Speaker Biographies
2020 Virtual Nonprofit Forum
June 29-30, 2020

Ronald Bartek

Ronald Bartek is the co-founder and President of Friedreich's Ataxia Research Alliance; Board of Directors and past Chairman of the National Organization for Rare Disorders; Board of Directors, Alliance for a Stronger FDA and Alliance for Regenerative Medicine; Member, National Center for Advancing Translational Sciences (NCATS) National Advisory Council; Vice Chair, NIH/NCATS Cures Acceleration Network Review Board; member, Food and Drug Administration Clinical Trials Transformation Initiative Patient Engagement Collaborative; and four-year member, NIH National Advisory Neurological Disorders and Stroke Council. He was the former partner and president of a business development/government affairs firm. Mr. Bartek's professional experience also includes 20 years of federal executive and legislative branch service in defense, foreign policy, and intelligence (six years on House Armed Services Committee staff; four years at the U.S. State Department; one year on the U.S. Delegation to Intermediate-Range Nuclear Forces Treaty talks, Geneva; and six years as CIA analyst, including a year as Intelligence Community representative to U.S. arms control committees). Following graduation from the United States Military Academy at West Point, Mr. Bartek spent four years as an Army officer, serving as a company commander in Korea and an Infantry and Military Intelligence officer in Vietnam. He has a master's degree in Russian Area Studies from Georgetown University.

Martina Bebin

Dr. Martina Bebin is a professor of neurology at the University of Alabama at Birmingham (UAB), and a pediatric neurologist at North Alabama Children's Specialists in Huntsville, AL. Serving on the Board of Directors for the Tuberous Sclerosis Alliance and as the co-director of the UAB Tuberous Sclerosis Complex (TSC) clinic, Dr. Bebin oversees the comprehensive care of individuals suffering from a rare, multi-system genetic disease that causes benign tumors to form in vital organs. She was named 2019 Woman of Impact by Yellowhammer News.

In 2013, Dr. Bebin's contribution to TSC research earned her the Manual R. Gomez award, an annual honor presented to the individual who has made significant breakthroughs in TSC research, from the Tuberous Sclerosis Alliance.

In addition to her incredible work with TSC, Dr. Bebin is a member of the Department of Neurology and Epilepsy Center at UAB and a member of the Child Neurology Society Legislative Affairs Committee. Her primary research interest is the early identification of epilepsy in infants, particularly discovering new therapies and treatments for these children. Dr. Bebin later earned her medical degree from the University of Mississippi School of Medicine, her MPA from Harvard University, and completed her residency work at the Mayo Graduate School of Medicine.

Geraldine Bliss

Geraldine Bliss is the parent of a 20-year-old son with Phelan-McDermid Syndrome (PMS), caused by a partial deletion of the SHANK3 gene. Because of the severity and refractory nature of her son's epilepsy, she made a promise to him to do everything she could to help him get better, which led her to become the research support committee chairperson of the Phelan-McDermid Syndrome Foundation (PMSF). She currently serves on PMSF's board of directors. Her vision and personal mission are to accelerate research that will lead to effective treatments and eventually cures for Phelan-McDermid Syndrome.

Kurt Fischbeck

Dr. Kurt Fischbeck is chief of the Neurogenetics Branch at the National Institute of Neurological Disorders and Stroke (NINDS) at National Institutes of Health (NIH) and received Bachelor of Arts and Master of Arts degrees from Harvard University and an M.D. degree from Johns Hopkins University. After a medical internship at Case Western Reserve University in Cleveland and a neurology residency at the University of California in San Francisco, he conducted postdoctoral research on muscular dystrophy at the University of Pennsylvania. In 1982, he joined the faculty in the Neurology Department at the University of Pennsylvania Medical School. In 1998, he came to the NINDS as Chief of the Neurogenetics Branch. He received the Cotzias Award from the American Academy of Neurology and the Jacoby Award from the American Neurological Association and was elected to the Institute of Medicine. His research group is identifying the causes and studying the mechanisms of hereditary neurological and neuromuscular diseases with the goal of developing effective treatment for these disorders.

Paul Gross

Paul Gross is an adjunct associate professor of Population Health Sciences at the University of Utah. He is a driving force in accelerating clinical and translational research in neuroscience for cerebral palsy and hydrocephalus. He is chairman and founder of the Cerebral Palsy Research Network – a 20 center effort in North America to conduct high quality clinical research for CP. He is the past chairman of the Hydrocephalus Association (HA), the co-founder of the Hydrocephalus Clinical Research Network (hcrn.org), and co-founder of the Adult Hydrocephalus Clinical Research Network (ahcrn.org). In his role as vice-chair of the HA

Research Committee, he has played a leadership role in the creation of the HA Network for Discovery Science – a virtual laboratory to accelerate basic science advancement through the collaboration of neuroscientists in hydrocephalus. In 2015, he received the "Making a Difference" award from the American Academy of Cerebral Palsy and Developmental Medicine. He recently completed his term as an Advisory Council Member for NINDS. Prior to ramping up his focus on advancing medical research, he was CEO of a web startup, a Senior Vice President with the Microsoft Corporation, and with Borland International.

Jill Jarecki

Dr. Jill Jarecki is the Chief Scientific Officer at Cure SMA, and she has worked there since 2005. In this capacity, she has overseen over $40 million in SMA research investments, including in basic research, preclinical drug development, clinical research, and regulatory work. In the area of preclinical drug development, Dr. Jarecki specifically managed a scientific project team for a Cure SM owned drug program, through a pre-IND meeting at the FDA, through designation of orphan disease status from the Orphan Products Office of the FDA, and finally through successful out-licensing of the program for clinical development to several industry partners. Prior to working at FSMA, she spent 5 years in the biotech industry, where she led project teams developing HTS assays, conducting high-throughput screens, and validating the bioactivity of small molecules in disease relevant assays. In this capacity she has worked on an SMA drug program, as well as several cancer-focused projects.

Petra Kaufmann

Dr. Petra Kaufmann is the Senior Vice President, Translational Medicine & Clinical Development at AveXis, Inc. Previously, she was the director of both the Office of Rare Diseases Research and the Division of Clinical Innovation at National Center for Advancing Translational Sciences (NCATS) at the NIH. Her work included overseeing NCATS' Rare Diseases Clinical Research Network, Genetic and Rare Diseases Information Center, and Clinical and Translational Science Awards Program as well as the NIH/NCATS Global Rare Diseases Patient Registry Data Repository/GRDR® program. Before joining NCATS, Kaufmann was the director of the Office of Clinical Research at NINDS, where she worked with investigators to plan and execute a large portfolio of clinical research studies and trials in neurological disorders, including many in rare diseases. She established NeuroNEXT, a trial network for Phase II trials using a central institutional review board, streamlined contracting, active patient participation in all project phases, and a scientific and legal framework for partnership with industry.

A native of Germany, Kaufmann earned her M.D. from the University of Bonn and her M.Sc. in biostatistics from Columbia University's Mailman School of Public Health. She completed an internship in medicine at St. Luke's/Roosevelt (now part of Mt. Sinai) in New York City, training in neurology and clinical neurophysiology at Columbia University, and a postdoctoral fellowship in the molecular biology of mitochondrial diseases at Columbia's H. Houston Merritt Clinical Research Center for Muscular Dystrophy and Related Diseases.

Walter Koroshetz

Dr. Walter Koroshetz was selected as Director of NINDS in 2015. Dr. Koroshetz joined NINDS in 2007 as Deputy Director, and he served as Acting Director from October 2014 through June 2015. He has held leadership roles in a number of NIH and NINDS programs including the NIH's BRAIN Initiative, the Traumatic Brain Injury Center collaborative effort between the NIH intramural program and the Uniformed Services University of the Health Services, and the multi-year work to develop and establish the NIH Office of Emergency Care Research to coordinate NIH emergency care research and research training. Before joining NINDS, Dr. Koroshetz served as Vice Chair of the neurology service and Director of Stroke and Neurointensive Care Services at Massachusetts General Hospital (MGH). He was a professor of neurology at Harvard Medical School (HMS) and led neurology resident training at MGH between 1990 and 2007. Over that same period, he co-directed the HMS Neurobiology of Disease Course with Drs. Edward Kravitz and Robert H. Brown.

A native of Brooklyn, New York, Dr. Koroshetz graduated from Georgetown University and received his medical degree from the University of Chicago. He trained in neurology at MGH, after which he did postdoctoral studies in cellular neurophysiology at MGH with Dr. David Corey, and later at the Harvard neurobiology department with Dr. Edward Furshpan, studying mechanisms of excitotoxicity and neuroprotection. He joined the neurology staff, first in the Huntington's Disease unit, followed by the stroke and neurointensive care service. A major focus of his clinical research career was to develop measures in patients that reflect the underlying biology of their conditions. With the MGH team he discovered increased brain lactate in HD patients using magnetic resonance (MR) spectroscopy. He helped the team pioneer the use of diffusion-and perfusion-weighted MR imaging and CT angiography-and-perfusion imaging in acute stroke.

Jane Larkindale

Dr. Jane Larkindale is the Vice President for Research Development at the Friedreich's Ataxia Research Alliance (FARA) and has concurrently led the Duchenne Regulatory Science Consortium (D-RSC) at the Critical Path Institute since 2015, serving as the consortium's Executive Director. In both positions, she is dedicated to accelerating drug development for these rare diseases, by promoting increased efficiency, increased cooperation, shared infrastructure, and supporting high quality science. Through D-RSC, she leads an international consortium dedicated to developing regulatory-ready drug development tools for Duchenne Muscular Dystrophy. At FARA, she supports research, develops infrastructure, and helps researchers source the tools and collaborators they need. She is a molecular biologist by training, having completed her D.Phil. (Ph.D.) in the Department of Plant Sciences at Oxford University in 2001, which she attended on a Rhodes Scholarship.

Peter Marks

Dr. Peter Marks is the director of the Center for Biologics Evaluation and Research (CBER) at the Food and Drug Administration (FDA). The center is responsible for assuring the safety and effectiveness of biological products, including vaccines, allergenic products, blood and blood

products, and cellular, tissue, and gene therapies. Dr. Marks and center staff are committed to facilitating the development of biological products and providing oversight throughout the product life cycle including reviewing and providing advice during product development, evaluating applications and making approval decisions based on safety and effectiveness data, monitoring the safety of biological products, and conducting research that supports product development and characterization.

Dr. Marks received his graduate degree in cell and molecular biology and his medical degree at New York University. Following this, he completed an Internal Medicine residency and Hematology/Medical Oncology fellowship at Brigham and Women's Hospital in Boston, where he subsequently joined the attending staff as a clinician-scientist and eventually served as Clinical Director of Hematology. He then worked for several years in the pharmaceutical industry on the clinical development of hematology and oncology products before returning to academic medicine at Yale University where he led the Adult Leukemia Service and served as Chief Clinical Officer of Smilow Cancer Hospital. He joined the FDA in 2012 as Deputy Center Director for CBER and became Center Director in 2016.

Melanie Quintana

Dr. Melanie Quintana is a Senior Statistical Scientist at Berry Consultants, where she specializes in designing Bayesian adaptive clinical trials across a wide range of therapeutic areas. Her work includes numerous examples of designing platform clinical trials in rare and progressive disease with a focus on developing models of disease progression to design better and more powerful clinical trials. Before joining Berry Consultants, she earned her Ph.D. in Statistics from Duke University and went on to pursue a Postdoc in Biostatistics at the University of Southern California (USC). While at Duke and USC, she worked closely with collaborators within multiple large studies and consortiums around the nation to develop and implement analytical strategies to assess genetic risk factors for various complex diseases.

Nina Schor

Dr. Nina Schor is Deputy Director of the NINDS and a Senior Faculty Associate at the University of Rochester. Dr. Schor graduated cum laude from Yale University with a B.S. degree in Molecular Biophysics and Biochemistry and as a Scholar of the House in Chemistry Research in 1975. She received her Ph.D. in Medical Biochemistry from Rockefeller University and the laboratory of Dr. Anthony Cerami in 1980 and her M.D. from Cornell University Medical College in 1981. Dr. Schor pursued residency training in Pediatrics at Boston Children's Hospital (1981-1983) under Dr. Mary Ellen Avery and Child Neurology at the Longwood Area-Harvard Neurology Program (1983-1986) under Dr. Charles Barlow. During residency, she also pursued a postdoctoral fellowship in the laboratory of Dr. Manfred Karnofsky at Harvard. During this time, she began her studies of neuroblastoma, aimed at understanding the neurobiology of this tumor and exploiting this understanding to design and test in preclinical models, novel strategies for the therapy of chemoresistant neuroblastoma. For the next 20 years, Dr. Schor rose through the academic and administrative ranks at the University of Pittsburgh, ultimately becoming the Carol Ann Craumer Professor of Pediatric Research, Chief of the Division of Child

Neurology in the Department of Pediatrics, and Associate Dean for Medical Student Research at the medical school. In 2006, Dr. Schor became the William H. Eilinger Chair of the Department of Pediatrics, and Pediatrician-in-Chief of the Golisano Children's Hospital at the University of Rochester, posts she held until January 2018, when she joined NINDS.

Eric Sid

Dr. Eric Sid joined the NIH/National Center for Advancing Translational Sciences (NCATS) as a Presidential Management Fellow (PMF) in the Office of Rare Diseases Research (ORDR) in September 2017. He since has become a program officer in ORDR, where he leads the Genetic and Rare Diseases (GARD) Information Center. GARD provides free, comprehensive, plain language information on rare and genetic diseases to the public and is accessible through both GARD's website and contact center. Dr. Sid is the lead for the Rare Diseases Registry (RaDaR) program, which offers guidance for establishing and maintaining patient registries. He also oversees the NCATS Toolkit for Patient-Focused Therapy Development, which disseminates best practices for patient-partnered research through collaborations between patients and caregivers, community organizations, researchers and NIH/U.S. Food and Drug Administration staff.

He received his M.D. and MHA degrees from the University of Washington's School of Medicine and School of Public Health, respectively. In 2019, he completed his PMF, which included a rotation with the VA Center for the Study of Healthcare Innovation, Implementation and Policy of the VA Greater Los Angeles Healthcare System.

Yael Weiss

Dr. Yael Weiss is Vice President of Business Development at Ultragenyx, a company specializing in the development of therapies for the treatment of rare genetic diseases. Prior to joining Ultragenyx, she spent 10 years at Merck, ultimately heading the group that evaluates the clinical, regulatory and safety aspects of Merck's regional deals globally. Her earlier roles at Merck included Director of External Scientific Affairs and World Wide Licensing, West Coast and Medical Director at Merck Israel, a role in which she oversaw the company's clinical trials and supported the sales and marketing organizations. Before joining Merck, Dr. Weiss served as Director of Medical Affairs and Business Development at Genzyme Israel and spent several years at Columbine Ventures. Dr. Weiss began her career practicing medicine at Sheba Medical Center. Dr. Weiss received her M.D. degree from Hadassah Medical School in Jerusalem, and her Ph.D. in molecular genetics from the Weizmann Institute of Science in Rehovot, Israel.

Monica Weldon

In November of 2012, Monica Weldon's twin son, Beckett, was one of the first six patients in the world to be diagnosed with the gene mutation SYNGAP1 (6p21.3). Ms. Weldon began a journey to find answers to help her son. She started to blog about his progress which ultimately led to building a community of parents and caregivers who now constitute a strong support group.

She retired in 2016 after 23 years in education teaching secondary science. Her new focus is on building the programs and mission of Bridge the Gap – SYNGAP Education and Research Foundation. She is the Primary Investigator on the SYNGAP1 (MRD5) Registry and Natural History Study. She is a life member of the Worldwide Association of Female Professionals and a member of the first class of 2017 Illumina Ambassadors, established in the US. In addition, she is an author, public speaker, and consultant who advocates for rare disease legislation at both the federal and state levels. Her scientific publications include *Nature Neuroscience*, the *Journal of Neurodevelopmental Disorders*, and the *Journal of Pediatrics*. She is the author of a book about her son Beckett's diagnostic journey called "My Special Boy - Slow Moving Stream." She is a graduate of East Texas Baptist University with a Bachelor of Science degree in biology and psychology (1991) and has a Secondary Certification in Education (1995). She is the wife of Chris Weldon and has five beautiful children, Haleigh (27), Taylor, USMC (24), Sawyer (22), and twins Beckett and Pyper (10).

Timothy Yu

Dr. Timothy Yu is an attending physician in the Division of Genetics and Genomics at Boston Children's Hospital and an assistant professor in pediatrics at Harvard Medical School. The Yu lab conducts genome-wide and worldwide searches to find the genes that are responsible for autism spectrum disorders (ASDs) and other neurodevelopmental conditions. The lab has a emphasis on biallelic/recessive mutations (i.e., impacting both copies of a gene) in ASD, and has uncovered a striking enrichment of biallelic gene disruptions in affected individuals, especially in girls. The lab also finds ways to bring genomic tools to the bedside to help care for patients with genetic disorders. These projects range from the deployment of genome sequencing in the neonatal intensive care unit to the design and delivery of genome-guided therapeutics for sick children.

Dr. Yu completed his undergraduate degree in biochemistry and molecular biology at Harvard College, his M.D. and Ph.D. degree (in Neuroscience) from the University of California at San Francisco, clinical neurology training at Massachusetts General Hospital and Brigham and Women's Hospital, and a fellowship in neurodevelopmental genetics at MGH and Boston Children's Hospital. He joined the faculty in the Division of Genetics and Genomics as Instructor in 2010 and Assistant Professor in 2013.