Curriculum Vitae

Donna M. Martin, MD, PhD
Associate Professor of Pediatrics
Associate Professor of Human Genetics
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Personal Data

Name:	Donna M. Martin, MD, PhD
Title:	Associate Professor of Pediatrics Associate Professor of Human Genetics
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Education and Training

8/83-5/87	Michigan Technological University, Houghton, Michigan, B.S. Mathematics/Foreign Language Certificate in German
6/87-6/88	Visiting Student, Universität Stuttgart, Deutscher Akademischer Austauschdienst
8/88-12/92	The University of Michigan, Ann Arbor, Michigan, Ph.D., Neuroscience
8/92-6/96	The University of Michigan Medical School, Ann Arbor, Michigan, M.D.
8/96-7/97	Intern in Pediatrics, Mott Children's Hospital, The University of Michigan, Ann Arbor, Michigan
8/97-6/99	Resident in Pediatrics, Mott Children's Hospital, The University of Michigan, Ann Arbor, Michigan
7/99-6/01	Medical Genetics Residency/Postdoctoral Research (mentor: Sally Camper), The University of Michigan, Ann Arbor, Michigan

Certification and Licensure

1994	United States Medical Licensing Exam Step 1-No. 40351538
1996	United States Medical Licensing Exam Step 2-No. 40351538
1997	United States Medical Licensing Exam Step 3-No. 40351538
10/19/99	American Board of Pediatrics- No. 067911, exp. 12/31/13
1999	State of Michigan Board of Medicine, No. 4301068270, exp 1/31/12
2000	State of Michigan Controlled Substance License, No. 4301068270, exp 1/31/12
9/01/02	American Board of Medical Genetics, Clinical Genetics, exp 12/31/2012
12/8/03	Drug Enforcement Administration Controlled Substance Registration Certificate, expiration 1/31/13
3/2/07	National Provider Identification Number 1477531028

Academic, Administrative, and Clinical Appointments

Academic Appointments

7/01—12/02	Lecturer in Pediatrics and Communicable Diseases, Division of Genetics, The University of Michigan School of Medicine
11/01—6/03	Research Investigator in The Department of Human Genetics, The University of Michigan Medical School
1/03--	Faculty Member of the Neuroscience Program, The University of Michigan

1/03—9/08 Assistant Professor of Pediatrics and Communicable Diseases, Division of Genetics, The University of Michigan Medical School
7/03—9/08 Assistant Professor of Human Genetics, The University of Michigan
9/08-- Associate Professor of Pediatrics and Communicable Diseases (with tenure) The University of Michigan
9/08-- Associate Professor of Human Genetics, The University of Michigan
7/08-- Faculty Member, Cellular and Molecular Biology Program, The University of Michigan

Administrative Appointments
11/06-- Director, Medical Genetics Residency Program, The University of Michigan

Clinical Appointments
07/01-- Staff Physician, Marquette General Hospital, Marquette, Michigan

Research Interests

Developmental brain disorders of mammalian neuronal function
Central and peripheral nervous system dysfunction in humans and mice

Current Projects
Pitx2 function in the mammalian central nervous system
Functional analysis of *Chd7*, encoding a chromodomain protein mutated in CHARGE syndrome
Genetic analysis of autism and disorders of neurodevelopment

Grants

Active

Principal Investigator, R01 NS054784-01A1
Pitx2 Function in Developing Mouse Brain Neurons
Feb. 1, 2007-January 31, 2011 ($1,285,255 total/$875,000 direct costs)-in no cost extension
This proposal aims to determine the effects of *Pitx2* deficiency on *Pitx2* lineage neuronal migration and axonal outgrowth, and to test the effects of *Pitx2* isoform specific deficiency on neuronal development.

Principal Investigator
NIH, 1R01DC009410-01A1
Developmental mechanisms of the chromodomain gene *Chd7*
12/1/2008-9/30/2013 ($1,851,345 total costs)
This grant aims to (1) characterize cellular proliferation, survival, and differentiation in Chd7 mutant mice, (2) determine the contributions of neural and otocyst derivatives to inner ear development, and (3) characterize cultured inner ear and neural stem cells from Chd7 mutant embryos and determine binding of CHD7 to target gene promoters.

ARRA Supplement to “Developmental Mechanisms of the Chromodomain Gene Chd7”
3R01DC009710-01A1S1, NIH/NIDCD
Martin (PI) 7/17/09–6/30/11 ($264,156 total costs)
Developmental mechanisms of the chromodomain gene *Chd7*

The equipment and personnel funded by this supplement to 1R01DC009410 were provided to accelerate the pace of experiments that fall within the scope of the parent grant.

NIH/NICHD K12HD28820 Castle (PI)
Child Health Research Career Development Award /Child Health Research Center
Advancing child health through cell and molecular biology
9/1/07–8/31/12 ($377,281 direct costs)
Goal: This grant provides support for junior investigators seeking research training in pediatrics.
Role: Recruitment Officer

Pending

March of Dimes
Analysis of CHD7, the Chromodomain Protein Mutated in CHARGE Syndrome, in GnRH Neurons
6/1/2012-5/31/2015 ($306,720 direct costs)
The goals of this grant are to generate and analyze mice with GnRH-specific loss of CHD7 and to characterize CHD7 binding sites in GnRH neurons.

NIH, R21
Retinoic acid signaling and chromodomain protein regulation of
inner ear morphogenesis
4/1/2012-3/31/2014 ($194,375.00 total costs)
The goals of this project are to identify novel mechanisms of retinoic acid and CHD7-mediated defects in the developing inner ear, using conditional Chd7 mutant mice, retinoic acid agonists and antagonists, and retinoic acid reporter mice.

Co-Investigator (PI: Matt State, Yale University)
Simons Simplex Collection Genetics Consortium
7/1/2011-6/30/2012 ($17,397 total costs)
Dr. Martin serves as the Genetics PI for the University of Michigan Simons Simplex collection. This subcontract is to cover effort for Dr. Martin's ongoing contributions to the genotype-phenotype analysis for the Simons Simplex Collection of 3000 individuals with autism spectrum disorders and their families.

Co-Investigator (PI: Raphael)
NIH, R01
Supporting Cells in Deaf Ears: A Substrate for Hearing Restoration Therapy
04/01/2012-03/31/2017 ($1,943,750.00 total costs)
The goals of this project are to characterize the mouse inner ear cellular substrates for potential trans-differentiation therapies using viral vectors. The emphasis in this project is on identifying novel gene expression patterns in supporting cells that are potentially receptive to induction of trans-differentiation.

Prior

1F31DC010955-01 Layman (PI)
NIH-NSRA

Role of CHD7 in neural development and maintenance
6/1/10–5/31/13 ($32,356 total costs)
The long term goal of this project is to determine the role of CHD7 in neural tissues to help improve diagnosis and treatment of olfactory and endocrine defects.
Role: Co-Investigator and Genetics Principal Investigator.

Co-investigator (PI: Lord)
Simons Simplex Family Collection Project
Sponsor: Simons Foundation
7/1/07 – 3/31/11 ($162,000 total costs)
The goals of this multi-center initiative are to collect DNA samples from families with one child affected by an autism spectrum disorder. My effort in the project is to assist with phenotypic characterization and genetic analyses.

Early Steps Study
National Institutes of Mental Health (NIMH)
University of Michigan/University of California-Davis
PI: Catherine Lord, PhD
8/1/08-8/31/2010 ($21,667)

"Microarray Capture and Rapid Sequencing to Identify the Genetic Cause of Martin-Probst Syndrome, an X-linked Disorder Affecting Hearing, Cognition, Craniofacial Development, Somatic Growth, and Aging"
The University of Michigan Center for Genetics in Health and Medicine
Endowment for the Basic Sciences
PI: Martin
2/1/2009-7/31/2010 ($40,000)

Feinberg Foundation Visiting Faculty Program
December 2009-January 2010
PI: Martin
Host: Orly Reiner, Weizmann Institute, Rehovot, Israel
This award covered travel and expenses for a 6 week visiting professorship at the Weizmann Institute in Rehovot, Israel

Clinical Translational Science Award (CTSA)
The University of Michigan
Microarray Capture and Rapid Sequencing to Find Genetic Causes of Autism.
PI: Martin
October 31, 08-November 1, 2009 ($35,000)

Amendt-Heller Pediatric Research Award
Mechanisms of chromodomain protein function in mammalian olfaction
The University of Michigan
PI: Martin
January 1, 2008-December 31, 2008 ($20,000)

Principal Investigator, The National Organization for Hearing Research Foundation
Analysis of Hearing and Ear Anomalies in *Chd7* Mutant Mice
Awarded 1/27/06 ($20,000)
This grant provided supplies and technical support for studying the role of Chd7, a chromodomain gene, in development and function of the mouse inner ear.

Principal Investigator, Pitx2 Function in Developing Mouse Brain Neurons
Biomedical Research Council Research Award
The University of Michigan ($20,000)
August 1, 2006-Dec. 31, 2006

Principal Investigator, Pitx2 Function in Developing Mouse Brain Neurons
Office of the Vice President for Research
The University of Michigan ($10,000)
August 1, 2006-Dec. 31, 2006

Principal Investigator, KO8HD40288, The Role of *Pitx2* in the Mammalian Central Nervous System
National Institute of Child Health and Human Development
Partial salary support and supplies
July 1, 2001-June 30, 2006 ($500,000)

Principal Investigator, The Janette Ferrantino Award
Analysis of *Pitx2*-dependent neuronal differentiation in mature neurons and embryonic stem cells
Department of Pediatrics, The University of Michigan Medical School
July 1, 2003 ($30,000)

Faculty Sponsor, Rackham Interdisciplinary Workshop
Experimental Approaches to Understanding Vertebrate Neurogenesis
Rackham School of Graduate Studies, The University of Michigan
October 30, 2003-June 14, 2008 ($6,000 annually)

Principal Investigator
Child Health Research Center,
The Role of *Pitx2* in the Mammalian Central Nervous System
National Institute of Child Health and Human Development
Concurrent support for supplies
Sept 1, 2001-August 1, 2004 ($90,000)

Honors and Awards

1983	Valedictorian, Wayland High School, Wayland Michigan
1983	Board of Control Scholar, Michigan Technological University
1984	Phi Eta Kappa, Michigan Technological University
1987	Magna cum Laude, Michigan Technological University
1987	Fellowship for Summer Research, Universität Saarbrucken, Deutscher Akademischer Austausch Dienst (DAAD)
1987–1988	Fellowship for Study abroad, Universität Stuttgart, DAAD

1989	University of Michigan Rackham Graduate School Fellowship to attend Learning and Memory, Cold Spring Harbor Laboratory, New York (Eric Kandel and John Byrne, course organizers)
1990	NIH National Research Service Award (Neuroscience Program)
1991–1992	NIH National Research Service Award (Neurology Department)
1999	Scholarship to attend the 13th Annual International Mouse Genome Conference annual meeting, Philadelphia, Pennsylvania
2003	Janette Ferrantino Award, Department of Pediatrics, The University of Michigan
2004	Top Resident Teacher Award, Department of Pediatrics, University of Michigan
2007	The University of Michigan Hartwell Foundation nominee
2007	The University of Michigan John Merck Scholar nominee
2007	Best Doctors in America (*Detroit Hour* Magazine)
2011	Elected Member, The American Society for Clinical Investigation (ASCI)
2010	Top Resident Teacher Award, Department of Pediatrics, University of Michigan

Grant Support/Honors Received for Trainees

2001	University of Michigan Undergraduate Research Opportunity Program Summer Biomedical Research Award (Steven T. Philips)
2003	Parkinson's Disease Foundation Summer Research Program (Steven T. Philips)
2002	University of Michigan Undergraduate Research Opportunity Program Summer Biomedical Research Award (Hemanth Ramaprakash)
2006	Summer Research Opportunity Program, The University of Michigan (Patrice Capers, Spelman College)
2006	Research Award, Annual Biomedical Research Conference for Minority Students (ABRCMS) in Anaheim, California (Patrice Capers, Spelman College)
2007	University of Michigan Undergraduate Research Opportunity Program Summer Biomedical Research Award (Katherine Cheng)
2007	Summer Research Opportunity Program, The University of Michigan (Erin Mortgart, Mount Union College, Alliance, Ohio)
2008	Graduate Student Award, 19th Annual Pediatric Research Symposium (Kaia Skaggs)
2008	Research Staff Award, 19th Annual Pediatric Research Symposium (Liz Hurd)
2009	Research Staff Award, 20th Annual Pediatric Research Symposium (Jennifer Skidmore)
2009-2010	Hearing, Balance, and Chemical Senses Training Grant, Wanda Layman (4th year Human Genetics student)
2010-2013	Individual NRSA award, "Role of CHD7 in neural development and maintenance", Wanda Layman (NICHD)
2010-2011	Regents Fellowship for Cell and Molecular Biology, Mindy Waite (4th year CMB grad student)
2010	Graduate Student Award, 21st Annual Pediatric Research Symposium (Jillian Lee Wiggins, 4th year Developmental Psychology Student)
2011-2012	Rackham Predoctoral Fellowship, Mindy Waite (5th year CMB grad student)

2011-2012 Hearing, Balance, and Chemical Senses T32 Training Grant, Joe Micucci 3rd year Biochemistry student)

Memberships and Offices in Professional Societies

1988-- Member, Society for Neuroscience (SFN)
1999-- Fellow, American Academy of Pediatrics (AAP)
1999-- Member, American Society for Human Genetics (ASHG)
1999-- Member, Association for Research in Otolaryngology (ARO)
2003 Member, American Society for Gene Therapy (ASGT)
2003--2005 Member, American Society for Cell Biology (ASCB)
2003-- Member, American Medical Association (AMA)
2003-- Fellow, American College of Medical Genetics (ACMG)
2005-2008 Member, International Mammalian Genome Society (IMGS)
2007 Member, The Endocrine Society
2008-- Member, The International Society for Developmental Neuroscience (ISDN)
2009-- Member, International Society for Autism Research (INSAR)
2011 Elected Member, The American Society for Clinical Investigation (ASCI)
2011 Member, The Association of Professors of Human and Medical Genetics

Editorial Positions, Boards, and Peer-Review Service

Editorial Positions
Review Editor, Editorial Board of Frontiers in Craniofacial Biology

Peer Reviewer:
American Journal of Human Genetics
American Journal of Medical Genetics
American Journal of Pathology
Autism Research
BMC Medical Genetics
Developmental Dynamics
Developmental Psychology
Endocrine Reviews
Experimental Neurology
FASEB Journal
Genesis
Hearing Research
Human Molecular Genetics
Indian Journal of Medical Research
International Journal of Medical Sciences
Investigative Ophthalmology & Visual Sciences
Journal of Cellular Cytogenetics
Journal of Clinical Investigation
Journal of Medical Genetics
Journal of Neuroscience
Molecular and Cellular Biology

Molecular Biology and Evolution
Molecular Endocrinology
Molecular Genetics and Metabolism
Mammalian Genome
Neurobiology of Disease
Neuroscience
PLoS Genetics

Study Sections/Grant Review Committees

1999	National Science Foundation
2004	Neurodegenerative Disorders, State of Pennsylvania Department of Health Merit Review
2005	*Ad hoc member*, Developmental Brain Disorders study section (DBD), National Institutes of Health, March 3-4, 2005
2006, 07	Proposals in Neuroscience, State of Pennsylvania Department of Health Performance Review, Oak Ridge Associated Universities, Oak Ridge Tennessee
2007-2010	Office of the Vice President for Research (OVPR) bridging funds reviewer
2007	Ad hoc member, Neurogenesis and Cell Fate Study Section (NCF), National Institutes of Health, June 14-15, 2007
2007	*Ad hoc member*, Brain Disorders and Clinical Neuroscience (BDCN) Member Conflict Special Emphasis Panel, National Institutes of Health, July 9, 2007
2008	*Ad hoc member*, Special Emphasis Panel/Scientific Review Group 2008/05 ZRG1 MDCN-J (02), National Institutes of Health, April 3, 2008
2008	Scientific Review Board, International Rett Syndrome Foundation
2008	Oak Ridge Associate Universities, Pennsylvania Department of Health, Collaborative Projects
2009	*Ad hoc member*, Neural Development, Plasticity and Repair Study Section (NDPR), National Institutes of Health, San Fransisco, Feb. 12-13, 2009
2009-2012	Scientific Review Board, International Rett Syndrome Foundation (3-year term)
2009	UM NIEHS P30 Core Center Pilot Project Program reviewer
2009	Medical Research Council Career Development Award reviewer
2009	*Ad hoc* member, Developmental Brain Disorders (DBD) NIH study section, June 8-9, 2009
2009	*Ad hoc* member, NIH BDCN-T Challenge Grant Panel 11
2009-2015	Permanent member, Developmental Brain Disorders (DBD) NIH study Section
2010	*Ad hoc* member, ZRG1 F09-E (20) L Oncological Sciences NIH study Section
2011	*Ad hoc* grant reviewer, Israel Science Foundation
2011	*Ad hoc* grant reviewer, Medical Research Council, Great Britain
2011	*Ad hoc* grant reviewer, Wellcome Trust, Great Britain
2011	*Ad hoc* grant reviewer, Italian Telethon Foundation

Teaching

Training Programs

7/01--	Faculty Member, The University of Michigan Graduate Program in Neuroscience

7/01--	Faculty Member, The University of Michigan Medical Genetics Residency Training Program
9/03--	Faculty Member, Michigan Comprehensive Cancer Center
9/03--	Faculty Member, Medical Scientist Training Program
3/04--	Full Faculty Member, The University of Michigan Center for Organogenesis
6/05--	Faculty Member, The University of Michigan Hearing, Balance, and Chemical Senses Training Program
2005--	Medical Scientist Training Program: MSTP Career Advisory Panel
2008--	Faculty Member, The University of Michigan Predoctoral Cell and Developmental Biology Training Program
2008--	Faculty Member, The University of Michigan Predoctoral Genetics Training Program

<u>Didactic Activities</u>

Oct 1995	Diabetes Short Course for Second-Year Medical Students, The University of Michigan Medical School, 4 one-hour lectures/small group sessions
Oct 2000	"Neurocristopathies", invited lecturer, 90 minute lecture/discussion, Organogenesis Seminar Series for graduate students on The Neural Crest
May 2001	"Syndromes that Affect the Craniofacial Complex", invited lecturer for 90 minute Dental Biology Course to Dental residents
Aug 2003-06	"Patients and Populations" Medical Genetics Component, Small Group Discussion Leader for M1 Medical Students (meets annually)
Fall 2004	Faculty Leader for NS700 seminar series (12-week course for teaching seminar presentations for Neuroscience graduate students) on "Transcriptional Regulation of Forebrain Development"
Nov 2004	"Transcription Factor Regulation of Hypothalamic Neuroendocrine Secretory Cells" 30 minute lecture/60 min discussion for CDB 680-682 Organogenesis of Complex Tissues and Organs
Dec. 2004	"Hyperammonemia", 90 min lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Feb. 2005	"Metabolic Acidosis" 90 min lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Jan. 2006	"Hyperammonemia", 90 min lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Feb. 2006	"Metabolic Acidosis" 90 min. lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Feb. 2006	"Genes and Development", 60 min lecture/discussion for M1 students, The University of Michigan Medical School (co-lecture with Dr. Vinod Misra).
Fall 2006	Co-Instructor of Neuronal Development Module for NS610/613, a first-year graduate course in Neuroscience. This included: "Regional Specialization in the Developing Nervous System", 60 minute lecture "Developmental Brain Disorders", 90 minute lecture Two 120-minute paper discussion sessions for graduate students Assistance with preparation and grading of exams for 18 enrolled students.
Dec. 2006	"Hyperammonemia", 90 min lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics

Feb. 2007	"Metabolic Acidosis" 90 min. lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Fall 2007	Co-Instructor of Neuronal Development Module for NS610/613, a first-year graduate course in Neuroscience. This included: "Developmental Brain Disorders", 90 minute lecture Two 120-minute paper discussion sessions for graduate students
Dec. 2007	"Metabolic Acidosis", 90 min lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Feb. 2008	"Hyperammonemia" 90 min. lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Aug 2008-11	"Patients and Populations" Medical Genetics Component, Small Group Discussion Leader for M1 Medical Students (meets annually)
9/08-4/09	Co-instructor for HG821/822, Human Genetics graduate student seminar course
9/08-4/09	Reviewer for CMB850, graduate student seminar course
9/15/08	"Mouse Models for Human CHARGE syndrome", 60 minute chalk talk for HG632, Genetic Strategies in Biomedical Research
10/3/08	"Hyperammonemia" 60 min. lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
Fall 2008	Co-Instructor of Neuronal Development Module for NS610/613, a first-year graduate course in Neuroscience. This includes: "Developmental Brain Disorders", a 90 minute lecture, and two 120-minute paper discussion sessions for graduate students
Feb. 2009	"Developmental Brain Disorders", a 60 minute lecture for HG649 Advanced Clinical Concepts in Medical Genetics
July 2009	"Introduction to Metabolic Disease", a 60 minute lecture for Pediatric Residents
Oct 2009	"Metabolic Acidosis" 60 min. lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics
11/2009	Co-Instructor of Neuronal Development Module for NS610/613, a first-year graduate course in Neuroscience. This includes: "Developmental Brain Disorders", a 90 minute lecture, and one 90-minute paper discussion session for graduate students
9/09-4/11	Co-instructor for HG821/822, Human Genetics graduate student seminar course
2/10-3/11	Developmental Brain Disorders" 60 min. lecture/discussion for HG649 Advanced Clinical Concepts in Medical Genetics, given once per year
9/09-current	"Introduction to Medical Genetics", 60 minute lecture for 3rd year Medical Students, given 2-3 times per year.

<u>Clinical Teaching</u>

9/04-7/05	Clinical educator in Medical Education: teaching first and second year medical students history and physical exam skills (40 half-days per academic year)
July 1999--	Supervising medical students, residents, and fellows in the Pediatric Genetics clinics and inpatient service.

<u>Seminars</u>

Aug. 1999	"Beckwith-Wiedemann Syndrome", 30 min. presentation, Medical Genetics Grand Rounds

Aug. 1999	"Breast Cancer", 20 min. presentation, Medical Genetics Grand Rounds
Oct. 1999	"Cystic Fibrosis", 20 min. presentation, Medical Genetics Grand Rounds
Dec. 1999	"Craniosynostosis", 20 min. presentation, Medical Genetics Grand Rounds
Dec. 1999	"Stickler syndrome", 20 min. presentation, Medical Genetics Grand Rounds
Oct 1999	"*Pitx2* in the Developing Brain", 20 minute presentation, Human Genetics Retreat
May, 2000	"SCAD vs SCHAD", 20 min. presentation, Medical Genetics Grand Rounds
Sept. 2000	"The Genetic Basis of Laterality", 60 min. presentation, Medical Genetics Grand Rounds
Sept. 2000	"Trisomy 5q31", 30 min. presentation, Medical Genetics Grand Rounds
Sept 2000	"Contiguous Gene Deletion Syndromes", 30 minute presentation, Cytogenetics Weekly Conference
Oct 2000	"The Role of *Pitx2* in Brain Development", 20 minute presentation, Dept of Pediatrics Annual Research Symposium
Apr 2001	"Evaluation of Organic Acidemias", 60 min. presentation, invited lecturer, Neurogenetics Journal Club
June 2001	"Nonketotic Hyperglycinemia", 20 min. presentation, Medical Genetics Grand Rounds
July 2001	"Pompe Disease", 20 min. presentation, Medical Genetics Grand Rounds
July 2001	"Introduction to Metabolic Disease", 60 min. presentation to Pediatric Residents
Sept. 2001	"Metabolic Disease", 60 min. presentation, 14th Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
May, 2002	"Contiguous Gene Deletion Syndromes", 60 min. presentation to Pediatric Residents
May, 2002	"Carnitine Palmitoyl Transferase Deficiency", 20 min. presentation, Medical Genetics Grand Rounds
June, 2002	"Craniosynostosis syndromes", 20 min. presentation, Medical Genetics Grand Rounds
June 2002	"Mapping an X-Linked Mental Retardation Syndrome", 60 min. presentation, Pediatric Grand Rounds, The University of Michigan.
July, 2002	"Crash Course in Metabolic Diseases", 60 min. presentation, Pediatric Residents
Sept. 2002	"Prenatal Diagnosis", 60 min. presentation, Pediatric Residents
Sept. 2002	"Metabolic Disease", 60 min. presentation, 15th Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
Dec. 2002	"Carnitine Palmitoyl Transferase Deficiency Type II", 20 min. presentation, Medical Genetics Grand Rounds
Jan. 2003	"Cockayne Syndrome", 20 min. presentation, Medical Genetics Grand Rounds
May, 2003	"Metabolic Diseases", 30 min. presentation, Pediatric ICU staff
Aug. 2003	"Crash Course in Metabolic Diseases", 60 min. presentation, Pediatric Residents
Sept. 2003	"GM2 Gangiosidosis", 20 min. presentation, Medical Genetics Grand Rounds
Sept. 2003	"Metabolic Disease", 60 min. presentation, 16th Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
March 2004	"X-linked Deafness", 30 min. presentation, Medical Genetics Grand Rounds
April 2004	"Sotos Syndrome", 30 min. presentation, Medical Genetics Grand Rounds
March 2004	"Overgrowth Syndromes" 60 min. lecture to Pediatric House Officers
March 2004	"A Revealing Case of X-Inactivation", 30 min. presentation, Medical Genetics Grand Rounds

Sept. 2004	"Metabolic Disease", 60 min. presentation, 17th Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
Sept. 2004	"Subthalamic Nucleus Genetics in Development and Disease", 60 min. presentation, hosted by the Neuroscience Program, The University of Michigan.
Sept. 2004	"Crash Course in Metabolic Diseases", 60 min. presentation, Pediatric Residents
Dec. 2004	"A Fragile Case of Autism", 30 min. presentation, Medical Genetics Grand Rounds
Feb. 2005	"*Ciona* Species for Studying Pitx2 Gene Function", 60 min. presentation, Organogenesis Journal Club
June, 2005	"Duplication 15q11q13 and schizophrenia", 30 min. presentation, Medical Genetics Joint Conference
July, 2005	"Crash course in metabolic diseases", 60 min. presentation to Residents, Mott Children's Hospital
Dec. 2005	"Using Mice to Dissect Multiple Anomaly Conditions", 20 min. presentation, Program in Biomedical Sciences
Jan. 2006	"*CHD7* in CHARGE", 60 min. presentation, Hearing and Biochemical Senses Seminar
Jan. 2006	"Molecular Genetics of CHD7 in Mutant Mice", 60 min. presentation, Research in Progress Seminar, Department of Human Genetics
June, 2006	"Beckwith Wiedemann Syndrome", 60 min. lecture to Pediatric Residents, The University of Michigan Mott Children's Hospital
June, 2006	"Aicardi Syndrome", 30 min. presentation, Medical Genetics Grand Rounds
Oct 2006	"Crash course in metabolic diseases", 60 min. presentation to Residents, Mott Children's Hospital
Jan. 2007	"Fatty Acid Oxidation Disorders, Lysosomal Storage Disorders, and Peroxisomal Diseases", 60 min. presentation to Residents, Mott Children's Hospital
March 2007	"CHARGE syndrome", 60 min. presentation, Medical Genetics Grand Rounds
July 2008	"Crash course in metabolic diseases", 60 min. presentation to Residents, Mott Children's Hospital
Sept. 2008	"Metabolic Disease", 60 min. presentation, 21st Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
July 2009	"Crash course in metabolic diseases", 60 min. presentation to Residents, Mott Children's Hospital
Sept. 2009	"Metabolic Disease", 60 min. presentation, 21st Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
Aug. 2010	"Management of Urea Cycle Disorders", 60 min. presentation to UM Pediatric ICU faculty and staff. Host: Dr. Kato Han.
April 2011	"Two cases of neurodevelopmental disorders", 30 min. presentation at Medical Genetics/Center for Genetics in Health & Medicine monthly conference

<u>Mouse Club Presentations</u>

Jan 2003	Exploring *Pitx2* Function in the Developing Mouse Brain
May 2004	*Pitx2* Function in Developing Neurons
April 2005	Analysis of *Pitx2* Isoforms in the Developing CNS (Christopher Chou)
March 2006	Analyzing *Chd7* in Mutant Mice

March 2007 Inner Ear Defects in *Chd7* Mutant Mice (Elizabeth A. Hurd)
January 2009 Midbrain Developmental Mechanisms, 30 min presentation
Nov 2010 *Pitx2* function in developing midbrain GABAergic neurons (Mindy Waite)

Neurogenesis Group Presentations
Feb 2003 The Role of *Pitx2* in Subthalamic Nucleus Development (Jennifer Skidmore)
Feb. 2004 *Pitx2* is required for STN and midbrain Neuronal Development
October 2004 *Pitx2* Function in Developing Neurons
October 2005 Mapping *Pitx2* Fates in Neuronal Development (Jennifer Skidmore)
March 2007 *Pitx2* Neuronal Fates in the Developing Hypothalamus and Midbrain
October 2007 Roles for CHD7 in Inner Ear Development (Liz Hurd)

Weekly Clinical Genetics Only Journal Club (My presentations listed below)
*journal club organizer from Dec. 2005-Nov. 2010
Jan. 2006 3-C syndrome, 20 min. presentation
June 2006 Beckwith Wiedemann Syndrome, 20 min. presentation
July 2006 *NOTCH2* in Alagille Syndrome, 20 min. presentation
Sept 2006 H19 hypomethylation in Silver Russell syndrome, 30 min presentation
April 2007 Autism and copy number variation, 20 min presentation
Aug. 2007 Jacobsen Syndrome, 20 min presentation
Jan. 2008 *POLG* mutations, 20 min presentation
March 2008 *BMP4* mutations in humans, 20 min presentation
Oct 2008 p63 mutations in human malformation syndromes; 20 min presentation
Jan 2009 CNTNAP2 mutations in autism; 20 min presentation
June 2009 TUBB2B mutations in polymicrogyria
Nov 2009 Massively parallel sequencing and Freeman-Sheldon mutations
July 2010 Skeletal dysplasias
Dec 2010 Whole exome sequencing in mental retardation
April 2011 Clinical genetics of autism and cognitive impairment

Life Sciences Institute Stem Cell Club Presentations
May 2009 Roles for the Chromodomain Protein CHD7 in Olfaction and Neural Stem Cell Proliferation (Wanda Layman)
Feb 2010 CHD7 function in Inner Ear Neurogenesis (Liz Hurd)
Sept. 2010 *CHD7* mutations and Hypogonadotropic Hypogonadism (Wanda Layman)

Inpatient Biochemical Genetics, consult service, and ward attending (2-4 week periods)
July, 2001, October, 2001, March, 2002, August, 2002, December, 2002
April 1-15, 2003, May 16-31, 2003 August, 2003, November, 2003, February, 2004, July, 2004, Sept, 2004, Dec, 2004, March, 2005, May, 2005, July, 2005, Oct. 2005, Dec. 2005, Feb. 2006, May 2006, July 2006, August 2006, October 2006, November 2006, Jan. 2007, March 2007, May 2007, July 2007, October 2007, Jan. 2008, March 2008, May 2008, Aug. 2008, Oct. 2008, Feb. 2009, April 2009, June 2009, Sept 2009, Nov. 2009, Feb 2010, March 2010, May 2010, July 2010, Sept. 2010, Oct. 2010, Dec 2010, Feb. 2011, April 2011, July 2011.

Ped Genetics Weekly Post-Clinic Teaching Conference

1 hr/week, residents, medical students, Genetic Counseling Students on Ped. Genetics rotation

Faculty Supervising Advisor, Medical Genetics Residency Program:

2001-2003	Stephanie Wechsler, M.D.
2001-2003	Sara Copeland, M.D.
2002-2003	Vinod K. Misra, M.D., Ph.D.
2005-2007	Tiffanee Lenzi, MD
2007-2009	Ryan Longman, MD
2007-2010	Jirair Bedoyan, MD, PhD
2011-2013	Shane Quinonez, MD

Postdoctoral Fellows Supervised:

1990-91	Caryn Vogel, Resident trainee; Current: Neurologist in Indianapolis
2000-03	Frank J. Probst, PhD, MD, Postdoctoral trainee; Current: Lecturer at Baylor College of Medicine
2006–curr	Elizabeth Hurd, PhD, Postdoctoral Research Fellow; Current: Research Associate
2008-2010	Jirair Bedoyan, MD, PhD, Medical Genetics Resident
2008-2010	Meredith Adams, MD, Fellow in Otology
2010--	Jirair Bedoyan, MD, PhD, Clinical Lecturer
2010	Martin P. Kracklauer, PhD, Postdoctoral Research Fellow

Undergraduate Students Supervised/current position:

1990-1992	Mihir Meghani, Freshman/Sophomore U of M Inteflex Program/Emergency Room physician, California
2000-2001	Sharon Fox, Freshman/Sophomore Summer research, Princeton University/Medical Student, Harvard University
2001	Joey Chang, Undergraduate Student, LS & A Freshman, UROP
2001--2002	Leni Morrison, Undergraduate Student LS & A Freshman, UROP
2000--2004	Anthony M. Sclafani, Honors Undergraduate Student, LS & A Freshman, UROP/Graduate Student in Biomedical Sciences, Yale University
2002--2004	Steven T. Philips, Honors Undergraduate Student LS & A Junior, SROP/MSTP student, The University of Michigan
2002--2003	Hemanth Ramaprakash, Undergraduate Student, College of Engineering Sophomore, UROP, SROP/PIBS student, The University of Michigan
2003-2004	Jamie Segel, Undergraduate Student LS & A Freshman, UROP
2003-2004	Robyn Vince, Undergraduate Student LS & A Freshman, UROP
2004-2008	John Cramer, Undergraduate Student LS & A Freshman, UROP, Honors program/Medical Student, Case Western University
2006	Patrice Capers, Undergraduate Student, SROP (Spelman College)
2006-08	Katherine Cheng, Undergraduate Student, LS & A Freshman
2006	Amrita Kaur, Undergraduate Student LS & A Freshman, UROP
2006	Karen Kao, Undergraduate Student LS & A Freshman, UROP
2007	Erin Mortgart, Undergraduate Student, SROP (Mount Union College)/Graduate student, University of Kentucky
2007	Renada Scott, Undergraduate Student, SROP (Alcorn State College)
2007-08	Ryan Hodges, Undergraduate Student, LS & A Freshman, UROP/MRC

2008-09 Arden Grace Trickey-Glassman, LS & A Sophomore, UROP/MRC
2008-10 Amanda Berry, LS & A Freshman, UROP
2008-- Elyse Reamer, LS & A Freshman, Honors Program in Biology
2008-11 Parisa Kaviany, LS & A Sophomore, Honors Program in Neuroscience

Medical Students supervised:
2009 Julie Kaplan, Summer Biomedical Research Program, University of Michigan

Graduate Students Supervised:
2005 Christopher Chou, MSTP student, The University of Michigan
2007-2011 Wanda Layman, PIBS PhD student, The University of Michigan
2007 Nicole Evans, PIBS PhD student, The University of Michigan
2007-2010 Kaia Skaggs, Neuroscience PhD student, The University of Michigan (co-mentor with Ben Novitch, UCLA)
2008 Allison Billi, rotating MSTP student, The University of Michigan
2008 Michael Mashiba, rotating MSTP student, The University of Michigan
2008-- Mindy Waite, Cell and Molecular Biology student, The University of Michigan
2010- Joshua Stowell, rotating Neuroscience student, The University of Michigan
2010- Joe Micucci, Biochemistry student, The University of Michigan
2010 Lindsey Cregan, rotating PIBS student, The University of Michigan
2011 Emily Maclary, rotating PIBS student, The University of Michigan

Thesis Committee membership:
2005-07 Kwan-Ho Chung, Neuroscience PhD student (thesis advisor: David Turner)
2005-2007 Chris Hart, Neuroscience PhD student (thesis advisor: David Turner)
2007-2011 Wanda Layman, PIBS PhD student, The University of Michigan (committee chair)
2007-2010 Kaia Skaggs, Neuroscience PhD student, The University of Michigan (co-mentor with Ben Novitch, UCLA)
2008-- Mindy Waite, Cell and Molecular Biology student, The University of Michigan (committee chair)
2008- Chris Chou, Human Genetics PhD student (thesis advisor: Tom Glaser)
2008- Lev Prasov, Human Genetics PhD student (thesis advisor: Tom Glaser)
2008-- Jillian Lee, Developmental Psychology PhD student (thesis advisor: Chris Monk)
2010- Valerie Schaibley, Human Genetics PhD student (thesis advisor: Jun Li)
2010- Randi Burns, CMB PhD student (thesis advisor: Margit Burmeister)
2010- Joe Micucci, Biochemistry student, The University of Michigan (committee chair)
2011- Danny Yang, Human Genetics PhD student (thesis advisor: John Kim)

Committee, Organizational, and Volunteer Service

Institutional

University of Michigan Medical School
2004 Stem Cell Faculty Search Committee, Department of Cell and Developmental Biology
2005 Medical Scientist Training Program Career Advisory Panel

2011-2014 Biomedical Research Council

Neuroscience Graduate Program

2003, 05, 06, 08, 09 Preliminary Exam Committee member
2005 Curriculum Evaluation Committee

Hearing, Balance, and Chemical Senses Training Program

2006--2010 Admissions Committee

Department of Human Genetics

2004-2006 James Neel Genetic Counseling Student Fellowship Award Committee,
2005 Faculty Selection Committee representing the Health Sciences Division, Rackham Predoctoral Graduate Student Award
2005-2011 Steering Committee and Permanent Member, Center for Genetics in Health and Medicine
2005, 08, 09 Preliminary Exam Committee member
2006 Program in Biomedical Sciences Graduate Student Admissions Committee,
2006 Preliminary Exam Committee chair
2007 Chair, Admissions Committee for Program in Biomedical Sciences Graduate Student Admissions
2007-08 Human Genetics Seminar Series Organizing Committee
2009-10 Cardiovascular Genetics Faculty Search Committee member, Dept of HG
2009-11 Faculty Search Committee
2011- Diane Baker Alumnae Lectureship Committee, Depart. of Human Genetics

Department of Pediatrics

2001 Participant in Proposals for funding for the Department of Pediatrics, the University of Michigan Health System Office of Medical Development and Alumni Relations
2002 Participant in Proposal to the Elizabeth E. Kennedy Fund, the University of Michigan Health System Office of Medical Development and Alumni Relations
2003 Resident Research Committee, Department of Pediatrics
2005-6 Pediatric Biochemical Genetics Recruitment Committee
2005-6 Pediatric Molecular Genetics Recruitment Committee
2006-09 Research Advisory Committee, Department of Pediatrics (elected position)
2007-- Program Committee for NIH-T32 Training Grant "Cell and Molecular Biology Training Program in Pediatrics".
2005-2010 Organizer, weekly Clinical Genetics Only Journal Club for Pediatric Genetics
2003-- Organizer, monthly Medical Genetics Grand Rounds for Pediatric Genetics
2001-2007 Organizer, 18-lecture series on Genetics topics for Pediatrics Residents
2007 Faculty organizer for Recruiting Pediatrics Residents Research Experience
2008 Chair Advisory Committee for Selection of Endowed Chair Positions
2009 Director, 19th Annual Pediatric Research Symposium, November 9-10, 2009
2010 Director, 20th Annual Pediatric Research Symposium, October, 2010
2011 Executive Committee Recruitment Officer, Child Health Research Center

Cell and Molecular Biology Training Program

2009-10 Co-chair, Retreat Planning Committee
2010-2013 Executive Committee faculty member, Human Genetics representative
2012 Prelim committee for Mie Kasanuki, PhD student (thesis advisor: Phil Gage)
2011-2014 Member, Program Committee

National

1999-- Member, Professional Advisory Board of the CHARGE Syndrome Foundation
2006-- Board Member, Michigan Association for the DeafBlind (DB Central)
2010 Abstract Reviewer, annual meeting of The American Society for Human Genetics

Volunteer Service

1999 Judge, Southeastern Michigan Science Fair
2005-2006 Volunteer Assistant, Girl Scout Troop #1539
Spring, 2005 Coach, Science Olympiad Map Reading, Logan Elementary School
Feb. 2006 Invited speaker for Annual Day, Greenhills High School, 45 min. lecture/discussion on "Raising Children with Special Needs".

Consulting Positions

March, 2006 Invited Member of the Advisory Board for the Simons Foundation Autism Research Fund (Host: Catherine E. Lord)

Visiting Professorships, Seminars, and Extramural Invited Presentations

Professorships

Dec, 2009-Jan 2010 Feinberg Foundation Visiting Faculty Program
Host: Orly Reiner, Weizmann Institute, Rehovot, Israel
This award covered travel and expenses for a 6 week visiting professorship at the Weizmann Institute in Rehovot, Israel

Seminars and Extramural Invited Presentations

June 1999 Alexander Graham Bell conference on hearing impairments; "Hearing Loss: The Parent and Physician Perspective", The University of Michigan Medical Center.
June, 2000 The 101st Annual Meeting of The Japanese Society for Otolaryngology, Tokyo Japan; "Genetic Approaches to Hereditary Deafness".
Jan. 2002 "Analysis of *Pitx2* Function in the Developing Mouse Diencephalon and Mesencephalon", Seminario de Año Nuevo, University of Murcia, Murcia, Spain. Host: Luis Puelles.
Feb. 2002 "Functional Analysis of *Pitx2* in Developing Neurons", 60 min. presentation, Cell and Developmental Biology Seminar, The University of Michigan.
Oct. 2002 "Adverse Developmental Effects of Alcohol", 21st Annual Michigan Statewide Conference on Child Abuse and Neglect, Ypsilanti, Michigan, sponsored by The University of Michigan Medical School.
Feb. 2003 "X-Linked Mental Retardation", Pediatric Grand Rounds, William Beaumont Hospital, Royal Oak, Michigan (Host: David Aughton).
March, 2003 "PITX2 is required for subthalamic nucleus development". *The Great Lakes Mammalian Development Meeting*, Toronto, March 14-16, 2003.

May 2004	Annual meeting of the Society for Pediatric Research, San Francisco, "Non-hematopoietic Stem Cells", session chair.
Dec. 2004	"The Role of *Pitx2* in Central Nervous System Neuronal Differentiation", Centre de Regulació Genòmica, Barcelona Spain. Host: Xavier Estivill
March 2005	"*Pitx2*-mediated disruption of mammalian neuronal development", The Weizmann Institute of Science, Rehovot, Israel. Host: Orly Reiner
July 2005	"*Pitx2* function in developing CNS neurons", Department of Pediatrics, The University of Iowa, Iowa City. Host: Val Sheffield.
Sept. 2005	"Introduction to Metabolic Diseases", 17th Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
Jan. 2006	"Genetics of Autism", Center for Autism and Communication Disorders, The University of Michigan. Host: Catherine Lord
March 2006	"Fate mapping *Pitx2* lineage neurons in the developing mouse brain", New York University Medical Center, The Skirball Institute. Host: Alex Joyner
Sept. 2006	"Introduction to Metabolic Diseases", 18th Annual Pediatric Board Review Course, sponsored by The University of Michigan Medical School.
Oct. 2006	"Loss of PITX2 disrupts migration and differentiation in distinct populations of neurons in the developing mouse midbrain and hypothalamus" *Annual Meeting of the Society for Neuroscience*, Atlanta, Georgia, October 14-18, 2006.
Feb, 2007	"Roles for CHD7 in CHARGE-related organ development", Nephrology Basic Science Seminar. Host: Ben Margolis
Oct. 2007	Moderator, Platform Session "Animal Models", *57th Annual Meeting of the American Society for Human Genetics*, San Diego, California, October 23-27, 2007.
Jan. 2008	"Mechanisms of Chromodomain Protein Function in Mammalian Inner Ear Development", University of Illinois-Champagne, Urbana. Host: Lori T. Raetzmann
Jan. 2008	"Pleiotropic roles for the Chromodomain protein CHD7 in human and mouse development". Cell and Developmental Biology Seminar, The University of Michigan. Host: Deneen Wellik
Feb. 2008	Moderator, Platform Session "Development", *31st Annual MidWinter Meeting of the Association for Research in Otolaryngology*, Phoenix, Arizona, February 16-21, 2008.
May, 2008	"Mouse Models of Human CHARGE Syndrome", Pediatric Endocrinology Seminar, The University of Michigan. Host: Vasantha Padmanabhan.
June, 2008	Roles for Chromodomains Proteins in Cellular Differentiation, Proliferation, and Survival", Pediatric Hematology/Oncology Seminar, The University of Michigan. Host: Larry Boxer.
Sept. 2008	"Pitx2 Function in Mammalian Development", Review of the field, oral presentation and Thesis Opponent for Johan Holmberg, Lund University, Sweden.
Sept. 2008	"Roles for Pitx2 in Mammalian Neuronal Migration and Cell Fate", Seminar at Lund University, Sweden. Host: Tord Hjalt
July 7, 2009	"Roles for the transcription factor PITX2 in mammalian neuronal migration and connectivity. Seminario de Alicante, Alicante, Spain. Host: Salvador Martinez.
Sept. 2009	"Chromodomain protein 7 functions in inner ear development". NIDCD

	Host: Doris Wu.
Oct. 2009	Invited Scientific Session Organizer, "CHARGE Syndrome and CHD7: Linking Chromatin Biology and Development with Variable Human Phenotypes", 59th Annual meeting of The American Society of Human Genetics, Honolulu, Hawaii, October 20-24, 2009.
Oct. 2009	Invited speaker, "Use of mouse models in exploring CHD7 functions." Oral presentation (20 min) at the 59th Annual meeting of The American Society of Human Genetics, Honolulu, Hawaii, October 20-24, 2009
Nov, 2009	Invited speaker, "Introduction to Genetics in Developmental Brain Disorders", University of Michigan Center for Autism and Communication Disorders. Host: Catherine Lord
Jan. 2010	*Pitx2* is Required for Neuronal Migration in Distinct Regions of the Developing Mouse Brain". The Weizmann institute, Rehovot, Israel. Host: Orly Reiner.
Jan. 2010	"Chromodomain protein 7 functions in inner ear development". The Weizmann Institute, Rehovot, Israel. Host: Avraham Yaron.
June 2010	"CHARGE Syndrome: from Bedside to Bench". Emory University, Atlanta, Georgia. Host: David Ledbetter.
June 2010	"Chromodomain Proteins in Development and Disease: Lessons from CHD7". Emory University, Atlanta, Georgia. Host: David Ledbetter.
Nov. 2010	Moderator, Platform Session, Craniofacial and Skeletal Disorders, *60th Annual Meeting of the Society for Human Genetics*.
March 2011	"Mechanisms of chromodomain protein-mediated neurogenesis". Neurology Research Conference, The University of Michigan. Host: Jack Parent.
March 2011	"Genetics of Autism". Joint OB-Gyn/Pediatrics Grand Rounds, St. Joseph Mercy Hospital, Ypsilanti, Michigan. Host: Robert Stager.
March 2011	"Genetics of Autism". Center for Human Growth and Development, The University of Michigan. Host: Chris Monk.
April 2011	"Genetics of Autism". Developmental/Behavioral Pediatrics conference, The University of Michigan. Host: Betsy Lozoff.
May 2011	"Chromodomain Proteins in Development and Disease". Invited seminar for the Department of Human Genetics, Cincinnati Children's Hospital. Host: Gregory Grabowski.
June 2011	"Recent Advances in Understanding the Genetics of Autism". Pediatrics Grand rounds, Michigan State University, Kellogg Center for Medical Studies. Host: Allan Lareau
July 2011	"Advances in the Use of Mouse Models of CHARGE Syndrome". Professional Day, 10th Bienniel meeting of the CHARGE Syndrome Foundation, Orlando, Florida, July 28, 2011.
July 2011	"Advances in Mouse Models of CHARGE Syndrome", Family Day, 10th Bienniel meeting of the CHARGE Syndrome Foundation, Orlando, Florida, July 29, 2011.

<u>Radio, Internet, and Newspaper interviews</u>

Dec 9, 2009 Interviewed by Lucy Ann Lance for WLBY 1290 am for evaluation of an article on the role of genetics in autism and obesity (Bochukova et al., *Nature* 2009; 20 min live show; podcast available at www.lucyannlance.com).

April 21, 2011 "Ask the Doctor" Podcast, interview "What is autism?" and "Is it treatable." (http://www.askthepodcastdoctor.org/)

May, 2011 http://www.uofmhealth.org/news/multimedia+gallery+news#/1270248827419

Bibliography

Original Publications in Peer-Reviewed Journals

1. **Martin DM**, Yee D, Feldman EL. Gene expression of the insulin-like growth factors and their receptors in cultured human retinal pigment epithelial cells. *Mol. Brain Res.* 12:181–186, 1992.

2. **Martin DM**, Yee D, Carlson RO, Feldman EL. Gene expression of the insulin-like growth factors and their receptors in human neuroblastoma cell lines. *Mol. Brain Res.* 15:241–246, 1992.

3. **Martin DM**, Carlson RO, Feldman EL. Interferon-γ inhibits DNA synthesis and insulin-like growth factor–II expression in human neuroblastoma cells. *J. Neurosci. Res* .34:489-501, 1993.

4. **Martin DM** and Feldman EL. Regulation of insulin-like growth factor–II expression and its role in autocrine growth of human neuroblastoma cells. *J. Cell. Physiol.* 155:290-300, 1993.

5. Carlson RO, **Martin DM**, Feldman EL, Agranoff BW. PKC activity and PKC–α mRNA content are reduced in serum-deprived human neuroblastoma cells without concomitant induction of differentiation. *Exp. Cell Res.,* 207:340-347, 1993.

6. **Martin DM** and Feldman EL. Reversibility of serum removal effects on IGF–II mRNA in human neuroblastoma cells. *Ann. N Y Acad. Sci.* 692:259-261, 1993.

7. Meghani, MA, **Martin, DM**, Singleton, JR, Feldman, EL. Effects of serum and insulin-like growth factors on human neuroblastoma cell growth. *Reg. Peptides*, 48:217-224, 1993.

8. **Martin, DM**, Singleton, JR, Meghani, MA, Feldman, EL. IGF receptor function and regulation in autocrine human neuroblastoma cell growth. *Reg. Peptides*, 48:225-232, 1993.

9. Anderson DW, Probst FJ, Belyantseva IA, Fridell RA, Beyer L, **Martin DM**, Wu D, Kachar B, Friedman TB, Raphael Y, Camper SA. The motor and tail regions of myosin XV are critical for development and function of the auditory and vestibular systems. *Human Molecular Genetics* 9:1729-1738, 2000.

10. **Martin, DM**, Probst FJ, Camper, SA and Petty, EM. Characterization and Genetic Mapping of a New X-Linked Deafness-Mental Retardation Syndrome, *Journal of Medical Genetics* 37: 836-841, 2000.

11. **Martin, DM**, Sheldon, S, and Gorski, JL. CHARGE Association in a Child with an Apparently Balanced *de novo* Chromosomal Translocation t(2;7)(p14;q21.11), *American Journal of Medical Genetics* 99(2):115-9, 2001.

12. **Martin, DM**, Gencyuz, CF, and Petty, EM. Systemic Lupus Erythematosis in a Man with Noonan Syndrome: A Brief Case Report and Literature Review. *American Journal of Medical Genetics* 102: 59-62, 2001.

13. **Martin, DM** and Gorski, JL. Ocular Malformations, Postaxial Polydactyly and Delayed Intramembranous Ossification: a New Autosomal Dominant Condition. *Journal of Medical Genetics* 38:547-55, 2001.

14. **Martin, DM**, Probst, FJ, Fox, SE, Schimmenti, LA, Semina, E, Belmont, JW, Camper, SA. Exclusion of *PITX2* Mutations as a Major Cause of CHARGE Association. *American Journal of Medical Genetics* 111:27-30, 2002.

15. Brown, DF, Kim, TB, Petty, EM, Downs, CA, **Martin, DM**, Strouse, PJ, Moroi, SE, Milunsky, JM, Lesperance, MM. Autosomal dominant stapes ankylosis, broad thumbs, hyperopia and skeletal anomalies caused by heterozygous nonsense and frameshift mutations in the noggin gene (*NOG*) *American Journal of Human Genetics* 71:618-624, 2002. PMC 379196.

16. **Martin, DM,** Skidmore, JM, Fox, SE, Gage, PJ, Camper, SA. *Pitx2* Distinguishes Subtypes of Terminally Differentiated Neurons in the Developing Mouse Neuroepithelium. *Developmental Biology* 252(1): 84-99, 2002.

17. **Martin, DM**, Mindell, MH, Kwierant, CA, Glover, TW and Gorski, JL. Interrupted Aortic Arch in a Child with Trisomy 5q31.1q35.1 Due to a Maternal (20;5) Balanced Insertion. *American Journal of Medical Genetics* 116A:268-271, 2003.

18. Keegan, CE, **Martin, DM**, Quint, DJ, Gorski, JL. Acute extrapyramidal syndrome in ornithine transcarbamylase deficiency: metabolic stroke involving the caudate and putamen. *European Journal of Pediatrics* 162: 259–263, 2003.

19. Brown DJ, Kim TB, Petty EM, Downs CA, **Martin DM**, Strouse PJ, Moroi SE, Gebarski SS, Lesperance MM. Characterization of a Stapes Ankylosis Family with a NOG Mutation. *Otology and Neurotology* 24(2):210-5, 2003.

20. **Martin, DM** and Raphael, Y. Gene-Based Diagnostic and Treatment Methods for Tinnitus. *Tinnitus* 9(1):3-10, 2003.

21. Treutelaar, MK, Skidmore, JM Dias-Leme, CL, Hara, M, Zhang, L Simeone, D, **Martin, DM**, Burant, CF, Nestin-lineage cells contribute to the microvasculature but not endocrine cells of the islet. *Diabetes* 52(10):2503-2512 (2003).

22. Karolyi, IJ, Probst, FJ, Beyer, L, Odeh, H, Dootz, G, Cha, KB, **Martin, DM**, Avraham, KB, Kohrman, D, Dolan, DF, Raphael, Y, Camper, SA. *Myo15* gene function is distinct from *Myo6*,

Myo7a, and *pirouette* genes in development of cochlear stereocilia. *Human Molecular Genetics* 12(21):2797-805 (2003).

23. **Martin, DM,** Skidmore, JM, Philips, ST, Vieira, C, Gage, PJ, Condie, BG, Raphael, Y, Martinez, S, Camper, SA. PITX2 is required for normal development of neurons in the mouse subthalamic nucleus and midbrain. *Developmental Biology* 267 (1):93-108 (2004).

24. Lalani, SR, Safiullah, A, Molinari, LM, Fernbach, SD, **Martin, DM**, Belmont, JW. *SEMA3E* mutation in a patient with CHARGE Syndrome. *Journal of Medical Genetics* 41, 41(7):E94 (2004).

25. FJ Probst, P Hedera, AM Sclafani, MG Pomponi, G Neri, J Tyson, JA Douglas, EM Petty, **DM Martin**. Skewed X-inactivation in Carriers Refines Genetic Mapping of a Novel X-linked Deafness Mental Retardation Syndrome. *American Journal of Medical Genetics* 131A: 209-212 (2004).

26. Kojic, J, Robertson, PL, Quint, DJ, **Martin, DM**, Pang, Y, Sundgren, PC. High Brain Glutamine by MR Spectroscopy in a Urea Cycle Disorder with Coma. *Pediatric Neurology* 32(2):143-6 (2005).

27. Philips, ST, Albin, RA, **Martin, DM**. Genetics of Subthalamic Nucleus in Development and Disease. *Experimental Neurology* 192:320– 330 (2005).

28. Vadlamudi, U, Espinoza, HM, Ganga, M, **Martin, DM,** Liu, X, Engelhardt, JF, Amendt, BA. PITX2, β-catenin and LEF-1 interact to synergistically regulate the *LEF-1* promoter. *Journal of Cell Science* Mar 15;118(Pt 6):1129-37 (2005).

29. Espinoza, HM, Ganga, M, Vadlamudi, U, **Martin, DM**, Brooks, BP, Semina, EV, Murray, JC, Amendt, BA. Protein C phosphorylation modulates N- and C-terminal regulatory activities of the PITX2 homeodomain protein. *Biochemistry* 2005 Mar 15;44(10):3942-3954 (2005).

30. Raphael Y and **Martin, DM**. "Deafness: Lack of regulation encourages hair cell growth". *Gene Therapy* Mar 31; Jul;12(13):1021-2 (2005).

31. Sclafani, AM, Skidmore, JM, Ramaprakash, H, Trumpp, A, Gage, PJ, **Martin, DM**. Nestin-Cre mediated deletion of *Pitx2* in the mouse. *Genesis,* 44:336-344 (2006).

32. Hurd, EA, Capers, PL, Blauwkamp, MN, Adams, ME, Raphael, Y, Poucher, HK, **Martin, DM.** Loss of *Chd7* function in gene trapped reporter mice is embryonic lethal and associated with severe defects in multiple developing tissues. *Mammalian Genome,* Feb;18(2):94-104 (with Cover figure) (2007).

33. Hurd, EA and **Martin, DM**. The 20th International Mammalian Genome Conference Meeting Report. *Mammalian Genome.* Mar;18(3):145-53. Epub 2007 Apr 10 (2007).

34. Adams, ME, Hurd, EA, Beyer, LA, Swiderski, DL, Raphael, Y, and **Martin, DM**. Defects in vestibular sensory epithelia and innervation in mice with loss of *Chd7* function: implications for human CHARGE syndrome. *Journal of Comparative Neurology* 504:519-532 (2007).

35. Vue, TY, Aaker, J, Taniguchi, A, Kazemzadeh, C, Skidmore, JM, **Martin, DM**, Martin, JF, Treier, M, Nakagawa, Y. Characterization of progenitor domains in the developing mouse thalamus. *Journal of Comparative Neurology* Nov 505(1):73-91 (2007).

36. Skidmore, JM, Cramer, JD, Martin, JF, and **Martin, DM**. *Cre* fate mapping reveals lineage specific defects in neuronal migration with loss of *Pitx2* function in the developing mouse hypothalamus and subthalamic nucleus. *Molecular and Cellular Neurosciences* 37: 696-707 (2008). PMC 2386145.

37. Ou, Z, **Martin, DM**, Cooper, ML, Chinault, AC, Stankiewicz, P, Cheung, SW. "Branchiootorenal Syndrome and Oculovertebral Spectrum Features with Duplication of SIX1, SIX6, and OTX2 Resulting from a Complex chromosomal Rearrangement. *American Journal of Medical Genetics* Oct 1;146A(19):2480-9 (2008). PMC requested.

38. D.R. Jensen, **D.M. Martin**, S. Gebarski, T. Sahoo, E. Brundage, C.A. Chinault, E. Otto, M. Chaki, F. Hildebrandt, S.W. Cheung, and M.. Lesperance. A Novel Chromosome 19p13.12 Deletion in a Child with Multiple Congenital Anomalies. *American Journal of Medical Genetics* Am J Med Genet Part A 149A:396–402 (2009). PMC 2872113.

39. Layman, WS, McEwen, DP, Beyer, LA, Lalani, SR, Fernbach, SD, Oh, E, Swaroop, A, Hegg, CC, Raphael, Y, Martens, JR, **Martin, DM**. Defects in neural stem cell proliferation and olfaction in *Chd7* deficient mice indicate a mechanism for hyposmia in human CHARGE syndrome. *Human Molecular Genetics*. 18(11):1909-1923 (2009). PMC 2678924.

40. Rosenfeld, JA, Ballif, BC, **Martin, DM**, Aylsworth, AS, Bejjani, BA, Torchia, BS, Shaffer, LG. Clinical characterization of individuals with deletions of genes in holoprosencephaly pathways by aCGH refines the phenotypic spectrum of HPE. *Human Genetics*, 127(4):421-440 (2010).

41. Zentner, GE, Layman, WS, **Martin, DM**, Scacheri, P. Molecular and phenotypic aspects of *CHD7* mutation in CHARGE syndrome. *American Journal of Medical Genetics*. Mar;152A(3):674-86 (2010). PMC 2918278.

42. Bedoyan, JK, Kumar, RA, Sudi, J, Silverstein, F, Ackley, T, Iyer, RK, Christian, SL, **Martin, DM**. Duplication at 16p11.2 in a Child with Infantile Seizure Disorder. *American Journal of Medical Genetics A.* Jun;152A(6):1567-74 (2010). PMC 3160635.

43. Layman, WS, Hurd, EA, **Martin, DM**. Chromodomain Proteins in Development: Lessons from CHARGE Syndrome. *Clinical Genetics* Jul;78(1):11-20 (2010) PMC 3079949.

44. **Martin, DM.** Chromatin remodeling in development and disease: focus on CHD7. *PLoS Genetics* Jul 15;6(7):e1001010 (2010). PMC 2904764.

45. Hurd, EA, Poucher, HK, Cheng, K, Raphael, Y, **Martin, DM**. The ATP-dependent chromatin remodeling enzyme CHD7 regulates proneural gene expression and neurogenesis in the inner ear. *Development* 137(18):3139-3150 (2010).

46. Zentner, GE, Hurd, EA, Schnetz, MP, Wang, Z, Tesar, PJ, **Martin, DM**, Scacheri, P CHD7 functions in the nucleolus as a positive regulator of ribosomal RNA biogenesis, *Human Molecular Genetics* Vol.19, No.18:3491-3504 (2010).

47. Viren C. Patel, Kajari Mondal, Amol Carl Shetty, Vanessa L. Horner, Jirair Bedoyan, **Donna M Martin**, Tamara Caspary, David J. Cutler and Michael E. Zwick, Microarray Oligonucleotide Probe Designer (MOPeD). *Open Access Bioinformatics* Vol. 2010:2 Pages 145 – 155 (2010). PMC 3048354.

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1. Cushman, LJ, Camper, SA, **Martin, DM**, Baker, DL, "About Pituitary Tumors: Information for Patients and Families." Published by the Pituitary Disorders, Education & Support group, T. Sullivan, Founding Director, Brighton, MI, with support from an educational grant from Pharmacia & Upjohn (1999).

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