

International Summit in Human Genetics and Genomics
National Human Genome Research Institute
National Institutes of Health
August 31st – September 29th, 2018

Agenda

Bldg.10 Room 4-B1C2015 (FAES Classroom)

	Topic	Speaker
Aug 31st (Fri)		
8:00 – 8:45 am	Registration	Please bring Passports and visas
8:45 – 9:00 am	SURVEY	
9:00 – 9:15 am	Welcome	Dr. Eric Green Director, NHGRI Dr. Max Muenke Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
9:15– 10:15 am	The Genomic Landscape Circa 2018	Dr. Eric Green Director, NHGRI
10:15 – 10:30 am	BREAK	
10:30 – 11:30 pm	Family History Collection & Assessment	Dr. Ben Solomon Managing Director, GeneDx
11:30 am – 12:30 pm	Traditional & Non-traditional patterns of inheritance	Dr. Ben Solomon Managing Director, GeneDx
12:30 – 1:30 pm	LUNCH	
1:30 – 2:30 pm	Dysmorphology	Dr. Ben Solomon Managing Director, GeneDx
2:30 – 3:30 pm	Hands on Dysmorphology Practice	Dr. Max Muenke Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
3:30 – 4:00 pm	BREAK	
4:00 – 4:45 pm	CDC, FDA, NIH	Dr. Martin Ruiz-Beltran CAPT, U.S. Public Health Service Director, Division of Management Operations, Office of Management / Office of Regulatory Affairs U.S. Food and Drug Administration
4:30 – 4:45 pm	Survey	

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 4th (Tues)		
8:00 – 8:15 am	SURVEY	
8:15 – 9:15 am	The Role of Race, Ethnicity and Socioeconomic Status in Clinical Genetics Research	Dr. Eliseo Perez-Stable Director, NIMHD
9:15 – 10:15 am	Inherited Pulmonary Disorders	Dr. Bernadette Gochuico Attending Pulmonologist, Medical Genetics Branch, NHGRI
10.15 - 10.45 am	BREAK	
10.45 - 11.45 am	Ophthalmic Genetics	Dr. Brian Brooks Clinical Director, Chief, Unit on Pediatric, Developmental & Genetic Ophthalmology, NEI
11.45 - 1.00 pm	LUNCH	
1:00 – 2:00 pm	Neuromuscular Disease	Dr. Carsten Bonnemann, Senior Investigator, Neuromuscular & Neurogenetic Disorders of Childhood Section, NINDS
2:00 – 3:00 pm	Sickle Cell Disease, The Global Health Burden	Dr. Vence Bonham, Senior Advisor to the NHGRI Director on Genomics and Health Disparities, NHGRI
3:00 – 3:30 pm	BREAK	
3.30- 4:30 pm	Using whole genome sequencing and genomics to understand the clinical impact of human sequence variation	Dr. Drew Michael Chief Clinical Genetics Fellow NIH/NHGRI, NIH/NIDCR
4:30 – 5:30 pm	Genetics of Lipid Disorders	Dr. Robert Shamburek, Clinician/Scientist, Lipid Section, Cardiovascular and Pulmonary Branch, NHLBI
5:30- 5.45 pm	SURVEY	

Meet at The Gateway Visitor's Center, NIH

	Topic	Speaker
Sept. 5th (Wed)		
8.00 – 8.15 am	Survey on the bus	
9:00 am – 5:30 pm	<p>Field Trip- NIH Intramural Sequencing Center (NISC) 9-11 am 5625 Fishers Lane Rockville, MD 20852</p> <p>GeneDX 12- 3 pm 207 Perry Pkwy, Gaithersburg, MD 20877</p> <p>National Center for Advancing Translation Sciences (NCATS) 3:30-5 pm 9800 Medical Center Dr., Rockville, MD 20850</p> <p>Survey on the bus</p>	

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 6th (Thurs)		
8:00 – 8:15 am	SURVEY	
8:15 – 9:15 am	Neurogenetics	Dr. Andrea Gropman, Division Chief, Neurodevelopmental Disabilities & Neurogenetics, CNHS
9:15 – 10:15 am	Human Microbiome Research	Dr. Julie Segre, Senior Investigator Translational and Functional Genomics NHGRI
10.15 - 10.45 am	BREAK	
10.45 - 11.45 am	Genetic Counseling	Mr. Don Hadley Certified Genetic Counselor Deputy Director, Medical Genetics & Genomic Medicine Residency, NHGRI
11.45 - 1.00 pm	LUNCH	
1:00 – 2:30 pm	Genetics of Skin Disorders	Dr. Gabriele Richard, Chief Medical Officer, GeneDx
2:30 – 3:30 pm	NIH Undiagnosed Disease Program	Dr. Bill Gahl, Clinical Director Head, Undiagnosed Disease Program, NHGRI
3:30 – 3:45 pm	BREAK	
3:45 – 4:00 pm	Walk to the Clinical Center	
4:00 – 6:00 pm	CLINICAL CENTER TOUR: 4:00 - 4:30 pm Metabolic Unit 4:45 - 5:15 pm Organoid Culture 5:30 – 6:00 pm Clinical Center	Drs. Kong Chen and Stephanie Chung NIDDK Dr. Peter McGuire Head, Metabolism, Infection and Immunity Section, NHGRI Ms. Laura M. Lee, MS, RN Director, Office of Patient Safety and Clinical Quality NIH Clinical Center
6:00- 6:15 pm	SURVEY	

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 7th (Fri)		
8:00 – 8:15 am	SURVEY	
8:15 – 10:15 am	Cytogenetics	Dr. Jeanne Meck, Director, Prenatal Diagnosis & Cytogenomics, GeneDx
10.15 - 10.45 am	BREAK	
10.45 - 11.45 am	Clinical Aspects of Cytogenetic Disorders	Dr. Clesson Turner, Clinical Geneticist, WRNMMC
11.45 - 1.00 pm	LUNCH	
1:00 – 2:00 pm	Variant Nomenclature	Dr. Christopher Lau, Clinical Molecular Geneticist, Undiagnosed Disease Program, NHGRI
2:00 – 3:00 pm	Variant Interpretation	Dr. Christopher Lau, Clinical Molecular Geneticist, Undiagnosed Disease Program, NHGRI
3:00 – 3:30 pm	BREAK	
3:30 – 4:30 pm	Genetics of Cardiac Structural Defects	Dr. Laura Olivieri, Pediatric Cardiologist CNHS
4:30 – 5:30 pm	Genetics of Arrhythmia and Cardiomyopathy	Dr. Elizabeth Sherwin, Pediatric Electrophysiologist, CNHS
5:30- 5.45 pm	SURVEY	

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 10th (Mon)		
8:00 – 8:15 am	SURVEY	
8:15 – 9:15 am	Nurse Scientists: Moving Precision Medicine Forward	Dr. Katy Meilleur, PhD, PPCNP-BC Tenure Track Investigator, Chief, Neuromuscular Symptoms Branch NINR
9:15 – 10:15 am	Clinical Trials	Dr. Cyndi Tifft Deputy Clinical Director, NHGRI
10.15 - 10.45 am	BREAK	
10.45 - 11.45 am	Noninvasive Prenatal Genomics: The Vanguard of Genomic Medicine	Dr. Diana W. Bianchi, Director, Eunice Kennedy Shriver National Institute of Child Health and Human Development
11.45 - 1.00 pm	LUNCH	
1:00 – 2:00 pm	Genetics of Hearing Loss	Dr. Isabelle Roux, Molecular Biology and Genetics Section, Otolaryngology Branch, NIDCD
2:00 – 3:00 pm	Genetic Databases (ClinVar, dbGAP, GTR)	Ms. Adriana Malheiro, Staff Scientist, Director, NIH Genetic Testing Registry, NCBI/NLM
3:00 – 3:30 pm	BREAK	
3:30 – 4:30 pm	What Makes Research Ethical?	Mr. Ben Berkman, Faculty, NIH Department of Bioethics Deputy Director, NHGRI Bioethics Core
4:30 – 5:30 pm	Ethical Issues in Genetic Research with Stored Samples	Dr. Sara Hull, Director, Bioethics Core, Office of the Clinical Director, NHGRI
5:30- 5.45 pm	SURVEY	

Meet at The Gateway Visitor's Center, NIH

	Topic	Speaker
Sept. 11th (Tues)		
7:30 – 7:45 am	Survey on the bus	
8:45 – 5:00 pm	Field Trip- Johns Hopkins University 8:45-11:30 733 N Broadway, Baltimore, MD 21205 Lunch at JHU 11:30-12 Kennedy Krieger Institute 12-2 Maryland Newborn Screening Laboratory 2:30-4:30 Survey on the bus	

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic		Speaker
Sept. 12th (Wed)			
8:00 – 8:15 am	SURVEY		
8:15 – 9:15 am	Diabetes: Insights from Genomics	Dr. Lori Bonnycastle Staff Scientist, NHGRI	
9:45 – 10:30 am	Genetic syndromes in diverse populations	Dr. Paul Kruszka, Staff Clinician, NHGRI	
10:30 – 11:00 am	BREAK		
11:00 – 12:00 pm	Genetics of Periodontal disease	Dr. Thomas Hart Senior Director ADAF Volpe Research Center	
12:00 – 12:15 pm	Welcome	Dr. Francis Collins, Director, NIH	
12:15 – 1:00 pm	Lunch		
	Group A*	Group B	
1:00 – 2:30 pm	Bioinformatics workshop (1-5 pm) - 5 th floor conference room, bldg. 50; Dr. Tyra Wolfsberg, Associate Director	Molecular Genetics	Dr. Suzanne Hart, Deputy Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
2:30 – 3:30 pm		Genetics of Genito-urinary Disorders	Dr. Melissa Merideth Attending Ob/Gyn and Geneticist Director, Medical Biochemical and Clinical Biochemical Training Programs
3.30 – 4:00pm		BREAK	
4:00 – 5:00 pm	Bioinformatics and Scientific Programming Core, NHGRI	How to Write a Scientific Paper	Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
5:00 – 5:15 pm	SURVEY		

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Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic		Speaker
Sept. 13th (Thurs)			
	Group A	Group B	
8:00 – 8:15 am	Survey	Survey	
8:15 – 9:45 am	Molecular Genetics	Bioinformatics workshop (8:30 am - 12:30 pm)- 5 th floor conference room, bldg. 50; Dr. Tyra Wolfsberg, Associate Director	Dr. Suzanne Hart, Deputy Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
9:45 – 10:45 am	Genetics of Genitourinary Disorders		Dr. Melissa Merideth Attending Ob/Gyn and Geneticist Director, Medical Biochemical and Clinical Biochemical Training Programs
10:45 – 11:15 am	Break		
11:15 – 12:15 pm	How to Write a Scientific Paper	Bioinformatics and Scientific Programming Core, NHGRI	Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
12:15 – 1:30pm			LUNCH
1:30 – 2:30 pm	Genetics of Diabetes and Insulin Resistance		Dr. Phillip Gorden Director Emeritus and Senior Investigator NIDDK
2:30 – 3:30 pm	Mitochondrial Genetics		Ms. Lynne Wolfe, Certified Registered Nurse Practitioner, NHGRI
3:30 – 4:00 pm			BREAK
4:00 – 5:30pm	Patient Panel		Moderator Mr. Don Hadley
5:30 – 5:45 pm			SURVEY

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 14th (Fri)		
7:45 – 8.00 am		SURVEY
8.00 – 12.00 pm	Grant Writing- Part 1	Dr. Michael Zigmond, Professor of Neurology, Neurobiology and Psychiatry, University of Pittsburgh
12:00 – 1:00 pm		LUNCH
1:00 - 2:00 pm	Small group discussion of specific aims)	
2.00 – 3.00 pm	Grant Writing- Part 2 (focusing on specific aims), Q & A	Dr. Michael Zigmond, Professor of Neurology, Neurobiology and Psychiatry, University of Pittsburgh
3:00 – 4:00 pm	Overview of Fogarty Extramural Programs	Dr. Ken Bridbord, Fogarty International Center/NIH
	The Emerging Global Leader Award (K43): A Career Development Award for LMIC Investigators	Dr. Christine Jessup, Fogarty International Center/NIH
4:00 – 4:30 pm		BREAK
4:30 – 5:30 pm	Life Sciences Research and Opportunities from USAID's US Global Development Lab	Jason Porter Senior Research Advisor, Center for Development Research, U.S. Global Development Lab, USAID
5:30 – 5:45 pm		SURVEY

Personalized Training at ICs

	Topic	Speaker
Sept. 17th– 20th (Mon-Thurs)		
8:00 am – 5:00 pm	Personalized Training at ICs	

Afternoon Activities at Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 21st (Fri)		
8:00 am – 12:30 pm	Personalized Training at ICs	
12:30 pm – 1:30 pm		LUNCH
1:30 pm – 2:00 pm		SURVEY
2:00 pm – 3:00 pm	Epigenetics	
3:00 pm – 4:00 pm	Skeletal Dysplasia	
4:00 pm – 4:15 pm		SURVEY

Meet at The Gateway Visitor's Center, NIH

	Topic	Speaker
Sept. 24th (Mon)		
8.00 – 8.15 am	Survey on the bus	
8.15 – 5.00 pm	<p>Field Trip- Children's National Health System 9 am- 12 pm 111 Michigan Ave NW, Washington, DC 20010</p> <p>MedStar Washington Hospital Center 12.30 pm- 4 pm 110 Irving St NW, Washington, DC 20010</p> <p>Survey on the bus</p>	

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 25th (Tues)		
8:00 – 8:15 am	SURVEY	
8:15 – 9:15 am	Mosaicism	Dr. Les Biesecker, Chief & Senior Investigator, Medical Genomics & Metabolic Genetics Branch, Head, Clinical Genomics Section, NHGRI
9:15 – 10:15 am	Trinucleotide Repeat Disorders	Dr. Kenneth (Kurt) Fischbeck NIH Distinguished Investigator, Neurogenetics Branch, NINDS
10.15 - 10.45 am	BREAK	
10.45 - 11.45 am	A Program of the National Center for Advancing Translational Sciences-The Genetic and Rare Diseases Information Center: 15 Years of Providing Access to Genetic and Rare Disease Information	Ms. Janine Lewis, Certified Genetic Counselor, Genetic and Rare Disease Information Center, NCATS Dr. Anne Pariser Director, Office of Rare Diseases Research NCATS
11.45 - 1.00 pm	LUNCH	
1:00 – 2:00 pm	Genetics of Pediatric Obesity	Dr. Jack A Yanovski. Chief, Section on Growth and Obesity Division of Intramural Research Eunice Kennedy Shriver National Institute of Child Health and Human Development
2:00 – 3:00 pm	Growing Success Stories of Genomics in African Ancestry Populations: H3 Africa and Beyond	Dr. Charles Rotimi, Director, Center for Research on Genomics and Global Health, Chief & Senior Investigator, Metabolic, Cardiovascular and Inflammatory Disease Genomics Branch, NHGRI
3:00 – 3:30 pm	BREAK	
3:30 – 4:30 pm	Genetics of Kidney Disorders	Dr. Meral Gunay-Aygun Associate Professor, JHU Staff Clinician, NHGRI
4:30 – 5:30 pm	Genetics of Craniofacial and Dental Anomalies	Dr. Janice Lee Clinical Director NIDCR
5:30- 5.45 pm	SURVEY	

Bldg. 45 (Natcher Conference Center), Room C1/C2

	Topic	Speaker
Sept. 26th (Wed)		
8:00 – 8:15 am		SURVEY
8:15 – 9:15 am	Neurocutaneous Syndromes	Dr. Dusica Babovic-Vuksanovic, Chair, Department of Clinical Genomics, Mayo Clinic
9:15 – 10:15 am	Auto-Inflammatory Disorders	Dr. Dan Kastner, Scientific Director, NHGRI
10:15 – 10:45 am		BREAK
10:45 – 11:45 am	Genetics of Complex Traits	Dr. Larry Brody, Director, Division of Genomics and Society, NHGRI
11:45 am – 1:00 pm		LUNCH
1:00 – 2:30 pm	Cancer Genetics	Ms. Tiffani DeMarco, Certified Genetic Counselor, Manager, Cancer Genetics Program, Division of Medical Genomics, INOVA
2:30 – 3:30 pm	Cancer Genomics	Dr. Kathy Calzone, Senior Nurse Specialist, Research, NCI
3:30 – 4:00 pm		BREAK
4:00 – 5:45 pm	Biochemical Genetics	Dr. Nick Ah Mew Medical Biochemical Geneticist CNHS
5:45 – 6:00 pm		SURVEY

Bldg. 45 (Natcher Conference Center), Room F1/F2

	Topic	Speaker
Sept. 27th (Thurs)		
8:00 – 8:15 am	SURVEY	
8:15 – 9:15 am	Societal Implications of Genomics	Dr. Dave Kaufman Program Director Division of Genomics and Society NHGRI
9:15 – 10:00 am	Harnessing the Power of “Big Data” to Improve Health and Healthcare	Dr. Nilay D. Shah, Chair, Division of Health Care Policy & Research, Department of Health Sciences Research; Associate Professor of Health Services Research, Mayo Clinic, Rochester, MN
10:00 – 10:30 am	BREAK	
10:30 – 11:15 am	The Sustainable Development Goals, Global Health Strategies, and Newborn Screening	Mr. Mark Engman, Senior Director, Public Policy and Advocacy, UNICEF USA
11:15 am – 12:00 pm	Introducing and Sustaining Newborn Screening in a Developing Nation	Dr. Bradford L. Therrell, Director, National Newborn Screening and Genetics Resource Center; International Newborn Screening Consultant
12:00 – 1:00 pm	LUNCH	
1:00 – 1:30 pm	Role of Professional Societies	Dr. Mike Watson, Executive Director, American College of Medical Genetics and Genomics
1:30 – 2:00 pm	The Translation of Research into Practice: Accelerating Implementation Through Newborn Screening Pilot Projects	Dr. Amy Brower, Newborn Screening Translational Research Network, American College of Medical Genetics and Genomics
2:00 – 2:30 pm	Leveraging Global Maternal Child Health Platforms	Dr. Linda Arnold, Chair of the American Academy of Pediatrics, Section on International Child Health
2:30 – 3:15 pm	Offline Wikipedia for Clinicians and Researchers	Lane Rasberry, Wikimedian in Residence, Data Science Institute at the University of Virginia. Dr. Samuel Zidovetzki
3:15 – 3:45 pm	BREAK	
3:45 – 4:30 pm	Building Medical Genetics Virtual Care & Capacity Building Network in the Developing World: reaching the target population	Dr. Saroj K. Mishra, Prof. & Head, Dept. of Endocrine Surgery; Nodal Officer SGPGI Telemedicine Program Sanjay Gandhi PG Inst. of Medical Sciences, Lucknow, India
4:30 – 5:30 pm	Challenges in setting up genetic services in low-resource settings	Dr. Carmencita D. Padilla, Professor and Chancellor University of the Philippines Manila
5:30 – 5:45 pm	SURVEY	

Bldg. 45 (Natcher Conference Center), Room A

	Topic	Speaker
Sept. 28th (Fri)		
8:00 – 9:00 am	Presentation by Candidates (4)	
9:00 – 10:00 am	Presentation by Candidates (4)	
10:00 - 10.15 am		BREAK
10.15 – 11:15 am	Presentation by Candidates (4)	
11:15 – 12:15 pm	Presentation by Candidates (4)	
12:15 – 1:15 pm		LUNCH
1:15 – 2:15 pm	Presentation by Candidates (4)	
2.15 – 3:15 pm	Presentation by Candidates (4)	
3:15 – 3:30 pm		BREAK
3.30 – 4:15 pm	Presentation by Candidates (3)	
4:15 – 5:00 pm	Graduation and Closing Remarks	Program Coordinators- Ms. Manjit Kaur, Program Administrator, NHGRI Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program; Senior Investigator and Chief, Medical Genetics Branch, NHGRI

Sept. 29th All candidates must depart