香港中文大學
The Chinese University of Hong Kong

香港中文大學醫學院
Faculty of Medicine
The Chinese University of Hong Kong

# Workshop on Application of Whole Exome Sequencing (WES) in Prenatal and Paediatric period

Date: **23 January 2016 (Sat)** Time: **09:00-13:00**

Tutorial Room, 3/F, Postgraduate Education Centre,
School of Public Health, Prince of Wales Hospital, Shatin

## Background

Whole Exome Sequencing is a newly developed test for the identification of mutations that are causative or related to the medical disease. In contrast to single gene sequencing tests, the Whole Exome Sequencing test analyze the exons or coding regions of thousands of genes simultaneously using next-generation sequencing techniques.

The utilization of Whole Exome Sequencing in Paediatrics has been successful for the identification of rare genetic diseases. The recent introduction of prenatal whole exome sequencing for pregnancies with structural anomalies could potentially allow Obstetricians to diagnose rare genetic diseases in the prenatal period.

## Organisers

Department of Obstetrics and Gynaecology, Prince of Wales Hospital,
The Chinese University of Hong Kong, Hong Kong

## Accreditation (Pending)

CME points accredited by HK College of Obstetricians and Gynaecologists
CME points accredited by HK College of Paediatricians
CME points accredited by Faculty of Medicine, CUHK for the practising doctors
PEM points accredited by Department of Obstetrics and Gynaecology, CUHK
CNE points accredited by The Nethersole School of Nursing, CUHK

## Language

English

## Registration and enquiry

Mr. Brown Mak
Tel: 852-2632 1534 Fax: 852-2636 0008
Email: cymak@cuhk.edu.hk
Dept. of O&G, 1E, Prince of Wales Hospital,
Shatin, Hong Kong

## Program Timetable

| Time | Topic |
|---|---|
| 09.00-09.30 | Understanding the technology of WES |
| 09.30-10.00 | How to determine pathogenicity of variants |
| 10.00-10.30 | Counseling and consent before WES |
| 10.30-10.45 | Break |
| 10.45-11.30 | Practical tips: How to interpret WES report with case illustration |
| 11.30-12.15 | Application of WES in Paediatric period with case sharing |
| 12.15-13.00 | Application of WES in Prenatal period: Practical issue and case sharing |

## Speaker

**CHRISTINE M. ENG, M.D.**
*Professor, Molecular and Human Genetics*
*Baylor College of Medicine*
*Chief Quality Officer, Vice President, Executive Lab*
*Director, Baylor Miraca Genetics Laboratories*
*Director, Storage Disorders Clinic*
*Texas Children's Hospital*

My research interests are directed towards translational medicine, specifically the application of molecular genetics to the diagnosis and treatment of genetic diseases. Recently, my efforts have been focused on laboratory and clinical aspects of genetic testing and clinical research in lysosomal storage diseases.

## Registration (HK$100)

Surname _____ First Name _____
Title Prof. / Dr. / Mr. /Ms
Specialty Obstetrician / Paediatricians / Trainee / Nurse /
Others: _____
Position _____
Department _____
Organization _____ (Hospital)
Tel _____ Fax _____
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