**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, 5thfloor 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  Newborn Screening in the Philippines: A Successful Model in a Developing Country | Dr. Amy Bower, National Coordinating Center, Newborn Screening Translational Research Network, American College of Medical Genetics  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.