Updated April 10, 2018

DAY ONE Wednesday, April 25		
Morning Focus – Shaping the Future: Transforming Pediatric Intensive Care		
8 – 9 am	Registration, Exhibit Hall	**Breakfast sponsored by Edico Genome**
9 – 9:10 am	Welcome	Stephen Kingsmore, MD, DSc, President and CEO Rady Children's Institute for Genomic Medicine (RCIGM)
9:10 – 10 am	Patient Story	A Life Changing Diagnosis: Rapid Whole Genome Sequencing Reveals a Treatable Disorder in a Neonate with Seizures David Dimmock, MD, Senior Medical Director, RCIGM, in conversation with Kristen Wigby, MD, RCIGM physician-scientist; mother and child
10 – 10:30 am	Presentation	Engineering Two-day Diagnosis with Rkurt apid Whole Genome Sequencing Shimul Chowdhury, PhD, DABMGG, CGMB Clinical Laboratory Director, RCIGM
10:30 – 11 am	Break	
11 – 11:30 am	Presentation	NICU Rapid Whole Genome Sequencing: The Sanford Genomic Medicine Consortium Experience Nancy Mendelsohn MD, Chief of Specialty Pediatrics, Children's Minnesota
11:30 – noon	Presentation	Use of Exome Sequencing for Infants in Intensive Care Units: The Experience at Baylor College of Medicine Linyan Meng, PhD, Laboratory Director, Baylor Genetics
12 – 1 pm	Networking	**Lunch sponsored by Fabric Genomics**
Afternoon Focus – Shaping the Future: Economic and Clinical Impact of Whole Genome Sequencing		
1 – 1:05 pm	Overview / Intro	Shimul Chowdhury, Clinical Lab Director, RCIGM
1:05 – 1:35 pm	Presentation	The Value Proposition in Genomic Diagnoses: Beyond Clinical Utility and Cost-Utility Scott Grosse, PhD, Senior Health Economist
1:35 – 2:05 pm	Presentation	Is Rare Disease Genomics Cost Effective? An Australian Perspective. Zornitza Stark, MA BMBCh DM Oxf, MBioeth FRACP Consultant Clinical Geneticist, Victorian Clinical Genetics Services Clinical Research Fellow, Australian Genomics Honorary Principal Fellow, Department of Paediatrics, University of Melbourne
2:05 – 2:30 pm	Break	
2:30 – 3 pm	Presentation	Improving and deploying clinical whole genome sequencing Ryan Taft, Phd, Senior Director Scientific Research, Clinical Genomics Research, Illumina
3 – 3:15 pm	Case Study	Molecular Diagnosis Where Rapid Whole Genome Sequencing Makes a Difference Kasia Ellsworth, Assistant Laboratory Director, RCIGM
3:15 – 3:45 pm	Presentation	Healthcare Economics in NICU and PICU Lauge Farnaes, MD, Hematololgy, Oncology, Infectious Diseases, Physician-Scientist RCIGM
3:45 – 4:15 pm	Discussion	Moderator: Shimul Chowdhury, Clinical Lab Director, RCIGM
4:15 – 7 pm	**Networking**	**Seaside DNA Day Reception sponsored by Illumina**

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DAY TWO Thursday, April 26		
Morning Focus – Shaping the Future: Discovery and Innovation		
8 – 9 am	Registration, Exhibit Hall	**Breakfast sponsored by DNAnexus**
9 – 9:05 am	Welcome	Shimul Chowdhury, Clinical Lab Director, RCIGM
9:05 – 9:45	Patient Story	David Dimmock, MD, Senior Medical Director RCIGM in conversation with physician, mother and child
9:45 – 10:15 am	Presentation	Value for Genomic Diagnosis in Biochemical Genetics and Beyond: A Pediatrician Perspective Sylvia Stockler MD, PhD, MBA, FRCPC, Professor Pediatrics, University of British Columbia, Head Division Biochemical Diseases, BCCH
10:15 – 10:40 am	Break	
10:40 – 10:55 am	Case Study	From Gene of Uncertain Significance to Diagnosis: Advancing Research and Providing Answers Meredith Wright, Clinical Genomics Analyst, RCIGM
10:55 – 11:15	Presentation	Genetic Susceptibility to Glioma Matthew Bainbridge, Msc, PhD, Clinical Investigator RCIGM
11:15 – 11:30	Case Study	Genome Sequencing in a Pediatric Patient with Recurrent Fevers and Colonic Ulceration Mari Tokita, MD, Asst. Clinical Lab Director, RCIGM
11:30 – 12:00	Presentation	VAAST, Phevor and the Utah Genome Project Mark Yandell, PhD, Department of Human Genetics, University of Utah, USTAR Center for Genetic Discovery, Salt Lake City, UT
12 – 1 pm	Networking	**Lunch sponsored by Amazon Web Services**
Afternoon Focus – Shaping the Future: Scaling Genomic Medicine with Artificial Intelligence		
1 – 1:10 pm	Topic Overview	Data Science Approaches to Rare-Disease Patient Diagnosis John Reynders, Vice President R&D Strategy, Program Management and Data Sciences, Alexion Pharmaceuticals
1:10 – 1:40 pm	Presentation	Human Phenotype Ontology-Driven Genomic Diagnostics Peter Robinson, MD, MSc, Professor of Computational Biology, The Jackson Laboratory for Genomic Medicine
1:40 – 1:55 pm	Presentation	Utilizing Natural Language Processing to Improve Phenotyping Girish Nadkarni, MD, Icahn School of Medicine, Mount Sinai
1:55 – 2:10	Presentation	Underdiagnosis of Monogenic Hypertension in a Multiethnic Clinical Cohort Ron Do, PhD Assistant Professor, Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai
2:10 – 2:40 pm	Break	
2:40 – 3:00 pm	Presentation	Collaborative Phenotyping with Your Patient Using the Human Phenotype Ontology Melissa Haendel, Director Ontology Development Group; Dept. of Medical Informatics & Clinical Epidemiology, Oregon Health & Science University
3 – 3:30 pm	Presentation	Artificial but Intelligent: Building an Accelerated Whole Genome Sequencing Pipeline with AI Stephen Kingsmore, Rady Children's Institute for Genomic Medicine
3:30 – 4 pm	Panel Discussion	Using AI to Automate and Expedite Diagnosis Moderator: John Reynders, Alexion

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DAY THREE Friday, April 27	CONFERENCE ENDS AT 12 pm	
Morning Focus – Shaping the Future: Newborn Sequencing in the Genomic Era		
8 – 9 am	Registration, Exhibit Hall	Breakfast sponsored by Karius
9 – 9:05 am	Welcome	Shimul Chowdhury, Clinical Lab Director, RCIGM
9:05 – 9:35 am	Presentation	Second-tier DNA testing in newborn screening for cystic fibrosis and metabolic disorders. Curt Scharfe, MD, PhD, Clinical Molecular Geneticist, Yale School of Medicine
9:35 – 9:40 am	Topic Overview	Insights from NSIGHT (Newborn Sequencing in Genomic Medicine and Public Health) Moderator: Anastasia Wise, PhD, Program Director, Division of Genomic Medicine, National Human Genome Research Institutes
9:40 – 10 am	Presentation	Newborn Screening in the NICU and the Nursery: The Critical Importance of DNA Sequence Renata Gallagher. University of California, San Francisco Robert Currier, former California Department of Public Health, chief statistician for Genetic Disease Screening Program
10 – 10:20 am	Presentation	Medically reportable outcomes and cost implications of newborn sequencing in the BabySeq project Ingrid Holm, MD, MPH, division of Genetics and Genomics Boston Children's Hospital Kurt Christensen, MPH, PhD, Brigham and Women's Hospital and Harvard Medical School
10:20 – 10:50 am	Break	
10:50 – 11:10 am	Presentation	North Carolina Newborn Exome Sequencing for Universal Screening: Findings from NC NEXUS decision aid data Cynthia Powell, MD, University of North Carolina Chapel Hill Megan Lewis, PhD, Program Director, Patient & Family Engagement Research at Research Triangle Institute (RTI) International
11:10 – 11:30 am	Presentation	Rady Children's Institute of Genomic Medicine : Clinical and Parental Perspectives Rapid Whole Genome Sequencing Results for Acutely Ill Newborns Nathaly Sweeney, MD, MPH Clinical Asst. Professor, Neonatal-Perinatal Medicine, UCSD; Physician Scientist, RCIGM
11:30 – 11:50 am	Panel Discussion	Moderator: Anastasia Wise, PhD, National Human Genome Research Institute
11:50 an – noon	Closing Remarks	New Frontiers in Pediatric Genomic Sequencing Stephen Kingsmore, RCIGM
12 pm	**Grab and Go boxed lunch**	**Sponsored by Clinithink**