Innovations in Diagnostics and Therapeutics: An Interdisciplinary Virtual Rare Disease Symposium

April 20, 2022

tinyurl.com/RareDisease2022

UCSF Innovation Ventures | Foundation Ipsen

Rare Diseases

affect more than 350 million people worldwide. The UCSF Catalyst Program, in partnership with Foundation Ipsen, welcomes you to our one-day virtual rare disease symposium. With a focus on the interdisciplinary development of new diagnostics, new therapeutics, and social advances for patients and their families affected by a rare disease, we hope all participants come away from this symposium with an expanded understanding of the field and the inspiration to

Make an Impact

ABOVE By Matthew N McCarroll: Lateral view of a 5 day old zebrafish larvae expressing human Tau-mEos3 in sensory neurons.

Wednesday, April 20, 2022

Morning

9:00 – 9:15	Welcome and Introductions
9:15 – 9:45	Rare Disease Therapeutics - Soup to Nuts Tiina Urv, PhD *Program Director, Rare Disease Clinical Research Network*
9:45 – 10:45	Case Study: Sphingosine Phosphate Lyase Insufficiency Syndrome (SPLIS) SPLIS: From Gene Discovery to Gene Therapy Julie Saba, MD, PhD *UCSF Professor of Pediatrics, Hematology/Oncology* Patient Story: Living with SPLIS Ehtesham Khalid, MD *Neurophysician at Ideal Medicare Clinic*
10:45 – 11:15	State of the Art Diagnostics Stephen Kingsmore, MD, DSc *President and CEO of Rady Children's Hospital*
11:15 – 11:45	Negotiating the Challenges of Therapeutics Discovery for Rare Diseases Emil D. Kakkis, MD, PhD *Founder, President, and CEO, Ultragenyx*
11:45 – 12:15	n-Lorem: Meeting the Needs of n-1 Patients One Patient at a Time Stanley Crooke, MD, PhD *Founder and CEO, n-Lorem*

Wednesday, April 20, 2022

Afternoon

12:15 – 1:00 Rare Voices: A Concert of Science
Featuring musicians and poets with a rare disease

1:00 – 2:20 Case Study: Sickle Cell Disease (SCD)

Hope for a Cure: Treating Sickle Cell Disease with CRISPR
Featuring Brooklyn Haynes

Curative Therapies for SCD
Mark C. Walters, MD
UCSF Professor of Pediatrics

Understanding and Addressing Health Disparities in SCD
Marsha J. Treadwell, PhD
UCSF Professor of Pediatrics, Hematology

2:20 – 2:55 Defining a New Future for People with Rare Diseases
Amrit Ray, MD, MBA
Chief Patient Officer, Biohaven Pharmaceuticals

2:55 - 3:00 Closing Remarks

Jim Levine, MD, PhD
President, Foundation Ipsen

Charles Hart, PhD
Director, UCSF Catalyst Program

Objectives

- Assemble a panel of experts on rare disease detection and treatment, that includes experts from the academic sector, biotechnology, health industries, social media, and public policy.
- Initiate concrete international cooperation within this network.
- Establish a framework in which key experts from diverse and critically important sectors in rare disease discuss innovations in diagnostics and therapeutics.
- Produce a roadmap in order to achieve objectives and framework on rare disease detection following the meeting on April 20, 2022.
- Provide long-term, tangible resources from this event, making content from the meeting freely available worldwide online via YouTube.

Overview

7,000 RARE DISEASES have an impact on 350 million people worldwide. This means that 1 in 11 Americans will have a rare disease in their lifetimes. Three-fourths of all patients with a rare disease are children, many of whom are affected for their whole lives. Out of these suffering patients, half will not receive an accurate diagnosis, and one in four patients will have to wait years for a diagnosis. The average delay in diagnosis is one-and-a-half years, during which time patients may either be misdiagnosed, incorrectly treated, and encounter unnecessary anguish.

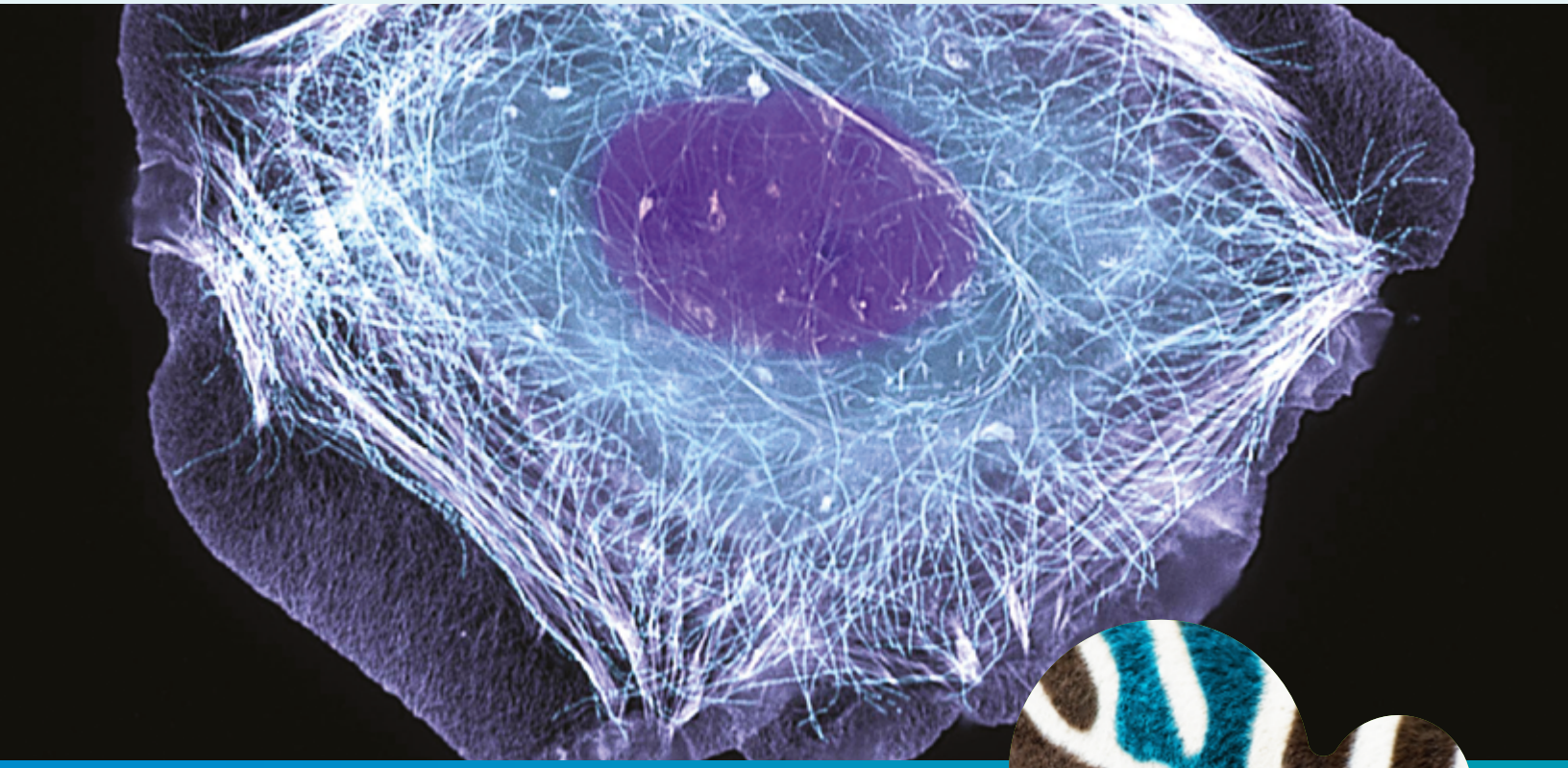
Rare disease detection affects people worldwide and by its nature presents great challenges including the limited availability of worldwide genetic testing, and the necessary research infrastructure needed to discover and develop treatments.

Nevertheless, the landscape is evolving with hundreds of new, rare diseases being discovered each year. More people are receiving genetic testing, driving a need for targeted therapies – sometimes these therapies are designed for only a few dozen, or even a single, patient.

We are holding this international, virtual meeting based at the University of California, San Francisco, to help improve innovations in diagnostics and therapeutics for rare diseases,

We thank you for your engagement.

Presentations



ABOVE By Torsten Wittman, PhD: Cytoskeleton in growth factor stimulated human keratinocytes.

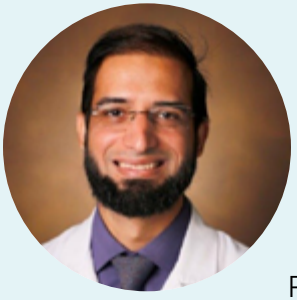
9:15 – 9:45am: Opening Address: Rare Disease Therapeutics - Soup to Nuts

Tiina Urv, PhD is the Program Director for the Rare Diseases Clinical Research Network (RDCRN), a multi-disciplinary international program in the Office of Rare Diseases Research (ORDR) at the NIH. As the lead for the RDCRN program, Tiina collaborates with 10 NIH Institutes to manage 22 consortia and a central Data Management Coordinating Center. The RDCRN has more than 200 participating sites in 17 countries and more than 100 Patient Advocacy Groups as research partners and conducts research on about 200 rare diseases. Before joining the ORDR, Urv was a program director in the Division of Clinical Innovation where she provided stewardship for multiple Clinical and Translational Science Awards Program hubs and worked with the Trial Innovation Network as well as NCATS' ORDR.

9:45 – 10:15am: SPLIS: From Gene Discovery to Gene Therapy

Julie Saba, MD, PhD is a pediatric oncologist at the University of California San Francisco where she is Professor of Pediatrics and holds the John and Edna Beck Chair of Cancer Research. Dr. Saba attended The Johns Hopkins University, University of Maryland medical school, and completed her residency, fellowship and obtained a PhD in Cell Biology at Duke Medical Center. During her graduate studies, while searching for new cancer-related genes, Dr. Saba identified the first sphingosine phosphate lyase gene using a genetic screen in budding yeast. She later found the human gene encoding this enzyme, called SGPL1, which is critical for metabolism of sphingolipids. In 2017, exactly 20 years after discovering this gene, she and her colleagues discovered SPLIS—an inherited disorder of sphingolipid metabolism caused by inactivating mutations in SGPL1. Dr. Saba is now focused on understanding the pathologic basis of SPLIS and finding a cure for this devastating disease.

10:15 – 10:45am: Patient Story: Living with SPLIS



Ehtesham Khalid, MD is a Senior Consultant, Neurophysician, and Department Head at Mukhtar A. Sheikh Hospital in Multan, Pakistan. Additionally, Dr. Khalid has served as an Adjunct Clinical Instructor of Neurology at Vanderbilt University where he completed a Neuromuscular Fellowship and Neurology Residency. Dr. Khalid areas of expertise include neuromuscular disorders, neuro-muscular junction problems, muscle diseases, polyneuropathies, and autonomic disorders among others.

10:45 – 11:15am: State of the Art Diagnostics

Stephen F. Kingsmore, MD, DSc is President/CEO of Rady Children's Institute for Genomic Medicine where he leads a multi-disciplinary team of scientists, physicians and researchers who are pioneering the use of rapid Whole Genome Sequencing to enable precise diagnoses for critically-ill newborns. Before being selected to lead RCIGM, he was the Dee Lyons/Missouri Endowed Chair in Genomic Medicine at the University of Missouri-Kansas City School of Medicine and Director of the Center for Pediatric Genomic Medicine at Children's Mercy Hospital, Kansas City. Dr. Kingsmore received MB ChB BAO BSc and DSc degrees from the Queen's University of Belfast. He trained in clinical immunology in Northern Ireland and did residency in internal medicine and fellowship in rheumatology at Duke University Medical Center.

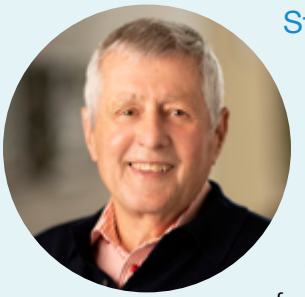
11:15 – 11:45am: Negotiating the Challenges of Therapeutics Discovery for Rare Diseases

Emil D Kakkis, MD, PhD

A medical geneticist by training, Dr. Kakkis is a pioneer in the development and commercialization of treatments for rare diseases. He is the founder and CEO at Ultragenyx, a biopharmaceutical company developing novel products for the treatment of rare and ultra-rare diseases.

Before founding Ultragenyx in 2010, Dr. Kakkis guided and contributed to the development of multiple approved rare disease products as chief medical officer at BioMarin Pharmaceutical. Dr. Kakkis also founded and serves on the Board of EveryLife Foundation for Rare Diseases, a non-profit dedicated to accelerating innovation for rare diseases. Dr. Kakkis has been recognized for his leadership by Biotechnology Innovation Organization (BIO), the National MPS Society, WORLDSymposium, and California Life Sciences.

11:45 – 12:15pm: n-Lorem – Meeting the Needs of n-1 Patients One Patient at a Time



Stanley Crooke, MD, PhD is a founder, chairman and chief executive officer of n-Lorem, a nonprofit foundation focused on providing treatments for patients with ultra-rare diseases. Prior to n-Lorem, Dr. Crooke founded Ionis Pharmaceuticals. During his tenure at Ionis, he has led the scientific development of a new platform for drug discovery, antisense technology and engineered the creation of one of the largest and more advanced development pipelines in the biotechnology industry. Early in Dr. Crooke's career, he led the creation of the first broad anticancer program in the industry at Bristol-Myers, bringing numerous anticancer drugs to the market. He went on to be responsible for worldwide R&D at SmithKline Beecham. Dr. Crooke has received several awards including the American Chemical Society's E.B. Hershberg Award for Important Discoveries in Medicinally Active Substances, the Lifetime Achievement Award presented by the Oligonucleotide Therapeutics Society, and the Scrip Lifetime Achievement Award. Dr. Crooke received his MD and PhD degrees at Baylor College of Medicine. In 2021, Dr. Crooke was named Distinguished Alumnus of both Baylor College of Medicine's Graduate and Medical schools.

12:15 – 1:00pm: Rare Voices: A Concert of Science

Featuring musicians and poets with a rare disease

"Sparsh" Shah (a.k.a Purhythm) is an 18-year-old prodigy, singer, songwriter, rapper, inspirational speaker, and philanthropist. He was born with an incurable disease called Osteogenesis Imperfecta, which makes his bones extremely fragile/brittle, due to which even a hard handshake can break his hand. He cannot bear his own weight and hence cannot walk or run like other normal kids. He has had more than 140 fractures and multiple rod surgeries so far and God knows how many more are yet to come. But he has been gifted with a divine voice and a brilliant mind. Sparsh has won prizes in 9 singing competitions and done over 300 live performances in 9 countries and at prestigious venues such as Madison Square Garden, Radio City Music Hall, and the National Center of Performing Arts, just to name a few. As a performer he has helped raise over $1.5 million dollars for various non-profit organizations supporting children and adults in need. Sparsh's vision in life is to inspire and touch everyone's hearts around the world.

Child Life Poetry Workshop

Senior Associate Director, Catalyst Program and InVent Fund Roopa Ramamoorthi, PhD, and Child Life Special Events Coordinator Elyse Cann with UCSF Benioff Children's Hospitals, initiated a poetry workshop for adolescents with rare diseases for them to develop language to express their fears, their courage, and sometimes their triumphs in the on-going fight against their disease. By bearing witness to their struggle, they hope to bring a greater understanding of life in the shadow of disease.

1:00 – 2:20pm: Hope for a Cure: Treating Sickle Cell Disease with CRISPR

Featuring Brooklyn Haynes

Alameda County native Brooklyn Haynes was diagnosed with sickle cell disease at birth. From the age of two-months she has been treated for this disease at UCSF Benioff Children's Hospital, Oakland. For almost three decades she has lived with the on-going cycles of disease related pain.

Curative Therapies for Sickle Cell Disease

Mark C. Walters, MD is the Jordan Family Director of the Blood and Marrow Transplantation Program at UCSF Benioff Children's Hospital, Oakland, and Professor of Pediatrics at UCSF School of Medicine. He is Program Director of the California Institute of Regenerative Medicine alpha stem cell clinic at UCSF. Dr. Walters received his AB with honors in Genetics from the University of California, Berkeley and his MD from the University of California, San Diego. He completed pediatric residency training at the University of Washington and hematology/oncology fellowship training at the University of Washington and the Fred Hutchinson Cancer Research Center in Seattle. He has been active in cooperative clinical transplantation trials and has led several NIH-supported investigations of hematopoietic cell transplantation for sickle cell anemia and thalassemia. He has a research interest in extending transplantation to young adults with hemoglobin disorders and other novel cellular therapies for hemoglobin disorders. Currently, research interests are focused on genomic editing and gene addition therapies as a strategy to extend curative therapy in all patients who inherit a clinically significant hemoglobinopathy.

Marsha J. Treadwell, PhD is a Professor of Pediatrics at UCSF in the Division of Hematology and is the Jordan Fund Endowed Chair at UCSF Benioff Children's Hospital Oakland. She is Co-Chair of the Diversity, Equity, Inclusion and Anti-Racism Council for the UCSF Benioff Children's Hospitals. Dr. Treadwell's research integrates physical, behavioral and psychological processes, allowing for the identification of risk and resiliency factors and the development of more effective interventions for populations made vulnerable by systems in both high and low resource settings. Dr. Treadwell has expertise with measurement development, community engagement, conducting health care quality improvement projects, evaluating programs and assessing health-related quality of life for individuals and communities. Dr. Treadwell is Co- PI and Regional Director for the Pacific Sickle Cell Regional Collaborative. She is co-PI and Director for a Sickle Cell Disease Implementation Consortium (SCDIC) site, funded by the National Heart, Lung and Blood Institute. The goal of the SCDIC is to use implementation science to improve outcomes for adolescents and adults with sickle cell disease by improving understanding of barriers to care and utilization of evidence-based guidelines. Dr. Treadwell received her doctorate in clinical child psychology from the University of Washington in Seattle, WA and advanced training in clinical research from UCSF.

UCSF Benioff Children's Hospital Oakland has one of the premiere centers in the nation for sickle cell disease treatment and has been at the forefront of advancing the research for better treatments and a cure for over 45 years. A sickle cell-CRISPR gene therapy clinical trial – a partnership with UCSF, UCLA and UC Berkeley – will be the first human trial using CRISPR gene editing technology to replace mutated blood cells with edited cells – a potential cure for sickle cell.

2:20 – 2:55pm: Defining a New Future for People with Rare Diseases

A globally-experienced industry leader, physician researcher, and biopharmaceuticals expert, Amrit Ray, MD, MBA's passion is advancing medical breakthroughs and championing healthcare access for patients. He serves as a board director and independent advisor to biopharmaceutical companies. Dr.Ray has held three C-level roles at major companies including as Global President of R&D at Pfizer, and as Chief Medical Officer at Johnson & Johnson pharmaceuticals. Most recently, Dr.Ray was an Executive Leadership Team member at Pfizer-Upjohn overseeing all aspects of R&D worldwide and the company's most senior decision-maker on patient matters. He has led teams of up to 3,500 professionals in 90 countries to advance pipelines and access for innovative medicines in Oncology, Neuroscience and Cardio-Metabolism, and sponsored >100 pioneering inter-sectoral healthcare partnerships. He is currently the Chief Patient Officer at Biohaven Pharmaceuticals

Our Organizers

Charles Hart, PhD is the director of the Catalyst program at the University of California, San Francisco.

charles.hart@ucsf.edu
innovation.ucsf.edu

Jennifer Janci, PhD is the program director of Diagnostics and Digital Health for the Catalyst Program, and also the Director of the UCSF LaunchPad.

jennifer.janci@ucsf.edu

Nathaniel Prorok, MHA is the senior program manager of the Catalyst program at the University of California, San Francisco. He runs the Digital Health program and is Co-Director of LaunchPad.

nathaniel.prorok
@ucsf.edu

James Levine, MD, PhD is President of Fondation Ipsen. He is also the Director of the Rare Disease Institute at Fondation Ipsen.

james.levine@ipsen.com
fondation-ipsen.org

Yannick Tanguy, PhD is the project manager at Foundation Ipsen.

yannick.tanguy
@ipsen.com

Roopa Ramamoorthi, PhD is the senior associate director for the Catalyst Program and the InVent Fund at the University of California, San Francisco.

roopa.ramamoorthi
@ucsf.edu

From Our Organizers

Translational technology development and commercialization can be a complex process because every project is unique. UCSF Innovation Ventures is dedicated to helping the University of California community navigate those complexities by working up-close with our research faculty and promising students to really understand their science and its potential. The Catalyst Program is UCSF's translational accelerator focused on advancing UCSF discoveries with clinical impact and commercial potential. The Catalyst Program aims to foster academic and industry collaboration as well as enhance education in early translational research and preparation for future entrepreneurship. The centerpiece of the program is the Catalyst Awards, our program that provides both industry advisor mentorship, and seed funding to UCSF investigators with promising projects. The Catalyst Awards are focused on the development of therapeutics, diagnostics, medical devices, and digital health.

Charles Hart, PhD
Director of Catalyst Program

innovation.ucsf.edu

For several decades, Fondation Ipsen has been working to improve the public's living conditions by disseminating scientific knowledge to the general public and promoting exchanges within the scientific community. Since its creation in 1983 under the aegis of the Fondation de France, Fondation Ipsen has contributed to major advances in biological and medical research by organizing numerous scientific conferences.

Today, our activities have been refocused on rare diseases in order to advance knowledge and improve the health and daily lives of patients and their families. Fondation Ipsen's ultimate ambition is to improve the early detection and diagnosis of rare diseases.

James Levine, MD, PhD
President of Fondation Ipsen

Director of the Rare Disease Institute at Fondation Ipsen

fondation-ipsen.org

Blood is so accessible and yet I lack its cells
If only people knew the struggles
That the disease entials
I've been laughed at, called names
And even called a liar
But nothing is worse than when you lose your fire
That beautiful fan that blows gently on your flame
Was taken away from me
But do not dismay
I plan to come back brighter.

Michael D.
Age 16
Aplastic Anemia patient

UCSF Innovation Ventures
innovation.ucsf.edu
innovation@ucsf.edu

Foundation Ipsen
fondation-ipsen.org