Final 11/1/2022

Biographies of Speakers

Thrive's 60th Celebration of PK Deficiency as a Genetic Anemia Conference:

Agnot, Celeystn:

Celestyn is a second-year genetic counseling student studying for her MSPH at the University of South Florida's Genetic Counseling Program. She graduated from the University of Florida in 2018 with a Bachelor of Science in Biology. She previously worked as a Study Coordinator at Children's Diagnostic and Treatment Center where she worked in research on the Pediatric HIV/AIDs and Cohort Study. There she worked with children and young adults perinatally infected and exposed to HIV.

Al-Samkari, Hanny:

Assistant Professor of Medicine at Harvard Medical School and clinical investigator in the Massachusetts General Hospital Division of Hematology Oncology.

Dr. Al-Samkari earned his medical doctorate at the Washington University School of Medicine and completed an internal medicine residency at the Hospital of the University of Pennsylvania, where he was selected as a Chief Medical Resident. He then completed his fellowship in hematology and medical oncology at the Dana-Farber/Mass General Partners program. He has published over 100 peer-reviewed manuscripts, including first-author original research manuscripts in the top journals in medicine and hematology. His clinical and research interests are in bleeding disorders and hemolytic anemias. With regards to the latter, he is a leading investigator in the use of pyruvate kinase activators to treat hereditary and acquired anemias and serves as principal investigator for many ongoing studies in this space. He also Co-Chairs the First International Guidelines for Diagnosis and Management of Pyruvate Kinase Deficiency, leading a global initiative of experts to develop the first evidence-based clinical guideline for Pyruvate Kinase deficiency, a genetic, hemolytic anemia.

Bassett, Bianca:

Bianca was born in 1980 in a country town in Victoria, Australia. At six weeks of age, she was diagnosed with PKD, and was transfusion dependent until the age of 36. Bianca began her iron chelation at the age of 8.

Growing up, Bianca played lots of sports including athletics, tennis, and racquetball. She completed her education until the age of 16 and left to follow a career in hairdressing. Bianca soon realised she preferred a career in the corporate sector and in 2008 she took a position in the Arts and Events sector in management, HR, marketing, and Project work for a not-for-profit Aboriginal Arts Organisation. She continued working in corporate roles as an Executive Officer, in Senior

HR roles, major projects, social marketing and events for 20 years. Bianca specialised in working for local government, community engagement and with First Nations communities.

In 2016 Bianca lost her leg following an illness and quickly realised there was so much for people to learn in the disability inclusion space. She saw an opportunity to use her lived experience in a positive way to help others understand the value of inclusion for people with disability and now works as a Disability Inclusion Consultant, to make a more accessible and inclusive world for people with disability.

In 2021 Bianca self-published her story 'Walking My Path', candidly sharing her life journey and how she overcame her many challenges of loss, grief, and trauma to become stronger, resilient, and joyful about her future.

Bianca is a passionate and active community volunteer and has been a volunteer for the Country Fire Authority for over 10 years. She strives to show her community and others that her disability and what she learns everyday through her lived experience, continues to be the enabler not the barrier for her striving to achieve her dreams and goals.

Blackman, Jason:

Jason Blackman is from New Albany, IN. He is forty-nine years old but claims to feel like he's forty-eight. He has a wonderful son, Mason, and wife that is way too good for him named Sasha. He was diagnosed with Congenital Dyserythropoietic Anemia (CDA) at birth. Three years ago, he was genetically tested and confirmed to have PK deficiency. He has been transfusion dependent his entire life and has been on mitipivat for three years. Jason was an athlete in his youth specializing in baseball and was a poker dealer but was forced to retire early because of chronic anemia. He is a wizard in Lego-building and hopes to make the future better for upcoming kids with PKD.

Cannon, MSc, Lily:

Thalassaemia's International Federation's Deputy Director, Lily Cannon holds a BSc in Psychology and MA in Human Resource and Knowledge Management from Lancaster University, and an MSc in Forensic Psychology from the University of Central Lancashire. Whilst in the UK, she worked on a voluntary basis at St Helen's Women's Aid, Therapeutic Services with victims and perpetrators of domestic violence. In addition, she was involved in the setting up of the National Committee of the European Youth Parliament in Cyprus, participating both as an organizer and a chair in national sessions.

Since December 2010, Lily has been a member of the Staff of TIF. She previously held the post of Policy, Public Health Affairs & European Organisations Senior Officer until 2015, when she assumed the post of TIF Operations Manager until August 2022. Her role deals primarily with the implementation of action plans and the attainment of strategic objectives of the Federation, the initiation and further collaboration with external partners, and the day-to-day monitoring of deliverables.

Eijenraam, Maaike:

Maaike is a person with PK deficiency. She lives with her partner Robert (also a PKD patient) and her youngest son in a small village in the north of the Netherlands. Her eldest son lives near them. She is a trained social worker, however about ten years ago she was unable to continue working.

Over the last few years, she has become active as a patient advocate working on several projects with Agios Pharma. She has also been speaking at conferences and played an advisory role in the organization of Thrive's 60Th Year Celebration of PKD conference.

She is very much interested in focusing on the psychological aspects of illness and medical trauma.

Garrett, Libby:

Libby is forty years old and lives in Georgia. She was diagnosed with Hemolytic Anemia at six months of age, and in 2009, through genetic testing, discovered she had PK deficiency. She is transfusion-dependent, and after starting a monthly regimen about a year ago, her quality of life has improved. However, after having her miracle baby, Bryson, seven years ago, she became much sicker with PK deficiency.

She had the honor of telling her PKD story in 2019 to the FDA at a Patient-Focused Drug Development Meeting where she had the enjoyable experience of meeting other people with PK deficiency for the first time.

Grace, MD, Rachel:

Dr. Rachael Grace, MD, MMSc, is a pediatric hematologist and clinical researcher at the Dana-Farber/Boston Children's Cancer and Blood Disorders Center. She is the medical director of both the Hematology Ambulatory Program and the Hematology Clinical Research Program. Dr. Grace enjoys clinical care of pediatric patients, and her research interests are focused on improving outcomes in the care and treatment of individuals with immune cytopenias.

Dr. Grace is the lead investigator for the Pyruvate Kinase Deficiency Natural History Study, a global registry for patients with PK deficiency. She is a site investigator of the DRIVE-PK and ACTIVATE mitapivat (AG-348) clinical trials and the PEAK registry.

Dr. Grace is an Associate Professor of Pediatrics at Harvard Medical School. She received her MD at Brown University and received her master's degree in clinical research at Harvard Medical School. She completed her residency at Boston Children's Hospital and completed her pediatric hematology/oncology fellowship at Dana-Farber/Boston Children's Cancer and Blood Disorders Center.

Herzog, Becky:

From Montana, Becky is mom to a sweet, caring, and strong six-year-old girl named Remy, who was diagnosed with PK deficiency at 3 months of age. Becky advocated for her daughter at FDA's Patient-Focused Drug Development Meeting in 2019. She is a Co-founder and Patient and Caregiver Outreach Director for Thrive with PK Deficiency. Her main goal is to help new caregivers understand and communicate with their little ones and to educate people on PKD.

Kellar, Jessica:

Jessica Kellar is a mother of three children from New Hampshire. Her oldest child Travis has PKD; Savannah and Addison are both carriers of PKD. She was a member of the original PKD group on Yahoo and is a founding member of the PKD Facebook group. She worked with the Facebook group members several years ago to create the original PK deficiency ribbon. She believes the knowledge we have together about our own successes and failures helps us all to better determine our PKD journeys and to develop care plans that help us reach our management goals.

Kellar, Travis:

Travis Kellar is a 15-year-old high school freshman. He was diagnosed with PKD when he was 6 months old, however, he did have symptoms starting within a few hours of birth. He enjoys playing with his PS5, watching The Green Arrow, and lawn care. He would like to share his story and continue to build relationships with others who are affected by PKD to help create a consistent standard of care.

King, Lillian:

Young Lillian is a patient advocate for PK deficiency who champions better treatment options and brings awareness to the physical and mental impact of living with PKD. Lillian is an online student currently in the 6th grade and is an artist.

King, Jennifer:

Jennifer is a full-time homemaker and caregiver for three children, two with PK deficiency. She advocates for treatment options that provide better long-term outcomes for patients with PKD. Prior to her family commitments, she was a Veterinarian Technician for an Equine Center.

Lander, RN, Carl:

Carl is a Registered Nurse in the UK and has worked within both critical care and interventional cardiology specialties. Carl moved into education and then onto organizational development and people advocacy roles; these roles crossed both healthcare management and, with the Royal College of Nursing, representing and advising nurses on both employment and professional issues. He currently works in roles that support NHS Trusts to improve or change systems and processes to resolve issues and effectiveness, along with patient advocacy in rare diseases. Carl manages PK deficiency with help from his wife. He is the Co-Founder and International Collaboration Director for Thrive with PK Deficiency Organization and serves on Agios' Advocacy Advisory Council, is the Patient Representative for the PEAK Registry and PKD Guidelines Committee.

Marquis, Madalyn:

Madalyn Haley Marquez and her older sister both have PK deficiency. They are similar in how it affects them. Madalyn has been explaining her illness to doctors since she was nine. She says she always freaked doctors out that don't know about PKD when they see her blood work. She's had to live a little differently than her peers most of her life. Even into adulthood and with the pregnancy of her daughter Mila Daisy, she never let it stop her. She didn't understand much about it when she was younger and believes she still has a lot to learn. Her goal is to raise awareness by posting more videos about PK deficiency. Madalyn is a singer and social media influencer.

McGuire, Carson:

Carson McGuire is a talented freelance artist, illustrator, designer, and vintage goods dealer living in Indiana. He was a person living with PK deficiency prior to being one of the first two people to undergo gene therapy to cure PKD in 2020. Carson's career as an artist was cut short due to complications related to PK deficiency. He is now living with a "normal" hemoglobin and enjoys being able to be more active.

Nathan, MD, David:

David G. Nathan, M.D. was born in Boston in 1929. He is a graduate of Harvard College (1951) and Harvard Medical School (1955). He was an intern and senior resident in medicine at the then Peter Bent Brigham Hospital and a Clinical Associate at the National Cancer Institute from 1955 to 1959. From 1959 to 1966 he was a hematologist at the Peter Bent Brigham Hospital and then became Chief of the Division of Hematology at Children's Hospital. In 1974 he established the combined Division of Pediatric Hematology at Children's Hospital and the Dana-Farber Cancer Institute. From 1985 to 1995 he was Physician-in-Chief of the Children's Hospital and from 1995 to 2000 was President of Dana Farber Cancer Institute.

Dr. Nathan's research has focused on the inherited disorders of red cells and granulocytes and particularly on thalassemia and sickle cell anemia. His contributions include the introduction of effective treatment of iron overload and of hydroxyurea, the only FDA approved drug for prevention of the symptoms of sickle cell anemia. He has been instrumental in the development of the prenatal diagnosis of the hemoglobinopathies. He has trained over 100 hematologists, many of whom hold leading positions in pediatrics and internal medicine. His textbook, begun with his colleague, Frank Oski, entitled Hematology of Infancy and Childhood is the leading text in the field. He is the author of

two popular books: Genes Blood and Courage published by the Harvard University Press in 1995, and The Cancer Treatment Revolution published by John Wiley and Sons in March 2007.

Dr. Nathan is a former trustee of the Boston Children's Hospital and of Rockefeller University and a member of the American Society for Clinical Investigation, the Association of American Physicians, the American Pediatric Society, the Institute of Medicine, the American Academy of Arts and Sciences and the American Philosophical Society. He is the recipient of numerous awards including the National Medal of Science, the Stratton medal of the American Society of Hematology (of which he was President), The Walker Prize of the Boston Museum of Science, The John Howland medal of the American Pediatric Society, The George M. Kober Medal of the Association of American Physicians, The John Stearns Medal for Lifetime Achievement in Medicine of the New York Academy of Medicine, Harvard University Honorary Doctor of Science, The Wallace Coulter Award of the American Society of Hematology, Clarkson University Honorary Doctor of Science, The Dr. von Hauner Medal, Dr. von Hauner Children's Hospital (Munich, Germany), Sultan Bin Khalifa Grand International Award (Abu Dhabi) and the Boston Children's Hospital's Inaugural Lifetime Impact Award.

Dr. Nathan remains on the faculty at Boston Children's and Dana-Farber where he teaches fellows in the training program he established and mentors young faculty members.

O'Conner, Leah:

Leah O'Connor, 21 years old from Cork, Ireland. She was diagnosed with PK deficiency in 2004 at 3 years of age. Leah currently practices as a Beauty Therapist/Cosmetologist.

Schryver, PhD, RD, Tamara:

Tamara Schryver, PhD, RD, practiced as Nutrition Scientist and Registered Dietitian before leaving the workforce because of health complications related to PK deficiency. She and her husband Rob have two children, three pets, and seven garden beds they nurture in the great State of MN. Tamara is a Co-Founder and President of Thrive with PK Deficiency Organization, a nonprofit dedicated to helping people with PK deficiency live a better quality of life. She also serves as a patient advocate on Agios' Advocacy Advisory Council and on Global Genes Global Advocacy Alliance Leadership Council. She is a patient representative for the PK deficiency guidelines committee and her goal is to improve the quality of life for people living with PKD.

Shah, MD, Ami:

Dr. Shah is a Clinical Professor of Pediatrics at Stanford Children's Hospital. She received her medical degree at University of North Carolina at Chapel Hill and completed her residency and fellowship in pediatric hematology oncology at Children's Hospital Los Angeles. She has been a pediatric bone marrow transplantation physician for over 25 years. Her research has been primarily involved with long term follow up and survivorship following transplantation and transplantation for non-malignant diseases. She has been involved with gene therapy for a variety of rare diseases. She is currently the site PI for the Lentiviral gene therapy trial for PKD, sponsored by Rocket Pharmaceuticals.

Shields, Andrea:

Andrea is a second-year genetic counseling student at the University of South Florida. She earned her undergraduate degree in psychology with a concentration in neuroscience from the University of Florida in 2016, and then her masters in neuroscience from the University at Buffalo in 2018. She has always loved genetics and biology, and originally planned to go into academia and research. However, she found herself wanting to work more closely with patients. She decided to change her career to genetic counseling and is so happy she did. Andrea loves this field and is extremely excited to see what the future holds.

Solis, Jacob:

Jacob was diagnosed with PKD at 9 months old when his family lived in New Jersey. His parents originally from Costa Rica, moved back when he was 4 years old giving Jacob the opportunity to experience care of a rare disease in two different countries. Jacob attended Keiser University and UCF where he obtained a Bachelor's in Biomedical Sciences. He currently works at a dermatology billing company and a restaurant and is excited to meet other people with PKD.

Tompkins, Lindsey:

Lindsey is a 23-year-old Registered Dental Assistant from Iowa living with PK deficiency. She likes to advocate for PK deficiency by talking about her story in the hopes of helping other people with rare diseases close in age.

Welle, Jill:

Jill currently works full-time managing sales for a software company while juggling her role as a PK deficiency patient advocate and grandmother to eight grandchildren. Jill was diagnosed with PKD when she was ten years old, and it is her goal to help children diagnosed with PK deficiency today live with better treatment options.