Niemann-Pick Type C Clinical Roundtable
Expert Attendees

Elizabeth Berry Kravis, MD, PhD
Rush University Medical Center
Principal Investigator, NPC Newborn Screening Initiative

Elizabeth Berry-Kravis MD, PhD is a Professor of Pediatrics, Neurological Sciences, and Biochemistry at Rush University Medical Center. She established the Fragile X Clinic and Research Program in 1992, and provides care to over 600 patients with FXS and additional patients with other NDDs and Neurogenetic diseases through specialty clinics and programs for Phelan McDermid syndrome (PMS), Neimann-Pick type C (NPC), Angelman syndrome (AS), Rett syndrome (RS), creatine transport deficiency (CTD), Battens disease (CLN), and pantothenate kinase-associated neurodegeneration (PKAN). She conducts clinical and basic research on FXS, other NDDs and neurodegenerative diseases, and her lab studies genetic and molecular mechanisms in FXS. She has studied phenotypes in FXS, has been PI on natural history, screening, outcomes and biomarker projects and clinical trials in FXS, NPC, AS, RS, PMS, CLN, CTD, PKAN, Down syndrome, autism spectrum disorder and ID populations, and has been a leader in the translational effort to develop targeted treatments for disease modification in FXS and other neurogenetic diseases.

Simona Bianconi, MD
National Institutes of Health

Simona Bianconi is an assistant research physician at NIH, Eunice Kennedy Shriver Institute for child health and development who evaluates patients with a number of rare diseases including Niemann Pick Disease at the NIH Clinical Center as part of a multidisciplinary team. She has been involved in a number of observational and clinical trials in NPC including the international phase 2/3 trial of Vts-270. She obtained her medical degree at the University of Vienna in 2003, completed pediatric residency in 2006 at New York- Presbyterian Brooklyn Methodist Hospital, and attained a fellowship in clinical genetics in 2011 at the National Genome Research Institute.

Nicole Yanjanin Farhat, NP
National Institute of Child Health and Human Development

Nicole Yanjanin Farhat is a pediatric nurse practitioner and has been involved in clinical research at the NIH since 2002. She joined the Porter lab in 2006 and has since worked on basic and clinical aspects of NPC research, with her role including patient care, clinical and translational research design and implementation, regulatory management of protocols as well as analysis and preparation of data for publication. Nicole was the Lead Associate Investigator on the NIH Phase 1 trial of intrathecal HPBCD in NPC1 and is the Central Blinded Rater in the multi-site VTS301 Phase 2/3 trial, using the NPC neurologic severity scale which she and her NIH colleagues developed. Additional current clinical studies include the NPC natural history study, in which over 115 individuals with NPC have enrolled, and which has provided biospecimens and clinical data leading to the collaborative development of a blood-based method of diagnosing NPC disease.

Caroline Hastings, MD
UCSF School of Medicine

Caroline Hastings received her M.D. from the University of California, Davis in 1986 and subsequently completed her Pediatric residency and fellowship in Pediatric Hematology and Oncology at Children's Hospital & Research Center Oakland, California. She is on the faculty at UCSF School of Medicine and Benioff Children's Hospital Oakland, and serves as the Director of the Pediatric Hematology/Oncology Fellowship Program. Her academic interests include clinical trial development in high risk and relapsed extramedullary leukemia as well as Niemann-Pick Type C disease. She was the first physician in the US to develop and use hydroxypropyl-β-cyclodextrin in children with NPC and developed the first FDA approved compassionate use protocol. Building on her experience with compassionate use of cyclodextrins, Dr. Hastings serves as the Principal Investigator for a US phase I clinical trial supported by CTD Holdings (CTDH) to test intravenous cyclodextrins formally in NPC. She also serves as the Senior Clinical Advisor to CTD's EU phase I/II clinical trial, Co-Chair of CTD's Family and Physicians Listening Circle, and member of CTD's Scientific Advisory Board. Dr. Hastings is currently developing future trials in Lysosomal Storage Diseases including Tay Sachs disease.

Dr. Hastings also has a special interest in Neuro-Oncology (patients with brain and spinal cord tumors). She is involved with advocacy on a national and regional level and serves on the Executive Board of Trustees for the American Society of Pediatric Hematology Oncology and chairs the Finance committee. She leads awareness and philanthropic events and has personally raised several million dollars to support local programs in research and clinical care as well as pediatric clinical trials in rare disease.

Dr. Hastings enjoys spending time with her husband and three daughters, travelling, all animals, reading about economics and creating the perfect garden.

Jean-Baptiste Le-Pichon, MD, PhD
Children's Mercy Kansas City

Dr. Le Pichon is a Child Neurologist at Children's Mercy Kansas City. He is Associate Professor in the Division of Neurology. He did an MD/PhD at Baylor College of Medicine and completed his Child Neurology residency at Texas Children's Hospital. He is board certified both in Neurology with Special Qualifications in Child Neurology and in Epilepsy. Dr. Le Pichon is especially interested in neurogenomics. He leads the Mitochondrial and the Neurogenomic clinics at Children's Mercy Kansas City. In addition to his clinical interests he continues to be active in research and has contributed to a number of publications, mostly in neurogenomics and epilepsy. He is the site Principal Investigator for the VTS-270 compassionate use of Intrathecal 2-Hydroxypropyl-β-cyclodextrin for NPC1 patients.

Professor Marc C. Patterson, MD, FRACP, FAAN, FANA
Mayo Clinic

Marc Patterson was born and educated in Australia, and trained in neurology, child neurology and neurometabolic disease at the University of Queensland, at Mayo Clinic, and at NINDS/NIH, the last mentioned under the guidance of Roscoe Brady, MD. He is currently Professor of Neurology, Pediatrics and Medical Genetics. He was Director of the Child Neurology Training program at Mayo 2008-2016), and Chair of the Division of Child and Adolescent Neurology (2008-2017) (Mayo Clinic administrative positions are term-limited). Dr Patterson had previously served as Professor and Director of Pediatric Neurology at Columbia University in New York (2001-2007). He has served as a member of the Neurology topic advisory group for revision of the ICD-10 of the World Health Organization, the Committee on Adverse Effects of Vaccines for the Institute of Medicine, and leads the Education Core of the NIH-funded Lysosomal Disease Network.

He has served in a number of positions in the Child Neurology Society, American Academy of Neurology, American Board of Psychiatry and Neurology and American Neurological Association. Dr Patterson has served on the editorial board of Neurology, on the oversight committee of Annals of Neurology and is currently an Editor for the Journal of Inherited Metabolic Disease. He became Editor-in Chief of the Journal of Child Neurology on January 1st, 2014, and subsequently Editor-in-Chief of its open-access sister journal, Child Neurology Open.

His research and practice has focused on rare diseases in children, including multiple sclerosis and neurometabolic disorders in general, with special interests in Niemann-Pick disease, type C (NPC), other lysosomal diseases (including glycoproteinoses), mitochondrial cytopathies and congenital disorders of glycosylation, areas in which he has published more than 230 peer-reviewed papers and book chapters. He has presented widely through the United States and internationally, both to professional and lay organizations. He serves on the scientific advisory boards of several rare disease foundations. Dr Patterson has received funding support from NIH, industry and private foundations.

Dr Patterson participated in the first controlled clinical trial of (potential) disease modifying therapy in NPC, using cholesterol-lowering agents at the National Institutes of Health; subsequently, he was principal investigator for the pivotal trial of miglustat which led to approval of this agent in 45 countries for the management of NPC. He is a participant in the current trial of arimoclomol in NPC, and is a member of the advisory board assisting the development of n-acetyl-leucine for NPC and other lysosomal disorders. He has led teams developing guidelines for the diagnosis and management of NPC, and currently leads the scientific advisory Board for the International Niemann-Pick disease registry. He has served the National Niemann-Pick Disease Foundation since its first meeting of the National institutes of Health in 1991, and also serves other national and international foundations devoted to NPC, including NPUK and the Niemann-Pick Selbsthilfegruppe eV (Deutschland). He has provided continuing care for many families with Niemann-Pick C, some for periods of more than 25 years.

Forbes D Porter, MD, PhD
National Institute of Child Health and Human Development

Dr. Forbes D. Porter is a Senior Investigator in the Intramural Research Program of the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD, NIH). He has also served as the Clinical Director for NICHD since 2010. Dr. Porter earned his MD and PhD from Washington University in St. Louis and subsequently trained in Pediatrics and Clinical Genetics at St. Louis Children's Hospital.

Dr. Porter formed his own basic and clinical research groups at the NIH starting in 1996. His research has been primarily focused on disorders of cholesterol homeostasis, specifically Smith-Lemli-Opitz syndrome (SLOS) and Niemann-Pick disease, type C (NPC). More recently his research group has also been involved in studies of CLN3 and creatine transport deficiency. His group's goal is to combine both basic and clinical science to understand the pathological processes contributing to these disorders and to develop and test therapeutic interventions. Dr. Porter's laboratory also works on basic and translational studies of NPC.

NPC1 clinical trials

1. NPC Natural history study
2. VTS-270 Phase 1/2 study
3. VTS-270 Phase 2/3 study
4. Combined intrathecal and intravenous VTS-270 study
5. Neonatal VTS-270 for Liver disease
6. NPC Vorinostat study
7. NPC NAC study

Cynthia M. Powell, MD, FAAP, FACMG
University of North Carolina at Chapel Hill School of Medicine

Dr. Cynthia Powell is a Professor of Pediatrics and Genetics at the University of North Carolina at Chapel Hill School of Medicine where she is also the Director for the Medical Genetics and Genomics residency program. She is a board-certified clinical geneticist, cytogeneticist, pediatrician and genetic counselor, current President of the Association of Professors of Human and Medical Genetics, Chair of the HRSA Advisory Committee on Heritable Disorders in Newborns and Children and a member of the Board of Directors of the American College of Medical Genetics and Genomics.

Latha Soorya, PhD
Rush University Medical Center

Dr. Latha Soorya is a clinical psychologist, behavior analyst, Associate Professor, and Director of the autism center in the Department of Psychiatry at Rush University Medical Center in Chicago, IL. Her expertise is in the diagnostic, neurocognitive, and behavioral characterization of neurodevelopmental conditions and application of these findings to genetics, imaging, and treatment studies. She is experienced evaluator and has served as an investigator on federally-funded autism and rare genetic disease networks. In this work, she has worked on measurement challenges related to diagnostic and outcome data collection in developmentally diverse populations. Her active studies are funded by NIMH, NICHD, Autism Speaks, and the Simons Foundation. Dr. Soorya's treatment research is focused on development of integrated behavioral-pharmacological interventions with capacity to improve cognitive and behavioral features of idiopathic autism and medically complex rare neurodevelopmental conditions.

Audrey Thurm, PhD
National Institute of Mental Health

Audrey Thurm, PhD, is Director of the Neurodevelopmental and Behavioral Phenotyping Service in the Office of the Clinical Director, part of the National Institute of Mental Health (NIMH)'s Intramural Research Program (IRP). After receiving a B.S. in human development from Cornell University, she received training in child clinical psychology at DePaul University, trained as an intern at Boston Children's Hospital/Harvard Medical School, and conducted a post-doctoral fellowship at Johns Hopkins School of Medicine. She has been at NIMH since 2002, serving in the extramural program until 2006, at which time she moved to the IRP to engage in research on autism spectrum disorder (ASD) and other related neurodevelopmental disorders. Among the many rare genetic disorders Dr. Thurm studies with respect to neurodevelopmental outcomes, she has been involved as an associate investigator and research clinician conducting neurodevelopmental assessment for several NPC-1 studies at NICHD, including a natural history study and studies of Cyclodextrin.

Allison Wainer, PhD
Rush University Medical Center

Dr. Wainer is a licensed clinical psychologist, Assistant Professor, and Research Director at the Autism Assessment, Research, Treatment, and Services (AARTS) Center in the Department of Psychiatry at Rush University Medical Center (RUMC). The AARTS Center program of research spans genetics to cognitive and behavioral phenotyping to the development and evaluation of intervention and service delivery models for individuals with autism spectrum disorder (ASD) and related neurodevelopmental disorders (NDDs). Dr. Wainer's own work focuses primarily on identifying ways to support the transportation of effective intervention to the "real world" to increase access to high quality intervention for children with NDDs. Dr. Wainer has supported clinical research efforts for individuals with Fragile X, Angelman Syndrome, and Phelan McDermid Syndrome, and currently conducts neuropsychological assessments for Dr. Berry-Kravis' Niemann-Pick Type C research program at RUMC.

Melissa Wasserstein, MD
Children's Hospital at Montefiore

Melissa Wasserstein, M.D. is the Chief of the Division of Pediatric Genetic Medicine at the Children's Hospital at Montefiore and an Associate Professor of Pediatric and Genetics at the Albert Einstein College of Medicine (AECOM). Dr. Wasserstein received her B.S. in biology from Cornell University and her medical degree from New York University School of Medicine. She completed her pediatrics residency and medical genetics fellowship at the Mount Sinai School of Medicine, where she subsequently remained as a faculty member before moving to Einstein-Montefiore in 2016.

In addition to diagnosing and managing patients with inborn errors of metabolism, Dr. Wasserstein is a clinical investigator. Her research focuses on expanding newborn screening for rare inherited disorders and evaluating the associated ethical, legal and social implications. She is a Principal Investigator of NYCKidSeq, a multisite NHGRI and NIMHD funded program studying the implementation of genomic medicine in underserved populations. She has been the Principal Investigator on numerous clinical trials evaluating novel therapeutics for rare genetic diseases.

Dr. Wasserstein is a Co-Chair of the Newborn Screening Translational Research Network's Steering Committee. She is on the Executive Committee of AECOM's Rose F. Kennedy Intellectual and Developmental Disabilities Research Center where she also serves as Associate Director of the Human Clinical Phenotyping Core. In addition, she is on the Scientific Advisory Boards of the International Niemann-Pick Disease Association, the MSUD family support group, and WylderNation.

Amy White, MD
Southwest Pediatric Associates

Originally from Wisconsin, Dr. White attended the University of Wisconsin-Madison for college and Medical School. She completed her residency in Pediatrics at the University of North Carolina at Chapel Hill.

After residency Dr. White was an attending physician in the Pediatric Emergency Department at UNC before moving to Austin, TX in 2004. She has practiced primary care pediatrics and urgent care medicine in Austin since then. She currently practices Pediatrics at Southwest Pediatric Associates in Austin and currently has 2 patients with NPC1. Dr. White served on the Executive Board of The Volunteer Healthcare Clinic in Austin, TX and is currently on the board of The Firefly Fund.

Kara Woolgar RN, BSN - Phoenix Children's Hospital

Kara Woolgar received her Bachelor of Science in nursing (BSN) from Kent State University School of Nursing in 2005. She has worked at Phoenix Children's Hospital as an RN for the past 14 years including 7 years in the pediatric intensive care unit (PICU) and 2 years in the post anesthesia care unit (PACU). She currently is the lysosomal program coordinator in outpatient Genetics. She is committed to lysosomal patients through care coordination, research, and outreach. She is the research nurse on several studies including Shire HGT-HIT-094 Extension study for intrathecal therapy in Hunter Patients, Genzyme rare disease registry, Shire Hunter Outcome Survey and Phenotypic-Genotypic-relationship in Cobalamin C disease. Past studies include Alexion LAL-CL 08 phase 2, open label study of sebelipase alfa in infants with lysosomal acid lipase deficiency. Kara is also very involved with NPC patients and is the research nurse for an infant patient who is receiving IV cyclodextrin VTS-270 through expanded access. Kara presented a poster at the Niemann-Pick type C research Conference in 2018 and a podium presentation at the WORLD symposium 2019 on "Intravenous 2-hydroxypropyl-beta-cyclodextrin for a Niemann-Pick disease type C1 infant with liver cirrhosis." In addition to the NPC presentations, she has presented several other posters at several conferences including the WORLD Symposium 2017 and 2019 and the International Symposium on MPS and related disease 2018. She is also involved in outreach for lysosomal patients and planned a regional MPS patient meeting in 2016. She is a current speaker for Genzyme. Kara was the recipient of the Daisy Award for extraordinary nurses in 2011 and has been nominated for Phoenix Children's Hospital nurse of the year in 2016, 2018 and 2019.