

International Summit in Human Genetics and Genomics
National Human Genome Research Institute
National Institutes of Health
September 1st – 30th, 2016

Agenda

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 1st		
8.00 - 9.00 am	Registration	Please bring Passports and visas
9.00 - 9.30 am	Welcome	Dr. Eric Green, Director, NHGRI Dr. Dan Kastner, Scientific Director, NHGRI Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program; Senior Investigator and Chief, Medical Genetics Branch, NHGRI
9.30 - 10.30 am	The Genomic Landscape Circa 2016	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, Chief, Div. of Medical Genomics, INOVA
12.00 - 1.00 pm	Lunch	
1.00 - 2.00 pm	Traditional & Non-traditional patterns of inheritance	Dr. Ben Solomon, Chief, Div. of Medical Genomics, INOVA
2.00 - 3.00 pm	Dysmorphology	Dr. Ben Solomon, Chief, Div. of Medical Genomics, INOVA
3.00 - 3.15 pm	Break	
3.15 - 4.45 pm	Most Common Syndromes and International Atlas	Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program; Senior Investigator and Chief, Medical Genetics Branch, NHGRI
4.45 - 5.00 pm	Survey	
6.00 - 7.30 pm	Welcome reception at the DoubleTree Hotel	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 2nd		
8.00 - 8.15 am	Survey	
8.15 - 10.15 am	Tools of Molecular Genetics	Dr. Suzanne Hart, Deputy Director, Medical Genetics & Genomic Medicine Training Program, NHGRI
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Variant Nomenclature	Dr. Christopher Lau, Clinical Molecular Geneticist, Undiagnosed Disease Program, NHGRI
11.30 - 12.30 pm	Variant Interpretation	Dr. Christopher Lau, Clinical Molecular Geneticist, Undiagnosed Disease Program, NHGRI
12.30 - 1.30 pm	Lunch	
1.30 - 2.30 pm	Biochemical Genetics: Small Molecules	Dr. Carlos Ferreira, Medical Biochemical Geneticist, Children's National Health System (CNHS)
2.30 - 3.30 pm	Biochemical Genetics: Large Molecules	Dr. Carlos Ferreira, Medical Biochemical Geneticist, CNHS
3.30 - 4.00 pm	Survey	

Meet at The Gateway Visitor's Center, NIH

	Topic	Speaker
Sept. 6th		
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 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 7th		
8.00 - 8.15 am	Survey	
8.15 - 10.15 am	Neurogenetics	Dr. Andrea Gropman, Div. Chief Neurodevelopmental Disabilities & Neurogenetics, CNHS
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Ophthalmic Genetics	Dr. Brian Brooks, Chief, Unit on Pediatric, Developmental & Genetic Ophthalmology, NEI
11.30 - 12.30 pm	NIH Undiagnosed Disease Program	Dr. Bill Gahl, Clinical Director & Head, Undiagnosed Disease Program and Human Biochemical Genetics Program, NHGRI
12.30 - 1.30 pm	Lunch	
1.30 - 2.30 pm	Cytogenetics I	Dr. Ludmila Matyakhina, Director, Cytogenetics, GeneDx
2.30 - 3.30 pm	Cytogenetics II	Dr. Ludmila Matyakhina, Director, Cytogenetics, GeneDx
3.30 - 3.45 pm	Break	
3.45 - 4.45 pm	Clinical Aspects of Cytogenetic Disorders	Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program; Senior Investigator and Chief, Medical Genetics Branch, NHGRI
4.45 - 5.00 pm	Survey	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 8th		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Neuromuscular Disease	Dr. Carsten Bonnemann, Senior Investigator, Neuromuscular & Neurogenetic Disorders Of Childhood Section, NINDS
9.15 - 10.15 am	Genetic Databases (ClinVar, dbGap, GTR)	Dr. Wendy Rubinstein, Senior Scientist, Director, NIH Genetic Testing Registry, NCBI/NLM
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Trinucleotide Repeat Disorders	Dr. Kurt Fischbeck, NIH Distinguished Investigator, Neurogenetics Branch, NINDS
11.30 - 12.30 pm	Genetics Disorders of the Heart: Structural Defects	Dr. Paul Kruszka, Staff Clinician, NHGRI
12.30 - 1.30 pm	Lunch	
1.30 - 3.30 pm	Genetics of Hearing Loss and Deafness	Dr. Kathleen Amos, Director of the Genetics Program, Gallaudet University
3.30 - 3.45 pm	Break	
3.45 - 4.45 pm	Genetics of Complex Traits	Dr. Lawrence Brody, Director, Division of Genomics and Society, NHGRI
4.45 - 5.00 pm	Survey	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 9th		
8.00 - 8.15 am	Survey	
8.15 - 10.15 am	Genetics Skin Disorders	Dr. Gabriele Richard, Chief Medical Officer, GeneDx
10.15 - 10.30 am	Break	
10.30 - 12.30 am	Cancer Genetics	Ms. Tiffani DeMarco, Certified Genetic Counselor, Manager, Cancer Genetics Program, Division of Medical Genomics, INOVA
12.30 - 1.30 pm	Lunch	
1.30 - 2.30 pm	Genetics of Dental and Craniofacial Disorders	Dr. Janice Lee, Clinical Director, NIDCR
2.30 - 3.30 pm	Cleft lip and palate	Dr. Sophia Hufnagel, Assistant Professor, CNHS
3.30 - 4.00 pm	Survey	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 12th		
8.00 - 8.15 am	Survey	
8.15 - 10.15 am	Sickle Cell Disease, The Global Health Burden	Dr. Caterina Minniti-Crouch, Senior Advisor to the NHGRI Director on Genomics and Health Disparities Dr. Vence Bonham, Senior Advisor to the NHGRI Director on Genomics and Health Disparities, NHGRI
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Prenatal Genetics	Dr. Huda Al-Kouatly, Genetics Fellow, NHGRI
11.30 - 12.30 pm	Neurocutaneous Syndromes	Dr. Dusica Babovic-Vuksanovic Chair, Dept. of Clinical Genomics, Mayo Clinic
12.30 - 1.30 pm	Lunch	
1.30 - 2.30 pm	Working up Undiagnosed Cases	Dr. Dusica Babovic-Vuksanovic Chair, Dept. of Clinical Genomics, Mayo Clinic Dr. Eric Klee Assoc. Program Director, Bioinformatics, Mayo Clinic
2.30 - 3.30 pm	Lipid Disorders	Dr. Robert Shamburek, Clinician/Scientist, Lipid Section, Translational Medicine Branch, NHLBI
3.30 - 3.45 pm	Break	
3.45 - 4.45 pm	Nurse Scientists: Moving Precision Medicine Forward	Dr. Ann Cashion, Scientific Director, NINR Dr. Katy Meilleur, CRNP, Assistant Clinical Investigator, Tenure Track Investigator, NINR
4.45 - 5.00 pm	Survey	

Building 45 (Natcher Conference Center), Room G1/G2

	Topic	Speaker
Sept. 13th		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Genetic Counseling	Mr. Don Hadley, Genetic Counselor, Deputy Director, Medical Genetics & Genomic Medicine Residency, NHGRI
9.15 - 10.15 am	Newborn Screening	Dr. Debra Regier, Assistant Professor, Director of Genetic and Genomic Education, CNHS
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Kidney Disorders	Dr. Meral Gunay-Aygun, Associate Professor, JHU Staff Clinician, NHGRI
11.30 - 12.30 pm	Lunch	
12.30 - 1.00 pm	Walk to Building 50	
1.00 - 3.00 pm	Bioinformatics Workshop Part 1	Dr. Baxevanis, Director, Bioinformatics and Scientific Programming Core, NHGRI
3.00 - 3.15 pm	Break, Bldg. 50, 5 th floor lobby	
3.15 - 5.00 pm	Bioinformatics Workshop Part 2	Dr. Baxevanis, Director, Bioinformatics and Scientific Programming Core, NHGRI
5.00 - 5.15 pm	Survey	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 14th		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	Whole Exome Sequencing in a Clinical Setting	Dr. Sherri Bale, Managing Director, GeneDx and Senior Vice President, BioReference Laboratories
9.15 - 10.15 am	Mitochondrial Disorders	Ms. Lynne Wolfe, CRNP, NHGRI
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Auto-inflammatory Disorders	Dr. Dan Kastner, Scientific Director, NHGRI
11.30 - 12.30 pm	Genetics of Genitourinary Disorders	Dr. Melissa Merideth, Attending Ob/Gyn & Geneticist; Director, Clinical and Medical Biochemical Genetics Training, NHGRI
12.30 - 1.30 pm	Lunch	
1.30 - 2.30 pm	Reducing barriers to healthcare in underserved populations and minorities	Dr. Eliseo Perez-Stable, Director, NIMHD
2.30 - 3.30 pm	Inherited Pulmonary Disorders	Dr. Bernadette Gochuico, Attending Pulmonologist, NHGRI
3.30 - 4.00 pm	Break	
4.00 - 5.00 pm	How to Write a Scientific Paper	Dr. Max Muenke, Director, Medical Genetics & Genomic Medicine Training Program; Senior Investigator and Chief, Medical Genetics Branch, NHGRI
5.00 - 5.30 pm	Break	
5.30 - 7.00 pm	Connective Tissue Disorders	Dr. Howard Levy, Associate Professor, JHU
7.00 - 7.15 pm	Survey	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 15th		
8.00 - 8.15 am	Survey	
8.15 - 10.15 am	A Clinical Approach to a Patient with A Suspected Genetic Disorder	Dr. Adebowale Adeyemo, Deputy Director, Center for Research in Genomics and Global Health, NHGRI
10.15 - 10.30 am	Break	
10.30 - 11.30 am	Mosaicism	Dr. Leslie Biesecker, Chief & Senior Investigator, Medical Genomics & Metabolic Genetics Branch Head, Clinical Genomics Section, NHGRI
11.30 - 12.30 pm	Genetics & Bayesian Inference Testing	Dr. Leslie Biesecker, Chief & Senior Investigator, Medical Genomics & Metabolic Genetics Branch Head, Clinical Genomics Section, NHGRI
12.30 - 1.30 pm	Lunch	
1.30 - 3.30 pm	Growing Success Stories of Genomics in African ancestry populations: H3Africa 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
3.30 - 3.45 pm	Break	
3.45 - 5.00 pm	Patient Panel	Mr. Don Hadley, Genetic Counselor, Deputy Director, Medical Genetics & Genomic Medicine Residency, NHGRI
5.00 - 5.15 pm	Survey	

Meet at The Gateway Visitor's Center, NIH

	Topic	Speaker
Sept. 16th		
9.00 - 9.15 am	Survey on the bus	
9.15 - 4.00 pm	Field Trip- NIH Intramural Sequencing Center GeneDX Survey on the bus	

Advanced Training at ICs

	Topic	Speaker
Sept. 19th		
8.00 - 5.00 pm	Advanced Training at ICs	

Meet at The Gateway Visitor's Center, NIH

	Topic	Speaker
Sept. 20th		
8.00 - 8.15 am	Survey	
8.15 - 4.00 pm	Field Trip- Johns Hopkins University	

Advanced Training at ICs

	Topic	Speaker
Sept. 21st – 23rd, Sept. 26th		
8.00 - 5.00 pm	Advanced Training at ICs	

Meet at The Gateway Visitor's Center, NIH

	Topic	Speaker
Sept. 27th		
12.00 – 4.00 pm	Field Trip- NIH Clinical Center Kennedy Krieger Institute	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 28th		
8.00 - 8.15 am	Survey	
8.30 - 9.00 am	Welcome	Dr. Francis Collins, Director, National Institutes of Health
9.00 - 10.00 am	What Makes Research Ethical?	Mr. Ben Berkman, Deputy Director, Bioethics Core, NHGRI
10.00 - 10.30 am	Break	
10.30 - 11.30 am	Ethical Issues in Genetic Research with Stored Samples	Dr. Sara Hull, Director, Bioethics Core, Office of the Clinical Director, NHGRI
11.30 - 12.30 pm	Role of Professional Societies	Dr. Mike Watson, Executive Director, American College of Medical Genetics and Genomics
12.30 - 1.15 pm	Lunch	
1.15 - 2.15 pm	Sustainable Developmental Goals And Global Health Platforms Leveraging Global Maternal Child Health Platforms	Mr. Mark Engman, Senior Director, Public Policy and Advocacy, U.S. Fund for UNICEF Dr. Linda D. Arnold, Chair of the American Academy of Pediatrics, Section on International Child Health
2.15 - 3.15 pm	A program of the National Center for Advancing Translational Sciences - The Genetic and Rare Diseases Information Center: 14 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
3.15 - 3.30 pm	Break	
3.30 - 4.30 pm	Essential Roles Of Parent-Advocates In Building And Supporting Newborn Screening Programs Prevention and Care of Congenital and Genetic Disorders in High Burden Countries: The Need for New Strategies in the SDG era	Ms. Jill Levy-Fisch, President, Save Babies Through Screening Foundation Dr. Christopher P. Howson, Vice-President Global Programs, March of Dimes Foundation
4.30 - 4.45 pm	Survey	

Building 31, C wing, 6th Floor, Room 5

	Topic	Speaker
Sept. 29th		
8.00 - 8.15 am	Survey	
8.15 - 9.15 am	The World Of Newborn Screening: The System And Its Global Impact	Dr. Bradford L. Therrell, Director, National Newborn Screening and Genetics Resource Center; International Newborn Screening Consultant
9.15 - 10.15 am	Translating Research into Practice: Establishing Newborn Screening Pilot Projects	Dr. Amy Bower, National Coordinating Center, Newborn Screening Translational Research Network, American College of Medical Genetics
	Newborn Screening in the Philippines: A Successful Model in a Developing Country	Dr. Bradford L. Therrell, Director, National Newborn Screening and Genetics Resource Center; International Newborn Screening Consultant
10.15 - 10.30 am	Break	
10.30 - 10.45 am	Survey	
10.45 - 11.45 am	Presentations by candidates (4)	
11.45 - 12.30 pm	Lunch	
12.30 - 1.30 pm	Presentations by candidates (4)	
1.30 - 2.30 pm	Presentations by candidates (4)	
2.30 - 3.15 pm	Presentations by candidates (3)	
3.15 - 3.30 pm	Break	
3.30 - 4.15 pm	Presentations by candidates (3)	
4.15 - 4.45 pm	Closing Remarks	Program Coordinator- Ms. Manjit Kaur, Program Specialist, NHGRI

Sept. 30th All candidates must depart

Agenda is subject to change every year.