



Genetics Clinical Grand Rounds

Presented by
Department of Genetics
Yale School of Medicine

Regions of Homozygosity in Our Genome: Consanguinity, Uniparental Disomy and Recessive Single Gene Mutations

Jiadi Wen, FACMG

Date: March 23, 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

Chromosomal microarray platforms with single nucleotide polymorphism (SNP) probes can detect regions of homozygosity. This confers a potential utility in revealing consanguinity, autosomal recessive diseases and uniparental disomy related disorders.

LEARNING OBJECTIVES

- 1) Introduction: Diagnostic Implications of regions of homozygosity: Consanguinity, Uniparental Disomy, and Recessive Single-Gene Mutations
- 2) The additive diagnostic value on reporting regions of homozygosity for the detection of pathogenic variants and uniparental disomy in pediatric patients
- 3) Practice guideline in interpreting and reporting ROH and the clinical implications.

DESIGNATION STATEMENT

The Yale School of Medicine designates this live activity for 1 AMA PRA Category 1 Credit(s)TM. Physicians should only claim the credit commensurate with the extent of their participation in the activity.

FACULTY DISCLOSURES

Jiadi Wen, FACMG - none
Dr. Rama Kastury, DO - none

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