

# 10th Annual Rare Disease Symposium

Topics include next generation technology, long-read genome sequencing, therapeutic sequencing, patient empowerment and health equity, panel discussions and more.

8:30-8:45

## Welcome/Introduction

**Bruce Korf, M.D., Ph.D.**

Associate Dean for Genomic Medicine  
Distinguished Professor, Department  
of Genetics | UAB Heersink School of  
Medicine, UAB | The University of  
Alabama at Birmingham

8:45-11:30

## Technology

8:45-9:30

## Next generation of evidence-based medicine for Rare Diseases: Power of Real-World Data and Genomics

**Praduman Jain**

Chief Executive Officer  
Vibrent Health

9:33-10:18

## Genomes for Everyone: The power and promise of personalized medicine

**Ryan Taft, Ph.D.**

Vice President, Scientific Research  
Illumina Inc.

10:20-11:05

## Long read genome sequencing for the diagnosis of neuro-developmental disorders

**Gregory Cooper, Ph.D.**

Faculty Investigator HudsonAlpha  
Institute for Biotechnology

11:05-11:30

## Panel Discussion

11:30-12:30

## Lunch/Networking

12:30-3:30

## Patient Empowerment and Health Equity

12:30-1:15

## KEYNOTE: Beyond Engagement: Patient Communities as Critical Drivers of Rare Disease Research

**Tania Simoncelli, M.S.**

Vice President, Science in Society  
Chan Zuckerberg Initiative

1:18-2:03

## The NORD Rare Disease Centers of Excellence Program: Collaborating to Address Unmet Needs

**Edward Neilan, M.D., Ph.D.**

Chief Medical and Scientific Officer  
National Organization for Rare Disorders

2:05-2:50

## Building a Community Engagement Model for Rare Disease: A Community Ambassador Approach

**Teneasha Washington, Ph.D., MPH, MBA**

Assistant Professor, UAB School of Public  
Health, Department of Health Behavior

2:50-3:15

## Panel Discussion

3:15

## Closing Remarks

## LEARNING OBJECTIVES

- 1 To discuss limitations of current DNA testing technologies that are in use for standard testing, newer "long-read" technologies and how they differ from current standard approaches, and the results from preliminary evaluation of long-read sequencing inpatients with undiagnosed neurodevelopmental disorders.
- 2 Share with the UAB+ community initial learnings from CZI's Rare As One Project that was launched in 2019 and has to date supported more than 80 patient-led organizations working to accelerate research in their disease areas.
- 3 Explain community based participatory research
- 4 Identify effective forms of community engagement

## CONTINUING EDUCATION

Children's of Alabama designates this live activity for a maximum of 2.5 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.

Questions? Contact Alli Romack at [alfaun@uab.edu](mailto:alfaun@uab.edu)