



Genetics Clinical Grand Rounds

Presented by
Department of Genetics
Yale School of Medicine

**“From genetic diagnosis to personalized genetic correction:
Analytical pipelines, data sharing and genetic therapies.”**

Monkol Lek, PhD

Date: November 5, 2019 10:15am
Location: SHM I-304

Course Director/Host: Dr. James McGrath, MD, PhD

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

Genomic sequencing is now in routine clinical practice. The current diagnosis rate is approximately 50% for most rare genetic diseases and will take a collaborative approach to improve this diagnosis rate. In this presentation we introduce infrastructure built at Yale that can be used for collaborative analysis and data sharing. In addition, highlighting our vision for implementing precision medicine using Duchenne muscular dystrophy as an example.

LEARNING OBJECTIVES

1. Broad overview of exome/genome sequencing analysis and interpretation including pipelines, tools and infrastructure available at Yale.
2. Matchmaker exchange node for the discovery of cases with a candidate novel gene. An introduction to scoring algorithms implemented in the Yale matchmaker exchange node, MatchY.
3. Case study of Duchenne muscular dystrophy mutations and the translation of genetic therapies.

DESIGNATION STATEMENT

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

FACULTY DISCLOSURES

Dr. Monkol Lek, PhD - none
Dr. James McGrath, MD, PhD - none

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