Second International Chordoma Research Workshop

April 3–5, 2008

Speaker Biographies

Christopher Austin, M.D.

Dr. Christopher Austin is Director of the NIH Chemical Genomics Center (NCGC) and Senior Advisor to the Director for Translational Research at the National Human Genome Research Institute (NHGRI). NCGC, part of the NIH Roadmap Molecular Libraries initiative, develops small-molecule probes for biological functions and new paradigms for high-throughput screening, chemistry, and cheminformatics. In his role as Senior Advisor for Translational Research, he initiated the Knockout Mouse Project, which is producing knockout mice for all mouse genes, and an in-depth transcriptome map of the mouse. Dr. Austin came to NIH from Merck and Co., where he directed research programs in genomics-based target discovery, pharmacogenomics, and DNA microarray technologies, with a focus on neuropsychiatric diseases. Dr. Austin received his A.B. from Princeton University and M.D. from Harvard University. He did clinical training in neurology at Massachusetts General Hospital, followed by a fellowship in genetics at Harvard.

Laurence Baker, D.O.

Dr. Laurence Baker is Professor of Internal Medicine and Pharmacology in the Departments of Internal Medicine and Pharmacology, Division of Hematology/Oncology, University of Michigan Medical School. He received his medical degree from the University of Osteopathic Medicine and Surgery in Des Moines, Iowa, in 1966. He did a medical residency at Detroit Osteopathic Hospital, which was followed by an oncology fellowship at Wayne State University. He joined the faculty of the Department of Oncology at Wayne State upon completion of his fellowship in 1972. In 1987, Dr. Baker became the Director of the Meyer L. Prentis Comprehensive Cancer Center of Metropolitan Detroit, where he remained until his recruitment to the University of Michigan in 1994. He served as the Deputy Director and Director for Clinical Research at the University of Michigan until May 2004.

Dr. Baker has 30 years' experience in the treatment of sarcoma and design of clinical trials for new sarcoma therapies. In April 2005, he became Chairman of the Southwest Oncology Group (SWOG), the largest cancer clinical trials organization in the world, after election by the membership. SWOG's membership lists nearly 4,000 of the nation's leading physicians and scientists at 283 institutions in the United States and Canada. Dr. Baker is also the Executive Director of Sarcoma Alliance for Research through Collaboration (SARC), a not-for-profit consortium that advocates for sarcoma medical research and for the conduct of clinical trials studying new treatment for sarcoma. He is a scientific consultant to the National Cancer Institute and several universities, a member of many prestigious organizations, and a nationally recognized expert in sarcoma treatment. He has published extensively in peer-reviewed

literature, has authored or co-authored 50 books and/or book chapters, and is regularly invited to speak as a guest lecturer both at the national and international levels.

Francis Collins, M.D., Ph.D.
Dr. Francis Collins is Director of the National Human Genome Research Institute (NHGRI) at NIH. He led the successful effort to complete the Human Genome Project (HGP), a complex multidisciplinary scientific enterprise directed at mapping and sequencing all human DNA and determining aspects of its function. A working draft of the human genome sequence was announced in June 2000, an initial analysis was published in February 2001, and a high-quality reference sequence was completed in April 2003. From the outset, the project ran ahead of schedule and under budget, and all data are now available to the scientific community without restrictions on access or use.

Dr. Collins received a B.S. from the University of Virginia, a Ph.D. in physical chemistry from Yale University, and an M.D. from the University of North Carolina at Chapel Hill. Following a fellowship in human genetics at Yale, he joined the faculty at the University of Michigan, where he remained until moving to NIH in 1993. His research has led to the identification of genes responsible for cystic fibrosis, neurofibromatosis, Huntington's disease, and Hutchison-Gilford progeria syndrome. He is a member of the Institute of Medicine and the National Academy of Sciences.

Thomas F. DeLaney, M.D.
Dr. Thomas DeLaney is Medical Director of the Francis H. Burr Proton Therapy Center at Massachusetts General Hospital (MGH) in Boston. He also is Co-Director of the Sarcoma Center and Associate Professor of Radiation Oncology at Harvard Medical School. His primary areas of clinical research interest include bone and soft-tissue sarcomas and charged-particle (proton) radiation therapy.

Dr. DeLaney obtained his B.A. in history at Harvard College in 1978 and his medical degree from Harvard Medical School in 1982. Dr. Herman Suit, who was Chief of Radiation Oncology at MGH while Dr. DeLaney was a Harvard medical student, was a major influence in his decision to enter the field of radiation oncology. After completing his internship at the Yale-New Haven Hospital in general surgery, Dr. DeLaney took three years of residency training at MGH in radiation oncology, completing training in 1986. He then spent six years as a senior investigator at the National Cancer Institute, where his interests were in the treatment of adult and pediatric sarcomas and translational research in photodynamic therapy with light-activated dyes. He returned to Boston in 1992 to become Chief of Radiation Oncology at Boston University Medical Center, where he also established a joint residency program in conjunction with MGH. He returned to MGH to head the sarcoma service in the Department of Radiation Oncology in 2000 and became medical director of the Proton Therapy Center in 2001. He is a co-editor of *Proton and Heavier Charged Particle Radiotherapy* (Lippincott, 2007).

George D. Demetri, M.D.
Dr. George Demetri received an undergraduate degree in biochemistry from Harvard University, followed by a Rotary Foundation Fellowship at the Université de Besancon, France. In 1983, Dr. Demetri received his medical degree from Stanford University School of Medicine. After completing an internal medicine residency and chief residency at the University of Washington Hospitals in Seattle, he pursued a fellowship in medical oncology at the Dana-Farber Cancer Institute and Harvard Medical School, where he has served as an attending physician since 1989.

Dr. Demetri's research and clinical interests have focused on the development of targeted therapies for sarcomas and other solid tumors, with a particular emphasis on molecularly defined disease subtypes, such as gastrointestinal stromal tumors. The contributions of Dr. Demetri and his colleagues at Dana-Farber/Harvard Cancer Center have led to the development of oral "smart drugs," such as Gleevec, Sutent, and other new therapies in development. Dr. Demetri serves as co-chair of the Medical Advisory Board for the Sarcoma Foundation of America and is a member of the Executive Committee of the Sarcoma Alliance for Research through Collaboration. With an interest in Internet-based patient support, he also serves on the Medical Advisory Board of American Society of Clinical Oncology's educational public site (www.cancer.net) and founded a nonprofit patient education website (www.sarcoma.net). Linking Dana-Farber to major research centers across the world, Dr. Demetri serves as Executive Director for Clinical and Translational Research in the Ludwig Institute for Cancer Research, the largest privately funded philanthropic organization supporting cancer research.

Soldano Ferrone, M.D., Ph.D.
Dr. Soldano Ferrone is a member of the Roswell Park Cancer Institute. Dr. Ferrone has focused his research program on the development and application of antibody-based immunotherapy targeting the human high–molecular weight melanoma-associated antigen (HMW-MAA). He has shown that HMW-MAA–specific antibodies have an antitumor effect both in animal model systems and in patients with melanoma.

Brian Harfe, Ph.D.
Dr. Brian Harfe's undergraduate degree is from the University of Glasgow in Scotland. After traveling across Europe, he returned to the United States and began a Ph.D. at Johns Hopkins University in 1993. His thesis was on muscle development in the nematode *Caenorhabditis elegans* in the laboratory of Dr. Andrew Fire (2006 Nobel Prize winner for his discovery of RNAi). After obtaining his Ph.D. in 1998, Dr. Harfe moved to Emory University and began a postdoctoral position in the laboratory of Dr. Sue Jinks-Robertson working on DNA-damage pathways in yeast. In 2000, he moved to Boston, where he began a second postdoctoral position in the laboratory of Dr. Cliff Tabin at Harvard Medical School working on the molecular pathways responsible for limb formation using the mouse and chick model systems. Since 2003, Dr. Harfe has been an Assistant Professor in the Molecular Genetics and Microbiology Department at the University of Florida College of Medicine in Gainesville.

Francis Hornicek, M.D. Ph.D.
Dr. Francis Hornicek is Chief of the Orthopaedic Oncology Unit at Massachusetts General Hospital and an Associate Professor in the Department of Orthopaedic Surgery at Harvard Medical School. Dr. Hornicek received his M.D. from the University of Pittsburgh School of Medicine and his Ph.D. from Georgetown University School of Medicine.

Amin B. Kassam, M.D., FRCS(C)
Dr. Amin Kassam completed his medical and undergraduate education at the University of Toronto and his residency and fellowship training at the University of Ottawa. He pursued additional postgraduate training in epidemiology and clinical outcomes. Dr. Kassam joined the faculty of the Department of Neurological Surgery at the University of Pittsburgh in February 1998. He spent the next year focusing on microvascular surgery. Dr. Kassam has performed more than 1,000 microvascular decompression procedures for cranial nerve neuropathy and has provided a unique perspective by using the endoscope to visualize and enhance difficult regions. Since his appointment, he has also focused on building a collaborative center to provide comprehensive care for complex pathology of the skull base. This center builds on the strength of combining the talents of surgeons from multiple specialties, allowing for the use of proven conventional approaches in conjunction with new minimally invasive endoscopic approaches to provide safe and effective treatment for patients. This work culminated in the development of the multidisciplinary Minimally Invasive Neurosurgical Center (MINC).

Dr. Kassam along with Dr. Carl Snyderman and Dr. Ricardo Carrau were directly involved with the development of the expanded endonasal approach (EEA). This approach represents an entirely new paradigm to remove complex lesions of the skull base and brain without incisions. The center, under the direction of Dr. Kassam, has pioneered and developed much of the technology and instrumentation used during EEA surgeries. With continued research and experience, he now uses the EEA surgery for most tumors affecting the skull base. Dr. Kassam has performed more than 3,000 neurosurgical procedures, including more than 700 minimally invasive endoscopic procedures. Dr. Kassam remains active in cerebrovascular surgery and has helped develop a program to better understand the genetic alterations that lead to the development of intracranial aneurysms.

In July 2006, Dr. Kassam was named Interim Chairman of the Department of Neurological Surgery. Since then, he has focused on increasing interdisciplinary activities among neurosurgery and radiology, medical, radiation and surgical oncology, anesthesiology, neurology, and otolaryngology. It is hoped that these cooperative ventures will lead to new innovations in care for patients with a variety of neurologic abnormalities. Dr. Kassam has more than 80 peer-reviewed publications, an additional 11 book chapters currently published or in press, and is funded by industry and NIH. He lectures nationally and internationally on surgery of the cranial nerves and skull base and on minimally invasive endoscopic techniques.

Michael Kelley, M.D.
Dr. Michael Kelley received his M.D. from the University of Michigan School of Medicine in 1985. He completed internal medicine residency training at Duke University in 1988 and completed training at the National Cancer Institute, including postdoctoral work in the laboratory of Dr. Stuart Aaronson and a clinical fellowship in medical oncology. He is currently Associate Professor of Medicine at Duke University and Chief of Hematology/Oncology at the Durham Veterans Affairs Hospital.

Dr. Kelley's primary research interest is development of molecularly targeted agents for prevention and treatment of cancer, especially in lung cancer. His interest in chordoma began more than a decade ago in collaboration with Dr. Dilys Parry and others in pursuit of familial chordoma gene(s).

Robert Maki, M.D., Ph.D.
Dr. Robert Maki is Co-Director of the Adult Sarcoma Program and Associate Member of the Department of Medicine at Memorial Sloan-Kettering Cancer Center in New York. His research focuses on novel drug development and clinical trial design for patients with sarcomas of soft tissue and bone. He is a graduate of Northwestern University in the Integrated Science Program and has an M.D. and Ph.D. in immunology from Cornell University Medical College.

Claudia Palena, Ph.D.
Dr. Claudia Palena received her B.S. and Ph.D. in biochemistry from the National University of Rosario, Argentina, in 2000 and did her postdoctoral training in the Laboratory of Tumor Immunology and Biology at the National Cancer Institute under the direction of Dr. Jeffrey Schlom. She is currently a Staff Scientist in the Laboratory of Tumor Immunology and Biology and serves as the Head of the Immunoregulation Group. Dr. Palena's research in tumor immunology focuses on the use of co-stimulation to enhance antitumor T-cell responses, the study of mechanisms to improve the potency of antigen-presenting cells for optimal T-cell activation, and the identification of novel tumor antigens that could be used to elicit immune responses against cancer cells.

Nadege Presneau, Ph.D.
Dr. Nadege Presneau completed her Ph.D. training in France at the University Blaise Pascal on predisposition to breast and ovarian cancer in French families. She did a postdoctoral training at McGill University in Montreal working on identifying a new tumor suppressor gene in sporadic ovarian cancer with Dr. Patricia Tonin. Then she moved to London to be a research fellow working on the molecular signature of sarcoma with Dr. Chris Boshoff and Dr. Adrienne Flanagan. She is now a Senior Research Fellow working on bone tumours with Dr. Flanagan.

Vijaya Ramesh, Ph.D.
Dr. Vijaya Ramesh is an Associate Professor of Neurology (Genetics) at Harvard Medical School and an Associate Neurologist at Massachusetts General Hospital (MGH). She obtained her Ph.D. from the University of Madras, India, and completed postdoctoral training in the laboratories of Dr. James Gusella and Dr. Vivian Shih at MGH. She is a member of the Molecular Neurogenetics

Unit and the Center for Human Genetic Research at MGH. She also directs a Monoclonal Antibody Core.

Dr. Ramesh's laboratory investigates tumor suppressor genes and their functions, particularly tumor suppressors related to neurofibromatosis 2 (NF2) and tuberous sclerosis 1 and 2 (TSC). The NF2 protein merlin is a cytoskeletal-associated protein with a variety of functions. Her laboratory is working on understanding merlin's functions through two of its interacting partners, NHERF and Magicin. NHERF is a multifunctional adaptor protein that links various ion channels and receptors to the actin cytoskeleton through merlin and its related family members ezrin, radixin, and moesin (ERMs). Magicin, a novel cytoskeletal protein, appears to have an essential role in signaling to the actin cytoskeleton as well as in transcription regulation.

Tuberous sclerosis complex (TSC), an autosomal dominant disease caused by mutations in either TSC1 or TSC2, is characterized by the development of hamartomas in a variety of organs. Dr. Ramesh's laboratory has shown distinct activation of Akt/MAPK pathways in the central nervous system tumors of TSC patients, which may contribute to some of the neurological manifestations seen in TSC. Their most recent work documents the binding of TSC2 protein tuberin with Pam, a huge protein originally identified as associated with c-My. Dr. Ramesh's laboratory is investigating whether Pam functions as an E3 ubiquitin ligase, regulating the stability of the TSC proteins and other key signaling molecules in neurons, thus playing a role in synaptic plasticity.

Josh Sommer

Mr. Josh Sommer is Vice President of the Chordoma Foundation, which he co-founded along with his mother, Dr. Simone Sommer, after he was diagnosed with a clival chordoma in 2006. He believes that patients should play an active role in bringing about treatments for their own conditions and that patients represent a largely untapped source of funding, energy, and know-how in the treatment development process. Mr. Sommer is currently a junior at Duke University, where he is a Trinity Scholar. He is conducting research in the laboratory of Dr. Michael Kelley to understand the unique biology of chordoma and search for therapeutic targets. His investigations include gene-expression microarray analysis, candidate gene knockdown using RNAi, *in vitro* drug screening, and analysis of known oncogenic pathways in chordoma. In high school, Mr. Sommer received numerous honors and awards, including the *USA Today* All-USA Academic First Team Award, Prudential Spirit of Community Award, Coca-Cola Scholarship, and AXA Achievement National Award.

Simone Sommer, M.D., M.P.H.

Dr. Simone Sommer is the President and Treasurer of the Chordoma Foundation, which she formed after her son and best friend, Josh, was diagnosed with a chordoma in 2006. Dr. Sommer is dedicated to improving the quality of life for people affected by chordomas and has devoted her full-time efforts to bringing about effective treatments and ultimately a cure for this disease. Under her direction, the Chordoma Foundation has initiated numerous collaborative research projects with scientists and physicians at institutions across the world.

Dr. Sommer's goal is to have the Chordoma Foundation serve as the focal point for a coordinated international chordoma research effort.

Dr. Sommer received her M.D. from George Washington School of Medicine and her master's degree in public health in epidemiology from the University of North Carolina School of Public Health. She was formerly Associate Clinical Professor at the University of North Carolina at Chapel Hill in the Department of Family Medicine and served as Medical Director of the Guilford County Health Department Chronic Disease Prevention Program. She is past president of Sommer Health Services of Greensboro, North Carolina, which delivered comprehensive on-site corporate health promotion, disease prevention, and targeted interventions for self-insured companies.