

# 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



Children's  
of Alabama®



UAB SCHOOL OF  
MEDICINE  
The University of Alabama at Birmingham



Center for Clinical and Translational Science



RAREDISEASEDAY.ORG

CENTER FOR  
GENOMIC MEDICINE  
UAB | HUDSONALPHA

**Friday: \$35 To register, visit [ChildrensAL.org/genetics](http://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](mailto: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 Initiative 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