POPULATION GENOMICS

SIXTH ANNUAL RARE DISEASE GENOMICS SYMPOSIUM

Friday, March 1, 2019

Bradley Lecture Center, Children's of Alabama

8 am — 3:45 pm Scientific Focus

Friday: $35 To register, visit ChildrensAL.org/genetics

FREE Parking in the 5th or 7th Avenue Children's Decks. Please follow the signs.

Questions? Contact Shaila Handattu at hande@uab.edu

LEARNING OBJECTIVES

Upon completion of this live activity, participants will be able to:

- Explain the goals of All of Us Research Program.
- Discuss the utility of genome sequencing in individuals affected with rare diseases.
- Identify the goals of the population cohort in the Alabama Genomic Health Initiative.
- Discuss genomic sequencing as a diagnostic tool for pediatric congenital disease.
- Discuss the potential and challenges of genomic diagnostics.
- Explain concepts of different mutation directed approaches and difficulties in their application.
- Explain the criteria of pathogenicity in variant interpretation.
- Learn about the use of zebrafish to model rare human disorders.
- Discuss the rationale and potential for applying yeast phenomics to rare disease.

CONTINUING EDUCATION

CME

Children's of Alabama designates this live activity for a maximum of *5.0 AMA PRA Category 1 Credits*™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Children's of Alabama is accredited by the Medical Association of the State of Alabama to provide continuing medical education for physicians.

BEYOND THE DIAGNOSIS ART EXHIBIT

Selected works from this traveling art exhibit, focusing on the rare disease patient, will be displayed at Children's of Alabama from mid-February through mid-March of 2019. Art has been used for thousands of years to successfully convey a message, whether it be a story or a glimpse into the human spirit. The purpose of this exhibit, presented by the Rare Disease United Foundation, is to encourage a look "beyond the diagnosis" to the patient.

POPULATION GENOMICS

SIXTH ANNUAL RARE DISEASE GENOMICS SYMPOSIUM

FRIDAY, MARCH 1, 2018

SESSION 1 ~ Bruce Korf, MD, PhD, Moderator
University of Alabama at Birmingham

8:30-8:45 Introduction and Overview
~ Bruce Korf, MD, PhD

8:45-9:20 AGHI-Affected Cohort
~ Anna C.E. Hurst, MD, MS

9:20-9:55 A Statewide Population Screening Initiate for Genetic Health Risk
~ Kelly East, MS, CGC

9:55-10:30 SouthSeq: Genomic Diagnosis for Ill Newborns Across the South
~ Greg Cooper, PhD

10:30-10:40 Break

10:40-11:15 All of Us Research Program
~ Bruce Korf, MD, PhD

11:15-12:15 KEYNOTE SPEAKER
Population Genomics, Precision Medicine and Rare Disease
~ David Goldstein, PhD

12:15-12:30 Break/Grab Lunch

SESSION 2

12:30-1:30 Parent Panel
~ Ashley Cannon, MS, PhD, Moderator
University of Alabama at Birmingham

SESSION 3 ~ Matthew Might, PhD, Moderator
University of Alabama at Birmingham

1:30-2:00 Development of Mutation Directed Therapeutics for NF1
~ Deeann Wallis, PhD

2:00-2:30 Fishing for Novel Treatments for Muscular Dystrophies
~ Matthew Alexander, PhD

2:30-2:40 Break

2:40-3:10 An Algorithm for Variant Interpretation
~ Mei-Jan Chen, PhD

3:10-3:40 Yeast Genetic Models to Repurpose Drugs for Individual Rare Diseases
~ John Hartmann, MD

3:40-3:45 Q & A and Wrap up

SPEAKERS

Matthew Alexander, PhD
University of Alabama at Birmingham

Mei-Jan Chen, PhD
University of Alabama at Birmingham

Greg Cooper, PhD
HudsonAlpha Institute for Biotechnology

Kelly East, MS, CGC
HudsonAlpha Institute for Biotechnology

David Goldstein, PhD
Columbia University Medical Center

John Hartmann, MD
University of Alabama at Birmingham

Anna C.E. Hurst, MD, MS
University of Alabama at Birmingham

Bruce Korf, MD, PhD
University of Alabama at Birmingham

Deeann Wallis, PhD
University of Alabama at Birmingham