**PRAMOD KUMAR MISTRY, MD, PhD**

**CURRENT POSITION**

Professor of Medicine,

Professor of Pediatrics,

Professor of Cellular & Molecular Physiology,

Director: Yale Gaucher Center for Lysosomal Diseases

Yale School of Medicine

Attending Physician,

Yale New Haven Hospital: Klatskin Liver Service and Liver Transplant Service.

Correspondence:

Department of Internal Medicine

(Digestive Diseases)

333 Cedar Street/LMP 1080

P.O. Box 208019

New Haven, CT 06520-8019

Campus & Fed-Ex Address:

One Gilbert Street, TAC Building, Room S227B

New Haven, CT 06519

Tel: 203 785 3412

Fax: 203 785 7273

Email: [Pramod.mistry@yale.edu](mailto:Pramod.mistry@yale.edu)

**PERSONAL STATEMENT:**

I am Professor of Medicine (Digestive Diseases), Pediatrics and Cellular & Molecular Physiology, at Yale School of Medicine. I completed PhD/MB BS at University of London and concluded my initial training in Internal Medicine and Gastroenterology at London’s UCL/Royal Free Hospital School of Medicine and Addenbrooke’s Hospital, Cambridge. In 2001, I was recruited as Associate Professor of Medicine in the Section of Digestive Diseases at Yale School of Medicine. From the beginning, my career has been dedicated to clinical research, advocacy, and education in Gaucher disease. My research is focused on comprehensive delineation of Gaucher disease through phenotype annotation, genomic annotation, modifier gene discovery, biomarker discovery/validation and therapies. My lab developed the first authentic mouse model of Gaucher disease. The advances in understanding disease mechanisms in the Gaucher mouse model informed clinical research on biomarker validation, osteopenia, and therapy. I run a dedicated inherited metabolic liver disease clinic offering the entire range of therapies from small molecules, recombinant enzyme replacement, and liver transplant.

Families afflicted with rare inherited disease are severely underserved having limited access to knowledgeable expert physicians and therapies; it is not uncommon for patients to have prolonged diagnostic odysseys while they suffer from severe complications. For last 2 decades, my advocacy work in this area includes playing a key role in building national centers of excellence in Gaucher disease in UK and US as well as humanitarian treatment programs for children with Gaucher disease in Egypt, China and India. I am Chairman of the International Gaucher Disease Registry, the largest rare disease registry in the world. I am most proud of having trained large number of physicians throughout the world to become experts in this rare disease area to treat a highly underserved patient population.

For past 10 years, biomarker discovery and validation has been a major focus of our research efforts to improve care of patients affected with this complex disease. I was an invited speaker at FDA meeting convened by The Office of Translational Sciences on September 20, 2010 on biomarkers of Gaucher disease. I developed the theme for biomarker discovery based on FDA Critical Path Opportunities List to identify candidate biomarkers in our mouse models followed by functional studies and validation in patient populations. In 2010, we reported in the Proceedings of National Academy of Sci (USA) (Mistry PK, Liu J, Yang M, Nottoli T, et al,Glucocerebrosidase gene-deficient mouse recapitulates Gaucher disease displaying cellular and molecular dysregulation beyond the macrophage. Proc Natl Acad Sci U S A. 2010;107(45): p. 19473-8. PMID: 20962279) wherein we described how glucosylsphingosine could be utilized as a biomarker. Indeed, as requested of each speaker at the meeting to present a final slide of ‘biomarker wish-list’ I included circulating levels of glucocosylsphingosine. In fact, this meeting set up the collaboration between Aerts and our labs that resulted in paper one year later reporting the properties of glucosylsphingosine to function as highly meaningful biomarker of Gaucher disease (Dekker N, …Mistry PK, Boot RG, Aerts JM., Elevated plasma glucosylsphingosine in Gaucher disease: relation to phenotype, storage cell markers, and therapeutic response. Blood. 2011; 118(16): p. e118-27. PMID: 21868580). Over the years, we have built a solid body of evidence in definitive papers linking glucosylsphingosine and new biomarkers to disease severity, response to therapy and various pathological aspects of Gaucher disease.

I was recently recognized by Expertscape World expert in Sphingolipidosis: <https://twitter.com/Daily_Experts/status/1142079734312050689> (accessed 6/20/19)

**EDUCATION**

08/1972 – 06/1975 B.Sc. (Hons) Queen Elizabeth College, University of London. Physiology/ Biochemistry

08/1975 – 10/1978 PhD, St Thomas’s Hospital School of Medicine, University of London

0/8/1978 – 06/1983 MB BS Royal Free UCL School of Medicine

1987 Member of Royal College of Physicians (by examination)

– elected Fellow of the Royal College of Physicians 1999

2007 MA (*privatum)* Yale University

2015 M.D. University of the State of New York, MD conferral by the Boards of Regents

### **POST-DOCTORAL TRAINING**

08/1983 – 07/84 Internship: House Officer in Medicine Royal Free Hospital and

House Officer in Surgery, Dewsbury General Hospital

08/1984 – 07/1986 Residency: Rotating Senior House Officer in Medicine (Nephrology, Cardiology,

Infectious diseases and General Internal Medicine)

08/1986-12/1989 Chief Resident: Registrar in Hepatology, Gastroenterology, Oncology and Internal Medicine, Royal Free Hospital, London

01/1990-10/1994 Fellowship: Senior Registrar Internal Medicine, Metabolic Medicine and Hepatology, Addenbrooke’s Hospital, Cambridge

**ACADEMIC APPOINTMENTS**

University of Cambridge Clinical School, Cambridge

1989-1994 Clinical Lecturer in Medicine

Department of Medicine

Royal Free UCL School of Medicine, London

1994 – 1998 Senior Lecturer (Hon Consultant) in Medicine

Department of Medicine

Mount Sinai School of Medicine

1998 - 2001 Associate Professor,

Departments of Human Genetics and Medicine

Yale University School of Medicine

2001 – 2005 Associate Professor of Medicine (Section of Digestive Diseases)

2005 – 2013 Professor of Pediatrics and Medicine, Chief, Section of Pediatric Gastroenterology and Hepatology

07/2013 – present Professor of Medicine (Digestive diseases) and Pediatrics (Gastroenterology) and Cellular & Molecular Physiology.

# HOSPITAL APPOINTMENTS

Royal Free Hospital, London

01/1994-04/1998 Attending Physician, Department of Medicine

Medical Co-Director, Liver Transplant Program

Founding Director, NHS-funded Gaucher disease center

Mount Sinai School of Medicine

05/1998 – 10/2001 Attending Physician

Director, Comprehensive Gaucher Disease Treatment Center

Yale New Haven Hospital

11/2001-10/2005 Attending Physician (Internal Medicine/GI)

Medical Director of Liver Transplant Program

11/2005 – 06/2013 Yale New Haven Children’s Hospital

Attending Physician, Chief, Pediatric GI

Medical Director of Pediatric Liver Transplant Program

Director of Inherited Metabolic Liver disease clinic

07/2013 – present Yale New Haven Hospital

Attending Physician/Transplant Hepatologist (Hepatology)

**HONORS AND AWARDS**

1980 Foulke's Foundation Fellowship

1995 Alan Gordon Memorial Award of the Gaucher Association (UK)

1997/8 Founding Director, National Center of Excellence for Gaucher Disease, Royal

Free Hospital, London

1998 Elected Fellow of the Royal College of Physicians (London)

1999 Physician of the Year Award, Genetic Disease Foundation, New York

2006-08 Chairman of Medical Advisory Board, American Liver Foundation (Connecticut Chapter

2002 Max Millman Memorial Lecturer, Baystate Medical Center, Springfield, MA

2003 Visiting Professor, University of Manitoba Medical School, Winnipeg. Ca

2004 Visiting Professor, University of Connecticut GCRC

2005/2010 Nominated for Alice Bohmfalk Teaching Award, Yale University.

2005/2008 Award from National Gaucher Foundation for contributions to Gaucher disease

2007 MA (*privatim*) Yale University

2010 Fellow of the Royal Society of Medicine

2004 – Present Listed in The Best Doctors in America

2015-present Chairman of MAB of Project HOPE Gaucher Initiative for children in Egypt

2017 Elected Fellow of the American Association for Study of the Liver

2020 Nominated for Leonard Tow Humanism in Medicine Award

**BOARD CERTIFICATION**

1994 Certificate of Specialist Training in Gastroenterology, General Medical Council of UK (inactive)

1998 ECFMG

1999 American Board of Internal Medicine (active)

**LICENSURE**

Admitted to the General Medical Council of United Kingdom,

License # 2818678

State of New Jersey, License # 25MA07679000 (Inactive)

The University of the State of New York, Medical License # 214606

DEA # BM 5991422 (Active)

State of Connecticut, License # 039656 (Active)

# EDITORIAL RESPONSIBILITIES

2010 – 2017 Editorial Board, Hepatology

2004 – Present Editorial Board, World Journal of Gastroenterology

2006 – Present Editorial Board, Expert Reviews in Gastroenterology and Hepatology

2017 - Co-chief Editor, Advances in Gaucher Disease

2018 - Editorial Board, Expert Reviews in Molecular Medicine

**NATIONAL AND INTERNATIONAL COMMITTEE ASSIGNMENTS**

1998-Present Elected to the Medical Advisory Board, National Gaucher Foundation, USA

1998-Present Elected to Medical Advisory Board, Gaucher Association, UK

1998-Present Regional coordinator, International Gaucher registry.

1999 Member of NIH site visit panel Cincinnati Children’s Hospital GCRC

2000 Referee for Welcome Trust, UK.

2000 Referee for Cochrane Cystic Fibrosis and genetic disorders group

2004 Medical Advisory Board, Children’s Fund for Glycogen Storage Disease type 1a.

2004 Medical advisory Board, WISDOM, Wilson Disease Association of India.

2004 Moderator for 2004 AASLD oral presentations: Metabolic and Inherited Disease

2007 Course director, AALSLD/NASPGHAN symposium ‘Fibrocystic diseases of the liver’

2007 Chairman, American Liver Foundation Medical Advisory Board, Connecticut Chapter

2001 - Present Gaucher Generation Program Review Committee

2008 Course Director NASPGHAN AASLD symposium ‘Advances in Inherited Cholestasis Syndromes’

2008 – 2015 Abstract Review Committee, Chair, DDW and AASLD

2009 Chairman of Project Hope Humanitarian Program for Gaucher disease (on medical advisory board since 2001 and Chairman since 2009)

2010 Member, DSMB, NIDDK Pediatric Acute Liver Failure Study Group (PALF)

2011/12/17 Member of Review Panel FDA Office of Orphan Drug Development

2012 External Reviewer for AXA Research Fund, Paris.

2012 External Reviewer for Medical Research Council (UK) RFA on Stratified Medicine

2014 Grant Reviewer, Telethon Foundation, Italy.

2017 Reviewer for Israel Science Foundation

2017 External Reviewer, Vici programme ZonMw, The Netherlands

NIH Study Section Member:

2002 Ad Hoc Member ZDK1 GRB-C (J1)

2005 ZDK1 GRB-8(M8)

2005 Special Emphasis Panel ZDK1 GRB-4 M1

2007 Special Emphasis Panel ZDK1 GRB-8 M3

2009/2011 Special Emphasis Panel

Chairman:

2014 NIDDK Special Emphasis Panel

Childhood Liver Disease Research Network ZDK1 GRB-7 M1 R

ZTR1 RD-8 01 1, Rare Disease Clinical Research

Date(s) of review: February 11, 2019 - February 22, 2019

2012 and 2017 FDA Grant Review Committee

2013 – current Review Committee Yale Clinical Scholars Program of Yale’s NIH Clinical Translational Research Award (CTSA)

2014 External Reviewer for Italian Telethon Research Foundation

2015 Chairman, R01 ancillary study section panel for the ChilDREN network

2014 - 2017 Member of American Liver Foundation Grants Committee

2016 – current Member of TAG (Therapeutic Advances in Genetic Diseases) study section

2017 – current External Scientific Consultant to NDDK for ChiLDReN

2017 Netherlands Organization for Scientific Research, NWO.

2017 – 2020 Member, Awards Committee of AALSD

2019 NIH NCAT Grant Review Panel, Washington DC. 02.20.2019

2020 Reviewer for The Wellcome Trust/ DBT India

2020 Reviewer for FWF Austrian Science Fund Erwin Schroedinger Programme

2021 Member of Parkinson disease genes Expert Curation panel

2021 Chairman, MAB, Internal Wilson Disease Registry

2021 Member of the Executive Committee of Yale Liver Center

**PROFESSIONAL SOCIETIES**

1992-Present Member, European Working Group on Gaucher Disease

1998-Present Member, American Association of Human Genetics

1998-Present Member, American Association of Study of the Liver

2006 Member, North American Society of Pediatric Gastroenterology, Hepatology and Nutrition.

2010 Member of Society for Study of Inborn Errors of Metabolism

# NATIONAL/INTERNATIONAL ORGANIZATIONS

American Society for the Study of the Liver, AASLD

North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (NASPGHAN)

American Society of Human Genetics ASHG

Society of Inborn Errors of Metabolism SSIEM

Internal Liver Transplant Society ILTS.

**DEPARTMENTAL, UNIVERSITY and HOSPITAL SERVICE**

Yale Medical School

2006 - 2015 Medical School Selection Committee

2007 Search Committee for Chief of Solid Organ Transplantation

2011 Search committee for Chief of Pediatric Surgery

2011 Search committee for Medical Director of Pediatric Liver Transplant Program

2013 Search committee for Chief of Transplantation

2010 – Present Appointment & Promotions Committee (Term) of Yale School of Medicine

2006 - 2012 Appointment & Promotions Committee, Department of Pediatrics, YSM.

2013 – current Appointments & Promotions Committee, Department of Medicine

**EDUCATIONAL ACTIVITIES**

YSM Teaching Responsibilities

2001 – Present Clinical correlations (first year medical students) 2/8 sessions

2005 – Present Bench to Bedside seminars (first year medical students)

2001 – Present Digestive Disease Module (second year medical students)

2013 – Present First Year Physiology class

2001 - Present Didactic program teaching of adult and pediatric GI fellows

# MAJOR RESEARCH INTERESTS

# Clinical phenotyping and genomics of inherited metabolic liver diseases

# Gaucher disease: natural history, modifier genes, disease mechanisms and therapies

* Wilson disease: natural history, genetic heterogeneity and natural history
* Glycogen storage disease type 1a: determinants of hepatocellular adenocarcinoma
* Genetic syndromes leading to acute liver failure in children
* Genetic basis of biliary atresia

**ACTIVE GRANT AND CONTRACT SUPPORT**

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| **Active**  **ACTIVE**  Genzyme-Sanofi  Mistry (PI) 1/01/13 – 6/31/2022 $599,639 4.8 Calendar Months  Center of Excellence in Clinical Translational Research in Gaucher Disease Major aims are phenotype stratification, transcriptome analysis and hypothesis-driven research in International Gaucher Registry.  Sanofi-Aventis  Mistry (PI) 2/6/2015 – 2/5/2020 $65,217 .12 Calendar Months  Gaucher Generation Program Established Investigator Award: Studies to Delineate Specific Phenotypes of Gaucher disease and decipher respective biomarker signatures and genetic underpinnings  NIH NINDS  S. Chandra (PI)  Role: Co-Investigator 2/1/19 – 11/30/2023 $40,000 0.6 Calendar Months  Role of Glucosylsphingosine in Parkinson Disease and Dementia with Lewy Bodies: Goal of this study is to investigate role of acid ceramidase and GBA2 enzyme in glucosylsphingosine-mediated alpha synuclein aggregation using mouse models and patient-derived induced PSC cell lines  Genzyme-Sanofi  Mistry (PI) 2018-2021 0.6 Calendar Months  Multicenter phase 2 clinical trial of glucosylceramide synthase for type 3 Gaucher disease |

**RECENT EXTRAMURAL LECTURES (SELECTED)**

* 4/26/21: International Gaucher Alliance: Managing bone complications of Gaucher disease
* 4/11/20: South Asia Lysosomal Disease Annual Conference: Covid19 and Gaucher disease
* Lysosomal disease WORLD conference 2021 Speaker and Chairman, Rare Disease Registries: Pioneering Real World Evidence’
* 10/24/20: Spanish Gaucher Association ‘Covid19 and Gaucher disease’
* 10/28/20: Virtual seminar for hematologists in Thailand. Malaysia and Singapore “Gaucher disease beyond splenomegaly and thrombocytopenia’
* 10/5/20: European Working Group on Gaucher Disease: EWGGD Outcomes after 4.5 years of eliglustat treatment in ENGAGE trial.
* June 15, 2020 International Cerdelga Advisory Board, Virtual Meeting, Chairman and speaker.
* May 23, 2020: Internal Gaucher Alliance, Virtual Meeting, Speaker: Clinical Trials in rare disease.
* February 5-6, WORLD Lysosomal Meeting, Orlando, Speaker, Moderator.
* December 6, 2019: Cedars Sinai Medical Center, Los Angeles: Annual Lysosomal Disease Conference key note speaker: Role of glucosylsphingosine in Gaucher disease.
* November 15-16: LATAM Gaucher bone disease conference: Scientific Committee Member, Moderator, speaker.
* July 12th, 2019: Jekei University School of Medicine, Tokyo. Fifth International Forum on Gaucher Disease. Clinical and Biochemical advances in Gaucher disease.
* Invited speaker, GD/ASMD Emerging Leaders Program, Tokyo. 5.18.2019. Current and new biomarkers.
* Invited speaker, WORLD Symposium Sphingolipids- opening a black box, Orlando, 2.06.2019 ‘Role of sphingolipids in Gaucher disease pathophysiology’
* Keynote speaker, Indian Society of Inborn Errors of Metabolism, ISIEM, Pune, India. 01.19.2019. Gaucher disease – a model for phenotypic diversity and therapeutics
* Grand Rounds speaker, Mayo Clinic Phoenix 12.13.2018 ‘Understanding the link between Gaucher disease and myeloma’
* Invited speaker at Amsterdam Future of Biomarkers in Gaucher disease 9.22.18 “ Glucosylsphingosine, a key biomarker of Gaucher disease.
* Presidential plenary speaker, SSIEM, Athens August 31, 2018 ‘Cancer risk in inborn errors of metabolism’
* Grand Rounds, Maimonides Hospital, June 8, 2018
* Invited speaker and co-organizer, FDA minisymposium ‘ Sphingolipids and cancer: Lessons from Gaucher disease’, April 23, 2018
* South Asia Lysosomal Disease Conference: Key note lecture. New Delhi. April 21m, 2018
* Grand Rounds, Beth Israel Deaconess Medical Center, Department of Hematology and Oncology. November 15, 2017
* Interurban Clinical Club, 216th Meeting. Yale School of Medicine. Rare Gaucher disease informing common diseases. November 3rd, 2017.
* London Health Science Center, Ontario. GCS inhibition as treatment of Gaucher disease. September 13, 2017
* BC Children’s Hospital, university of British Columbia, Vancouver. Advances in Treatment of Gaucher disease. September 12, 2017.
* Gaucher Leadership Academy (organizer, co-moderator with Professor Timothy Cox. Eliglustat: real world experience. Mainz. June 2nd and 3rd.
* July 5, 2017: Yale Partnership for global Health Program. Invited speaker: Clinical Translational Research in Gaucher disease, relevance to common diseases.
* April 7 and 8, 2017: ILTS Alex Mowat Symposium, London: Invited Lecture: Can the need for liver transplantation be eliminated in 25 years’ time – gene therapy and small molecules in the management of liver disease
* November 19, 2016: 4th Gaucher disease forum: Tokyo Special Lecture

“Past, Present and Future of Gaucher Disease management”

Chair: Director of Advanced Clinical Research Center, Institute of Neurological Diseases

Emeritus Professor, The Jikei University School of Medicine, Yoshikatsu Eto

Speaker: Yale University School of Medicine, Professor, Pramod K. Mistry, MD, PhD, FRCP,

* October 14-15: Second Middle East North Africa Rare Disease Summit, Dubai.

Meeting Chair and Speaker for following talks:

Therapeutic advances in Gaucher disease and novel oral SRT

Developing and implementing regional guidelines for diagnosis and treatment of Gaucher disease around the world.

* September 24-25, 2016: New Delhi.

2nd South Asia LSD Symposium, 2016 “25 years of treatment with Enzyme replacement therapy: Learnings and New frontiers”

Plenary lecture: Gaucher Disease: Recent advances in Gaucher Disease

Debate: BMT in Gaucher disease - against

* 3rd Latin American Gaucher disease Conference, Buenos Aires. Argentina.

August 4-6, 2016

Lectures:

The lysosome: New pathophysiological perspectives and roll of glucocerebrosidase. What have we learned from stem cell models?

Bone disease markers: biomarkers for chronic inflammation

The role of substrate reduction therapy on the treatment of Gaucher patients

* Invited speaker at Cambridge – Yale Transplant Course, Yale club, NYC: NKT cells in liver immunity, October 2, 2015.
* Invited speaker, Boston Children’s Hospital, Division of Genetics and Genomics. June 16, 2015. Neuronopathic Gaucher disease.
* Invited speaker DDW May 19, 2015, Washington DC. Exome Analysis in Diagnosis and Management of Pediatric Liver Failure in Translations Symposium: Challenges and Opportunities in Pediatric Liver Diseases
* Invited speaker, moderator: LATAM Gaucher Conference, Rio de Janeiro. May 7,8, 2015
* Invited speaker, Nordic Gaucher Association 20th Anniversary Symposium, Stockholm, March 17th, 2015.
* Invited speaker Gaucher Leadership Forum, Berlin March 27 and 28, 2015: Novel substrate reduction therapy for Gaucher disease.
* Invited speaker to Lysosomal Disease Network CME Program ‘Advancing the Management of Gaucher disease’ February 11, 2015, Orlando.
* Key note speaker at Joel Barlow High School 2014 Science Symposium: Advice for a Young Scientist
* Invited speaker: Italian Gaucher Association 10th Anniversary Keynote Address, Genoa, October 13, 2014: Pathophysiologic Basis of phenotypic diversity in Gaucher disease and modifier genes.
* Invited speaker, 2nd International Bone Disease Conference in Gaucher disease, Seoul, S Korea. Pathophysiologic basis of skeletal manifestations in Gaucher disease: insights from mouse models.
* Invited speaker at Armed Medical College, Pune, India on Golden Jubilee of AFMC Department of Pediatrics Symposium, September 31, 2014 ‘Advances in Gaucher disease’
* Invited speaker to AECM Liver Center Seminar Series May 7, 2014: Rare disease Research in Gaucher disease – Beyond Macrophage Lipidosis.
* Invited speaker 11th European Working Group on Gaucher disease, Haifa, Israel. Role of GBA2 in Gaucher disease
* Asian Society of Inherited Metabolic Disease and Japanese Society of Inherited Metabolic Diseases Joint Meeting, Chiba, Japan Invited speaker “Future treatment of Gaucher disease. November 2013

15th Asia LSD Symposium, Chiba, Japan. Gaucher disease: Translating clinical and basic research into improved outcomes. November 2013

* European Gaucher Leadership Forum, Madrid, Member of Scientific Advisory Board, Speaker and Moderator, November 2013
* 1st International Conference on Bone Involvement in Gaucher disease, Rio de Janeiro: Chair, Invited speaker and Moderator July 2013
* Catholic University of Chile, Clinical Hospital, Santiago. Grand Rounds: Inherited Metabolic Liver Diseases Presenting with Liver Failure. 7/2/13
* Clinica Las Condes, Santiago. Grand Rounds: Metabolic Liver Diseases. 7/1/13
* 14th North American Lysosomal Disease Registries Meeting: Towards value-based medicine in treatment of lysosomal diseases. Chair, Speaker and Moderator. June 2013
* UCLA Genetics Symposium: Invited speaker March 2013
* Department of Molecular & Cellular Physiology, Yale School of Medicine: Invited research lecture 2012
* Gaucher Leadership Forum: Chair of Scientific Advisory Board, Speaker and Moderator, Munich, Germany, 2012
* International Symposium on Lysosomal Storage Diseases, London, Moderator, Speaker; Immunogenicity in Lysosomal Diseases, 2012
* Indian Association for the Study of the Liver, Mumbai. Invited lectures: Hemochromatosis; Fatty liver disease in children, 2012
* Royal Free UCL School of Medicine; Invited Research Talk: New Developments in Gaucher disease 2010
* Department of Medicine, Yale School of Medicine, Grand Rounds, 2010
* Primary Sclerosing Cholangitis Seeking a Cure Annual Meeting: Organizer and Speaker: Natural Course of PSC, Hartford, 2010
* **FDA Biomarker Qualification Workshop, Invited Speaker, 2010**
* Brigham and Women Hospital, Boston: Invited Research Talk: Syndromes due to GBA1 mutations, 2010
* Dean’s Workshop, Yale School of Medicine. Construction and characterization of genetically engineered mice. Invited Talk. 2010
* National Institutes of Health, Speaker, Parkinson disease in Gaucher disease, 2010
* National Gaucher Foundation Meeting, Invited speaker, 2010
* Value Based Medicine in Hepatology, University of Milano-Bicocca. Invited Talk on Value Based Medicine for inherited metabolic liver diseases, Milan. 2009
* Max Millman Symposium Speaker, Baystate Hospital, Springfiled, MA, 2009
* Latin American Symposium on Lysosomal Storage Diseases, Invited Research Talk, Cancum, Mexico. 2009
* European Society of Pediatric Gastroenterology and Hepatology Translational Medicine Symposium, London. Invited Speaker: How to create a KO mouse model; Small molecule and enzyme therapies.
* Invited speaker 1st and 2nd Gaucher Leadership Forum, Budapest and Milan. 2009 and 2010
* Rhode Island Hospital: Grand Round 2008
* Lysosomal Storage Diseases Club of Canada, Invited Research Talk, Vancouver, 2011
* Cleveland Clinic Grand Rounds, 2008
* CME University of Puerto Rico (with YSM), San Juan. Liver Transplantation for inherited Metabolic Liver Diseases, 2008
* Nordic Gaucher Association, Invited Talk. Norway. 2008
* NYU Medical Center, Grand Round. 2010

# STUDENTS, RESIDENTS, FELLOWS MENTORED (AT YALE)

|  | Training Period | Title of Research Project | Current Position of Past Trainees / Source of Support of Current Trainees |
| --- | --- | --- | --- |
| Mohsen Basiri | 2021 -2022 | Biomarkers and modifier genes in Gaucher disease | Lysosomal Disease Fellowship Training Award. |
| Praveena Narayanan | 2020 - 2023 | Immune response to lysolipids in liver disease | Fellow in Investigative Hepatology T32 program |
| Nathaniel Kleytman | 2020 - 2021 | Biomarker response to eliglustat | Medical student |
| Yuliya Afinogenova | 2016 - current | Biomarker validation in Gaucher disease | Fellow in Rheumatology, YSM, YNHH |
| Yumiko Taguchi | 2014-current | Mechanisms underlying GBA mutation associated Parkinson disease | PhD student, Department of Cell Biology, Yale University. Co-mentor |
| Silvia Vilarinho | 01/14 – present  (Primary Mentor K08;  Primary Mentor Doris Duke CSD Award) | Genomic analysis of biliary atresia and other childhood liver diseases | Yale Physician-Scientist Postdoctoral Program: Fellow  Assistant Professor |
| Vagishwari Murugeshan | 20117-2018 | Biomarker validation in Gaucher disease | Fellow, Rheumatology, Boston University Hospital |
| Sarah Lo  (YSM, Dept of Pediatrics) | 7/09-6/11 | Discovery of genetic modifiers of Gaucher disease/cancer phenotype | Assistant Professor of Pediatrics, Cornell Weil Medical College, NYC.  Investigative Hematology T32 Yale School of Medicine and Lysosomal Disease Network NIH Fellowship |
| Philip Stein  (YSM, Dept of Pediatrics) | 7/09-6/11 | Biomarker discovery and validation in Gaucher disease | Assistant Professor of Pediatrics, Director of Inflammatory Bowel Disease Program, St Christopher’s Hospital for Children.  Lysosomal disease Training Fellowship, Genzyme Corporation |
| Sarah Wardman  (YSM, Dept of Pediatrics) | 7/11-11/11 | Genetic basis of cancer in inborn errors of metabolism using whole exome sequencing | Visiting Scholar, King’s College Hospital, National Health Service, UK |
| Daniel Pearson  (YSM, Dept of Pediatrics) | 6/07-9/07 and 6/08-9/08 | Risk of Parkinson’s disease in Gaucher disease | MD/PhD program Harvard Medical School  Resident MGH |
| Gilberto Bultron  (YSM, Dept of Pediatrics) | 7/05-6/08 | Risk of Parkinson’s disease in Gaucher disease | Assistant Professor of Pediatrics, Loma Linda University School of Medicine |
| Sam Cheng  YSM, Pediatric GI Fellow | 7/09-6/12 | Role of calcium sensing receptor in inflammatory bowel disease | Assistant Professor, University of Florida, Department of Pediatric GI |
| Mohini Patel  YSM, pediatric GI Fellow | 7/11- 2013 | Biomarkers of IBD and exome capture sequencing in IBD/primary sclerosing cholangitis | Pediatric Gastroenterology, Morristown, NJ |
| Anthony Porto | 5/10- 2012 | Molecular genetic studies and enzyme therapy for lysosomal lipase deficiency | Associate Professor of Pediatrics (GI) and Vice chair. |
| Aileen Raizner (YSM, Dept of Pediatrics) | July 2010 - current | Annotating phenotypes in inborn errors of metabolism | Post-doctoral GI Trainee/T32 Investigative Hepatology Training Program, YSM  Assistant Professor, Children’s Hospital of Montefiore. |
| Tamar Taddei | July 2003- June 2006 | Natural history of N370S Gaucher disease | Associate Professor of Internal Medicine and Director of Liver Cancer Program, VA, West Haven, |
| Sohail Husain | 2004-2011 | Mechanisms of acute pancreatitis | Section Chief, Stanford Pediatric GI Section |
| Francis Chan | July 2005-July 2009 | Clinical spectrum of pulmonary vascular disease in Gaucher disease | Gastroenterologist, Connecticut Gastroenterology PC, New Haven |
| Uma Phatak | July 2007-June 2010 | Clinical and genetic spectrum of Wilson disease | Associate Professor of Pediatrics (GI), YSM |

PEER-REVIEWED PUBLICATIONS

1. Turner P, Miller N, Chrystie I, Coltart J, **Mistry P**, Nicoll A, Lewis B. Splanchnic production of discoidal plasma high-density lipoprotein in man. *Lancet*. 1979;1(8117):645-6. PMID: 85875
2. **Mistry P**, Miller NE, Laker M, Hazzard WR, Lewis B. Individual variation in the effects of dietary cholesterol on plasma lipoproteins and cellular cholesterol homeostasis in man. Studies of low density lipoprotein receptor activity and 3-hydroxy-3-methylglutaryl coenzyme A reductase activity in blood mononuclear cells. *J Clin Invest*. 1981;67(2):493-502. PMID: 6257763
3. Owen JS, Goodall H, **Mistry P**, Harry DS, Day RC, McIntyre N. Abnormal high-density lipoproteins from patients with liver disease regulate cholesterol metabolism in cultured human skin fibroblasts. *J Lipid Res*. 1984;25(9):919-31. PMID: 6491539
4. Desai K, **Mistry P**, Bagget C, Burroughs AK, Bellamy MF, Owen JS. Inhibition of platelet aggregation by abnormal high-density lipoprotein particles in plasma from patients with hepatic cirrhosis. *Lancet*. 1989 Apr 1;1(8640):693-5. PMID: 2564508
5. Mistry PK, Smith SJ, Ali M, Hatton CS, McIntyre N, Cox TM., Genetic diagnosis of Gaucher's disease. *Lancet*. 1992; 339(8798): p. 889-92. PMID: 1348297
6. **Mistry PK**, Davies S, Corfield A, Dixon AK, Cox TM., Successful treatment of bone marrow failure in Gaucher's disease with low-dose modified glucocerebrosidase. *Q J Med*, 1992. 83(303): p. 541-6. PMID: 1484931
7. Newman CM, Bruun BC, **Mistry PK**, Weissberg PL, Shanahan CM. High expression of osteopontin mRNA in human macrophages but not human vascular smooth muscle cells in culture. *Ann N Y Acad Sci*. 1995; 760:381-2. PMID: 7785923
8. Newman CM, Bruun BC, Porter KE, **Mistry PK**, Shanahan CM, Weissberg PL Osteopontin is not a marker for proliferating human vascular smooth muscle cells. *Arterioscler Thromb Vasc Biol*. 1995;15(11):2010-8. PMID: 7583583
9. Byrne CD, Wareham NJ, **Mistry PK**, Phillips DI, Martensz ND, Halsall D, Talmud PJ, Humphries SE, Hales CN. The association between free fatty acid concentrations and triglyceride-rich lipoproteins in the post-prandial state is altered by a common deletion polymorphism of the apo B signal peptide. *Atherosclerosis*. 1996;127(1):35-42. PMID: 9006802
10. **Mistry PK**, Wraight EP, Cox TM., Therapeutic delivery of proteins to macrophages: implications for treatment of Gaucher's disease. *Lancet*. 1996; 348(9041): p. 1555-9. PMID: 8950883
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