



## Genetics Clinical Grand Rounds

*Presented by*  
**Department of Genetics**  
**Yale School of Medicine**

### **“Recent updates on variant interpretation/genotype-phenotype correlation for congenital recessive Titinopathy”**

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**Date: April 27, 2021 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**

Titin, or connectin, is by far the longest known peptide in nature, expressed in cardiac and skeletal muscles. It is well known that truncating variants of TTN especially in the A-band account for 20% patients with dilated cardiomyopathy. However, interpretation of TTN variants pose several challenges, reflecting the sheer size of the TTN gene and the substantial spontaneous genetic variations even in healthy individuals. In the advent of next generation sequencing technology, mutations in TTN have emerged as a major cause of both dominantly and recessively inherited myopathies covering a wide and still expanding spectrum. Growing evidence indicates that many of the biallelic congenital presentations form part of a continuum of Titinopathy rather than distinct entities. Recent updates will be provided on clinical features, TTN-variant interpretation and genotype-phenotype correlation for congenital recessive Titinopathy in order to increase the disease awareness and also to improve the molecular-based diagnosis.

#### **LEARNING OBJECTIVES**

1. General clinical features of congenital recessive Titinopathy
2. Improved criteria for TTN-variant interpretation
3. Current understanding of genotype-phenotype correlations for congenital recessive Titinopathy

#### **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. Deqiong Ma, MD, PhD, FACMG - none  
Dr. Marco Savarese, PhD- none  
Dr. Rama Kastury, DO - none

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