

Defining a Clinical Data Ecosystem for Genomic Health

Wednesday, April 10th, 2024

Venue: The Line Hotel DC, 1770 Euclid St NW, Washington, DC 20009

Meeting Room: Carina

8:00 - 8:30 AM Breakfast | *Carina East*

8:30 - 9:15 AM Introduction | 45 minutes (15 minute presentations)

- Overview of the current issue and objective | **Nephi Walton** (NHGRI)
- Open the Genome to All | **Laura Duque Lasio** (Washington Univ.)
- Chief medical information officer (CMIO) perspective | **Kevin O'Bryan** (Washington University)

9:15 - 10:45 AM Real World Genomic Data in Clinical Care | 1 hour, 30 min (15 min presentations; 45 min discussion)

- Germline Mendelian Disease Data | **Ian Campbell** (CHOP)
- Systematic Review of Genomic Clinical Decision Support | **Darren Johnson** (Geisinger)
- Pharmacogenomics Data/External Systems | **Mark Dunnenberger** (North Shore)
- Informatics Challenges Open Discussion | Moderator: **Travis Osterman** (VUMC)

10:45 - 11:05 AM Break | 20 min.

11:05 - 12:50 PM Real Time and Rapid Genetic Diagnosis | 1 hour, 45 min (15 min presentations; 1 hour discussion)

- Real-Time Genetic Diagnosis at the Point of Care | **Marc Williams** (Geisinger)
- Expanding Rapid NICU Sequencing | **Luca Brunelli** (Univ. of Utah)
- Rapid NICU Sequencing Infrastructure | **Valerie Willis** (Indiana Univ.)
- Implementation Challenges Open Discussion | Moderator: **Ken Wiley** (NCATS)

12:50 - 1:20 PM Working Lunch | 30 min. *Carina*

1:20 - 3:00 PM AI and Polygenic Risk Scores | 1 hour, 40 min (15 min presentations; 55 min discussion)

- Data Availability | **Iftikhar Kullo** (Mayo Clinic)
- FHIR Genomics Operations | **Bob Dolin** (Elimu/HL GenomeX)
- Maximizing Genomic Clinical Utility: Context and Learning | **Pradeep Natarajan** (Broad)
- Implementation Challenges Open Discussion | Moderator: **Josh Peterson** (VUMC)

3:00 - 3:20 PM Break | 20 min.

3:20 - 4:50 PM Delivering Genomics Information to Patients and Providers | 1 hour, 30 min (15 min presentations; 45 min discussion)

- Primary Care Perspective | **Sharice Wood** (University of Cincinnati)
- Chatbots for Patient Outreach, Education, and Access to Genetic Testing | **Guilherme Del Fiol** (Univ. of Utah)
- Implementation of Genomic Information Resources | **Casey Overby Taylor** (Johns Hopkins)
- Implementation Challenges Open Discussion | Moderators: **Alanna Rahm** (NHGRI), **Juliann Savatt** (Geisinger)

4:50 - 5:00 PM Closing Remarks

5:00 PM Adjourn

**Dial-In
Information**

General Session Meeting ID: 929 2476 1966 Passcode: 988705
Meeting Link: <https://zoom.us/j/92924761966?pwd=SFFsZnhGZFFFUG1SdnFhNzVFbjg2QT09>
Dial-In: +1 346 248 7799 or +1 253 215 8782 or +1 669 900 6833 or +1 646 558 8656 or
+1 312 626 6799 or +1 301 715 8592

Defining a Clinical Data Ecosystem for Genomic Health

Thursday, April 11th, 2024

Venue: The Line Hotel DC, 1770 Euclid St NW, Washington, DC 20009

Meeting Room: Carina

8:00 - 8:30 AM Breakfast | *Carina East*

8:30 - 9:45 AM **Models for Genomic Data Sharing** | 1 hour, 15 min (5-15 min presentations; 30 min discussion)

- Brief Day One Recap | **John David Nolen** (*Children's Mercy Hospital Kansas City*)
- ACMG Points to Consider: Designated Record Set | **Marc Williams** (*Geisinger*)
- Models for Data Sharing | **Bret Heale** (*Humanized Health Consulting*)
- Is PACS a Model for FHIR Genomics? | **Srikar Chamala** (*University of Southern California*)
- Group Discussion | Moderator: **John David Nolen** (*Children's Mercy Hospital Kansas City*)

9:45 - 10:05 AM Break | 20 minutes

10:05 - 11:50 AM **Model Design Workshop** | 1 hour, 45 minutes

- Moderator: **Bret Heale** (*Humanized Health Consulting*)

11:50 AM - 12:20 PM Working Lunch | 30 minutes (*Carina*)

12:20 - 1:05 PM **Evaluating the Model: Data and Technical Standards** | 45 minutes

- **Robert Freimuth** (*Mayo Clinic*)
- **Robert Carroll** (*VUMC*)

1:05 - 1:50 PM **Evaluating the Model: Governance, Legal, and Ethical Challenges** | 45 minutes

- **Jorge Contreras** (*Univ. of Utah*)
- **Kevin O'Bryan** (*Washington Univ.*)
- **Casey Overby Taylor** (*Johns Hopkins*)

1:50 - 2:45 PM **Evaluating the Model: Sustainability** | 55 minutes

- **Bryce Christensen** (*Univ. of Utah*)
- **Michael Murray** (*Mt. Sinai*)
- **Kevin O'Bryan** (*Washington Univ.*)

2:45 - 3:20 PM **Path Forward and Action Plans** | **Nephi Walton** (NHGRI) 35 minutes

- Synthesis of Key Points from Discussions
- Setting Priorities: Identifying the Most Critical Barriers
- Closing Remarks and Future Collaboration

3:20 PM Adjourn