



Presented by: Genetics

Heterozygous variants in MYH10 associated with neurodevelopmental disorders and congenital anomalies with evidence for primary cilia-dependent defects in Hedgehog signaling

5/17/2022 10:15:00 AM – 11:15:00 AM | SHM I-304

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: 29223

Also [via Zoom](#)

Meeting ID: 92732690431

Zoom passcode: 941427

Faculty:

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Program Goal:

- 1 To recognize a novel autosomal dominant cause of neurodevelopmental disorders and multiple congenital anomalies associated with the MYH10 gene to facilitate diagnosis of patients and families with this disorder.
- 2 To understand that altered primary cilia biology and disrupted Hedgehog signaling is a key feature of the mechanism of this disorder.
- 3 To appreciate an emerging link between actomyosin dynamics and primary cilia length control with implications for vertebrate organogenesis and disease states.

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

Planners for this activity: Yong-hui Jiang, MD/DO and James Long

Financial Disclosure Information:

None of the faculty/planners for this educational activity have relevant financial relationship(s) to disclose with ineligible companies.

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)**[™]. Physicians should only claim credit commensurate with the extent of their participation in the activity.*

For questions, email james.long@yale.edu, janet.hernandez@yale.edu.