
# PRELIMINARY AGENDA

| **DAY ONE** | Wednesday, April 19, 2023 | 8:00 am to 4:30 pm PDT |
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| **MORNING FOCUS: rapid Whole Genome Sequencing™** | | |
| 8:00 am | Registration/Exhibit Hall/Breakfast | |
| 9:00 am | Welcome | Charlotte Hobbs, MD, PhD<br>VP and Chief Research & Clinical Management Officer, Rady Children's Institute for Genomic Medicine (RCIGM)<br>Stephen Kingsmore, MD, DSc<br>President & CEO, RCIGM |
| 9:15 am | Genomic Medicine for Ill Neonates and Infants (GEMINI Study) | Kristen Wigby, MD<br>Physician Investigator, RCIGM |
| 9:45 am | Streamlining Clinical Genetic Testing: The Promise of Long-Read Sequencing | Danny Miller, MD, PhD<br>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<br>Research & Project Manager, RCIGM |
| 11:15 am | Family Panel | Moderator: Jerica Lenberg, MS, CGC<br>Licensed Clinical Genetic Counselor, RCIGM<br>Patient families |
| Noon | Lunch & Networking | |
| **AFTERNOON FOCUS: Newborn Genomic Screening** | | |
| 1:00 pm | Introduction to Newborn Genomic Screening | Stephen Kingsmore, MD, DSc<br>President & CEO, RCIGM |
| 1:05 pm | The Genomics England Perspective | David Bick, MD, PhD<br>Principal Clinician, Genomics England |
| 1:40 pm | Screen4Care EU-IMI Project:<br>Accelerating Rare Disease Diagnosis by Genetic Newborn Screening and Digital Technology | Alessandra Ferlini, MD, PhD<br>Professor & Head of the Medical Genetics Unit, University of Ferrara / Project Scientific Coordinator, Screen4Care<br>Nicolas Garnier, PhD<br>Head of Patient Advocacy, Pfizer Global Product Development / EFPIA Lead, Screen4Care |



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| 2:15 pm | BREAK – Exhibit Hall Open | |
| 2:35 pm | The BeginNGS Perspective | Tom DeFay, PhD<br>Deputy Head of Diagnostics Strategy & Development, Alexion / Deputy Chair, BeginNGS Consortium |
| 3:10 pm | The Early Check Perspective | Holly Peay, PhD, MS, CGC<br>Senior Research Scientist, RTI International |
| 3:40 pm | Panel: Newborn Genomic Screening Programs Across the Globe | David Bick, MD<br>Principal Clinician, Genomics England<br>Alessandra Ferlini<br>Professor & Head of the Medical Genetics Unit, University of Ferrara / Project Scientific Coordinator, Screen4Care<br>Nicolas Garnier, PhD<br>Head of Patient Advocacy, Oncology & Rare Disease, Pfizer / EFPIA Lead, Screen4Care<br>Holly Peay, PhD, MS, CGC<br>Senior Research Scientist, RTI International<br>Robert Green, MD, MPH<br>Director, G2P (Genomes to People)<br>Petros Tsipouras, MD<br>CEO, PlumCare / Scientific Director, First Steps Greece<br>Moderator: Tom DeFay, PhD<br>Deputy Head of Diagnostics Strategy & Development, Alexion / Deputy Chair, BeginNGS Consortium |
| 4:30 – 7:00 pm | Reception/Networking | Heavy hors d'oeuvres and cocktails |

| **DAY TWO** | Thursday, April 20, 2023 | 8:00 am to 4:30 pm PDT |
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| **MORNING FOCUS: Newborn Genomic Screening** | | |
| 8:00 am | Registration/Exhibit Hall/Breakfast | |
| 8:45 am | Welcome | Charlotte Hobbs, MD, PhD<br>VP and Chief Research & Clinical Management Officer, RCIGM |
| 8:50 am | Panel: Addressing the Therapeutic Odyssey Through Pharma Participation in NBS-by-WGS programs | Catherine Nester, RN, BSN<br>Vice President, Physician & Patient Strategies, Inozyme Pharma<br>Charlotte Chanson, MSc<br>Senior Director, Global Diagnostics/Newborn Screening, Orchard Therapeutics<br>Steve Rodems, PhD<br>Vice President, Research, Travere Therapeutics<br>Tom DeFay, PhD<br>Deputy Head Diagnostics Strategy & Development, Alexion / Deputy Chair, BeginNGS Consortium<br>Nicole Miller, PhD<br>VP, Molecular Diagnostics, Ultragenyx Pharmaceuticals<br>Moderator: Walter Kowtoniuk, PhD<br>Venture Partner, Third Rock Ventures |
| 9:30 am | Newborn Genomic Screening in Greece | Petros Tsipouras, MD<br>CEO, PlumCare / Scientific Director, First Steps Greece |
| 10:05 am | The BabySeq Perspective | Robert Green, MD, MPH<br>Director, G2P (Genomes to People) |
| 10:40 am | BREAK – Exhibit Hall Open | |
| 11:00 am | Next-Generation Advocacy Driving Forward Collaborative Progress in Rare Disease | Charlene Son Rigby, MBA<br>CEO, Global Genes |
| 11:30 am | Panel: Bringing Together the Science and Medicine: Laboratory and Geneticists' Perspectives | Laurie Smith, MD, PhD<br>Consultant<br>Gunter Scharer, MD<br>Chief Medical Officer, RPRD Diagnostics<br>Kasia Ellsworth, PhD, FACMG, CGMB<br>Senior Director of Clinical Operations, RCIGM<br>Jennifer Schleit, PhD, FACMG<br>Laboratory Director, RCIGM<br>Moderator: Mei Baker, MD, FACMG<br>Newborn Screening Laboratory Director, Wisconsin State Laboratory of Hygiene |

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| Noon | Lunch & Networking | |
| **AFTERNOON FOCUS: Diagnostic rWGS®, Effectiveness & Reimbursement** | | |
| 1:00 pm | Welcome Back | Charlotte Hobbs, MD, PhD<br>VP and Chief Research & Clinical Management Officer, RCIGM |
| 1:05 pm | Lightning Talks<br><br>Pediatric Precision Psychiatry: Leveraging Genomics to Advance Youth Mental Health | <br><br>Matthew Bainbridge, PhD<br>Assistant Director of Translational Research, RCIGM<br><br>Nathaly Sweeney, MD, MPh, MS<br>Physician Investigator, RCIGM<br><br>Aaron Besterman, MD<br>Physician Investigator, RCIGM |
| 1:35 pm | Unraveling the Diagnosis of ENPP1 Deficiency | Catherine Nester, RN, BSN<br>Vice President, Physician & Patient Strategies, Inozyme Pharma |
| 2:00 pm | Bears, Manatees, and Deer, Oh My! – Michigan's Implementation of rWGS Affirms Strong Clinical and Economic Utility | Caleb Bupp, MD FACMG<br>Geneticist, Division Chief, Corewell Health & Helen DeVos Children's Hospital |
| 2:30 pm | Economic Utility of First Line rWGS in Commercial Payor Model Empowers Coverage and Implementation | Christy Moore, MS, CGC<br>Genetics Program Manager, Blue Shield of California |
| 3:00 pm | BREAK – Exhibit Hall Open | |
| 3:20 pm | Panel: rWGS — Michigan's Journey to Coverage and Implementation | Andrea Scheurer-Monaghan, MD<br>Neonatologist, Southwestern Michigan Neonatology, P.C.<br>Laura Appel<br>Executive VP, Michigan Health & Hospital Association<br>David Dimmock, MD<br>Chief Medical Officer, Creyon Bio<br>Moderator: Caleb Bupp, MD FACMG<br>Division Chief - Medical Genetics & Genomics, Spectrum Health & Helen DeVos Children's Hospital |
| 4:00 – 4:30 pm | Closing Discussion | Drs. Stephen Kingsmore and Charlotte Hobbs, RCIGM |