New England Regional Genetics Group

NERGG Annual Educational Conference 2020:
Learning Objectives

DAY 1

Considering the Field of Population Health … from the Genetics Perspective
Eden Haverfield, PhD, FACMG

1. State the role genetics can play in population health.
2. Summarize the current landscape of access to genetic health information.
3. Illustrate examples of how to implement a population health genetics program.

Ataxia Telangiectasia
Timothy Yu, MD, PhD/Victoria Suslovitch, MS, CGC/Jennifer Thornton/ Mehmet Kuzu

1. Identify antisense oligonucleotide therapies and translational medicine for neurogenetic diseases.
2. Examine the role of the genetic counselor in teaching and supporting families of a child with AT.
3. Summarize family life experience of a child with AT.

The Genetics of Epilepsy
Shelly Meitzler/Mustafa Sahin, MD, PhD/Annapurna Podouri, MD

1. Summarize the components of the clinical trials pipeline.
2. Examine the diagnostic process in the evaluation of epilepsy.

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DAY 2

Facioscapulohumeral Dystrophy
Madhuri Hegde, Ph.D., FACMG/June Kinoshita

1. State the clinical features and inheritance patterns of FSHD.
2. Indicate technological processes for diagnosis of FSHD.
3. Identify issues facing individuals with FSHD.

Prenatal Diagnosis: Bridging the Gap between the Laboratory and the Clinic
Christele du Souich, MSc, CGC, CCGC/Diane Allingham-Hawkins, Ph.D., FCCMG, FACMG

1. List some of the gaps in prenatal diagnosis between the laboratory and the clinic.
2. Describe some of the ethical, legal, and social issues facing the laboratory and clinic when it comes to prenatal diagnosis.
3. Apply creative solutions to common prenatal testing requests.

Our Journey with Justin: Embracing Life with a Rare Disease Mucopolysaccharidosis Type II (Hunter Syndrome)
Steven Gentile, MBA/Kim Gentile, MBA

1. List the clinical features of Hunter Syndrome.
2. Examine the diagnostic odyssey which can occur in identifying Hunter Syndrome.
3. Identify inclusionary activities to enhance the life experiences of an individual with a rare disorder.