FINAL AGENDA

DAY ONE	Wednesday, April 19, 2023	8:00 am to 4:00 pm PDT
MORNING FOCUS: rapid Whole Genome Sequencing™		
8:00 am	**Registration/Exhibit Hall/Breakfast**	
9:00 am	Welcome	Charlotte Hobbs, MD, PhD VP and Chief Research & Clinical Management Officer, Rady Children's Institute for Genomic Medicine (RCIGM) Stephen Kingsmore, MD, DSc President & CEO, RCIGM
9:15 am	Genomic Medicine for Ill Neonates and Infants: Lessons Learned from the GEMINI Study	Kristen Wigby, MD Assistant Professor of Pediatrics, University of California, San Diego / Physician Investigator, RCIGM
9:45 am	Streamlining Clinical Genetic Testing: The Promise of Long-Read Sequencing	Danny Miller, MD, PhD Assistant Professor of Pediatrics & Genome Sciences, University of Washington School of Medicine
10:15 am	**BREAK – Exhibit Hall Open**	
10:45 am	Infant Mortality Associated with Monogenic Disorders	Liana Protopsaltis, MS, CGC Research & Project Manager, RCIGM
11:15 am	Family Panel	Moderator: Jerica Lenberg, MS, CGC Licensed Clinical Genetic Counselor, RCIGM Patient families
Noon	**Lunch & Networking** Presentation by lunch sponsor Fabric Genomics: Rapid Diagnosis in the NICU and PICU: Using AI to Overcome the Challenges of Genomic Variant Interpretation.	Mark Yandell, PhD Professor of Human Genetics, Co-Director Utah Center for Genetic Discovery School of Medicine, University of Utah
AFTERNOON FOCUS: Newborn Genomic Screening		
1:00 pm	Introduction to Newborn Genomic Screening	Stephen Kingsmore, MD, DSc President & CEO, RCIGM
1:05 pm	Genomics England Newborn Genomes Programme	David Bick, MD, PhD Principal Clinician, Genomics England
1:40 pm	Genome Sequencing in Newborns: A Public Health Perspective	Laurie Smith, MD, PhD RCIGM Consultant

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		Gunter Scharer, MD Chief Medical Officer, RPRD Diagnostics Jennifer Schleit, PhD, FACMG Laboratory Director, RCIGM Moderator: Mei Baker, MD, FACMG Professor of Pediatrics, University of Wisconsin-Madison / Newborn Screening Laboratory Director, Wisconsin State Laboratory of Hygiene
2:15 pm	**BREAK – Exhibit Hall Open**	
2:45 pm	The Path to Universal Newborn Sequencing	Robert Green, MD, MPH Professor of Medicine (Genetics), Harvard Medical School and Mass General Brigham
3:15 pm	Challenges and Opportunities in Screening Newborns and Young Children for Complex Conditions using Genetic Risk Scores	Holly Peay, PhD, MS, CGC Early Check / Senior Research Scientist, RTI International
3:45 pm	Closing Remarks	Drs. Stephen Kingsmore and Charlotte Hobbs, RCIGM
4:30 – 7:00 pm	**Reception/Networking** Heavy hors d'oeuvres and cocktails Brief sponsor presentation: Illumina Opening Remarks and Role in Whole Genome Sequencing	Chris Kunard, Sr. Director, Open Innovation Customer Collaboration, Illumina

DAY TWO	Thursday, April 20, 2023	8:00 am to 4:30 pm PDT
MORNING FOCUS: Newborn Genomic Screening		
7:30 am	**Registration/Exhibit Hall/Breakfast**	
8:00 am	Welcome	Charlotte Hobbs, MD, PhD VP and Chief Research & Clinical Management Officer, RCIGM
8:05 am	Screen4Care EU-IMI Project: Accelerating Rare Disease Diagnosis by Genetic Newborn Screening and Digital Technology	Alessandra Ferlini, MD, PhD Professor & Head of the Medical Genetics Unit, University of Ferrara / Project Scientific Coordinator, Screen4Care Nicolas Garnier, PhD EFPIA Lead, Screen4Care / Head of Patient Advocacy, Pfizer Global Product Development
8:40 am	Newborn Genomic Screening in Greece	Petros Tsipouras, MD Scientific Director, First Steps Greece / CEO, PlumCare
9:15 am	The BeginNGS Perspective	Tom DeFay, PhD Deputy Chair, BeginNGS Consortium / Deputy Head of Diagnostics Strategy & Development, Alexion
9:50 am	Panel: Newborn Genomic Screening Programs Across the Globe	David Bick, MD Principal Clinician, Genomics England Alessandra Ferlini, MD, PhD Professor & Head of the Medical Genetics Unit, University of Ferrara / Project Scientific Coordinator, Screen4Care Nicolas Garnier, PhD EFPIA Lead, Screen4Care / Head of Patient Advocacy, Pfizer Global Product Development Holly Peay, PhD, MS, CGC Senior Research Scientist, RTI International Robert Green, MD, MPH Professor of Medicine (Genetics), Harvard Medical School and Mass General Brigham Petros Tsipouras, MD Scientific Director, First Steps Greece / CEO, PlumCare Moderator: Tom DeFay, PhD Deputy Chair, BeginNGS Consortium / Deputy Head of Diagnostics Strategy & Development, Alexion
10:30 am	**BREAK – Exhibit Hall Open**	
11:00 am	Unraveling the Diagnosis of ENPP1 Deficiency	Catherine Nester, RN, BSN VP, Physician & Patient Strategies, Inozyme Pharma

11:30 am	Panel: Addressing the Diagnostic Odyssey Through Pharma Participation in NBS-by-WGS programs	Catherine Nester, RN, BSN VP, Physician & Patient Strategies, Inozyme Pharma Charlotte Chanson, MSc Senior Director, Global Diagnostics/Newborn Screening, Orchard Therapeutics Steve Rodems, PhD VP, Research, Travere Therapeutics Tom DeFay, PhD Deputy Chair, BeginNGS Consortium / Deputy Head Diagnostics Strategy & Development, Alexion Nicole Miller, PhD VP, Molecular Diagnostics, Ultragenyx Moderator: Walter Kowtoniuk, PhD Venture Partner, Third Rock Ventures
12:10pm	**Lunch & Networking**	
AFTERNOON FOCUS: Diagnostic rWGS®, Effectiveness & Reimbursement		
1:10 pm	Welcome Back	Charlotte Hobbs, MD, PhD VP and Chief Research & Clinical Management Officer, RCIGM
1:15 pm	Lightning Talks DNA Methylation Detection Using PacBio Sequencing Improving Outcomes of Critical Congenital Heart Disease Through Genome-Informed Management Pediatric Genomic Psychiatry: Precision Medicine for Youth Mental Health	 Matthew Bainbridge, PhD Assistant Director of Translational Research, RCIGM Nathaly Sweeney, MD, MPh, MS Assistant Clinical Professor of Pediatrics, University of California, San Diego / Physician Investigator, RCIGM Aaron Besterman, MD Assistant Clinical Professor of Psychiatry, University of California, San Diego / Physician Investigator, RCIGM
1:45 pm	Next-Generation Advocacy Driving Forward Collaborative Progress in Rare Disease	Charlene Son Rigby, MBA CEO, Global Genes
2:15 pm	Bears, Manatees, and Deer, Oh My! – Michigan's Implementation of rWGS Affirms Strong Clinical and Economic Utility	Caleb Bupp, MD FACMG Assistant Clinical Professor of Pediatrics and Human Development, Michigan State University / Geneticist, Division Chief, Corewell Health & Helen DeVos Children's Hospital
2:45 pm	Economic Utility of First Line rWGS® in Commercial Payor Model Empowers Coverage and Implementation	Christy Moore, MS, CGC Genetics Program Manager, Blue Shield of California
3:15 pm	**BREAK – Exhibit Hall Open**	

3:45 pm	Panel: rWGS — Michigan's Journey to Coverage and Implementation	Andrea Scheurer-Monaghan, MD Clinical Assistant Professor of Pediatrics, Western Michigan University / Neonatologist, Southwestern Michigan Neonatology, P.C. Laura Appel Executive VP, Michigan Health & Hospital Association David Dimmock, MD Staff Physician, Valley Children's Hospital Moderator: Caleb Bupp, MD FACMG Assistant Clinical Professor of Pediatrics and Human Development, Michigan State University / Geneticist, Division Chief, Corewell Health & Helen DeVos Children's Hospital
4:25 – 4:40 pm	Closing Discussion	Drs. Stephen Kingsmore and Charlotte Hobbs, RCIGM