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GENERAL INFORMATION

Conference Venue: 30 Biopolis Street, MATRIX, Level 2 Singapore 138671

Nearest MRT station: Buona Vista (EW21/CC22). 5-minutes walk via Metropolis (Exit D).

Carpark: Visitors' Parking is at Basement 3 (Cash Card Parking). Park at B3 in the Orange zone at Matrix Building and take Lift D to Level 1 lobby. See parking rates at Biopolis 1 - Car Park.



Housekeeping Matters



In case of emergency or fire, please exit via the Emergency Exit nearest to you.



Restrooms are located outside the Auditorium. See floor plan on next page.

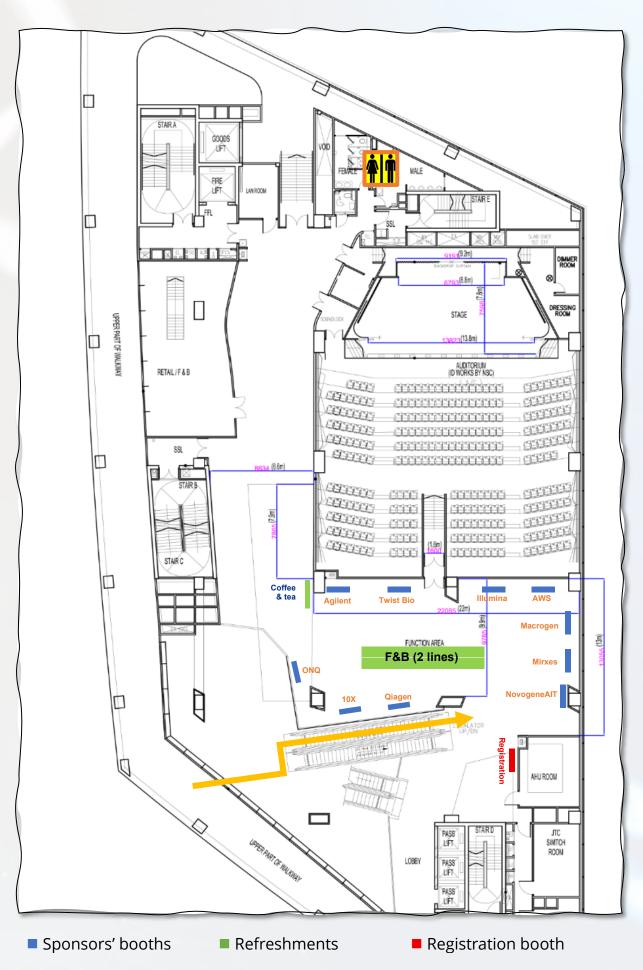


Food and drinks are not allowed inside the Auditorium.

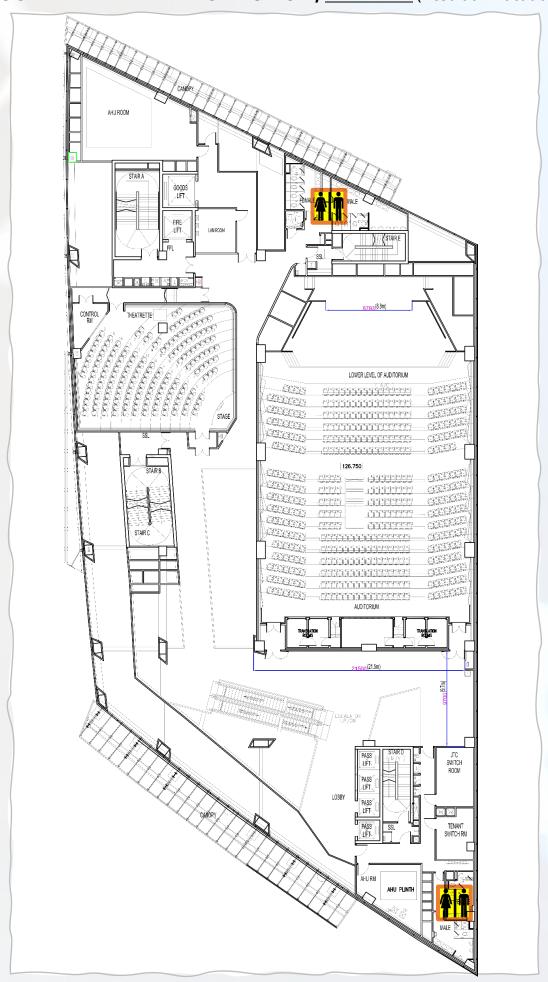


Please silence your digital devices when you are inside the Auditorium.

FLOOR PLAN - MATRIX AUDITORIUM, LEVEL 2



FLOOR PLAN - MATRIX AUDITORIUM, <u>LEVEL 2M</u> (Restroom locations)



WELCOME MESSAGE

Dear Participants,

This year, A*STAR Genome Institute of Singapore (A*STAR GIS) marks its 25th year of establishment in Singapore. As part of our anniversary celebrations, we welcome you to join us at our GIS25: Genome Innovation and Precision Medicine Conference from 11 to 12 September 2025 at Matrix, Biopolis, Singapore.

We are expecting over 400 local and international attendees at this conference, which will bring together a diverse group of researchers, practitioners, and thought leaders from around the world, where we will discuss and explore the latest advancements and innovations in the field of genomic sciences.

We are honoured to have an exceptional lineup of speakers and presenters who will share their expertise and groundbreaking research on RNA/DNA technologies, Spatial and Single Cell technologies, AI, Population Health and Precision Medicine. We hope you will have the opportunity to engage in stimulating discussions, attend insightful presentations, and network with peers who share your passion for scientific research and discovery and to foster collaborations for the future.

On behalf of the organising committee, we look forward to your active participation to contribute to the success of this conference and the future of genomic research.

Warm regards,

Dr WAN Yue and **Dr TAM Wai Leong**Co-Chairman, Conference Organising Committee
A*STAR Genome Institute of Singapore

ORGANISING COMMITTEE

Committee Co-Chairs



Dr. WAN Yue



Dr. TAM Wai Leong

Scientific Committee



Dr. KHOR Chiea Chuen



Dr. Shyam PRABHAKAR



Dr. Mile SIKIC



Dr. LIU Jinyue



Dr. Tim STUART



Dr. CHIA Minghao

Organising Committee



Ms. Madeline SHEE



Ms. Eliza LIM



Ms. Debby CHUA



Ms. Winnie LIM

PROGRAMME

TIME	DAY 1 – 11 SEPTEMBER				
8:00AM					
9:00AM	Day 1 – Session A Chair: Dr WAN Yue				
9:05AM	<u> </u>				
9:10AM	mRNA Stability Control: Lessons from Viruses and RNA Therapeutics				
9. IUAIVI	·				
	Professor V. Narry KIM Seoul National University				
9:55AM	Enhancing RNA Structure Prediction with Data-Driven Molecular Dynamics				
9.33AIVI	Simulations				
	Professor Shi-Jie CHEN				
	University of Missouri				
10:20AM					
10.20AW	Day 1 – Session B Chair: Dr LIU Jinyue				
11.00 4 14					
11:00AM	Single-cell RNA Sequencing of Peripheral Blood Links Cell-Type-Specific Regulation of				
	Splicing to Autoimmune and Inflammatory Diseases Assistant Professor LIU Boxiang				
	Department of Pharmacy and Pharmaceutical Sciences, National University of				
11:25AM	Singapore Targeted Gene Insertion Using CRISPR-Associated Transposases				
I I.ZSAW	Dr Leslie BEH				
	A*STAR Institute of Molecular and Cell Biology (A*STAR IMCB)				
11.500.00					
11:50AM	Associate Professor Yvonne TAY				
	Cancer Science Institute of Singapore, National University of Singapore (NUS)				
12:15PM	Lunch				
12. 137 101	Lunchtime Talks				
	• 12:20pm: Illumina				
	• 12:40pm: Twist Bioscience				
	• 1:00pm: Agilent				
	Day 1 – Session C Chair: Dr Tim STUART				
1:45PM	Reprogramming The Human Epigenome				
	Professor Ryan LISTER				
	Harry Perkins Institute of Medical Research, The University of Western Australia				
2:15PM	Spatial Architecture of Autism Pathogenesis During Early Development				
	Dr LIU Jinyue				
	A*STAR Genome Institute of Singapore (A*STAR GIS)				
2:40PM	Genome Stability and Cancer Therapy				
	Prof Kristijan RAMADAN				
	Lee Kong Chian School of Medicine, Nanyang Technological University Singapore				
3:05PM	Tea break				
	Day 1 – Session D Chair: Dr KHOR Chiea Chuen				
3:40PM	Histone Variant MacroH2A1 Shapes Long-Ranged Chromatin Architecture				
	Dr Arnold OU				
	The Rockefeller University				
4:05PM	From Genomes To Metagenomes: A Systems View of Precision Medicine				
	Associate Professor Sunny WONG				
	Lee Kong Chian School of Medicine, Nanyang Technological University Singapore				
4:30PM	A Two-Decade Odyssey in Genomics: From Discovery to Innovation				
	Professor NG Huck Hui				
	Agency for Science, Technology and Research				
5:00PM	End of Conference Day 1				

PROGRAMME

TIME	DAY 2 – 12 SEPTEMBER
8:00AM	Registration
9:00AM	Day 2 - Session E Chair: Assoc Prof TAM Wai Leong
9:00AM	Building More Representative Global Genomic Resources
	Professor Daniel MacArthur
	Centre for Population Genomics
	A joint initiative of Garvan Institute of Medical Research and Murdoch Children's
9:45AM	Research Institute The Singapore National Precision Program: Powering Research, Innovation and
9.43AW	Enterprise on a National Scale
	Professor Patrick TAN
	Precision Health Research Singapore (PRECISE)
	Duke-NUS Medical School (Duke-NUS)
	A*STAR Genome Institute of Singapore (A*STAR GIS)
10:10AM	Genetic Determinants of Leukocyte Telomere Length and Their Impact on Mortality,
	Disease Risk and Late-Life Health in Singaporean Chinese
	Professor KOH Woon Puay
	Healthy Longevity Translational Research Programme, Yong Loo Lin School of
	Medicine, National University of Singapore
	A*STAR Institute for Human Development and Potential, Singapore
10:35AM	Tea break
	Day 2 - Session F Chair: Dr Mile SIKIC
11:00AM	Automated Real-World Data Integration Improves Cancer Outcome Prediction
	Dr Nikolaus SCHULTZ
44.05414	Memorial Sloan Kettering Cancer Center
11:25AM	The Human Pangenome Project: A Global Resource to Map Genomic Diversity
	Associate Professor Karen MIGA
11:50AM	University of California, Santa Cruz 18 Years After the First Breast Cancer GWAS – Where Are We Now?
I I.SUAIVI	Dr LI Jingmei
	A*STAR Genome Institute of Singapore (A*STAR GIS)
12·15PM	Population Genomics and Public Health Perspectives of Indian Subcontinent
12.131 10	Dr Kumarasamy THANGARAJ
	CSIR-Centre for Cellular and Molecular Biology (CCMB), Hyderabad, India
12:40PM	Closing Remarks - Conference Chair
12:45PM	Lunch
12:45PM 2:00PM	Lunch Science Day @ GIS - Proceed to GIS, Genome, Level 2



Professor Narry KIM

- Professor, School of Biological Sciences, Seoul National University, Republic of Korea
- Director, Center for RNA Research, Institute for Basic Science, Republic of Korea

Narry Kim is a Professor in the School of Biological Sciences at Seoul National University and a founding Director of the RNA Research Center at Institute for Basic Science (IBS). Kim graduated from Seoul National University in 1992 and received her Ph.D. in 1998 from the University of Oxford where she studied lentiviruses and gene delivery. For postdoctoral training, she joined the Gideon Dreyfuss lab at the University of Pennsylvania to study mRNA surveillance. Kim moved back to Seoul National University in 2001 to set up her own group and has been investigating how genes are regulated at the RNA level. The Kim lab delineated the microRNA pathway, identified key factors including DROSHA, and revealed their action mechanisms and structures. Her group also uncovered the roles of noncanonical RNA tailing such as uridylation and mixed tailing in the control of microRNAs, mRNAs, and viral RNAs. She is a recipient of the L'Oreal-UNESCO Women in Science Award, the Hoam Prize, and the Asan Prize; was elected members of KAS, NAS, EMBO, and the Royal Society; and serves on the editorial boards of Science, Cell, Molecular Cell, GD, and EMBO J.

MRNA STABILITY CONTROL: LESSONS FROM VIRUSES AND RNA THERAPEUTICS

RNAs of external origin, such as viral RNAs and therapeutic mRNAs, rely on cellular machinery, but at the same time, they encounter multiple cellular barriers that inhibit their functions. In this presentation, I will discuss two recent studies focused on the regulatory mechanisms of exogenous RNAs. In the first part, I will talk about our recent work on the regulatory mechanisms of therapeutic mRNAs. To comprehensively explore cellular regulators of therapeutic mRNAs, we conducted genome-wide screens on in vitrotranscribed (IVT) mRNAs. We identified both positive and negative cellular regulators that impact IVT mRNA effectiveness. By comparing mRNAs with or without N1methylpseudouridine, we delineate the mechanism how N1-methylpseudouridine enhances protein production of mRNAs. In the second part of my presentation, I will discuss functional viromic screens from which we have uncovered numerous viral regulatory elements that can improve mRNA therapeutics. By examining libraries of viral segments located in the 3' UTR, we identified RNA elements that increase or reduce RNA abundance and translation. Remarkably, a substantial portion of positive elements are regulated by the terminal nucleotidyl transferases, TENT4 family members. These enzymes elongate poly(A) tails with mixed sequences, delaying deadenylation and extending mRNA half life. These findings demonstrate an interesting example of convergent evolution and highlight the importance of TENT4 in mRNA stability control. Our research provides a unique resource for virus and RNA research and demonstrates the potential of the virosphere for biological discoveries and RNA therapeutics development.



Professor Shi-jie CHEN

• Professor, University of Missouri

Shi-Jie Chen is a Curators' Distinguished Professor of Biophysics, Biochemistry, and Data Science & Informatics at the University of Missouri. He received a B.S. degree from Zhejiang University. Through T.D. Lee's CUSPEA program, he entered the graduate school at the University of California, San Diego, where he earned a Ph.D. in Physics. He then conducted postdoctoral research with Ken Dill in the Department of Pharmaceutical Chemistry at the University of California, San Francisco. Chen was elected a Fellow of the American Physical Society (APS) and a Fellow of the American Association for the Advancement of Science (AAAS) for his research on RNA structure and dynamics. He serves as an Associate Editor for PLOS Computational Biology, MLHealth, and RNA NanoMedicine, and as an Editorial Board Member for The Journal of Biological Chemistry. He is a Founding Council Member of the International Society of RNA Nanotechnology and Nanomedicine. Chen studies the computational biology of RNA folding and therapeutics. He is known for his work on RNA structure prediction; modeling of RNA folding stability, kinetics, and metal ion effects; and the computational design of RNAtargeted drugs, RNA aptamers, RNA-based nanomedicine, and CRISPR gene editing systems.

ENHANCING RNA STRUCTURE PREDICTION WITH DATA-DRIVEN MOLECULAR DYNAMICS SIMULATIONS

Emerging biomedical advances such as precision medicine and synthetic biology highlight RNA as a central regulator and information carrier. We are interested in predicting RNA structure, stability, and kinetics from nucleotide sequences, as well as designing molecules for therapeutic applications. We aim to answer questions such as: How can we build the native fold from the sequence? For a given RNA target, how can we predict RNA-small molecule interactions and identify potential drug candidates? Developing computational tools to address these questions is challenging due to the limited availability of RNA structural and binding data. By applying physical and chemical principles, and integrating RNA structural data with Bayesian statistics and molecular dynamics simulations, we have developed Vfold, an RNA structure prediction pipeline. Vfold was officially ranked #1 in the RNA category of the recent international competition, the Critical Assessment of Structure Prediction (CASP16), for biomolecular structure predictions. This new model provides a much-needed tool for structure-based understanding and the rational design of RNA therapeutics.



Assistant Professor LIU Boxiang

Department of Pharmacy and Pharmaceutical Sciences,
 National University of Singapore

As a PI and the director of the Genomic Data Science Lab in the National University of Singapore, Boxiang has dedicated his career to advancing the understanding of complex human diseases through innovative genetic, single-cell, and spatial transcriptomic analyses. A significant facet of my academic endeavour includes a pivotal role in the Genotype-Tissue Expression (GTEx) Project, contributing to a comprehensive understanding of genetic effects on molecular phenotype across diverse tissues. Additionally, his group spearheaded the single-cell splicing analysis within the Asian Immune Diversity Atlas project, and provided the first cell-type-specific sQTL map using over 1 million PBMC single cells. He has been awarded the Presidential Young Professorship (Singapore), President's Award in Natural Sciences and Mathematics (US), Stanford University CEHG fellowship (US), Charles B. Carrington Memorial Award (US), and the Chinese National Award for Outstanding Overseas Ph.D. Students.

SINGLE-CELL RNA SEQUENCING OF PERIPHERAL BLOOD LINKS CELL-TYPE-SPECIFIC REGULATION OF SPLICING TO AUTOIMMUNE AND INFLAMMATORY DISEASES

High-throughput genotyping and sequencing have led to the discovery of thousands of disease-associated variants. Because most of these variants lie in non-coding regions, their functional mechanisms remain unclear. To identify genetic effects underlying complex diseases, it has become increasingly important to investigate the proper cell types and contexts. Previous cell-type-specific studies have focused on gene expression, and we argue that alternative splicing plays an equally important role in disentangling complex diseases. Here we describe cell-type-specific, sex-biased and ancestry-biased alternative splicing in ~1 M peripheral blood mononuclear cells from 474 healthy donors from the Asian Immune Diversity Atlas. We identify widespread sex-biased and ancestrybiased differential splicing, most of which is cell-type-specific. We identify 11,577 independent cis-splicing quantitative trait loci (sQTLs), 607 trans-sGenes and 107 dynamic sQTLs. Colocalization between cis-eQTLs and trans-sQTLs revealed a cell-type-specific regulatory relationship between HNRNPLL and PTPRC. We observed an enrichment of cis-sQTL effects in autoimmune and inflammatory disease heritability. Specifically, we functionally validated an Asian-specific sQTL disrupting the 5' splice site of TCHP exon 4 that putatively modulates the risk of Graves' disease in East Asian populations. Our work highlights the impact of ancestral diversity on splicing and provides a roadmap to dissect its role in complex diseases at single-cell resolution.



Dr. Leslie BEH

 Principal Investigator, A*STAR Institute of Molecular and Cell Biology (A*STAR IMCB)

Leslie Beh is a Principal Investigator at the Institute of Molecular and Cell Biology (IMCB), Agency for Science, Technology and Research (A*STAR) since September 2022. Prior to joining IMCB, Leslie pursued a career in industry, joining Illumina to lead a research group for developing novel epigenetics assays. Driven by the desire to do biological research and make an impact on students, Leslie joined IMCB, A*STAR as a Principal Investigator. Leslie earned an A.B. in Biology at Harvard University as a John Harvard scholar, where he trained with Nicole Francis on the biochemistry of Polycomb group proteins. He then obtained his A.M. and PhD in Biology from Princeton University as a Petrie fellow with Laura Landweber and Tom Muir, where he developed methods for assembling synthetic chromosomes with custom epigenetic modifications. Using genomics and biochemical fractionation approaches, he identified a novel DNA methyltransferase complex with evolutionary links to the canonical RNA N6-methyladenosine (m6A) methyltransferase, METTL3/14. Following this, Leslie embarked on a short postdoctoral stint with Sam Sternberg at Columbia University, where he used genomic, structural, and biochemical approaches to study CRISPR-Cas systems that mediate RNA-guided DNA integration.

TARGETED GENE INSERTION USING CRISPR-ASSOCIATED TRANSPOSASES

CASTs (CRISPR-associated transposases) mediate programmable insertion of large DNA cargoes, holding promise as a safe and effective tool for gene therapy. All CAST systems encode a conserved AAA+ ATPase, TnsC, that is essential for transposition. Yet, it remains poorly understood how TnsC regulates CAST activity. To address this, we performed a protein-wide saturation mutagenesis screen of Scytonema hofmannii TnsC, quantitatively determining the impact of 22,000 TnsC mutations on transposition efficiency. Top-performing mutants exhibit a 5-10 fold increase in transposition efficiency, >98% specificity, and are enriched at three functional interfaces in TnsC: the ATP binding site, the transposase binding site, and between adjacent TnsC subunits. Molecular dynamics (MD) simulations reveal that specific TnsC mutations favour ontarget CAST protein complex formation while disfavouring the off-target state, leading to a dual increase in efficiency and specificity. Our study delineates TnsC as an attractive target for the engineering of highly active and specific CAST systems.



Associate Professor Yvonne TAY

Principal Investigator, Cancer Science Institute / National University of Singapore

Yvonne began her research career in Bing Lim's lab at the Genome Institute of Singapore, where she studied miRNA function and mechanisms of action (Tay et al, Nature 2008). She then pursued her postdoctoral training in the Pandolfi lab at Harvard Medical School, where she investigated how transcripts can co-regulate each other by competing for shared miRNAs (Tay et al, Cell 2011). Now based at the Cancer Science Institute of Singapore and National University of Singapore, Yvonne's research group studies noncoding RNAs as well as the non-coding regions of protein-coding mRNAs (untranslated regions, UTRs; Chan et al, Nat Cell Biol 2022). As many mRNA populations comprise transcripts with different UTRs, and these UTRs control key processes such as stability, localization and transport, a better understanding of their function may lead to insights into the regulation of key cancer genes. Her long-term goal is to translate these basic research discoveries into new avenues for the development of novel RNA-based anticancer diagnostics and therapeutics.

MESSENGER RNA HETEROGENEITY AND CANCER PROGRESSION

The majority of human mRNAs generate alternative 3' untranslated regions (UTRs) through various processes, including RNA modifications such as RNA editing, m6A methylation, and alternative polyadenylation, with 3'UTR splicing as an emerging mechanism. Multiple factors, ranging from the genome to transcriptome level, regulate these processes and contribute to 3'UTR heterogeneity. Genomic variants in 3'UTR regions as well as aberrant 3'UTR processing alter the transcriptomic landscape and are associated with cancer. Increasing evidence, aided by high-resolution sequencing technologies and large-scale computational analyses, points towards potential crosstalk between these processes, whose deregulation may further contribute to cancer pathogenesis. We have integrated short- and long-read scRNA-seq of colorectal cancer (CRC) samples to build an isoform-resolution CRC transcriptomic atlas. We identified dysregulated transcript structures in tumor epithelial cells and characterized isoforms associated with epithelial lineages and subpopulations exhibiting distinct prognoses.

Illumina

LUNCHTIME TALK



Dr Yin Nah TEODirector, Scientific Research, Molecular Sciences Department,

Yin Nah Teo is a Director in Scientific Research in the Illumina Core R&D team. She leads R&D project teams with a mission to deliver technologies that advance the fields of genomics and multiomics technology. During her 10 years at Illumina, her teams have contributed various innovations to Illumina's pipeline of products such as the NextSeq2000, NovaSeqX and MiSeqi100 sequencers. She has also been recognized as a champion inventor with more than 25 patent families in the company and more than 70 invention disclosures. Yin Nah is also passionate about bridging research innovations and operations, working cross-functionally to accelerate research to product.

Yin Nah received her PhD in Chemistry at Stanford University, studying the design and synthesis of modified nucleic acids. After her PhD, she worked as a Senior Research Fellow with Dr Sydney Brenner.

ILLUMINA INNOVATION ROADMAP: YOUR GUIDE TO MULTIOMIC INNOVATION

At Illumina, innovation is in our DNA. For more than 20 years, we've been at the forefront of next-generation sequencing, enabling scientists, clinicians and researchers across more than 140 countries to unlock the power of the genome. Our latest sequencing platforms - the NovaSeq™ X, NextSeq™ 2000 and MiSeq™ i100 – are transforming the way genomic data is generated. From advanced patterned flow cell engineering to streamlined onboard data analysis, we've made sequencing more accessible more accurate and more affordable than ever before – helping accelerate discovered that improve lives around the world.

Now, that same innovation is driving the future of multiomic research. Illumina's NGS technology has become the foundation for exploring biology at multiple levels - such as genomic, transcriptomic and epigenomic - providing a more complete view of health and disease. Through our innovation roadmap, we're developing tools that deliver deeper resolution, faster turnaround and seamless data integration. From single-cell analysis to spatial transcriptomics and proteomics, we are creating the technologies and partnerships that will make multiomic discovery not just possible – but truly transformative.

Join us to learn more about where we're heading next as we introduce upcoming products designed to enable comprehensive multi-omic data analysis.

LUNCHTIME TALK



Assistant Professor Jungjoon Kempthorne LEE Assistant Professor, Synthetic Biology for Clinical and Technological Innovation (SynCTI) and Department of

Biochemistry, National University of Singapore

ALPHA-FOLD GUIDED RATIONAL MUTAGENESIS AND MULTIPARAMETRIC DIRECTED EVOLUTION ENHANCE COMPACT BASE EDITOR PERFORMANCE BEYOND BE4MAX

Abstract: The search for compact and efficient cytidine base editors has accelerated the discovery of alternative deaminases beyond APOBEC1, yet many candidates suffer from low activity, cytotoxicity, and high indel formation. Here, we introduce an integrated optimization pipeline that combines Al-assisted rational design with multiplexed directed evolution in E. coli to overcome these limitations. Using SsdAtox, a compact deaminase from Pseudomonas syringae, we applied AlphaFold3-guided single-residue mutagenesis and identified a key lysine (K31) whose substitution enhanced DNA accessibility and catalytic performance. Subsequent screening with our newly developed Trinity-Screen, a three-in-one bacterial assay that concurrently selects for high activity, low indel formation, and reduced cytotoxicity, yielded optimized SsdAtox variants. These evolved editors demonstrated a 10.8-fold increase in editing efficiency, a 2-fold reduction in indels, a 10fold reduction in cytotoxicity, and up to a 31-fold overall improvement compared to BE4max across diverse genomic targets. To our knowledge, this is the first experimental validation of AlphaFold3-driven rational single-residue design leading to gain-of-function in a DNA-modifying enzyme, and the first evolution platform to simultaneously address safety and efficacy in base editor engineering. This work provides a blueprint for nextgeneration therapeutic genome editing tool development.

LUNCHTIME TALK



Dr LEE Chee Yang

 Senior Field Application Scientist, Genomics solutions, South Asia Pacific, Agilent Technologies Singapore (Sales) Pte Ltd

Chee Yang joined Agilent since 2010 and is part of the South Asia Pacific (SAP) Applications Team for the Diagnostics and Genomics Group. His work as a Senior Field Applications Scientist includes providing applications support for the Genomics portfolio of products, such as targeted NGS and microarray for research and clinical research in cancer and human health. He has also been involved in many projects, one of which was utilizing systems that combine liquid chromatography-mass spectrometry with PCR-based assays for viral/microbial detection. Chee Yang completed his Ph.D. at Curtin University in Western Australia and was involved in a wide range of research projects. These include the characterization of the ovine MHC class II region and its association with gastrointestinal parasite resistance, the effects of IgA on parasite resistance in sheep, identification of microsatellites in pedigree testing in Alpacas, as well identifying microsatellites in several crustacean species in environmental studies.

ADVANCING CANCER GENOMICS WITH THE AGILENT AVIDA TECHNOLOGY FOR LIQUID BIOPSY SAMPLES

Liquid biopsy has revolutionized oncology research by enabling ultra-sensitive detection of cancer-associated genomic alterations from circulating cell-free DNA (cfDNA) through a minimally invasive approach. However, challenges such as low DNA input, PCR bias, and labor-intensive workflows continue to limit its broader adoption. The Agilent Avida target enrichment technology is purpose-built to overcome these barriers—delivering high-sensitivity variant detection with a streamlined, efficient workflow. Furthermore, Avida enables simultaneous capture of methylation targets from the same sample, unlocking powerful multiomic insights from a single assay.



Professor Ryan LISTER

Professor, Harry Perkins Institute of Medical Research,
 The University of Western Australia

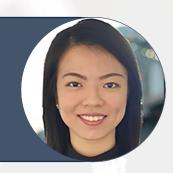
Ryan Lister leads a research group investigating the epigenome and cell identity, at the University of Western Australia and the Harry Perkins Institute of Medical Research. After receiving his PhD from UWA in 2005, Ryan undertook postdoctoral studies at The Salk Institute for Biological Studies, where he developed and applied new techniques to map the epigenome and transcriptome. Having returned to UWA in 2012, Ryan's laboratory is focused upon understanding the role of the epigenome in regulation of cell identity, and developing molecular tools to precisely edit the epigenome and transcriptional activity to control cell state and functions.

REPROGRAMMING THE HUMAN EPIGENOME

The ability to reconfigure the human epigenome is fundamental to controlling cell identity and function, with applications in regenerative medicine, disease modelling, and cell-based therapies. However, approaches to reprogram the epigenome, from local to global scales, frequently suffer from suboptimal accuracy, fidelity, or efficacy.

In this presentation, I will describe our recent advances in large-scale epigenome reconfiguration during induced pluripotent stem cell (iPSC) reprogramming and the development of novel targeted epigenome editing tools. Using an integrative multi-omics approach, we have mapped the dynamic changes in the human epigenome throughout reprogramming and identified key barriers that prevent full erasure of somatic cell epigenetic memory. Building on these insights, we have devised a strategy that more completely resets the epigenome, yielding iPSCs that more closely resemble embryonic stem cells at the molecular and functional levels.

Beyond global reprogramming, precise epigenome engineering holds great promise for controlling gene expression and modifying cell states. I will highlight our work developing new epigenome editing constructs and present findings from a large-scale screen exploring novel tools for targeted epigenetic modification in human cells. Together, these advances provide new avenues for both global and local epigenome reprogramming to control genome activity and cell function.



Dr. LIU Jinyue

 Principal Scientist, A*STAR Genome Institute of Singapore (A*STAR GIS)

Dr Liu Jinyue is a neuroscientist and Principal Investigator at the Genome Institute of Singapore, where she leads the Single-Cell Spatial Neuromics Laboratory. Her research integrates classical biology with omics technologies to unravel the complex organization of the human brain in health and disease. She holds a Ph.D. in Neurobiology from Harvard University, with work cited in Kandel's Principles of Neural Science. In 2021, she was named Singapore's 100 Women in Tech for her contributions to STEM.

SPATIAL ARCHITECTURE OF AUTISM PATHOGENESIS DURING EARLY DEVELOPMENT

Autism is a neurodevelopmental condition that is clinically and etiologically diverse. How does autism occur to result in behavioural hallmarks that are so unmistakable yet varied? By combining spatial and single-cell transcriptomics with patient-derived organoid models, we reconciled this paradox by showing that there is more heterogeneity than previously thought in the way patients' brains develop. In my talk, I will present a new model for autism pathogenesis alongside the ongoing hypothesis of molecular convergence.



Professor Kristijan RAMADAN

 Professor of Cancer and Stem Cell Biology and Director of Cancer Discovery and Reg. Medicine Programme, Lee Kong Chian School of Medicine, Nanyang Technological University Singapore

Kristijan Ramadan is a Croatian British scientist who has recently taken on the role of Director of the Cancer Discovery and Regenerative Medicine Program and Professor of Cancer and Stem Cell Biology at the Lee Kong Chian School of Medicine (LKCMedicine) at Nanyang Technological University, Singapore. Before this, he served as a full professor of Molecular Medicine at the University of Oxford, where he also holds the position of honorary MRC Investigator. Since 2020, he has been elected a member of the Croatian Academy of Science and Arts. He also holds an honorary position as a UK Medical Cancer Investigator at the University of Oxford.

GENOME STABILITY AND CANCER THERAPY

We and others have recently identified that a central component of the ubiquitin degradation system, p97 (also known as VCP in humans or Cdc48 in yeast), plays an essential role in chromatin-associated degradation (CHROMAD). We have also demonstrated that p97/CHROMAD repairs two chemotherapy-relevant DNA lesions, Topoisomerase 1-cleavage complex and trapped PARP1, induced by topoisomerase and PARP inhibitors, respectively. During my talk, I will discuss the role of CHROMAD in genome stability and how to utilise this knowledge for cancer therapy. Finally, I will share our recent results on the role of nucleophagy (selective autophagy of nuclear material) in CHROMAD and how nucleophagy contributes to chemotherapy resistance.



Dr Arnold OU

Postdoctoral Associate, The Rockefeller University

Arnold Ou earned his PhD in Chemistry from the University of Western Australia. He then joined the Laboratory of Genome Architecture and Dynamics (Risca Lab) at the Rockefeller University for his postdoctoral research.

HISTONE VARIANT MACROH2A1 SHAPES LONG-RANGED CHROMATIN ARCHITECTURE

MacroH2A is an enigmatic histone variant characterized by a tripartite structure consisting of a histone-fold domain, a lysine-rich linker, and a globular macrodomain, making it approximately three times larger than canonical H2A. Although MacroH2A and its isoforms (MacroH2A1.1, MacroH2A1.2, and MacroH2A2) have been implicated in a wide range of biological processes, such as maintenance of transcriptionally repressed chromatin states, being a tumor suppressor, regulating chromatin in DNA repair, and regulating a subset of enhancers. It has been postulated that these functions may, in part, be mediated through its role in shaping genome organization and chromatin compaction. Using Micro-C, a high-resolution 3C protocol, we show that macroH2A1 does not contribute to local chromatin fiber compaction at the kilobase scale, but instead regulates 3D genome architecture through coordinated changes in chromatin looping and compartmentalization, revealing a previously unrecognized architectural role for macroH2A.



Associate Professor Sunny WONG

- Clinician-Scientist and Associate Professor of Nutrition,
 Digestion and Metabolism and Assistant Dean, Academic Medicine, Lee Kong Chian School of Medicine
- Associate Professor, Lee Kong Chian School of Medicine, Nanyang Technological University Singapore

Dr Sunny Wong is Associate Professor at the Lee Kong Chian School of Medicine, Nanyang Technological University, and Director of the Centre for Microbiome Medicine. He also serves as a Consultant in Gastroenterology and Hepatology at Tan Tock Seng Hospital. Trained in medicine at the Chinese University of Hong Kong and did his PhD research in Oxford, his work focuses on the gut microbiome and its role in digestive and metabolic diseases. He has contributed to over 200 publications in leading journals and is actively involved in teaching, mentoring and service to the community. His research has been recognised by awards including the Clarivate Highly Cited Research Award and the NHG-LKCMedicine Clinician Scientist Award.

FROM GENOMES TO METAGENOMES: A SYSTEMS VIEW OF PRECISION MEDICINE

Abstract: The past two decades have seen remarkable progress in precision medicine, largely driven by advances in human genomics. Yet, health and disease cannot be fully understood through the host genome alone. Increasingly, the metagenome—the collective genetic repertoire of our microbial ecosystems—is emerging as a critical dimension in shaping physiology, disease susceptibility, and therapeutic response. This talk will present a systems view of precision medicine that integrates both genomes and metagenomes, highlighting how microbial functional capacity complements host genetics to influence clinical outcomes. I will explore recent insights into how metagenomic profiling informs diagnostics, drug metabolism, and immunotherapy response, and how coupling metagenomics with host multi-omics and artificial intelligence enables predictive modeling of disease trajectories. Beyond discovery, I will discuss translational pathways that bring metagenomic insights to the clinic, including microbiota-directed interventions, metagenome-informed therapeutics, and adaptive clinical trial designs. Finally, I will address current challenges—standardization, data harmonization, and clinical integration—while envisioning a future where host and microbial genomics coalesce to create a truly holistic, systems-level approach to individualized care.



Professor NG Huck Hui

Assistant Chief Executive • Research and Talent Development,
 Agency for Science, Technology and Research

Professor Huck-Hui NG is the Assistant Chief Executive of Research and Talent Development (R&TD), under the Agency for Science, Technology and Research. Professor Ng is renowned in the field of gene regulation and genomics. He had held several administrative positions. He served as the Executive Director of the Genome Institute of Singapore, the Executive Director of the A*STAR Graduate Academy and the Assistant Chief Executive of Biomedical Research Council. Professor Ng helped to setup and implement several national funding initiatives such as the Singapore Food Story, Prenatal and Early Childhood for Human Potential and Nucleic Acids Therapeutics. Professor Ng is active in research and sits on several boards such as Science Center Singapore and NUS High School, and R&D funding steering committees. In recognition of his scientific contributions, Professor Ng has received numerous local and international honours and awards.

A TWO-DECADE ODYSSEY IN GENOMICS: FROM DISCOVERY TO INNOVATION

This presentation will chronicle Professor Ng Huck Hui's 22-year journey at the forefront of genomics and functional genomics. Beginning with foundational research into the intricate mechanisms of gene regulation and stem cell biology, the talk will illustrate how the GIS-powered discovery platform has been instrumental in uncovering novel biological insights. Professor Ng will highlight key milestones, from his early work at the Genome Institute of Singapore, including spearheading efforts in human genomics and stem cell research, to his current work on driving innovations in therapeutics.

The narrative will delve into the evolution of genomic technologies and how their application has enabled groundbreaking research. This includes the use of next-generation sequencing to understand complex diseases like neurodegenerative and metabolic disorders, and the development of innovative approaches such as 3D organoid models for disease modeling and therapeutic discovery. Professor Ng will discuss the critical transition from fundamental discoveries to tangible innovations, showcasing examples of how insights into gene networks and cellular function are being translated into potential diagnostics and therapies for pressing health challenges.

Furthermore, the talk will address the importance of fostering a vibrant research ecosystem, nurturing talent, and forging collaborations to drive scientific advancement. Professor Ng will reflect on the growth of Singapore's biomedical sciences initiative and the strategic imperatives for future progress. Attendees will gain a comprehensive perspective on the power of sustained, integrated genomic research to not only expand our understanding of life but also to pioneer new solutions that can impact human health and society. This journey underscores a relentless pursuit of knowledge and its application, offering a compelling vision for the future of genomic science and medicine.



Professor Daniel MACARTHUR

 Director, Centre for Population Genomics
 A joint initiative of Garvan Institute of Medical Research and Murdoch Children's Research Institute

Professor Daniel MacArthur is a human genomicist with over two decades of experience at the interface between human biomedicine, large-scale genomics, and data science. From 2012-2019 he served as Co-Director of the Medical and Population Genetics Program at the Broad Institute of MIT and Harvard, and led the development of the Genome Aggregation Database (gnomAD), the world's largest accessible resource of human genetic data. In 2020 he returned to Australia as the inaugural Director of the Centre for Population Genomics, a joint initiative between the Garvan Institute of Medical Research, Sydney and Murdoch Children's Research Institute, Melbourne, focused on leveraging large-scale genomic and analytical tools to build a more equitable foundation for genomic medicine. He also leads the Australian Alliance for Secure Genomics and Al in Rare Disease (AASGARD) consortium, a national effort to validate and apply new analytical tools in the diagnosis of severe genetic diseases.

BUILDING MORE REPRESENTATIVE GLOBAL GENOMIC RESOURCES

Genomic medicine relies on the ability to compare data from patients with very large population reference datasets - making it possible, for instance, to accurately identify very rare genetic variants, or molecular signatures that are associated with disease. I will review the development of large-scale reference databases of human genetic variation, such as the Genome Aggregation Database (gnomAD), and their impact on the diagnosis of rare genetic disorders, as well as our understanding of human biology and variation. I will also discuss the challenges of incomplete representation of human genetic diversity in these databases, which remain predominated by individuals of European ancestry, including the effects on the diagnosis, prediction, and treatment of disease. Finally I will discuss global efforts to address this representation challenge, including many national genomics programs, the federated gnomAD network, and the OurDNA program in Australia.



Professor Patrick TAN

- Executive Director of Precision Health Research Singapore (PRECISE)
- Dean (Designate) and Senior Vice Dean for Research at Duke-NUS Medical School (Duke-NUS)
- Distinguished Principal Scientist, A*STAR Genome Institute of Singapore (A*STAR GIS)

Prof. Patrick Tan is Dean (Designate) and Provost's Chair in Cancer and Stem Cell Biology at Duke-NUS Medical School, Singapore. He is also Executive Director of PRECISE (Precision Health Research Singapore) coordinating Singapore's National Precision Medicine program, and Professor (adjunct) at Duke University, USA. He received his B.A. (summa cum laude) from Harvard University and MD PhD degree from Stanford University, where he received the Charles Yanofsky prize. Other awards include the President's Scholarship, Loke Cheng Kim scholarship, Young Scientist Award (A-STAR), Singapore Youth Award, Chen New Investigator Award (Human Genome Organization), President's Science Award, Japanese Cancer Association International Award, Public Administration Medal (Silver), Exemplary Public Service Award, and NUS University Research Recognition Award. He has received the American Association for Cancer Research (AACR) Team Science Award as Team Leader, and Genome Valley Excellence Award by the Government of Telangana (India). He is a member of the American Society for Clinical Investigation (ASCI), Association of American Physicians (AAP), the Singapore Bioethics Advisory Committee (BAC), a Board Member of the International Gastric Cancer Association, Global Alliance for Genomics and Health (GA4GH), Board of Editors for Science and Cancer Discovery, and an advisory member for Qatar Precision Health Institute.

THE SINGAPORE NATIONAL PRECISION PROGRAM: POWERING RESEARCH, INNOVATION AND ENTERPRISE ON A NATIONAL SCALE

Precision Medicine represents a transformative opportunity to improve patient outcomes and advance population health. When deployed at scale, precision medicine initiatives have the potential to reveal disease mechanisms, catalyse economic development, and strengthen a nation's biomedical and digital health ecosystem.

The Singapore National Precision Medicine (NPM) program aims to develop scalable, evidence-based solutions tailored to Asia's diverse populations. Precision Health Research, Singapore (PRECISE) is the central entity coordinating NPM, through a three phase multi-year strategy to translate research into economic and societal impact. We complement current global research efforts with the PRECISE-SG100K dataset, one of Asia's largest and most deeply phenotyped multi-ethnic cohorts. The PRECISE-SG100K resource provides high-resolution insights to enable precision health applications and commercial opportunities including strategic industry partnerships. Underpinned by strong government coordination and anchored in the nation's Research, Innovation, Enterprise master plan, the NPM programme offers integrated, ethical, and secure infrastructure supporting national-scale healthcare innovation and applications in data science. High levels of public trust and regulatory excellence further augment Singapore's potential as a credible and reliable partner in global precision medicine research and development.



Professor KOH Woon Puay

- Professor, Healthy Longevity Translational ResearchProgramme
- Yong Loo Lin School of Medicine, National University of Singapore
- Senior Principal Investigator, A*STAR Institute for Human Development and Potential, Singapore

Dr Koh is Professor in Healthy Longevity Translational Research Programme at Yong Loo Lin School of Medicine, National University of Singapore (NUS). She received her MBBS (Honours) from NUS, her PhD in immunology from the University of Sydney, Australia, and postdoctoral training in epidemiology from the University of Southern California in USA. Being a population health scientist, Prof Koh's research is in studying the epidemiology of common chronic diseases in the population. She is the Principal Investigator of the Singapore Chinese Health Study, and has co-authored about 500 scientific papers on diet, lifestyle and genes in relation to risk of diseases such as cancer, cardiovascular diseases, diabetes, osteoporosis and osteoarthritis. More recently, she has also published on factors that influence important ageing outcomes such as physical frailty, cognitive impairment and aging-related depression. She is regularly listed among the world's top 2% most cited scientists by Stanford University and has received over \$35 million dollars in funding from National Institutes of Health (NIH) in USA and the National Medical Research Council in Singapore.

GENETIC DETERMINANTS OF LEUKOCYTE TELOMERE LENGTH AND THEIR IMPACT ON MORTALITY, DISEASE RISK AND LATE-LIFE HEALTH IN SINGAPOREAN CHINESE

Our study investigated the genetic determinants of leukocyte telomere length (LTL) in Singaporean Chinese, providing insight into novel loci and polygenic risk prediction for East Asians. We conducted a genome-wide association study (GWAS) on 23,096 Singapore Chinese Health Study (SCHS) participants, identifying ten significant LTL loci, including five novel ones (PARP1, TERF1, ATM, POT1, and TINF2) linked to telomere maintenance. A trans-ethnic meta-analysis with Singaporean Chinese (n=23,096) and Europeans (n=37,505) identified six additional loci, three of which were novel. Further analysis of lowfrequency, Asian-specific variants in 25,533 participants revealed four significant variants absent in non-East Asians. Notably, a POT1 missense variant exhibited strong deleterious effects, while a TERF1 variant correlated with increased colon cancer risk and mortality. Longer LTL was associated with reduced respiratory and cardiovascular mortality but indicated a trend towards increased cancer mortality. Polygenic risk scores for LTL suggested elevated lung adenocarcinoma risk. Additionally, a weighted Genetic Risk Score (wGRS) based on 15 LTL-associated SNPs in East Asians was significantly associated with shorter midlife LTL in SCHS. Though wGRS did not directly predict later-life handgrip strength (β = -0.006, p = .094), mediation analysis indicated that about 33.3% of its effect on handgrip strength was mediated through LTL, implying genetic influence via telomere biology. Recently, the SG70 sub-study (1,163 SCHS participants aged 68-80) was launched to assess aging phenotypes through interviews, exams, and imaging. We plan to evaluate age-related markers, like telomere attrition and somatic mutations, using advanced sequencing to understand their impact on late-life health. Genomic and phenotypic integration will clarify how genetics shape telomere biology and health in Asians.



Dr Nikolaus SCHULTZ

 Director, Cancer Data Science Initiative, Memorial Sloan Kettering Cancer Center

Dr Nikolaus Schultz trained as a biochemist with a focus on molecular biology, then transitioned into bioinformatics and computational biology. He has extensive experience in the analysis of complex genomic data, especially in the context of signaling pathways. He is currently the Director of the Cancer Data Science Initiative at Memorial Sloan Kettering Cancer Center, where he oversees the implementation of tools for abstraction, analysis and visualization of cancer genomics data, including the cBioPortal for Cancer Genomics and OncoKB, a knowledge base for Precision Oncology. Nikolaus was a cofounder of the cBioPortal software and has been involved in guiding its development since its inception, especially the design of features that are useful for researchers and clinicians – all influenced by his own strong interest in cancer genomics research. He also runs a research laboratory that explores the genomic alterations that underlie human cancer.

AUTOMATED REAL-WORLD DATA INTEGRATION IMPROVES CANCER OUTCOME PREDICTION

The digitization of health records and growing availability of tumour DNA sequencing provide an opportunity to study the determinants of cancer outcomes with unprecedented richness. Patient data are often stored in unstructured text and siloed datasets. To overcome this limitation, we have combined natural language processing annotations with structured medication, patient-reported demographic, tumour registry and tumour genomic data at Memorial Sloan Kettering Cancer Center (MSK) to generate a clinicogenomic, harmonized oncologic real-world dataset (MSK-CHORD). MSK-CHORD includes data for >100,000 cancer patients and enables discovery of clinicogenomic relationships not apparent in smaller datasets. Leveraging MSK-CHORD to train machine learning models to predict overall survival, we find that models including features derived from natural language processing, such as sites of disease, outperform those based on genomic data or stage alone as tested by cross-validation and an external, multiinstitution dataset. MSK-CHORD also uncovers predictors of metastasis to specific organ sites, including a relationship between SETD2 mutation and lower metastatic potential in immunotherapy-treated lung adenocarcinoma corroborated in independent datasets. We demonstrate the feasibility of automated annotation from unstructured notes and its utility in predicting patient outcomes. Parts of the resulting data are provided as a public resource for real-world oncologic research through the cBioPortal for Cancer Genomics.



Associate Professor Karen MIGA

• University of California, Santa Cruz

Dr. Miga is an Associate Professor in the Biomolecular Engineering Department at UCSC, an Associate Director of the UCSC Genomics Institute, and the Director of the UCSC Sequencing Technology Center. In 2019, she co-founded the Telomere-to-Telomere (T2T) Consortium, an open, community-based effort to generate the first complete assembly of a human genome. Additionally, Dr. Miga is the Director of the Genome Center for the Human Pangenome Reference Consortium (HPRC). Central to Dr. Miga's research program is the emphasis on satellite DNA biology and the use of long-read and new genome technologies to construct high-quality genetics and epigenetic maps of human peri/centromeric regions.

THE HUMAN PANGENOME PROJECT: A GLOBAL RESOURCE TO MAP GENOMIC DIVERSITY

The human reference genome is the most widely used resource in human genetics and is due for a major update. Its current structure is a linear composite of merged haplotypes from more than 20 people, with a single individual comprising most of the sequence. It contains biases and errors within a framework that does not represent global human genomic variation. A high-quality reference with global representation of common variants, including single-nucleotide variants, structural variants and functional elements, is needed. The Human Pangenome Reference Consortium aims to create a more sophisticated and complete human reference genome with a graph-based, telomere-totelomere representation of global genomic diversity. Here we leverage innovations in technology, study design and global partnerships with the goal of constructing the highest-possible quality human pangenome reference. Our goal is to improve data representation and streamline analyses to enable routine assembly of complete diploid genomes. With attention to ethical frameworks, the human pangenome reference will contain a more accurate and diverse representation of global genomic variation, improve gene-disease association studies across populations, expand the scope of genomics research to the most repetitive and polymorphic regions of the genome, and serve as the ultimate genetic resource for future biomedical research and precision medicine.



Dr LI Jingmei

 Principal Scientist, A*STAR Genome Institute of Singapore (A*STAR GIS)

Dr. Jingmei Li is a woman in science doing science for women. Her research centers on breast cancer genetics and proactive health, with a mission to make science more inclusive, empathetic, and accessible. As Group Leader at the Genome Institute of Singapore, she leads interdisciplinary work combining genomics, epidemiology, and qualitative research to better predict breast cancer risk - especially in Asian populations.

She is the co-principal investigator of the BREATHE study, a pioneering risk-based breast cancer screening initiative, and has co-led the development of Singapore's largest breast cancer cohort, comprising over 10,000 patients. Dr. Li is a strong advocate for translating science into action and engaging the public in shaping research that matters.

Her contributions have earned her numerous accolades, including the Young Scientist Award (Singapore National Academy of Science), the National University of Singapore Outstanding Young Alumni Award, and Great Women of Our Time (Women's Weekly). Her career has been supported by prestigious fellowships such as the UNESCO-L'Oréal International Fellowship and the National Research Foundation Singapore Fellowship.

18 YEARS AFTER THE FIRST BREAST CANCER GWAS - WHERE ARE WE NOW?

In 2007, the first breast cancer GWAS drew me to do a PhD in the field. Since then, the field has grown exponentially. To such an extent that we are now seeing papers published by PhD students of the PhD students who worked on the early discoveries. This generational shift speaks to the scientific maturity of breast cancer genetics, but also invites us to reflect: What have we achieved, and what lies ahead?

To date, hundreds of common germline susceptibility loci have been identified. Polygenic risk scores (PRS), once experimental, and in combination with other known breast cancer risk factors, are now robust enough (although still debatable) to inform population-level screening strategies and risk-reducing interventions in certain contexts. Yet questions remain. Have we reached the limits of discovery in common variant GWAS? We've found the variants. Maybe we need the variants to find their way into clinics now (or not).

In this talk, I will highlight recent work from global consortia and emerging efforts in multiancestry PRS, explore what's left to discover (and whether it matters), and discuss the "implementation holy grail".



Dr Kumarasamy THANGARAJ

 CSIR Bhatnagar Fellow, CSIR-Centre for Cellular and Molecular Biology (CCMB), Hyderabad, India

Dr K. Thangaraj is presently a CSIR Bhatnagar Fellow at the CSIR-Centre for Cellular and Molecular Biology (CCMB), Hyderabad, India. He was the Director of the Centre for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad during 2019 - 2023. His main research interest for the last three decades has been in population and medical genomics. He is an elected Fellow of - Indian National Science Academy, Indian Academy of Sciences, and National Academy of Sciences. He is a recipient of several awards, including J C Bose Fellowship, Sun Pharma Research Award in Medical Sciences, Raman Research Fellowship, Life-time achievement Award, Excellence in Science Award, Distinguished Scientist Award, Sir CV Raman Memorial Lecture Award, Sir Dr. UN Brahmachari Award, and delivered several Orations. He is a Board Editor of Mitochondrion; Associate Editor of BMC Medical Genomics; Tropical Medicine and International Health, and Editorial Board member of – Human Genetics and Clinical Genetics. He was the President of the "Indian Society of Human Genetics" (2011 - 2015); and founder & President of the "Society for Mitochondrial Research and Medicine".

POPULATION GENOMICS AND PUBLIC HEALTH PERSPECTIVES OF INDIAN SUBCONTINENT

Indian subcontinent / South Asia is a region of remarkable cultural, linguistic, and genetic diversity with over 4,500 anthropologically well-defined groups. We have been studying various South Asian populations to understand their origin, affinities, and impact of endogamy. One of our studies established that the contemporary Indian populations have descend from two divergent groups: (1) Ancestral South Indians, & (2) Ancestral North Indians (Nature, 2009), and these two founding groups have admixed during the past 2000 - 4000 years (Am. J. Hum. Genet., 2013). Since then, almost all the populations in Indian subcontinent have been practicing endogamy. To assess the impact of endogamy, we have analysed 275 distinct South Asian groups and found that 81 out of 275 groups have strong founder event than the one that occurred in both Finns and Ashkenazi Jews (Nat. Genet., 2017). Further, we went back to the populations that have strong founder event and found that they have high prevalence of population-specific diseases. Notably, one of the populations has high frequency of Junctional Herlitz Epidermolysis Bullosa disease, characterized by vesicobullous skin lesions, oral mucositis, congenital heart disease, and premature death. Subsequently, we performed exome sequencing and found a novel mutation in the LAMB3 gene of the patients, whereas the parents were heterozygous for this deletion. CRISPR/Cas9 mediated knockout of the above mutation and iPSC derived cell lines exhibits the same phenotype in mouse (C57BL/6NI) (unpublished data). Our continuing effort is to identify recessive mutations in the populations, provide prenatal and premarital counseling, which would help in eliminating the pathogenic mutation(s) from the population and reduce disease burden.

GIS Science Day @ Genome Level 2

- Take the lift at Matrix from Level 2 to Level 5 and cross the skybridge to Genome.
- Take the lift at Genome from Level 6 to Level 2.



GIS SCIENCE D

GROWING STRONG FOR 25 YEARS - IT'S IN OUR DNA

12 September 2025

2pm - 5pm



Location Genome Building L2

Join us to celebrate 25 years of Genomic Discovery and Innovation!

INTERACTIVE BOOTHS: *EXPERIENCE THE SCIENCE*

Real Time Immune Profiling



A rapid end-to-end platform for single-cell RNA-seq from a finger-prick for immune profiling and diagnosis

From Lab to Life (CGD)



Clinical diagnostics in a nutshell: from development to application

The Al Molecular Pathologist



Predicting Gene Expression and Pathology from Standard Tissue

Edit to Fold (Eterna): Molecular **Puzzles for RNA Research**



Interactive platform powered by logic and collective curiosity to solve RNA design challenges

Biodiversity for Food Security and Health



Embracing biodiversity research to protect Singapore's natural capital, enhance nutrition, and support human health

Next-Generation **Cancer Diagnostics**



Scalable liquid biopsies and the implementation of transcriptomics in Molecular Tumour Board

Powering Precision Medicine for Asians: AIDA and SG100K





Profiling Asian genetic gand immune diversity to advance precision medicine in prognostics, diagnostics, and therapeutics

Our GIS domains:



Genomic Tech



Health

Al & Compute

BREAKOUT DIALOGUE SESSIONS: MISSION IN MOTION

Session 1 (2:15pm - 2:45pm)

Rewriting Health -The Genomic Revolution at GIS



Dr Wan Yue Executive Director, GIS



Dr Tam Wai LeongDeputy Executive
Director, GIS

Session 2 (3:15pm - 3:45pm)

Intelligence in Scale: Turning Genomic Data into Health Decisions



Dr Anders Skanderup Assistant Director, GIS



Dr Claire Bellis

Session 3 (4:15pm - 4:45pm)

Genomics that Matter **Tools Changing Lives**



Chayaporn, Nok Senior Scientist, GIS / ASTRID

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GROWING STRONG FOR 2	25 YEARS - IT'S IN OUR DNA
POSTER SESSIONS: COM	MUNICATING OUR SCIENCE
Get insights directly from Joh repres	entatives across all three GIS domains
Genomic Tech	Health Al & Compute
Odd Session (2:15pm - 3:30pm)	Even Session (3:30pm - 4:45pm)
P1. Dr Gokce Oguz Your Data, Our Expertise: Unlocking Biological Insights - Bioinformatics Consulting & Training Platform (PI: Dr Adaikalavan Ramasamy)	P2. Ling Min Hao SG-NEx: A Long-Read RNA Sequencing Resource for Five Cancer Cell Lines (PI: Dr Jonathan Göke)
P3. Dr Sui Yue Isoform-level discovery, quantification and fusion analysis from single-cell and spatial long-read RNA-seq data with	P4. Joel Bonnie From Base Pairs to Functions: Rich RNA Representations via Multimodal Language Modeling (PI: Dr Mile Sikic)
Bambu-Clump (Pl: Dr Jonathan Göke) P5. Maxime Rochkoulets From Current to Real: Deep Learning for Real-Time Segmentation of	P6. Dr Pierre-Alexis Vincent Goy GRIDS-NPM's beyond-data-delivery infrastructure & PRECISE-SG100K genomic releases' creation (PI: Dr Nicolas Bertin)
Nanopore Signals (PI: Dr Mile Sikic) P7. Justin Jeyakani GA4GH Whole Genome Sequencing (WGS) Quality Control (QC) Standards (PI: Dr Nicolas Bertin)	P8. Li Zhihui A Catalogue of Structural Variation across Ancestrally Diverse Asian Genomes (PI: Dr Nicolas Bertin)
P9. Gu Zhenhao Efficient trace reconstruction in DNA storage systems using Bidirectional Beam Search (PI: Dr Niranjan Nagarajan)	P10. Amy Rose Birss Exploring the Symbiotic Dance: The Impact of Marine Shipping Pollution on Algae-Bacteria Communities (PI: Dr Niranjan Nagarajan)
P11. Dr Aarthi Ravikrishnan Age-Related Variations in Skin Structure, Physiology, and Microbiome Influence Post-Stress Recovery (PI: Dr Niranjan Nagarajan)	P12. Devika Menon Data-driven detection of translation signatures in unannotated ORFs using Ribosome profiling (PI: Dr Sonia Chothani)
P13. Dr Nigel Chou BANKSY unifies cell typing and tissue domain segmentation for scalable spatial omics data analysis (Pl: Dr Chen Kok Hao)	P14. Dr Larry Loo Engineering alpha-Gal-Free Cultivated Meat: A CRISPR-edited Hypoallergenic Porcine Cell Line for Safer Future Foods (PI: Dr Chew Wei Leong)
P15. Dr Amine Meliani Safety-Enhanced AAV Variants Overcome Pre-Existing Immunity and Reduce Immunogenicity for Repeat Dosing (PI: Dr Chew Wei Leong)	P16. Matthew Dowson Exploring the Mechanisms Underlying DNA Damage and Repair in Regulatory Regions (PI: Dr Jay W Shin)
P17. Umar Bin Mohamad Sahari An ALXI Craniofacial Enhanceropathy: Functionally Dissecting a Deeply Conserved Enhancer Cluster	P18. Dr Cao Yiqun Elaine Capturing Multiple Facets of Blood Cancer Using Single-Cell Long- Read RNA-sequencing (PI: Dr Jay W Shin)
(PI: Dr Jay W Shin) P19. Jane Seng Single-Cell Transcriptomics Reveals Cell-Type Specific Targeting Preferences of Therapeutic Drugs and Peptides (PI: Dr Jay W Shin)	P20. Dr Quyen Do Molecular vulnerability in human midbrain-like organoid model of Parkinson's Disease (Pl: Dr Liu Jinyue)

FOLLOW A*STAR GIS: TO STAR-GIS X ASTAR_GIS



GIS SCIENCE DA

GROWING STRONG FOR 25 YEARS - IT'S IN OUR DNA

POSTER SESSIONS (CONTINUED): COMMUNICATING OUR SCIENCE					
Get insights directly from lab representatives across all three GIS domains					
Genomic Tech	Health Al & Compute				
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Odd Session cont'd (2:15pm - 3:30pm)	Even Session cont'd (3:30pm - 4:45pm)				
P21. Dr Mohua Das Targeted probing of b-catenin-TCF responsive transcription activity affects cellular programs to alter the tumour-immune microenvironment in colorectal cancers (Pl: Dr Ramanuj DasGupta) P23. Dr Shalu Singh Engineering a T7 RNA Polymerase for reusable In Vitro Transcription system (Pl: Dr Seow Yiqi) P25. Dr Jagadish Sankaran Multimodal inference of morphogenomic phenotypes using morphology and expression data optimal clustering (MEDOC) (Pl: Dr Shyam Prabhakar) P27. Tan Le Min Single-cell analysis of human diversity in circulating immune cells (Pl: Dr Shyam Prabhakar) P29. Dr Leung Jia Yu High throughput methods for clonal tracing in single cells (Pl: Dr Tim Stuart) P31. Dr Ashley Aw BiG-SPLASH: Identifying tertiary RNA-RNA Interaction Mapping with spatial proximity (Pl: Dr Wan Yue) P33. Dr Deepa Subramanian NGS-Powered Clinical Workflows: Development & Validation (Pl: Dr Alexander Lezhava)	P22. Dr Daniel Muliaditan Stable bridge amplifications of Chr11q13/q22 advance squamous cancers (PI: Dr Ramanuj Dasgupta) P24. Chua Yi Jing Direct Lysis and Lyophilised LAMP for Point-of-Care Diagnostics (PI: Dr Seow Yiqi) P26. Dane Marc Yap Bagaoisan Extracellular Cartography of CRC Reveals Immunomodulatory Axes (PI: Dr Shyam Prabhakar) P28. Dynn Sim Leveraging space mutagenesis to develop improved cultivars for sustainable indoor farming (PI: Prof Teh Bin Tean) P30. Chrysania Lim Scaling single-cell chromatin analysis with regulatory element modules (PI: Dr Tim Stuart) P32. Dr Han Jian RNA structural heterogeneity in a eukaryotic cell influences its heat shock response (PI: Dr Wan Yue) P34. Ken Liou Next-Gen Diagnostics: Multiplex LAMP for Pathogen Identification (PI: Dr Alexander Lezhava) P36. Dr Sinem Kadioglu Tumor transcriptome deconvolution identifies genes associated with matagtatic progression in colorsociated synthematagtatic progression in colorsociated synthematagtatic progression in colorsociated with matagtatic progression in colorsociated with				
somatic variant calling from tumor-only sequencing using weakly supervised deep learning (Pl: Dr Anders Skanderup)	metastatic progression in colorectal cancer (Pl: Dr Anders Skanderup)				
P37. Dr Guo Yu Amanda Transcriptomic meta- analysis identifies robust, tissue-agnostic gene expression signature of immune checkpoint blockade response (PI: Dr Anders Skanderup)	P38. Denis Odinokov A deep-learning model for quantifying circulating tumour DNA from the density distribution of DNA-fragment lengths (PI: Dr Anders Skanderup)				
P39. Yiamunaa M Gut Microbiota Modulation of Regulatory DNA Elements revealed by Massively Parallel Functional Characterization (Pl: Dr Benson Chen)	P40. Dr Li Zhe Profiling DNA Modification Landscape for Cardiomyocyte Development and Reprogramming in Human and Mouse (Pl: Dr Jianjun Liu)				

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GIS SCIENCE DAY

GROWING STRONG FOR 25 YEARS - IT'S IN OUR DNA						
POSTER SESSIONS (CONTINUED): COMMUNICATING OUR SCIENCE						
Get insights directly from lab represe	entatives across all three GIS domains					
Genomic Tech	Health AI & Compute					
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Odd Session cont'd (2:15pm - 3:30pm)	Even Session cont'd (3:30pm - 4:45pm)					
P41. Jia Qi Tandem Repeat Variation in Asian Populations and Impact on Complex Traits (PI: Dr Liu Jianjun)	P42. Dr Yao Fei Characterizing EBV genome diversity by Long Read Sequencing (Pl: Dr Jianjun Liu)					
P43. Prasad Sarashetti A Complete Telomere- to-Telomere Diploid Reference Genome for Indian Population (Pl: Dr Liu Jianjun)	P44. Tung Ru Jing Ecosystem Perspectives on Implementing Population-Wide Risk-Based Screening: A Qualitative Study of In-Depth Interviews with Healthcare, Industry, and Policy Stakeholders (PI: Dr Li Jingmei)					
P45. Ryan Lim Coexisting Background Breast-Lesion Features and Prognosis in Stage I—						
III Invasive Breast Cancer: A Retrospective Cohort Study (Pl: Dr Li Jingmei)	P46. Dr Lee Yong-An MASLD Therapeutic Discovery Pipeline Identifies Gene X as a siRNA Candidate (PI: Dr Ng Huck Hui)					
P47. Gao Mingtong Targeting AMPK Deficiency: Geroprotective Interventions to Reverse Cellular Aging (Pl: Dr Rajkumar Dorajoo)	P48. Lau Zhi Chng Arterial Wall Tissue Gene Expression Associated with Telomere Length Attritions in Coronary Artery Disease (PI: Dr					
P49. Dr Wu Hao Epigenetic Age Acceleration in novel subtypes of Type 2 diabetes: Opportunities	Rajkumar Dorajoo)					
for precision medicine in diabetes care (Pl: Dr Rajkumar Dorajoo)	P50. Dr Arshia Naaz Dysfunction in BLM increases telomere length and affects the DNA damage response pathway (PI: Dr Rajkumar					
P51. Dr Ma Shijun Reprogramming T Cell Fate for Durable and Systemic Anti-Cancer Immunity (PI: Dr Yu Qiang)	Dorajoo)					

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Illumina Innovation Roadmap

Your Guide to Multiomic Innovation

Thursday 11 September | 12:20-12:35pm

At Illumina, innovation is in our DNA. For more than 20 years, we've been at the forefront of next-generation sequencing, enabling scientists, clinicians and researchers across more than 140 countries to unlock the power of the genome. Our latest sequencing platforms - the NovaSeq™ X, NextSeq™ 2000 and MiSeq™ i100 - are transforming the way genomic data is generated. From advanced patterned flow cell engineering to streamlined onboard data analysis, we've made sequencing more accessible more accurate and more affordable than ever before - helping accelerate discovered that improve lives around the world.

Now, that same innovation is driving the future of multiomic research. Illumina's NGS technology has become the foundation for exploring biology at multiple levels - such as genomic, transcriptomic and epigenomic - providing a more complete view of health and disease. Through our innovation roadmap, we're developing tools that deliver deeper resolution, faster turnaround and seamless data integration. From single-cell analysis to spatial transcriptomics and proteomics, we are creating the technologies and partnerships that will make multiomic discovery not just possible – but truly transformative.

Join us to learn more about where we're heading next as we introduce upcoming products designed to enable comprehensive multi-omic data analysis.



Yin Nah Teo Director, Scientific Research | Molecular Sciences Dept.

Yin Nah Teo is a Director in Scientific Research in the Illumina Core R&D team. She leads R&D project teams with a mission to deliver technologies that advance the fields of genomics and multiomics technology. During her 10 years at Illumina, her teams have consistently delivered innovative solutions to the company pipeline of products such as the NextSeq1000, NovaSeqX and MiSeqi100 sequencers. She has also been recognized as a champion inventor with more than 25 patent families in the company and more than 70 invention disclosures. Yin Nah is also passionate about bridging research innovations and operations, working cross-functionally to accelerate research to product.

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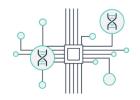


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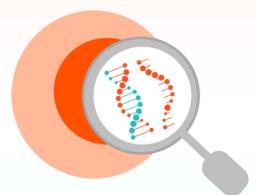
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