

**Public Workshop**  
**Complex Issues in Developing Drug and Biological Products for Rare Diseases**  
*January 6, 2014*

**Speakers' and Panelists' Biographies**

*\*Alphabetically ordered by last name*

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**NURIA CARRILLO-CARRASCO, M.D., National Center for Advancing Translational Sciences (NCATS), NIH**

Dr. Carrillo is the Head of the Clinical Group of the Therapeutics for Rare and Neglected Diseases Program at the NIH. The group is responsible for conducting natural history studies and early-phase clinical trials needed to advance promising therapies for rare diseases, developing biomarkers and identifying appropriate outcome measures. Currently, she is the Principal Investigator of the Natural History study of GNE myopathy and the Phase Ia clinical trial of ManNAc. Prior to joining the TRND program, her research focused on clinical translational aspects of inborn errors of metabolism, biomarker development using LC-MS/MS and gene therapy studies. She earned her MD degree from the National Autonomous University of Mexico (UNAM) and completed her pediatrics residency at Georgetown University Hospital. She is board-certified in Pediatrics, Medical Genetics and Biochemical Genetics.

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**EDWARD COX, M.D., M.P.H, Center for Drug Evaluation and Research, FDA**

Edward Cox received his undergraduate degree in chemistry from the University of North Carolina at Chapel Hill and his medical degree from the University of North Carolina School of Medicine. He completed an internship and residency in internal medicine at the Hospital of the University of Pennsylvania in Philadelphia, and he went on to complete a fellowship in infectious diseases at the National Institute of Allergy and Infectious Diseases at the National Institutes of Health in Bethesda, MD. Dr. Cox is board certified in internal medicine and infectious diseases.

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**DIANE DORMAN, National Organization for Rare Disorders**

Ms. Dorman is the Vice President for Public Policy for the National Organization for Rare Disorders (NORD) and leads NORD efforts in its relationship with the federal government and Congress. She is the primary DC representative for more than 30 million Americans who have one of the 7,000 known rare diseases. Her overriding mission is to improve the plight of patients with rare diseases and increase incentives for the development of orphan drugs, devices, and diagnostics.

Since joining NORD in October 2000, Ms. Dorman's advocacy has been instrumental in the passage of three public laws and has been influential in the adoption of numerous programs, regulations and guidances that touch the lives of patients with rare diseases.

On behalf of NORD and coalitions in which NORD participates, Ms. Dorman leads education and outreach programs to gain policymaker support for increased research into rare diseases and greater development of orphan products. She sits on the Board of Directors and Executive Committee of the Alliance for a Strong FDA, and currently serves as President of that organization.

Ms. Dorman sits on the Advisory Committee of the Keck Graduate Institute Center for Rare Disease Therapies based in Claremont, CA. Ms. Dorman has also been appointed to the Council of the Convention of the US Pharmacopeia, and served as a member of the Strategic Team for the Entrepreneurs in Residence Program house in the Center for Devices Radiological Health at the FDA.

She is responsible for ensuring that patients continue to have access to life-saving orphan therapies through Medicare, Medicaid and private insurance. She also serves as NORD's primary liaison to the Food and Drug Administration, the National Institutes of Health, Social Security Administration, and the Center for Medicare and Medicaid Services, as well as the biopharmaceutical and medical device industries. Ms. Dorman has been appointed as a consumer representative to the Medicare Evidence Development Coverage Advisory Committee.

Ms. Dorman develops and maintains relationships with other healthcare voluntary agencies and patient groups. She provides technical assistance and legislative analysis to NORD's member agencies on government-related matters, as well as the training of staff and volunteers of member organizations. Her leadership efforts have led to introduction and passage of the *Rare Diseases Act* (P.L. 107-281), and the *Rare Diseases Orphan Product Development Act* (P.L. 107-281). She was also influential in the introduction of *House Concurrent Resolution 147*, commemorating the 20<sup>th</sup> Anniversary of the *Orphan Drug Act*. Recently, Ms. Dorman played an influential role in key provisions related to rare diseases and orphan product development in the *Food and Drug Administration Safety & Innovation Act* (P.L. 112-144).

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**STEPHEN GROFT, Pharm.D, National Center for Advancing Translational Sciences, NIH**

Steve is the Director of the Office of Rare Diseases Research (ORDR) in the National Center for Advancing Translational Sciences at the National Institutes of Health (NIH). His major focus is on stimulating research with rare diseases and developing information about rare diseases and conditions for health care providers and the public. To help identify research opportunities and establish research priorities, the Office has co-sponsored over 1200 rare diseases-related scientific conferences with the NIH research Institutes and Centers and the extramural research community, including patient advocacy groups. Current and recent activities include establishing common and unique data elements and patient registries for rare diseases, developing an inventory of available bio-specimens from existing bio-repositories, maintaining

a public information center on genetic and rare diseases, developing an international rare diseases research consortium, maintaining the Rare Diseases Clinical Research Network, and assisting in the development of a special emphasis research network with senior clinical staff for patients with undiagnosed diseases at NIH's Clinical Research Center Hospital and at academic centers in the United States.

Steve received the B.S. degree in Pharmacy in 1968 and the Doctor of Pharmacy degree from Duquesne University in 1979.

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### **NICOLE HAMBLETT, Ph.D., University of Washington & Seattle Children's Research Institute**

Nicole Mayer Hamblett, Ph.D. is an Associate Professor in the Departments of Pediatrics and Adjunct Associate Professor in the Department of Biostatistics at the University of Washington. Dr. Hamblett is Co-Director of the Cystic Fibrosis Foundation (CFF) Therapeutics Development Network (TDN) Coordinating Center at Seattle Children's Hospital, supporting a network of over 70 clinical sites dedicated towards evaluating the safety and effectiveness of CF therapies. In this role, Dr. Hamblett has engaged in the design of multiple CF trials supported by both industry and academia, and has overseen the coordination of over 30 clinical trials supported by the TDN Coordinating Center. She is currently the Principal Investigator of the Data Coordinating Center for the NIH-funded OPTIMIZE clinical trial in CF, which aims to evaluate the efficacy of a combined antibiotic approach to reduce pulmonary symptoms among children with CF with newly acquired *Pseudomonas aeruginosa*.

Dr. Hamblett is also the Director of the Seattle Children's Core for Biomedical Statistics, providing biostatistical and data management support for local investigators in collaboration with the NIH's Clinical and Translational Science Award (CTSA) program. Dr. Hamblett's academic interests include the efficient design and analysis of clinical trials, with emphasis in the pediatric and orphan disease setting. She is involved in epidemiologic research using large patient registries and archived clinical trial data, in addition to the development of new clinical outcome measures for CF, including the validation of biomarkers to enable early evaluation of new therapies. Dr. Hamblett received her PhD in Biostatistics from the University of Washington, and has an undergraduate degree in Mathematics from Santa Clara University.

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### **JOHN HYDE, M.D., Ph.D., Center for Biologics Evaluation and Research, FDA**

Dr. Hyde has worked at the FDA for more than 19 years, and his experience has encompassed the medical review of drugs, devices, and biologics. He has been involved as medical team leader in the review and approval of several drug products for rare metabolic diseases and other products using small study populations. He is currently working in the Center for Biologics Evaluation and Research, where his responsibilities include advising researchers and biologics companies on the development of cellular and gene therapies for a variety of rare non-oncologic diseases.

## **EMIL KAKKIS, M.D., Ph.D., EveryLife Foundation for Rare Diseases & Ultragenyx**

Dr. Kakkis is best known for his work over the last 18 years to develop novel treatments for rare disorders. He began his work developing an enzyme replacement therapy (Aldurazyme®) for the rare disorder MPS I, with minimal funding and support. The struggle to get the therapy translated from a successful canine model to patients succeeded due to the critical financial support of the Ryan Foundation, a patient organization formed by Mark and Jeanne Dant for their son Ryan.

Aldurazyme development was later supported by BioMarin Pharmaceutical and eventually their partner Genzyme leading to FDA approval in 2003. During his tenure at BioMarin, Dr. Kakkis guided the development and approval of two more treatments for rare disorders, MPS VI and PKU, and has contributed to the initiation of 7 other treatment programs for rare disorders, three of which are now in clinical development.

After 11 years at BioMarin, Dr. Kakkis left industry to initiate an effort to improve the regulatory and clinical development process for rare diseases. In early 2009, Dr. Kakkis launched and funded the Kakkis EveryLife Foundation to accelerate biotech innovation for rare diseases. The Foundation initiated a campaign to improve the regulatory and clinical development process for rare diseases. In just over a year, 160 patient organizations and physician society partners have endorsed the Campaign.

Dr. Kakkis has founded Ultragenyx™ to return to development of drugs for rare diseases. For many rare diseases, reasonable science exists that needs to get translated to patients. He will build on his previous experiences and will assemble an experienced team to efficiently develop treatments for rare diseases.

Dr. Kakkis is board certified in both Pediatrics and Medical Genetics. He graduated from Pomona College, magna cum laude and received combined M.D. and Ph.D. degrees from the UCLA Medical Scientist Program and received the Bogen prize for his research. He completed a Pediatrics residency and Medical Genetics Training Fellowship at Harbor- UCLA Medical Center. He became an assistant professor of Pediatrics at Harbor-UCLA Medical Center from 1993 to 1998 where he initiated the enzyme therapy program for MPS I. In 1998, he joined BioMarin where he remained for 11 years in various titles eventually as Chief Medical Officer, before leaving in 2009.

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## **MWANGO KASHOKI, M.D., M.P.H., Center for Drug Evaluation and Research, FDA**

Dr. Kashoki is the Associate Director for Safety in the Office of New Drugs (OND), in the Center for Drug Evaluation and Research (CDER) at FDA. Dr. Kashoki's responsibilities include ensuring OND's implementation of the policies and processes related to CDER's various safety initiatives, including the Safety First and Sentinel Initiatives. She also leads OND's implementation of FDA's new authorities to require safety labeling changes, postmarketing investigations, and risk evaluation and mitigation strategies, as provided under the FDA Amendments Act (FDAAA).

Dr. Kashoki joined OND in 2002 as a primary medical officer in the former Division of Anesthetic, Critical Care and Addiction Drug Products, and then served as a clinical team leader in that division for several years. As a team leader, she supervised primary medical officers in reviewing investigational and new drug applications, as well as in providing guidance to individual researchers and pharmaceutical companies regarding addiction and analgesic drug development programs. Prior to her current position, Dr. Kashoki served as Associate Director for Special Projects in the former Division of Anesthesia, Analgesia and Rheumatology Products, leading the development and conduct of research projects under FDA's Critical Path Initiative and in collaboration with external groups. Dr. Kashoki is board certified in Preventive Medicine and Public Health.

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**MICHAEL KOSOROK, Ph.D., M.S., University of North Carolina at Chapel Hill**

Michael R. Kosorok, Ph.D., is W. R. Kenan, Jr. Distinguished Professor and Chair of Biostatistics and Professor of Statistics and Operations Research at the University of North Carolina at Chapel Hill (UNC). His biostatistical expertise includes survival analysis, regression modeling, clinical trials, dynamic treatment regimens, personalized medicine and adaptive intervention trials, and he has written a major text on the theoretical foundations of these and related areas in biostatistics (Kosorok, 2008). He has over 110 peer-reviewed publications in biostatistical methodology and theory as well as in the application of biostatistics to cystic fibrosis and cancer. More recently he has pioneered statistical methods based on machine learning for dynamic treatment regimens and Sequential Multiple Assignment Randomized Trials (SMARTs). Dr. Kosorok is also the contact Principal Investigator for an NIH Program Project Grant (P01 CA142538) from the National Cancer Institute on statistical methods for cancer clinical trials. This is a joint endeavor involving UNC, Duke and NC State University and studies many aspects of the design and analysis of clinical trials. He also directs the Biostatistics Core of the Clinical and Translational Science Award (CTSA) located at UNC. He is an honorary fellow of both the American Statistical Association and the Institute of Mathematical Statistics and is an associate editor of both the Annals of Statistics and the Journal of the American Statistical Association Theory.

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**ROBERT KOWALSKI, Pharm.D., Novartis Pharmaceuticals**

Rob Kowalski was appointed Senior Vice President and Global Head of Drug Regulatory Affairs for Pharmaceuticals at Novartis in August 2009. Rob has also been the US Head of Development for Pharmaceuticals since November 2011 and is a member of the Executive Committee for Novartis Pharmaceuticals Corp in the USA.

Rob began his regulatory career at Sandoz and has held subsequent positions in the regulatory area at Pharmacia (now Pfizer) and Schering-Plough (now Merck) before returning to Novartis.

Under his leadership, Rob has successfully brought dozens of new products to the market, including multiple new chemical entities across a wide array of therapeutic areas. Rob is a Board Member and Vice-Chairman of the R&D Council of New Jersey and is also active in multiple professional organizations, including a member of the PhRMA RACC (regulatory committee).

Rob attended the University of Wisconsin–Madison where he received both a Bachelor of Science in Pharmaceutical Sciences and a Doctorate in Pharmacy. Rob completed his post-doctoral training at Rutgers University / Sandoz Pharmaceuticals.

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**MATTHIAS KRETZLER, M.D., University of Michigan Medical School**

Dr. Kretzler is the Warner-Lambert/Parke-Davis Professor of Internal Medicine/Nephrology and Computational Medicine and Bioinformatics. He is the PI of the U54 Nephrotic Syndrome Research Network (NEPTUNE) in the Rare Disease Clinical Research Network II. He is the director of the Applied Systems Biology Core in the Director of the O'Brien Renal Center at UMichigan and a PI in the R24 "Integrated Systems Biology Approach to Diabetic Microvascular Complications". He has 15 years of experience in integration of bioinformatics, molecular and clinical approaches in more than 120 collaborative studies on molecular analysis of renal disease. He has a tract record on interdisciplinary data integration of large-scale data sets and has assembled collaboration with numerous leading consortia across four continents. He has initiated the European renal cDNA bank, the largest biobank of renal tissues for molecular analysis (>2500 biopsies). He focuses with his research team on the analysis of molecular mechanism of glomerular damage. Using integrated biology approaches he defines transcriptional networks in glomerular diseases in human cohorts and integrates them with genetic information, complex clinical data sets and metabolomic information. The large-scale data integration across the genotype-phenotype continuum allows to reach a more holistic understanding of renal disease and has already resulted in the initiation of trials with targeted therapies in glomerular diseases.

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**SANDRA KWEDER, M.D., F.A.C.P, Center for Drug Evaluation and Research, FDA**

Rear Admiral (retired) Sandra L. Kweder graduated from the University of Connecticut in 1979 and attended the University of North Carolina's School of Public Health in Chapel Hill. She was commissioned in the U.S. Public Health Service in 1980 upon entering the Uniformed Services University of Health Sciences (USUHS). She was awarded her M.D. in 1984, along with the Surgeon General's Award for Outstanding PHS Graduate. She completed her internship and residency in Internal Medicine at the Walter Reed Army Medical Center, where she also was the Chief Resident. She is certified by the American Board of Internal Medicine. In 1988 RADM Kweder joined the U.S. Food & Drug Administration (FDA) as a medical reviewer in the Division of Antiviral Drugs, to address the growing field of HIV drug development. She has since held a number of positions, including leadership of the Division of Postmarketing Surveillance & Epidemiology and the Office of Antimicrobial Products and spent two years on

sabbatical at Brown University as a clinical fellow in Obstetric and Consultative Medicine. She has been Deputy Director, Office of New Drugs (OND) in FDA's Center for Drug Evaluation & Research (CDER) since 2002.

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**PAUL LASKO, Ph. D., McGill University**

Prof. Lasko received his A. B. from Harvard and his Ph. D. from the Massachusetts Institute of Technology, and joined McGill in 1990 after a postdoctoral period at the University of Cambridge. Using the *Drosophila* system, Dr. Lasko's research concerns regulatory processes that control gene expression at the levels of mRNA stability or translation. He has authored over 100 research papers in this area. Since 2010 Prof. Lasko has served as Scientific Director of the CIHR Institute of Genetics. He oversees the Institute's strategic research funding initiatives, many of which involve fostering international partnerships. In April 2013 he also assumed the position of Chair of the Executive Committee of the International Rare Diseases Research Consortium, a worldwide consortium of over 30 public and private research funders who collectively have pledged over USD 1 billion for research in rare diseases. More information about this consortium is available at [www.irdirc.org](http://www.irdirc.org).

In the past Dr. Lasko worked extensively for the Human Frontiers of Science Program Organization (HFSP), serving on its program grant panel from 2001-2005, and then as one of two Canadian representatives on the Council of Scientists. He chaired the HFSP Council of Scientists from 2007-2010. Dr. Lasko also served as President of the Genetics Society of Canada from 2007-2010.

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**JOHN MCKEW, Ph.D., National Center for Advancing Translational Science, NIH**

Dr. McKew is the Acting Scientific Director of the Division of Preclinical Innovation at the National Center for Advancing Translational Sciences (NCATS) within the NIH. His responsibilities include developing the Therapeutics for Rare and Neglected Disease (TRND) program and the Bridging Interventional Development Gaps program (BrIDGs; former NIH-RAID program). Both of these programs focus on novel public/private partnerships to advance collaborative drug discovery projects through pre-clinical development into early clinical development. These collaborative projects are advanced using a combination of internal scientific resources as well as government contracts. Prior to joining the NIH, Dr. McKew held a Director level position at Wyeth Research in Cambridge, Massachusetts where he spent a total of 17 years. One role at Wyeth he held comprised leading a hit-to lead chemistry group supporting cardiovascular, musculoskeletal and metabolic disease therapeutic areas. Prior to that Dr. McKew spent 10 years working in the inflammation therapeutic area resulting in multiple compounds entering clinical evaluation. His research interests include rare and neglected disease research, medicinal chemistry, synthetic methodology, and tool compounds to probe biology. These interests have resulted in >35 publications, 10 Granted US Patents and >60 invited presentations. John also enjoys sharing his passion for science with others. This has prompted him to become course director and lecturer in GMS PM 881 "Drug Discovery and Development"

a graduate level course in the Department of Pharmacology and Experimental Therapeutics which resulted in his appointment as an Adjunct Associate Professor Boston University School of Medicine. He has also taken an active role in the Northeastern Section of the American Chemical Society and has served as the Chair-Elect, Chair and the Immediate Past Chair.

Dr. McKew graduated from State University of New York at Stony Brook with B.S. degrees in Chemistry and Biochemistry. He completed his Ph.D. in Organic Chemistry at University of California, Davis and held post-doctoral research positions at the University of Geneva and Firmenich, SA.

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**RICHARD MOSCICKI, M.D., Center for Drug Evaluation and Research, FDA**

Richard (Rich) A. Moscicki (Mo-shis-ke), M.D., joined the U.S. Food and Drug Administration's (FDA) Center for Drug Evaluation and Research (CDER), as Deputy Center Director for Science Operations in April 2013. A nationally recognized expert in clinical research and development, Dr. Moscicki is bringing his extensive scientific expertise and executive leadership skills to Center operations and direction and to effective development and implementation of CDER programs.

Before joining CDER, Dr. Moscicki served as senior vice president (SVP), Head of Clinical Development at Genzyme Corporation. He joined Genzyme in 1992 as Medical Director, becoming Chief Medical Officer and SVP of Biomedical and Regulatory Affairs in 1996 and holding that post until 2011. During that time, Dr. Moscicki was responsible for worldwide global regulatory and pharmacovigilance matters and oversaw all aspects of clinical research and medical affairs for the company.

Dr. Moscicki received his medical degree from Northwestern University Medical School. He is board certified in internal medicine, diagnostic and laboratory immunology, and allergy and immunology. He completed his residency with a focus on immunology, followed by a four-year fellowship at Massachusetts General Hospital (MGH) in immunology and immunopathology. He remained on staff at MGH and on the faculty of Harvard Medical School from 1979 until 2013.

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**ANDREW MULBERG, M.D., F.A.A.P., C.P.I., Center for Drug Evaluation and Research, FDA**

Andrew is currently the Division Deputy Director of Gastroenterology and Inborn Errors Products, Center for Drug Evaluation and Research (CDER), U.S. Food and Drug Administration (FDA) since 2010. Before joining FDA, Andrew has served as Portfolio Leader in Established Products responsible for providing worldwide leadership in support of GI and diverse Internal Medicine products within the Established Products Therapeutic Area of Johnson and Johnson from 2000-2010. He has served as Attending Physician in Gastroenterology and Hepatology at Children's Hospital of Philadelphia from 1993-2010. Andrew is a graduate of

Columbia College of Columbia University and of the Mount Sinai School of Medicine. He completed his residency in Pediatrics at the Children's Hospital of Philadelphia followed by a Pediatric Gastroenterology Clinical Fellowship and a Post-Doctoral Fellowship in Cellular and Molecular Physiology at New England Medical Center. Andrew is Adjunct Professor of Pediatrics at the University of Maryland School of Medicine, Adjunct Associate Professor of Pediatrics in the University of Pennsylvania School of Medicine and Associate Professor of Pharmacy at the University of the Sciences in Philadelphia. He has served as Principal Editor of Pediatric Drug Development: Concepts and Applications published April 2009 with Wiley-Blackwell and now in its 2<sup>nd</sup> edition released August 19, 2013. He is a member of multiple professional medical societies including Alpha Omega Alpha Honor Medical Society, American Gastroenterological Association and the North American Society for Pediatric Gastroenterology and Nutrition.

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**ANNE PARISER, M.D., Center for Drug Evaluation and Research, FDA**

Anne Pariser is the Associate Director for Rare Diseases in the Office of New Drugs at the US Food and Drug Administration Center for Drug Evaluation and Research. She established the Rare Diseases Program in OND in 2010, where she is currently working to support, facilitate and accelerate the development of therapeutics for rare diseases. The Rare Diseases Program concentrates on the development of biomedical and regulatory science, rare disease-specific training and education, and policy and guidance generation for rare disease product review and regulation. Dr. Pariser is also actively involved in numerous collaborations within FDA and with drug developers, other governmental agencies, advocacy groups and other stakeholders to further the development of treatments for rare diseases. Dr. Pariser has worked at FDA since 2000. Prior to founding the Rare Diseases Program, she was a Medical Officer and Team Leader in OND where she worked almost exclusively on the review and regulation of products for rare genetic disorders. Her research interests include the development of regulatory, translational and biomedical science for rare diseases and Orphan products.

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**GAYATRI RAO, M.D., J.D., Office of Orphan Products Development, FDA**

Gayatri R. Rao, is the Director for the Office of Orphan Products Development (OOPD) at FDA. The Office's mission is to advance the development of promising products, including drugs, biologics, devices, and medical foods, for rare diseases. As Director, she oversees a number of programs created to promote the development of such products, including the Orphan Drug Designation Program, the Humanitarian Use Device Designation Program, the Orphan Products Grants Program, and the Pediatric Device Consortia Grant Program. In addition, she coordinates cross-Agency efforts on rare disease issues, is actively engaged in a number of internal and external collaborations to promote the development of products for rare diseases, and oversees the Office's extensive outreach efforts to patients, sponsors, and other stakeholders. Prior to joining OOPD, Dr. Rao worked in FDA's Office of the Chief Counsel where she provided advice on a wide range of issues related to medical devices, combination products, clinical trials, and human subject protection.

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**IRA SHOULSON, M.D., Georgetown University**

Ira Shoulson, MD is Professor of Neurology, Pharmacology and Human Science and Director of the Program for Regulatory Science and Medicine (PRSM) [<http://regulatoryscience.georgetown.edu>] at Georgetown University, Washington, DC. From 1990 until 2011, Dr Shoulson was the Louis C. Lasagna Professor of Experimental Therapeutics and Professor of Neurology, Pharmacology and Medicine at the University of Rochester School of Medicine & Dentistry in Rochester, New York, where he currently holds adjunct appointments as Professor of Neurology, Pharmacology & Physiology. He received his MD degree (1971) and postdoctoral training in medicine (1971-73) and neurology (1975-77) at the University of Rochester and in experimental therapeutics at the National Institutes of Health (1973-75). Dr. Shoulson founded the Parkinson Study Group ([www.parkinson-study-group.org](http://www.parkinson-study-group.org)) in 1985 and the Huntington Study Group ([www.huntington-study-group.org](http://www.huntington-study-group.org)) in 1994 -- international academic consortia devoted to research and development of treatments for Parkinson disease, Huntington disease and related neurodegenerative and neurogenetic disorders. He has served as principal investigator of the National Institutes of Health-sponsored trials "Deprenyl and Tocopherol Antioxidative Therapy of Parkinsonism" (DATATOP), the "Prospective Huntington At Risk Observational Study" (PHAROS), and more than 25 other multi-center clinical research studies. He was formerly a health policy fellow in the US Senate, a member of the National Institute of Neurological Disorders and Stroke Council, and president of the American Society for Experimental NeuroTherapeutics (ASENT). He is currently principal investigator of the FDA-Georgetown University Collaborating Center of Excellence in Regulatory Science and Innovation (CERSI - FD004319), associate editor of *JAMA Neurology* and an elected member of the Institute of Medicine of the National Academy of Sciences. He has authored more than 300 scientific reports.

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**MARSHALL SUMMAR, M.D., Children's National Medical Center**

Dr. Summar is internationally known for his pioneering work in caring for children diagnosed with rare diseases. He joined Children's National in 2010 from Vanderbilt University. At Children's National he leads the Division of Genetics and Metabolism, currently the largest clinical division in the world seeing over 7000 patients a year with rare diseases. He also directs the Clinical Research Center and is Vice-Chair of Special Projects. Dr. Summar's laboratory works on both devices and treatments for patients with genetic diseases and adapting knowledge from rare diseases to mainstream medicine. His work has resulted in new drugs in FDA trials for patients with congenital heart disease and premature birth. His laboratory is best known for its work in the rare diseases affecting nitrogen and ammonia metabolism. Dr. Summar has also organized and led a large number of international work groups to develop standards of care and treatment for rare diseases resulting in significant improvements in outcomes. Dr. Summar developed a program with NIH where very young children can benefit from NIH research programs at Children's National. Dr. Summar is board-certified in Pediatrics, Clinical Genetics, and Biochemical Genetics and has been listed with Best Doctor's in America since 2004. He

serves on the board of directors of the National Organization of Rare Diseases and the Society of Inherited Metabolic Diseases. He directs the National Organization of Rare Diseases Scientific Advisory Committee and is spearheading an effort to develop national standards for families to collect information about poorly understood rare diseases affecting 25 million Americans. He is very active in newborn screening issues developing testing and follow-up systems.

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**ELLIS UNGER, M.D., Center for Drug Evaluation and Research, FDA**

Dr. Ellis F. Unger is the Director, Office of Drug Evaluation-I, Office of New Drugs (OND), Center for Drug Evaluation and Research (CDER), U.S. Food and Drug Administration. Dr. Unger is a cardiologist who obtained his medical degree from the University of Cincinnati, and received post-doctoral training at the Medical College of Virginia (internal medicine) and The Johns Hopkins Hospital (clinical cardiology). Dr. Unger was a Senior Investigator in the Cardiology Branch, National Heart, Lung, and Blood Institute, National Institutes of Health, from 1983 to 1997, where he directed a translational research program in angiogenesis, developing new approaches for the treatment of coronary artery disease and peripheral vascular disease. From 1997 to 2003, Dr. Unger served as a Medical Officer, Team Leader, and subsequently Branch Chief in the Office of Therapeutics Research and Review, Center for Biologics Evaluation and Research (CBER), FDA. When regulatory authority for therapeutic biologics was transferred from CBER to CDER in 2003, Dr. Unger assumed the responsibilities of Deputy Director, Division of Therapeutic Biological Internal Medicine Products, Office of Drug Evaluation-VI, OND, CDER. With the dissolution of the therapeutic biologics division in 2005, Dr. Unger became Deputy Director of the Division of Cardiovascular and Renal Products. From November, 2006 until October, 2007, Dr. Unger served as the Acting Deputy Director of the Office of Surveillance and Epidemiology in CDER. Dr. Unger became Deputy Director, Office of Drug Evaluation-I, in July, 2009, and its Director in July, 2012. He has served on numerous working groups, including the Risk Assessment Working Group - PDUFA III Implementation, Council for International Sciences (CIOMS) Working Group VII, the International Conference on Harmonization (ICH) Expert Working Groups on E2F and E2C(R2). Dr. Unger has authored, co-authored, and edited numerous scientific articles, and is a co-holder of two patents.

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**MARC K. WALTON, M.D., Ph.D., Center for Drug Evaluation and Research, FDA**

Dr. Walton received his graduate degrees from the University of Chicago. Following a neurology residency at the University of Rochester, Rochester NY, he moved to the National Institute of Neurological Disorders and Stroke, NIH researching neurotransmitter responses in embryonic spinal cord.

Dr. Walton joined the Center for Biologics Evaluation and Research (CBER) at FDA in 1993 as a Medical Officer working on clinical trials of biological products for neurological disorders. His appointment as Branch Chief in the Division of Clinical Trial Design and Analysis brought additional clinical areas of experience. Dr. Walton became the Division Director with clinical

oversight for all non-oncology uses of biological proteins during with the transfer of jurisdiction for biological protein products from CBER to CDER. A subsequent move to the Office of Policy in the Office of the Commissioner gave involvement in a broad range of agency-wide issues. His position in the Office of Translational Sciences in CDER focuses on fostering both internal and external science and policies to support innovative approaches to therapeutic development, and includes biomarkers, clinical study design and analysis methods, liaison to external consortia and other agencies, and rare diseases.

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**PAMELA WILLIAMSON, R.A.C., F.R.A.P.S., Biotechnology Industry Organization**

Pamela M. Williamson, RAC, FRAPS is the Senior Vice President and Global Head of Regulatory Affairs and Compliance for Genzyme Corporation, a Sanofi Company. Ms. Williamson has over 25 years of bio-pharmaceutical industry experience including Regulatory Affairs, Quality Assurance & Compliance, Clinical & Product Development and Manufacturing Operations. She currently leads Genzyme's global regulatory & compliance organization. In her role, she is responsible for developing registration strategies for Genzyme's early and late stage product portfolio which includes a range of therapies for uncommon and underserved medical conditions such as lysosomal storage disorders, rare cancers, and genetic cardiovascular disease and encompasses an array of medical technology platforms, including protein-based therapies, therapeutic polymers, small molecules, biomaterials and gene therapies. She is a member of the Genzyme Executive Leadership Team and serves on multiple internal senior management and governance committees. Externally, Pam represents Genzyme on several industry committees including PhRMA's Regulatory Affairs Coordinating Committee (RACC) and BIO's Regulatory Affairs Committee (RAC) and serves as an industry spokesperson and rare disease expert at various conferences, workshops and liaison meetings. Her additional affiliations include the Massachusetts Biotechnology Council, the Food and Drug Law Institute, the Drug Information Association and the Regulatory Affairs Professionals Society where she was awarded the distinction of Fellow for her contributions to the regulatory profession. Prior to joining Genzyme in 2007, Ms. Williamson was Vice President of Regulatory Affairs and Quality Assurance for Serono. She holds an MBA from Northeastern University in Boston, MA and a BA degree in Psychology from Skidmore College in Saratoga Springs, NY.

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