

CURRICULUM VITAE

Name: Pramod Kumar Mistry, M.D., Ph.D., FRCP

School Assignment: School of Medicine

Education:

1975 B Sc (Honors), Biochemistry & Physiology
Queen Elizabeth College, University of London.
1979 Ph D, St Thomas' Hospital Medical School, University of London
1983 MB BS
Royal Free Hospital School of Medicine, University of London
1987 MRCP, Royal College of Physicians, U.K.

Career:

1983-1984 House Officer in Medicine, Royal Free Hospital,
House Officer in Surgery, Dewsbury General Hospital
1984-1986 Rotating Senior House Officer in Medicine (Nephrology, Cardiology,
Infectious diseases and General Internal Medicine)
1986-1990 Registrar in Hepatology, Gastroenterology, Oncology and Internal
Medicine, Royal Free Hospital, London
1990-1994 Senior Registrar Internal Medicine, Metabolic Medicine and
Hepatology, Addenbrooke's Hospital, Cambridge
Clinical Lecturer in Medicine, University of Cambridge
Supervisor of MB-PhD Program, University of Cambridge
1994- 1998 Senior Lecturer in Medicine
Consultant Physician to the Hepatobiliary and Liver Transplant Unit
Royal Free Hospital, London
Director, Supra-Regional Gaucher Clinic, Royal Free Hospital
1998-2001 Associate Professor Human Genetics and Internal Medicine,
Mount Sinai School of Medicine, NYC
Director of Comprehensive Gaucher Disease Treatment Center,
Associate Attending Physician, Mount Sinai Medical Center, NYC
10/01 – 9/05 Associate Professor Internal Medicine (Digestive Disease Section),
Yale University School of Medicine, New Haven.
10/01-9/05 Associate Professor Internal Medicine (Digestive diseases),
Attending Physician, Yale-New Haven Hospital.
9/05– Pres Professor of Pediatrics, Chief, Section of
Pediatric Gastroenterology, Hepatology and Nutrition, Department
of Pediatrics, Yale University.

Medical Director, Pediatric Liver Transplant Program, Yale New Haven Hospital.
Director, National Gaucher Disease Treatment Center

Board Certification

1994 General Medical Council (UK) specialist certification in Internal Medicine and Gastroenterology
1997 ECFMG Certification Valid Indefinitely
1999 American Board of Internal Medicine Diplomate
2004 UNOS Certified Liver Transplant Physician

Licensure:

1983-present General Medical Council of United Kingdom
1998 University of The State of New York, Medical

2000 State of Connecticut
2003 State of New Jersey

Professional Honors or Recognition:

1980 Foulke's Foundation Fellowship to study medicine
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 Fellow of the Royal College of Physicians (London)
1999 Physician of the Year Award, Genetic Disease Foundation, NYC
2002 Max Millman Memorial Lecturer, Baystate Medical Center, Springfield, MA – 4/17/02.
2003 Visiting Professor, University of Manitoba Medical School, Winnipeg, Ca
2004 Visiting Professor, University of Connecticut General Clinical Research Center.
2004 Voted best grand round of the year by Department of Pediatrics, SUNY Downstate University Medical Center (4/7/04): Interventional Genetics in Lysosomal Diseases
2005 Named in Top Doctors of America
2005 National Gaucher Foundation (USA) award for contributions to Gaucher's disease
2005 Nominated for Bohmfalk Teaching Award in Clinical Science, Yale University School of Medicine.

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.

Lectures, Courses:

- 1994 International Liver Update, Royal Free Hospital: 'Liver transplantation for Inherited Metabolic Liver Diseases'.
- 1995 International Liver Update, Royal Free Hospital: 'Wilson's Disease'
- 1996 NIH Technology Assessment Conference on Gaucher Disease – current issues in Diagnosis and Treatment: In vivo distribution of mannose-terminated glucocerebrosidase in patients with type 1 Gaucher disease, P.K.Mistry and T. M. Cox. Published in JAMA, 1996, 275, 548-553.
- 1995 European Association for the study of the liver post-graduate course 'Gaucher Disease – a model for modern management of genetic disease'. Published J Hepatol, 1999, suppl 1, 30, 1-5.
- 1996 Medical Grand Round: Mount Sinai Hospital, NY, 'Diagnostic dilemmas in Wilson's Disease'.
- 1997 Fifth International Symposium on mucopolysaccharidosis and related disorders, Vienna, March 18-21, 1999: Enzyme replacement therapy for Gaucher disease.
- 1998 Fourth European Working Group on Gaucher Disease, September 6-9, 2000, Jerusalem: Pulmonary Hypertension in type 1 Gaucher disease.
- 1999 National Gaucher Foundation, USA : Gaucher disease in the new millenium, November 12-13, 2000, Arlington, VA. Pulmonary hypertension in type 1 Gaucher disease.
- 2002 Albert Einstein College of Medicine, Liver Research Center, October 25, 2000: 'The liver and cholelithiasis in Type 1 Gaucher disease'.
- 2002 National organization for Rare Diseases, Washington DC, 10/10/02. Changing spectrum of type 1 Gaucher disease.
- 2002 Society of inborn errors of metabolism, Dublin 9/3/02. A decade of enzyme replacement therapy for type 1 Gaucher disease.
- 2003 Australian Gaucher Treaters Annual Meeting, Sydney, 2003 – 2 presentations (5/3/03) 1. Natural History of Gaucher's disease – impact of genetic modifiers and enzyme therapy 2. Monitoring the response to enzyme therapy.
- 2003 Murdoch Institute, Melbourne, 5/6/03 Gaucher's disease

- Royal Brisbane Hospital, Hematology and Genetics Seminar
5/8/03
- Member International Faculty Indian Society of
Gastroenterology 2003 Meeting CME Program 'Treatment of
Inherited Metabolic Liver Diseases – Prospects for Gene
Therapy' 11/20/03
- Chennai Plenary speaker Indian Association for the Study of
the Liver, Chennai (11/21/03) 'Inherited Metabolic Liver
Diseases'
- 2004 University of Alberta, Medical Genetics Seminar: Modifiers of
Gaucher's phenotype (3/8/04)
- 2004 University of British Columbia, Vancouver General Hospital,
Department of Hematology Seminar: 'Hematologic aspects
of Gaucher disease' (3/9/04)
- Voted best grand round of the year by Department of
Pediatrics, SUNY Downstate University Medical Center
(4/7/04): Interventional Genetics in Lysosomal Diseases.
Annual Lysosomal Storage disease symposium Madrid:
Pulmonary hypertension in Gaucher disease' (4/21/04)
- Nordic Gaucher Disease Network, Copenhagen, Gaucher
disease – state of the art (11/5/04)
- Tenth Anniversary of enzyme therapy in France symposium:
Gaucher disease – therapeutic goals, Paris (12/16/04)
- Keynote speaker Latin America Symposium on Lysosomal
Diseases, Santiago 'Therapeutic goals for Gaucher's
disease' (8/7/04)
- Invited speaker 2004 (10/26/04) American Society of Human
Genetics Symposium, Toronto "Challenges of Lysosomal
Storage Disorders: A Unifying Approach to Unique
Diseases". My presentation: Biomarkers of Lysosomal
Storage Diseases.
- 2005 Second European symposium on
Lysosomal Storage Disorders, Athens 'Developing
therapeutic goals for Gaucher disease' (3/4/05)
- NYU Department of Gastroenterology seminar: Hepatic
aspects of Gaucher disease (3/29/05)
- World Organization for Research on Lysosomal Diseases, Phoenix,
Biomarkers of lysosomal diseases' (5/2/05).
- University of Mexico 7th Annual Pediatric and Neonatal Heme/Onc
Symposium, 8/20/05 'Gaucher disease; current management
model'
- Invited plenary speaker to Portuguese Society of Metabolic
diseases 3rd International Symposium, 9/24/05: Therapeutic Goals
in Gaucher Disease.

Invited speaker to Second International Gaucher disease Management Board meeting, Paris, 12/19/05: Effect of splenectomy in natural history of Gaucher disease'.

INTERNET BASED EDUCATIONAL MATERIALS:

1. National Gaucher Foundation web site <http://www.gaucherdisease.org> Lecture Therapeutic goals in Gaucher's disease
2. Web ex meeting 'The challenge of untreated Gaucher patient' 1/12/06 <http://www.gaucherdisease.org>

Professional Service:

- | | |
|-------|---|
| 1999- | Member, Medical Advisory Board, National Gaucher Foundation (USA) |
| 1999 | Member Medical Advisory Board, Gaucher Association (UK) |
| 1999 | Regional coordinator, Internal Gaucher Registry (ICGG) |
| 2001 | Member of Expert Committee, Project Hope Gaucher Initiative |
| 2000 | Member of Medical Advisory Board, American Liver Foundation (CT chapter) |
| 2000 | Member ad hoc NIH site visit team to General Clinical Research Center, Children's Hospital Research Foundation, Cincinnati. |
| 2002 | Ad Hoc Member NIDDK study section NIDDK ZDK1 GRB-C (J1) |
| 2002- | Preceptor, Post-doctoral T32 Yale program in Investigative Hepatology |
| 2004 | Member, Scientific Advisory Board, Children's Fund for Glycogen Storage Disease Research |
| 2004 | Associate Director, Molecular Bases of Disease module on protein folding diseases (#601), YSM |
| 2004 | Moderator for 2004 AASLD oral presentations session: Metabolic and Inherited Diseases |
| 2005 | Member, Intern Selection Committee, Yale School of Medicine |
| 2005 | Member, NIH NIDDK study section ZDK1 GRB-8(M8) |
| 2005 | Member, NIH NIDDK Special Emphasis Panel, ZDK1, GRB-4 M1 |

PUBLICATIONS

Original Research

1. Mistry P, Miller NE, Laker M, Hazzard WR and 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 in blood mononuclear cells. J Clin Invest 1981, 67,493-502.

2. 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, I, 693-695
3. Mistry PK, Smith SJ, Ali M, Hatton CSR, McIntyre N, Cox TM. Genetic diagnosis of Gaucher's disease. *Lancet* 1992, 339:889-892
4. Mistry PK, Davis S, Corfield A, Dixon AK, Cox TM. Successful treatment of bone marrow failure in Gaucher disease with low-dose modified glucocerebrosidase. *Quarterly J Med* 1992, 83(303): 541- 546
5. Newman CM, Bruun BC, Mistry PK, Weissberg PL and Shanahan CM. High expression of osteopontin mRNA in human macrophages but not human vascular smooth muscle cells in culture. *Ann NY Acad Sci* 1995, 760: 381-2.
6. Byrne CD, Wareham NJ, Mistry PK, Phillips DIW, Martinez ND, Halsall D, Talmud PJ, Humphries SE and 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: 35-42.
7. Mistry PK, Wraight EP and Cox TM. Therapeutic delivery of proteins to macrophages: implications for treatment of Gaucher's disease. *Lancet* 1996, 348:1555-1559.
8. Van't Hoff WG, Dixon M, Taylor J, Mistry P, Rolles KR, Rees L and Leonard JV. Combined liver-kidney transplantation in methylmalonic acidemia. *J Pediatrics* 1998, 132, 1043-1044.
9. Diaz, G, 12 authors and Mistry, P., Desnick, R.J. The origins of the Ashkenazi Jewish N370S and 84 GG acid beta glucosidase mutations. *Amer J Hum Genet*, 2000, 66, 1821-1832.
10. Gu, M., Cooper J.M., Butler, P., Walker A.P., Mistry, P.K., Dooley, J., Shapira A.V.H. Oxidative phosphorylation defects in Wilson's disease. *Lancet*, 2000, 356,469-473.
11. Charrow, J., Anderson, H.C., Kaplan P., Kolodny E.H., Mistry, P., Pastores G., Rosenbloom B., Scott, R., Wappner R., Weinreb, N., Zimran A. The Gaucher registry: demographics and disease characteristics of 1,698 patients with Gaucher disease. *Arch Int Med*, 2000, 160, 2835-43.
12. Butler P, McIntyre N, Mistry PK. Molecular diagnosis of Wilson Disease. *Molec Genet & Metab*, 2001, 72:223-30.
13. Weinreb NJ, Charrow J, Andersson HC, Kaplan P, Kolodny E, Mistry P, Pastores G, Rosenbloom B, Scott R, Wappner R, Zimran A (2002) Effectiveness of enzyme replacement therapy in 1,028 patients with type 1 Gaucher disease: A report from the Gaucher registry. *Amer J Med*, 113, 112-9.
14. Mistry PK, Sirrs S, Chan A, Pretzker M, Grace M, Duffy TP, Goldman M (2002). Pulmonary hypertension in type 1 Gaucher disease: genetic and epigenetic modifiers of phenotype and response to therapy. *Molecular Genetics and Metabolism*, 77, 91-98.
15. Sirrs S, Irving J, McCauley G, Gin K, Munt B, Pastores G and Mistry P (2002). Failure of echocardiography and cardiac catheterization to identify pulmonary hypertension in 2 patients with type 1 Gaucher's disease. *J Inherited Metab Dis*, 25,131-132.

16. Xu R, Mistry P, McKenna G, Emre S, Schiano T, Bu-Ghanim M, Levi G, Fiel MI. (2005) Hepatocellular carcinoma in type 1 Gaucher disease: a case report with review of the literature. *Seminars Liver Dis.* 25(2):226-9.
17. El-Beshlawy A, Ragab L, Yaoussry IY, Mansour IM and Mistry PK (2006) Skeletal response to enzyme replacement therapy in Egyptian children with Gaucher disease. *J Inher Met Diseases*, in press.

Reviews

1. Mistry PK and Cox TM. The glucocerebrosidase locus in Gaucher disease: molecular analysis of a lysosomal enzyme. *J Med Genetics* 1993, 30: 889-94
2. Mistry PK. Genotype/phenotype correlations in Gaucher's disease. *Lancet* 1995, 346: 982-3.
3. Mistry PK. Gaucher's disease: a model for modern management of a genetic disease. *J Hepatology*, 1999, suppl 1, 30, 1-5.
4. Mistry, PK. Treatment of Gaucher's disease with OGT 918. *Lancet.* 2000; 356: 676-7.
5. Charrow, J., Andersson HC, Kaplan P, Kolodny EH, Mistry P, Pastores G, Prakash-Cheng A, Rosenbloom BE, Scott CR, Wappner RS, Weinreb NJ, *Enzyme replacement therapy and monitoring for children with type 1 Gaucher disease: consensus recommendations.* *J Pediatr*, 2004. 144(1): p. 112-20.
6. Pastores GM, Weinreb NJ, Aerts H, Andria G, Cox TM, Giralto M, Grabowski GA, Mistry PK, Tylki-Szymanska A., *Therapeutic goals in the treatment of Gaucher disease.* *Seminar in Hematology*, 2004; 41(4 Suppl 5):4-14.
7. Weinreb NJ, Aggio MC, Andersson HC, Andria G, Charrow J, Clarke JT, Erikson A, Giraldo P, Goldblatt J, Hollak C, Ida H, Kaplan P, Kolodny EH, Mistry P, Pastores GM, Pires R, Prakesh-Cheng A, Rosenbloom BE, Scott CR, Sobreira E, Tylki-Szymanska A, Vellodi A, vom Dahl S, Wappner RS, Zimran A. *Gaucher disease type 1: revised recommendations on evaluations and monitoring for adult patients.* *Semin Hematol.* 2004 Oct; 41(4 Suppl 5):15-22.
8. Andersson HC, Charrow J, Kaplan P, Mistry P, Pastores GM, Prakesh-Cheng A, Rosenbloom BE, Scott CR, Wappner RS, Weinreb NJ; International Collaborative Gaucher Group U.S. Regional Coordinators. Individualization of long-term enzyme replacement therapy for Gaucher disease. *Genet Med.* 2005 Feb; 7(2):105-10.
9. Weinreb NJ, Barranger JA, Charrow J, Grabowski GA, Mankin HJ and Mistry PK. 2005, Guidance on the use of miglustat for treating patients with type 1 Gaucher disease. *Amer J Hematol.*, 80, 223-9
10. Mistry P. A. and Germain D.P., 2006. Therapeutic goals in Gaucher disease. *Revue de Medecine Interne*, 27, S34-S38.
11. Germain D.P. and Mistry P. 2006. Phenotype variations in Gaucher disease *Revue de Medecine Interne*, 27, S7-S10.

Book Chapters etc:

1. Mistry PK and Hoffbrand AVH. The liver in intracellular and extracellular lipidosis. Chapter 19.7 in *Oxford Textbook of Clinical Hepatology Second Edition 1999* Eds Bircher J, Benhamou J-P, McIntyre N, Rizzetto M and Rodes J.
2. Mistry PK and Abrahamov. A practical approach to diagnosis and management of Gaucher disease. In *Gaucher's Disease Ed A Zimran : Bailliere's Clinical Hematology* 1997.
3. McGovern MM and Mistry PK Lysosomal storage diseases In *Liver Disease in Children* EdS FJ Suchi, RJ Sokol, WF Balistreri. 2000. Lippincott Williams & Wilkins.
4. Schilsky M and Mistry P Inherited metabolic liver diseases in *Curr Opinion Gastro: Liver Diseases*, Ed: Moseley, R.H., Lippincott, Williams & Wilkins, 2001: 221-231.
6. Mistry PK and Zimran A: Clinical features and natural history of Gaucher disease in 'Gaucher disease' Eds Futerman and Zimran A, CRC Press (2006). In press
7. Mistry, P.K., Wilson disease for *Genetic Diseases of the Kidney*, Eds Lifton R, Somlo S, Giebisch G and Seldin D. Elsevier Inc (2005) – due March 2006.
8. Grabowski GA, Kolodny EH, Weinreb NJ, Rosenbloom BE, Prakash-Cheng A, Kaplan P, Charrow J, Pastores GM, Mistry PK. Gaucher disease: Phenotypic and Genetic Variation. *Metabolic and Molecular Basis of Inherited disease*, Eds Scriver CM, Beudet AL, Sly WS and Valle D, McGraw Hill (2006) in press.