

## Scientific Programme

# 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: AI 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

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# 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 & Autism**

- 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)***

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### *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.*