Professional Advisory Board (PAB)

PROFESSIONAL ADVISORY BOARD 2017-2019	
Karen Gripp, MD - Co-Chair	Jennifer Katzenstein, PhD, ABPP
Katherine "Kate" Rauen, MD, PhD - Co-Chair	Bronwyn Kerr, MD
David Stevenson, MD - Co-Chair	Suma Shankar, MD, PhD, FACMG
Marni E. Axelrad, PhD, ABPP	Dawn Siegel, MD
Emma Burkitt-Wright, MBChB	Mihir Thacker, MD
Kathryn Chatfield, MD, PhD	Heather Thompson, PhD CCC-SLP
Dan Doyle, MD	K. Nicole Weaver, MD

CO-CHAIRS

Karen Gripp, MD, FAAP; FACMG
Professor of Pediatrics, Sidney Kimmel Medical School at T. Jefferson University
Chief, Division of Medical Genetics
A.I. duPont Hospital for Children

Dr. Karen W. Gripp is the Chief of the Division of Medical Genetics and the Medical Director of the Molecular Diagnostic Laboratory at the A.I. du Pont Hospital for Children/Nemours in Wilmington, DE. She is a Professor of Pediatrics at the S. Kimmel Medical College at T. Jefferson University. She is board certified in pediatrics and clinical and clinical molecular diagnostic genetics. Her areas of particular clinical expertise include craniofacial malformations and rasopathies. Costello syndrome is the focus of her research and she is closely involved with the Costello Syndrome Family Network as the co-chair of the Professional Advisory Board. In addition to co-authoring the Handbook of Physical Measurements, Dr. Gripp has >100 peer reviewed publications. Her professional activities include membership in the ClinGen panel on rasopathies, an active role in ACMG committees, and organizing the "D.W. Smith Meeting on Malformation and Morphogenesis" and the "Diagnostic Dilemmas" session at the annual ASHG meeting. She is on the scientific advisory board for FDNA, the parent company for Face2Gene.

Katherine Rauen, MD, PhD
Professor, Department of Pediatrics
Chief, Division of Genomic Medicine
Albert Holmes Rowe Endowed Chair in Human Genetics II
Director, NF/Ras Pathway Clinic
UC Davis MIND Institute

Dr. Katherine (Kate) Rauen is a Professor in the Department of Pediatrics, Division of Genomic Medicine at the UC Davis where she currently serves as the Chief of Genomic Medicine and holds the Albert Holmes Rowe Endowed Chair in Human Genetics. She received a MS in Human Physiology and a PhD in Genetics from UC Davis doing research on gene dosage compensation and genetic evolution. She obtained her MD at UC Irvine where she also did research in cancer genetics. Dr. Rauen did her residency training in Pediatrics and fellowship in Medical Genetics at UC San Francisco.

Dr. Rauen is internationally known for her pioneering work in the early application of microarray technology in clinical genetics and as a leader and major contributor to the understanding of the "RASopathies", the Ras/MAPK pathway genetics syndromes. Her research program involves the clinical and basic science study of cancer syndromes with effort to identify underlying genetic abnormalities affecting common developmental and cancer pathways. Dr. Rauen led the research team, including the CFC International Family Support Group that discovered the genetic cause of cardio-facio-cutaneous syndrome.

Dr. Rauen is committed to academic medicine, medical education, and advancing best practices for patients with RASopathies. She has successfully obtained both intramural and extramural funding for her research activities, and currently holds a 5-year NIH grant studying skeletal myogenesis in Costello syndrome and CFC. She is the innovator of the world-renowned NF/Ras Pathway Clinic which she initiated in 2007 and this clinic has now been emulated around the globe. She serves on the medical advisory board of CFC International, is a Co-Director for the Costello Syndrome Family Network Professional Advisory Board, and serves on the advisory broads for RASopathies Network USA and Global Genes. Dr. Rauen was recently awarded the Presidential Early Career Award for Scientists and Engineers (PECASE) on her work for CFC and Costello syndrome. Additionally, in June 2014, Dr. Rauen won an International Advocacy Award, the "Global Genes RARE Champion of Hope in Science".

David Stevenson, MD
Associate Professor, Division of Medical Genetics
Stanford University

Dr. David Stevenson is a pediatrician and medical geneticist at Stanford University. He completed his residency in Pediatrics at the University of New Mexico and a 3-year fellowship in medical genetics at the University of Utah. He was on faculty at the University of Utah for 10 years before joining the faculty in the Division of Medical Genetics as an Associate Professor at Stanford University. His initial research focused on neurofibromatosis type 1 (NF1), and since that time he has expanded his research to RASopathies focusing on the musculoskeletal problems and genotype-phenotype correlations. He has received grant funding from the NIH, Doris Duke Charitable Foundation, Thrasher Research Fund, and Department of Defense to investigate the musculoskeletal system in syndromes of the Ras/MAPK pathway. He currently serves as a co-chair of the Costello Syndrome Family Network Professional Advisory Board, is the co-chair of the Children's Tumor Foundation International NF1 Bone Abnormalities Consortium, and is a member of the National Prader-Willi Syndrome Association (PWSA) USA Scientific Advisory Board. He has published over 100 scientific articles, and 2 GeneReviews focused primarily on Ras/MAPK syndromes. Dr. Stevenson first attended the Costello conference in Portland, Oregon in 2007 and has attended all the conferences since that time.

MEMBERS

Marni E. Axelrad, PhD, ABPP
Board Certified in Clinical Child and Adolescent Psychology
Clinical Program Director, Clinical and Pediatric Psychology
Psychology Service, Texas Children's Hospital
Associate Professor, Department of Pediatrics
Baylor College of Medicine

Dr. Marni Axelrad is a board certified clinical child and adolescent psychologist at Baylor College of Medicine/Texas Children's Hospital in Houston, Texas. She has been involved in studying the cognitive, behavioral, and adaptive skills in Costello syndrome since 2003. She has published numerous articles on Costello syndrome, including a review paper about the developmental course in Costello syndrome. Dr. Axelrad looks forward to learning more about Costello syndrome at the current conference, as well as sharing what she knows.

Emma Burkitt-Wright, MRCP, PhD
Consultant Clinical Geneticist
Manchester Centre for Genomic Medicine
Manchester Academic Health Sciences Centre
University of Manchester & Central Manchester University Hospitals NHS Foundation Trust

Dr. Burkitt-Wright graduated in medicine from the University of Liverpool (2002), and undertook house jobs and a general internal medicine rotation in Liverpool, gaining Membership of the Royal College of Physicians (2005). After a year of paediatric experience, she started training in clinical genetics in Manchester (2006), as the first academic clinical fellow in the UK's integrated clinical academic training scheme. During that post, she joined Professor Mariano Barbacid's group at the Spanish National Cancer Centre (CNIO) on secondment, to characterise their B-Raf mouse model of CFC syndrome. Awarded a Manchester Biomedical Research Centre PhD training fellowship in 2009, she secured a Wellcome Trust Research Training Fellowship (2010-2013). Her PhD thesis, '*De novo* germline disorders of the Ras-MAPK pathway: clinical delineation, molecular diagnosis and pathogenesis' was awarded in summer 2014. She obtained her certificate of completion of specialist training in November 2014, having trained with Professor Bronwyn Kerr (seeing patients with Costello, CFC and Noonan syndromes). As a consultant, she offers clinics for patients with these disorders within the Manchester Centre for Genomic Medicine and the nationally commissioned highly specialised service for complex NF1.

Kathryn Chatfield, MD, PhD
Assistant Professor of Pediatrics, Cardiology
University of Colorado School of Medicine
Director, Cardiac Genetics Clinic
Division of Cardiology

Dr. Chatfield is an Assistant Professor of Pediatrics, Cardiology, University of Colorado School of Medicine and the Director of the Cardiac Genetics Clinic. Her degree is from Dartmouth Medical School and she completed her residency at Children's Hospital of Philadelphia. Dr. Chatfield's clinical interests include Noonan-spectrum disorders, Marfan syndrome and related connective tissue disorders, familial congenital heart disease, metabolic and inherited forms of cardiomyopathy. Her research focus is the role of mitochondrial energy metabolism in pediatric cardiomyopathy and heart failure. She is also currently doing research related to the role of cardiac transplantation in children with Noonan-spectrum disorders and hypertrophic cardiomyopathy.

Dan Doyle, MD
Associate Professor of Pediatrics, Thomas Jefferson University
AI duPont Hospital for Children
Division of Endocrinology

Dr. Doyle is an Associate Professor of Pediatrics at Thomas Jefferson University and AI dupont Children's Hospital. His interest is in the natural history of growth and the use of growth hormone in Costello syndrome. His previous measurements of the children with CS have resulted in the recent publication of "Normative Growth Curves in Children with Costello Syndrome" in the American Journal of Medical Genetics. He will discuss the use and limitations of these curves as well as the natural history of growth and the use of growth hormone in CS children. Dr. Doyle will share his section of the Costello syndrome consensus guidelines upcoming publication.

Jennifer Katzenstein, PhD, ABPP
Board Certified Subspecialist in Pediatric Neuropsychology
Pediatric Neuropsychologist
Johns Hopkins AII Children's Hospital

Dr. Jennifer Katzenstein is a board certified clinical neuropsychologist at Johns Hopkins All Children's Hospital in St. Petersburg, Florida. She has been involved in studying the cognitive, behavioral, and adaptive skills in Costello syndrome since 2009. She has worked with Marni Axelrad, PhD since 2009 to continue to develop our knowledge base.

Bronwyn Kerr, MD
Consultant Clinical Geneticist/Honorary Senior Lecturer
Associate Medical Director
Clinical Genetics Service
Manchester Centre for Genomic Medicine

Professor Bronwyn Kerr is a consultant clinical geneticist who has worked in Manchester in the UK since 1995. Her principal research interest has been Costello syndrome, and more recently, other disorders of the RAS/MAPK pathway. She is a member of the Medical Advisory Boards of the International Costello Syndrome Support Group (UK), the Costello Syndrome Family Network (USA) and the Noonan syndrome association (UK). She has a number of key publications in this area, and is frequently consulted by national and international colleagues for advice on diagnosis and management in this group of disorders. A particular interest is developing an evidence base for management of rare disorders. She is a member of the steering group of GenIDA (Genetically determined Intellectual Disabilities and Autism Spectrum Disorders), an on-line registry for patients, families and professionals. She is the lead for the Guideline Development work-stream of the European Reference Network project ITHACA (HP-ERN-2016; 739516), a five-year project commencing April 2017.

Suma Shankar, MD, PhD, FRCS, FACMG
Associate Professor of Clinical Pediatrics
Director of Precision Genomics
Genomic Medicine Division, MIND Institute
University of California, Davis

Dr. Suma Shankar is an Associate Professor in the Department of Pediatrics, Division of Genomic Medicine at UC Davis Medical Center. She is the Director of Precision Genomics and will lead the effort in bringing genomics to every day clinical practice. She is working on integrating genomic medicine with multiple specialties in pediatrics and adults for rare Mendelian diseases and complex disorders. Dr. Shankar is a clinician scientist who received her medical degree from Bangalore Medical College, Bangalore and PhD in Molecular Genetics from University of Iowa, USA. She is an ophthalmologist trained in United Kingdom and is a Fellow of the Royal College of Surgeons (FRCS), Edinburgh and Member of the Royal College of Ophthalmologists (MRCOphth), London. She also completed a fellowship in Medical Genetics at the University of California, San Francisco and is board certified in Medical Genetics by the American Board of Medical Genetics (FACMG). She was faculty in the Departments of Human Genetics and Ophthalmology at Emory University, School of Medicine, Atlanta, USA where she initiated Ocular Precision Health Initiative with biobank and genetic studies for inherited eye diseases for the first time in Georgia. She served as Medical Director for Emory Genetics Laboratory and was primary investigator on many clinical trials investigating novel therapies for rare genetic disorders. She has won the Western scholar and Henry Christian Award and Certificate of Excellence in Research from the American Federation Medical Research for research on ocular manifestations in RASopathies. She has a number of publications and several book chapters on genetic testing, ocular genomics, novel enzyme replacement, substrate, and chaperone therapies. Her chief research interests are gene discovery projects and translational research for genetic diseases. A link to her publications can be found at this url.
http://scholar.google.com/citations?hl=en&user=u6FQarEAAAAJ&view_op=list_works&sortby=pubdate

Dawn Siegel, MD
Dermatologist
Medical College of Wisconsin

Dr. Dawn Siegel is a pediatric dermatologist from Medical College of Wisconsin and Children's Hospital of Wisconsin in Milwaukee. Dr. Siegel received her medical degree from the University of Wisconsin, Madison and then went on to do a pediatric residency at Children's Hospital Oakland California. Dr. Siegel did her dermatology residency and pediatric dermatology fellowship at the University of California, San Francisco. Dr. Siegel has a particular interest and expertise in genetic dermatologic syndromes, including Costello syndrome, NF and the RASopathies. She is co-director of the Neurofibromatosis and RASopathy clinic at Children's Hospital of Wisconsin. In addition, she has spent the last several years working on gene discovery for a hemangioma syndrome called "PHACE syndrome". Dr. Siegel has been actively involved in studying the skin issues in Costello syndrome and cardio-facio- cutaneous syndrome since 2006. The paper on CFC was published in 2010 and the Costello paper was published in 2011 in the British Journal of Dermatology. Today Dr. Siegel will teach you about the updates in the skin section of the Costello syndrome consensus paper and share some general skin care guidelines with you.

Mihir Thacker, MD
Pediatric Orthopedic Surgeon, Orthopedic Oncologist
Alfred I. duPont Hospital for Children
Department of Orthopaedic Surgery

Dr. Mihir M. Thacker **is** an orthopaedic surgeon at the Nemours/Alfred I. duPont Hospital for Children. He went to the Seth G. S. Medical College in Mumbai, India and completed his orthopaedic residency at the Lokmanya Tilak Municipal General Hospital in Mumbai. He has had extensive subspecialty training through pediatric orthopaedic fellowships at the National University Hospital in Singapore, Hospital for Joint Diseases/New York University in New York, and the duPont Hospital for Children. He also completed a fellowship in musculoskeletal oncology at the Jackson Memorial Hospital/University of Miami.

Dr. Thacker's major clinical interest is bone and soft tissue tumors. He performs limb salvage surgery for tumors, including using noninvasive expandable (growing) prostheses. He is also interested in hip disorders in children and performs complex hip reconstruction (including surgical hip dislocations and peri-acetabular osteotomies), treatment of complex congenital anomalies, correction of complex deformities and limb lengthening. He directs the clubfoot clinic and practices the Ponseti technique for clubfoot treatment.
Dr. Thacker is active in teaching medical students, residents, fellows and international visiting observers. He is invited to lecture nationally and internationally, and is a member of several societies including the Pediatric Orthopaedic Society of North America, Children's Oncology Group, Musculoskeletal Tumor Society and Lower Limb Reconstruction Society. He is actively involved in research and has more than 30 peer-reviewed publications, five book chapters and several national and international presentations at scientific conferences. Dr. Thacker has been selected by *Delaware Today 2014* as one of the best surgeons in Delaware among both pediatric surgeons and surgical oncologists. He received the Patient Choice Award and Compassionate Doctor Award and was listed in *Best Doctors in America (2009-11)*.

Heather Thompson, PhD CCC-SLP
Assistant Professor, SLPA Program Coordinator
California State University, Sacramento
Department of Communication Sciences and Disorders

Heather L. Thompson, Ph.D is an assistant professor and coordinator for the speech-language pathology assistant program in the Department of Communication Sciences and Disorders at California State University, Sacramento. She trained at Western University in London, Canada and completed her PhD at the University of Utah. She has taught numerous classes in speech-language pathology at the undergraduate and graduate levels. Her line of research is focused in the areas of developmental speech and language disorders. She has a specific interest in examining speech and language outcomes in children with cleft palate and developmental syndromes of Ras/MAPK pathway dysregulation. She has published in the area of neurofibromatosis and has coauthored publications in the areas of specific language impairment/ADHD and pediatric feeding. She has received many University grants as well as funding from the Canadian Institutes of Health Research (CIHR). She is on the advisory board for District 2 of the California Speech Hearing Association and is part of the Response Evaluation in Neurofibromatosis and Schwannomatosis (REiNS) International collaboration.

K. Nicole Weaver, MD
Assistant Professor of Pediatrics, University of Cincinnati College of Medicine
Division of Human Genetics
Cincinnati Children's Hospital Medical Center

Dr. K. Nicole Weaver is a clinical geneticist at Cincinnati Children's Hospital Medical Center in Cincinnati, Ohio. She is the geneticist for the Cardiovascular Genetics Clinic at CCHMC, which provides diagnostic evaluations and ongoing management for children and adults with connective tissue disorders such as Marfan syndrome, as well as Noonan and related syndromes. Dr. Weaver met Dr. Gripp and became interested in Costello syndrome research through taking care of a very special little girl with Costello syndrome caused by the rare G12E mutation. Dr. Weaver attended the 2015 Costello Syndrome Family Conference and conducted research on the medical concerns affecting adults with Costello syndrome.