

*Presented by: Genetics*

## Rare Disease Day Speaker Series 2022: Homocystinuria

2/8/2022 10:15:00 AM – 11:15:00 AM | Medical Campus

Participants will learn the latest clinical and molecular advances in human genetics. The participants will learn to recognize abnormal molecular, cytogenetics and biochemical laboratory test results. The participant will learn the clinical management of biochemical and storage disorders. The participant will recognize key dysmorphic features that define individual syndromes.

**Texting code for today's session: 29209**

### [Zoom Link](#)

Zoom passcode: 941427

#### **Faculty:**

Mark Korson, MD  
Director  
VMP Genetics

Danae Bartke, BSED  
Executive Director  
HCU Network America

#### **Program Goal:**

- 1 Outline a metabolic differential diagnosis for homocysteinemia.
- 2 Review some important points about the diagnosis and management of cystathionine-beta-synthase deficiency.
- 3 Describe some real-life challenges facing patients with homocystinuria and their families.

**Target Audience:** Cardiovascular Disease, Endocrinology, Internal Medicine, Neurology, Nutrition, Ob/Gyn, Oncology, Ophthalmology, Pediatrics, Rheumatology, Urology, Multiple Specialties, Pathology, Otolaryngology, Orthopedic, Surgery / Transplant, Digestive Diseases, Vascular Medicine, Pulmonology, Hematology

#### *Financial Disclosure Information:*

*Dr. Mark Korson, faculty for this educational event, receives grant or research support from Recordati, and consulting fees from HemoShear & Tessera.  
All of the relevant financial relationships listed for these individuals have been mitigated.*

*Accreditation Statement: Yale School of Medicine is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians.*

*Designation Statement: Yale School of Medicine designates this Live Activity for a maximum of **1.00 AMA PRA Category 1 Credit(s)**<sup>™</sup>. Physicians should only claim credit commensurate with the extent of their participation in the activity.*

**For questions, email [james.long@yale.edu](mailto:james.long@yale.edu).**