

**FRONTIERS  
IN PEDIATRIC  
GENOMIC  
MEDICINE**



**HYBRID EVENT**

May 1 –2, 2024  
8am–4:30pm PDT  
Scripps Seaside Forum  
8610 Kennel Way  
La Jolla, CA 92037



## **WELCOME**

### **Welcome to 2024 Frontiers in Pediatric Genomic Medicine**

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

The annual Frontiers in Pediatric Genomic Medicine Conference brings leaders in research, industry and healthcare together to explore advances in translating genomic diagnostics into targeted treatment for patients in neonatal and pediatric intensive care. This year's program will focus on the topics of Rapid Whole Genome Sequencing in Behavioral Health, Neuro-Oncology and Newborn Genomic Sequencing.

You won't want to miss the sessions with patient parents. For additional learning and networking, visit the Exhibit Hall and drop into the Lounge to chat with presenters and other attendees. There's a lot to do. Enjoy!

## ACCREDITATION



### **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 live activity for a maximum of **10.00 AMA PRA Category 1 Credits™**. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

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## Disclosure Statement

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Faculty Member Name	Role in Activity	Name of Ineligible Company(ies)/Nature of Relationship(s)
Stephen F Kingsmore, MBBS, FRCPATH	Course Director	Membership on Advisory Committees or Review Panels, Board Membership, etc.-Springer-Nature Membership on Advisory Committees or Review Panels, Board Membership, etc.- Grant or research support-Alexion Pharmaceuticals Grant or research support-Amgen, Inc. Grant or research support-Chiesi Farmaceutici Grant or research support-Fabric Genomics Grant or research support-Genomenon Inc. Grant or research support-Helix Inc. Grant or research support-Horizon Therapeutics Grant or research support-Illumina Inc.  Grant or research support-Ionis Pharmaceuticals Grant or research support-Inozyme Pharma Grant or research support-

		Mahzi Therapeutics Grant or research support-Nest Genomics Grant or research support-Orchard Therapeutics Grant or research support-Rocket Pharma Grant or research support-Sanofi S.A. Grant or research support-Sarepta Therapeutics Grant or research support-Sentiny  Therapeutics Grant or research support-TileDB Inc. Grant or research support-Travere Therapeutics Inc. Grant or research support-Ultragenyx
Jonathan Douglas Santoro, MD	Faculty	Consulting Fee-UCB Consulting Fee-Cycle Pharmaceuticals Consulting Fee-TG Therapeutics
Diana Azzam, PhD	Faculty	Scientific Advisor - First Ascent Biomedical
Rob Knight, PhD	Faculty	Advisor-Biota technology Advisor-GenCirq, Inc.  Advisor-DayTwo, Ltd. Consulting Fee-Cybele Microbiome, Inc.  Consulting Fee-BiomeSense,Inc.  Advisor-Micronoma, Inc.
Jennifer H Yang, MD	Faculty	Grant or research support-Astellas (Any division) Grant or research support-Stealth

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## **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 leaders in research, industry and healthcare together to explore advances in translating genomic diagnostics into targeted treatment for patients in neonatal and pediatric intensive care. This year's program will focus on the topics of Rapid Whole Genome Sequencing in Behavioral Health, Neuro-Oncology and Newborn Genomic Sequencing.

Day 1:

- MORNING FOCUS: BeginNGS®
- AFTERNOON FOCUS: Behavioral Health

Day 2:

- MORNING FOCUS: RCIGM Case Reports
- AFTERNOON FOCUS: Neuro-Oncology

### **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 healthcare approach that utilizes various technologies to accurately identify patients with 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 of acutely-ill babies, a pediatric genomic medicine knowledge gap currently exists between many healthcare providers in the United States.

Genomic medicine is a relatively recent development in the healthcare sphere introduced only 15 years ago with the completion of the first human genome. Incorporating genomics into clinical decision making has only been feasible with the advent of improved technologies allowing cost-effective genomic 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.

# AGENDA

DAY ONE	Wednesday, May 1, 2024	8:00 am to 4:30 pm PDT
<b>MORNING FOCUS: BeginNGS®</b>		
8:00 am	Registration/Exhibit Hall/Breakfast	
9:00 am	Welcome/Keynote	Stephen Kingsmore, MD, DSc President & CEO, Rady Children’s Institute for Genomic Medicine (RCIGM)
9:45 am	Lightning Talks: Leveraging Diverse Population Databases to Scale Whole Genome Newborn Sequencing BeginNGS Prospective Trial Update: Beyond the Beginning	Meredith Wright, PhD Genomics Integration Scientist, RCIGM Rebecca Reimers, MD/MPH, FACOG, FACMG Maternal-Fetal Medicine & Clinical Geneticist, San Diego Perinatal Center & RCIGM/RCHSD
10:15 am	BREAK – Exhibit Hall Open	
10:45 am	BeginNGS Consortium Panel: Beginning to Scale – Barriers to Success and Opportunities to Innovate	Terri Finkel, MD, PhD Professor and Associate Chair, Department of Pediatrics, College of Medicine, University of Tennessee Health Science Center Thomas May, PhD Floyd and Judy Rogers Endowed Professor, Washington State University, Vancouver, Elson S. Floyd College of Medicine Jessie Conta, MS Laboratory Stewardship Consultant, PLUGS Moderator: Kathy Lynch, Esq. Board Member, Rady Children’s Institute for Genomic Medicine
11:15 am	Family Panel	RCIGM Patient Family Moderator: Jerica Lenberg, MS, LCGC Lead Genetic Counselor, RCIGM
Noon	Lunch & Networking	
<b>AFTERNOON FOCUS: Behavioral Health</b>		

1:00 pm	Keynote: Microbiome and Its Impact on Adolescent Treatments for Depression	Rob Knight, PhD Professor of Pediatrics and Computer Science & Engineering, UC San Diego / Wolfe Family Endowed Chair in Microbiome Research, Rady Children's Hospital San Diego (RCHSD)
1:45 pm	Case: Genomics-Informed Pediatric Precision Mental Health	Aaron Besterman, MD Assistant Clinical Professor of Psychiatry, Department of Psychiatry, UC San Diego / Clinical Investigator, RCIGM/RCHSD
2:15 pm	BREAK – Exhibit Hall Open	
2:45 pm	Lightning Talk: Mitochondrial Disorders and Genetic Mimickers of Neuroinflammatory Disorders	Jennifer Yang, MD Assistant Clinical Professor of Neurosciences, Division of Pediatric Neurology at UC San Diego / RCIGM / RCHSD
3:10 pm	Lightning Talk: Neuroinflammation in Down Syndrome Regression Disorder	Jonathan Santoro, MD Director, Neuroimmunology & Demyelinating Disorders Program, Director of Research, Neurologic Institute at Children's Hospital Los Angeles / Associate Professor of Neurology and Pediatrics, USC Keck School of Medicine
3:35 pm	Clinical Panel Discussion	<ul style="list-style-type: none"> <li>• Aaron Besterman, MD, UC San Diego / RCIGM</li> <li>• Rob Knight, PhD, UC San Diego / RCHSD</li> <li>• Jennifer Yang, MD, UC San Diego / RCIGM</li> <li>• Jonathan Santoro, MD, USC Keck School of Medicine</li> </ul>
4:00 pm	Wrap Up of Day 1	Stephen Kingsmore, MD, DSc President & CEO, RCIGM
4:30 – 7:00 pm	Reception/Networking	Heavy hors d'oeuvres and cocktails

DAY TWO		Thursday, May 2, 2024	8:00 am to 4:30 pm PDT
MORNING FOCUS: RCIGM Case Reports			
8:00 am	Registration/Exhibit Hall/Breakfast		
9:00 am	Expanding the Breadth of Clinical Rapid WGS Test at RCIGM	Katarzyna Ellsworth, PhD, FACMG, CGMB Sr. Director of Clinical Operations, RCIGM	
	Identification of Trinucleotide Repeat Expansions in DMPK Using rapid Whole Genome Sequencing	Lucia Guidugli, PhD, FACMG, CGMB Clinical Lab Director, RCIGM Jennifer Schleit, PhD, FACMG Lab Director, RCIGM	
9:30 am	Lightning Talks: Beyond the Clinical urWGS Result: Integration of a Translational Research Program	Whitney Thompson, MD, Mayo Clinic Eric Klee, PhD, Mayo Clinic	
	Long Reads	Matthew Bainbridge, PhD, RCIGM	
10:15 am	BREAK – Exhibit Hall Open		
10:45 am	Keynote: The Impact of rWGS® in RCHSD’s NICU & PICU	Gail Knight, MD Sr. Vice President & Chief Medical Officer, RCHSD	
11:15 am	Clinician Panel: Beyond the NICU	Panelists: <ul style="list-style-type: none"> <li>Paige Hazelton, MS, Mayo Clinic</li> <li>Joseph Fakhoury, MD, Bronson Children’s Hospital</li> <li>Sabrina Jenkins, MD, Neonatologist, University of Utah / Primary Children’s Hospital</li> <li>Kristen Suhrie, MD, Riley Children’s Health</li> </ul> Moderator: Rebecca Reimers, MD/MPH, FACOG, FACMG Maternal-Fetal Medicine & Clinical Geneticist, San Diego Perinatal Center & RCIGM/RCHSD	
Noon	Lunch & Networking		
AFTERNOON FOCUS: Neuro-Oncology			
1:00 pm	Keynote: Functional Precision Medicine to Guide Individualized Treatments for Relapsed/Refractory Pediatric Cancers	Diana Azzam, PhD Assistant Professor, Lab Director, Environmental Health Sciences, Florida International University	
1:45 pm	Case: Molecular Tumor Board (MTB): Advancing Precision Medicine for Children	Lukas Chavez, PhD Scientific Director, RCHSD Pediatric Neuro-Oncology	

	with Brain Tumors  A Light in the Dark: Using Molecular Profiling to Unveil Novel Treatments for Pediatric Neuro-Oncology Patients	Molecular Tumor Board / RCIGM / Sanford Burnham Prebys Medical Discovery Institute / UC San Diego  Megan Paul, MD Clinical Co-lead, RCHSD Pediatric Neuro-Oncology Molecular Tumor Board
2:15 pm	BREAK – Exhibit Hall Open	
2:45 pm	Lightning Talk: Leveraging Pediatric Neuro-oncology Genomics for Therapeutic Benefit	Parent / Father of patient Praveen B Raju, MD, PhD, Associate Physician Diplomate, Neurosciences, UC San Diego
3:15 pm	Panel: Challenges and Opportunities of Genomic Medicine in Pediatric Neuro-oncology	Panelists: <ul style="list-style-type: none"> <li>• Parent</li> <li>• Diana Azzam, PhD</li> <li>• Megan Paul, MD, RCIGM</li> <li>• Lukas Chavez, PhD, RCIGM</li> <li>• Praveen B Raju, MD, PhD, UC San Diego</li> </ul> Moderator: Jerica Lenberg, MS, LCGC, RCIGM
4:00 – 4:30 pm	Frontiers 2024 Closing	Stephen Kingsmore, MD, DSc President & CEO, RCIGM

## FACULTY LISTING

### Planning Committee

Stephen Kingsmore, MB ChB BAO DSc  
President and Chief Executive Officer  
Rady Children's Institute for Genomic Medicine

### Visiting Faculty

Diana Azzam, PhD  
Assistant Professor, Lab Director, Environmental Health  
Sciences, Florida International University

Kathleen Lynch, Esq.  
Board Director  
Rady Children's Institute for Genomic Medicine

Matthew Bainbridge, PhD  
Head of Research  
Rady Children's Institute for Genomic Medicine

Sabrina Malone Jenkins, MD  
Neonatologist  
University of Utah/ Primary Children's Hospital

Aaron Besterman, MD  
Assistant Professor, Clinical Investigator  
Assistant Clinical Professor of Psychiatry,  
Department of Psychiatry, UC San Diego / Clinical  
Investigator, RCIGM/RCHSD

Thomas May, PhD  
Floyd and Judy Rogers Endowed Professor  
Washington State University,  
Vancouver Elson S. Floyd College of Medicine

Lukas Chavez, PhD  
Assistant Professor, Scientific Director, RCHSD  
Pediatric Neuro-Oncology Molecular Tumor Board /  
RCIGM / Sanford Burnham Prebys Medical  
Discovery Institute / UC San Diego

Megan Paul, MD  
Pediatric Neuro-Oncologist Clinical Co-lead  
RCHSD Pediatric Neuro-Oncology Molecular Tumor  
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Tennessee Health Science Center

Jonathan Santoro, MD  
Director of Neuroimmunology and Associate Professor  
of Neurology, Director, Neuroimmunology &  
Demyelinating Disorders Program, Director of  
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Kristen Suhrie, MD  
Director of Perinatal Genomics  
Indiana University School of Medicine and Riley  
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Gail Knight, MD, MMM  
Senior Vice President & Chief Medical Officer  
Rady Children's Hospital San Diego

Whitney Thompson, MD, MPhil  
Fellow Physician  
Mayo Clinic

Rob Knight, PhD  
Professor/PI, Professor of Pediatrics and Computer  
Science & Engineering, UC San Diego / Wolfe Family  
Endowed Chair in Microbiome Research,  
Rady Children's Hospital San Diego

Meredith Wright, Ph.D.  
Genomics Development Scientist  
Rady Children's Institute for Genomic Medicine

Jerica Lenberg MS, LCGC  
Lead Genetic Counselor  
Rady Children's Institute for Genomic Medicine

Jennifer Yang, MD  
Assistant Clinical Professor of Neurosciences, Division  
of Pediatric Neurology  
UC San Diego / RCIGM / RCHSD

### **Commercial Support Acknowledgement**

We would like to thank the following company for providing educational support of this activity:  
Takeda

### **Exhibitor Acknowledgement:**

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

- Illumina
- Kwoya Kirin
- Orchard
- Fabric Genomics
- UCB
- Mirum
- Chiesi
- Ultragenyx
- Inozyme
- AWS
- RCHSD
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