

Center for Pediatric Genomic Medicine Short Course Agenda, 2021

(Note: Times are in CST)

Monday, June 7, 2021		
8:00-8:30 a.m.	Welcome, Introduction, and pre-course assessment	Emily Farrow, PhD, CGC
8:30-9:00 a.m.	Introduction to Genomics and Human Variation	Emily Farrow, PhD, CGC
9:00-10:00 a.m.	Next Generation Sequencing Fundamentals and Laboratory Approach	Emily Farrow, PhD, CGC
10:00-10:30 a.m.	Alignment and the Reference Genome	Neil Miller
10:30-10:45 a.m.	Break	
10:45-11:30 a.m.	Variant calling and Annotation	Neil Miller
11:30 a.m.-12:00 p.m.	Changes to the Regulations: Implications for Genomic Studies	Ryan McDowell
12:00-1:00 p.m.	Lunch Break	
1:00-2:00 p.m.	Bioethics: Bridging the Tension between Research and Clinical Testing	John Lantos, MD
2:00-3:30 p.m.	Bioethics- Ethical and Societal Impacts of Germline Gene Editing: How Can We Secure Human Rights?	Jodi Halpern, PhD
3:30-4:00 p.m.	Case Study Discussion: Evaluate Ethical Considerations in Research and Genomic Sequencing	Courtney Berrios, CGC Emily Farrow, PhD, CGC

Tuesday, June 8, 2021		
8:00-9:00 a.m.	Introduction to Variant Interpretation	Joe Alaimo, PhD
9:30-10:00 a.m.	Introduction to Analysis Workshop 1 (Orientation to Software, access)	Joe Alaimo, PhD Emily Farrow, PhD, CGC
10:00-11:30 a.m.	Workshop 1: Variant Analysis	Analysis Team
11:30 a.m.-12:00 p.m.	Workshop 1: Wrap-up and Discussion	Analysis Team
12:00-1:00 p.m.	Lunch	
1:00-2:00 p.m.	Introduction to RNAseq- Studies and Analysis	Jeff Johnston
2:00-3:00 p.m.	Environmental Trigger or Genetic Predisposition: Reading Disease Risk from the Epigenome	Elin Grundberg, PhD
3:00-4:00 p.m.	Workshop 2: Personalized Genomic Education Testing (PGET) Analysis and Genetic Counseling	Analysis and GC Teams



Wednesday, June 9, 2021

8:00-9:00 a.m.	Parent of Origin Effects: Mitochondrial Genome and Monoallelic Expression	Isabelle Thiffault, PhD, FACMG
9:00-11:00 a.m.	Using the UCSC Genome Browser to Interpret Human Genetic Variants	Robert Kuhn, PhD
11:00 a.m.-12:30 p.m.	Precision Medicine in Oncology -Interpretation and Bioinformatic challenges	Midhat Farooqi, MD, PhD
12:30-1:00 p.m.	Lunch Break	
1:00-2:00 p.m.	Architecture of Variation in Gene Regulation and Human Disease	Tomi Pastinen, MD, PhD
2:00-2:45 p.m.	Approaches to Functional Validation	Scott Younger, PhD
2:45-3:00 p.m.	Break	
3:00-4:00 p.m.	Pharmacogenetics- Challenges and Applications	Andrea Gaedigk, PhD

Thursday, June 10, 2021

8:00-9:15 a.m.	Epilepsy Genetics--from Patients to Models	Ann Poduri, MD, MPH
9:15-9:45 a.m.	Discussion Session/Mentor Match	
9:45-10:15 a.m.	Intro to Trio Analysis	Joe Alaimo, PhD
10:15 a.m.-12:15 p.m.	Workshop 3: Trio Analysis and complex cases	Analysis Team
12:15-12:45 p.m.	Workshop 3: Wrap-up and Discussion	Analysis Team
12:45-1:00 p.m.	Course wrap-up	Emily Farrow, PhD, CGC

