



FRONTIERS  
IN PEDIATRIC  
GENOMIC  
MEDICINE  
CONFERENCE

Virtual Conference

Sept. 24-25, 2020  
9am-4:30pm PDT

Rady  
Children's  
Institute  
Genomic Medicine

UC San Diego  
SCHOOL OF MEDICINE

# Welcome

## Welcome to 2020 Frontiers in Pediatric Genomic Medicine

Dr. Stephen Kingsmore and the team at Rady Children's Institute for Genomic Medicine welcome to the Fifth Annual Frontiers in Pediatric Genomic Medicine conference.

We have a content-rich agenda featuring leaders in Rapid Precision Medicine™ from children's hospitals and research centers around the world. You'll hear about their innovations, case studies and lessons on how multi-state and international genomic alliances are transforming patient care. You won't want to miss the sessions with patient parents.

For additional learning and networking, visit the Exhibit Hall and Poster Sessions. And drop into the Lounge to chat with presenters and other attendees. There's a lot to do. Enjoy!

# Accreditation

## UC San Diego SCHOOL OF MEDICINE

### Continuing Medical Education (CME) for Physicians

The University of California, San Diego School of Medicine is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians.

- The University of California, San Diego School of Medicine designates this Internet live activity for a maximum of **11.0 AMA PRA Category 1 Credits™**. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

### Cultural and Linguistic Competency:

California Assembly Bill 1195 requires continuing medical education activities with patient care components to include curriculum in the subjects of cultural and linguistic competency. It is the intent of the bill, which went into effect on July 1, 2006, to encourage physicians and surgeons, CME providers in the State of California, and the Accreditation Council for Continuing Medical Education to meet the cultural and linguistic concerns of a diverse patient population through appropriate professional development. The planners, speakers and authors of this CME activity have been encouraged to address issues relevant in their topic area. In addition, a variety of resources are available that address cultural and linguistic competency, some of which are included in your syllabus or handout materials. Additional resources and information about AB1195 can be found on our website at [cpd.ucsd.edu](http://cpd.ucsd.edu)

## Disclosure Statement

It is the policy of the University of California, San Diego School of Medicine to ensure balance, independence, objectivity and scientific rigor. All persons involved in the selection, development and presentation of content are required to disclose any real or apparent conflicts of interest. All conflicts of interest will be resolved prior to an educational activity being delivered to learners through one of the following mechanisms 1) altering the financial relationship with the commercial interest, 2) altering the individual's control over CME content about the products or services of the commercial interest, and/or 3) validating the activity content through independent peer review. All persons are also required to disclose any discussions of off label/unapproved uses of drugs or devices. Persons who refuse or fail to disclose are disqualified from participating in the CME activity. Participants will be asked to evaluate whether the speaker's outside interests reflect a possible bias in the planning or presentation of the activity. This information is used to plan future activities.

The 2020 Frontiers in Pediatric Genomic Medicine Conference speakers listed below have no relevant financial relationships to disclose:

Stephen Kingsmore	Erica Sanford	Art D'Harlingue
Shimul Chowdhury	Kobayashi	Jason Knight
Nathaly Sweeney	Anthony Magit	Fiona Lynch
Caleb Bupp	Shareef Nahas	Andrea Scheurer
Elsebet Oestergaard	Joseph Gleeson	Zornitza Stark
Sophia Ceulemans	Jerica Lenberg	
Katarzyna Ellsworth	Danny Miller	

The following conference faculty member discloses two commercial relationships:

Name	Name of Commercial Interest	Nature of Relevant Relationship
David Dimmock, MD	Audentes	Consultant
	Biomarin	Consultant

- The CME staff, meeting planners, planning committee and CME committee reviewers do not have any relevant financial relationships to disclose.
- This educational activity may contain discussion of unlabeled and/or investigational uses of agents that are not approved by the FDA. Please consult the prescribing information for each product.
- The views and opinions expressed in this activity are those of the faculty and do not necessarily reflect the views of the University of California, San Diego School of Medicine.

## Other Accrediting Agencies

### **Continuing Education Unit (CEU) for Genetic Counselors**

The National Society of Genetic Counselors (NSGC) has authorized the Rady Children's Institute for Genomic Medicine to offer up to 1 CEU or 10 Category 1 contact hours for the activity RCIGM Frontiers in Pediatric Genomic Medicine Conference 2020. The American Board of Genetic Counseling (ABGC) will accept CEUs earned at this program for the purposes of genetic counselor certification and recertification.

### **Continuing Education for Nurses**

Rady Children's Hospital-San Diego is a Continuing Education Provider approved by the California Board of Registered Nursing, BRN Provider #00111 for 10 (ten) total continuing education contact hours.

# About this Conference

## Audience

This event is for a multidisciplinary professional audience from the fields of Cellular & Molecular Medicine, Neurosciences, Pediatrics, Genetics, Neonatology, and Cardiology.

## Course Description

The annual Frontiers in Pediatric Genomic Medicine Conference brings together leaders in research and healthcare to explore advances in translating next generation molecular diagnostics into targeted treatment for critically-ill patients hospitalized in neonatal, pediatric and cardiac intensive care. The 2020 conference theme is "Integrating Genome Medicine Across Systems". Presentations will focus on the topics of:

- Next Generation Diagnostics
- Improving NICU/PICU Implementation
- Quality Improvement
- Multi-site Genomic Alliance

## Learning Objectives

At the completion of the conference, participants should be able to:

- Describe the principles of genomic medicine and whole genome sequencing
- Discuss the uses, benefits, and limitations of genomic testing technologies
- Recognize the implications of implementing genomic medicine in NICUs and PICUs
- Recognize when and how to refer patients for whole genome sequencing
- Identify tools and other practical resources for implementation of whole genome sequencing
- Determine early ascertainment for patients who may benefit from whole genome sequencing
- Recognize how to collaborate with an interdisciplinary team of health professionals

## Needs Assessment and Practice Gap

Genomic medicine is a new approach to healthcare that utilizes advanced technologies to accurately identify specific variations in human genes that result in disease and mortality. It is a large, somewhat diffuse field of study that encompasses diverse disciplines. While the practice of whole genomic sequencing could be a key technology leveraged by NICUs and PICUs to assist in making treatment decisions for acutely-ill babies, a pediatric genomic medicine knowledge gap currently exists among many healthcare providers in the United States.

Genomic medicine is a relatively recent development in the healthcare sphere introduced only 17 years ago with the decoding of the first human genome. Incorporating genomics into clinical decision making has only been feasible with the advent of improved technologies allowing cost-effective genome analysis. Genomic diagnostic and decision-making tools are being developed at a higher rate now than ever before. Unfortunately, didactic coursework in healthcare curricula that prepares healthcare professionals to utilize genomic medicine has been slow to catch up. This has left many practicing healthcare professionals in the workforce without formal training in pediatric genomic medicine, which is hampering the adoption of genomics in the care of acutely-ill children.

Weil, A. R. (2018). Precision Medicine.

# Day One

Thursday, September 24, 2020 | 9:00 am to 4:30 pm PDT

## Morning Focus: Next Generation Diagnostics

<b>9 am</b>	Welcoming Comments: Integrating Genomic Medicine Across Systems	Stephen Kingsmore, MD, DSc President and CEO, RCIGM
<b>9:30 am</b>	Expanding the Impact of Rapid Diagnostics	David Dimmock, MD Senior Medical Director, RCIGM; Associate Physician, UC San Diego Health
<b>10:30 am</b>	Break – Exhibit Hall Open	
<b>11:00 am</b>	New Findings, New Future  WT1 - Management Impact	<b>Facilitator:</b>  Shimul Chowdhury, PhD, FACMG, CGMB Clinical Laboratory Director, RCIGM  Erica Sanford Kobayashi, MD Pediatric Intensivist, Rady Children’s Hospital and Cedars Sinai Medical Center; Clinical Investigator RCIGM  Katarzyna Ellsworth PhD, FACMG Associate Laboratory Director, RCIGM  Jerica Lenberg, MS, CGC Licensed Certified Genetic Counselor, RCIGM
<b>Noon</b>	Exhibit Hall & Poster Presentations	



# Day One

Thursday, September 24, 2020 | 9:00 am to 4:30 pm PDT

## Afternoon Focus: Next Generation Diagnostics

<b>1 pm</b>	Using Clinical rWGS: Lessons Learned From a Quality Improvement Initiative	Shareef Nahas, PhD, FACMG, CGMB Senior Director of Clinical Operations  Jerica Lenberg, MS, CGC Licensed Certified Genetic Counselor, RCIGM
<b>1:45 pm</b>	The Impact of rapid Whole Genome Sequencing™ on Medical Management and Resource Utilization in the Care of Critically-ill Infants with Congenital Heart Disease	Nathaly Sweeney, MD Pediatric Cardiologist, Rady Children's Hospital; Assoc. Professor, UC San Diego School of Medicine; Clinical Investigator, RCIGM
<b>2:30 pm</b>	Break – Exhibit Hall Open	
<b>3:00 pm</b>	Molecular Diagnosis in Recessive Pediatric Neurogenetic Disease Can Help Reduce Disease Recurrence in Families	Joseph Gleeson, MD, PhD Director Neurodevelopmental Genetics, RCIGM; Professor Neuroscience and Pediatrics, UC San Diego School of Medicine  Anthony Magit, MD Chief of Physician Integration, RCHSD; Professor Neuroscience and Pediatrics, UC San Diego School of Medicine  Sophia Ceulemans, MS, CGC Licensed Certified Genetic Counselor, RCHSD
<b>4:00-4:30 pm</b>	Open Q&A with Stephen Kingsmore	Stephen Kingsmore, MD, DSc President & CEO, RCIGM

# Day Two

Friday, September 25, 2020 | 9 am to 4:30 pm PDT

## Morning Focus: Multi-Site Collaborations

<b>8 am</b>	Exhibit Hall Opens	
<b>9:00 am</b>	Opening Remarks with Dr. Patrick Frias, President & CEO, Rady Children's Hospital-San Diego	Stephen Kingsmore, MD, DSc President and CEO, RCIGM
	Baby Nathan video	
<b>9:30 am</b>	Project Baby Bear – The Final Report	David Dimmock, MD Senior Medical Director, RCIGM
<b>10:30 am</b>	Break – Exhibit Hall Open	
<b>11:00 am</b>	Lending a Hand – Telemedicine and Project Baby Deer in Michigan	Caleb Bupp, MD, FACMG Section Chief, Medical Genetics and Genomics, Spectrum Health – Helen DeVos Children's Hospital  Andrea Scheurer, MD Vice President for Research and Education, Southwest Michigan Neonatology, P.C.
<b>11:30 am</b>	Before and After: Our Family's Search for Answers in Rare Disease	Danny Miller Founder and CEO, MEPAN Foundation
<b>Noon</b>	Exhibit Hall & Poster Presentations	

## Afternoon Focus: International Genomic Alliances

<b>1:00 pm</b>	Introduction	Stephen Kingsmore, MD, DSc President and CEO, RCIGM
<b>1:15 pm</b>	Targeted Genome Sequencing for Potentially Treatable Conditions in Critically Ill Patients	Elsebet Oestergaard, MD Chief Physician, Associate Professor Copenhagen University Hospital Rigshospitalet
<b>2:00 pm</b>	Break – Exhibit Hall Open	
<b>2:30 pm</b>	Implementing rapid Genomics at National Level: The Australian Experience (7:30am, Saturday, September 26)	Zornitza Stark, BM BCh MA DM, FRACP Clinical Geneticist, Victorian Clinical Genetics Services & Clinical Research Fellow, Australian Genomics Health Alliance
<b>3:30 pm</b>	Genetic Counseling and the Parent Experience in Rapid Acute Care Genomics	Fiona Lynch, BSc MGenCouns PhD Candidate, Murdoch Children’s Research Institute
<b>4:00-4:30 pm</b>	Wrap Up	Stephen Kingsmore, MD, DSc President & CEO, RCIGM

# Faculty Listing

## Planning Committee

### **Stephen Kingsmore, MD, DSc**

President and Chief Executive Officer  
Rady Children's Institute for  
Genomic Medicine

### **Karen Garman, EdD, MAPP, BCC**

Director of Education and Engagement,  
Rady Children's Institute  
for Genomic Medicine

## UC San Diego Faculty

### **David Dimmock, MD**

Senior Medical Director,  
Rady Children's Institute for Genomic  
Medicine; Associate Physician,  
UC San Diego Health

### **Joseph Gleeson, MD, PhD**

Director, Neurodevelopmental Genetics,  
Rady Children's Institute for Genomic  
Medicine;  
Professor Neuroscience and Pediatrics,  
UC San Diego School of Medicine

### **Erica Sanford Kobayashi, MD**

Pediatric Intensivist,  
Rady Children's Hospital-San Diego;  
Cedars Sinai Medical Center;  
Associate Physician, UC San Diego  
Health;  
Clinical Investigator,  
Rady Children's Institute for  
Genomic Medicine

### **Anthony Magit, MD**

Chief of Physician Integration  
Rady Children's Hospital-San Diego;  
Clinical Professor,  
UC San Diego School of Medicine

### **Nathaly Sweeney, MD**

Pediatric Cardiologist  
Rady Children's Hospital;  
Assistant Professor, UC San Diego  
School of Medicine;  
Clinical Investigator, Rady Children's  
Institute for Genomic Medicine

## Visiting Faculty

### **Caleb Bupp, MD, FACMG**

Section Chief, Medical Genetics and Genomics  
Spectrum Health – Helen De Vos Children’s Hospital

### **Sophia Ceulemans, MS, CGC**

Licensed Clinical Genetic Counselor  
Rady Children’s Hospital of San Diego

### **Shimul Chowdhury, PhD, FACMG, CGMB**

Clinical Laboratory Director  
Rady Children’s Institute for Genomic Medicine

### **Art D’Harlingue, MD**

Director/Chief Neonatology  
UCSF Benioff Children’s Hospital Oakland

### **Katarzyna Ellsworth, PhD, FACMG**

Associate Lab Director  
Rady Children’s Institute for Genomic Medicine

### **Jason Knight, MD**

Division Chief – Critical Care, Medical Director, PICU  
CHOC Children’s Hospital

### **Jerica Lenberg, MS, CGC**

Licensed Certified Genetic Counselor  
Rady Children’s Institute for Genomic Medicine

### **Fiona Lynch, BSc, MGenCouns**

PhD Candidate, Graduate Researcher  
Murdoch Children’s Research Institute

### **Danny Miller**

Founder and CEO  
MEPAN Foundation

### **Andrea Scheurer, MD**

Vice President for Research and Education  
Southwest Michigan Neonatology, P.C.

### **Shareef Nahas, PhD, FACMG, CGMB**

Senior Director of Clinical Operations  
Rady Children’s Institute for Genomic Medicine

### **Elsebet Oestergaard, MD**

Chief Physician, Associate Professor  
Copenhagen University Hospital Rigshospitalet

### **Zornitza Stark, BM, BCh ,MA, DM, FRACP**

Clinical Geneticist, Victorian Clinical Genetics Services & Clinical Research Fellow  
Australian Genomics Health Alliance

# Exhibitor Acknowledgement

Rady Children's Institute for Genomic Medicine would like to acknowledge the following companies and organizations for exhibiting at this conference:

- Illumina
- Alexion
- Fabric Genomics
- L7 Informatics
- March of Dimes
- Rady Children's Hospital-San Diego

NOTE: Cancellations must be made at least 2 weeks prior to the conference date in order to receive a full refund. After this date, no refunds will be granted, and the fee is non-transferable. Rady Children's Institute for Genomic Medicine reserves the right to cancel this course, in which case a full refund will be given.