CSCB Seminar Series

DUKE NUS GRADUATE MEDICAL SCHOOL SINGAPORE

Title:

"Modeling the Morbid Human Genome."

Abstract:

Genome-wide sequencing is emerging as a powerful tool as a first-pass diagnostic test. This has heightened the need for interpretive assays to determine the pathogenic potential of genetic variation. To address these challenges, and to capitalize on the opportunity to shorten the time to secure molecular diagnoses, we have created the Task Force for Neonatal Genomics at Duke University. The Task Force targets a uniquely vulnerable patient cohort: infants and neonates whose developmental anomalies are more likely to be within a timeframe for treatment. Our efforts harness the full spectrum of clinical, genetics and cellular biology expertise, including the use of transient model organisms (primarily zebra fish). I will discuss: 1) the interdisciplinary nature of our efforts; 2) our methodology for recruitment, data generation and analysis, and communication strategies between researchers and clinicians; 3) our analysis progress to date; and 4) our evolving approach to returning primary and secondary molecular findings to clinicians and family members. In phenotype-appropriate patients, we couple whole Exome sequencing of trios, a multi-tiered bioinformatics prioritization strategy, and functional modeling of novel variants in physiologically relevant vertebrate and cell-based models to inform allele pathogenicity. Strikingly, in our first year, we have achieved definitive diagnoses for 41% of our patients and strong candidate diagnoses that typically involved novel disease genes and/or complex genetic interactions in 78% of our cohort. This initiative provides an unprecedented model for communication across an interdisciplinary research/clinical team with the ultimate goal of responsible and timely integration of new genetic technologies into clinical care.

Date:

14 January 2013 (Tuesday)

Time:

12:00 PM to 1:00 PM

Venue:

Amphitheatre, Level 2
Duke-NUS Grad Med Sch
8 College Road, S169857

(Opposite Singapore General Hospital, Block 6/7)

Host:

David Virshup, M.D.
Professor & Director
Program in Cancer & Stem Cell Biology
Duke-NUS Graduate medical School Singapore

"No registration is required."
Any enquiry, pls contact:
Jamie Liew (Tel: 6516 6954)

Speaker:

Nicholas Katsanis, Ph.D.
Director
Center for Human Disease Modeling
Jean and George W. Brumley Professor of Cell Biology and Pediatrics

Biography:

Dr. Katsanis obtained his first degree in Genetics from UCL in London in 1993 and his doctorate from Imperial College, University of London in 1997. He then joined the laboratory of Dr. Lupski at Baylor College of Medicine, where he initiated his studies on Bardet-Biedl syndrome. In 2002, he relocated to the Institute of Genetic Medicine, Johns Hopkins University where he led studies that unified several allied conditions under the ciliopathy umbrella. In 2009, he moved to Duke University to establish the Center for Human Disease Modeling, where he is the Director; this new structure aims to facilitate collaboration across disciplines and to develop physiologically relevant tools to study variation found in human patient genomes. As part of that effort, Dr. Katsanis leads the Taskforce for Neonatal Genomics. This multidisciplinary group of physicians and basic scientists strives to synthesize genomic and biological data for the faster diagnosis, improved/focused clinical care, and potential therapeutic paradigms, for infants and neotates with genetic conditions. In parallel, the Katsanis lab pursues questions centered on the signaling roles of vertebrate cilia, the translation of signaling pathway defects on the causality and possible treatment of ciliary disorders, and the dissection of second-site modification phenomena as a consequence of genetic load in a functional system. In recognition of his work, Dr. Katsanis was awarded the Young Investigator Award from the American Society of Nephrology in 2009, the E. Mead Johnson Award from the Society for Pediatric Research in 2012 and has delivered several Distinguished lectures. Dr Katsanis is a Professor in the Departments of Cell Biology and Pediatrics and holds the Brumley Distinguished Professorship. He has published over 190 research papers, reviews, and book chapters, serves on several advisory, editorial, and organizational boards and has delivered over 140 lectures in 18 countries.