

## Transforming Lives with Rapid Precision Medicine™

Preliminary agenda as of April 8, 2021

DAY ONE:	Wednesday, April 21, 2021	8:00 am to 1:00 pm PDT		
Day One Theme: The Impact of RPM® in Clinical Practice				
8:00 am	Welcome	Stephen Kingsmore, MD, DSc President & CEO, RCIGM		
	A Fireside vChat: Transforming Lives with Rapid Precision Medicine™	Stephen Kingsmore, MD, DSc President & CEO, RCIGM		
		Eric Topol, MD Founder & Director, Scripps Research Translational Institute; Executive VP, Scripps Research; Professor of Molecular Medicine, Scripps Research; Gary & Mary West Endowed Chair of Innovative Medicine, Scripps Research		
9:00 am	13.5 Hour Automated Diagnosis and Clinical Decision Support of Genetic Diseases by Whole Genome Sequencing	Mallory Owen, MD Postdoctoral Fellow, RCIGM		
9:30 am	Closing the Evidence-to-Practice Gap in Rapid Whole Genome Sequencing	Linda Franck, RN, PhD, FAAN Professor, Department of Family Health Care Nursing, UC San Francisco		
10:00 am	BREAK – Exhibit Hall Open			
10:30 am	Advancing Scientific Approaches Through Administrative Data	Gretchen Bandoli, PhD Assistant Professor, Pediatrics, UC San Diego		
	Genomic Determinants of Infant Death in a Retrospective Cohort from San Diego County	Mallory Owen, MD Postdoctoral Fellow, RCIGM		
11:00 am	Payors' Perspective of RPM®: A Panel Discussion Facilitated by Russell Nofsinger, PhD Senior Director of Business Development, RCIGM	Bruce Quinn, MD, PhD, MBA Principal, Bruce Quinn Associates LLC  Jennifer Malin, MD, PhD Senior Medical Director, Oncology and Genetics, United Healthcare  Robert Plass, MD, MMM Medical Director Policy and Technology Assessment, Blue Shield of California  James Uli Senior Vice President and Chief Financial Officer, Rady Children's Hospital-San Diego		
11:45 am	Day One Insights	Stephen Kingsmore, MD, DSc President & CEO, RCIGM		
12:00 - 1:00 pm	Selected Scientific Poster Presentations:  Successful utilization of prenatal samples for diagnostic whole genome sequencing (WGS) in a case of recurrent pregnancy loss  Whole-genome sequencing identifies novel variants in MCM6 in siblings with Meier-Gorlin Syndrome phenotype  Building the Foundation of a Precision Genomic Medicine Clinic for Rare and Undiagnosed Disorders at Rady Children's Hospital  Rapid whole genome sequencing in the NICU: Lessons from a pilot study	Facilitated by Cody Sargent, MSc Clinical Research Associate, RCIGM		
12:00 - 1:00 pm	Exhibit Halls Open			



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DAY TWO Th	nursday, April 22, 2021	8:00 am to 1:00 pm PDT		
Day Two Theme: Research Findings That Impact RPM®				
8:00 am	Exhibit Hall Opens			
8:00 am	Illumina Presentation (Non-CME)			
9:00 am	Clinical Aspects of the Wales Infants' and Children's Genome Service for Unwell Children	Oliver Murch, BSc, MRes, PhD, MBBCh Clinical Geneticist, All Wales Medical Genomics Service, University of Wales, Cardiff, UK		
9:30 am	Developing the Infrastructure to Deliver Rapid Whole Genome Sequencing	Sian Corrin, PhD, FRCPath Clinical Scientist and Deputy Head, Institute of Laboratory, NHS Wales, All Wales Medical Genomics Service, University of Wales, UK		
10:00 am	BREAK – Exhibit Hall Open			
10:30 am	The Hopes and Challenges of Integrating Genomic Testing Platforms into Newborn Care	Jill Maron, MD, MPH Executive Director, Mother Infant Research Institute at Tufts Medical Center; Vice Chair of Pediatric Research, Tufts Children's Hospital; Professor of Pediatrics Adjunct Professor of Obstetrics and Gynecology, Tufts University School of Medicine		
11:00 am	Personalized Genomic Therapy in the Era of Antisense Oligonucleotides	Joseph Gleeson, MD, PhD Director of Neuroscience Research, RCIGM and Professor of Neuroscience and Pediatrics, UC San Diego School of Medicine		
11:30 am	RNA-seq for Rare Disease Diagnosis Overcomes Limitation of Conventional Genomic Testing	David Murdock, MD, FACMG Assistant Professor, Molecular and Human Genetics, and Assistant Director Human Genome Sequencing Center Clinical Laboratory, Baylor College of Medicine		
12:00 pm	Day Two Insights	Stephen Kingsmore, MD, DSc President & CEO, RCIGM		
12:05 — 1:00 pm	Selected Scientific Poster Presentations:  Identifying deleterious variants of overgrowth syndromes related genes in pediatric brain tumor patients' genomes  Whole genome sequencing in patients with Kabuki and CHARGE syndromes  WINGS – Wales Infants and Children's Genome Service. Rapid and accurate clinical analysis of variants from Whole Genome Sequencing assays using a user-friendly web application.  Identification of a novel FOXF1 gene variant by rapid whole genome sequencing: support for aggressive care of atypical alveolar capillary dysplasia  Rapid whole genome sequencing in the pediatric intensive care unit is cost-effective	Facilitated by Cody Sargent, MSc Clinical Research Associate, RCIGM		
12:05 – 1:00 pm	Exhibit Halls Open			



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DAY THREE F	riday, April 23, 2021	8:00 am to 1:00 pm PDT	
Day Three Theme: How RPM® Improves the Patient Family Experience			
8:00 am	Exhibit Hall Opens		
8:00 am	Bringing Novel Therapies to Market for Rare Diseases (Non-CME)	Dorothee Diogo, PhD Director of Statistical Genetics, Takeda Pharmaceuticals Company	
9:00 am	Genetic Sequencing for SCID: Linking Newborn Screening and Rapid Precision Therapy	Lori Broderick, MD, PhD Immunologist, Rady Children's Hospital-San Diego and Assistant Professor, Pediatrics and Allergy/Immunology, UC San Diego Shimul Chowdhury, PhD, FACMG, CGMB	
		VP of Laboratory Operations, BlueStar Genomics	
40.00	PDEAK ELITHING	Patient Family	
10:00 am	BREAK – Exhibit Hall Open		
10:30 am	Universal Screening for Genetic Cancer Predispositions in Pediatric Oncology	Dennis Kuo, MD, MS Hematologist/Oncologist, Rady Children's Hospital-San Diego and Clinical Professor, Pediatrics, UC San Diego  Sapna Mehta Clinical Researcher, Rady Children's Hospital-San Diego	
11:00 am	Parental Perceptions of Genomic Research in the Neonatal Intensive Care Setting	Julie Cakici, RN Researcher, Family Medicine and Public Health, UC San Diego; RCIGM; San Diego State University, School of Public Health	
11:30 am	Caregiver Experiences of Rapid Precision Medicine A Panel Discussion facilitated by Jerica Lenberg, MS, LCGC, Licensed Certified Genetic Counselor, RCIGM	Four patient families invited by the RPM® Family Partnership Program	
12:00 – 12:30 pm	Virtual Q&A and Conference Wrap Up	Stephen Kingsmore, MD, DSc President & CEO, RCIGM	