FGFR Syndromes Collaborative Research Network Conference

March 17th & 18th, 2022

Speaker Bios

Cassandra Aspinall, MSW, LICSW, ACSW

Social Worker, Craniofacial Center, Seattle Children's

Cassy Aspinall has worked at Seattle Children's Hospital for over 3 decades, providing psychosocial support to children and families throughout the childhood of each patient. She has been active both nationally and internationally in the field of Craniofacial Care. The focus of her work is on making sure that all aspects of care are supported including a child's psychological health and the wellbeing of the family. This means making sure to understand a family's story and their specific communication styles and needs. Assessing for financial, work, educational and other community resource needs is a critical part of supporting success and the best medical / surgical outcomes. As a social worker, she is an active member of the Craniofacial team, doing work to help make sure to understand the child and family, so they have their best understanding of the medical information. This work extends into the community to connect with a child's school, community medical providers and any relevant family resource elements such as the workplaces of the family members, insurance companies, therapists, and support resources. Individual intervention plans are updated over time, to assure that any needs and their resources are updated.

Scott P. Bartlett, MD

Attending surgeon in the Division of Plastic, Reconstructive and Oral Surgery and Director of the Craniofacial Program at Children's Hospital of Philadelphia

Dr. Bartlett is well known for his work in facial plastic surgery in children and adults, treating them for both reconstructive and cosmetic concerns. In his reconstructive work, Dr. Bartlett treats patients with facial trauma, facial palsy, congenital abnormalities and defects of the face following skin cancer surgery. In the area of cosmetic surgery, he has special interests in the face and neck, concentrating on nasal, eyelid and facial rejuvenation procedures. By combining his expertise in infant and adult reconstruction with aesthetic procedures, he is able to marry form and function in restoration.

Dr. Bartlett is a member of the Edwin and Fannie Gray Hall Center for Human Appearance, a team of specialists dedicated to improving ways of treating appearance related problems. He is also the holder of the Peter Randall Endowed Chair in Pediatric Plastic Surgery and of the Friends of Brian Endowed Chair in Pediatric & Reconstructive Surgery at the Children's Hospital of Philadelphia. He is widely published in specialty journals and books.

Dr. Bartlett lectures worldwide and is a member of prestigious plastic surgery societies such as the American Association of Plastic Surgeons, the American Society of Plastic Surgeons and the International Society of Craniomaxillofacial Surgery, of which he is a former President. He is a fellow

of the American College of Surgeons and the American Academy of Pediatrics. He is the Section Editor in Pediatric / Craniofacial for Plastic and Reconstructive Surgery, the most widely read journal in the specialty. Each year he travels to Poland in a volunteer capacity where he performs surgery and trains Polish physicians in methods of facial reconstruction for children and adults.

Dr. Bartlett is the Mary Downs Endowed Chair in Craniofacial Surgery at Children's Hospital of Philadelphia and is a Professor of Plastic Surgery at the University of Pennsylvania Medical Center. He completed his pre-medical studies at Weber State College in Utah and his medical studies at Washington University School of Medicine in St. Louis, Missouri. He interned and did his surgical and plastic surgery residencies at Massachusetts General Hospital in Boston. At Harvard University, he was a research fellow in surgical immunology and later a craniomaxillofacial and pediatric fellow at the University of Pennsylvania Medical Center and Children's Hospital of Philadelphia. He is board certified in Plastic Surgery.

Lauren Beckett, MS, CCC-SLP

Pediatric Speech Language Pathologist, Child Development and Rehabilitation Center/Institute on Development and Disability, Oregon Health & Science University

Lauren works as a team member of the multi-disciplinary Craniofacial and Cleft Palate Program and the Pediatric Feeding and Swallowing Disorders Program at OHSU.

Lauren's primary clinical interest is in working with children with cleft palate and/or velopharyngeal dysfunction because she is fascinated by the role anatomy plays in speech production abilities. Lauren is active in her field's professional organizations and her research interests include speech outcomes after palate repair, patient satisfaction after velopharyngeal insufficiency surgery, and therapies for young children with cleft palate.

Jannett Barton

Parent Advocate

Avery Cooper

Pfeiffer Syndrome Advocate

Avery is 13 years old. She loves doing theatre camp in the summer. Last year she was Kaa the Snake in The Jungle Book. Avery loves to sing and dance to music. It makes her happy. She has a cat that she loves so much, named Oliver.

Delaney Cunha

Pfeiffer Syndrome Advocate

Delaney Cunha, age 10, is a current fourth grader at Deer Park Elementary School in Tampa, FL, where she resides with her dad, Steve and golden retriever, Rosie. She is also a St. Petersburg, FL, resident with her Mom, Kelly, stepdad, Brett, siblings Kylie, Ethan and Zan, Jack Russell puppy, Jake, and cat, Lucy. Delaney loves gymnastics, creating videos, playing with her Rainbow High and Barbie dolls, practicing make-up looks with her favorite influencers, gaming on Roblox, hair colors and fairy hair, and watching Gilmore Girls on Netflix. Delaney was diagnosed with Pfeiffer Syndrome at 2 years old, wears hearing aids and will have her midface at Boston Children's this year which she will overcome with her trusted security blanket friend, Lucy. A proud member of the craniofacial community, Delaney and her family plan to author children's books among other entrepreneurial

ideas to show more kids with hearing aids, share her journey and help kids. She was also the cover feature of Tampa Bay Parenting magazine and recently guest starred in a TV show with a favorite local non-profit role model airing this Spring.

Carl Dambkowski, MD

Dr. Dambkowski received his medical degree at Stanford University School of Medicine. His work focuses on the evaluation of genetic anomalies and treatment regimens for various genetically-driven conditions, including skeletal dysplasias.

John Dahl, MD, PhD, MBA

Otolaryngologist, Seattle Children's; Assistant Professor, Department of Otolaryngology – Head and Neck Surgery, University of Washington

Prior to joining the faculty at the University of Washington, Dr. Dahl spent three years on the faculty of the Indiana University School of Medicine and served as the Surgical Director of the Aerodigestive Program at Riley Hospital for Children. Dr. Dahl has a significant interest in basic and clinical scientific research related to vascular anomalies, tracheal malformations, airway management in patients with complex craniofacial disorders, and patient quality/safety.

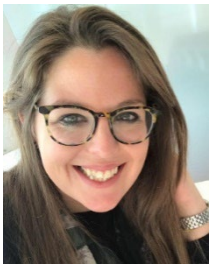
Dr. Dahl obtained a PhD in Pharmacology and an MBA from The Pennsylvania State University and earned his medical degree from Sidney Kimmel Medical College of Thomas Jefferson University. He completed residency training in Otolaryngology- Head and Neck Surgery at the University of North Carolina, Chapel Hill, and a fellowship in Pediatric Otolaryngology - Head and Neck Surgery at Seattle Children's Hospital.

Sarah Hofman DeYoung, MD

Dr. Hofman DeYoung received her medical degree at the University of Washington School of Medicine, Seattle campus, followed by pediatrics residency at Seattle Children's/University of Washington. She is now a second year pediatric pulmonary fellow at Seattle Children's Hospital, with professional interests in airway anomalies, acute and chronic complex ventilation including respiratory management of critically ill children, bronchoscopy, and healthcare disparities in children with chronic respiratory conditions.

Claire Ellis

Claire Ellis is the Newborn Screening & Diagnostics Policy Fellow with the EveryLife Foundation for Rare Diseases. Claire advocates for newborn screening legislation at the state and federal level, creates advocacy resources and tools to facilitate patient and organization engagement within the newborn screening ecosystem, and assists in the management of the Foundation's Community Congress Newborn Screening working group. Previously, Claire worked with the Indiana House of Representatives as a Legislative & Constituent Services Intern and with Zimmer Biomet as a Government Affairs Intern. Claire graduated from the O'Neill School of Public and Environmental Affairs at Indiana University with a degree in Law & Public Policy and a concentration in Healthcare Policy.



Jessica L. Giordano, MS, CGC

Assistant Professor of Genetic Counseling (OBGYN), Columbia University Irving Medical Center

Jessica is an ABGC board-certified genetic counselor and assistant professor of genetic counseling with 13+ years of clinical, research, and industry related experience in prenatal genetic counseling and testing. In her clinical work, she has witnessed how non-invasive aneuploidy screening, diagnostic testing via microarray, and pan-ethnic screening has transformed prenatal practice, making complex genetics test part of primary obstetrical practice. Recently, she successfully led Columbia's fetal exome sequencing study published in Lancet, the stillbirth exome study published in the NEJM, and is the primary manager for the NICHD multi-center study PrenatalSEQ, aiming to study the use of genomic sequencing in pregnancy to change post-natal management. In these research roles, she consents patients, curates variants, and assesses genotype-phenotype correlations with the goal of ultimately communicating results to patients to impact clinical management. Additionally, Jessica has been integral to the recent development and launch of Columbia's new Genetic Counseling Graduate Program and currently serves as a course director and scholarly projector mentor to their graduate students.

Elvire Gouze, PhD

Elvire Gouze is the Founder and CEO of Innoskel, a pioneering platform biotechnology company developing therapies for rare skeletal diseases. Dr. Gouze is an experienced scientist, serial entrepreneur and expert in skeletal disorders.

She holds a PhD in Molecular Pharmacology and has over 20 years of experience in bone and cartilage diseases. Dr. Gouze has a proven track record of progressing the development of an innovative therapeutic pipeline, as seen with her first venture Therachon, a biotech company focusing on achondroplasia, a rare bone disease, which was acquired at Phase 1 stage for $810M by Pfizer in May 2019. Dr. Gouze holds a tenured track at INSERM and directs a team from which Innoskel is the spin-off. She is focused on the development of innovative biotherapies for skeletal dysplasia.

Anne Hing, MD

Anne V. Hing, MD, is attending physician at Seattle Children's Hospital, professor in the Department of Pediatrics at the University of Washington School of Medicine, and adjunct faculty member in the Division of Medical Genetics. Her clinical interests include the diagnosis and management of infants, children, and adolescents with craniofacial and genetic conditions.

Dr. Hing works in the Craniofacial and Craniofacial Genetics Clinics and also serves as a genetics consultant in outreach clinics throughout the state of Washington. She coordinates the Craniofacial resident elective (and selective) rotation and provides bedside teaching for medical students and residents. She is program director of the Craniofacial Medicine fellowship and coordinates the annual multidisciplinary Craniofacial Center Educational Retreat.

Dr. Hing completed her medical training, pediatric residency and genetics fellowship from Washington University in St. Louis, MO. She is Board Certified in Pediatrics and Medical Genetics.

Ellie Hollingshead

Pfeiffer Syndrome Advocate

Ellie is a creative young woman who can play piano by ear and loves to draw unicorns, her spirit animal. She is the oldest of four children and loves spending time with her siblings. Born with Pfeiffer Syndrome, she has conquered 11 surgeries, and recently shared a video on how to prepare for the midface advancement surgery. She enjoys reading, swimming and playing with friends.

Whitni Hollingshead

Parent Advocate

Whitni Hollingshead is a homemaker and homeschool mom. She has been homeschooling for the past 6 years. Whitni has been happily living in Pocatello, Idaho, for 4 ½ years with her husband, Ryan, and four children, Ellie (11), Ammon (9), Maggie (6), and Hyrum (4). She loves going on long walks with an exciting audiobook.

Falesha Johnson

Parent Advocate, Director of Development at the Foster School of Business at the University of Washington

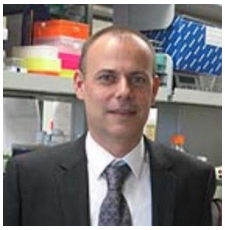
Falesha wears many hats, including being mom and caregiver to her daughter Caliyah who is two years old and has Pfeiffer Syndrome. Cali has had her trach since she was six weeks old, and Falesha and her husband have overcome many obstacles while learning how to care for Caliyah. She is looking forward to sharing what she's learned and aspects of life with a trach from a parent's perspective.

Synthiea Kaldi

Despite claiming that her cats raised her child, Synthiea Kaldi's background in project management and customer service on cruise ships are what most equipped her to be the mother of a child with Pfeiffer Syndrome. Other rare parents appreciate her candor and share her frustration with the health insurance system and DDA. She single-handedly coined the term "MomGyver" and is completely extra when it comes to her family's Halloween costumes.

Synthiea earned her MBA at L'ecole de Management in Lyon France and holds a bachelor's degree in Speech Communication (aka Public Speaking) from the University of Washington. While she does not (yet) hold a medical degree, she often claims that she, "plays a doctor on TV," and believes that the gene mutations involved in Pfeiffer Syndrome also contribute to a great sense of comedic timing and having beautiful, "bee sting" lips.

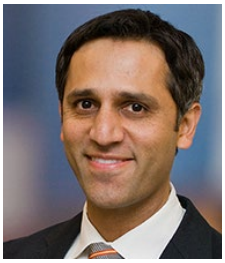
Synthiea has very strong opinions on spica casts and how to manage them, so your child doesn't smell. She also has a unique way of explaining craniosynostosis to kids, using Halloween skulls, panty hose and balloons. If you're lucky, you might get to see that today!



Ivo Kalajzic, MD, PhD

Professor, Director, Center for Regenerative medicine and Skeletal Development, University of Connecticut Health Center

Dr Kalajzic's research has been focused on the enhancement of bone repair utilizing BMP2 and PDGF growth factors with the aim of improving healing. He has worked extensively on evaluating the effects of Jagged Notch signaling modulation for improving fracture healing and direct intrabone transplantation of mesenchymal stem cells as a therapeutic approach for osteogenesis imperfecta. Dr. Kalajzic received his MD from Zagreb University School of Medicine and his PhD from Split University School of Medicine. He has received numerous awards including Outstanding Researcher Award from the School of Dental Medicine-Colgate, as well as awards from the Osteogenesis Imperfecta Foundation, the Children's Brittle Bone Foundation, the American Society of Bone and Mineral Research, and the East Coast Connective Tissue Society.



Hitesh Kapadia, DDS, PhD

Chief of Craniofacial Orthodontics, Seattle Children's; Director, Fellowship Training Program in Craniofacial Orthodontics, Assistant Professor, Orthodontics, University of Washington School of Dentistry and Plastic Surgery and University of Washington School of Medicine

Dr. Kapadia has been at the Craniofacial Center at Seattle Children's Hospital since 2010. He is passionate about helping children and their families understand the complexities of cleft and craniofacial care and guiding them through various phases of treatment. He works closely with Craniofacial team members to manage the orthodontic needs of children with craniofacial differences.

He completed his DDS and residency training in Orthodontics at The University of Texas Health Science Center- Houston. He received a PhD from The University of Texas – Graduate School of Biomedical Sciences. He was then on faculty at Baylor College of Dentistry. He subsequently completed fellowship training in Craniofacial Orthodontics at New York University Medical Center, Institute for Reconstructive Plastic Surgery.

Elisa Landmann

Pfeiffer Syndrome Advocate

Elisa is 15 years old and in the 9th grade. She has Pfeiffer Syndrome and is Hard of Hearing. She is fluent in Sign Language.

Elisa is smart, fun, witty, spunky and "impressive", according to her doctors. She loves playing the piano, watching Marvel movies, playing board games with her sister and dad and traveling locally and internationally with her family. She also enjoys watching movies & TV Shows.

Ioannis Mavridis, MD, PhD, FNYAM

Assistant Professor of Neurosurgery, Head, Department of Neurosurgery, Medical School, Democritus University of Thrace, University General Hospital of Alexandroupolis, Greece

Dr. Mavridis was born in Greece and received his medical degree from the University of Athens. He was involved with neuroanatomical research and earned his PhD in stereotactic neurosurgical anatomy from the same University. After completion of his Neurosurgery residency, he received further training in neuroendoscopy (Germany) and functional neurosurgery (USA). He completed his

Clinical Fellowship in Pediatric Neurosurgery at Birmingham Children's Hospital (United Kingdom). His is author of several articles and book chapters on human stereotactic and neurosurgical anatomy, as well as various neurosurgical and neuroscientific topics. He has also authored a book on 'Stereotactic Brain Microanatomy' published in New York. Among his research achievements are the determination of the most reliable stereotactically standard part of the human nucleus accumbens and discovery of its atrophy in Parkinson's disease. His research interests include brain anatomy, stereotaxy, functional neurosurgery and pediatric neurosurgery.

Erin Mackintosh, MD

Acting Assistant Professor, Pediatric Pulmonary and Sleep Medicine, University of Washington, Seattle Children's

Dr. MacKintosh is a pediatric pulmonologist and sleep medicine physician at Seattle Children's Hospital and the University of Washington. Her clinical and research interests include care of critically ill children and the use of technology to improve comfort and breathing for kids with chronic illness.

Dr. MacKintosh worked in the field of implantable devices before attending medical school at UC San Diego. She completed her Pediatrics residency, Pediatric Pulmonary fellowship, and Sleep Medicine fellowship at the University of Washington.

Amy Merrill-Brugger, PhD

Associate Professor and Chair, Department of Biomedical Sciences, Ostrow School of Dentistry, University of Southern California

Dr. Amy Merrill received her Ph.D. in Biochemistry and Molecular Biology in 2005 from the University of Southern California. During her doctoral studies she used mouse genetics to discover a novel role for cellular boundaries in the pathogenesis of craniosynostosis. From 2005-2007 she did a postdoctoral fellowship at University of California, San Francisco where she uncovered the unique potential of cranial neural crest cells to autonomously control the timing of bone formation in the developing face. Prior to joining the faculty in the Department of Biochemistry and Molecular Biology at USC in 2010, she completed a fellowship in Medical Genetics at University of California, Los Angeles/Cedars Sinai Medical Center. Her studies in human genetics identified the first disease-causing mutations for Short-rib polydactyly syndrome and establish this lethal skeletal disorder as a ciliopathy. Currently Dr. Merrill's laboratory studies the disease mechanism for Bent Bone Dysplasia Syndrome.

Dr. Merrill was the recipient of the Basil O'Connor Starter Scholar Research Award and Gene Discovery and Translational Research Grant from the March of Dimes and currently holds two NIH R01 and an R21 grants. In 2019, Dr. Merrill was awarded the Marylou Buyse Excellence in Craniofacial Research Award from the Society for Craniofacial Genetics and Developmental Biology and currently serves as the society's Vice President.

Ellen Morgan, MS

Founder and President, PROS Foundation; Board Member, Born a Hero Research Foundation

Ellen is the founder of the Pathways for Rare and Orphan Studies (PROS) Foundation, which is a 501(c)(3) non-profit organization whose mission is to accelerate the development of effective treatments for rare diseases. Using our wealth of clinical development experience, PROS is

committed to guiding Rare Patient Groups with their registries, natural history studies, identifying disease endpoints, and preparing them for the clinical trial phase.

Ellen has 25 years of drug development experience, in large pharma, small biotech and CRO. She founded and led two successful Clinical Research Organizations – Synteract and Agility Clinical – most recently serving as President, Rare Diseases at Precision for Medicine. Agility was dedicated to clinical trials in rare diseases and supporting patient organizations. Ellen received a BS in Chemistry and MS in Management and Industrial Engineering from Rensselaer Polytechnic Institute, and post graduate studies in statistics and genetics.

Cindy Ola, PhD

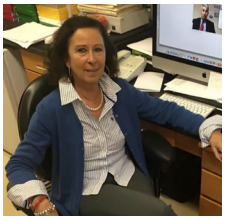
Dr. Ola holds clinical roles in the Seattle Children's Hospital (SCH) Craniofacial Center and other specialty clinics at Seattle Children's. Dr. Ola completed her graduate training at the University of Washington and her psychology residency training at the University of New Mexico Health Sciences Center, before returning to Seattle Children's to complete her 2-year postdoctoral fellowship. She has collaborated as a researcher and clinician on studies evaluating a culturally adapted parent-child intervention for Latino fathers and collaborated with others from the SCH/UW Craniofacial team on several projects related to the neurodevelopmental assessment of infants and young children with craniofacial anomalies.

David Ornitz, MD, PhD

Alumni Endowed Professor, Developmental, Regenerative and Stem Cell Biology Program, Neurosciences Program, Washington University in Saint Louis

Dr. Ornitz graduated from the MD-PhD program at the University of Washington in 1987. He was at the forefront of developing transgenic mouse technology for in vivo models of cancer and as tools to identify transcriptional regulatory elements. As a postdoctoral fellow in the Department of Genetics at Harvard Medical School, Dr. Ornitz developed a binary genetic system to model cancer and other lethal diseases in mice. He also discovered that heparan sulfate proteoglycans are required for Fibroblast Growth Factor (FGF) signaling. This discovery linked cell-surface and extracellular matrix molecules to growth factor signaling pathways. Dr. Ornitz is now an Alumni Endowed Professor in the Department of Developmental Biology at Washington University School of Medicine. He is also an elected fellow of both the American Association of Anatomy (AAA) and the American Association for the Advancement of Science (AAAS). Over the past 30 years, his research has focused on the in vivo function of FGFs in development, physiology, response to injury, and cancer, and he has made significant contributions to cardiovascular, inner ear, pulmonary, and skeletal system biology.

In the skeletal system, the Ornitz lab discovered that the mutation in FGF receptor 3 (FGFR3) that causes Achondroplasia activates receptor signaling. His laboratory created the first transgenic mouse model for Achondroplasia and showed that activation of FGFR3 decreased the proliferation of growth plate chondrocytes. This mouse model had been instrumental in demonstrating the potential efficacy of drugs that are now being used to treat this disease. Stemming from these early studies on FGFR3 signaling in the growth plate, the Ornitz lab has more recently pursued studies directed at understanding the function of FGF ligands and receptors in skeletal development, and in osteoblast and endothelial lineages, and how these cells interact with growth plate chondrocytes to regulate bone growth and homeostasis.



Natalina Quarto, PhD

Senior Scientist, Stanford University School of Medicine

Dr. Quarto is a Senior Scientist at Stanford University and leader of the Craniofacial research group in the Hagey Laboratory for Pediatric Regenerative Medicine directed by Dr. Michael Longaker. She received her PhD in Biological Sciences from the University of Napoli Federico II and Specialization in Medical Genetics from the University of Roma "La Sapienza." Natalina pursued her postdoctoral research at the New York University, Medical School with a focus on FGF-2 biology where she identified the three forms of Fibroblasts Growth Factor-2 (FGF-2). She was able to unveil and define how these forms promote distinct biological outcomes. She continued her research on FGF-2 signaling pathways at the University of Paul Sabatier in Toulouse, France and joined Dr. Michael Longaker at Stanford University in 2000, focusing on skeletal biology and regenerative medicine.

Her current research is investigating skeletal stem/progenitor cells resident in normal and craniosynostotic calvarial sutures, and their transcriptomic and molecular signature. Her group is currently studying key-signaling pathways governing the stem-ness of the calvarial suture mesenchyme with the ultimate goal to develop translational applications by using skeletal stem/progenitor cells to prevent re-synostosis upon ablation of a craniosynostotic cranial suture

Cory M. Resnick, MD, DMD, FACS

Attending Physician, Oral & Maxillofacial Surgery Program, Boston Children's Hospital; Associate Professor, Harvard Medical School

Dr. Resnick practices pediatric oral and maxillofacial surgery at Boston Children's Hospital, with an emphasis on craniofacial anomalies and syndromes, orthognathic (jaw) surgery, management of Robin Sequence and pediatric obstructive sleep apnea, management of maxillofacial cysts and tumors, and the surgical management of temporomandibular joint disorders. Dr. Resnick is an active member of the multidisciplinary Craniofacial and Vascular Anomalies teams.

He attended the University of Pennsylvania in Philadelphia for undergraduate school and continued at Penn to earn his dental degree. He then moved to Boston to earn his medical degree at Harvard Medical School and complete an Oral and Maxillofacial Surgery residency program at the Massachusetts General Hospital. Dr. Resnick is certified by the American Board of Oral and Maxillofacial Surgery and holds a Certificate of Added Qualification in Pediatric Craniomaxillofacial Surgery, and he is a Fellow of the American College of Surgeons.

Joan Richtsmeier, PhD

Distinguished Professor, Pennsylvania State University

Joan Richtsmeier is Distinguished Professor of Anthropology at the Pennsylvania State University. She received her PhD from Northwestern University in 1985 and joined the faculty of the Department of Cell Biology and Anatomy, Johns Hopkins University School of Medicine in 1986. There, she focused on establishing new quantitative methods for studying change in biological shape through time, especially in primates, with Professor Subhash Lele. In 1999 she became the 55th woman to achieve the rank of Professor at Johns Hopkins University School of Medicine since the school opened in 1893. In 2000, Dr. Richtsmeier moved her lab to the Pennsylvania State University. There, her focus turned to joining developmental biology with evolutionary biology, and with collaborators and students, she has worked to integrate the study of mouse models carrying known genetic variants with understanding the biological basis of patterns of evolutionary change. She is particularly interested in early formation of the chondrocranium and how and why cells

decide to become osteoblasts and make bone. Dr. Richtsmeier was elected Fellow of the American Association of Anatomists (AAA) in 2018, received the Henry Gray Scientific Achievement Award of the AAA in 2019, and the David Bixler Excellence in Craniofacial Research Award of the Society for Craniofacial Genetics and Developmental Biology in 2019, and the Rohlf Medal for Excellence in Morphometric Methods and Applications. She was elected Fellow of the AAAS (Section on Biological Sciences) in 2020. Her work is supported by grants from the National Science Foundation, the National Institutes of Health, and the Wellcome Trust.

Hoa Schneider, PhD

Pediatric Neuropsychologist, BCH Cleft and Craniofacial Center, Instructor Harvard Medical School/Boston Children's Hospital

Dr. Hoa Lam Schneider is a pediatric neuropsychologist in the Boston Children's Hospital Center for Neuropsychology who specializes in the care of children with craniofacial anomalies and developmentally based learning disabilities. Dr. Schneider also conducts research within the Department of Plastic and Oral Surgery investigating the neurocognitive, psychosocial, and developmental outcomes of children with nonsyndromic and syndromic craniosynostosis.

Dr. Schneider is originally from the Boston area, but she completed her undergraduate studies in Psychology at Trinity College before receiving her Ph.D. in Clinical Psychology from the University of Miami. Afterwards, she completed her internship with a focus in pediatric neuropsychology at the University of Florida, followed by two years of post-doctoral training in pediatric neuropsychology at Boston Children's Hospital, where she joined the faculty in 2019.

Carolina Sommer

Host of the FGFR Syndrome Collaborative Research Network Virtual Conference; Founder & CEO of Born a Hero Research Foundation; Founder of Seattle Rare Disease Fair; author of the Lucy's Journey book series; Co-founder of the "ABC Kind Program" with Gerry Ghanooni (a national curriculum that teaches kids about loving differences, including medical differences)

Carolina is a member of the Rare Disease Access Working Group with EveryLife Foundation, We Work For Health, Voters for Cures, and WA Health Access Network. Carolina is currently working to launch a Rare Disease Council in Washington State with Max Brown from We Work for Health. She has also partnered with a few organizations around the country in hopes to start a K.I.N.D Initiative Council.

Carolina was born in Medellin, Colombia and has lived in the Seattle area since age 8. She received a BS in Theoretical Math from the University of Washington and was an intern at NASA's Jet Propulsion Laboratory in Pasadena, California. Carolina is a public speaker, a certified Wedding and Event Planner, and a Refit Revolution dance instructor. She is an artist, and a former actress. In 2012

Carolina gave birth to Mariana, who has Pfeiffer Syndrome. She is now a stay home mom of two and homeschools her kids.

Srinivas Susarla, DMD, MD, MPH

Associate Professor, Plastic Surgery, University of Washington School of Medicine, Oral and Maxillofacial Surgery, University of Washington School of Dentistry

Dr. Susarla completed his undergraduate education at Princeton University. He completed his dental degree at the Harvard School of Dental Medicine, a master of public health degree at the Harvard School of Public Health, and his medical degree at Harvard Medical School. He completed surgical residencies in Oral and Maxillofacial Surgery at the Massachusetts General Hospital and Plastic Surgery at the Johns Hopkins Hospital. This was followed by fellowship training in Craniofacial Surgery at Seattle Children's Hospital/University of Washington Harborview Medical Center.

Dr. Susarla's clinical practice focuses on pediatric and adult craniomaxillofacial reconstructive surgery, including management of craniosynostosis, cleft lip and palate, dentofacial deformities, maxillofacial pathology, and primary and secondary reconstruction following facial trauma. He has an active research portfolio, with over 200 peer-reviewed publications and membership on the editorial boards of several premier journals in craniomaxillofacial surgery.

Klane K. White, MD, MSc

Dr. White is an internationally recognized expert and advocate in the care of mucopolysaccharidosis and skeletal dysplasia. He serves on the Medical Advisory Board of Little People of America, the Scientific Advisory Board of the National MPS Society and is an executive founding member of the Skeletal Dysplasia Management Consortium. In addition to skeletal dysplasia and the mucopolysaccharidoses, Dr. White's clinical and research interests also include the diagnosis and management of early onset scoliosis, metabolic bone disease, and complex spine deformity. Dr. White has authored more than 90 peer-reviewed publications, articles and book chapters, serves as reviewer for multiple medical journals and is principal investigator in several multicenter research studies for rare disease.

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