EXHIBIT 1

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CURRICULUM VITAE

ROBERT LUKE NUSSBAUM

Address
150 Woodland Avenue
San Francisco, CA 94117

Business Phone
(415) 472-3200
(415) 476-1356 (FAX)
E-mail: nussbaumr@humgen.ucsf.edu

Current Academic Rank and Series
Professor of Medicine In Residence, UCSF 2006 to present
Holly Smith Distinguished Professor of Science and Medicine
Professor of Neurology, UCSF (Joint Appointment)
Member, UCSF Institute for Human Genetics

Department
Medicine

Administrative Title(s)
Chief, Division of Medical Genetics

EDUCATION

1967-1971	Harvard College cum laude	A.B. Applied Mathematics
1971-1975	Harvard Medical School & Harvard-MIT Program in Health Sciences and Technology.	M.D. Medicine
1975-1976	Intern in Internal Medicine, Barnes Hospital, Washington University, St. Louis, Mo.	
1976-1977	Junior Assistant Resident, Barnes Hospital, Washington University, St. Louis, Mo.	
1977-1978	Senior Assistant Resident, Barnes Hospital, Washington University, St. Louis, Mo. (Chief Resident, John Cochran VA Hospital, Wash University service)	
1978-1981	Fellow in Medical Genetics, Baylor College of Medicine, Houston, TX	
1981-1983	Associate, Howard Hughes Medical Institute, Baylor College of Medicine.	

LICENSES, CERTIFICATIONS

4/26/06	California Medical License C52306, exp. 2/28/2010 (active)

12/31/94	Maryland Medical License D46971, exp. 9/30/07 (Inactive)
12/3/76	Missouri Medical License (Inactive)
2/21/81	Texas Medical License (Inactive)
10/8/84	Pennsylvania Medical License (Inactive)
9/13/78	Certified, American Board of Internal Medicine #65308, 9/13/78
3/19/82	Certified, American Board of Medical Genetics in Clinical Genetics #1367
9/24/93	Certified American Board of Medical Genetics, Clinical Molecular Genetics #93220; Recertified, 8/15/2002
7/28/94	Basic Project Officer Course, DHHS

EMPLOYMENT

Principal Positions Held

1981-1984	Assistant Professor of Medicine, Baylor College of Medicine
1983-1984	Associate Investigator, Howard Hughes Medical Institute at Baylor
1984-1993	Associate Investigator, Howard Hughes Medical Institute at UPenn
1984-1989	Assistant Professor of Human Genetics, University of Pennsylvania, School of Medicine (Primary)
1989-1993	Associate Professor of Human Genetics University of Pennsylvania School of Medicine
1993-1994	Professor of Genetics, University of Pennsylvania School of Medicine
1993-2006	Chief, Genetic Disease Research Branch, NHGRI
1995-2006	Acting Chief, Inherited Disease Research Branch, NHGRI
2006-	Professor of Medicine and Chief, Division of Medical Genetics, Department of Medicine, UCSF
2007	Professor of Neurology (Secondary), UCSF

Ancillary Positions Held

1984-1989	Assistant Professor of Pediatrics, University of Pennsylvania, School of Medicine (Secondary)
1984-1993	Physician, Division of Human Genetics, Children's Hospital of Philadelphia
1989-1993	Associate Professor of Pediatrics, University of Pennsylvania School of Medicine
1991-1993	Associate Professor of Medicine, University of Pennsylvania School of Medicine
1991-1994	Director, Molecular Biology Graduate Group, University of Pennsylvania
1993-1994	Professor of Pediatrics, and Medicine, University of Pennsylvania School of Medicine
1994-2000	Adjunct Professor of Genetics, University of Pennsylvania School of Medicine
1994-2006	Clinical Staff, NIH Clinical Center
1996-2006	Project Officer, Center for Inherited Disease Research
2006-	Member, Executive Committee, UCSF Institute for Human Genetics
2006-	Faculty, Biomedical Sciences Graduate Group, UCSF

2006- Consultant in Genetics, UCSF Medical Center
2008- Director, Molecular Medicine Pathway to Discovery, UCSF School of Medicine

Clinical Training and Experience

1975-1976 Intern in Internal Medicine, Barnes Hospital, Washington University, St. Louis, Mo.
1976-1977 Junior Assistant Resident, Barnes Hospital, Washington University, St. Louis, Mo.
1977-1978 Senior Assistant Resident, Barnes Hospital, Washington University, St. Louis, Mo.
(Chief Resident, John Cochran VA Hospital, Wash University service)
1978-1981 Fellowship, Medical Genetics, Baylor College of Medicine, Houston, TX

HONORS AND AWARDS

1975 Alpha Omega Alpha
1977 Outstanding Medical Resident, Barnes Hospital
1978-80 Arthritis Foundation Research Fellowship
1986 Medical Research Award, Lowe's Syndrome Association
1987 Elected Member, American Society for Clinical Investigation
1989 Dean's Award for Excellence in Basic Science Teaching, University of Pennsylvania
1989 Master of Arts (honorary), University of Pennsylvania
1989 Member, Human Genome Organization
1990 Leonard Berwick Memorial Teaching Award, University of Pennsylvania
1991 Sir Clavering Fison Visiting Professor, Institute for Child Health, Hospital for Sick Children, Great Ormond Street, London, UK
1993 Medical Research Award, Lowe's Syndrome Association
1994 Morehouse School of Medicine Human Genome/Molecular Medicine Symposium Lecturer
1996 Medical Research Award, Lowe's Syndrome Association
1996 Elected Member, Association of American Physicians
1996 NHGRI Director's Award
1996 NIH Director's Award
1998 NHGRI Director's Award (Group award)
1999 G. Burroughs Mider Lectureship, National Institutes of Health
2000 Special Service Award, NIMH
2003 NHGRI Director's Award (Group Award)
2003 Forbes Lectureship, Medical College of Virginia
2004 Elected Member, Institute of Medicine of the National Academies of Science.
2004 NHGRI Director's Distinguished Service Award
2008 Legacy Award, Lowe's Syndrome Association, awarded in recognition of over two decades of service and commitment to patients and families with Lowe Syndrome.

PROFESSIONAL ACTIVITIES

Membership in Professional Organizations

1980- American Society of Human Genetics
1986- Society for Inherited Metabolic Disease
1989- Human Genome Organization
1991- American College of Medical Genetics
2003- American Society of Cell Biology
2006- Society for Neuroscience

Work for Professional Societies and Organizations

Completed

1988-1991 Co-Chair, Xq committee, International Human Gene Mapping Conferences HGM 9.5, (1988), 10 (1989), 10.5 (1990) and 11 (1991)
1990-1992 American Society of Human Genetics Annual Meeting Program Committee (Chairman of Program Committee, 1992 meeting)
1992-1996 American Society of Human Genetics Board of Directors
1998-2000 American Society of Human Genetics Awards Committee
1995, 1997 Gordon Conference on Molecular Genetics (Co-Chair 1995, Chair 1997)
1999-2002 Molecular Genetic Pathology Test Committee, American College of Pathology
1997-2000 American College of Medical Genetics, Board of Directors (Chairman of Program Committee, 1999 meeting)
2000-2006 Board of Scientific Overseers, The Jackson Laboratory
2004 President, American Society of Human Genetics
2005-2008 American Society of Human Genetics Awards Committee
2006 ACCE Guidelines Committee on Warfarin Dosing, American College of Medical Genetics
2006-2008 Association of Professors of Human and Medical Genetics representative for UCSF
2006-2008 Scientific Review Committee, the Parkinson's Study Group
2007 Personalized Health Care Expert Panel, Dept. of Health and Human Services

Ongoing

1992- March of Dimes/Birth Defects Foundation Clinical Advisory Committee
1995- Medical Advisory Board, The Lowe Syndrome Association
2006- External Advisory Committee, The Parkinson's Institute
2006- dbGaP Working Group, NCBI, NLM
2007- Member, Institute of Medicine Roundtable on Translating Genomic Based Research for Health.
2009-OMIM Scientific Advisory Board
2009-Scientific Advisory Board, Institute of Genetics, Canadian Institutes for Health Research.

Service to Professional Publications

1987-89	Editorial Board, American Journal of Human Genetics
1989-1993	Editorial Board, Cytogenetics and Cell Genetics
1992-1998	Communicating Editor, Human Mutation
1994-1996	Editorial Board, Human Genetics
1997-	Editorial Board, Journal of Clinical Investigation
1997-2003	Editor-in-Chief, Human Genetics (for Asia and Americas)
1998-	Editorial Board, Human Molecular Genetics, American Journal of Medical Genetics, Functional and Integrative Genomics, Journal of Clinical Investigation, Genome Medicine.
2006-	Section Editor, Genetics in Medicine

Ad hoc Reviewer for Nature, Nature Genetics, Nature Neuroscience, Neurology, Journal of Neuroscience, Neurogenetics, American Journal of Human Genetics, Molecular and Cellular Biology, Journal of Cell Biology

INVITED PAPERS, LECTURES, PRESENTATIONS (Since 2004)

2004

Columbia College of Physicians and Surgeons, New York, NY	Molecular Genetic Approach to Parkinson Disease
Fox-Chase Cancer Center, Philadelphia, PA	Cancer Genetics
University of Western Ontario, London, ON, Canada	Sodium-dependent vitamin c transporter
Thomas Jefferson University School of Medicine, Philadelphia, PA	Molecular Genetic Approach to Parkinson Disease
Bruce Rapaport Symposium, Maimonides Medical Center, Haifa, Israel	Human Genome Project
Jackson Laboratory 45th Annual Short Course in Medical and Mammalian Genetics (invited speaker).	Molecular Genetic Approach to Parkinson Disease
American Medical Writers' Association.	"Genetics 101" – Introduction to Human Genetics for medical writers
University of California Program in Human Genetics, San Francisco, CA	Molecular Genetic Approach to Parkinson Disease
Lowe Syndrome Trust International Meeting on Lowe Syndrome	Plenary Talk on Lowe Syndrome
University College London and Guy's and St. Thomas' Hospital Medical School	Molecular Genetic Approach to Parkinson Disease

2005

Astra-Zeneca Neurodegeneration meeting Scottsdale, AZ	Molecular Genetic Approach to Parkinson Disease

University of California, Irvine	Molecular Genetic Approach to Parkinson Disease
Children's Hospital of Philadelphia	Molecular Genetic Approach to Parkinson Disease
The Jackson Laboratory 46th Annual Short Course on Medical and Experimental Mammalian Genetics	Parkinson Disease
Gordon conference on Human Molecular Genetics and Genomics	Molecular Genetic Approach to Parkinson Disease
Brigham and Women's Hospital	Molecular Genetic Approach to Parkinson Disease
Harvard Medical School continuing Medical Education Course in Genetics	Keynote Address: Personalized Medicine
Brigham and Women's Hospital Medical Grand Rounds	Genetic Diseases, Mendelian and Complex
Cold Spring Harbor Laboratory Banbury Conference on mitochondria in neurodegeneration and aging	Role of alpha-synuclein in lipid metabolism

2006

Parkinson Disease and Diffuse Lewy Body Disease Symposium, World Parkinson;s Congress, Washington DC February 22, 2006	Molecular Genetics of Parkinson's Disease
World Parkinson's Disease Congress Washington, DC, February 23, 2006	Finding genes implicated in Parkinson Disease
The Parkinson's Institute Annual Retreat Asilomar, CA April 21, 2006	Biology of Alpha-Synuclein
Short Course in Medical and Mammalian Genetics, Bar Harbor, ME. July, 2006	Molecular Genetics of Parkinson's Disease
National Genetic Policy Summit Washington, D.C. September 20, 2006	Direct to Consumer Genetic testing
BMS Graduate Program Retreat Granlibakken, Lake Tahoe, CA, October 14, 2006	Studies on Vitamin C Transporter knock-out mice
President's Council on Bioethics November 16, 2006	Genetic Testing
UCSF Medical Grand Rounds December 7, 2006	Personalized Medicine

2007

Destination	Title
UCSF Department of Neurology Frontiers (Grand Rounds), February 7, 2007	Molecular Genetics of Parkinson's Disease
University of New Mexico Cancer Center, March 2007	Molecular Genetic Approach to Parkinson's Disease

Gladstone Institutes, May 2007	A Geneticist looks at Parkinson disease
San Francisco Veterans Administration, Medical Grand Rounds May 2007	Personalized Medicine
Alzheimer's Association Medical Scientific Advisory Council Research Symposium.	What Human Genetics has taught us about Alzheimer Disease
Stanford University, June 5, 2007	
Short Course in Medical and Mammalian Genetics, Bar Harbor, ME. July, 2007	1. Molecular Genetics of Parkinson's Disease 2. Individualized Medicine
GME Grand Rounds, UCSF, Sept. 1, 2007	Personalized Genetic Medicine
Italian Lowe Syndrome Association Formiggiana, Italy, Sept. 6-9, 2007	Lowe Syndrome
American Bar Association Continuing Education, South San Francisco, CA, October 2, 2007	Molecular Genetic Testing
Van Andel Institute, Grand Rapids, MI, October 31, 2007.	Molecular Genetic Approach to Parkinson Disease
Inositide Signalling, Janelia Farms (HHMI) November 4-7, 2007	Towards a mouse model for Lowe Syndrome
International Lowe Syndrome Meeting, London, UK, December 7, 2007	Towards a mouse model for Lowe Syndrome
NHGRI Workshop on Personalized Medicine, Bethesda, MD, December 19, 2007	Genetic Personalized Medicine
UCSF Research Retreat on Parkinson Disease	Phosphorylation of α-synuclein
2008	
Gallo Institute, Emeryville, CA, April 16, 2008	Molecular Genetic Approach to Parkinson Disease
Lowe Syndrome Association, Orlando, FL, June 26-28, 2008	1. Status of Lowe Syndrome Research 2. Molecular Genetics of Lowe Syndrome
Short Course in Medical and Mammalian Genetics, Bar Harbor, ME. July, 2008	Individualized Medicine
National Human Genome Research Institute, Bethesda, MD, September 2008	Genomics in Social and Behavioral Research
Michael J. Fox Foundation Therapeutics Conference, Chicago, IL, Sept., 2008	A progressive mouse model for Parkinson disease
Kaiser Office of Research, Oakland, CA, Sept., 2008	Pharmacogenomics and Personalized Medicine
University of California, San Diego, October 2008	A geneticist's view of Parkinson disease and α-synuclein
University of Texas Southwestern Dallas, TX, October 2008	A geneticist's view of Parkinson disease and α-synuclein

2009

San Francisco Veterans Hospital Medical Grand Rounds, February 2009	Personalized Medicine
American College of Medical Genetics, Invited Platform Presentation, March 2009	Creating a mouse model for Lowe syndrome
UCSF Pediatric Grand Rounds, April 2009	Personalized Medicine
Invited Keynote Speaker, Bowes Genetics Award, Harvard Partners, Boston, June 2009	Personalized Medicine: Where do we stand?
Short Course in Medical and Mammalian Genetics, Bar Harbor, ME. July, 2009	Individualized Medicine
Michael J. Fox Foundation Conference, New York, NY, September 2009	A progressive mouse model for Parkinson disease
American Society for Cell Biology Satellite Meeting on Cell Biology of Lowe Syndrome, San Diego, December 2009	Creating a mouse model for Lowe syndrome

ADMINISTRATIVE RESPONSIBILITIES AND SERVICE

Department or Institute

Chair, Committee on Appointments and Promotions, NHGRI/DIR

1990-2006	Executive Committee and Faculty, NHGRI/Johns Hopkins Genetic Counseling Training Program
1990-2006	Project Officer, Center for Inherited Disease Research
1998-2006	Medical Director NHGRI/Johns Hopkins Genetic Counseling Training Program

University or NIH Wide

1988-1994	Admissions Committee, Molecular Biology Graduate Group, University of Pennsylvania
1995-1999	Chair, Small Animal Users' Committee, Building 49
1995-2004	Head, Clinical Molecular Genetics Training Program, NHGRI
1996-2003	Executive Committee, Metropolitan Washington Medical Genetics Residency Training Program.
1999-2002	Committee on Appointments and Promotions, NIMH/DIR
2006-	Search Committee for Joint Pharmaceutical Sciences/IHG recruitment
2006-	Search Committee for Division Chief, Pediatric Genetics, Dept. of Pediatrics
2006-	Co-Chair, Search Committee for Psychiatric/Behavioral Genetics recruitment
2007-	Search Committee for Division Chief, Maternal-Fetal Medicine, Dept. of Ob/Gyn
2007-	Search Committee for Genetics in Clinical Pharmacy, School of Pharmacy
2007-	Executive Committee, Biomedical Sciences Graduate Group, UCSF

2007 Search Committee for Nephrology Division Chief, Department of Medicine.
2007- Search Committee for Pediatric Neurologist, Department of Neurology
2007- MSTP Council, UCSF
2007- Interviewer for internal medicine residency applicants, Department of Medicine, UCSF
2008- Director, Molecular Medicine Pathway to Discovery, UCSF School of Medicine
2008-09 Co-Director, Demystifying Medicine Lecture Series, UCSF
2008- Chair, Search Committee for Pediatric Genetics Faculty, Department of Pediatrics
2008- UCSF Chancellor's Council
2008- Search Committee for Statistical Geneticist, the Gallo Institute and Dept. of Neurology, UCSF.
2009- Search Committee for new Chairman of Pediatrics
2009- Search Committee for geneticists for the Cardiovascular Research Institute (Co-Chair)

Government Service

NIH Regular Initial Review Group Membership

1987-91 Genetic Basis of Disease, NIGMS.
1992-96 Human Genome Research Study Section (DRG)
1998-00 Mammalian Genetics (CSR).

Ad Hoc Review Committees

NIGMS
1986 Camden Cell Repository Advisory Committee
1988 RFA: Mapping and Sequencing Human and Representative Genomes
1987 RFA: Data Management of Mapping and Sequencing Information

National Institute of Child Health and Development
1987 RFA: Program Projects on Rett's Syndrome.

National Institute of Aging
1985 Special Study Section on Linkage Analysis in Alzheimer's Disease
2003 Site Visit, NIA DIR.

National Center for Human Genome Research
1989 RFA: SBA applications for Genome Analysis
1990 RFA: SBA applications for Genome Analysis
1990 Special Study Section, Division of Research Grants
1991 Site Visitor, Genome Center Application, Duke University
1994 Chair, Site visit, Genome Center Application, Washington University
2002 Haplotype Mapping RFA Review Committee
2004 Haplotype Mapping RFA Review Committee

1998 Model Organisms Database Workshop
2001 Chair, Subgroup on Genotyping Methods, SNP Mapping Advisory Committee
2000 Member, Advisory Panel on Genomics for Office of AIDS Research
2001 GRASPP panel on sequencing other whole genomes
2005- Working Group, National Chemical Genomics Center
2006- NCBI/NLM Working Group, dbGAP Implementation,

National Institute of Mental Health
1997 NIMH Genetics and Mental Disorders Planning Committee
1998 NIMH Intramural Research Planning Group on Genetics.

National Institute of Neurological Disorders and Stroke
1998 Neurogenetics Planning Group
2003 Udall Center Review
2006- Technical Contract Review for Pharmacological agents in Parkinson disease (CINAPS)

Community/Public Service

Grant Reviews for Private Foundations

2006	Michael J. Fox Foundation Fast-Track Grant Reviews
2006	Parkinson's Study Group Clinical Grant Applications Reviews
2007	Simons Foundation Reviews for grants in Autism Research

Education and Outreach

'96, '99, '03	Organized and taught courses for the public on genetics and genomics in the Smithsonian Associates Program, Washington DC.
2005	Advances in Parkinson Disease Genetics, NINDS-sponsored press conference
2003-2006	DNA Day Outreach Participant
2007	Judge, Biotech Educator Awards
2008	Wellness Lecture for the UCSF Development Office on Personalized Medicine and Genetics
2008	Numerous interviews with the lay press on Direct-to-Consumer Genetics testing
2008	Science Café Interview for UCSF Public Affairs

TEACHING and MENTORING

Courses taught

At the University of Pennsylvania

1986-1988	Genetics 100: Introduction to Human Genetics for Medical Students, University of Pennsylvania; I gave 6-8 hours of lecture
1989-1994	Genetics 100: Introduction to Human Genetics for Medical Students, University of Pennsylvania; I was Course Director and gave 8 hours of

	lectures to ~140 students and led 16 hours of small group discussions for ~20 students
1995-1999	Genetics 100: Introduction to Human Genetics for Medical Students, University of Pennsylvania, I gave 4 hours of lecture per year

At NHGRI

1999-2006	Introduction to Human Genetics for Genetic Counseling Students NIH/Johns Hopkins Joint Training Program in Genetic Counseling; I lead 32 hours of small group discussion for 4 or 5 students.
1998-2003	Introduction to Clinical Molecular Genetics; Course Director and I gave three or four hours of lecture for ~15 students.

At UCSF

2005-2006	Small group discussion leader for Genetics in Prologue, Brain and Behavior and Life Cycle blocks, UCSF (6 hours + 15 hours prep time)
2006-2007	Two lectures in Organ block, UCSF School of Medicine Medical School Curriculum (2 hours)
2006-2007	Small group discussion leader for Genetics in Prologue, Organ Block, Brain and Behavior and Life Cycle blocks, UCSF (8 hours + 20 hours prep time)
2007	Internal Medicine Resident Education Sessions on Genetics at Moffett-Long, SFGH and SFVA Hospitals (3 hours)
2007	Education Sessions on Genetics and Individualized Medicine for Fellows in the Division of General Internal Medicine (2 hours)
2007-2008	Two lectures in Organ block, UCSF School of Medicine Medical School Curriculum (2 hours)
2007-2008	Small group discussion leader for Genetics in Prologue, Brain and Behavior, Organs Block, Cancer, Life Cycle blocks, UCSF (10 hours + 15 hours prep time)
2007-2008	Genetics 224 in BMS, co-taught with Dr. Jane Gitschier, 22 hours
2007-2008	Supervise Journal Club presentations in BMS 255 Genetics course (4 hours)
2007-2008	Resident teaching through regular attendance at the weekly M&M Conference in the Department of Medicine (30 hours)
2007-2008	NS219, seminar for Neuroscience graduate students, 8 graduate students (2 hours + 2 hours prep time)
2008-2009	Supervise Teaching course for graduate students, 4 hours, 4 students
2008-2009	Lecture in CME Course in Electrophysiology (1 hour plus 3 hours prep time)
2007-2008	Three lectures in Organ block, UCSF School of Medicine Medical School Curriculum (2.5 hours)

2008-2009 Small group discussion leader for Genetics in Prologue, Brain and Behavior, Organs Block, Cancer, Life Cycle blocks, (10 hours + 15 hours prep time)

2008-2009 NS219, seminar for Neuroscience graduate students, 8 graduate students (2 hours + 2 hours prep time)

2009-2010 Resident teaching through regular attendance at the weekly M&M Conference in the Department of Medicine (30 hours)

Three lectures in Organ block, UCSF School of Medicine Medical School Curriculum (2.5 hours)

Small group discussion leader for Genetics in Prologue, Organs Block, M3 (6 hours + 9 hours prep time)

(Anticipated)

One lecture in Metabolism on Inborn Errors of Metabolism (2.5 hours)

Small group discussion leader for Genetics in Brain and Behavior, Life Cycle blocks, (4 hours + 6 hours prep time)

Genetics 224 in BMS, co-taught with Dr. Jane Gitschier, 22 hours

Introduction to the Laboratory for the Molecular Medicine Pathway (120 hours)

Predoctoral Students Supervised

1995(summer)	Laura Pomerance	Sidwell Friends School
1997(summer)	Rachel Perline	High School in St.Louis, Missouri
99-01	Sotiria Sotiriou	College, University of Edinburgh
05-07	Susan Bothwell	College, Lafayette
06	Adam Hoagland	College, Colorado College

Graduate Students Supervised

Years	Name	Graduate School
84-89	Dorothy Reilly	University of Pennsylvania
85-91	Diane Merry	University of Pennsylvania
91-96	Isabelle Olivos	University of Pennsylvania
92-95	Pasi Janne	MD/PhD University of Pennsylvania
89-92	Jeannie Lee	MD/PhD University of Pennsylvania
93-96	Sudha Srinivasan	University of Pennsylvania

01-05	Leslie (Whitty) Farber	George Washington University
03-	Sotiria Sotiriou	Johns Hopkins University
07	Karen Ring	Rotation in BMS, UCSF
09	Mike Andos	Rotation in BMS, UCSF

Postdoctoral Fellows Supervised

Yrs	Name	Current Position
85-88	Anne Maddalena	Scientist, GeneDx
87-90	Max Muenke	Chief, Medical Genetics Branch, NHGRI/NIH
86-90	Rhonda Schnur	Attending Genetics, Cooper Hospital, Camden, New Jersey
89-91	Alessandra Murgia	Professor Pediatrics, University of Padova, Italy
91-93	Anne-Marie Leahey	Asst. Professor of Oncology Children's Hospital of Philadelphia (Currently homemaker caring for children)
92-94	Rebecca Oakey	Lecturer in Genetics, Department of Medical & Molecular Genetics, Guy's King's & St Thomas' School of Medicine, London, UK
93-96	Christian Lavedan	Head of Discovery, Vanda Pharmaceuticals
93-97	Sharon Suchy	Staff Scientist, NHGRI
94-95	Ichiro Okabe	Dept of Pediatrics Haga Red Cross Hospital, Japan
95-00	Ti Lin	Staff Scientist, NIA
96-98	Marlene Dressman	Scientist, Vanda Pharmaceuticals
96-98	Roger Bascom	Research Fellow, University of Alberta
96-99	Lei Bi	Research Scientist, FDA
96-01	Elina Hellsten	Corning Sciences
99-01	Suzana Gispert	Staff Scientist, Goethe University, Frankfurt
02-05	Felicia Eason Forbes	Public School Science Teacher
99-06	Deborah Cabin	Assistant Professor, McLaughlin Research Institute, Great Fall, MT
99-06	Ornit Chiba-Falek	Assistant Professor, Duke University Medical School
99-06	Nelson Cole	Research Associate, NHLBI Division of Intramural Research

00-	Christopher Ellis	Current
04-	Melinda McFarland	Current
05-	Valerie Drews	Current

Medical Students supervised
2009 Samuel Brondfield
summer

BMS Prelim and Thesis Committees at UCSF

8/06 Sue-Ann Lee (Muchowski Lab) Prelim Committee and Thesis Committee
5/07 Beth Theusch (Gitschier Lab) (Chair of thesis Committee)
9/07 Angela Sia (Muchowski Lab) Prelim Committee
12/09 Ryan Devon (Ptacek lab) Thesis Committee

Teaching Awards

1989 Dean's Award for Excellence in Basic Science Teaching, University of Pennsylvania School of Medicine
1989 Commendation for Outstanding Teaching, University of Pennsylvania School of Medicine, Class of 1992
1990 Leonard Berwick Award for Excellence in Combining Basic Science and Clinical Teaching, University of Pennsylvania School of Medicine
1990 Commendation for Outstanding Teaching, University of Pennsylvania School of Medicine, Class of 1993
1991 Commendation for Outstanding Teaching, University of Pennsylvania School of Medicine, Class of 1994
2007 Certificate of Appreciation from the NHGRI for Teaching and Mentoring in the NIH/Johns Hopkins Genetic Counseling Training Program
2009 Excellence In Teaching Award, UCSF Academy of Medical Educators

CLINICAL RESPONSIBILITIES

In addition to my own research protocols, I served from 1994-2004 as a consultant in genetics in the NIH Clinical Center, serving either for two months or one month. I would see an average of one consult per week, usually an outpatient. Evaluation Scores are not generated at the NIH.

Currently on staff at Moffett-Long Hospitals. I provide Medical Genetics consultation for the Program in Cardiovascular Genetics Clinics for Marfan syndrome and primary cardiomyopathies (3 ½-day per month), the Mt. Zion Cancer Risk Clinic (2 ½-day /month), and the General Genetics Clinic in Pediatrics (1 ½-day per month). Total in-clinic time: 6 ½-days per month.

RESEARCH PROGRAM

Dr. Nussbaum's laboratory studies genetic contributions developmental and neurodegenerative disorders. His two major areas of concentration are Lowe syndrome and Parkinson disease. Lowe syndrome, formally known as Lowe oculocerebrorenal syndrome (OCRL), is a rare X-chromosome-linked disorder that can cause mental retardation, seizures, cataracts, and kidney disease in young children. Most Lowe syndrome patients die in their teens or twenties. Parkinson disease is a slowly progressive disease of the nervous system, which strikes an estimated 50,000 mostly older Americans each year. It is second only to Alzheimer's disease among the most common neurodegenerative diseases in the developed world.

In 1992, Dr. Nussbaum identified a defective gene that causes Lowe syndrome. The gene, *OCRL1*, codes for phosphatidylinositol-4, 5-bisphosphate 5-phosphatase-an enzyme that acts primarily in the Golgi apparatus of the cell and may be involved in protein processing and transport. Dr. Nussbaum's lab developed a clinically useful enzyme test for Lowe Syndrome, carried out the first prenatal diagnosis of the condition by enzyme assay, and pioneered the delivery of genetic services and counseling, including carrier testing, to families of Lowe syndrome patients. Determining the enzyme's normal function and why disabling it affects so many apparently unrelated organ systems could point to possible treatments. Interestingly, *OCRL1* knockout mice do not develop Lowe syndrome manifestations. Dr. Nussbaum is investigating the role of an autosomal paralog for *OCRL1*, *INPP5B,* as a gene encoding a possible compensating gene product in mice.

In his Parkinson disease work, Dr. Nussbaum's laboratory seeks to understand the range of genes that can contribute to this disorder. Scientists long believed that Parkinson was not an inheritable disease. In 1997, Dr. Nussbaum and collaborators within and outside NHGRI identified a missense mutation in the alpha-synuclein gene (*SNCA*) as the cause of hereditary, early onset Parkinson disease in an Italian-American family. In 2003, collaborating with researchers John Hardy and Andy Singleton at the National Institute on Aging, Dr. Nussbaum and his colleague Dr. Amalia Dutra helped the NIA researchers identify a triplication of the *SNCA* in one large family affected by early-onset Parkinson disease. When Lewy bodies- protein aggregates that are a defining characteristic of Parkinson disease- were found to be composed primarily of alpha-synuclein, the link between mutations in *SNCA* and Parkinson disease was clearly established and provide strong evidence that mutations that change alpha-synuclein's properties or cause it to be overexpressed may be involved in Parkinson disease pathogenesis. Finally, Dr. Nussbaum's laboratory has demonstrated a defect in phospholipid metabolism in mouse brains lacking alpha-synuclein. In particular, a deficiency of cardiolipin was seen, associated with a mild defect in mitochondrial electron transport chain function. The normal function of alpha-synuclein has been obscure and these results point to a definitive link between this protein and mitochondrial function, which has long been implicated in Parkinson disease.

Dr. Nussbaum's group has engineered several transgenic mice that express mutant human alpha-synuclein. Interestingly, mice that express only mutant human alpha-synuclein develop severe, rapid neurological deterioration, particularly in the spinal cord; mice that express both the mouse and the human protein develop mild deterioration. The investigators still do not know why and how the mouse protein protects against neurodegeneration.

CLINICAL RESEARCH PROGRAM

N/A

RESEARCH SUPPORT

2006-2008 Sandler Family Foundation (PI)
Total Direct Costs $200,000

2006-2008 Lowe Syndrome Trust(PI)
Total Direct Costs $176, 508

2006-2008 Michael J. Fox Foundation(PI)
Total Direct Costs $294,784

2009-2012 NIH/NIA 1R21AG033941-01(PI)
Total Direct Costs $450,000
"Glucocerebrosidase mutations in a mouse synucleinopathy model"

PATENTS

Cloning of a gene mutation for Parkinson's disease. United States Patent 7001720.

CONSULTING ACTIVITIES

Government

1997	NIMH Genetics and Mental Disorders Planning Committee
1998	NIMH Intramural Research Planning Group on Genetics
1998	NINDS Neurogenetics Planning Group
1998	Model Organisms Database Workshop
2000	Member, Advisory Panel on Genomics for Office of AIDS Research
2001	Chair, Subgroup on Genotyping Methods, SNP Mapping Advisory Committee
2001	GRASPP panel on sequencing other whole genomes, NHGRI
2002	Canadian Institutes for Health Research, Genetics Strategy meeting
2005-	Working Group, National Chemical Genomics Center, NHGRI
2007-	dbGaP Working Group, NCBI, NLM
2008	Site visit to review Dr. Eric Green, Scientific Director of NHGRI

Academia

2001	Site Visit to Review Department of Human Genetics, University of Michigan
2002	Consultant for Department of Genetics, University of North Carolina
2003-08	External Advisory Board, COBRE Program in Neuroscience, University of North Dakota

2004-2006	External Scientific Advisory Board, Carolina Center for Genome Sciences University of North Carolina
2007	Site Visit to Review Department of Genetics, University of Alabama, Birmingham
2007	Bioethics of Biobank Studies, Ontario Genome Center, Toronto, Canada
2008	California Technology Assessment Forum of Blue Cross-Blue Shield Foundation on utility of genotype-directed warfarin dosing

Private Industry
None

BIBLIOGRAPHY

Original Papers (peer reviewed only, meeting abstracts not included).

1. **Nussbaum RL**, and Puck JM: Recurrence risks for retinoblastoma: Model for autosomal dominant disorders with complex inheritance. J. Pediatr. Ophthalmology 13:89-98, 1976.

2. Caskey CT, **Nussbaum RL**, Cohan LC, and Pollack L: Sporadic occurrence of Duchenne Muscular Dystrophy: Evidence for new mutation. Clin. Genet. 18:329-341, 1980.

3. **Nussbaum RL**, and Caskey CT: Purification and characterization of hypoxanthine-guanine phosphoribosyltransferase from Saccharomyces cerevisiae. Biochemistry 20:4584-4590, 1981.

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