

*Presented by: Genetics*

## **RASopathies at The Level of Ras Activation: A Group of Genetic Disorders with Overlapping Phenotypes and Unique Mechanisms**

10/26/2021 10:15:00 AM – 11:15:00 AM | Zoom

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: 29194**

### **Zoom Link**

passcode: 941427

#### **Faculty:**

Chen Zhao, PhD, ABMGG  
Assistant Director, DNA Lab  
Department of Genetics

#### **Program Goal:**

- 1 Review **Rasopathies** and variable syndromes caused by deregulation of RAS-MAPK activity at different steps of disruptions.
- 2 Discuss clinical cases of Rasopathies at the level of Ras activation step caused by disease-causing variants in GTPase such as NF1 or guanine nucleotide exchange factor such as SOS1.
- 3 Understand the the negative (GAPs) and positive regulators (GEFs) at the step of Ras activation and the corresponding loss-of-activity or gain-of-activity germline variants of these factors in Rasopathies.

**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: Rama Kastury, 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](mailto:james.long@yale.edu).**