Advisory Panel on Rare Diseases Biographies

Scott Berns, MD, MPH, FAAP

Chair, Advisory Panel on Rare Disease
Stakeholder Affiliation: Clinicians

Chief Executive Officer, National Institute for Children's Health Quality

Email: sberns@nichq.org

Term Ends August 2021

Scott Berns is the President and CEO of the National Institute for Children's Health Quality (NICHQ), a nonprofit organization that works to improve children's health, and a co-Founder of The Progeria Research Foundation (PRF), which aims to discover treatments and the cure for Hutchinson-Gilford progeria syndrome and its aging-related conditions.

As Chair of the PRF Board, Berns ensures that PRF is a key force behind the discovery of the progeria gene, and he has developed programs and services to aid both those affected by progeria and the scientists who conduct progeria research. Berns has made multiple media appearances as an expert in perinatal health, and his son Sam and family were featured in an HBO Emmy Award–winning documentary titled *Life According to Sam*.

Berns is a board-certified pediatrician and pediatric emergency physician. He is a Clinical Professor of Pediatrics at the Warren Alpert Medical School of Brown University in Providence, Rhode Island, and a Clinical Professor of Health Services, Policy, and Practice at the Brown School of Public Health. Berns received his bachelor's and medical degrees from Boston University. He completed his residency in pediatrics at Rhode Island Hospital and completed a fellowship in pediatric emergency medicine at Children's National Medical Center in Washington, DC. He earned a Master of Public Health with a concentration in health, policy, and management from the Harvard School of Public Health.

At PCORI's fifth Annual Meeting in September 2019, Berns and his wife Leslie Gordon, MD, PhD, presented the Opening Keynote.

Doug Lindsay, BS
Co-Chair, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers and Patient Advocates

Personal Medical Consultant and Founder, Doug Says LLC.

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Term Ends August 2022

Doug Lindsay became ill at age 21 and spent the next 11 years home- and bedbound until he figured out what was wrong, developed novel treatments to keep his rare autonomic-adrenal condition at bay, and eventually developed the innovative surgery used to treat him.

During Lindsay's 14-year medical odyssey, he worked with 35 senior faculty at 28 institutions, developed new uses for five existing prescription drugs, won a national first court case protecting patients' rights, and developed the concept for the two successful, innovative adrenal surgeries used to treat his case.

Now, Lindsay is a nationally recognized speaker on the role of hope and character in innovation and in life. He is a Community Advisory Board member for Washington University's Institute of Clinical and Translational Sciences and Institute for Public Health. He also provides an in-depth personal medical consultant service to a small number of rare disease and complex condition patients who have found themselves trapped in the medical system.

Sarah Bacon, MS
Member, Advisory Panel on Rare Disease

Stakeholder Affiliation: Patients, Caregivers and Patient Advocates

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Term Ends August 2023

Sarah Bacon is a rare disease patient, advocate, and writer. Shortly after her 2013 diagnosis with lymphangioleiomyomatosis (LAM), and after the sequester cuts to the National Institutes of Health, she wrote an op-ed in The Atlantic, "Medical Research Cuts Have Immediate Health Effects." Since then, she has applied her 15 years of professional communications, advocacy, and issue-related campaign design experience to advancing rare disease research and policy.

Bacon suggested and raised funds for a novel study on the link between LAM and melanoma, which share several biomarkers, including proteins. Melanoma took her father's life and threatened hers. Fortunately, hers was caught early. The study bore meaningful results on the high incidence of melanoma family histories in LAM patients. When the LAM Lab faced losing a commitment from Novartis of a $200,000 in-kind donation for a pilot trial, Bacon advocated with Novartis executives and restored the donation.

Bacon secured bipartisan sponsors of The OPEN Act in the House of Representatives with Rare Disease Legislative Advocates, spoke on patient-driven medicine for Global Genes and The Milken Institute, and served on Global Genes' grant application review committee. She has written on rare diseases for The Atlantic, Fast Company, The Washington Post, and New York Magazine, and is currently working on her first book, Living with Zebras.

Roxanna Bendixen, PhD, OTR/L, FAOTA

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Researchers

Rehabilitation Scientist, Associate Professor, and Program Director, Department of Occupational Therapy at the University of Pittsburgh

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Terms Ends August 2021

Roxanna Bendixen is a Rehabilitation Scientist and Associate Professor at the University of Pittsburgh, Department of Occupational Therapy, School of Health and Rehabilitation Sciences. She also holds a secondary appointment in the Department of Neurology at the University of Pittsburgh. Dr. Bendixen's clinical expertise is in the area of pediatrics and early-onset neuromuscular disorders. Her principal responsibilities at the University of Pittsburgh are in research and teaching. Her current primary research interests include infants and children with early onset neuromuscular disorders; participation and quality of life; validity and reliability of pediatric sleep outcome measures in neuromuscular disorders; and community-based activity monitoring through mobile tracking devices. Her secondary line of research focuses on research engagement with healthcare, community, family, and policy stakeholders to develop organization-level interventions in support of individuals with Intellection and Developmental Disability. Bendixen has served on numerous rare-disease work groups, such as the American Academy of Neurology Muscular Dystrophy Measure Development Work Group and has provided expert scientific review for clinical guidelines in both Duchenne muscular dystrophy and congenital muscular dystrophies. She is a clinical evaluator, scientific reviewer, and member of the Cooperative International Neuromuscular Research Group. Bendixen is a graduate of the University of Florida, receiving her bachelor's and master's degrees in Occupational Therapy and her Doctorate in Rehabilitation Sciences.

Vanessa Boulanger, MSc

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers and Patient Advocates

Director of Strategic Partnerships, Amyloidosis Research Consortium

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Terms Ends August 2021

As the Director of Strategic Partnerships at the Amyloidosis Research Consortium (ARC), Vanessa Boulanger works with stakeholders to develop innovative programs and collaborative research initiatives. Prior to joining ARC, Boulanger was the Director of Research at the National Organization for Rare Disorders (NORD), where she led the strategic development, growth, and implementation of NORD's research and scientific activities, developing evidence-informed programs to support the rare disease community. She brings over 15 years of experience addressing health and social inequalities from a range of health and development perspectives to her position at ARC.

Boulanger holds an MSc degree in Global Health and Population from Harvard University and a BA in International Development and Social Change from Clark University.

Danielle Boyce, MPH
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers, and Patient Advocates

Senior Research Data Analyst, COPD Foundation

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Term Ends August 2023

Danielle Boyce is an award-winning rare epilepsy advocate, researcher, public speaker, and writer. Her work has appeared in dozens of scientific journals and her children's book, Charlie's Teacher, is used in children's hospitals throughout the country. She has served on several patient and caregiver advisory panels for academic institutions, pharmaceutical companies, nonprofits, the Food and Drug Administration, the American Academy of Pediatrics, and PCORI. Boyce currently works for the COPD Foundation and advises rare disease organizations on data science-related matters. She previously worked for Johns Hopkins Schools of Medicine and Nursing. She holds a master's in public health with a concentration in epidemiology and is currently working toward a doctorate in public administration.

After representing the caregiver voice on a PCORI-funded study, Boyce realized that serving on a PCORI advisory panel is the perfect marriage of her research and advocacy skills and is excited to serve patients through her continued involvement with PCORI. She lives outside of Philadelphia with her husband, Jim, and their four children.

Mathew Edick, PhD
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers, and Patient Advocates

Director of the Center for Strategic Health Partnerships, Michigan Public Health Institute

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Term Ends August 2023

Mathew J. Edick is the Director of the Center for Strategic Health Partnerships at Michigan Public Health Institute. He is an organizational PCORI Ambassador and has been a principal investigator, co-investigator, project manager, and programmatic/technical assistance lead on four PCORI-funded projects. Edick works with local, regional, and national-scale projects that engage diverse stakeholders to identify and implement innovative solutions for improving health outcomes. He received his doctorate in pharmacogenomics from the University of Tennessee.

Julie Gortze, RN

Member, Advisory Panel on Rare Disease Stakeholder Affiliation: Patients, Caregivers and Patient Advocates

Founder, Rare New England

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Term Ends August 2021

Julie Gortze is a registered nurse and Founder of Rare New England, a nonprofit patient advocacy organization focused on improving the lives of those living and working with rare and complex disorders. She is helping to create a rare-disease advisory council initiative that is forming through Connecticut legislation, and she is active on a similar bill in Massachusetts that would allow a group of rare-disease stakeholders to form a council to investigate issues in the rare-disease community and find solutions.

Through Rare New England, Gortze is a member of the Regional Genetics Network Steering Committee, Global Genes Alliance, and North Attleboro Commission on Disability. She is a 2017 Health Resources and Services Administration Regional Genetics Collaborative Advocate Leaders Partnership Program Recipient. She has been on various conference planning committees, including the New England Regional Genetics Group and Mito Hope and Help, and has helped organize Rare Disease Day State House events.

Gortze has personal experience with a complex disease and has learned firsthand the barriers patients and families must overcome while searching for a cause for symptoms. As a nurse, she also understands that most medical personnel do not have the familiarity to recognize, diagnose, and treat complex medical disorders, so they need help and support as well.

Salman Hussain, MPH

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Industry

Consultant, Charles River Associates

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Term Ends August 2023

Salman Hussain is a life sciences consultant at Charles River Associates (CRA), and a member of the Board of Directors for the National Alopecia Areata Foundation (NAAF). He has more than seven years of experience in healthcare research and has advised leading pharmaceutical and biotechnology companies across many therapeutic areas (with a specific focus in rare disease), both globally and in the United States.

Prior to joining CRA, Hussain conducted research at Massachusetts General Hospital and The Dartmouth Institute for Health Policy & Clinical Practice, specializing in the rare disease alopecia areata universalis and the interface between dermatology and mental health. While working with NAAF on a PCORI-funded project, he developed a patient-centered outcomes research and comparative clinical effectiveness research training program for patients. He was so inspired after this project that he became a PCORI Ambassador and has continued to advise his pharmaceutical clients and academic researchers on how they can meaningfully engage with patients to inform their research.

In addition to his work in alopecia areata, Hussain has advised and conducted substantial research for pharmaceutical companies to better understand the patient experience in many rare diseases, such as Duchene muscular dystrophy, spinal muscular atrophy types 3 and 4, acute lymphoblastic leukemia, maple syrup urine disease, nemaline myopathy, GM3-synthase deficiency, aromatic 1-amino acid decarboxylase deficiency, and acute hepatic porphyria

Tilicia Mayo-Gamble, PhD, MPH, MA

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers and Patient Advocates

Assistant Professor, Georgia Southern University

Email: dr.mayogamble@gmail.com

Term Ends August 2021

Tilicia Mayo-Gamble is an Assistant Professor for the Department of Health Policy and Community Health in the Jiann-Ping Hsu College of Public Health at Georgia Southern University. She conducts research on community-engaged approaches to improving protective health behaviors in patients with one or more chronic conditions. In her home life, Dr. Mayo-Gamble is the spouse to a husband with sickle cell disease. Much of her research interests are guided by her and her husband's experiences interacting with healthcare providers on sickle cell disease management and challenges with managing the disease at home. She recognizes that books and online text do not address her husband's needs—this information can be gained only through lived experience. She also understands the difficulties of providing input to healthcare providers and researchers about personal health. Dr. Mayo-Gamble became an advisory panel member to share the voices of individuals with sickle cell disease. She sees this as a unique opportunity to bring patient and caregiver perspectives into the discourse.

Nancy Rose, MD

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Clinicians

Adjunct Professor of Obstetrics and Gynecology, University of Utah

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Term Ends August 2023

Currently a Medical Consultant at the National Coordinating Center for the Regional Genetics Networks, Nancy Rose was previously the Director of Reproductive Genetics at Intermountain Healthcare in Salt Lake City Utah for 16 years, with the goal to decrease healthcare disparities and disseminate information prior to and during pregnancy as well as in the newborn period throughout the state. She has served as the President of the American Board of Medical Genetics and Genomics and Chair of the Committee on Genetics for the American College of Obstetricians and Gynecologists, and continues to serve on committees for the American College of Medical Genetics and Genomics, the American College of Obstetricians and Gynecologists, the American Board of Medical Genetics and Genomics, and the American Board of Medical Specialties. She remains an adjunct Professor of Obstetrics and Gynecology at the University of Utah.

Sherene Shalhub, MD, MPH

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Clinicians

Vascular Surgeon and Translational Researcher; Director of the Vascular Genetic Clinic, University of Washington

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Term Ends August 2021

Sherene Shalhub is a vascular surgeon and translational researcher at the University of Washington (UW) and the Director of its Multidisciplinary Vascular Genetics Clinic. She is a former NIH T32 Postdoctoral Fellow and recently completed the two-year Rising Star Program at the Clinical and Translation Science Awards (CTSA)-funded UW Institute of Translational Health Sciences. Her work aims to provide high-quality care to patients suffering from rare vascular conditions and to improve the success of surgical repair and long-term survival for these high-risk patients.

Saira Sultan, JD

Member, Advisory Panel on Rare Disease Stakeholder Affiliation: Policy Makers

CEO, Connect4Strategies

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Term Ends August 2022

Saira Sultan, JD, has been at the nexus of patient groups, the medical community, drug/device manufacturers, and governmental organizations, for more than two decades. Her passion for rare and ultrarare disease issues culminated in her founding of the Haystack Project in 2016. Through Haystack, Sultan has given voice to the experiences and perspectives of patients with rare and ultrarare conditions to influence research and policy making. She also continues to work with patient groups suffering from more common conditions such as cancer, sickle cell disease, pain, and addiction as the President and CEO of Connect 4 Strategies, a boutique reimbursement, value, and access consulting firm.

Her history with the cancer community stretches back nearly 20 years to when she directed government affairs for the Association of Community Cancer Centers. During her career, she also held leadership positions with Pfizer, Sanofi, and Medtronic. Sultan holds a juris doctor from the University of Virginia in Charlottesville, Virginia.

Laura Tosi, MD

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Clinicians

Founder and Director of the Bone Health Program, Children's National Hospital

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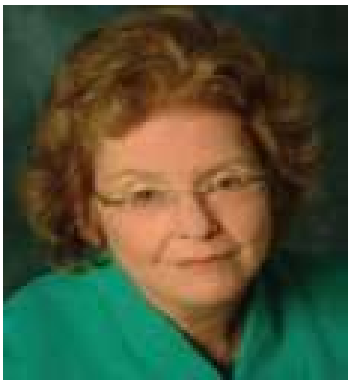
Term Ends August 2023

Laura L. Tosi serves on the Board of Directors of the Osteogenesis Imperfecta Foundation (OIF) and the US Bone & Joint Initiative. She serves as site principal investigator for the Brittle Bone Disease Consortium's Osteogenesis Imperfecta Longitudinal Study and has particularly supported the study's patient-reported outcomes research. Tosi served as the principal investigator for the OIF's first patient centered outcomes research project and is currently serving as a co-lead for an OIF PCORI-funded project on improving patient-centered outcomes. She is chair of the Rare Bone Disease Alliance Steering Committee and faculty chair of the Rare Bone Disease TeleECHO program.

Tosi is the Founder and Director of the Bone Health Program at Children's National Hospital and Associate Professor of orthopaedics and pediatrics at George Washington University in Washington, D.C. Her clinical practice focuses on the orthopedic care of children and young adults with physical disabilities and/or issues related to bone health/rare skeletal disease. She is a graduate of Harvard Medical School and received her orthopaedic training at the Columbia Presbyterian Hospital in New York and the Hospital for Sick Children in Toronto.



Naomi Aronson, PhD

Ex-Officio Member from PCORI's Methodology Committee

Executive Director, Clinical Evaluation, Innovation, and Policy, Blue Cross Blue Shield Association (BCBSA)

Email: naomi.aronson@bcbsa.com

Dr. Naomi Aronson leads BCBSA clinical effectiveness and policy engagement with government, regulatory agencies, and policy consortia. Her areas of leadership include comparative effectiveness, patient centered research, safety surveillance, regulatory science, and methodological standards. Previously, Dr. Aronson led the development of the BCBSA Technology Evaluation Center (TEC), now Evidence Street™ (ES), as a nationally recognized technology assessment program and an Evidence- based Practice Center (EPC) of the Agency for Healthcare Research and Quality (AHRQ).

She is a member of the Methodology Committee of the Patient-Centered Outcomes Research Institute (PCORI), the Health Technology Assessment International Health Policy Forum, the National Academy of Medicine Genomics Roundtable, the New Drug Development Paradigms (NEWDIGS) initiative of the MIT Center for Biomedical Innovation, and the Medical Device Innovation Consortium (MDIC) National Evaluation System for Health Technology (NEST) Governing Committee.

Prior to joining BCBSA, Dr. Aronson was a member of the Northwestern University faculty, specializing in the sociology of science and medicine. She also was a post-doctoral fellow in the Science, Technology and Society Program at the Massachusetts Institute of Technology.

RDAP Spring 2021 Presenters

Rare Disease Research Awards: Study Highlights and Discussion (1:15-2:10 PM)

Penny Mohr, MA
Acting Program Director, Research Infrastructure, PCORI
Email: pmohr@pcori.org

Jason Gerson, PhD
Senior Program Officer, Clinical Effectiveness and Decision Science, PCORI
Email: jgerson@pcori.org

Project Title: Comparative Effectiveness of Palliative Surgery vs. Additional Anti-seizure medications for Lennox-Gastaut Syndrome

Anne Berg, PhD
Role: PI

Research Professor of Pediatrics (Neurology and Epilepsy), Northwestern University Feinberg School of Medicine
Email: atberg@luriechildrens.org

Sandi Lam, MD, MBA
Role: Dual PI

Division Head, Neurosurgery; Professor of Neurological Surgery, Northwestern University Feinberg School of Medicine
Email: slam@luriechildrens.org

Tracy Dixon Salazar, PhD
Role: Stakeholder Partner

Executive Director, Lennox-Gastaut Syndrome (LGS) Foundation
Email: tracy@lgsfoundation.org

Project Title: Preserving Kidney Function in Children with Chronic Kidney Disease (PRESERVE)

Michelle Denburg, MD, MSCE
Role: Dual PI

Associate Professor of Pediatrics, Division of Nephrology, The Children's Hospital of Philadelphia & Perelman School of Medicine at the University of Pennsylvania
Email: denburgm@chop.edu

Anita Jovonokvska
Role: GLEAN Parent Partner; Co-Investigator

Email: anitaj@hotmail.com

Rebecca Levondosky
Role: GLEAN Parent Partner; Co-Investigator

Email: levondoskyfamily@gmail.com

Project Title: Utilizing PCORnet to Support Transition from Pediatric to Adult Centered Care and Reduce Gaps in Recommended Care in Patients with Congenital Heart Disease

Thomas Carton, PhD, MS
Role: PI

Chief Data Officer, Louisiana Public Health Institute
Email: tcarton@lphi.org

Anitha John, MD, PhD
Role: Dual PI

Medical Director, Washington Adult Congenital Heart Program, Children's National Hospital
Email: AnJohn@childrensnational.org

Scott Leezer, BA
Role: Patient Partner

Vice President of Government Relations, CURA Strategies
Email: scott.leezer@curastrategies.com

Project Title: Comparative Effectiveness Research for Neuroendocrine Tumors (CER-NET)

Michael O'Rorke, PhD
Role: PI

Assistant Professor, Department of Epidemiology, University of Iowa
Email: michael-ororke@uiowa.edu

Josh Mailman, MBA
Role: Stakeholder Partner

President of NorCal CarciNET Community
Email: josh@nextobject.com

Project Title: Development of methods to improve identification of patients with rare or complex diseases

Daniel Herman, MD, PhD
Role: Principal Investigator (PI)

Assistant Professor of Pathology and Laboratory Medicine, University of Pennsylvania
Email: daniel.herman2@uphs.upenn.edu

Jinbo Chen, PhD
Role: Dual PI

Professor of Biostatistics in Biostatistics and Epidemiology, University of Pennsylvania
Email: jinboche@upenn.edu

Engagement Awards and Rare Disease Organizations: An Update and Project Highlights (2:25-3:10 PM)

Karen Martin, MIA
Program Director, Engagement Awards, PCORI
Email: kmartin@pcori.org

Jennifer Canvasser, MSW
Founder and Director of Necrotizing Enterocolitis (NEC) Society
Email: jennifer@necsociety.org

Identifying our National Priorities: Relevance for Rare Disease Populations (3:10-3:40 PM)

Laura Lyman Rodriguez, PhD
Interim Chief Program Support Officer, Senior Advisor to the Executive Director, PCORI
Email: llrodriguez@pcori.org

An Update on the Cost-Data Provision (3:40-3:55 PM)

Andrew Hu, MPP
Director, Public Policy and Government Relations, PCORI
Email: ahu@pcori.org

COVID Connect: PCORI's Response to the COVID-19 Pandemic (4:10-4:40 PM)

Claudia Grossmann, PhD
Senior Program Officer, Research Infrastructure, PCORI
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PCORI Rare Disease Advisory Panel Staff Team

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