

**Dr. Richard Gibbs, AC Ph.D.**

1. Title of talk:

AHDC1 Update

2. Short bio:

**FOUNDING DIRECTOR, BAYLOR COLLEGE OF MEDICINE HUMAN GENOME SEQUENCING CENTER,  
WOFFORD CAIN PROFESSOR OF HUMAN AND MOLECULAR GENETICS,**

Richard Gibbs was born in Australia and graduated from Melbourne University in 1986 in Genetics and Radiation Biology. He moved to the USA to Baylor College of Medicine, where he performed key work on the molecular characterization of human genetic disease and provided an early leadership role for the Human Genome Project (HGP). In 1997 he founded the BCM Human Genome Sequencing Center (HGSC), one of five main groups to complete the HGP. The HGSC then led many genome projects for model species and contributed to the Mammalian Gene Collection, the Hapmap Project, the 1000 Genomes Project and the Cancer Genome Atlas. Since 2005, the group pioneered Personal Genomics, beginning with the personal genome sequences of James Watson and James Lupski and then developing the now ubiquitously used DNA exome capture technologies to create the first Genetics Diagnostics Laboratory to be based on whole gene sequencing. The group has made innumerable Mendelian disease gene discoveries and is now focused on accelerating clinical translation of genomics in adult care, via cardiovascular disease risk testing as well as being one of three centers to generate the data for the national All of Us Program.

Gibbs was elected to the National Academy of Medicine in 2011 and was recognized as one of the 400 most influential biomedical scientists in the world in 2013 (Eur J. Clin. Invest. 2014).

3. Brief description the talk:

Presentation will summarize developments in XGS and AHDC1 research.

**Dr. Jennifer Posey**

1. Title of talk:

The Therapeutic Landscape: challenges and opportunities

2. Short bio:

Dr. Posey is an Assistant Professor in the Department of Molecular and Human Genetics at Baylor College of Medicine. She completed her MD/PhD training at Baylor College of Medicine, and then went to Columbia University Medical Center in New York to train in Internal Medicine. She then returned to Baylor College of Medicine to do additional training in Medical Genetics and has since built her clinical and research programs at Baylor. Dr. Posey's research interests lie in the relationship between genomic variation and clinical (phenotypic) expression and how this can be leveraged to develop individualized treatment approaches. With Dr. Gibbs and Dr. Jim Lupski, she leads the Baylor College of Medicine Genomic Research to Elucidate the Genetics of Rare (BCM-GREGoR) research program.

3. Brief description the talk:

This talk will address the current status of therapeutics for rare disease, including gene therapy. The talk will highlight recent successes and important challenges and considerations in development of precision therapeutics.

### **Dr. Davut Pehlivan**

1. Title of talk:

Epilepsy in Xia-Gibbs syndrome

2. Short bio:

Dr. Pehlivan is an Assistant Professor in the Department of Pediatrics/Neurology at Baylor College of Medicine (BCM). He graduated from University of Istanbul and completed his genetic residency training at the same institute. He then moved to USA and joined the laboratory of James Lupski at BCM to study identifying genetic etiologies of many neurological conditions. He completed second residency in Child Neurology at BCM. He is a physician scientist at Texas Children's Hospital Rett Center and working to bring genetic-based treatments for neurogenetic disorders. He has co-authored over 120 articles and received numerous awards from national/international organizations.

3. Brief description the talk:

He will mention epilepsy and seizure in general, and seizures in Xia-Gibbs Syndrome.

### **Dr. Jianhong Hu**

1. Title of talk:

## The Xia-Gibbs Syndrome Registry: A Valuable Resource for Researchers and the Community

### 2. Short bio:

Dr. Hu is an associate professor at the Human Genome Sequencing Center at Baylor College of Medicine. She received her Ph.D. from Case Western Reserve University and did her postdoctoral training at University of Florida. She joined HGSC at Baylor College of Medicine in 2013. She is one of the key personnel to set up the HGSC Clinical Laboratory and she oversees the clinical lab. She has managed the XGS Registry ever since July 2019. Her research interest is in large cohort genomics and the study of genetic variants and the associated human diseases.

### 3. Brief description the talk:

Dr. Hu will talk about the importance of XGS Registry and current enrollment status.

## **Dr. Adam Hansen**

### 1. Title of talk:

The Xia-Gibbs Syndrome Registry App and Platform: Geneial

### 2. Short bio:

Dr. Adam Hansen is cofounder and CEO of Geneial. Ten years ago he dedicated his career to relieving human suffering through entrepreneurship in precision medicine, a path which led him to Houston for PhD training under Dr. Richard Gibbs, where he led the discovery of more than 150 candidate genetic disorders. In 2022 he was the first recipient of the Small Business Transition Grant for Early Career Scientists from the National Human Genome Research Institute, and has since received multiple additional NIH grants in support of Geneial's vision of private, community-led precision and genetic medicine. He was previously a digital health fellow at the Texas Medical Center in Houston and director of consulting at Enventure, a Houston-based grassroots innovation nonprofit supporting life sciences innovation.

### 3. Brief description the talk:

An overview of the what, where, when, why, who, and how of participating in the Xia-Gibbs Syndrome Registry via the Geneial app. An update on relevant Geneial progress since the September 2023 XGS Society webinar will also be shared.

## **Dr. Anthony Oro**

### 1. Title of talk:

Why does Xia-Gibbs Syndrome affect so many parts of the body

### Short bio:

Anthony (Tony) Oro MD/PhD is the Eugene and Gloria Professor of Dermatology at Stanford University School of Medicine and Co-Director of the Maternal and Child Health Research Institute. Dr. Oro is a trained developmental geneticist and clinician. He studied Drosophila genetics while a graduate student at the Salk Institute in La Jolla with Ronald Evans, and human genetics as a post-doctoral fellow in Matthew Scott's lab at Stanford. His research interests focus on human developmental genetics, tissue regeneration, and carcinogenesis.

### Brief description the talk:

Small changes in a person's DNA can have effects all over the body in ways that seemingly don't make sense or lead to specific therapy suggestions. Our work on the gene affected in XGS suggests that it regulates the mesoderm, the middle layer of the embryo, that interacts with the other parts of the developing body, explaining why so many tissues are affected. Our mechanistic studies suggest that AHDC1 reads local positional and timing information that can affect both development and ongoing health issues in adult patients.

## **Dr. Bo Yuan**

### Title of talk:

What is genetic variation and what does it mean to your health?

### Short bio:

Bo Yuan is an Associate Professor with the Human Genome Sequencing Center at Baylor. He is a board certified clinical molecular geneticist who has spent most of his career in understanding genetic variations and uses evidence-based medicine to interpret their role in our health. He has discovered and delineated mechanisms contributing to diseases such as Yuan-Harel-Lupski syndrome, Alzami-Yuan syndrome, BAFopathies, chromatinopathies, etc. He leads clinical genomic interpretation in the HGSC focusing on a variety of topics such as Cardio-metabolic conditions, epilepsies, and general health screening.

### Brief description the talk:

In his presentation, Bo will go over the genetic variations and how they contribute to our health.