

Newborn Screening in the Genomic Era: Setting a Research Agenda

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Presenter and Moderator Biographies



Arthur Beaudet, M.D., received his medical degree from Yale University, did pediatric residency training at Johns Hopkins Hospital, and was a research associate at the National Institutes of Health before joining Baylor College of Medicine (BCM) where he has remained to the present. Dr. Beaudet has made diverse contributions in the field of mammalian genetics, including the discovery of uniparental disomy in humans and publishing over 250 original research articles. He has argued for the importance of epigenetics in human disease since 2002 (PMID: 12394355) and proposed a mixed epigenetic and genetic and mixed *de novo* and inherited model for oligogenic inheritance in autism (PMID:

15389703). In 2004, Dr. Beaudet and a BCM team of investigators were the first in the U.S. to introduce array comparative genomic hybridization into the clinical lab, and they have gone on to play a leadership role in the transformative impact of this technology on clinical genetics. His current work is focused on the role of genomic copy number abnormalities, epigenetic changes in neurobehavioral disabilities, and especially on the importance of the *CHRNA7* gene in mental retardation, autism, and schizophrenia. Dr. Beaudet is currently the Henry and Emma Meyer Distinguished Service Professor and Chair of the Department of Molecular and Human Genetics at BCM.



Susan A. Berry, M.D., is professor of pediatrics and genetics, cell biology, and development at the University of Minnesota. She is the director of the Division of Genetics and Metabolism in the Department of Pediatrics and the Institute of Human Genetics. She received a bachelor's degree in biochemistry from Rice University in 1975 and an M.D. from the University of Kansas in 1978. She did her residency in pediatrics (1978–1981) and fellowship in medical genetics (1981–1984) at the University of Minnesota and has been on the faculty there since 1984. Like many genetics professionals, she sees adults and children with genetic conditions of all kinds. She has a particular interest in providing management for persons with inborn errors of metabolism and has a

longstanding interest in improvement in their care through early diagnosis and treatment. She is the co-leader of the Region 4 Genetics Collaborative's Priority 2 (long-term follow-up) Project implementing the Inborn Errors of Metabolism Information System.



Jeffrey R. Botkin, M.D., M.P.H., is a professor of pediatrics at the University of Utah and an adjunct professor of human genetics. He is chief of the Division of Medical Ethics and Humanities in the Department of Internal Medicine. He obtained his B.A. from Princeton University, his M.D. from the University of Pittsburgh, and his M.P.H. from Johns Hopkins University. Dr. Botkin is the associate vice president for research integrity at the University of Utah with oversight responsibilities for the institutional review board, conflict of interest, responsible conduct of research, biosafety, and research ethics education. His research and publications are focused on the ethical, legal, and social implications of genetic technology with a particular emphasis on research ethics, genetic testing for cancer susceptibility, newborn screening, and prenatal diagnosis. Dr. Botkin formerly was chair of the Committee on Bioethics for the American Academy of Pediatrics and a member of the Secretary's Advisory Committee on Human Research Protections at the U.S. Department of Health and Human Services. Dr. Botkin is currently a member of the Secretary's Advisory Committee on Heritable Diseases in Newborns and Children. He chairs the National Institutes of Health Embryonic Stem Cell Working Group and is a member of the U.S. Food and Drug Administration's Pediatric Ethics Advisory Committee. Dr. Botkin is an elected fellow of the Hastings Center.



Wendy Chung, M.D., Ph.D., is a clinical and molecular geneticist who directs the clinical genetics program at Columbia University and performs human genetic research. She is currently the Herbert Irving assistant professor of pediatrics and medicine. She received her B.A. in biochemistry and economics from Cornell University, her M.D. from Cornell University Medical College, and her Ph.D. in genetics from The Rockefeller University.

Clinically, she directs programs in genetic risk assessment for cancer, cardiomyopathy, arrhythmias, and diabetes. She studies families with mysterious undiagnosed genetic conditions and uses the powerful tools of genomic analysis to identify the underlying cause of disease. Dr. Chung directs a clinical molecular diagnostics laboratory and develops novel genetic tests to improve clinical testing, including preimplantation genetic diagnosis to help couples have healthy children by selecting embryos free of genetic disease. For her contributions in this area, she received the Medical Achievement Award from Bonei Olam.

Dr. Chung directs NIH-funded research programs in human genetics of obesity and birth defects, including congenital diaphragmatic hernia and congenital heart disease. She serves as the molecular genetics core for the New York Obesity Research Center and the Diabetes and Endocrine Research Center at Columbia University. She was the recipient of the American Academy of Pediatrics Young Investigator Award, the Medical Achievement Award from Bonei Olam, and a career development award from Doris Duke. She has authored more than 95 peer reviewed articles and 20 chapters and reviews.

Dr. Chung is renowned for her teaching and mentoring. She is a member of the Glenda Garvey Teaching Academy and has won many awards for teaching, including the Charles W. Bohmfalk Award for Distinguished Contributions to Teaching, the American Medical Women's Association Mentor Award, and the Columbia University Presidential Award for Outstanding Teaching.

Dr. Chung also lectures and writes on the legal and social issues raised by genetic testing, disparities in access to genetic care, confidentiality and privacy of genetic information, and gene patents and intellectual property. She is a member of the Columbia Center for BioEthics. Dr. Chung enjoys the challenges of genetics as a rapidly changing field of medicine and strives to facilitate the integration of genetic medicine into all areas of health care in a medically, scientifically, and ethnically sound, accessible, and cost effective manner.



Ronald W. Davis, Ph.D., is considered to be a world leader in biotechnology and the development and application of recombinant DNA and genomic methodology to biological systems. Consequently, his laboratory has developed many of the techniques currently used in academic and industrial biotechnology laboratories. Dr. Davis also is considered to be a world expert in the electron microscopy of nucleic acids and has developed many of the mapping methods. For this, he received the Eli Lilly Award in Microbiology in 1976. His laboratory also was instrumental in the development of lambda vectors, which were commonly used for the primary cloning of DNA molecules in *E. coli*. His laboratory developed many of the yeast vectors and helped to develop yeast as a host for recombinant DNA. For this, Dr. Davis received the United States Steel Award, presented by the National Academy of Sciences, in 1981. In 1983, he became a member of the National Academy of Sciences. He also has helped develop the policy of the United States on recombinant DNA; was a co-signer of the 1973 letter alerting researchers to the potential hazards of recombinant DNA; participated in writing the NIH guidelines for recombinant DNA; and was very active in the downgrading of these guidelines. Dr. Davis participated in the dissemination of recombinant DNA techniques by teaching a course in bacterial genetics at the Cold Spring Harbor Laboratory from 1976 to 1981, and he wrote the first manual on genetic engineering techniques, which was published by the Cold Spring Harbor Laboratory Press. He was a co-author on a publication that first described a new approach for conducting human genetics and for the construction of a human genetic linkage map. For this, he received the Lewis S. Rosenstiel Award for Distinguished Work in Basic Medical Research. His laboratory is now conducting genomic analysis of *Saccharomyces cerevisiae*, for which he received the 2004 Lifetime Achievement Award from the Yeast Genetics Society. His laboratory is developing many new technologies for the genetic, genomic, and molecular analysis of model organisms and humans with a focus on clinical medicine, for which he received the 2004 Sober Award from the American Society for Biochemistry and Molecular Biology.



Geoffrey Duyk, M.D., Ph.D., joined TPG in 2004. Prior to that, he served on the board of directors and was president of research and development at Exelixis, where he led a 550+ person group focused on the discovery and development of small molecule therapeutics. Prior to Exelixis, he was one of the founding scientific staff at Millennium Pharmaceuticals. As Vice President of Genomics at Millennium, Dr. Duyk was responsible for building and leading the informatics, automation, DNA sequencing, and genotyping groups as well as the mouse and human genetics group.

Prior to his tenure at Millennium, Dr. Duyk was an assistant professor at Harvard Medical School (HMS) in the Department of Genetics and assistant investigator of the Howard Hughes Medical Institute. While at HMS, Dr. Duyk was a co-principal investigator in the

National Institutes of Health (NIH) funded Cooperative Human Linkage Center. Dr. Duyk has been and continues to be a member of numerous NIH panels and oversight committees focused on the planning and execution of the Human Genome Project. Dr. Duyk's interests include translational “bench to bedside” research, orphan diseases, and the application of biotechnology to problems of environmental conservation and sustainability.

Dr. Duyk graduated from Wesleyan University (1980) with a bachelor’s degree in biology and was elected to Phi Beta Kappa. He holds a Ph.D. and an M.D. from Case Western Reserve University and completed his medical and fellowship training at the University of California, San Francisco (UCSF). While at UCSF, Dr. Duyk was a fellow of the Lucille P. Markey Foundation and also was awarded a post-doctoral fellowship from the Howard Hughes Medical Institute.

Dr. Duyk serves on the board of directors and/or scientific advisory board of a number of public and private companies as well as advisory boards for a number of orphan disease foundations.



Alan R. Fleischman, M.D., is senior vice president and medical director of the March of Dimes Foundation and clinical professor of pediatrics and epidemiology and population health at the Albert Einstein College of Medicine in New York.

Born in New York City, Dr. Fleischman graduated Phi Beta Kappa from the City College of New York and Alpha Omega Alpha from the Albert Einstein College of Medicine. He continued his education in pediatrics at the Johns Hopkins Hospital in Baltimore, Maryland and completed a fellowship in perinatal physiology at the National Institutes of Health and through a Royal Society of Medicine Foundation Scholarship at Oxford University in England. He joined the faculty at the Albert Einstein College of Medicine and the Montefiore Medical Center in 1975, where he became professor of pediatrics and professor of epidemiology and social medicine and served as director of the Division of Neonatology until 1994.

In 1994, he became senior vice president of the New York Academy of Medicine, where he catalyzed the academy’s growth into a research intensive institution in areas related to urban health, medical education, public policy, bioethics, and public health. In 2004, Dr. Fleischman became ethics advisor to the National Children’s Study at the National Institutes of Health and was chair of the Federal Advisory Committee to the study from 2005–2010.

In the academic area, he has published and lectured extensively in many areas of perinatal medicine and has been a pioneer in the field of bioethics, emphasizing the rights of individual patients and the responsibilities of health care professionals and organizations. This work has resulted in more than 150 publications in peer reviewed journals and book chapters, including a book edited with Robert Cassidy entitled *Pediatric Ethics--From Principles to Practice*, published by Harwood Press.

Dr. Fleischman was a member of the American Academy of Pediatrics Bioethics and AIDS Committees, a member of the National Human Research Protections Advisory Committee for the Office for Human Research Protections of the U.S. Department of Health and Human Services (DHHS), an expert advisor to the Institute of Medicine’s Committee on Ethical Conduct of Clinical Research Involving Children, and a member of the National Research Council/Institute of Medicine Committee on Ethical Issues in Housing-Related Health Hazard Research Involving Children, Youth, and Families. He was a founding member and is currently a member of the New York State Governor’s Task Force on Life and the Law, and a

member of the DHHS Secretary's Advisory Committee on Human Research Protections' Subcommittee on Research Involving Children.

Richard Gibbs, Ph.D., was born in Australia and graduated from Melbourne University and the Peter MacCallum Cancer Institute in 1986. He moved to the U.S., where he performed key work on the molecular characterization of human genetic disease and the development of methods for mutation detection. He provided an early leadership role for the Human Genome Project (HGP) and in 1997 founded the Baylor College of Medicine Human Genome Sequencing Center (HGSC) in Houston, Texas. Under his leadership, the HGSC was one of five main groups to complete the HGP, and since has completed the genomes of many other species. Subsequently the group has both contributed to major international large scale genetics projects and focused on the analysis of individual personal human genomes, including the Watson, Tutu, and Lupski genome projects. These have been key models for the use of DNA sequencing in medical diagnostics. In 2007, the HGSC began to systematically analyze genes and genomes involved in somatic mutations in cancer. Since then, the group has been a major data producer for the national cancer genome programs.



David B. Goldstein, Ph.D., is a professor of molecular genetics and microbiology and director of the Center for Human Genome Variation at Duke University School of Medicine. He received his Ph.D. in Biological Sciences from Stanford University in 1994 and from 1999–2005 was Wolfson Professor of Genetics at University College London. In April 2007, he was appointed honorary professor at the Institute of Neurology at University College London.

Dr. Goldstein is the author of more than 150 scholarly publications in the areas of population and medical genetics. His principal interests include human genetic diversity, the genetics of disease, and pharmacogenetics. He is the recipient of one of the first seven nationally awarded Royal Society Wolfson Research Merit Awards in the U.K. for his work in human population genetics. Dr. Goldstein was awarded the Triangle Business Journal Health Care Heroes Award in March 2008 for his work on host determinants of control of HIV-1. Most recently, he was appointed co-chair and chair of the Gordon Research Conference meeting on human genetics and genomics for 2011 and 2013.



Eric D. Green, M.D., Ph.D., is the director of the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH) in Bethesda, Maryland, a position he has held since late 2009. Previously, he served as the NHGRI scientific director (2002–2009), chief of the NHGRI Genome Technology Branch (1996–2009), and director of the NIH Intramural Sequencing Center (1997–2009). Dr. Green received his B.S. degree from the University of Wisconsin—Madison in 1981 and his M.D. and Ph.D. degrees from Washington University in 1987. He subsequently trained in clinical pathology. Since the early 1990s, Dr. Green has been extensively involved in efforts to map, sequence, and understand eukaryotic genomes. His work included significant

start-to-finish involvement in the Human Genome Project and more recently has involved several major efforts that utilize large-scale DNA sequencing to address important problems in genomics, genetics, and biomedicine.



Alan E. Guttmacher, M.D., became the director of the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development in August 2010. Previously, he served as acting director of the institute beginning in December 2009. A pediatrician and medical geneticist, Dr. Guttmacher came to the National Institutes of Health (NIH) in 1999 to work at the National Human Genome Research Institute (NHGRI), where he served in a number of roles, including 7 years as the deputy director and, from August 2008 to December 2009, as the acting director. In those roles, he oversaw the institute's efforts to advance genome research, integrate that research into health care, and explore the ethical, legal, and social implications of human genomics.

Dr. Guttmacher also previously served as the senior clinical advisor to the director of NHGRI and as director of NHGRI's Office of Policy, Communications, and Education, leading the institute's health affairs, public policy, communications, community outreach, and public education functions. He led the institute's involvement in educating both the public and health professionals about genomics and genomic health care. Among Dr. Guttmacher's areas of expertise is the development of new approaches for translating the findings of the Human Genome Project into better ways of diagnosing, treating, and preventing disease. He oversaw NIH's involvement in the U.S. Surgeon General's Family History Initiative, an effort to encourage all Americans to learn about and use their families' health histories to improve health. He served as co-editor for a series of articles about genomic medicine that was published in the *New England Journal of Medicine* in 2002–2003 and is co-editing a similar series to appear in the same journal starting in early 2010.

Dr. Guttmacher came to NIH from the University of Vermont, where he directed the Vermont Regional Genetics Center and Pregnancy Risk Information Service, the Vermont Newborn Screening Program, and the Vermont Cancer Center's Familial Cancer Program; founded Vermont's only pediatric intensive care unit; and was the principal investigator for an NIH-supported initiative that was the nation's first statewide effort to involve the general public in discussion of the Human Genome Project's ethical, legal, and social implications. He also conducted research, taught, and had a busy practice in clinical genetics.

A graduate of Harvard College and of Harvard Medical School, Dr. Guttmacher completed an internship and residency in pediatrics and a fellowship in medical genetics at Harvard and the Children's Hospital of Boston. He is a member of the Institute of Medicine and a Fellow of the American Academy of Pediatrics.



R. Rodney Howell, M. D., is a professor of pediatrics and chairman emeritus at the University of Miami's Leonard M. Miller School of Medicine. He is certified by the American Board of Pediatrics as well as the American Board of Medical Genetics in the area of Clinical Biochemical Genetics.

He is the author of more than 175 original articles, numerous abstracts, and other materials dealing largely with serious genetic diseases in childhood. He was recognized in 2007 with the Lifetime Achievement Award from the Duke Medical Alumni Association. Recently named lectureships include the Klauber Lecture (Greenwood Genetics Center), the Jimmy L. Simon, M.D. Distinguished Lecturer (Wake Forest University), and the Butterfield Lecturer at the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD)/University of Colorado Annual Conference in Aspen, Colorado.

He serves at the NICHD as senior advisor to the director, focusing on the institute's research efforts in newborn screening. He is a member of the Association of Public Health Laboratories' Newborn Screening and Genetics in Public Health Committee.

Dr. Howell is chair of the Secretary's Advisory Committee of Heritable Disorders in Newborns and Children, the congressionally-mandated committee that advises the Secretary of Health and Human Services on newborn genetic screening and other issues concerning genetics in children.



Petri Huhtinen, Ph.D., is business manager in PerkinElmer's Newborn Screening business unit. Dr. Huhtinen joined PerkinElmer as a research and development chemist in 2006 and moved to the strategic business team in 2008. During his career at PerkinElmer, Dr. Huhtinen has been developing novel assay applications as well as assessing new business opportunities in emerging markets within the newborn screening arena.

Prior to PerkinElmer, Dr. Huhtinen worked as a research scientist at the University of Turku, Finland. His research addressed developing and applying nanoparticle label technology in bioaffinity assays. Huhtinen also was involved in the early phase of setting up the Finnish Microarray and Sequencing Centre, a functional genomics core facility providing research technologies and services.

Dr. Huhtinen received his M.Sc. in molecular biology from University of Oulu and his Ph.D. in biotechnology from University of Turku. He also holds a B.B.A. from the Turku School of Economics.



Bruce R. Korf, M.D., Ph.D., received his M.D. degree from Cornell University Medical College and his Ph.D. degree in genetics and cell biology from The Rockefeller University. He then completed training in pediatrics, pediatric neurology, and genetics at Children's Hospital Boston. He served as clinical director in the Division of Genetics at Children's Hospital from 1986 to 1999 and as the medical director of the Harvard-Partners Center for Genetics and Genomics from 1999–2002. Currently, he holds the Wayne H. and Sara Crews Finley Chair in Medical Genetics and is professor and chair in the Department of Genetics and director of the Heflin

Center for Genome Sciences at the University of Alabama at Birmingham. In his previous appointment at Harvard Medical School, he served as co-director of the course Genetics, Developmental and Reproductive Biology, taught to all first year students at Harvard Medical School. His book based on this course, *Human Genetics: A Problem-Based Approach*, published by Blackwell Science, is currently in its third edition. He is also co-author of *Medical Genetics at a Glance*, co-editor of the fifth edition of *Emery and Rimoin's Principles and Practice of Medical Genetics*, and co-editor of *Current Protocols in Human Genetics*. Dr. Korf is president of the American College of Medical Genetics and is a member of the Board of Scientific Counselors of the National Human Genome Research Institute. He has completed terms as president of the Association of Professors of Human and Medical Genetics, member of the boards of directors of the American College of Medical Genetics and the American Society of Human Genetics, member of the Liaison Committee on Medical Education, and the National Cancer Institute Board of Scientific Counselors.



David H. Ledbetter, Ph.D., F.A.C.M.G., is executive vice president and chief scientific officer at Geisinger Health System. He came to Geisinger from Emory University School of Medicine, where he was the Robert W. Woodruff Professor and director of the Division of Medical Genetics in the Department of Human Genetics.

Dr. Ledbetter previously held academic and leadership positions at the University of Chicago, the National Center for Human Genome Research (now NHGRI) at NIH, and Baylor College of Medicine. He is a graduate of Tulane University and earned his doctorate at the University of Texas at Austin. After his early discovery of the genetic cause of Prader-Willi syndrome and Miller-Dieker syndrome, Dr. Ledbetter has focused his research efforts on discovering the underlying etiology of childhood developmental disabilities such as autism and the translation of new genomics technologies into clinically useful genetic tests for early diagnosis and intervention. His current research interests include leveraging the massive amount of genomics data generated during routine patient care for knowledge generation and integration of this information into electronic health records in a clinically useful manner.

Michele Lloyd-Puryear, M.D., Ph.D., is a pediatrician and geneticist. She has held academic appointments and has worked in pediatric clinics at the local and international levels. In her present position, she has administrative responsibilities for the federal Genetic Services Program at the Health Resources and Services Administration. She serves as chief of the Genetic Services Branch, Maternal and Child Health Bureau, is the executive secretary of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, and is responsible for policy and public affairs concerning issues and activities surrounding the use of genetic medicine and technology, including program planning and education activities at her agency and bureau. Dr. Lloyd-Puryear has served in an advisory capacity about genetics and newborn screening to her bureau, her agency and other government agencies, and to nongovernmental organizations.



Fred Lorey, Ph.D., received his bachelor's degree from the College of William and Mary and his master's and Ph.D. from the University of California Davis. Prior to coming to the Genetic Disease Screening Program in California, he was an assistant professor of biological anthropology at the University of Minnesota. He has been the acting director of both the California Newborn Screening Program and the California Prenatal Screening Program for the past 3 years. His publications include studies on hemoglobinopathies, metabolic disorders, endocrine disorders, prenatal screening, and other newborn screening-related subjects. He is currently serving on the standing committee for the Newborn Screening Translational Research Network and participating in the NIH-sponsored Severe Combined Immunodeficiency pilot project.



Andre Marziali, Ph.D., P.Eng., received his B.A.Sc. in engineering physics from the University of British Columbia (UBC) in 1989 and his Ph.D. in physics from Stanford University in 1994. He subsequently worked for several years with Dr. Ron Davis in the Stanford DNA Sequencing Technology Center developing instruments for DNA sequencing and sample purification. He returned to Canada in 1998 as an assistant professor at UBC in the Department of Physics and Astronomy, where he formed the Applied Biophysics Laboratory. Shortly after his return to Canada, Dr. Marziali formed

the GenomeBC Technology Development Platform, which he continues to lead as co-director. In 2005, he was appointed director of the Engineering Physics Program at UBC, a position he continues to hold in parallel with research and commercialization activities.

In 2004, Dr. Marzali co-invented the concept of using synchronous mobility perturbations to create divergent velocity fields for selectively focusing nucleic acids. This technology, termed SCODA, is the basis of a spin-off company, Boreal Genomics, Inc., founded in 2007 by Dr. Marzali and colleagues to commercialize high performance instruments for DNA and RNA purification. In the last few years he has been awarded the 2003 Killam Prize for Excellence in Teaching, the 2004 BC Innovation Council – Young Innovator award, the 2005 Canadian Association of Physicists Medal for excellence in teaching, and the 2007 Association for Lab Automation Innovation Award.

Boreal Genomics employs more than 20 people in leased space on the UBC campus. It has recently been recognized by the National Research Council of Canada as a Canadian Innovation Leader, and by the Association for Lab Automation as one of its top ten breakthroughs of 2009.



Jeff Murray, M.D., is a pediatrician and human molecular geneticist with a longstanding career commitment to understanding the genetic and environmental causes of complex perinatal disorders. He trained at the Massachusetts Institute of Technology, Tufts University, and the University of Washington. He is an attending physician in the newborn nurseries and birth defects clinics at the University of Iowa. He has used human gene mapping approaches and epidemiology to discover the underlying causes of birth defects, especially cleft lip/palate and preterm birth. He directed the human genome center at the University of Iowa in the 1990s and served on both the scientific review and the Ethical, Legal, and Social Implications panels for NIH. He has been the director of the University of Iowa's Neonatal Research Biorepository since 1999 and oversees preterm birth and birth defect projects in Denmark, India, Brazil, the Philippines, and Argentina. The focus of these efforts has been on using genome-wide association, linkage, and sequencing approaches to identify genetic causes and then coupling these to environmental covariates. With his postdoctoral fellow Kelli Ryckman, Dr. Murray is currently overseeing a project examining the role of genetic variation in determining analyte levels from newborn screening cards and looking at the role of environmental and demographic variables in a sample of 250,000 newborn screening results. He has also published on health outcomes related to term infants and on the genetic components of the complications of prematurity. Dr. Murray has a strong commitment to genetic education and has twice been named the Mentor of the Year at the University of Iowa. He is an elected member of the Institute of Medicine and a former E. Mead Johnson and Curt Stern awardee from the American Academy of Pediatrics and American Society of Human Genetics, respectively.



Bent Nørgaard-Pedersen, M.D., got his medical degree from Aarhus University in 1965. After 5 years of clinical work, he was employed at Department of Clinical Biochemistry, Rigshospitalet, University of Copenhagen, where he got his specialist license in laboratory medicine and received his Doctorate of Medical Science in 1976. In 1996, he became the first professor of biochemical screening at the University of Copenhagen.

After working as a consultant and head of the Department of Clinical Biochemistry at Sonderborg Hospital (1976–1983), Dr. Nørgaard-Pedersen was offered a position as head of the Department of Clinical Biochemistry at the Statens Serum Institut in Copenhagen (1984–2008).

Dr. Nørgaard-Pedersen has been in charge of the Danish Neonatal Screening program since 1984, and has also overseen the Prenatal Screening program since the early 1970s. Since then, his main research has been in fetal and maternal pathophysiology, as documented by his more than 320 peer-reviewed publications.

In neonatal screening, Dr. Nørgaard-Pedersen has developed many new immunoassays and has been a pioneer in biobank studies looking for new markers for old and new disorders. He has been a key person for use and regulation of the Danish Newborn Screening Biobank, and his setup has been a model worldwide. Dr. Nørgaard-Pedersen retired in 2008, but he is still employed part-time at the Statens Serum Institut and is actively involved in several national and international studies.



P. Pearl O'Rourke, M.D. is the director of Human Research Affairs at Partners HealthCare Systems in Boston and an associate professor of pediatrics at Harvard Medical School. She is responsible for the systems that support the regulatory and ethical oversight of human research and the responsible conduct of research. She is also chair of the Partners Healthcare System Embryonic Stem Cell Research Oversight Committee.

Dr. O'Rourke has worked as a pediatric critical care physician at Children's Hospital Boston and at the Seattle Children's Hospital, where she was the director of the Pediatric Intensive Care Unit. She was active in clinical research in extracorporeal membrane oxygenation, liquid ventilation, high frequency ventilation, and pediatric resuscitation. In Seattle, she served many years as a member of the institutional review board. Dr. O'Rourke also has been involved in international medical care, serving in China and Indonesia with Project HOPE.

In 1995–1996, Dr. O'Rourke did a Robert Wood Johnson Health Policy fellowship working for Senator Edward Kennedy. Following this fellowship, she became the deputy director of the Office of Science Policy in the Office of the Director at NIH, where she worked on issues such as privacy, gene therapy (transfer), embryonic stem cells, and genetic discrimination.



Vamsee K. Pamula, Ph.D., is a co-founder and chief technology officer of Advanced Liquid Logic, Inc. which develops digital microfluidics products for clinical diagnostics, screening, and other research applications. His current research interests include development of inexpensive and accessible diagnostic devices. He serves as a principal investigator on several projects funded by NIH, has given numerous talks on digital microfluidics, has published more than 50 articles, authored 3 book chapters and a book, and has more than 100 issued and pending patent applications. He has a Ph.D. in electrical and computer engineering from Duke University. He also serves on the graduate faculty at the

Department of Electrical and Computer Engineering at Duke University.



Piero Rinaldo, M.D., Ph.D., graduated summa cum laude from the University of Padova School of Medicine (Italy) in 1982 and enrolled in the pediatric residency program there. After completion of his residency, Dr. Rinaldo completed a research doctorate (Ph.D.) program in developmental sciences. In 1987, he joined the laboratory of Dr. Kay Tanaka in the Department of Human Genetics at Yale University School of Medicine. He later enrolled in the Biochemical Genetics fellowship training program, which he completed in 1992. Upon the retirement of Dr. Tanaka, he was offered a faculty position at Yale and became the director of the Biochemical Disease Detection Laboratory. In 1998, Dr. Rinaldo joined Mayo Clinic as director of the Biochemical Genetics Laboratory and professor in the Department of Laboratory Medicine and Pathology (DLMP), with joint appointments in the Department of Medical Genetics and in the Department of Pediatric and Adolescent Medicine, where he is the T. Denny Sanford Professor of Pediatrics. He has served as Division Chair in DLMP from 2001 to 2007. Since 2007, he has been the DLMP Vice Chair of Academic Affairs and Intramural Practice.

Dr. Rinaldo has authored more than 170 articles and reviews/chapters and given more than 200 grand rounds, seminars, and lectures on clinical and laboratory aspects of metabolic disorders. His current primary focus is on the expansion and quality improvement of newborn screening programs. He serves on the board of directors of professional and patient support organizations, including the American College of Medical Genetics, where he is the Vice President of Laboratory Genetics.



Jeffery A. Schloss is program director for technology development coordination in the Division of Extramural Research at the National Human Genome Research Institute (NHGRI), a component of the National Institutes of Health (NIH). At NHGRI, he manages a grants program in technology development for DNA technologies and in particular the program to develop technologies with which to sequence an entire human genome for \$1000. He led the team that launched and continues to coordinate the Centers of Excellence in Genomic Science. Dr. Schloss represented NHGRI on the NIH Bioengineering Consortium, BECON, from its establishment in 1997 until 2008 and served as the chair of BECON from 2001–2004. Among his numerous BECON activities, he co-organized the BECON 2000 symposium on nanotechnology in biomedicine. He represents the NIH on the National Science and Technology Council's subcommittee on Nanoscale Science, Engineering, and Technology, planning for the National Nanotechnology Initiative. He also co-chairs the NIH Nanomedicine Roadmap Initiative and was co-chair of the Trans-NIH Nano Task Force from 2006–2008. He serves on the NIH Roadmap Human Microbiome Project implementation group, leading the HMP technology development initiative. Dr. Schloss earned a B.S. degree in biology with honors from Case Western Reserve University, earned a Ph.D. in cell biology from Carnegie Mellon University, conducted postdoctoral research at Yale University, and served on the biology faculty at the University of Kentucky.



Keld Sorensen, Ph.D., has a track record of developing assays and assay technologies. From his Ph.D. work at the University of Copenhagen, where he developed assays for enzymes involved in fibrinolysis and blood clotting, to his first postdoctoral research in Switzerland, where his work on brain enzymes lead to an assay used in prenatal diagnosis of neural tube defects. Later, he

moved to the U.S., where he worked at NTD Laboratories, a New York service laboratory which was recently acquired by PerkinElmer. Dr. Sorensen developed in-house assays and methods for pre-natal screening.

His technical skills as well as his leadership skills led him to the position of director of research and development at Sigma-Aldrich in St. Louis, MO, where he was responsible for growing the research and development teams from a handful of scientists in 1997 to more than 100 scientists in 2005. In 2005, when Luminex was looking for an expert to initiate a commercial assay development effort, they turned to Dr. Sorensen, who in 2005 moved to Austin and started the Luminex Bioscience Group, which developed assays both for Dx and for life science. Most recently, Dr. Sorensen was asked to start a Luminex team focused on development of new assay technologies. This team is focused on expanding the Luminex technology and adopting new methods and technologies for the continued growth of the company. Dr. Sorensen holds a number of patents, publications, and is a sought-after speaker for events.



Richard M. Weinshilboum, M.D., received B.A. and M.D. degrees from the University of Kansas, followed by residency training in internal medicine at Massachusetts General Hospital, a Harvard teaching hospital in Boston. He was also a pharmacology research associate at the National Institutes of Health in Bethesda, Maryland, in the laboratory of Nobel laureate Dr. Julius Axelrod. Dr. Weinshilboum began his affiliation with the Mayo Medical School and Mayo Clinic in Rochester, Minnesota, in 1972. He is presently professor of molecular pharmacology and experimental therapeutics and internal medicine and Mary Lou and John H. Dasburg Professor in Cancer Genomics Research. Dr.

Weinshilboum's research has focused on pharmacogenetics and pharmacogenomics, and he has authored more than 320 scientific manuscripts which address these topics. A major area of investigation has been the pharmacogenetics of drug metabolism, with a focus on methylation and sulfation, but in recent years his research has increasingly applied genome-wide pharmacogenomic techniques rather than candidate gene or candidate pathway-based approaches. Dr. Weinshilboum has been the recipient of many awards and honors including an Established Investigatorship of the American Heart Association, a Burroughs Wellcome Scholar Award in Clinical Pharmacology, the Oscar B. Hunter Award of the American Society for Clinical Pharmacology and Therapeutics, the Harry Gold Award of the American Society for Pharmacology and Experimental Therapeutics, the Catecholamine Club Julius Axelrod medal, the U.S. Food and Drug Administration William B. Abrams Lectureship Award, and the Edvard Poulsson Award from the Norwegian Pharmacology Society.

Planning Committee

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