





Scientific Programme Page 1 6th BCM-CUHK-ZJU Joint Symposium in Clinical Genetics 2025 15 – 16 November 2025 | Zhejiang Hotel, Hangzhou, China Day 1 - Saturday, 15 November 2025

Plenary Session 1 (08:45 – 10:30)

- Prof. Brendan Lee Uncovering Skeletal Development Related Disorders Through Animal Modeling
- Prof. Tak Yeung Leung In Utero Therapy for Fetal Genetic Diseases
- Prof. Hefeng Huang Genetic Risk Scores for Chronic Diseases and the Prevention

Parallel Sessions 1A & 1B (11:00 - 12:30)

Session 1A: NIPS (Singleton & Twins)

- Prof. Jinglan Zhang NIPS for Single Gene Disorders
- Prof. Richard Choy Multiple Pregnancies
- Dr. Ye Cao Incidental Findings of Maternal Disorders During Cell-Free DNA
 Screening
- Abstract Presentations

Session 1B: Genetic Evaluation & Counselling

- Dr. Josephine Chong Amyotrophic Lateral Sclerosis: Treatable and Non-Treatable, From Genetic Complexity to Challenging Counselling
- Dr. Matthew Chau Prospective Evaluation of Optical Genome Mapping, Mate-Pair Sequencing, and Chromosomal Microarray Analysis in Prenatal Genetic Diagnosis
- Dr. Taosheng Huang New Insights in Mitochondrial Medicine
- Abstract Presentations







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Lunch Symposium 1 & 2 (12:30 - 13:30)

Parallel Sessions 2A & 2B (13:30 - 15:00)

Session 2A: Expanded Carrier Screening

- Prof. Tak Yeung Leung Overview of Expanded Carrier Screening in Hong Kong
- Dr. Yan Lu The practice and exploration of expanded carrier screening in China
- Dr. Ye Cao Disclosure of Personal Health Risks from Carrier Screening
- Abstract Presentations

Session 2B: Al and Bioinformatics

- Prof. Daryl Scott A Machine Learning Approach to identifying low-penetrance genes
- Dr. Tzung-Chien Hsieh Integrating Next-Generation Phenotyping & Exome Sequencing for Rare Disorders
- Dr. Matthew Chau —Artificial intelligence-Assisted Karyotyping Analysis and Its Clinical Applications in the Cytogenetics Diagnostic Laboratory
- Abstract Presentations







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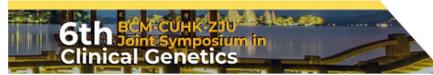
Parallel Sessions 3A & 3B (15:30 – 17:00)

Session 3A: In Utero Treatment for Genetic Diseases

- Prof. Tak Yeung Leung In Utero Therapy for Thalassemia
- Dr. Honglei Duan RhD Blood Group Screening and Perinatal Management Strategies in East Asian Populations
- Dr. Hui Yang Gene Editing-Mediated Gene Therapy for Genetic Diseases
- Abstract Presentations

Session 3B: IEM and Endocrinology

- Prof. Brendan Lee Treating Urea Cycle Disorders as a Model for Approaching Inborn Errors of Metabolism
- Prof. Fernando Scaglia Use of Metabolomics in Inborn Errors of Metabolism
- Dr. Minxin Guan Mitochondrial Deafness: Genetics, Prevention and Treatment
- Abstract Presentations









Scientific Programme 6th BCM-CUHK-ZJU Joint Symposium in Clinical Genetics 2025 15 – 16 November 2025 | Zhejiang Hotel, Hangzhou, China Day 2 - Saturday, 16 November 2025

Parallel Sessions 4A & 4B (09:00 - 10:30)

Session 4A: Genetics in Reproduction

- Dr. Dan Zhang Ovarian Aging: Genetic Background and Micro-Environmental Factors
- Dr. Matthew Chau Concurrent Preimplantation Genetic Testing for Aneuploidies, Structural Rearrangements, and Monogenic Conditions, Integrating the Detection of Triploidy and Genome-Wide Absence of Heterozygosity
- Prof Richard Choy Mosaicism
- Abstract Presentations

Session 4B: New Genetic Disorders

- Prof. Daryl Scott Gene Discovery in the Clinic: Seeing Things for What They Are
- Prof. Pengfei Liu The Development and Validation of Antisense Oligonucleotide Therapies for Genetic Disorders
- Prof. Fung Zhang The Genes Specific for Human Reproduction: Beyond the Murine Models
- Abstract Presentations







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Parallel Sessions 5A & 5B (11:00 – 12:30)

Session 5A: Genetics in Sex Development Disorders

- Dr. Lei Wang Spindle Assembly and Female Fertility
- Dr. Elvis Dong Loss-of-Function in Genes From Chromosome X and Its Role in Infertility
- Dr. Chunyu Liu Genetic Pathogenesis of Asthenoteratozoospermia
- Abstract Presentations

Session 5B: Microbiome & Autisum

- Dr. Ivan Lo Deciphering the Genetics of Autism Spectrum Disorder
- Prof. Ting Fan Leung Genetics of Childhood Food Allergy
- Dr. Qing Zhou Autoinflammatory and Autoimmune Diseases: Diagnosis and Targeted Therapy
- Abstract Presentations

Lunch Symposium 3 & 4 (12:30 - 13:30)







Scientific Programme Page 6 6th BCM-CUHK-ZJU Joint Symposium in Clinical Genetics 2025 15 – 16 November 2025 | Zhejiang Hotel, Hangzhou, China Day 2 - Saturday, 16 November 2025

Parallel Sessions 6A & 6B (13:30 - 15:00)

Session 6A: Genetics in Macrosomia & FGR

- Dr. Luming Sun The Genetic Etiologies of Fetal Growth Restriction
- Prof. Tak Yeung Leung Genetic Diagnosis of Fetal Macrosomic Syndromes
- Dr. Liqun Sun Prenatal Imaging Findings Associated With Genetic Conditions of Heterogeneous Etiology
- Abstract Presentations

Session 6B: Genetics of CAKUT

- Prof. Richard Choy Contribution of Genomic Imbalance in Prenatal Congenital Anomalies of the Kidney and Urinary Tract: A Multi-Center Cohort Study
- Prof. Fernando Scaglia Paediatrics in CAKUT
- Dr. Jianhua Mao High-Risk and Family Screening for Fabry Disease
- Abstract Presentations

Plenary Session 2 (15:30 – 17:00)

- Prof. Fernando Scaglia Clinical Trials in Mitochondrial Disease
- Prof. Ting Fan Leung Gut Microbiome and Childbirth Defects
- Prof. Jian Yang From Genetic Associations to Genes and Spatial Cellular Contexts for Human Complex Diseases

Note: The programme is subject to change without prior notice at the discretion of the organizers.