

Presented by: Genetics

Identification of Structural Variants Using Next-Generation Sequencing Data for Rare Disease Diagnosis

12/6/2022 10:15AM – 11:15AM | 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: 34323

[Zoom Link](#)

Zoom passcode: 080122

Faculty:

Yong-hui Jiang, MD/DO

Professor

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Laboratory Genetics and Genomics Fellow

Mayo Clinic

Program Objective(s):

- 1 Review the methods of structural variant detection in clinical genetic testing
- 2 Learn how and what types of structural variants can be detected using sequencing methods
- 3 Understand the limitations of structural variant calling and how it can be improved

Target Audience: Cardiovascular Disease, Endocrinology, Gastroenterology, General Surgery, Internal Medicine, Neurology, Neurosurgery, Ob/Gyn, Oncology, Pediatrics, Rheumatology, Urology, Otolaryngology, Infectious Diseases, Nephrology, Digestive Diseases, Vascular Medicine, Pulmonology, Hematology

Planners for this activity: Yong-hui Jiang, MD and Jennifer Essandoh

Financial Disclosure Information:

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

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For questions, email Jennifer.Essandoh@yale.edu.