia, thyroid hemiplasia and biliary atresia. A new syndrome? Annual Clinical Genetics Meeting. Kissimmee, FL. March, 2004.

65. Ehlers JAC, Ghisolfi ES, Maegawa GHB, Zanardo AP, Strimitzer Im, Propiuk AS, Pereira ML, Carvalho T, JArdim LB, Lara DR. Impaired P50 gating in Machado-Joseph disease. 19th Brazilian Meeting of Clinical Neurophysiology and 28th Brazilian Epilepsy League Meeting. Rio de Janeiro, RJ, Brazil. October, 2003.
66. Maegawa GHB**,** Pires R, Farret L, Gurgel J, Goncalves S, Ferreira C, Rocha R, Silveira T, Holme E, Lindstedt S. Treatment of tyrosenima type I with N.T.B.C.. 15th Brazilian Congress in Clinical Genetics. Porto Alegre, Brazil. June 2003.
67. Disorders of cholesterol metabolism: report of two cases. Milan T, Maegawa G, Zandona D, Scholz AP, Burin M, Kratz L, Kelley RI, Metzenberg A, Leite JCL. 15th Brazilian Congress in Clinical Genetics. Porto Alegre, Brazil. June, 2003.
68. Giugliani R, Wajner M, Barschak A, Cecchin C, Zandon D, Ferreira G, Maegawa G, Jardim L, Goulart L, Sirtori LR, Chiochetta M, Deon M, Vargas CR. X-linked adrenoleukodystrophy : clinical and biochemical findings in Brazilian patients. 52nd Annual Meeting of the American Society of Human Genetics, Baltimore, MD. October 15th – 19th, 2002.
69. Vargas CR, Wajner M, Sirtori LR, Deon M, Chiochetta M, Goulart L, Sippel FK, Maegawa G, Jardim L, Giugliani R. Clinical and biochemical findings in 7 patients with X-linked adrenoleukodystrophy treated with Lorenzo`s oil. 40th Symposium of Society for the Studies of Inborn Errors of Metabolism (SSEIM). Dublin, Ireland. September 3rd-6th, 2002.
70. Sanseverino MT, Maegawa GHB, Magalhaes A, Leite JCL. Prenatal diagnostic of skeletal dysplasia in Medical Genetics Unit of Hospital de Clinicas de Porto Alegre. 21th Scientific Meeting of Hospital de Clinicas de Porto Alegre. Porto Alegre, Brazil. September, 2002.
71. Vargas CR, Sirtori L, Domingues GS, Goulart L, Luft AP, Barschak A, Pulrolnick V, Jardim L, Cecchin C, Maegawa GHB, Giugliani R. Biochemical effect of Lorenzo's oil intake and/or lovastatin in patients with X-linked adrenoleukodystrophy. 20th Scientific Meeting of Hospital de Clinicas de Porto Alegre. Porto Alegre, Brazil. September, 2002.
72. Zandona DI, Silveira I, Pereira ML, Ferro A, Alonso I, Moreira MC, Mendonça P, Ferreirinha F, Maegawa GHB, Cechin C, Sequeiros J, Jardim LB. Spino-cerebellar ataxias in southern of Brazil – up-date report: 81 familial cases with Machado.-Joseph Disase (SCA 3), SCA1, SCA 2, SCA 6, SCA 7, SCA 8 or unidentifiable mutations. 14th Brazilian Congress in Clinical Genetics. Ribeirao Preto, Brazil. May, 2002.
73. Zandoná DI, Maegawa GHB, Pires RF. Myxiploid diploid-tetraploid. A case report. 14th Brazilian Congress in Clinical Genetics. Ribeirao Preto, Brazil. May, 2002.
74. Maegawa GHB**,** Zandoná DI, Nonemacher K, Matte U, Leistner S, Felix TM. Clinical study of 153 cases of mental retardation who undergone to molecular analysis for *FRAXA* and *FRAXE* gene. 14th Brazilian Congress in Clinical Genetics. Ribeirao Preto, Brazil. May, 2002.
75. Sirtori RL, Domingues GS, Luft AP, Goulart LS, Barschak, Pulrolnik V, Jardim L, Cecchin C, Maegawa GHB, Wajner M, Giugliani R. Effect of Lorenzo' s oil in very long chain fatty acids of X-linked adrenoleukodystrophy patients. 13rd Scientific Meeting of Students of Universidade do Rio Grande do Sul. Porto Alegre, Brazil. March, 2002.
76. Domingues GS, Sirtori RL, Luft AP, Goulart LS, Barschak, Pulrolnik V, Jardim L, Cecchin C, Maegawa GHB, Wajner M, Giugliani R. Administration of glyceryl trioleate/glyceryl trierucate im X-linked adrenoleukodystrophy patients. 13rd Scientific Meeting of Students of Universidade do Rio Grande do Sul. Porto Alegre, Brazil. March, 2002.
77. Souza AHF, Maegawa GHB, Fraga JC, Knihs PR, Miura C, Takamato EE, Termignoni R. Acute supurative appendicitis in a 10-month-old child. Case report. 20th Scientific Meeting of Hospital de Clinicas de Porto Alegre. Porto Alegre, Brazil. June, 2001.
78. Leite JC, Maegawa GHB, Passos-Bueno, MR Beare-Stevenson Syndrome: a case report. 13rd Brazilian Congress of Clinical Genetics. São Pedro, Brazil, April, 2001.
79. Maegawa GHB, Werneck LC, Scola RH. Dystrophin project – molecular analysis of patinets with Duchenne and Becker muscular dystrophy. Annals of 7th Meeting of Students of the Universidade Federal do Paraná. Curitiba, Brazil. September, 1999.

80. Skudlarek DAC, Skudalarek CD, Toregiani JF, Maegawa GHB, Oliveira FC, Mukai MM, Lima LC. Pregnancy risk in adolescence women in outskurts of Curitiba. Annals of 23rd Congress of Interns of Hospital Nossa Senhora das Graças. Curitiba, Brazil. September, 1999.
81. Souza RLR, Maegawa GHB, Furtado L, Akel C, Castro RMV, Chautard Freire Maia, EA. Frequency of F2 mutation of human butyrylcholinesterase. Annals of 43rd Brazilian Congress of Genetics and 3th Meeting of Brazilian Society of Mutagenesis, Carcinogenesis and Enviromental Teratogenesis. Curitiba, Brazil. August, 1999.
82. Maegawa GHB, Scola RH, Raskin S, Werneck LC. Molecular analysis of 42 patinets with Duchenne and Becker muscular dystrophy. Annals of XI Scientific Congress of Hospital de Clínicas da UFPR Curitiba, Brazil. April 1999.
83. Pescador MVB, Zanchet ACB, Sandrini F, de Lacerda L, Maegawa GHB, Carreiro JE, Sandrini R. Report of 227 in-patients with diabetic cetoacidosis. Annals of 3rd Brazilian Congress of Pediatric Endocrinology and Metabolism. Curitiba, Brazil. April, 1999.
84. Werneck LC, Scola RH, Maegawa GHB, Werneck MCM. Duchenne and Becker muscular dystrophy. Molecular analysis. 1st Scientific meeting of Neurology department of Hospital de Clinicas de Curitiba Universidade Federal do Parana. Curitiba, Brazil. June 1999.
85. Scola RH, Werneck LC, Faoro LN, Maegawa GHB, Iwamoto FM. True neurogenic thoracic outlet syndrome. Report of two cases. Annals of 18th Brazilian Congress of Neurology. Curitiba. September, 1998.
86. Iwamoto FM, Maegawa GHB, Werneck LC, Scola RH. Inflammatory myopaties of infancy. A report of 24 cases. Annals of 6th Science Meeting of Students of Universidade Federal do Parana. Curitiba. April, 1998.
87. Souza RLC, Maegawa GHB, Furtado L, Akel C, Castro RMV, Chautard Freire Maia, EA. Frequency of F2 mutation of human butyrylcholinesterase in a random population sample from southern Brazil. The 6th International Meeting on Cholinesterase. La Jolla, CA, USA. March, 1998.
88. Souza RLR, Diniz ACP, Furtado L, Maegawa GHB, Pereira L, Freund AA, Culpi L, Chatard Freire Maia, EA. Frequency of the K mutation of Human Butyrycholinesterase in caucasians and blacks from Curitiba. *Braz J Hum Genetics* 1998;19(3) 243.
89. Souza RLR, Maegawa GHB, Furtado L, Akel C, de Castro RMV, Chautard Freire Maia, EA. Frequency of F2 mutation of Human Butyrylcholinesterase. *Braz J Hum Genetics* 1997;20(3) :316.
90. Werneck LC, Maegawa GHB, Scola RH, Iwamoto FM, Watanabe M. Clinical, Biochemical and electrophysiological study in 24 patients with infantil inflammatory myopathy. Annals of 15th Brazilian Congress in Clinical Neurophysiology and 22nd Meeting of Brazilian Society of Epilepsy. Curitiba, Brazil. November, 1997.
91. Maegawa GHB, Furtado L, Diniz ACP, Chautard Freire Maia, EA Studies in Human Butyrylcholinesterase. Annals of 4th Science Meeting of Students of Universidade Federal do Parana. Curitiba, Brazil. September, 1996.
92. Chautard Freire Maia, EA, Alcantara, VM, Souza RL, Rodrigues LCR, Furtado, L, Maegawa GHB, Diniz ACP. Association of genetic variability of Human Butyrylcholinestearase with height and weight. Annals of Parana Meeting of Genetics. Curitiba, Brazil. March, 1996.
93. Souza RLR, Maegawa GHB, Furtado L, Chautard Freire Maia, EA. Genetic Variability of Human Butyrylcholinesterase. Annals of Parana Meeting of Genetics. Curitiba, Brazil. March, 1996.

Panel board meetings, presentations and special lectures

1. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J(18) - NIH Study Section. February 27, 2020. Los Angeles, CA.
2. “Natural history of GM2 gangliosidosis" and "Development high throughput assays for neurological Lysosomal Disorders’. Two talks. Neurological Outcomes in Lysosomal Storage Disorders GRIDS 2019 Fairfax, Virginia on November 24th – 25th, 2019

3. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Meeting. Grant Panel Reviewer. TC - FDA, Silver Spring, MD. November 17, 2019.
4. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J(18) - NIH Study Section. October 24-25, 2018. Pentagon City, VA.
5. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Meeting. Grant Panel Reviewer. FDA, Silver Spring, MD. June 24, 2019.
6. Department of Defense Congressionally Directed Medical Research Programs (CDMRP). Peer-Reviewed Medical Research Program (PRMRP) Neuro Disorders Panel. Review Panel Member. June 7-12, 2019.
7. Department of Defense Congressionally Directed Medical Research Programs (CDMRP). Peer-Reviewed Medical Research Program (PRMRP) Frontotemporal Degeneration (FTD) panel. Review Panel Member. April 27-29, 2019
8. 2019 Global Leukodystrophy Initiative Conference Interdisciplinary Partnerships for Clinical Development. Krabbe disease. Introduction Talk and Chariing Session. Philadelphia. May 2-3 2019.
9. NATIONAL CENTER FOR ADVANCING TRANSLATIONAL SCIENCES ZTR1 RD-8 (01). NIH Study Section for Rare Disease Clinical Research. Panel member. February 11-22, 2019. Bethesda, MD.
10. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J(18) - NIH Study Section. Panel Member. February 21, 2019. Alexandria, VA.
11. Moderna Fabry Disease Advisory Board. Boston, MA. January 23rd, 2019.
12. Protalix Fabry Advisory Board Meeting. Dallas, TX. November 9 and 10, 2018.
13. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Meeting. Grant Panel Reviewer. FDA, Silver Spring, MD. November 26, 2018.
14. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J(18) - NIH Study Section. October 28, 2018. Alexandria, VA.
15. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J(18) - NIH Study Section. June 28, 2018. San Diego, CA.
16. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Meeting. Grant Panel Reviewer. FDA, Silver Spring, MD. June 1, 2018.
17. Clinical Applications of Metabolomics. Discussion Leader: Gustavo Maegawa, MD, PhD. CPSA Metabolomics. March 21st , 2018. Gainesville, FL.

18. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J(18) - NIH Study Section. February 22, 2018. New Orleans, LA.

19. Krabbe Disease. Leukodystrophy Symposium. 46th Child Neurology Society (CNS) Meeting. Kansas City, MO. October 4-7, 2017.

20. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J - NIH Study Section. October 26 and 27, 2017. Baltimore, MD.

21. Krabbe Disease. Leukodystrophy Symposium. 46th Child Neurology Society (CNS) Meeting. Kansas City, MO. October 4-7, 2017.

22. Pathogenesis Aspects of Leukodystrophies. Leukodystrophy Symposium. 46th Child Neurology Society (CNS) Meeting. Kansas City, MO. October 4-7, 2017.

23. Neurological Sciences and Disorders B. National Institute of Neurological Disorders and Stroke Initial Review Group. NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE. ZNS1 SRB-J - NIH Study Section – 323137. June 22 and 23, 2017. Chicago, IL.

24. Developing Small Molecule Therapies for Lysosomal Storage Diseases. Shanghai International Summit Forum on Pediatric Endocrine and Metabolic Diseases. Shanghai, China. June 3-6, 2017.

25. Nemours Pediatric Research Lectures 2017. Small Molecule as Therapeutic Approaches for Lysosomal Storage Diseases. April 3rd 2017. Nemours Biomedical Research/CPASS. Wilmington, DE.

26. Clinical Applications of Metabolomics and It's Role on Diagnosis and Therapeutic Response and Monitoring. Discussion Leader: Gustavo Maegawa, MD, PhD. CPSA Metabolomics. March 13-15th, 2017. Gainesville, FL.

27. Lysosomal Storage Disease Network (LDN) WORLD Symposium. Identification of psychosine-reducing small molecule agents for Krabbe disease. February 13-17, 2017. San Diego, CA.

28. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Meeting. Grant Panel Reviewer. FDA, Silver Spring, MD. December 6, 2016.

29. NIH Center for Scientific Review. Therapeutic Approaches to Genetic Diseases Study Section [TAG] Meeting. TAG Grant Review Panel Meeting. Grant Panel Reviewer. San Francisco, CA. October 27 and 28, 2016.

30. Updates and Emerging Therapies for Leukodystrophies. Krabbe Disease and Metachromatic Leukodystrophy (MLD). Speaker. 2016 United Leukodystrophy Foundation. Family Conference. July 27 – 30, 2016. Omaha, NE.

31. Cell-Based Assay of Psychosine Reduction. Speaker. 2016 Hunter Hope Foundation Medical Symposium. Speaker. July 25-27, 2016. Ellicottville, NY.

32. Southeastern Regional Genetics Group (SERGG) Annual Meeting. Panel Moderator. July 14-16, 2016. Ponte Vedra, FL.

33. NIH Center for Scientific Review. Therapeutic Approaches to Genetic Diseases Study Section [TAG] Meeting. TAG Grant Review Panel Meeting. Grant Panel Reviewer. Ritz Carlton Hotel in Pentagon City, VA. June 27 and 28, 2016.

34. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Meeting. Grant Panel Reviewer. FDA, Silver Spring, MD. June 3rd, 2016.
35. Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD). Intramural Reviewer Panel. June 1-2, 2016. NICHD Intramural Site Visit. NIH Clinical Research Center, Bethesda, MD.
36. Drug screening for Lysosomal storage disorders. Emerging and translational biology of leukodystrophies. Speaker. May 12-14th, 2016. Moser Center for Leukodystrophies, Kennedy Krieger Institute, Baltimore, MD.
37. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Meeting. Grant Panel Reviewer. FDA, Silver Spring, MD. December 8th, 2015.
38. Advisory Board Meeting on Lysosomal Acid Lipase Deficiency. Adviosry Board. Alexion Pharmaceuticals Inc. Boston, MA. October 15, 2015.
39. Fabry Disease: Place in Treatment of Current and Emerging Trends. Advisory Board Member. Genzyme Advisory Board Meeting. Baltimore, MD. October 10, 2015.
40. Therapeutic Development in Lysosomal Diseases. Department of Pathology, University of Florida, Gainesville, FL. October 7, 2015
41. Development of Small Molecule Therapies for Lysosomal Storage Diseases. Department of Neuroscience, University of Florida, Gainesville, October 1, 2015.
42. Late Onset Pompe Advisory Board Meeting. Wahsington, D.C.. Biomarin Pharm. Inc. Washington, DC. July 10th, 2015.
43. The Food and Drug Administration (FDA) Office of Orphan Products Development (OOPD). Endocrinology and Metabolism Grant Review Panel Member. FDA, Silver Spring, MD. May 29th, 2015.
44. Developing Small Molecules Therapeuties for Lysosomal Storage Diseases. Department of Pediatrics. University of Arizona College of Medicine. Tucson, AZ. May 11th 2015.
45. Developing Small Molecules Therapeutic Approaches to Lysosomal Storage Diseases. Clinical Translational Research Center. State University of New York at Buffalo. Buffalo, NY. May 5th 2015.
46. Small Molecule Therapeutic Strategies for Lysosomal Storage Diseases. Genetics Seminar, Division of Medical Genetics. Department of Pediatrics, University of Utah, Salt Lake City, UT. April 30th 2015.
47. Development of Therapies based on Small Molecules for Lysosomal Diseases. Molecular Medicine and Genetics Seminar, Division of Molecular Medicine and Genetics. Department of Internal Medicine. University of Michigan Medical School. Ann Arbor, MI. April 20th 2015.
48. NTSAD's Scientific Symposium & Workshop and 37th Annual Family Conference. Leader of workshop entitled 'Small molecule approaches that could be disease-modifying for our diseases. Are there opportunities in repurposing therapies? What is the path to translation?'. April 16-19, 2015, Reston, VA.

49. Developing Small Molecule Therapies for Lysosomal Storage Diseases. Genetics Seminar, Division of Genetics and Metabolism, Department of Pediatrics, University of Florida, Gainesville, FL. April 10th 2015.

50. Small Molecule Therapies for Lysosomal Storage Diseases. Research Seminar. Division of Clinical Pharmacology. Department of Medicine. Johns Hopkins University School of Medicine. Baltimore, MD. April 8th 2015.

51. Development of cell-based high-throughput screening assays for lysosomal diseases. Gordon Research Conferences – Lysosomal Diseases. March 15-20, 2015. Galveston, TX, US.

52. Small Molecule Therapeutic Strategies for Lysosomal Storage Diseases. Genetics Seminar, Division of Medical Genetics, Department of Pediatrics, Stanford University, Palo Alto, CA. February 10th 2015.

53. Gaucher Disease Prescribers Advisory Board. December 12th 2014. Tampa, FL.

54. Development of Small Molecule Therapeutic Strategies for Lysosomal Storage Diseases. Genetics Seminar, Division of Medical Genetics, Department of Medicine, University of Washington, WA. October 31st 2014.

55. Lysosomal storage diseases: clinical aspects and therapeutic challenges. Kennedy Krieger Institute Educational Lectures. Kennedy Krieger Institute, Baltimore, MD. July 2nd 2014.

56. Updates on small molecule therapy discovery for globoid-cell and metachromatic leukodystrophies. Scientific talk at United Leukodystrophy Meeting, July 31st 2014, Baltimore, MD.

57. Lysosomal storage diseases: molecular pathogenic mechanisms and therapeutic investigations. Human Genetics Graduate Course Lecture. Johns Hopkins School of Medicine. January 31, 2014.

58. Johns Hopkins Harriet Lane Case Conference – Acute Care Clinic. A case of 5-year old with recurrent abdominal pain. January 13rd 2014.

59. Food and Drug Administration (FDA) - Endocrinologic and Metabolic Drugs Advisory Committee Meeting, November 14, 2013.
    FDA Advisory Committee members met to attend presentations, discuss and vote the biologics license application (BLA) 125460, for Vimizim (elosulfase alfa), manufactured by BioMarin Pharmaceutical, Inc., for the treatment of Mucopolysaccharidosis Type IVA (Morquio A syndrome). Morquio A syndrome is a lysosomal storage disease caused by the deficiency an lysosomal enzyme, *N*-acetylgalactosamine-6 sulfatase, which is important metabolic pathway, leading to problems with bone development, growth and movement.
    Meeting location: Center for Drug Evaluation and Research Food and Drug Administration (FDA), Spring, MD 20993.

60. Eliglustat Advisory Board Meeting. Genzyme Center. Cambridge, MA. July 26, 2013.

61. Johns Hopkins Pediatric Case Conference. A case of mucolipidosis type I/II. April 5, 2013.

62. Best Practices in the Treatment of Slow Progressive MPS-VI. Scientific Advisory Board Meeting. Advisory Board Member. Participants: Clinicians and scientists from academina and pharma. Washington, DC. May 12, 2012.

63. Late onset Tay-Sachs Disease. Questions and Answers. National Tay-Sachs Disease and Allied Disorders Association. April 19, 2012. Orlando. FL. Dr. Gustavo Maegawa, MD, PhD. Patients and families.

64. Development of a Psychosine Assay to Identify Therapeutic Small Molecules for Krabbe Disease - Lysosomal Disease Network (LDN) 8th Annual WORLD Symposium. February 8-10, 2012, San Diego, California, USA. platform presentation.

65. Shire HGT North American Outcome Surveys HOS Investigator Meeting. San Diego, CA. February 7, 2012. Investigators of HOS Study.

66. Studies on Small Molecule Therapies for MLD and Krabbe Disease. United Leukodystrophy Foundation (ULF) Family Conference. July 15, 2011. Deblak, IL Patients and families.

67. High throughput screening assays for identification of small molecule therapies for lysosomal storage diseases. United Leukodystrophyy Foundation – Scientific Meeting, July 2011. Chicago, IL.

68. Small Molecule Therapies in Metachromatic Leukodystrophy and Krabbe Disease. United Leukodystrophy Foundation – Family and Patients Meeting. July 2011. Chicago, IL.

69. Gaucher Disease in Children. National Gaucher Foundation and Children's National Medical Center. Ronald Reagan Bldg. and International Trade Center. Washington DC. March 27 2011.

70. Developing Small Molecule Therapies for LSDs. Research Seminar presented in the Research & Development Department at BioMarin Pharmaceutical Inc. Novato, CA. March 1st 2011.

71. Lysosomal storage diseases: molecular pathophysiology and therapeutic investigations. Human Genetics Graduate Course Lecture. Johns Hopkins School of Medicine. November 18,2010.

72. High-throughput screening for the discovery of small molecules as therapeutic agents for lysosomal storage diseases. Molecular Genetics Laboratory Meeting. Johns Hopkins Hospital. November 8, 2010.

73. Cardiac Syndromes in Inherited Metabolic Dsieases – Pediatric Cardiology Review Course. Johns Hopkins Hospital Pediatric Cardiology Felowship Program. Johns Hopkins Hospital. Baltimore, MD. October 25, 2010.

74. High-throughput in vitro studies for drug discovery in Krabbe Disease. Inaugural "Sweet Baby Grace" Symposium on Krabbe Leukodystrophy. Kennedy Krieger Institute, Baltimore, MD. September 8, 2010.

75. Enzyme replacement therapy, mutant protein stabilization in Lysosomal Storage Diseases. Advanced topics in Human Genetics. Human Genetics Graduate Course lecture. Johns Hopkins University School of Medicine, Baltimore, MD. April 26, 2010.

76. Case of 13-year old boy with chronic hypertension and eye problems. Genetic perspective of nephronophthisis. Pediatric Case Conference. Children's Medical Center, Johns Hopkins Hospital, Baltimore, MD. April 23, 2010.

77. Identification and characterization of two small molecules as potential enzyme enhancement agents for two lysosomal storage diseases. National Institutes of Health – National Chemical Genomics Center (NIH/NCGC). Rockville, MD. December 14, 2009.

78. Juvenile GM-1/GM-2 Case Studies of Early and Later Onsets. Diagnosis, Management & Treatment of Progressive Neurological Disease from Infancy to Adult using Tay-Sachs Disease as a Model. CME conference for specialized clinicians and general practitioners. The Joseph B. Martin Conf. Ctr. Harvard Medical School, Boston, MA, US. September 21, 2009.

79. Late Onset Tay-Sachs Phenotypic Variations Among Siblings. Diagnosis, Management & Treatment of Progressive Neurological Disease from Infancy to Adult using Tay-Sachs Disease as a Model. CME conference for specialized clinicians and general practitioners. The Joseph B. Martin Conf. Ctr. Harvard Medical School, Boston, MA, US. September 21, 2009.

80. Cleaning house-disorders of intracellular protein processing. Centre for Brain & Behaviour Cross Talk Seminar. Division of Neurology, Department of Paediatrics, The Hospital for Sick Children, Toronto, ON, Canada. GM2 gangliosidosis May 29, 2008.

81. Lecture for residents and fellows from The Hospital for Sick Children (Toronto, ON, Canada) as Royal College of Physicians & Surgeons (RCPS(C)) exam preparation. Title: Mucopolysaccharidosis.: clinical and biochemical aspects. The Hospital for Sick Children, Toronto, ON, Canada. April 7, 2008.

82. Lecture for residents and fellows from The Hospital for Sick Children (Toronto, ON, Canada) as Royal College of Physicians & Surgeons (RCPS(C)) exam preparation. Title: Genetic syndromes: "gestald" diagnosis based on patients photos and brief clinical description. The Hospital for Sick Children, Toronto, ON, Canada. April 1, 2008.

83. Genetic Metabolic Rounds. Title: Treatment of Lysosomal Storage Diseases: juvenile GM2 gangliosidosis as a model. The Hospital for Sick Children, Toronto, ON, Canada. February 15, 2008.

84. Genetic Metabolic Rounds. Title: Studies for novel therapies in late-onset forms of GM2 gangliosidosis. The Hospital for Sick Children, Toronto, ON, Canada. April 11, 2007.

85. 28th National Tay-Sachs and Allied Diseases Annual Meeting. Substrate reduction therapy in juvenile GM2 gangliosidosis. Research Updates. Alexandria, Virginia, USA. April 6-9, 2006.

86. Genetic Grand Rounds. Title: Clinical, molecular and new therapeutic aspects in juvenile GM2 gangliosidosis. The Hospital for Sick Children, Toronto, ON, Canada. March 3, 2006.

87. Genetic Grand Rounds. Title: Natural History and Molecular Analysis of Juvenile GM2 gangliosidosis. The Hospital for Sick Children, Toronto, ON, Canada. December 2, 2004.

C. Medical certifications and examinations

United States Medical License Examination (USMLE) Step 3, July 2008

Diplomate, Brazilian Society of Clinical Genetics. Issued in June 2002

Education Commission for Foreign Medical Graduates (ECFMG) certificate. Issued in February 6, 2002

Clinical Skills Assessment (CSA), January 2002

United States Medical License Examination (USMLE) Step 2, December 1999

United States Medical License Examination (USMLE) Step 1, October 1998

D. Language skills and certifications

English, French and Portuguese (fluent); Spanish (basic)

Test of Language as Foreign Language (T.O.E.F.L.), 2005

Diplome D'études en Langue Française ' D.E.L.F. – 1er Degré, 1997

Diplome D'études en Langue Française ' D.E.L.F. – 2ème Degré, 1999

University of Cambridge - First Certificate in English Examination, 1991

The Oxford Examination of English as a Foreign Language, 1991