

## April 30 - Genomics 101

Half-Day Preconference at Hotel La Jolla		Noon to 5:00 pm
Noon	Lunch	
1:00 pm	Pre-Session Assessment	Karen Garman, EdD
1:15 pm	Intro to Precision Medicine	Lauge Farnaes, MD
1:30 pm	Types of Genetic Tests: Benefits and Limitations of Each	Chelsea Gatcliffe, MD
2:00 pm	How to Phenotype a Patient	Erica Sanford, MD
2:30 pm	How to Classify a Variant	Kasia Ellsworth, PhD
3:00 pm	BREAK	
3:15 - 4:45 pm  Station 1 3:15 pm  Station 2 3:45 pm  Station 3 4:15 pm	<ul> <li>Case-Based Learning:</li> <li>Principles of Genomic Analysis</li> <li>Reporting Results To The Clinical Team</li> <li>Communicating Results to the Family</li> </ul>	Jerica Lenberg, LCGS Shimul Chowdhury PhD  Kasia Ellsworth PhD Chelsea Gatliffe, MD & Erica Sanford, MD  Lisa Salz, MS, LCGC Kelly Watkins, MS, LCGC
4:45 pm	Post Session Assessment and Evaluations	Karen Garman, EdD

5 pm - Reception for all Preconference Attendees & Conference Speakers



DAY ONE	Wednesday, May 1	8 am to 4:30 pm	
Morning Focus – Implementing rWGS: Changing the Standard of Care			
8:00 am	Registration, Exhibit Hall Opens Breakfast		
9:00 am	Pioneering Rapid Whole Genome Sequencing in the NICU	Stephen Kingsmore, MD, DSc President and CEO, RCIGM	
9:15 am	Patient Case Presentation	Jennifer Friedman, MD	
9:45 am	Exploring New Frontiers: How Rady Children's Clinical Collaborators are Deploying rWGS	Regan Veith, CGC Children's Minnesota Austin Larson, MD, Children's Colorado Magaly Diaz Barbosa, MD, Nicklaus Children's	
10:30 am	BREAK		
11:00 am	So Who Is A Doctor Going To Sequence?	David Dimmock, MD Senior Medical Director, RCIGM	
11:30 am	The Potential and Challenges: Where Do We Go From Here? Panel: Rady Children's, Minnesota, Colorado, Nicklaus, Sanford-Fargo and Sanford-Sioux Falls	Facilitator: Lauge Farnaes, MD Assistant Medical Director, RCIGM	
Noon	Lunch		

## Afternoon Focus: Genomic Research that is Leading the Way to Implementation

1:00 pm	Overview	Stephen Kingsmore, MD, DSc
1:05 pm	Harnessing the Power of Genomics To Improve the Health of a Population	Richard Scott, MD Clinical Lead for Rare Disease, 100,000 Genomes Project, Genomics England
1:35 pm	Using Allelic Expression To Extend Rare Disease Diagnosis Beyond Coding Mutations	Pejman Mohammadi, PhD Scripps Research
2:05 pm	BREAK	
2:35 pm	Rapid Variant Testing In A Clinically Relevant Time Frame	Matthew Bainbridge, PhD Principal Investigator, RCIGM
3:05 pm	Towards an Automated Monogenic Patient Diagnostic System	Gill Bejerano, PhD Associate Professor of Developmental Biology, Stanford University
3:35 – 4:15 pm	Decoding Genetic Variants with Deep Learning	Kyle Kai-How Farh Director of Bioinformatics, Illumina



DAY TWO	Thursday, May 2	8 am to 4:30 pm	
Morning Focus – How Big Data Influences Implementation			
8:00 am	Registration, Exhibit Hall Opens		
9:00 am	Opening Remarks	Stephen Kingsmore, MD, DSc President & CEO, RCIGM	
9:15 am	Genomics England: Big Data in Genomics	Richard Scott, MD Clinical Lead for Rare Disease, 100,000 Genomes Project, Genomics England	
10:15 am	BREAK		
10:45 am	How Much More Can We Explain With Genetics? EHR Mining And The Undiagnosed Patient	Lisa Bastarche, MS Vanderbilt University	
11:30 am	Utilizing Machine-Learning and Clinical Language Processing to Diagnose Genetic Diseases in Record Time	Stephen Kingsmore, MD, DSc and Michelle Clark, PhD RCIGM	
Noon	Lunch		

## Afternoon Focus – Implementing rWGS in the NICU: Lessons Learned

1:00 pm	Project Baby Bear: California Tests the Waters of Public Health Coverage for WGS	Kathleen Lynch, JD Global Affairs & Public Policy Illumina
1:15 pm	The Project Baby Bear Experience at Participating Children's Hospitals	Jason Carmichael, LCGC Valley Children's Hospital Denise Suttner, MD Rady Children's Hospital
2:00 pm	BREAK	The system of the system
2:30 pm	The Project Baby Bear Experience at Participating Children's Hospitals	Suma Shankar, MD UC Davis Children's Hospital Jason Knight, MD, Children's Hospital Orange County
3:00 pm	Legislative Leadership and the Quest to Expand Access	California Assemblyman Brian Maienschein
3:30 pm	Maximizing Lab Results Through Collaboration and Consultation with NICU Teams	Shimul Chowdhury, PhD
4–4:30 pm	Panel Discussion/Audience Q &A	Facilitator: Kathleen Lynch, JD



DAY THRE	E Friday, May 3	8 am to noon	
Morning Focus – Genomic Sequencing at the Population Level			
8:00 am	Registration, Exhibit Hall Opens Breakfast		
9:00 am	The Case That Changed Our Entire Culture	Caleb Bupp, MD Medical Geneticist Spectrum Health Helen DeVos Children's Hospital	
9:30 am	Mining California's Statewide Birth Data: Study of Outcomes in Mother and Infants (SOMI)  Potential of Genomic Sequencing as a Preventative Diagnostic Tool for SIDS	Christina Chambers, PhD, MPH Director of Clinical Research Department of Pediatrics, UC San Diego and Rady Children's Hospital Matthew Bainbridge, PhD Principal Investigator, RCIGM	
10:15 am	BREAK		
10:30 am	From a Trillion Points of Data to Discovery	Atul Butte, MD, PhD UCSF School of Medicine	
11:15 am	Audience Q&A Wrap Up	Facilitator: Stephen Kingsmore, MD, DSc President & CEO, RCIGM	
Noon	Box Lunch		