

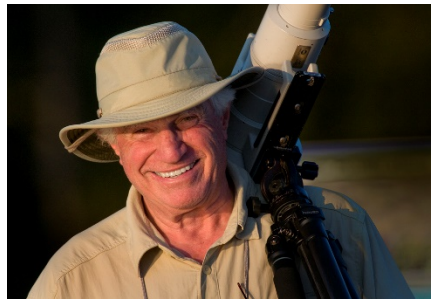


Finestone Visiting Professor in Medical Genetics

October 4, 2019, 10 - 11 am
MUHC Glen site, Room B 02.9390

Emerging Concepts in Glutaric Acidemias

Dr. Stephen I. Goodman
Distinguished University Professor
University of Colorado



Learning Objectives

Participants will be able to:

1. Recognize GA1 and GA2 in the absence of expanded newborn screening
2. Better understand where current concepts of biochemistry and disease pathogenesis fall short

Dr. Goodman and his colleagues at the University of Colorado have had a major impact on the field of inborn errors of metabolism. Accomplishments have included: (a) the development of gas chromatography-mass spectrometry (GC-MS) systems to screen for and characterize organic acidemias; (b) the first description of several inborn errors, most importantly glutaric aciduria type 1; and (c) the clinical, biochemical and molecular characterization of many inborn errors, including glutaric aciduria types 1 and 2 and several disorders of fatty acid oxidation.