Global Science Summit: Focus on Angelman Syndrome Translational Research

Visit our vendors and sponsors throughout the day - located in the Expo Hall and on the virtual platform

AGENDA

All times are CST (Austin, Texas)

9:00 AM - 9:50 AM

KEYNOTE SPEAKER

Jan A. Nolta, Ph.D.,
Director of the Stem Cell Program at University of California, Davis School of Medicine
Director of the Institute for Regenerative Cures

STEM CELL AND GENE THERAPY PLATFORMS

The Stem Cell Program and Gene Therapy Center at the University of California Davis Medical Center works with a wide range of stem cell therapies, gene therapy and gene editing approaches. The different platforms that could apply to neurological disorders like Angelman syndrome will be discussed.

9:55 AM - 10:55 AM

Allyson Berent, DVM, DACVIM
Chief Science Officer, FAST

OVERVIEW OF THE THERAPEUTIC LANDSCAPE FOR ANGELMAN SYNDROME

Dr. Berent will give an overview of FAST's mission and vision through an aggressive drug development roadmap. She will lay out FAST's robust efforts involving deep collaborations with academic, industry and regulatory partners and share an overview of the current therapeutic landscape for Angelman syndrome.

11:00 AM - 11:15 AM

BREAK

11:15 AM - 11:55 AM

Kyle Fink, Ph.D.; David Segal, Ph.D.; Jill Silverman, Ph.D.
University of California, Davis

THE CREATION OF A ROBUST INFRASTRUCTURE INCLUDING MOLECULAR, NEUROBEHAVIOR, AND BIOMARKER TESTING FOR PRE-CLINICAL DRUG EVALUATION IN ANGELMAN SYNDROME

A panel from UC Davis will discuss how an AS pre-clinical infrastructure was created for testing new therapeutic compounds.

12:00 PM - 12:20 PM

Yong-Hui Jiang, M.D., Ph.D.
Professor of Genetics, Neuroscience, and Pediatrics at Yale University School of Medicine and Chief of Medical Genetics at Yale Medicine

UPDATE ON ANGELMAN SYNDROME HUMAN IPSC BIOREPOSITORY PROJECT AND THE ANGELMAN SYDROME LARGE DELETION MOUSE MODEL.

Dr. Jiang will present an update on the progress of the Angelman syndrome IPSC (induced pluripotent stem cell) biorepository and the creation of a large deletion AS Mouse model.

12:25 PM - 12:45 PM

Albert Keung, Ph.D.
Assistant Professor of Chemical and Biomolecular Engineering at North Carolina State University

ENGINEERING HUMAN STEM CELL MODELS FOR MULTIPLE ANGELMAN SYNDROME (EPI)GENOTYPES

Dr. Keung will describe his efforts to create human stem cell lines and organoids that can capture the different types of genetic changes that can lead to Angelman syndrome. These models may help to better understand potential differences in responses to therapeutics and to find therapies tailored to each specific form of Angelman syndrome.

12:50 PM - 1:35 PM

LUNCH

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1:35 PM - 1:50 PM

Ryan Butler, Ph.D.
Assistant Professor of Chemical and Biomolecular Engineering at University of Texas Southwestern Medical Center

GENE THERAPY FOR ANGELMAN SYNDROME THROUGH RNA INTERFERENCE

This presentation will examine the potential for vectorized shRNA to induce paternal expression of *UBE3A* through the "stop-the-stop" approach. We have tested on mouse and human cell lines while concurrently testing in a mouse model of Angelman syndrome.

1:55 PM - 2:05 PM

Jessica Duis, M.D., M.S.
Pediatrician and Medical Geneticist at Children's Hospital of Colorado

WE'RE THERE WHEN YOU NEED US: A 24-7 EMERGENCY CARE HOTLINE FOR THE AS COMMUNITY

In July 2021, a 24/7 Emergency Care Hotline for the management of Angelman syndrome launched, offering provider-to-provider consultation for individuals with any Angelman syndrome-related concerns from seizures to GI conditions. Dr. Duis will discuss the structure of the hotline service, the team's experiences to date, and some of the barriers to ensure acccess to providers is made simple.

2:10 PM - 2:35 PM

Scott Dindot, Ph.D.
Associate Professor of Genomics and EDGES Fellow at Texas A&M University
Executive Director of Molecular Genetics at Ultragenyx Pharmaceutical

TRANSLATIONAL RESEARCH IN A LARGE ANIMAL MODEL OF ANGELMAN SYNDROME

A pig model was developed to advance and understand the pathophysiology of Angelman syndrome, facilitating the development of translational therapies to human patients. The learnings to date will be discussed.

2:40 PM - 3:20 PM

Jim Wilson, M.D., Ph.D.
Director, Gene Therapy Program and Orphan Disease Center, Professor in the Perelman School of Medicine at the University of Pennsylvania

GENETIC APPROACHES FOR TREATING ANGELMAN SYNDROME

Dr. Wilson will describe 3 different platforms his team is investigating to treat Angelman syndrome in a murine model. This includes AAV gene replacement, AAV-mediated expression of microRNAs, and genome editing.

3:25 PM - 3:40 PM

AFTERNOON BREAK

3:40 PM - 4:25 PM

Joseph Anderson, Ph.D.
Associate Professor at the University of California, Davis Department of Internal Medicine
UC Davis Stem Cell Program
Mehrdad Abedi, M.D.
Professor Hematology and Oncology, University of California, Davis

HEMATOPOIETIC STEM CELL GENE THERAPY FOR ANGELMAN SYNDROME: PROGRESS AND PROCESS

The proof-of-concept work on utilizing a lentiviral vector hematopoietic stem cell gene therapy approach for Angelman syndrome has proven effective at both preventing and reversing phenotypes in an immunodeficient newborn and adolescent animal model of AS. This talk will focus on the path forward toward a human clinical trial for both adult and pediatric patients living with Angelman syndrome.

4:30 PM - 5:30 PM

PANEL DISCUSSION WITH RESEARCHERS

DRS. JAN NOLTA, KYLE FINK, DAVID SEGAL, JILL SILVERMAN, YONG-HUI JIANG, ALBERT KEUNG, RYAN BUTLER, JESSICA DUIS, SCOTT DINDOT, JIM WILSON, JOSEPH ANDERSON, MEHRDAD ABEDI

Global Science Summit: Focus on Pharma

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AGENDA

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8:30 AM - 9:10 AM

KEYNOTE SPEAKER

David Segal, Ph.D.
University of California, Davis
NOW IS THE TIME FOR MOLECULAR THERAPIES FOR ANGELMAN SYNDROME
Dr. Segal will discuss therapies targeting the molecular basis of Angelman syndrome and why hope for this community has never been higher.

9:15 AM - 9:30 AM

Laurent Servais, M.D, Ph.D.
Professor of Pediatric Neuromuscular Diseases at the University of Oxford,
Invited Professor at the University of Liege, Belgium
UPDATE ON THE UK NATURAL HISTORY STUDY AND WEARABLE DEVICES
Dr. Servais will present the set up and basline data of the first patients included in the UK AS natural history study.

9:35 AM - 9:55 AM

Emil Kakkis, M.D., Ph.D.
CEO of Ultragenyx Pharmaceutical Inc.
DEVELOPMENT OF RARE DISEASE THERAPIES: OVERCOMING CHALLENGES
Ultragenyx Pharmaceutical has partnered with GeneTx Biotherapeutics on the development of GTX-102 for Angelman syndrome and have had an eventful year. Dr. Kakkis will talk about the prospect of benefit and the challenges in drug development for rare diseases like Angelman syndrome.

10:00 AM - 10:20 AM

Brenda Vincenzi, M.D.
Senior Medical Director for the Angelman Syndrome program, Roche Pharmaceuticals
ROCHE PHARMACEUTICALS ANGELMAN SYNDROME PROGRAM UPDATE TO THE FAST COMMUNITY
This session will focus on a general update to the FAST community on TANGELO and FREESIAS.

10:25 AM - 10:45 AM

Scott Stromatt, M.D.
Chief Medical Officer at GeneTx Biotherapeutics
Elizabeth Berry-Kravis, M.D., Ph.D.
Professor of Pediatrics, Neurological Sciences, and Biochemistry at Rush University Medical Center in Chicago
GTX-102 PHASE 1/2 CLINICAL TRIAL UPDATE, DEVELOPMENT OF AN ASO FOR ANGELMAN SYNDROME: SCIENCE AND REGULATION
GeneTx Biotherapeutics will provide an update on the Phase 1/2 clinical trial of GTX-102, an intrathecally delivered antisense oligonucleotide (ASO) for the potential treatment of Angelman syndrome.

10:50 AM - 11:10 AM

Becky Crean, Ph.D.
Executive Director, Clinical Development at Ionis Pharmaceuticals
IONIS PHARMACEUTICALS ANGELMAN SYNDROME PROGRAM UPDATE
Ionis Pharmaceuticals will provide an update on their Angelman syndrome program status and preliminary clinical trial design.

11:15 AM - 11:30 AM

BREAK

11:30 AM - 11:40 AM

Kyle Fraser, Ph.D.
Biomarker Lead Scientist for the Angelman Syndrome Program at Biogen Inc.
UPDATE ON BIOGEN INC. PRE-COMPETITIVE CSF COLLECTION STUDY
Biogen Inc. will provide an update on their pre-competitive CSF Collection Study.

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11:45 AM - 12:05 PM

Larry Glass,
Chief Science Officer, Neuren Pharmaceuticals Ltd.
Nancy Jones, Ph.D.
Vice President, Clinical Development
Neuren Pharmaceuticals Ltd.

NNZ-2591 AS A POTENTIAL TREATMENT FOR ANGELMAN SYNDROME

NNZ-2591 is a novel drug being developed by Neuren Pharmaceuticals for the treatment of Angelman syndrome. This presentation will provide an overview of NNZ-2591, why it might be a potentially useful treatment for Angelman syndrome, and give an update on the development program.

12:10 PM - 12:30 PM

Emily Mcginnis,
Chief Patient Officer and Head of Government Affairs, Taysha Gene Therapies
Suyash Prasad, M.D.
Chief Medical Officer and Head of Research and Development, Taysha Gene Therapies

PUTTING PATIENTS AT THE CENTER

Taysha Gene Therapies, a patient-centric company focused on discovering and developing novel gene therapies for devastating disorders of the central nervous system (CNS), will share its investigational approaches to treating Angelman syndrome.

12:30 PM - 1:30 PM

Panel Discussion with Pharma

ASK YOUR QUESTIONS TO OUR SPEAKERS IN A PANEL DISCUSSION.

Ultragenyx Pharmaceutical Inc., GeneTx Biotherapeutics, Ionis Pharmaceuticals, Biogen Inc., Roche Pharmaceuticals, Taysha Gene Therapies, PTC Therapeutics and Neuren Pharmaceuticals Ltd.

1:30 PM

GLOBAL EDUCATIONAL SUMMIT ON DEMAND

ON DEMAND

Amy Bereiter, MS CCC-SLP, ATP

AAC AND LANGUAGE DEVELOPMENT

Many children with Angelman Syndrome utilize AAC for communication. This session will specifically focus on AAC users and provide an overview of language development, considerations for language intervention, and ways to monitor language growth.

ON DEMAND

Hilda Tourians & Yuji Oka

YOGA & MOVEMENT FOR EVERY ABILITY

Individuals living with Angelman syndrome need stimulation and exercise to assist with mobility and body/mind connection. Learn techniques for yoga and movement for every ability.

ON DEMAND

Meagan Cross
FAST Australia chairperson

THE IMPORTANCE OF THE GLOBAL ANGELMAN SYNDROME REGISTRY

Why sharing data on your experiences with Angelman syndrome is important and how your information can inform research and clinical trials. Meagan demonstrates the registry's new features, functionality, ease of use and value of joining the 1700+ participants worldwide.

AGENDA

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GLOBAL EDUCATIONAL SUMMIT ON DEMAND

ON DEMAND

Michele Speckman; M.Ed, Justin Paul; Amanda Black-Ingersol M.Ed; Margie Borawska Ph.D.
TRANSITION: PREPARING FOR SUCCESS THROUGH PLANNING, TEAMWORK, AND NATURAL OPPORTUNITIES FOR LEARNING
The panel will discuss how to successfully plan and implement transition into high school. Additionally, they will discuss programming and planning for life after high school.

ON DEMAND

Tami Hicks, M.Ed.
FAQ ABOUT ANGELMAN SYNDROME AND EDUCATION
Within the realm of education in the United States, there are an abundance of terms, laws, and questions that arise pertaining to children with Angelman Syndrome. Everything from safety, goals, communication, rights, how to voice frustrations, paraprofessionals and so on. Tami Hiks will shed light on these popular topics and provide suggestions for how to go about finding answers to your questions within your child's educational journey.

ON DEMAND

Kate Ahern, M.S.Ed.
AIMING HIGH
Starting in March 2020 AAC Voices began offering online classes and groups to children and young adults who use augmentative and alternative communication (AAC). We will explore the progress made in communication, literacy, and numeracy, as well as social and emotional learning by learners with AS who attend. Time will be taken to discuss effective educational strategies for teaching these amazing students.