INFORMATICS APPLICATION IN RARE DISEASES

Friday, February 21, 2020

Bradley Lecture Center, Children's of Alabama

8 am — 3:30 pm Scientific Focus

SEVENTH ANNUAL RARE DISEASE GENOMICS SYMPOSIUM

Fee is $35 To register, visit ChildrensAL.org/genetics by Tuesday, February 18, 2020

FREE for students, Department of Genetics and Children's of Alabama professionals

FREE Parking in Children's 5th or 7th Avenue parking decks. Follow signs to Bradley Lecture Center.
Questions? Contact Shaila Handattu at hande@uab.edu

GLOBAL LEARNING OBJECTIVES

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

- Summarize the latest genomic discoveries and technology innovations driving rare disease research and precision medicine.
- Discuss approaches for applying genomics and bioinformatics to rare disease molecular diagnosis.
- Identify key barriers to connecting patients and providers with genetic counselors and keeping them engaged.
- Review deep phenotyping software for clinical genetics evaluation.
- Discuss practical strategies for integrating software into clinic.
- Review emerging trends how deep learning and bioinformatics strategies can be combined to bring new hope for devising cost-effective solutions to treat rare human genetic disorders.
- Discuss how AI can help with therapeutic identification in precision medicine.

CONTINUING EDUCATION

CME

Children's of Alabama designates this live activity for a maximum of 5.25 *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.

Children's of Alabama®

CCTS Center for Clinical and Translational Science

Alabama Genomic HEALTH INITIATIVE

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 2020. 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.

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FRIDAY, FEBRUARY 21, 2020
AGENDA

8:00-8:30 **Registration and Breakfast**
Doors open at 8:00 for Breakfast

SESSION 1
~ Matthew Might, PhD, Moderator

8:30-9:15 **Genome-Guided Rare Disease Molecular Diagnosis and Drug Prioritization**
~ Brittany N. Lasseigne, PhD

9:15-9:45 **Integration of Facial Analysis Software in Genetics Clinic**
~ Anna C.E. Hurst, MD, MS

9:45-10:30 **Leveraging Technology to Increase Genetic Counseling Access and Engagement**
~ Meagan B. Farmer, MS, MBA

10:30-10:45 **Break**

10:45-11:45 **KEYNOTE SPEAKER**
Computable Phenotyping for Diagnostics and Discovery
~ Melissa Haendel, PhD

11:45-Noon **Break/Pick Up Lunch**

SESSION 2
~ Ashley Cannon, MS, PhD, Moderator

Noon-1:00 **Parent Panel**
(CME credit is not offered for this session)

SESSION 3
~ Bruce Korf, MD, PhD, Moderator

1:00-1:45 **Identification and Interpretation of Molecular Variation in Rare and Not So Rare Human Disease**
~ Elizabeth Worthey, PhD

1:45-2:30 **Artificial Intelligence in Precision Medicine**
~ Matthew Might, PhD

2:30-3:15 **AI for Rare Disease Drug Repositioning**
~ Jake Y. Chen, PhD

3:15 **Q&A and Wrap Up**

SPEAKERS/MODERATORS

Ashley Cannon, MS, PhD
University of Alabama at Birmingham

Jake Y. Chen, PhD
University of Alabama at Birmingham

Meagan B. Farmer, MS, MBA
University of Alabama at Birmingham

Melissa Haendel, PhD
Oregon Health and Science University, Portland, OR

Anna C.E. Hurst, MD, MS
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Bruce Korf, MD, PhD
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Brittany N. Lasseigne, PhD
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Matthew Might, PhD
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Elizabeth Worthey, PhD
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