Approved by the Regents
May 15, 2008

RECOMMENDATION FOR PROMOTION
UNIVERSITY OF MICHIGAN MEDICAL SCHOOL
DEPARTMENT OF PEDIATRICS AND COMMUNICABLE DISEASES
DEPARTMENT OF HUMAN GENETICS

Donna M. Martin, M.D., Ph.D., Assistant Professor of Pediatrics and Communicable Diseases, Department of Pediatrics and Communicable Diseases, and Assistant Professor of Human Genetics, Department of Human Genetics, Medical School, is recommended for promotion to Associate Professor of Pediatrics and Communicable Diseases, with tenure, Department of Pediatrics and Communicable Diseases, and Associate Professor of Human Genetics, without tenure, Department of Human Genetics, Medical School.

Academic Degrees:

M.D.	1996	University of Michigan
Ph.D.	1992	University of Michigan
B.S.	1987	Michigan Technological University

Professional Record:

2003-Present	Assistant Professor of Pediatrics and Communicable Diseases, University of Michigan
2003-Present	Assistant Professor of Human Genetics, University of Michigan
2001-2003	Research Investigator, Department of Human Genetics, University of Michigan
2001-2002	Lecturer, Department of Pediatrics and Communicable Diseases, University of Michigan

Summary of Evaluation:

Teaching: Dr. Martin teaches in multiple venues in both the Department of Pediatrics and the Department of Human Genetics, including Medical Student training in human genetics, didactic genetics lectures for Pediatric residents and Medical Genetics residents, Pediatrics Grand Rounds, and Medical Genetics Grand Rounds at the University of Michigan, and an Annual Pediatric Board Review Course on Metabolic disease, which she has given for the last five years. Students have given her outstanding evaluations for her well-prepared sessions. In her laboratory, she mentors several undergraduate students, Ph.D. candidate graduate students, one postdoctoral fellow, and one senior Research Associate, exploring developmental disorders of the nervous system.

Dr. Martin is the Director of the Medical Genetics Residency Program, a two-year program that provides Clinical Medical Genetics training for physicians to achieve board certification through the American Board of Medical Genetics. This post, which she has held since November, 2006, involves responsibilities for the education, recruitment and administrative oversight of Medical Genetics residents. She also mentors Genetic Counseling students in the Pediatric Genetics clinic, and has assisted Human Genetics graduate students in seminar preparation as part of their coursework requirements. Dr. Martin mentors rotating Pediatrics

residents and M4 medical students during their elective months in Pediatric Genetics, and she trains weekly rotating M3 medical students in the Pediatrics Genetics and Biochemical Genetics clinics. In 2004-05, 10% of her effort was devoted to Clinical Education for M1 and M2 medical students learning clinical skills (history taking and physical exams). She received outstanding evaluations from students for her efforts in this course. In 2006, Dr. Martin and a divisional colleague, Dr. Vinod Misra, co-developed and presented a 60 minute lecture on genetics of brain development for M1 medical students.

Research: Research in Dr. Martin's laboratory has focused on mouse models of human developmental disorders of the nervous system. Her laboratory defined a critical role for *Pitx2*, a paired like homeodomain transcription factor mutated in Rieger syndrome, in mouse brain development. This work resulted in two publications in *Developmental Biology*, one in *Genesis*, and a review article on subthalamic nucleus development and function in *Experimental Neurology*. During this time, her laboratory was also supported by a NIH funded Child Health Research Center grant in the Department of Pediatrics, and Dr. Martin received the first Janette Ferrantino Award in 2003 from the Department of Pediatrics.

Dr. Martin has established a very strong, funded research program in developmental disorders of the nervous system. Recent efforts include developing a new initiative to identify genes involved in autism. This work, directed by Dr. Martin, is being done in collaboration with Dr. Cathy Lord at the University of Michigan Autism & Communication Disorders Center (UMACC), Drs. Innis and Ram Iyer of the Michigan Medical Genetics Laboratories in Pediatrics, and Dr. Bob Lyons of the DNA Sequencing Core. This project illustrates the outstanding energy and commitment Dr. Martin brings to the genetics of neurological disorders, and highlights her ability to assemble teams of researchers around common goals. Her long-term goals are to gain critical insights into the basic neurobiology of disorders of the human nervous system that will ultimately lead to rational therapies and interventions for these children. Clearly, she has established a national and international reputation as a stellar academic physician who will continue to make important contributions to our understanding of nervous system function and its relevance for children with developmental disabilities.

Recent and Significant Publications:

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* 18(2):94-104, 2007.

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.

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.

Probst FJ, Hedera P, Sclafani AM, Pompon MG, Neri G, Tyson J, Douglas JA, Petty EM, Martin DM: 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.

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.

Service: Dr. Martin provides extensive service in both the Department of Pediatrics and the Department of Human Genetics. At the national level, Dr. Martin has participated in three NIH Study Sections as a temporary member. She has served as a reviewer for the Oak Ridge Laboratories, the National Science Foundation, and for numerous peer-reviewed journals. She was recently appointed as a scientific consultant for the National Organization for Hearing Research. Dr. Martin is on medical advisory boards for the CHARGE Syndrome Foundation, a national support organization, and for Deaf-Blind Central, the State-sponsored agency whose mission is to provide care and support for children with vision and hearing impairments and their families. Dr. Martin has served as a representative for Human Genetics on the Faculty Selection Committee for the Rackham Predoctoral Graduate Student Award. In winter, 2006, she began serving as a faculty representative for Human Genetics on the PIBS Admissions Committee. In 2005, she joined the Steering Committee for the Center for Genetics in Health and Medicine, based in the Department of Human Genetics. She is an active, contributing member in the Hearing, Balance, and Chemical Senses program, the Neuroscience Program, the Center for Organogenesis, and the Cancer Center. She and her laboratory staff are regular contributors to Mouse Club, an interdisciplinary group that meets monthly to discuss scientific advances in mouse genetics. Dr. Martin is also a faculty sponsor for the Rackham Interdisciplinary Workshop entitled "Experimental Approaches to Understanding Vertebrate Neurogenesis." This workshop consists of 12 principal investigators and their laboratories who meet monthly to discuss recent advances in Developmental Neurobiology. Since October 2003, Dr. Martin has received annual funding from Rackham to cover administrative costs, support for a student coordinator, and monies to bring in outside speakers to the University. Numerous collaborations and joint efforts have arisen from these meetings. This workshop is a vital component of the educational contribution to the University and is an example of Dr. Martin's outstanding efforts to foster inter-departmental communication to promote scientific collaboration.

Professional Work: Dr. Martin is a superb clinician. She has developed nationally recognized expertise in evaluating and caring for children with a variety of genetic and developmental disorders. She participates in weekly Pediatric Genetics clinics, monthly Biochemical Genetics clinics, staffs the inpatient Pediatric Genetics service, and shares call responsibilities for the Pediatric Genetics service and for biochemical genetics patients an average of 10 weeks per year. Currently 10% of her effort is devoted to patient care. An additional 10% of her effort includes clinical evaluations for children in the University of Michigan Autism Center for Communication Disorders, where she was recruited to help based on her knowledge and expertise with children who have developmental disorders affecting the nervous system. Her clinical skills were recently acknowledged by *Detroit Hour Magazine,* where she was listed as one of the area's top doctors. Dr. Martin participates actively 3-4 times per year in several two-day Genetics Outreach Clinics in Marquette, Traverse City and Gaylord, Michigan.

External Review:

Reviewer A: "...she has clearly developed substantial expertise in an area that is nationally recognized as valuable and hard to find, and would allow her to take her pick of positions across the country."

Reviewer B: "She exemplifies the successful physician-scientist who maintains a strong commitment to clinical care while building an outstanding research career."

Reviewer C: "Donna's work has focused primarily on the *PITX2* gene and, in particular, in the role that it plays in neurodevelopment. Donna has built a strong publication record in this arena and it would be entirely fair to say that she is now regarded as one of the world's experts in this particular area."

Reviewer D: "Dr. Martin is one of an unfortunately small and high-demand subspecialist groups, the translational clinical molecular-genetics researcher. There are fewer of these researchers in the country than there are available positions and well-regarded university medical centers compete vigorously for these candidates."

Reviewer E: "She has established a productive research laboratory in the area of developmental genetics which beautifully compliments her interest in clinical genetics and medical genetics. She has undertaken major administrative responsibilities in running the residency training program and this also contributes to her increasingly high profile nationally and internationally. Someone of Dr. Martin's stature would certainly achieve the rank of Associate Professor here..."

Summary of Recommendation:

Dr. Martin is a highly productive physician scientist who has an excellent record of scientific achievement, teaching and academic service. I enthusiastically support her promotion to Associate Professor, with tenure, in the Department of Pediatrics and Communicable Diseases, and Associate Professor, without tenure, in the Department of Human Genetics.

James O. Woolliscroft, M.D.
Dean
Lyle C. Roll Professor of Medicine

May 2008