FGFR Syndromes
Collaborative Research Network Virtual Conference

October 9, 2020

Agenda and Event Access Links

All times are listed in Pacific time

7:30 a.m.	Join the Conference
7:45 a.m.	Orientation to Webex and Conference Flow Brittany Richey, *Manager, Administration, Seattle Children's Research Institute*
8:00 a.m.	An Introduction Carolina Sommer, *Founder and CEO, Born a Hero Research Foundation*

Laboratory Approach Presentations

8:10 a.m.	Introductions and Moderation by Dr. Aris Economides, *Vice President of Research, Regeneron Pharmaceuticals*
8:11 a.m.	Fibroblast Growth Factors: Essential signaling pathways for skeletal development and homeostasis Dr. David Ornitz, *Alumni Endowed Professor of Developmental Biology, Washington University in St. Louis*
8:27 a.m.	Craniosynostosis and Dwarfism Syndrome Mutations Hijack the Physiological Mechanisms of FGF Receptor Regulation Dr. Moosa Mohammadi, *Professor of Biochemistry and Molecular Pharmacology at New York University*
8:42 a.m.	Laboratory Approach Discussion and Q&A
8:57 a.m.	*Break*
9:13 a.m.	Patient Video Presentation: *Fearless,* as recited by Abby McGowan

Translational Approach Presentations

9:15 a.m.	Introductions and Moderation by Dr. Devaveena Dey, *Senior Staff & Assistant Professor in the Department of Surgery, Uniform Services University of the Health Sciences*
9:17 a.m.	A Lesser-Known FGFR Syndrome: Bent Bone Dysplasia Dr. Amy Merrill-Brugger, *Assistant Professor of Biomedical Sciences, Biochemistry, and Molecular Biology, University of Southern California*
9:33 a.m.	Selfish spermatogonial selection and the origins of FGFR syndromes Dr. Andrew Wilkie, *Nuffield Professor of Pathology, Oxford University*
9:49 a.m.	Animal Models of FGFR Syndromes Dr. Ethylin Jabs, *Mount Sinai Professor of Developmental Genetics, Icahn School of Medicine*
10:04 a.m.	Translational Approach Discussion and Q&A *Dr. Gregory Holmes, Assistant Professor of Genetics and Genomic Sciences, Mt Sinai, will represent Dr. Ethylin Jabs*
10:19 a.m.	*Break*
10:31 a.m.	Patient Video Presentation: *Lucy has a Baby Brother*, by Mariana Sommer

Clinical Approach Presentations

10:40 a.m.	Introductions and Moderation by Dr. Jesse Goldstein, *Associate Professor of Plastic Surgery, University of Pittsburgh*
10:42 a.m.	Treating Syndromic Craniosynostosis: A Surgeon's Experience Dr. Jeffrey Fearon, *Director of the Dallas Craniofacial Center, Medical City Dallas Children's Hospital*
10:58 a.m.	Airway Management in Patients with FGFR Related Syndromes: A Translational Perspective Dr. John Dahl, *Otolaryngologist, Seattle Children's;* *Assistant Professor, Department of Otolaryngology - Head and Neck Surgery, University of Washington*
11:18 a.m.	Caring for a Children with FGFR Related Syndromes: Working as a team Dr. Kelly Evans, *Craniofacial Physician, Seattle Children's;* *Assistant Professor, Department of Pediatrics, University of Washington*
11:38 a.m.	Clinical Approach Discussion and Q&A
12:00 p.m.	*Break*
12:12 p.m.	Patient Video Presentation: Hula hoop performance from Madisyn Bowers

Keynote Presentation

12:15 p.m.	Introduction Dr. Scott Mellis, *Vice President of Translational Medicine, Regeneron Pharmaceuticals*
12:20 p.m.	The Collaborative Network Approach: how it accelerated discoveries for Castleman disease and can speed up discovery in FGFR syndromes Dr. David Fajgenbaum, *Assistant Professor of Medicine in Translational Medicine & Human Genetics, University of Pennsylvania; Associate Director of Patient Impact, Penn Orphan Disease Center; Founding Director; Center for Cytokine Storm Treatment & Laboratory; Co-Founder & Executive Director, Castleman Disease Collaborative Network*
12:40 p.m.	Q&A
12:50 p.m.	Breakout Sessions Leave Meeting and join your breakout session: Laboratory Translational Clinical Advocacy
1:50 p.m.	*Break; "Leave Meeting" and return to Conference*
	Re-Join the Conference
1:56 p.m.	Patient Video Presentation: "Fight Song," performed by Elisa Landmann
2:00 p.m.	Breakout Summary Reports and Q&A Moderated by Dr. Joan Richtsmeier, *Distinguished Professor of Anthropology, Pennsylvania State University*
3:05 p.m.	Open Discussion: How to move forward as a collaborative network
3:55 p.m.	Final remarks Carolina Sommer, *Founder and CEO, Born a Hero Research Foundation*
4:00 p.m.	Adjourn

Conference Mission

To bring together laboratory, translational, and clinical researchers as well as patients and families to launch an innovative and impactful initiative to develop management and treatment for the complex and multisystem FGFR syndromes, including but not limited to Apert, Crouzon, Pfeiffer, and Muenke syndromes. Professionals from multiple disciplines will come together and will share and discuss how research can advance treatment approaches. International collaborative discussions will generate a strategic plan to improve research, treatment, care, and resources for individuals with FGFR syndromes.

Guiding Questions for Discussion

1. What research questions are most important to answer?
2. What studies are most important to conduct to answer these key research questions?
3. What will be the impact of these studies in improving patient outcome?
4. What resources are needed to perform these key studies?
5. What collaborative groups of researchers are needed to perform these studies?
6. What is the order in which the research studies should be undertaken?
7. How can these studies be funded for targeted studies?

Tips to Prepare

- Download the Webex meetings app desktop version
- Plan to participate in the event from a laptop or desktop computer. Do not download the mobile version.
- If you are joining from an international location or from a location that has high security firewalls in place, we recommend working with your IT department to ensure the Webex app will function properly.

How to Join the Event

- Join a few minutes early to ensure proper connectivity and audio.
- Click the "Join Event" link from your computer, NOT on your mobile device or tablet.
- Use your computer audio, or select the option to receive a call after you join on your computer. If you do call in separately using your phone, ensure you enter your unique participant ID (available in the "I will call in" menu in Audio/Video connection). This ensures your video and audio will be linked and you will have a greatly improved attendee experience.

Information for During the Event

- Your camera and audio will be automatically off during the main event session. You will be able to use the chat function at any time.
- During the Q&A you can chat your questions or raise your hand and the host will unmute you so you can speak your question.
- When the breakout session begin you will leave the main session and join the breakout with the link and information provided. In the breakout you can turn on your camera and speak freely. (Please mute yourself when you are not speaking.)
- After the breakouts, re-join the main event using the same link you used to join at the beginning.
- Should you need assistance at any point throughout the event, please email SCRI@seattlechildrens.org or send a chat to Brittany Richey via the Webex chat feature.

Expanded Virtual Event Access Information

Main Conference – 7:30 a.m. – 4 p.m. (Pacific)

URL: https://seattlechildrens.webex.com/seattlechildrens/onstage/g.php?MTID=ed634fb2e72ce2a9894c0b5acdf2d51e5
Event number: 133 204 7238
Event password: conference

Breakout Sessions – 12:50 – 1:50 p.m. (Pacific)

Laboratory
URL: https://seattlechildrens.webex.com/seattlechildrens/j.php?MTID=m37083408b965786a7a44d8e9ca03b414
Meeting Number (access code): 133 755 8089
Meeting password: laboratory

Translational
URL: https://seattlechildrens.webex.com/seattlechildrens/j.php?MTID=mf7075f9d9dd6e54cc2edf8913d1cdd70
Meeting number (access code): 133 227 4920
Meeting password: translational

Clinical
URL: https://seattlechildrens.webex.com/seattlechildrens/j.php?MTID=m46f738264965ff59570cadf6e32ef4c4
Meeting number (access code): 133 945 2605
Meeting password: clinical

Advocacy
URL: https://seattlechildrens.webex.com/seattlechildrens/j.php?MTID=m7de9acca42542640250dd5d0e00687fd
Meeting Number (access code): 133 558 6092
Meeting password: advocacy

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VIRTUAL CONFERENCE – FGFR SYNDROMES, COLLABORATIVE RESEARCH NETWORK

SPEAKERS TALK TITLES AND DESCRIPTIONS

Carolina Sommer

Talk Title: An introduction

Talk Description: Why we started Born a Hero, what we have done, and what we hope to accomplish at the conference.

Dr. David Ornitz

Talk Title: Fibroblast Growth Factors: Essential signaling pathways for skeletal development and homeostasis

Talk Description: This talk will discuss the functions of FGF and FGF receptor signaling pathways in the developing skeleton, in growing bone, and in homeostatic maintenance of bone.

Dr. Moosa Mohammadi

Talk Title: Craniosynostosis and Dwarfism Syndrome Mutations Hijack the Physiological Mechanisms of FGF Receptor Regulation

Talk Description: I will begin by giving a succinct overview of key structural insights into the mechanism of FGF signaling regulation. I will then describe how craniosynostosis and dwarfism syndrome mutations confer a gain-of-function by hijacking various physiological control mechanisms of FGFR regulation.

Dr. Amy Merrill-Brugger

Tentative Talk Title: A Lesser-Known FGFR Syndrome: Bent Bone Dysplasia

Talk Description: I will highlight the progress made in understanding the mechanisms of bent bone dysplasia syndrome and discuss the future studies needed to advance patient care.

Dr. Ethylin Jabs

Talk Title: Animal Models of FGFR Syndromes

Talk Description: An overview of animal models available for translational research into FGFR syndromes will be present. An update of preclinical therapeutics that have been tested and potential strategies to be considered will be discussed.

Dr. Andrew Wilkie

Talk Title: Selfish spermatogonial selection and the origins of FGFR syndromes

Talk Description: Germline mutation rates for FGFR-associated craniosynostosis disorders, especially Apert and Muenke syndromes, are the highest known across all genetic diseases. This is driven by a process occurring in the testes of all men as they get older, termed selfish spermatogonial selection. Abnormal FGF signalllng during early embryogenesis underlies the major skeletal features of these disorders, posing significant challenges for prevention or therapy.

Dr. Jeffrey Fearon

Talk Title: Treating Syndromic Craniosynostosis: A Surgeon's Experience

Talk Description: This presentation will briefly review a single center's experience with treating syndromic craniosynostosis. Also presented will be an overview of our past clinical research, followed by a discussion of future areas for study, particularly those having the potential to impact treatment.

Dr. John Dahl

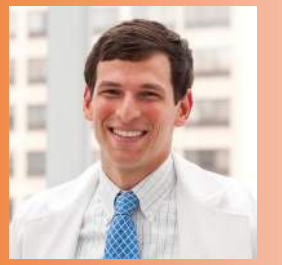
Talk Title: Airway Management in Patients with FGFR Related Syndromes: A Translational Perspective

Talk Description: I will review basic scientific data on tracheal cartilaginous sheath formation in mouse models of FGFR related craniosynostosis syndromes. I will then review our experience with airway management in patients with FGFR related craniosynostosis syndromes. I will end with a discussion on the importance of interdisciplinary management of these patients.

Dr. Kelly Evans

Talk Title: Caring for a Children with FGFR Related Syndromes: Working as a team.

Talk Description: I will review the clinical features of FGFR related syndromes at diagnosis and across the lifespan. I will focus on the need for a collaborative and interdisciplinary approach. I will also discuss opportunities to partner with patients and parents to provide holistic and patient-centered care.



Dr. David Fajgenbaum

Talk Title: The Collaborative Network Approach: how it accelerated discoveries for Castleman disease and can speed up discovery in FGFR syndromes

Talk Description: Dr. David Fajgenbaum, co-founder of the Castleman Disease Collaborative Network, physician-scientist at UPenn, and author of the national bestselling book, Chasing My Cure: A Doctor's Race to Turn Hope Into Action, will share lessons learned from his journey chasing a cure for Castleman disease, which he was diagnosed with during his third year of medical school. Dr. Fajgenbaum will share tangible steps and advice for adopting aspects of the Collaborative Network Approach. He is currently in his longest remission ever thanks to a drug that he identified and is testing in other patients.

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GET TO KNOW OUR PANEL OF EXPERTS

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, founded 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. She has lived in the Seattle area since she was 8 years old. She graduated from the University of Washington with a degree in Theoretical Math 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. Her hobbies include being with her family, painting, playing basketball, tennis, hiking, watching movies, and playing the guitar.

Dr. Aris Economides- Moderator and Vice President of Research at Regeneron Pharmaceuticals

Dr. Aris N. Economides received his Ph.D. in Biochemistry from Michigan State University in 1992, and promptly joined Regeneron Pharmaceuticals. He currently holds the position of Vice President, leading two groups: Genome Engineering Technologies, and Skeletal Diseases Therapeutic Focus Area. In addition, he is a co-founder of Regeneron Genetics Center (RGC), where he is also Head of Functional Modeling. Dr. Economides co-invented Cytokine Traps, VelociGene®, and VelocImmune®, all part of an integrated methodology for target discovery, validation, and the generation of biologic drugs such as the IL1 and VEGF traps, as well as therapeutic antibodies. More recently, he has been developing a new method for Enzyme Replacement Therapy (ERT), one that addresses two of the main limitations of current ERT, namely immunogenicity and inefficient uptake by the tissues most affected in the corresponding Lysosomal Diseases. As part of his involvement with the RGC, Dr. Economides has been working to elucidate the molecular pathophysiology of genetically-driven disorders. An example is his work in Fibrodysplasia Ossificans Progressiva, where he and his team discovered a novel mechanism that explains important aspects of FOP's pathophysiology and pinpoints a new potential route to therapy.

Dr. David Ornitz- Speaker

Alumni Endowed Professor of Developmental Biology at Washington University

He is a graduate student in the MD-PhD program at the University of Washington. 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, I developed a binary genetic system to model cancer and other lethal diseases in mice. I 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. Over the past 27 years, I have led an independent research laboratory at Washington University School of Medicine. My research has primarily focused on the in vivo function of FGFs in development, physiology, response to injury, and cancer. My laboratory has made significant contributions to cardiovascular, inner ear, pulmonary, and skeletal system biology. My laboratory has designed and engineered knockout and conditional knockout alleles, tetracycline regulatory alleles, and transgenic mouse lines for several FGF ligands and receptors, allowing us to probe gene function, understand mechanisms regulating organogenesis, model human disease, and develop genetic tools to model and test therapeutic strategies. I have also been very interested in understanding how FGF signaling pathways interact with other signaling pathways and transcription factors to coordinate complex developmental, injury response, regenerative processes, and cancer.

Dr. Moosa Mohammadi- Speaker

Professor of Biochemistry and Molecular Pharmacology at New York University

Dr. Mohammadi received his Ph.D. in Biochemistry from University of Zurich in Switzerland and completed his postdoctoral training in the laboratory of Dr. Joseph Schlessinger in the Department of Pharmacology at New York University School of Medicine. In 1997, he established his own research group in the same department where he is currently an Associate Professor. Dr. Mohammadi's group investigates the mechanisms of fibroblast growth factor (FGF) signaling using a variety of methods including x-ray crystallography, surface plasmon resonance, steady-state kinetic analysis and cellular signaling in cultured cells.

Dr. Devaveena Dey- Moderator

Senior Staff and Assistant Professor in the Department of Surgery at Uniform Services University of the Health Sciences, Postdoctoral Fellow at Brigham & Women's Hospital, and Research Fellow at Harvard Medical School.

Dr. Dey is a stem cell scientist, who is currently pursuing her research in a pediatric rare genetic musculoskeletal disorder, 'Fibrodysplasia Ossificans Progressiva' (FOP). This disorder is caused by a mutation in the bone morphogenetic protein (BMP) type I receptor, ALK2. The BMP pathway is the major signaling pathway underlying bone formation and development. This mutation results in abnormal bone formation within skeletal muscles, joints, and the rib cage, resulting in premature death due to cardiothoracic insufficiency. This ectopic bone formation is often triggered by trauma and injury.

As part of her research efforts, Dr. Dey is trying to understand the effect of the mutation and injury on abnormal bone formation in different tissues. She demonstrated that distinct stem cell populations mediate this pathological bone formation in different tissues, like muscles and tendons under the effect of the mutation. She demonstrated that while injury appears to be critical for intramuscular ossification, tendons and ligaments ossify spontaneously under the effect of the ALK2 mutation, without induction of injury.

Dr. Dey has extensive experience in the study of stem cell function in different tissues under various physiological and pathological conditions. Prior to her work on FOP, she was involved in cardiac and bone marrow stem cell research at the Stanford University School of Medicine. Her graduate work focused on understanding the link between normal stem cells and breast cancer. Dr. Dey has authored multiple research articles, reviews and book chapters on stem cells. She has also played a pivotal role in drafting this Natural History Study Grant. Her research expertise, coupled with her recent work on abnormal bone formation in the rare genetic disorder, FOP (which might have important links with Pfeiffer's), makes her an ideal candidate in our scientific team, to work collaboratively with Dr. Jabs.

She is highly motivated to help out the broader rare disease community through her scientific expertise. Currently she also serves as a volunteer writer-editor with the National Organization for Rare Disorders (NORD).

Dr. Amy Merrill-Brugger - Speaker

Assistant Professor of Biomedical Sciences, Biochemistry, and Molecular Biology at 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 were 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. Jesse Goldstein- Moderator

Associate Professor of Plastic Surgery at University of Pittsburgh

Dr. Jesse Goldstein is an attending surgeon in the Department of Plastic Surgery at University of Pittsburgh Medical Center and the Division of Plastic Surgery at the Children's Hospital of Pittsburgh (CHP) of UPMC. He specializes in the treatment of children and adolescents with cleft lip, cleft palate, and disorders of the craniofacial skeleton including craniosynostosis and Pierre Robin sequence. He serves as Director of the Pediatric and Craniofacial Surgery Fellowship at CHP.

Dr. Goldstein was born in the San Francisco Bay Area and moved to Pennsylvania for his undergraduate and medical training at the University of Pennsylvania. While at Penn, Dr. Goldstein completed a Doris Duke clinical research fellowship in Pediatric Trauma and outcomes research. His plastic surgery residency was completed at Georgetown University hospital before returning to Philadelphia for a fellowship in Craniofacial and Pediatric Plastic surgery at the Children's Hospital of Philadelphia and the University of Pennsylvania Medical Center.

Dr. Goldstein's clinical focus is centered around improving the function and appearance of children born with craniofacial disorders. He specializes in rhinoplasty, cleft lip/palate repair, orthognathic surgery, cranial reconstruction for craniosynostosis, and cranio-maxillofacial distraction osteogenesis.

Additionally, Dr. Goldstein is actively involved in research focused on outcomes in cleft and craniofacial surgery and he is founding member of several multi-centered outcomes studies. His work is funded by societal, national, and international scientific agencies.

Dr. Ethylin Jabs- Speaker

Mount Sinai Professor of Developmental Genetics at Icahn School of Medicine

Dr. Jabs is a clinical geneticist, with expertise in medical genetics, pediatrics, and craniofacial biology. In addition to her appointment as the vice chair of the Department of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai Medical Center, she also heads the Interdisciplinary Training in Systems and Developmental Biology in Birth Defects at the Mount Sinai Graduate School of Biomedical Sciences. Before joining Mount Sinai, Dr. Jabs was at the John Hopkins School of Medicine, where she was the Dr. Frank V Sutland Professor of Pediatric Genetics, Director of the Center for Craniofacial Development and Disorders, and Director of the International Collaborative Genetics Research Training Program. She still holds the position of adjunct professor in pediatrics, medicine, and surgery at John Hopkins.

Dr. Jab's research and clinical practice have focused on developmental genetics and patients with birth defects. Her clinical areas include prenatal diagnosis, birth defects, multiple congenital anomalies, craniofacial and limb anomalies, achondroplasia, macrosomia, micro- and macrocephaly, oral facial clefts, short stature, spina bifida and multiple associated syndromes like Crouzon's, DiGeorge, Down's, Fetal Alcohol, Fragile X and Turner's syndromes. Dr. Jabs is an advisor to several parent support groups, including 'Smile Train'.

Dr. Jab's research focuses on unraveling the mechanisms underlying the mutations which cause birth defects and malformations. Her laboratory was responsible for the identification of the first human mutation in a homeobox-containing gene, MSX2, an important regulatory gene in development. In her studies on craniosynostosis, she discovered that similar mutations in the gene, fibroblast growth factor receptor 2 (FGFR2), cause both Jackson-Weiss syndrome and Crouzon syndrome. Dr. Jab's team also validated novel genetic links underlying 'sagittal craniosynostosis', a common birth defect, where the bones on the side and top of the skull fuse prematurely. This genome-wide association study was published in a reputed medical journal, Nature Genetics in November, 2012. The group also demonstrated in a mouse model that inhibiting the protein p38 resulted in prevention of craniosynostosis in a rare genetic disorder, Beare-Stevenson cutis Gyrata syndrome (BSS).

For some of these conditions, Dr. Jabs demonstrated the association of advanced paternal age at conception. She has studied the increased frequency of spontaneous mutations arising in sperm with aging. She also initiated a database of clinical and genetic data for people with craniofacial disorders including those with Möbius syndrome, a rare neurological disorder, to help identify the genetic root of the condition. Dr. Jabs has published close to hundred research articles and book chapters on the genetics underlying craniofacial development and defects. Her research has been funded through multiple grants from the NIH, CDC, patient advocacy groups and Icahn School of Medicine

Dr. Andrew Wilkie- Speaker

Nuffield Professor of Pathology at Oxford University

I've been employed as an Honorary Consultant in Clinical Genetics in Oxford since 1993, and my work has always been driven by the desire to give patients and families better answers to the questions they ask me in clinic. Working with plastic surgeons, my primary interest is in craniofacial malformations in children especially craniosynostosis, the premature fusion of the cranial sutures of the skull. By identifying the molecular genetic basis of these conditions, not only can we give families the answers they seek, we also gain fundamental knowledge about the details by which a human skull is built. A key early discovery (1995) was that Apert syndrome, in which the craniosynostosis occurs together with fusions of the fingers and toes, is caused by highly localized, recurrent mutations in the fibroblast growth factor receptor type 2 (FGFR2) gene. From this, two major research themes developed discovering other genetic causes of craniosynostosis, and finding out why certain genetic misprints such as the Apert FGFR2 mutations occur up to a thousand times more frequently than they should. The work on craniosynostosis has led to many important disease gene discoveries, for which genetic testing has been translated into the NHS. Work on the origins of the mutations led to the recognition of a novel process occurring in the testes, which we termed 'selfish spermatogonial selection', that provides a link between the origins of germline and somatic mutation. Current efforts focus on harnessing the technological revolution provided by next generation sequencing to identify even more new genetic causes of craniosynostosis. Using this information, we can explore the complex mechanisms by which a population of stem cells is maintained within the sutures to keep the suture open, yet continuously turns over to promote continued growth of the skull.

Dr. Jeffrey Fearon- Speaker

Director of the Craniofacial Center at Children's Medical Center

Dr. Jeffrey Fearon was raised in London, England and New Canaan, Connecticut. After graduating the Mt. Hermon School in Massachusetts, he received his B.A. from Brown University. He subsequently pursued post-baccalaureate studies at Columbia University and then attended medical school at the University of Cincinnati. After completing a full general surgery residency at the Harvard Fifth surgical service in Boston, he pursued a plastic surgery residency at the Massachusetts General Hospital, with Harvard Medical School. Following a one-year craniofacial fellowship at the Children's Hospital of Philadelphia at the University of Pennsylvania, he joined Dr. Ian Munro at the Dallas Craniofacial Center as a director. Today, Dr. Fearon has an international practice that is limited to craniofacial surgery; specifically, the treatment of pediatric congenital birth defects.

Dr. Fearon is President Emeritis of the Texas Society of Plastic Surgeons, as well as President Emeritus of the American Society of Craniofacial Surgeons. He has also functioned as both Chief of Plastic Surgery and Chief of Pediatrics at Medical City Dallas Hospital. In addition to serving on the editorial board of the Journal of Plastic and Reconstructive Surgery, he has authored many scientific articles and book chapters, has received a U.S. patent for a bone distraction device, and remains actively engaged in clinical research. Dr. Fearon and his wife have three daughters. When not at work, he enjoys both surfing and heli-skiing.

Dr. John Dahl- Speaker

Pediatric Otolaryngologist at Seattle Children's Hospital and Assistant Professor in the Department of Otolaryngology- Head and Neck Surgery at the University of Washington

Following his undergraduate education at Villanova University, he obtained a PhD in Pharmacology and an MBA from The Pennsylvania State University. Dr. Dahl subsequently worked as a basic scientist including positions in the pharmaceutical industry and the University of Pennsylvania. Dr. Dahl earned his medical degree from Sidney Kimmel Medical College of Thomas Jefferson University, 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. 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. Kelly Evans- Speaker

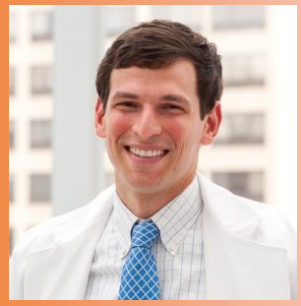
MD is an attending physician at Seattle Children's Hospital and an assistant professor in the Department of Pediatrics at the University of Washington School of Medicine.

Dr. Kelly Evans is an Assistant Professor of Pediatrics in the Division of Clinical Medicine at the University of Washington. Dr. Evans's scholarship focus includes working with diverse teams to develop pathways to facilitate evidence-based care for children with rare conditions. e.g. Robin sequence. Her research focuses on developing models to understand and treat airway obstruction and sleep apnea in infants and children with craniofacial conditions. She is dedicated to developing and nurturing collaborative partnerships to improve care delivery and outcomes for children with craniofacial conditions.

Dr. Scott Mellis- Moderator

Vice President, Early Clinical Development and Experimental Sciences, Rare Diseases at Regeneron Pharmaceuticals

Dr. Mellis received medical scientist training at Washington University School of Medicine and postgraduate training in Internal Medicine and Rheumatology at Columbia University College of Physicians and Surgeons. He joined Pfizer Pharmaceuticals Group in 1990 with a focus on Zithromax® (azithromycin), including research on B. burgdorferi in collaboration with academic colleagues. Dr. Mellis joined Regeneron Pharmaceuticals in 2001, and led clinical development for ARCALYST® (rilonacept), a medication for an ultra-rare auto-inflammatory disease. He then led Regeneron's translational and precision medicine initiatives across the general medicine portfolio. He is a co-founder of the Regeneron Genetics Center. Dr. Mellis is now leading an effort to optimize Regeneron's development of new medications for patients with rare diseases.



Dr. David Fajgenbaum- Keynote Lecturer

MD, MBA, MSc, FCPP, Assistant Professor of Medicine in Translational Medicine & Human Genetics at the University of Pennsylvania, Director of the Penn Center for Cytokine Storm Treatment & Laboratory (CSTL), Executive Director of the Castleman Disease Collaborative Network (CDCN), and Associate Director, Patient Impact for the Penn Orphan Disease Center.

David Fajgenbaum, MD, MBA, MSc, is a groundbreaking physician-scientist, disease hunter, speaker, and bestselling author of the acclaimed memoir, Chasing My Cure: A Doctor's Race to Turn Hope Into Action.

Dr. Fajgenbaum went from being a beast-like college Quarter-back to receiving his last rites while in medical school and nearly dying four more times battling Castleman disease. To try to save his own life, he spearheaded an innovative approach to research through the Castleman Disease Collaborative Network (CDCN) and discovered a treatment that is saving his life and others.

Now, he is spreading this approach to other diseases such as COVID19 and sharing lessons he learned about life, hope, and resilience from nearly dying through Chasing My Cure, which has been translated into five languages and named one of the "Best Non-Fiction Books of 2019" by Next Big Ideas Club.

One of the youngest individuals ever appointed to the faculty at Penn Medicine and the top 1 percent youngest grant awardees of a leading NIH grant (R01), Dr. Fajgenbaum has been recognized on the Forbes 30 Under 30 list, as a top healthcare leader by Becker's Hospital Review, the Global Genes RARE Champion of Hope: Science awardee, and one of three recipients--including Vice President Joe Biden--of a 2016 Atlas Award from the World Affairs Council of Philadelphia. He has published scientific papers in high-impact journals such as Blood, Lancet Hematology, and the Journal of Clinical Investigation, including a paper selected as one of the top innovations in science and medicine by STAT News in 2020. Before co-founding the CDCN, Dr. Fajgenbaum co-founded and led the Actively Moving Forward Support Network, a non-profit organization dedicated to supporting grieving college students. Dr. Fajgenbaum has been profiled in a cover story by The New York Times as well as by Good Morning America, CNN, and the Today Show, among others.

Dr. Joan Richtsmeier- Moderator

Joan Richtsmeier is Distinguished Professor of Anthropology at the Pennsylvania State University with faculty appointments in the Graduate Programs in Genetics and the Graduate Program Option in Bioinformatics and Genomics of the Huck Institutes of the Life Sciences. 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. In 1999 she became the 55th woman to achieve the rank of Professor at Johns Hopkins School of Medicine since the school opened in 1893. In 2000, Dr. Richtsmeier moved her lab to the Pennsylvania State University where she works with undergraduate students, graduate students, postdoctoral scholars and colleagues to understand the developmental basis of disorders of the head. Dr. Richtsmeier was the State of Maryland's Outstanding Young Scientist in 1990, received the W. Barry Wood Award for Outstanding preclinical teaching at Johns Hopkins in 1993, the Distinction in the Life Sciences Award from the College of the Liberal Arts Penn State in 2012, and the Faculty Scholar Medal for Outstanding Achievement in the Life and Health Sciences at Penn State in 2014. Her work is supported by grants from the National Science Foundation, the National Institutes of Health, and the Wellcome Trust.

Dr. Richtsmeier's research focuses on changes in developmental processes that occur in craniofacial diseases like craniosynostosis and Down syndrome. She and collaborators use laboratory mice to model disease mechanisms and have established precise parallels in human and mouse craniofacial disease phenotypes. Her team seeks to understand the complex genetic and developmental basis of variation in head shape in development, especially changes that occur prenatally. Her recent work focuses on the chondrocranium, a transient, cartilaginous endoskeleton of the head that protects the brain and principal sense organs before skull bones mineralize. Working with bioengineers, her team seeks to understand how mechanical stimuli of the cells of the formative brain, skull, and other tissues contribute significantly to the process of craniofacial development. Understanding the interactions of genetic signaling systems and biomechanical forces fundamental to the functional and structural association of developing tissues in normal craniofacial development and conserved over evolutionary time is essential to understanding how changes in development cause disease.

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