

Lecture Abstract

Event: Update on Newborn Metabolic Screening: Clinical, Laboratory and Counselling

Date: June 25, 2022

Time: 09:30 – 12:30 (HKT)

Format: Zoom Webinar

Moderator: Prof. Jessie LAI

This educational seminar provides a comprehensive update on newborn metabolic screening, bringing together a multidisciplinary panel of experts in medical genetics, chemical pathology, and nursing to address the full spectrum of care—from clinical recognition to laboratory testing and family counselling. Designed for obstetricians, paediatricians, pathologists, clinical laboratory scientists, nurses, midwives, and allied healthcare professionals, this session aims to enhance participants' understanding of the evolving landscape of newborn metabolic screening and its integration into clinical practice.

The program is structured to cover three essential pillars of newborn metabolic screening, each delivered by a recognized expert in the field.

Topic 1: Clinical Presentation of Newborn Metabolic Conditions

Dr. CHONG Shuk Ching, Josephine

Associate Professor of Practice in Medical Genetics, Deputy Clinical Director

The Chinese University of Hong Kong – Baylor College Joint Center for Medical Genetics

The Chinese University of Hong Kong

Dr. Chong will open the seminar with an in-depth exploration of the clinical recognition of inborn errors of metabolism (IEM). This session will focus on the early signs and symptoms that may indicate an underlying metabolic disorder, the differential diagnosis, and the critical importance of timely recognition to prevent irreversible morbidity and mortality. Attendees will gain practical insights into the clinical approach to newborns presenting with nonspecific symptoms such as poor feeding, lethargy, vomiting, or metabolic crises, and the red flags that should prompt immediate investigation.

Topic 2: Newborn Screening for Inborn Errors of Metabolism and Beyond

Dr. MAK Miu, Chloe

Consultant, Chemical Pathology Laboratory, Department of Pathology
Hong Kong Children's Hospital

Dr. Mak will provide an update on the laboratory aspects of newborn screening, focusing on the principles, methodologies, and technological advancements that underpin screening programs for inborn errors of metabolism. The session will cover the expansion of screening panels beyond traditional targets, the interpretation of screening results, and the challenges associated with false positives, false negatives, and the identification of incidental findings. Participants will gain an understanding of how laboratory medicine contributes to early diagnosis and the initiation of timely interventions.

Topic 3: Role of Nurses in Newborn Screening and Genetics

Ms. MAN Wing Yan, Diana

Registered Nurse, Department of Paediatrics
Prince of Wales Hospital

Ms. Man will address the pivotal role of nurses in the newborn screening process, from specimen collection and coordination to the delivery of results and family support. This session will explore the counselling aspects of newborn screening, including how to communicate abnormal results to families, provide psychosocial support, and facilitate referrals to specialist services. The critical role of nurses in patient education, informed consent, and long-term follow-up will be emphasized, highlighting the importance of a compassionate and informed approach to family-centred care.

Following each presentation, a moderated Q&A session will allow attendees to engage directly with the speakers, fostering interactive discussion on clinical scenarios, laboratory interpretation, and counselling challenges.

This seminar is made possible through the support of the Professional Services Advancement Support Scheme (PASS) of the Government of the Hong Kong Special Administrative Region. By integrating clinical, laboratory, and nursing perspectives, the event aims to strengthen the multidisciplinary collaboration essential for effective newborn screening programs, ultimately contributing to improved

outcomes for newborns and their families.