5th BCM-CUHK-UMJoint Symposium in CLINICAL GENETICS

Programme | 21 Sep 2024 | Day 1

08:00 - 08:45	Registration	**Reception**
08:45 - 10:30	**Plenary Session 1** **Chairpersons: LEUNG Tak Yeung; THONG Meow-Keong**	**Summit 1**
08:45 - 09:20	Opening and Diagnosis and Therapies for Brittle Bone Diseases	**LEE Brendan**
09:20 - 09:55	Facilitating the diagnosis of rare genetic disorders by artificial intelligence through facial imaging analysis	**HSIEH Tzung-Chien**
09:55 - 10:30	Implementation of prenatal genome sequencing: benefits and challenge	**CHOY Richard**
10:30 - 11:00	Tea Break	
Parallel Session 1A: Thalassemia **11:00 - 12:30**	**Chairpersons: LIM Karen; SETHI Neha**	**Summit 1**
11:00 - 11:20	Long-read sequencing in expanded carrier screening: thalassemia	**CHOY Richard**
11:20 - 11:40	In utero transfusion for Hb Barts fetuses	**HUI Annie**
11:40 - 12:00	Stem cells and gene therapy for thalassemia	**LEUNG Tak Yeung**
12:00 - 12:20	Genetic Diagnosis of Thalassemia and Complex Monogenetic Diseases with LongRead Sequencing	**PENG Zhiyu**
12:20 - 12:30	MGI product launch	**GUO Shirley**
Parallel Session 1B: Advances in Genomic Diagnosis in Rare and Undiagnosed Diseases **11:00 - 12:30**	**Chairpersons: DONG Elvis; MOEY Lip Hen**	**Summit 2**
11:00 - 11:20	Diagnosing the Undiagnosed: A Malaysian Experience in Genomic Testing	**ONG Winnie Peitee**
11:20 - 11:40	Clinical validation of RNAseq for rare disease diagnosis	**LIU Pengfei**
11:40 - 12:00	Value of bioinformatics re-analysis in patients with negative exome findings	**MOHD Khalid**
12:00 - 12:20	Whole genome sequencing in detecting multiple molecular diagnoses in a pediatric setting	**CHAU Matthew**
12:20 - 12:30	Discussion	
12:30 - 13:30	**Lunch Symposium by Xcelom**	**Summit 1**
	Clinical Solution for Complex Monogenic Diseases Based on SMRT Technology	**ZHAN Jiahan**
12:30 - 13:30	**Lunch Symposium by Norvatis**	**Summit 2**
	• Real-world experience of gene therapy with Zolgensma for patients with SMA in Malaysia	**TAE Sok Kun**
	• Gene therapy access: challenges and opportunities in developing country	**THONG Meow-Keong**

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Parallel Session 2A: Genetics in Reproductive Medicine		**Summit 1**
13:30 - 15:00	**Chairpersons: CHAN David; RAHMAH Binti Saaid**	
13:30 - 13:50	Structural variant as an underappreciated factor to human infertility	**DONG Elvis**
13:50 - 14:10	All-in-one PGT testings	**CHOY Richard**
14:10 - 14:30	Preconception workup in consanguineous couples	**KRISHNAN Vivek**
14:30 - 14:50	Abstract Presentation	
	1. Molecular genetic characterization of premature ovarian insufficiency through patients with X;autosome translocation	**CHEN Lin**
	2. Identification of cryptic chromosomal rearrangements through Optical Genome Mapping	**ZHOU Chunxiang**
14:50 - 15:00	Discussion	
Parallel Session 2B: Inborn Errors of Metabolism		**Summit 2**
13:30 - 15:00	**Chairpersons: CHONG Shuk Ching; LEONG Huey Yin**	
13:30 - 13:50	Use of metabolomics in inborn errors of metabolism	**SCAGLIA Fernando**
13:50 - 14:10	Antenatal presentation of IEM cases	**CHONG Josephine**
14:10 - 14:30	Clinical utility of urine metabolomic profiling in diagnosis of patients with suspected IEM	**LAW Eric**
14:30 - 14:50	Cholestatic liver disease in childhood	**CHEW Hui Bein**
14:50 - 15:00	Discussion	
15:00 - 15:30	Tea Break	
Parallel Session 3A: Spinal Muscular Atrophy		**Summit 1**
15:30 - 17:00	**Chairpersons: DONG Elvis; THONG Meow-Keong**	
15:30 - 15:50	Genetic carrier screening for Spinal Muscular Atrophy	**CAO Ye**
15:50 - 16:10	Prenatal USG and genetic diagnosis of Spinal Muscular Atrophy	**WAH Isabella**
16:10 - 16:30	Neonatal screening for Spinal Muscular Atrophy	**CHONG Shuk Ching**
16:30 - 16:50	Gene therapy for Spinal Muscular Atrophy	**BACINO Carlos**
16:50 – 17:00	Discussion	
Parallel Session 3B: Mitochondrial Diseases		**Summit 2**
15:30 - 17:00	**Chairpersons: LO Ivan; NGU Lock Hock**	
15:30 - 15:50	Emerging Therapies on Mitochondrial Disease	**SCAGLIA Fernando**
15:50 - 16:10	Mitochondrial replacement	**CHAN David**
16:10 - 16:30	Metabolic approach to hypoglycaemia	**LEONG Huey Yin**
16:30 - 16:50	**Abstract Presentation**	
	1. Genome Sequencing to Overcome Diagnostic Challenges in Paediatric Patients with Rare Diseases	**SHI Meng Meng**
	2. Bi-allelic pathogenic variants in the TMPRSS7 gene cause neurodevelopmental disorders	**LU Weiliang**
16:50 – 17:00	Discussion	

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Parallel Session 4A: Non-Invasive Prenatal Testing		**Summit 1**
09:00 - 10:30	**Chairpersons: HUI Annie; NARAYANAN Vallikannu**	
09:00 - 09:20	NIPT for multiple pregnancies	**CHOY Richard**
09:20 - 09:40	NIPT for microdeletion syndromes	**KWAN Angel**
09:40 - 10:00	NIPT and Mosaicism	**LEUNG Tak Yeung**
10:00 - 10:20	Discordant gender result between NIPT and USG	**LIM Karen**
10:20 – 10:30	Discussion	

Parallel Session 4B: Genetic Hearing Loss		**Summit 2**
09:00 - 10:30	**Chairpersons: LIU Pengfei; HANIFFA Muzhirah**	
09:00 - 09:20	Genetic carrier screening for hearing loss	**CAO Ye**
09:20 - 09:40	Neonatal screening for hearing loss	**KULASEGARAH Jeyanthi**
09:40 - 10:00	Gene therapy for genetic deafness	**SHU Yilai**
10:00 - 10:20	Contributions of common genetic variants to spoken language learning	**WONG Patrick**
10:20 – 10:30	Discussion	

10:30 - 11:00	Tea Break

Parallel Session 5A: Skeletal Dysplasia		**Summit 1**
11:00 - 12:30	**Chairpersons: WAH Isabella; ONG Winnie Peitee**	
11:00 - 11:20	New Therapies for Skeletal Dysplasias	**LEE Brendan**
11:20 - 11:40	Chondrodysplasia punctata binder's and beyond	**KRISHNAN Vivek**
11:40 - 12:00	Challenges in prenatal diagnosis of Achondroplasia	**LEUNG Tak Yeung**
12:00 - 12:20	**Abstract Presentation**	
	1. Case series with de novo variants of dominant monogenic disorders detected by non-invasive prenatal test (NIPT) in Vietnam	**TANG Sang Hung**
	2. Prenatal Diagnosis of Fetuses with Congenital Anomalies of Kidney and Urinary Tract (CAKUT)	**LIU Fangzi**
12:20 - 12:30	Discussion	

Parallel Session 5B: Advances in Therapy for Genetic Diseases		**Summit 2**
11:00 - 12:30	**Chairpersons: BACINO Carlos; CHEW Hui Bein**	
11:00 - 11:20	Design and validation of antisense oligonucleotides for rare disease therapy	**LIU Pengfei**
11:20 - 11:40	Genetic and targeted therapy of vascular malformations	**LEONG Kin Fon**
11:40 - 12:00	Update on solid organ transplantation in inborn errors of metabolism	**SCAGLIA Fernando**
12:00 - 12:20	Ethical issues of gene therapy in low/middle include country	**THONG Meow-Keong**
12:20 - 12:30	Discussion	

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12:30 - 13:30	**Lunch Symposium by Zhejiang Biosan Biochemical Technologies Co., Ltd**	**Summit 1**
	Biosan Services contribute to the High-quality Development of Maternal and Child Medical Genetics	**YOU Ying**

12:30 - 13:30	**Lunch Symposium by Astrazeneca**	**Summit 2**
	Real-world experience of selumetinib in neurofibromatosis Type 1 – plexiform neurofibroma in the Asian population	**LEE Beom Hee**

Parallel Session 6A: Cyto-genomic Diagnosis		**Summit 1**
13:30 - 15:00	**Chairpersons: LIU Pengfei; TAE Sok Kun**	
13:30 - 13:50	Methodologies and applications in cyto-genomic laboratories	**DONG Elvis**
13:50 - 14:10	Detection of clinically relevant monogenic copy-number variants by comprehensive genome-wide microarray with exonic coverage	**CHAU Matthew**
14:10 - 14:30	The Nile Delta of Precision Medicine: Prenatal Genomic Care in Highly Inbred Population	**AL-OWAIN Mohammed**
14:30 - 14:50	Incontinentia pigmenti: a glimpse of gene conversion as a disease mechanism	**LO Ivan**
14:50 - 15:00	Discussion	

Parallel Session 6B: Caner Genomics & others		**Summit 2**
13:30 - 15:00	**Chairpersons: SCAGLIA Fernando; MAZLAN Rifhan**	
13:30 - 13:50	Rare paediatric solid tumours	**FOO Jen Chun**
13:50 - 14:10	Malaysia experience in mainstreaming genetic testing in cancer management	**YOON Sook Yee**
14:10 - 14:30	Malaysia Experience in Diagnosing Hereditary Aortopathy	**HANIFFA Muzhirah**
14:30 - 14:50	**Abstract Presentation**	
	1. Pathogenic and likely pathogenic variants in familial breast cancer and the implication of cancer genetic counseling in patients and healthy family members: a study of 53 Indonesian familial cancer cases	**MUNIROH Muflihatul**
	2. Genomic and Neural Encoding Tests in Early Identification of Autism	**LAM Wai Fan Fanny**
14:50 - 15:00	Discussion	

15:00 - 15:30	Tea Break	

15:30 - 17:00	**Plenary Session 2** **Chairpersons: LEE Brendan; CHOY Richard**	**Summit 1**
15:30 - 16:00	Expanded parental carrier screening: are you ready?	**LEUNG Tak Yeung**
16:00 - 16:30	Cancer genomics and recent advances in screening, diagnosis and therapeutics	**CHEONG Sok Ching**
16:30 - 17:00	Advances in the diagnosis and treatment of achondroplasia	**BACINO Carlos**