



# **Genetics Clinical Grand Rounds**

Presented by

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

# "Exome sequencing in newborn screening for inborn errors of metabolism"

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Date: November 24, 2020 10:15am

via Zoom

**Zoom Link** – passcode: 781657

Course Director/Host: Dr. Rama Kastury, DO

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**

Newborn screening (NBS) is a public health programme that identifies infants with heritable disorders before the onset of clinical signs, allowing for early and life-saving intervention. Screening for metabolic disorders using tandem mass spectrometry (MS/MS) identifies most affected newborns, along with a large number of false-positive infants.

#### **LEARNING OBJECTIVES**

- 1. Review the NBS approach and challenges
- 2. Evaluate exome sequencing as a methodology for NBS
- 3. Considerations for implementation in second-tier testing

#### **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. Curt Scharfe, MD, PhD, FACMG - none Dr. Rama Kastury, DO - none

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