Genomic Medicine Symposium Schedule
Friday, September 6, 2019 (cs-8814)

7:00-7:50 **Registration and Continental Breakfast**

7:50-8:00 **Welcome and Review of the Day: Dr. Gene Hoyme**

8:00-9:00 **KEYNOTE SPEAKER: 100,000 Genomes Project – Transforming Healthcare: Professor Sir Mark Caulfield, MD, FRCP, FESC, FBHS, FMedSci, Genomics England and Queen Mary University of London; Chief Executive and Chief Scientist for Genomics England**

Mark Caulfield graduated in Medicine in 1984 from the London Hospital Medical College and trained in Clinical Pharmacology at St Bartholomew's Hospital where he developed a research program in molecular genetics of hypertension and translational clinical research.

In 2007, 2009 and 2011 his research has been independently rated among the top ten scientific discoveries in his field. In 2009 he won the Lily Prize of the British Pharmacology Society, in 2015 he won the Genome Valley Award at BioAsia and in 2016 the Bjorn Folkow Award of the European Society of Hypertension. Since 2008, he has directed the NIHR Biomedical Research Centre at Barts. He was appointed Director of William Harvey Research Institute in 2002, was elected to the Academy of Medical Sciences in 2008, and was President of the British Hypertension Society (2009-2011).

He is an NHS consultant in the Barts Blood Pressure Clinic within the Barts/William Harvey European Society of Hypertension Centre of Excellence. He raised £25m toward the William Harvey Heart Centre which created a translational clinical research center and was the academic leader that created the Barts Heart Centre bringing 3 hospitals together in 2015 to create the UK's largest heart center (includes UCLH Heart Hospital, the London Chest Hospital and Barts). He served on the 2011 NICE Guideline Group for hypertension and leads the Joint UK Societies' Working Group and Consensus on Renal Denervation. Since 2014, he has been one of the top 200 most highly cited researchers in the world in genomics according to Thomson Reuters. In 2013, he became an NIHR Senior Investigator.

In 2013 he became an NIHR Senior Investigator and was appointed as the Chief Scientist for Genomics England (100,000 Genomes Project). He was appointed Interim Chief Executive for Genomics England in January 2019. Sir Mark was awarded a Knighthood in the June 2019 Queen's Birthday Honours List for services to the 100,000 Genomes Project.

9:00-10:00 **Genomic Determinants of Macrocephaly and its Association with Autism, Intellectual Disability, and Cancer Predisposition: Julian Martinez-Agosto, MD, PhD, FACMG, Department of Human Genetics, Division of Medical Genetics, Department of Pediatrics, Department of Psychiatry, Semel Institute for Neuroscience and Human Behavior, David Geffen School of Medicine at UCLA; Director, Developmental Neurogenetics Clinic, Co-Director, Pediatric Cancer Predisposition Clinic, UCLA Undiagnosed Diseases Program**

Dr. Martinez-Agosto is Associate Professor with tenure at the University of California Los Angeles with a laboratory focused on dissecting the role of molecular pathways in regulating progenitor and organism growth. His laboratory utilizes model organisms and human genetic disorders associated with overgrowth/undergrowth to identify new genes and characterize signaling pathways regulating tissue growth. Their translational studies focus on identifying the genetic basis of autism and disorders that affect brain growth. He and his colleagues have described novel syndromes, extended the phenotypic findings for established ones, and identified their genetic basis. Using next generation sequencing technologies and modeling of human disease-associated genetic variants in *Drosophila*, they dissect the mechanistic basis for these genetic disorders and model these rare human genetic disorders by using gene editing technologies in human neural stem cells and their progeny.

10-10:15 **BREAK**

10:15-11:15 **The Genetic Basis of Mendelian Conditions: Discoveries, Challenges and Opportunities: Michael Bamshad, MD, Professor and Chief, Division of Genetic Medicine, Department of Pediatrics, Seattle Children's and the University of Washington School of Medicine**

Michael J. Bamshad, MD, is Professor and Chief of Genetic Medicine in the Department of Pediatrics at the University of Washington. He also holds the Allan and Phyllis Treuer Endowed Chair in Genetics and Development. He has studied rare genetic conditions for more than 25 years and he and his colleagues at the University of Washington were the first scientists to use exome sequencing to discover the genetic basis of a rare condition: "Exome sequencing identifies the cause of a mendelian disorder" (2010). He is a principal investigator of the University of Washington Center for Mendelian Genomics, part of a NIH program to identify the genes underlying all rare, Mendelian conditions and has developed innovative ways to share data (i.e., MyGene2) and manage return of genetic testing results (i.e., My46) from exome and whole genome sequencing in both research and clinical settings. He co-authors a popular textbook entitled *Medical Genetics*.

11:15-12:15 **Implementation of a Clinical Pharmacogenomics Service in a Pediatric Health-System: David Gregornik, BA, BS, PharmD, Medical Genetics and Genomics, Children's Minnesota**

David Gregornik is the Pharmacogenomics Program Director at Children's Minnesota. Dr. Gregornik attended the University of Minnesota College of Pharmacy where he earned his Bachelor of Science in Pharmacy and his PharmD. Under the direction of John Rodman, PharmD, he completed a 12 month post-doctoral training program at St. Jude Children's Research Hospital in Memphis TN. Dr. Gregornik was a member of the St Jude Pharmaceutical Department for a total of 18 years providing advanced clinical pharmacy care to children with cancer and undergoing bone marrow transplant. From 2009-2013 David moved to Memorial-Sloan Kettering Cancer Center, in New York City where he served as Manager of Pediatric Clinical Pharmacy Programs. Dr. Gregornik joined Children's Minnesota in August 2016 where he has established a Clinical Pharmacogenomics consult service and specialty clinic. The goal of the Pharmacogenomics Program at Children's Minnesota is to expand the use of genomic medicine in children to optimize medication use, prevent adverse drug reactions and avoid treatment failures.

12:15-1:15 **LUNCH**

1:15-2:15 **Gene Transfer and Genome Editing for Treating Human Disease: Mark A. Kay, MD, PhD, Dennis Farrey Family Professor, Departments of Pediatrics and Genetics, Associate Chair for Basic Research (Pediatrics), Stanford University School of Medicine**

Mark Kay, MD, PhD is the Dennis Farrey Family Professor in Pediatrics and Professor of Genetics. He is the head of the Division of Human Gene Therapy at Stanford University. His work spans basic discovery through clinical trials. Over the years, he has focused on gene therapy vector development and new genome editing approaches. He has made seminal contributions in RNAi based therapeutics. His lab has a general interest in how non-coding RNAs regulate the gene expression and has recently started to purse a novel class of tRNA, derived small RNAs, and their potential target for cancer therapeutics.
Dr. Kay has received many awards and has served on many national/international committees. Notably, he was a founding board member of The American Society for Gene and Cell Therapy and served as its President in 2004-2006. He also served on the Board of Directors of the Oligonucleotide Therapeutic Society for 10 years.

2:15-3:15 **Ethical Issues in the Era of Genomic Medicine: Curtis R. Coughlin II, M.S., M.Be., CGC, Associate Professor, Department of Pediatrics, Associate Faculty, Center for Bioethics and Humanities, University of Colorado Anschutz Medical Campus**

Curtis Coughlin II is a genetic counselor in the Section of Clinical Genetics and Metabolism where he focuses on the treatment of metabolic encephalopathies and clinical applications of genomic sequencing. He received his ethics training at the University of Pennsylvania.

Since joining the faculty at the University of Colorado, he has served as a member of the ethics consultation team at the Children's Hospital of Colorado (2011-present). His main interest is ethical issues within the field of genetics. He has served as the chair of the ethics advisory group for the National Society of Genetic Counselors (2010-2015) and as the ethics ad hoc content expert for Colorado Department of Public Health and Environment's Newborn Screening Program (2015-present).

His current research interest includes the return of results and secondary findings identified through genomic sequencing, and he is currently a member of the Clinical Genetics Resource's (ClinGen) Consent and Disclosure Recommendation Committee (2014-present).

3:15-3:30 **BREAK**

3:30-4:30 **The Emerging Role of Chromosomal Copy Number Variation in Clinical Psychiatry: David Isum Ward, MD, Assistant Professor, Department of Pediatrics, the University of South Dakota Sanford School of Medicine; Clinical Geneticist, Sanford Imagenetics and Sanford Children's Hospital, Sioux Falls**

D. Isum Ward, MD, provides a variety of services in medical genetics. He commonly treats metabolic disease, and offers individual care for those with rare conditions. His services include genetic evaluation of all health conditions and diagnostic evaluation of rare and undiagnosed conditions. Dr. Ward serves as an Assistant Professor in the Department of Pediatrics of the University of South Dakota Sanford School of Medicine. His current research interest includes the principal investigator in "The Attention Deficit Hyperactivity Disorder Family Study. He is also a participating investigator in "Achondroplasia Behavior Study" and the "Post-Traumatic Stress Disorder Study."

4:30-4:40 **Wrap-Up and Summary: Dr. Gene Hoyme, Sanford Health**