



#### Genetics Clinical Grand Rounds

Presented by

## **Department of Genetics Yale School of Medicine**

# "Genomic medicine: A Hong Kong Story on how international collaboration has resulted in improved diagnosis, better patient management and discovery of new syndromes"

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**Date: October 22, 2019 10:15am Location: SHM I-304** 

Course Director/Host: Dr. James McGrath, MD, PhD

There is no corporate support for this activity

This course will fulfill the licensure requirement set forth by the State of Connecticut

#### **ACCREDITATION**

The Yale School of Medicine is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians.

#### TARGET AUDIENCE

Attending physicians, clinical and basic scientists, nurses, residents, fellows, medical students, and other health care providers.

#### NEEDS ASSESSMENT

Genomic sequencing is now in routine clinical practice. Medical geneticists need to understand the different diagnostic options following an initial negative evaluation. We describe a collaborative study between Yale and the University of Hong Kong on exome re-analysis that has resulted in positive outcomes. It is important for providers and personnel to document the clinical utility of genomic testing beyond diagnosis to provide strong evidence for funding support.

#### **LEARNING OBJECTIVES**

- 1. Understand the different approaches in tackling undiagnosed genetic diseases following a negative WES.
- 2. Understand the need of longitudinal studies to evaluate the clinical benefits of genomic sequencing. Discuss the history and scope of NIH undiagnosed disease network.
- 3. Understand how international collaboration can lead to the discovery of new syndromes, e.g. MCTT syndrome.

#### **DESIGNATION STATEMENT**

The Yale School of Medicine designates this live activity for 1 AMA PRA Category 1 Credit(s)<sup>TM</sup>. Physicians should only claim the credit commensurate with the extent of their participation in the activity.

#### FACULTY DISCLOSURES

Dr. Brian Chung, MD - none Dr. James McGrath, MD, PhD - none

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