

**International Summit in Human Genetics and Genomics**  
**National Human Genome Research Institute**  
**National Institutes of Health**  
**September 1<sup>st</sup> – 30<sup>th</sup>, 2017**

**Agenda**

**Building 35A, Ground Floor, Room 610**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 1<sup>st</sup> (Fri)</b>		
8:00 – 9:00 am	Registration	Please bring Passports and visas
9:00 – 9:30 am	Welcome	Dr. Eric Green, Director, NHGRI Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
9:30 – 10:30 am	The Genomic Landscape Circa 2017	Dr. Eric Green, Director, NHGRI
10:30 – 10:45 am	Break	
10:45 – 11:00 am	Survey	
11:00 – 12:00 pm	Family History Collection & Assessment	Dr. Ben Solomon, Managing Director, GeneDx
12:00 – 1:00 pm	Lunch	
1:00 – 2:00 pm	Traditional & Non-traditional patterns of inheritance	Dr. Ben Solomon, Managing Director, GeneDx
2:00 – 3:00 pm	Dysmorphology	Dr. Ben Solomon, Managing Director, GeneDx
3:00 – 3:15 pm	Break	
3:30 – 4:30 pm	Clinical Center Tour	Mr. Donovan Kuehn, Lead Communication Specialist, Office of Communications and Media Relations NIH Clinical Center
4:30 – 4:45 pm	Survey	

**Building 45 (Natcher Conference Center), Room C1/C2**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 5<sup>th</sup> (Tues)</b>		
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	Prenatal Genetics	Dr. Huda Al-Kouatly, NHGRI
11.45 - 1.00 pm	Lunch	
1:00 – 2:00 pm	Genetics of Dental and Craniofacial Disorders	Dr. Janice Lee, Clinical Director, NIDCR
2:00 – 3:00 pm	Genetics of Connective Tissue Disorders	Dr. David Murdock, NHGRI
3:00 – 3:30 pm	Break	
3.30- 4:30 pm	Clinical Aspects of Cytogenetic Disorders	Dr. Clesson Turner, Clinical Geneticist, WRNMMC
4:30- 4.45 pm	Survey	

**Building 45 (Natcher Conference Center), Room C1/C2**

	<b>Topic</b>		<b>Speaker</b>
<b>Sept. 6<sup>th</sup> (Wed)</b>			
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	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
10:15 – 10:45 am	Break		
10:45 – 11:45 am	Trinucleotide Repeat Disorders		Dr. Kurt Fischbeck, NIH Distinguished Investigator, Neurogenetics Branch, NINDS
11:45 – 1:00pm	Lunch		
	Group A*	Group B	
1:00 – 2:00 pm	Bioinformatics workshop (1-5 pm) - 5 <sup>th</sup> floor conference room, bldg. 50; Dr. Tyra Wolfsberg, Associate Director Bioinformatics and Scientific Programming Core, NHGRI	Molecular Genetics	Dr. Suzanne Hart, Deputy Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
2:00 – 3:00 pm		Break	
3:00 – 3:30 pm		How to Write a Scientific Paper	Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
3:30 – 4:30pm		Survey	
4:30 – 4:45 pm			
5:00 – 5:15 pm	Survey		

**\*Manjit has list**

**Building 45 (Natcher Conference Center), Room C1/C2**

	<b>Topic</b>		<b>Speaker</b>
<b>Sept. 7<sup>th</sup> (Thurs)</b>			
	Group A	Group B	
8:00 – 8:15 am	Survey	Survey	
8:15 – 10:15 am	Molecular Genetics	Bioinformatics workshop (8:30 am - 12:30 pm)- 5 <sup>th</sup> floor conference room, bldg. 50; Dr. Tyra Wolfsberg, Associate Director Bioinformatics and Scientific Programming Core, NHGRI	Dr. Suzanne Hart, Deputy Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
10:15 – 11:00 am	Break		
11:00 – 12:00 pm	How to Write a Scientific Paper		Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
12:15 – 1:30pm	Lunch		
1:30 – 3:30 pm	Genetics of Hearing Loss and Deafness		Dr. Kathleen Arnos, Director of the Genetics Program, Gallaudet University
3:30 – 4:00 pm	Break		
4:00 – 5:00pm	A Clinical Approach to a Patient with a Suspected Genetic Disorder		Dr. Adebowale Adeyemo, Deputy Director, Center for Research in Genomic and Global Health, NHGRI
5:00 – 5:15 pm	Survey		

**Building 45 (Natcher Conference Center), Room C1/C2**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 8<sup>th</sup> (Fri)</b>		
7:45 – 8.00 am	Survey	
8.00 – 1.00 pm	Grant Writing	Dr. Beth Fischer, Assistant Professor of School of Education, University of Pittsburgh Dr. Michael Zigmond, Professor of Neurology, Neurobiology and Psychiatry, University of Pittsburgh
1:00 – 2:00 pm	Lunch	
2:00 - 3:30 pm	Overview of Fogarty Extramural Programs; K43 career development program for foreign scientists from eligible LMICs	Drs. Ken Bridbord and Christine Jessup, Fogarty International Center/NIH
3:30 – 4:30 pm	Seeding Labs: Catalyzing Research in Emerging Scientific Communities	Mr. Tim Fenton, Chair of the Board, Seeding Labs; & Thermo Fisher Scientific, Vice President, Global Government Relations
4:30 – 4:45 pm	Break	
4:45 – 5:45 pm	International Research Partnerships	Dr. Callie Raulfs- Wang- Program Manager, US Agency for International Development
5:45 – 6:00 pm	Survey	

**Building 45 (Natcher Conference Center), Room C1/C2**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 11<sup>th</sup> (Mon)</b>		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Variant Nomenclature	Dr. Christopher Lau, Clinical Molecular Geneticist, Undiagnosed Disease Program, NHGRI
9.15 - 10.15 am	Variant Interpretation	Dr. Christopher Lau, Clinical Molecular Geneticist, Undiagnosed Disease Program, NHGRI
10.15 - 10.45 am	Break	
10.45 - 11.45 am	NIH Undiagnosed Disease Program	Dr. Bill Gahl, Clinical Director & Head, Undiagnosed Disease Program, NHGRI
11.45 – 1.00pm	Lunch	
1.00 – 2.00 pm	Genetics of Genitourinary Disorders	Dr. Melissa Merideth, Attending Ob/Gyn and Geneticist; Director, Medical Biochemical and Clinical Biochemical Training Programs
2:00 – 3:00 pm	Exome Sequencing in the Clinical Lab	Dr. Jane Juusola, Director, Clinical Genomics Program, GeneDx
3:00 - 3.30 pm	Break	
3.30 - 4.30 pm	Genetic Counseling	Mr. Don Hadley, Certified Genetic Counselor, Deputy Director, Medical Genetics & Genomic Medicine Residency, NHGRI
4.30 – 4:45 pm	Survey	

**Meet at The Gateway Visitor's Center, NIH**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 12<sup>th</sup> (Tues)</b>		
8.00 – 8.15 am	Survey on the bus	
8.15 – 4.00 pm	Field Trip- MedStar Washington Hospital Center  Children's National Health System  Survey on the bus	

**Building 45 (Natcher Conference Center), Room C1/C2**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 13<sup>th</sup> (Wed)</b>		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Mitochondrial Genetics	Ms. Lynne Wolfe, Certified Registered Nurse Practitioner, NHGRI
9.15 - 10.15 am	Neurocutaneous Syndromes	Dr. Dusica Babovic-Vuksanovic, Chair, Dept of Clinical Genomics, Mayo Clinic
10.15 - 10.45 am	Break	
10.45 - 11.45 am	Noninvasive Prenatal Genomics: The Vanguard of Genomic Medicine	Dr. Diana W. Bianchi, Director, <i>Eunice Kennedy Shriver</i> , National Institute of Child Health and Human Development
11.45 – 1.00pm	Lunch	
1.00 – 3:00 pm	Cancer Genetics	Ms. Tiffani DeMarco, Certified Genetic Counselor, Manager, Cancer Genetics Program, Division of Medical Genomics, INOVA
3:00 - 3.30 pm	Break	
3.30 – 4:30 pm	Cancer Genomics	Dr. Kathy Calzone, Senior Nurse Specialist, Research, NCI
4:30 – 5:30 pm	Genetic Disorders of the Heart: Structural Defects	Dr. Paul Kruszka, Staff Clinician, NHGRI
5:30 – 5.45 pm	Survey	

**Building 45 (Natcher Conference Center), Room C1/C2**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 14<sup>th</sup> (Thurs)</b>		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Biochemical Genetics: Large Molecules	Dr. Carlos Ferreira, Medical Biochemical Geneticist, CNHS
9:15 – 10:15 am	Biochemical Genetics: Small Molecules	Dr. Carlos Ferreira, Medical Biochemical Geneticist, CNHS
10.15 - 10.45 am	Break	
10.45 - 11.45 am	Genetics of Kidney Disorders	Dr. Meral Gunay-Aygun, Associate Professor, JHU, Staff Clinician, NHGRI
11.45 – 1.00pm	Lunch	
1:00 – 3:00 pm	Neurogenetics	Dr. Andrea Gropman, Division Chief, Neurodevelopmental Disabilities & Neurogenetics, CNHS
3:00 - 3.30 pm	Break	
3.30 – 3.45 pm	Survey	

**Meet at The Gateway Visitor's Center, NIH**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 15<sup>th</sup> (Fri)</b>		
9.15 - 9.30 am	Survey on the bus	
9.30 – 4.00 pm	Field Trip-  NIH Intramural Sequencing Center  GeneDx	

**Advanced Training at ICs**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 18<sup>th</sup>, 20<sup>th</sup> – 22<sup>nd</sup> (Mon, Wed-Fri)</b>		
8.00 - 5.00 pm	Advanced Training at ICs	

**Meet at The Gateway Visitor's Center, NIH**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 19<sup>th</sup> (Tues)</b>		
8.00 – 8.15 am	Survey on the bus	
8.15 – 4.00 pm	Field Trip-  Johns Hopkins University  Kennedy Krieger Institute	



**Building 35A, Ground Floor, Room 610**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 25<sup>th</sup> (Mon)</b>		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Auto-Inflammatory Disorders	Dr. Dan Kastner, Scientific Director, NHGRI
9.15 – 9:45 am	Sickle Cell Disease, The Global Health Burden	Dr. Vence Bonham, Senior Advisor to the NHGRI Director on Genomics and Health Disparities, NHGRI
9:45 – 10:15 am	Examining Sickle Cell Disease Leg Ulcers Through a Skin Microbiome Lens	Anitra Persaud, B.S., NHGRI
10.15 - 10.45 am	Break	
10.45 - 11.45 am	Sickle Cell Disease, Bone Marrow Transplant and Gene Therapy	Matthew Hsieh, MD, Staff Clinician, NHLBI
11.45 – 1.00 pm	Lunch	
1:00 – 2:00 pm	Ophthalmic Genetics	Dr. Brian Brooks, Clinical Director, NEI Chief, Unit on Pediatric, Developmental & Genetic Ophthalmology
2:00 – 3:00 pm	Genetics of Skin Disorders, Part I	Dr. Gabriele Richard, Chief Medical Officer, GeneDx
3:00 - 3.30 pm	Break	
3.30 – 4:30 pm	Genetics of Skin Disorders, Part II	Dr. Gabriele Richard, Chief Medical Officer, GeneDx
4:30 – 4.45 pm	Survey	

**Building 35A, Ground Floor, Room 610**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 26<sup>th</sup> (Tues)</b>		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Nurse Scientists: Moving Precision Medicine Forward	Dr. Katy Meilleur, Certified Registered Nurse Practitioner, Assistant Clinical Investigator, Tenure Track Investigator, NINR
9.15 - 10.15 am	Neuromuscular Disease	Dr. Carsten Bonnemann, Senior Investigator, Neuromuscular & Neurogenetic Disorders of Childhood Section, 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. Henrietta Hyatt-Knorr Senior Program and Policy Analyst, Genetic and Rare Disease Information Center  Ms. Janine Lewis, Certified Genetic Counselor, Genetic and Rare Disease Information Center
11.45 – 1.00pm	Lunch	
1.00 – 2:00 pm	What Makes Research Ethical?	Mr. Ben Berkman, Deputy Director, Bioethics Core, NHGRI
2:00 – 3:00 pm	Ethical Issues in Genetic Research with Stored Samples	Dr. Sara Hull, Director, Bioethics Core, Office of the Clinical Director, NHGRI
3:00 - 3.30 pm	Break	
3.30 – 4:45 pm	Patient Panel	Led by Don Hadley, CGC
4:45 – 5:00 pm	Survey	

**Building 35A, Ground Floor, Room 610**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 27<sup>th</sup> (Wed)</b>		
8.00 - 8.15 am	Survey	
8.30 - 9.00 am	Welcome	Dr. Francis Collins, Director, National Institutes of Health
9.15 - 10.15 am	Mosaicism	Dr. Les Biesecker, Chief & Senior Investigator, Medical Genomics & Metabolic Genetics Branch, Head, Clinical Genomics Section, NHGRI
10.15 - 10.45 am	Break	
10.45 - 11.45 am	Inherited Pulmonary Disorders	Dr. Bernadette Gochuico, Attending Pulmonologist, Medical Genetics Branch, NHGRI
11.45 – 1:00pm	Lunch	
1:00 – 2:00 pm	Genetics of Lipid Disorders	Dr. Robert Shamburek, Clinician/Scientist, Lipid Section, Cardiovascular and Pulmonary Branch, NHLBI
2:00 – 3:00 pm	Genetics of Complex Traits	Dr. Larry Brody, Director, Division of Genomics and Society, NHGRI
3:00 - 3.30 pm	Break	
3.30 – 4:30 pm	Genetic Databases (ClinVar, dbGAP, GTR)	Ms. Adriana Malheiro, Staff Scientist, Director, NIH Genetic Testing Registry, NCBI/NLM
4:30 – 4.45 pm	Survey	

**Building 35A, Ground Floor, Room 610**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 28<sup>th</sup> (Thurs)</b>		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Genetic syndromes in diverse populations	Dr. Paul Kruszka, Staff Clinician, NHGRI
9.15 - 10.15 am	Metabolic Medicine: Beyond the Ivory Towers of North America	Dr. Mark Korson Medical Director, Genetic Metabolic Center for Education
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Making Newborn Screening Work in a Challenging Environment	Dr. Bradford L. Therrell, Director, National Newborn Screening and Genetics Resource Center; International Newborn Screening Consultant
11.30 – 12.30pm	SDGs, Global Health, and Newborn Screening	Mr. Mark Engman, Senior Director, Public Policy and Advocacy, UNICEF USA
12.30 – 1.30 pm	Lunch	
1.30 - 2.00 pm	Role of Professional Societies	Dr. Mike Watson, Executive Director, American College of Medical Genetics and Genomics
2.00 - 2.30 pm	Translating Research into Practice: Establishing Newborn Screening Pilot Projects	Dr. Amy Brower, Newborn Screening Translational Research Network, American College of Medical Genetics and Genomics
2.30 - 3.00 pm	Leveraging Global Maternal Child Health Platforms	Dr. Linda Arnold, Chair of the American Academy of Pediatrics, Section on International Child Health
3.00 – 3:30 pm	Break	
3:30 – 4:30 pm	Challenges in setting up genetic services in low-resource settings	Dr. Carmencita D. Padilla, Professor and Chancellor University of the Philippines Manila
4:30 - 4:15 pm	Survey	

**Building 35A, Ground Floor, Room 610**

	<b>Topic</b>	<b>Speaker</b>
<b>Sept. 29<sup>th</sup> (Fri)</b>		
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 Specialist, NHGRI  Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program; Senior Investigator and Chief, Medical Genetics Branch, NHGRI

**Sept. 30<sup>th</sup>**      All candidates must depart