Rare Disease Genomics Symposium 2016

Friday, February 26

8 am-3:30 pm
Bradley Lecture Center
4th Floor, Children's Harbor Building

$15 to attend. To register, visit ChildrensAL.org/genetics

Questions? Please contact Shaila Handattu at hande@uab.edu

Children's of Alabama®

UAB MEDICINE GENETICS

CENTER FOR GENOMIC MEDICINE UAB | HUDSONALPHA

Heflin Center for Genomic Sciences

CCTS Center for Clinical and Translational Science

Agenda

8:00-8:30 am
Registration and Breakfast

8:30-8:35 am
Introduction and Welcome
Bruce Korf, MD, PhD
University of Alabama at Birmingham

8:35-9:20 am
Review of Genome Sequencing Efforts at Children's of Alabama *(.75 CME credits)*
*Bruce **Korf, MD, PhD***

9:25-10:10 am
Empirical Data on the Path to Genomic Medicine *(.75 CME credits)*
Robert Green, MD, MPH
Harvard Medical School

10:10-10:25 am
Break

10:25- 11:10 am
Rare (and not-so-rare) Tumors Associated with the DICER1 Syndrome *(.75 CME credits)*
Douglas R. Stewart, MD
National Cancer Institute

11:15 am-12:00 pm
Primary Immune Deficiency Update *(.75 CME credits)*
Thomas Prescott Atkinson, MD, PhD
University of Alabama at Birmingham

12:05-12:50 pm
Pediatric Cancer Genetic Counseling *(.75 CME credits)*
Meagan Farmer, MS
University of Alabama at Birmingham

12:50-1:50 pm
Lunch

1:50-2:35 pm
Whole Genome Sequencing in the Clinic *(.75 CME credits)*
David Bick, MD
HudsonAlpha

2:40-3:25 pm
Tuberous Sclerosis Update *(.75 CME credits)*
Martina Bebin, MD
University of Alabama at Birmingham

3:25-3:30 pm
Questions and Wrap Up

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Objectives

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

Review of Genome Sequencing Efforts at Children's of Alabama

1. Describe approaches to whole genome sequencing.
2. Explain process of referral of children to Undiagnosed Diseases Program.
3. Explain how mutations are validated as pathogenic.

Empirical Data on the Path to Genomic Medicine

1. List key knowledge gaps on the path to implementation of genomic medicine.
2. Cite the example of APOE genotype disclosure as a paradigm for understanding genetic risk disclosure.
3. Describe ongoing clinical trials in genomic medicine with adults and newborns.

Rare (and not-so-rare) Tumors Associated with the DICER1 Syndrome

1. Name rare and common cardinal features and tumors of the DICER1 syndrome.
2. Describe screening modalities for tumors associated with the DICER1 syndrome.
3. Recognize the importance of genetic testing in the DICER1 syndrome.

Primary Immune Deficiency Update

1. Recognize the problems posed in the genetic diagnosis of patients with complex immune deficiencies.
2. Differentiate different basic categories of primary immune deficiency.
3. Recognize the basic types of testing strategies in current use for diagnosis of patients with suspected primary immune deficiency.

Pediatric Cancer Genetic Counseling

1. Differentiate between hereditary, familial, and sporadic cancer.
2. Review hereditary cancer genetics basics.
3. Identify medical and family history characteristics that indicate need for genetics evaluation.
4. Describe role of cancer genetic counselor.

Whole Genome Sequencing in the Clinic

1. Select patients appropriate for Whole Genome Sequencing.
2. Discuss the limitations of Whole Genome Sequencing as a clinical test.

Tuberous Sclerosis Update

1. Recognize the consensus guidelines for diagnosing patients (infants, children and adults) with TSC.
2. Recognize the important diagnosis and management decisions for infants less than 24 months if diagnosed with TSC.
3. Recognize the potential therapeutic interventions available for patients diagnosed with brain or renal lesions related to TSC.

Continuing Education

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.

Rare Disease Day

February 29, 2016

rarediseaseday.org