Framework for Developing a New Taxonomy of Disease

Committee

Susan Desmond-Hellmann

Co-Chair

Dr. Susan Desmond-Hellmann, MD, MPH, is chancellor of the University of California, San Francisco. She assumed the post August 3, 2009.

UCSF is a leading university dedicated to promoting health worldwide through advanced biomedical research, graduate-level education in the life sciences and health professions, and excellence in patient care. UCSF is the only campus in the 10-campus UC system devoted exclusively to the health sciences.

Desmond-Hellmann previously served as president of product development at Genentech, a position she held from March 2004 through April 30, 2009. In this role, she was responsible for Genentech's pre-clinical and clinical development, process research and development, business development and product portfolio management. She also served as a member of Genentech's executive Committee, beginning in 1996. She joined Genentech in 1995 as a clinical scientist, and she was named chief medical officer in 1996. In 1999, she was named executive vice president of development and product operations. During her time at Genentech, several of the company's patient therapeutics (Lucentis, Avastin, Herceptin, Tarceva, Rituxan and Xolair) were approved by the U.S. Food and Drug Administration, and the company became the nation's No. 1 producer of anti-cancer drug treatments.

She completed her clinical training at UCSF and is board-certified in internal medicine and medical oncology. She holds a bachelor of science degree in pre-medicine and a medical degree from the University of Nevada, Reno, and a master's degree in public health from the University of California, Berkeley.

Prior to joining Genentech, Desmond-Hellmann was associate director of clinical cancer research at Bristol-Myers Squibb Pharmaceutical Research Institute. While at Bristol-Myers Squibb, she was the project team leader for the cancer-fighting drug Taxol.

Desmond-Hellmann also has served as associate adjunct professor of epidemiology and biostatistics at UCSF. During her tenure at UCSF, she spent two years as visiting faculty at the Uganda Cancer Institute, studying HIV/AIDS and cancer. She also spent two years in private practice as a medical oncologist before returning to clinical research.

In January 2009, Desmond-Hellmann joined the Federal Reserve Bank of San Francisco's Economic Advisory Council for a three-year term. In July 2008, she was appointed to the California Academy of Sciences board of trustees. Dr. Desmond-Hellmann was named to the Biotech Hall of Fame in 2007 and as the Healthcare Businesswomen's Association Woman of the Year for 2006. She was listed among Fortune magazine's "top 50 most powerful women in business" in 2001 and from 2003 to 2008. In 2005 and 2006, the Wall Street Journal listed Desmond-Hellmann as one of its "women to watch." From 2005 to 2008, Desmond-Hellmann served a three-year term as a member of the American Association for Cancer Research board of directors, and from 2001 to 2009, she served on the executive Committee of the board of directors of the Biotechnology Industry Organization. She served on the corporate board of Affymetrix from 2004-2009.

Charles Sawyers

Co-Chair

Dr. Charles Sawyers, M.D., is an Investigator of the Howard Hughes Medical Institute and the inaugural Director of the Human Oncology and Pathogenesis Program (HOPP) at Memorial Sloan-Kettering Cancer Center (MSKCC), where he is building a program of lab-based translational researchers across various clinical disciplines and institutional infrastructure to enhance the application of global genomics tools to clinical trials.

Sawyers' laboratory is currently focused on characterizing signal transduction pathway abnormalities in prostate cancer, with an eye toward translational implications. His research is best demonstrated through his earlier studies of BCR-ABL tyrosine kinase function in chronic myeloid leukemia, his work with Brian Druker and Novartis in the development of the kinase inhibitor imatinib/Gleevec as primary therapy for CML, and his discovery that imatinib resistance is caused by BCR-ABL kinase domain mutations. This discovery led Dr. Sawyers to evaluate second generation Abl kinase inhibitors, such as the dual Src/Abl inhibitor dasatinib, which received fast track approval at the FDA in June 2006.

Dr. Sawyers' work in prostate cancer has defined critical signaling pathways for disease initiation and progression through studies in mouse models and humane tissues. This preclinical work led to the development of a novel antiandrogen MVD3100, a small molecule inhibitor discovered in collaboration with UCLA Chemist Michael Jung, which targets the increased levels of androgen receptor found in the hormone refractory disease. Based on impressive clinical results in a phase I/II study, MDV3100 is currently in phase III registration trial. Dr. Sawyers is past President of the American Society of Clinical Investigation and served on the National Cancer Institute's Board of Scientific Councilors. He has won numerous honors and awards, including: the Richard and Hinda Rosenthal Foundation Award; the Dorothy Landon Prize from the American Association of Cancer Research and the David A. Karnofsky Award from the American Society of Clinical Oncology; and the 2009 Lasker DeBakey Clinical Medical Research Award. He is a member of the Institute of Medicine and in 2010 was elected to the National Academy of Sciences.

David R. Cox

Member

Dr. David R. Cox M.D. Ph.D. serves as Chief Scientific Officer for the Applied Quantitative Genotherapeutics Unit of Pfizer's Worldwide Research & Development. This new unit brings together human genetics, systems biology, and cell biology, combining internal capabilities with outside collaborations, to focus on increasing preclinical target validation with the aim of significantly improving clinical survival. David is a co-founder of Perlegen, and was most recently Chief Scientific Officer of the company since its formation in 2000. David was Professor of Genetics and Pediatrics at the Stanford University School of Medicine as well as the co-director of the Stanford Genome Center. He obtained his A.B. and M.S. degrees from Brown University in Rhode Island and his M.D. and Ph.D. degrees from the University of Washington, Seattle. He completed a Pediatric Residency at the Yale-New Haven Hospital in New Haven, Connecticut and was a Fellow in both genetics and pediatrics at the University of California, San Francisco. David is certified by the American Board of Pediatrics and the American Board of Medical Genetics. He was an active participant in the large scale mapping and sequencing efforts of the Human Genome Project while carrying out research involving the molecular basis of human genetic disease. David has been a member of several commissions and boards, including the National Bioethics Advisory Commission (NBAC) and the Health Sciences Policy Board of the Institute of Medicine. He has also served on a number of international Committees, including the Council of the Human Genome Organization (HUGO). He has authored over 100 peer-reviewed scientific publications and has served on numerous editorial boards. Dr. Cox's honors include election to the Institute of Medicine of the National Academy of Sciences.

Claire M. Fraser

Member

Dr. Claire M. Fraser-Liggett is Director of the Institute for Genome Sciences and a Professor of Medicine at the University Of Maryland School Of Medicine in Baltimore, Maryland. Previously she was the President and Director of The Institute for Genomic Research in Rockville, Maryland. Dr. Fraser-Liggett has played a role in the sequencing and analysis of human, animal, plant and microbial genomes to better understand the role that genes play in development, evolution, physiology and disease. She led the teams that sequenced the genomes of several microbial organisms, including important human and animal pathogens, and as a consequence helped to initiate the era of comparative genomics. She has served on a number of National Research Council Committees on counter-bioterrorism, domestic animal genomics, polar biology, and metagenomics. Dr. Fraser-Liggett has more than 220 scientific publications, and has served on Committees of the National Science Foundation, Department of Energy and National Institutes of Health. She received her PhD in pharmacology from State University of New York at Buffalo.

Stephen J. Galli

Member

Stephen J. Galli received his BA and MD from Harvard, in 1968 and 1973, respectively, and completed a residency and chief residency in Anatomic Pathology at Massachusetts General Hospital (MGH) in 1977. After postdoctoral work with Harold F. Dvorak at MGH, he served on the faculty at Harvard Medical School from 1979 until 1999, when he moved to Stanford as Chair of the Department of Pathology, Chief of Pathology at Stanford Hospital & Clinics, Professor of Pathology and of Microbiology and Immunology, and the Mary Hewitt Loveless, MD Professor. He is also Co-Director of the Stanford Center for Genomics and Personalized Medicine. Dr. Galli's research focuses on the development and function of mast cells and basophils (key players in anaphylaxis, allergies, asthma and many other biological responses), and on developing new animal models to study the diverse roles of these cells in health and disease. Dr. Galli serves on the editorial boards of several medical journals and is a co-editor of The Annual Review of Pathology: Mechanisms of Disease. He received a MERIT Award from the National Institutes of Health (1995), Scientific Achievement Awards from the International Association of Allergy & Clinical Immunology (1997) and the World Allergy Organization (2011), and is an Honorary Fellow of the College of American Pathologists. He was President of the American Society for Investigative Pathology (2005-2006) and has been elected to the Pluto Club (Association of University Pathologists), the Collegium Internationale Allergologicum (he began a four year term as President in 2010), the American Society for Clinical Investigation, the Association of American Physicians, the Institute of Medicine of the National Academies (USA), and the Accademia Nazionale dei Lincei (the National Academy of the Lynxes) in Rome, considered the oldest secular scientific society in the Western world. In 2006-2007, the last year of a three year elected term, Dr. Galli was the Chair of the Advisory Board to the President and Provost of Stanford University.

David B. Goldstein

Member

Dr. David B. Goldstein is currently Professor of Molecular Genetics & Microbiology and Director of the Center for Human Genome Variation at Duke University. He received his Ph.D. in Biological Sciences from Stanford University in 1994, and from 1999 to 2005 was Wolfson Professor of Genetics at University College London. Dr. Goldstein is the author of over 150 scholarly publications in the areas of population and medical genetics. His work focuses on the genetics of human disease and treatment response, with a concentration on neuropsychiatric disease and host determinants of response to infectious diseases. He is the recipient of one of the first seven nationally awarded Royal Society / Wolfson research merit awards in the UK for his work in human population genetics and was awarded the Triangle Business Journal Health Care Heroes Award in 2008 for his work on host determinants of control of HIV-1. Most recently, he was appointed the co-chair and chair of the Gordon Research Conference meeting on human genetics and genomics for 2011 and 2013.

David J. Hunter

Member

Dr. David J. Hunter is currently the Dean for Academic Affairs at the Harvard School of Public Health and the Vincent L. Gregory Professor in Cancer Prevention in the Departments of Epidemiology and Nutrition. His research interests include cancer epidemiology and molecular and genetic epidemiology. Dr. Hunter analyzes inherited susceptibility to cancer and other chronic diseases using molecular techniques and studying molecular markers of environmental exposures. He is Co-Chair of the NCI Breast and Prostate Cancer Cohort Consortium and Co-Director of the NCI Cancer Genetic Markers of Susceptibility (CGEMS) Special Initiative.

Isaac S. Kohane

Member

Isaac (Zak) S. Kohane is the director of the Children's Hospital Informatics Program and is the Henderson Professor of Pediatrics and Health Sciences and Technology at Harvard Medical School (HMS). He is also co-director of the HMS Center for Biomedical Informatics and Director of the HMS Countway Library of Medicine. Dr. Kohane leads multiple collaborations at Harvard Medical School and its hospital affiliates in the use of genomics and computer science to study diseases (particularly cancer and autism). He has developed several computer systems to allow multiple hospital systems to be used as "living laboratories" to study the genetic basis of disease while preserving patient privacy. Among these, the i2b2 (Informatics for Integrating Biology and the Bedside) National Computing Center has been deployed at over 52 academic health centers internationally.

Dr. Kohane has published over 180 papers in the medical literature and authored a widely used book on microarrays for Integrative Genomics. He has been elected to multiple honor societies including the American Society for Clinical Investigation, the American College of Medical Informatics, and the Institute of Medicine. He leads a doctoral program in genomics and bioinformatics at the Division of Health Sciences and Technology at Harvard and MIT. He is also a practicing pediatrics endocrinologist and father of three energetic children.

Manuel Llinas

Member

Dr. Manuel Llinás is an Assistant Professor of Molecular Biology and a member of the Lewis-Sigler Institute for Integrative Genomics at Princeton University. Dr. Llinás earned a Ph.D. in molecular and cell biology from the University of California-Berkeley and did postdoctoral work in the lab of Joseph DeRisi at the University of California-San Francisco. He joined the Princeton faculty in 2005. Dr. Llinás' laboratory studies the deadliest of the four human Plasmodium parasites, Plasmodium falciparum. His research combines tools from functional genomics, molecular biology, computational biology, biochemistry, and metabolomics to understand the fundamental molecular mechanisms underlying the development of this parasite. The focus is predominantly on the red blood cell stage of development, which is the stage in which all of the clinical manifestations of the malaria disease occur. His research has focused on two major areas: the role of transcriptional regulation in orchestrating parasite development, and an in-depth characterization of the malaria parasite's unique metabolic network. On the transcription side, Dr. Llinás' lab works on the characterization of the first family of DNA binding proteins to be identified in the Plasmodium falciparum genome, the Apicomplexan AP2 (ApiAP2) proteins. The metabolomics work has begun to identify unique biochemical pathway architectures in the parasite including a novel branched TCA cycle. These two approaches explore relatively virgin areas in the malaria field with the goal of identifying novel strategies for therapeutic intervention.

Bernard Lo

Member

Bernard Lo, M.D., is Professor of Medicine and Director of the Program in Medical Ethics at UCSF. He is also National Program Director for the Greenwall Faculty Scholars Program in Bioethics, a career development award for bioethics researchers. He directs the Regulatory Knowledge Support Component of the NIH-funded Clinical and Translational Science Institute at UCSF and is co-Director of the Policy and Ethics Core of the Center for AIDS Prevention Studies. He chairs the UCSF Stem Cell Research Oversight Committee. He is co-chair of the Standards Working Group of the California Institute of Regenerative Medicine, which recommends regulations for stem cell research funded by the state of California. He is a member of the Centers for Disease Control and Prevention (CDC) Ethics SubCommittee of the Advisory Committee to the Director. He serves on DSMBs for NIH-sponsored HIV vaccine trials and for the Long-Term Oxygen Treatment Trial (LOTT) and on the Ethics Working Group of the HIV Prevention Trials Network. He also serves on the Board of Directors of the Association for the Accreditation of Human Research Protection Programs. He is a member of the Institute of Medicine (IOM), served on the IOM Council and as chair of the IOM Board on Health Sciences Policy. He chaired a 2009 IOM Committee on conflicts of interest in medicine and several earlier reports. Dr. Lo is author of Resolving Ethical Dilemmas: A Guide for Clinicians (4th ed., 2010) and of Ethical Issues in Clinical Research (2010).

Tom Misteli

Member

Dr. Tom Misteli is a Senior Investigator and Head of the Cell Biology of Genomes group at the National Cancer Institute, NIH. Dr. Misteli obtained his PhD from the University of London, UK, and joined the NCI after postdoctoral work at the Cold Spring Harbor Laboratory, New York. He has pioneered the field of genome cell biology by developing live-cell microscopy approaches to study the nuclear organization of the genome and gene expression in intact cells, and his laboratory aims to apply this knowledge to the development of novel diagnostic and therapeutic strategies for cancer and aging. Dr Misteli has received numerous awards for his work, and currently serves as Editor-in-Chief of The Journal of Cell Biology and of Current Opinion in Cell Biology.

Sean J. Morrison

Member

Dr. Sean J. Morrison, Ph.D. is an HHMI investigator as well as Henry Sewall Professor in Medicine at the University of Michigan Medical School, Professor of Internal Medicine and Cell and Developmental Biology at the University of Michigan, Research Professor at the University of Michigan Life Sciences Institute, and Director of the University of Michigan Center for Stem Cell Biology. He received his Ph.D. in Immunology at Stanford University. Dr. Morrison began his pioneering stem cell work only after a brief stint as a biotech entrepreneur. As a graduate student, Morrison identified key markers that distinguish hematopoietic stem cells, which give rise to blood and immune system cells, from other immature hematopoietic cells. He determined that stem cells are fundamentally different from other immature cells, and his results also suggested that certain factors are involved in regulating stem cell self-renewal. Later, as a postdoctoral fellow in the Caltech laboratory of David Anderson, a fellow HHMI investigator, Morrison became the first to isolate uncultured neural crest stem cells, which give rise to the peripheral nervous system. This led to his discovery that stem cells persist throughout adult life in the peripheral nervous system, where they were not previously believed to exist.

Today, Morrison's research focuses on neural stem cells and hematopoietic stem cells. By studying both, he hopes to understand the extent to which mechanisms that control self-renewal and other critical functions are conserved among stem cells in different tissues. Morrison has discovered that there are mechanistic differences between the self-renewal of normal stem cells and the proliferation of cancer cells that can be exploited to kill the cancer cells without harming the normal stem cells. He also has traced a potentially fatal birth defect that causes Hirschsprung disease to defects in the generation and migration of neural crest stem cells in the developing intestines. And, using techniques he developed as a graduate student, Morrison identified a family of cell surface receptors that scientists can use to separate hematopoietic stem cells from other, less primitive, hematopoietic progenitors. Each of these studies has the potential to change the way in which patients are treated. Ultimately, Morrison hopes to identify new treatments for diseases caused by stem cell defects, including cancer, degenerative disease, and birth defects.

David G. Nichols

Member

Dr. Nichols is a professor of anesthesiology/critical care medicine and pediatrics and the Mary Wallace Stanton Professor of Education. Since joining the School of Medicine faculty in 1984, he has held numerous leadership posts in both the Department of Anesthesiology and Critical Care Medicine and school-wide. Named vice dean for education in 2000, Dr. Nichols oversees undergraduate, graduate, residency, postdoctoral and continuing medical education programs, as well as the Welch Medical Library. He has led a wide variety of significant initiatives to improve the school of medicine's innovative use of technology in education; update the medical school's curriculum; improve faculty development by revising tenure and promotion guidelines; restructure graduate medical education; oversee the design of a new $50 million medical education building; and enhance diversity throughout Johns Hopkins Medicine.

From 1984 to 1987, Dr. Nichols was associate director of the residency education program in the Department of Anesthesiology and Critical Care Medicine. He became director of the Division of Pediatric Critical Care and of the pediatric intensive care unit (PICU) in 1988. The division was merged with pediatric anesthesiology under Dr. Nichols' leadership in 1997. During this period, he trained and mentored more than 50 postdoctoral fellows, many of whom now are professors or directors of PICUs in the United States and abroad. Dr. Nichols became a full professor of anesthesiology/critical care medicine and pediatrics in 1998 and became the recipient of the Mary Wallace Stanton Professorship for Education in 2005. He has written more than 80 professional journal articles and abstracts, held 17 guest professorships, headed more than 20 symposia and delivered more than 115 guest lectures. He also has been editor in chief of the leading textbooks in pediatric critical care medicine and edited Rogers Textbook of Pediatric Intensive Care and Critical Heart Disease in Infants and Children.

Maynard V. Olson

Member

Dr. Maynard Olson is Professor Emeritus of Medicine and Genome Sciences, at the University of Washington. He received his PhD in Chemistry at Stanford University in 1970 and a BS in Chemistry from the California Institute of Technology in 1965. His research interests focus on studies of natural genetic variation in both bacteria and humans. This research involves activities in human genetics, genomics, molecular genetics, analytical biochemistry, and computational biology. Dr. olson has a special interest in interdisciplinary research, particularly at the interfaces between chemistry, computer science and biology.

Dr. Olson was involved in shaping scientific policy toward the Human Genome Project, serving on the National Research Council Committee on Mapping and Sequencing the Human Genome, the Program Advisory Committee of the National Center for Human Genome Research Institute. In recognition of his research in genetics and genomics, he received the Genetics Society of America Medal in 1992, the City of Medicine Award in 2000, the Gairdner International Award in 2002, and the Gruber Prize in Genetics in 2007.

Charmaine Royal

Member

Dr. Charmaine Royal is an Associate Research Professor in the Institute for Genome Sciences & Policy and the Department of African and African American Studies at Duke University. She received her MS in Genetic Counseling and PhD in Human Genetics from Howard University. She subsequently completed her postdoctoral training in the Bioethics and Special Populations Research Program at the National Human Genome Research Institute of the National Institutes of Health, and in the Division of Epidemiology and Behavioral Medicine at the Howard University Cancer Center.

Prior to joining the Duke faculty in 2007, Dr. Royal was Assistant Professor of Pediatrics and Director of the GenEthics Unit in the National Human Genome Center at Howard University. She serves on the: Bioethics Advisory Committee of the March of Dimes Foundation; Social Issues Committee of the American Society of Human Genetics; Editorial Board of the American Journal of Bioethics; and various other professional Committees and boards.

Dr. Royal’s research and scholarship focus primarily on ethical, psychosocial, societal, and biomedical issues at the intersection of genetics/genomics and concepts of “race”, ancestry, ethnicity, and identity. Her specific interests include genetic variation and the (re)conceptualization of race, use of race and ancestry in research and clinical practice, gene-environment interactions in health and health disparities, genetic ancestry inference, involvement of historically marginalized and underrepresented groups in genetic and genomic research, and genomics and global health. She has taught, presented, published, and received funding in these and other related areas. A key objective of her research program is to advance a more holistic and ethical approach to understanding and improving human health and well-being through increased integration of genetic and genomic research with behavioral, social science, and humanities research.

Keith R. Yamamoto

Member

Dr. Keith Yamamoto, Ph.D., is Professor of Cellular and Molecular Pharmacology and Executive Vice Dean of the School of Medicine at the University of California, San Francisco. He has been a member of the UCSF faculty since 1976, serving as Director of the PIBS Graduate Program in Biochemistry and Molecular Biology (1988-2003), Vice Chair of the Department of Biochemistry and Biophysics (1985-1994), Chair of the Department of Cellular and Molecular Pharmacology (1994-2003), and Vice Dean for Research, School of Medicine (2002-2003). Dr. Yamamoto's research is focused on signaling and transcriptional regulation by intracellular receptors, which mediate the actions of several classes of essential hormones and cellular signals; he uses both mechanistic and systems approaches to pursue these problems in pure molecules, cells and whole organisms. Dr. Yamamoto was a founding editor of Molecular Biology of the Cell, and serves on numerous editorial boards and scientific advisory boards, and national Committees focused on public and scientific policy, public understanding and support of biological research, and science education; he chairs the Coalition for the Life Sciences (formerly the Joint Steering Committee for Public Policy) and for the National Academy of Sciences, he chairs the Board on Life Sciences. Dr. Yamamoto has long been involved in the process of peer review and the policies that govern it at the National Institutes of Health, serving as Chair of the Molecular Biology Study Section, member of the NIH Director's Working Group on the Division of Research Grants, Chair of the Advisory Committee to the NIH Center for Scientific Review (CSR), member of the NIH Director's Peer Review Oversight Group, member of the CSR Panel on Scientific Boundaries for Review, member of the Advisory Committee to the NIH Director, Co-Chair of the Working Group to Enhance NIH Peer Review, and Co-Chair of the Review Committee for the Transformational R01 Award. Dr. Yamamoto was elected as a member of the American Academy of Arts and Sciences in 1988, the National Academy of Sciences in 1989, the Institute of Medicine in 2003, and as a fellow of the American Association for the Advancement of Sciences in 2002.