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2024

global science summit

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Global Science Summit
Focus on
Angelman Syndrome
Translational Research

Friday, November 8, 2024
8:30 AM — 5:45 PM

Global Science Summit
Focus on Clinical Trials

Saturday, November 9, 2024
8:30 AM — 1:00 PM

Event App

For more information, including speaker bios and a list of exhibitors, or to watch the event live, download the Global Science Summit & Gala app by following the steps below:

1. Scan the QR code below to download the "Cvent Events" app.
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Friday, November 8, 2024
8:30 AM – 5:45 PM

Focus on Angelman Syndrome Translational Research

Time	Talk Title	Pillar	Speaker
8:30 AM	Making the Future: FAST's Mission and Strategy		Alana Newhouse Ryan Jacob *FAST*
9:05 AM	From Benchside to Bedside: FAST's Roadmap to a Cure		Allyson Berent, DVM, DACVIM *FAST and AS²Bio*
10:00 AM	Angelman Syndrome: From Discovery to Transformative Treatments		Arthur Beaudet, MD *Baylor College of Medicine*
10:30 AM	**Break**		
10:45 AM	Therapeutics 101	1,2,3	Edwin Weeber, PhD *FAST*
	Pillar 1		
11:25 AM	Backstage Passes to the Brain for Therapeutic Delivery	1	Barbara Bailus, PhD *Keck Graduate Institute*
11:45 AM	Hematopoietic Stem Cell Gene Therapy for the Potential Treatment of Angelman Syndrome	1	Christopher Luthers *University of California, Los Angeles*
12:05 PM	CRISPR Activation: How We are Addressing the Genes Outside of UBE3A for Individuals with Deletions	1	Nadav Ahituv, PhD *University of California, San Francisco*
	Pillar 2		
12:15 PM	The Tale of Two Therapies for Angelman Syndrome	2	David J. Segal, PhD *University of California, Davis*
12:35 PM	Non-Viral Gene Editing for Angelman Syndrome: Progress and Updates	2	Yong-Hui Jiang, MD, PhD and Jiangbing Zhou, PhD *Yale School of Medicine*
12:55 PM	**Lunch**		

Friday, November 8, 2024
8:30 AM – 5:45 PM

	Pillar 3		
1:40 PM	New Insights into Angelman Syndrome: Exploring UBE3A's Diverse Role in Brain Development and Health	3	Yu-Wen Alvin Huang, MD, PhD *Brown University*
2:00 PM	BDNF Signaling in Angelman Syndrome: Impact and Next Steps	3	John Marshall, PhD *Brown University*
2:20 PM	How You CAN Get More Involved in the FAST Mission		Elizabeth O'Connor and Meghan Edberg *FAST*
	Pillar 4		
2:35 PM	iPSC Biorepository: How Patient Contribution Supports Drug Development	4	Xiaona Lu, MD, PhD *Yale School of Medicine*
2:50 PM	Contribution of GABA-A Receptor Subunit Deletion to Angelman Syndrome Pathophysiology	4	Eric Levine, PhD *University of Connecticut* *School of Medicine*
3:05 PM	ABOM Goal/Purpose/Mission: What Matters, What is Meaningful, What is Measurable?	4	Allyson Berent, DVM, DACVIM
3:25 PM	Updates on the Angelman Syndrome Natural History Study	4	Anjali Sadhwani, PhD *Boston Children's Hospital*
3:45 PM	**Break**		
4:00 PM	Clinical Trials: Participant's Rights and Responsibilities	4	Niki Armstrong, MS, CGC *FAST*
4:15 PM	Hope in Action: Update on Advocacy Efforts	4	Ryan Fischer *FAST*
4:40 PM	Your Data Matters: GASR and LADDER Synergies	4	Niki Armstrong, MS, CGC Anne C. Wheeler, PhD *RTI International*
4:55 PM	FAST Across Borders - Highlights from FAST Affiliates Worldwide		Ellen Koekoeckx *FAST* Global Leads
5:15 PM	Panel Discussion		

Saturday, November 9, 2024
8:30 AM – 1:00 PM

Focus on Clinical Trials

Time	Talk Title	Pillar	Speaker
8:30 AM	Keynote: Breaking New Ground: Exploring Therapeutic Strategies for AS		Edwin Weeber, PhD *FAST*
9:05 AM	Controlled Clinical Trials: What Does it Mean and Why is it Often Needed		Jennifer Panagoulias, RAC *FAST and AS²Bio*
	Pillar 1		
9:20 AM	GTP-220: A Gene Replacement Therapy for Angelman Syndrome	1	James M. Wilson, MD, PhD *GEMMA Biotherapeutics*
	Pillar 2		
9:50 AM	ETX201: A Vectorized miRNA-Based Approach for the Potential Treatment of Angelman Syndrome	2	Sirika Pillay, PhD *Encoded Therapeutics*
10:10 AM	Ionis' Program Update of ION582 for Individuals Living with Angelman Syndrome	2	Rebecca Crean, PhD *Ionis Pharmaceuticals*
10:45 AM	A GTX-102 Clinical Development Update: Results from the Phase 1/2 Open-Label Study and an Overview of the Pivotal Phase 3 Study	2	Kimberly Goodspeed, MD *Ultragenyx*
11:20 AM	Break		

Time	Talk Title	Pillar	Speaker
11:35 AM	Industry Update from Roche: Rugonersen Tangelo Study and Alogabat Alderbaran Study	2, 3	Joerg Hipp, PhD *Roche Pharmaceuticals*
	Pillar 3		
12:10 PM	Results of the Phase 2 Study of NNZ-2591 for Angelman Syndrome	3	Nancy E. Jones, PhD *Neuren Pharmaceuticals*
12:25 PM	Panel Discussion		

Have questions?

Wondering how everything you learned applies to your loved one?

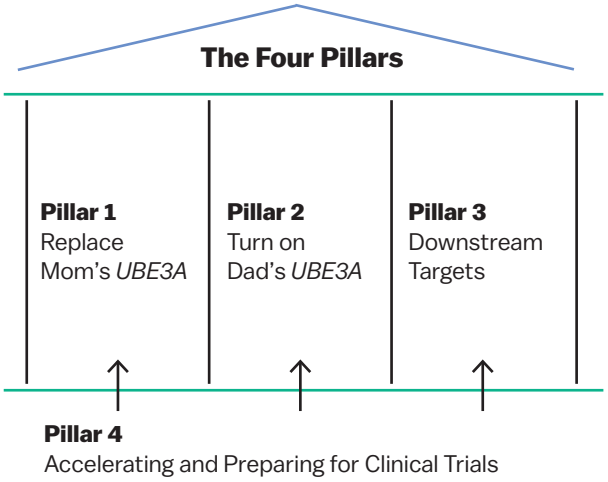
If you reside in the USA,
you can schedule a 1:1 meeting
with FAST's certified genetic counselor.

Scan the QR code below
to make an appointment.

SCAN ME

Our Strategic Roadmap to a Cure

In order to achieve a future in which there are multiple approved therapeutics—which is what will be needed for a syndrome in which there are many genotypes, not to mention unknown ways that different people will respond to different treatments—we need as many shots on goal as possible. And we need them to move through that pipeline quickly, but also safely. We have divided our work into four pillars.



Pillar 1

Replacing the *UBE3A* gene or protein in neurons

Programs in the pipeline:

AAV-GT
Adeno-Associated Virus Gene Therapy

HSC-GT
Hematopoietic Stem Cell Gene Therapy using a Lentivirus

ERT
Enzyme Replacement Therapy

CRISPRa
Clustered Regularly Interspaced Short Palindromic Repeats Activation

Pillar 2

Activating the silent paternal copy of the *UBE3A* gene in neurons

Programs in the pipeline:

ASO
Antisense Oligonucleotides

ATF
Artificial Transcription Factors / Zinc Fingers

shRNA/miRNA
Short-hairpin RNA / micro-RNA

CRISPR
Clustered Regularly Interspaced Short Palindromic Repeats

Small Molecule
Small molecules that inhibit the long noncoding RNA *(UBE3A-ATS)*

Pillar 3

Downstream targets to treat symptoms

Focus on different molecular pathways and effector proteins impacted by the missing UBE3A protein. These drugs generally aim to improve the communication of neurons at the synapse (junction between the two neurons) having a "downstream" effect from the gene or protein itself.

Learn More

This Roadmap to a Cure 2.0 was designed by Dr. Allyson Berent, Chief Science Officer of FAST.

Mother of three daughters—one of whom, Quincy, lives with Angelman syndrome

Check out the AS Glossary in the back!

Pillar 4

Accelerating and preparing for clinical trials

Often overlooked but wildly important! These are the actions that allow all of the work from Pillars 1 through 3 to reach individuals living with Angelman syndrome to ensure transformative therapeutics are advanced.

Creation of animal models and cell lines for each genotype

Angelman Syndrome Biomarker and Outcome Measure Consortium (A-BOM)

Global Angelman Syndrome Registry (GASR)

AS Natural History Study

FAST Search & Rescue

The RUSH F.A.S.T. Center for Translational Research

Newborn Screening

Angelman Syndrome (AS)
Glossary

Angelman Syndrome Biomarker and Outcome Measure Consortium (A-BOM)
A precompetitive consortium that brings together industry, clinicians, patient advocacy groups and researchers to advance the regulatory science for Angelman syndrome (AS) in order to understand, develop, test, and validate different tools that can be used to assess meaningful clinical outcomes in clinical trials for individuals living with AS.

Adeno-Associated Virus - Gene Therapy (AAV-GT)
A therapeutic approach where a healthy copy of a virus, Adeno-Associated Virus (AAV), is used to carry a healthy copy of a gene to a target organ. This is most commonly delivered in-vivo (inside the body). For Angelman syndrome (AS) the healthy copy of the missing or non-functional *UBE3A* gene is packaged inside the AAV, and is injected into the fluid that surrounds the brain, called the cerebrospinal fluid (CSF). Once in the fluid, the virus and gene can directly reach important cells of the brain, called neurons, and replace the non-functional or missing copy of the gene.

Allele
A specific version of a gene. Each individual has two alleles for each gene, one inherited from their mother and one from their father.

Angelman Syndrome (AS)
A rare neurogenetic disorder that affects approximately 1 in 15,000 people – or an estimated 500,000 individuals worldwide; a single-gene disorder caused by loss of function of the *UBE3A* gene on the maternal allele located in the 15q11.2-13.1 region.

Antisense Oligonucleotides (ASO)
A therapeutic approach that uses modified RNA or DNA molecules that bind to the RNA of the *UBE3A-ATS* (*UBE3A* antisense transcript). The *UBE3A-ATS* is responsible for silencing the paternal *UBE3A* gene through a process called imprinting. In binding to the targeted antisense transcript, the ASO stops the *UBE3A-ATS* from silencing the expression of the paternal *UBE3A* gene.

Artificial Transcription Factors/Zinc Fingers (ATF-ZF)
A therapeutic approach that consists of using engineered proteins designed to regulate gene expression in a highly specific manner. They are constructed to bind at specific DNA sequences and can either activate or repress the expression of target genes. ATF-Zinc fingers are small proteins that use zinc ions to stabilize their structures giving them a finger-like appearance. ATF-ZFs can bind to the *UBE3A-ATS* and prevent the silencing of the paternal copy of the *UBE3A* gene.

Biomarker
A biomarker (biological marker) is a measurable indicator in the body that helps assess the presence of a disease or monitor the effectiveness of a treatment. Examples in AS could include changes in brain activity (i.e. EEG) or changes in blood or other body fluids (e.g. levels of certain proteins like UBE3A in the CSF). To be used in a clinical trial, biomarkers must be validated to prove that they are reliable, associated with a specific symptom or condition, and work in many different individuals living with the condition. Validated biomarkers can provide objective evidence that a potential therapy is improving a condition or symptom.

Blood Brain Barrier (BBB)
A tightly locked layer of cells that acts as a filter and protects your brain from harmful substances, infections (e.g. virus) and other things that could cause damage. It also helps to keep beneficial chemicals inside the brain. It's a key part of maintaining brain health and isolating the brain for chemicals and substances that travel throughout your blood. Due to this protective layer, it is difficult to get large substances across it from the blood and into the brain.

Brain-Derived Neurotrophic Factor (BDNF)
A protein known to be reduced in AS that plays a crucial role in synaptic function and is essential for the process underlying learning and memory. BDNF exerts its effects by binding to specific receptors on the surface of neurons, mainly the TrkB (Tropomyosin receptor kinase B) receptor. The BDNF-TrkB receptor represents a potential downstream therapeutic target.

Clinical Global Impression (CGI)
A scale, typically completed by an expert health care provider, that provides a rating of overall illness severity, improvement and response to treatment.

Central Nervous System (CNS)
The CNS is the majority of the nervous system including the brain and spinal cord.

Clinical Trial
A clinical trial is a prospective research study, generally on human participants, designed to answer specific medical questions. This includes both interventional trials to evaluate the safety, effectiveness, and side effects of any form of a medicine in human patients or observations trials to evaluate the function of individuals over time (e.g. natural history study [NHS]).

Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR)
A gene editing tool that utilizes a guide RNA (gRNA) to recognize a specific section of DNA or RNA and directs an enzyme (nuclease) to cut at a specific section in the DNA or RNA. Designing a CRISPR that affects the UBE3A-ATS could potentially allow the paternal *UBE3A* gene to be turned on. It could also be used to edit a mutation in a gene, known as base editing.

CRISPRa or CRISPR-activation
A gene modulation technology that utilizes a nuclease-deactivated Cas9 protein that binds to the target genomic region with the same efficiency as Cas9, but does not cut the DNA and instead can exert RNA-directed transcriptional control of the targeted gene with a goal to upregulate a specific gene.

Cerebrospinal Fluid (CSF)
A clear colorless body fluid found in the space that surrounds the brain and spinal cord. CSF is produced by specialized cells, called ependymal cells, in the blood vessels (choroid plexus) of the ventricles of the brain. This fluid is being turned over and replenished throughout the day.

Delivery
The mechanism in which a therapeutic is transported to the central nervous system of the body; delivery includes the route of administration (e.g. brain, spinal cord, blood vessels, bone marrow, etc.) and how the therapeutic reaches its target location (e.g. carrier of the therapeutic like viral vector, bone marrow stem cells, RNP, etc.).

DNA (deoxyribonucleic acid)
The molecule that carries genetic information for the growth, development and function of an organism. DNA is composed of four chemicals, which are abbreviated A, T, C, and G (A=adenine, T=thymine, C=cytosine, and G=guanine). Segments of the DNA, called genes, provide the instructions for proteins, with the ATCG chemicals making up the code for the protein. DNA has two strands of the chemicals that wind around themselves to form a double helix.

Downstream Targets
A therapeutic approach in Angelman syndrome that focuses on different molecular pathways and effector proteins impacted by the missing or non-functional UBE3A protein.

Dystonia
A neurological movement disorder characterized by uncontrolled (involuntary) muscle contractions that cause repetitive or twisting movements or abnormal postures.

E3 Ubiquitan Protein-Ligase Gene (Ube3a) Rodent
The gene that codes for the Ube3a protein in rodents. *Ube3a* is generally expressed from both the maternal and paternal alleles throughout the body, but in neurons, the cells of the brain, only the maternal copy of the *Ube3a* gene is expressed. This is due to the imprinting phenomenon where the *Ube3a-ATS* is silencing the paternal *Ube3a* gene from being expressed. Please note that you will see this gene in italics (whereas the protein is not in italics) and the gene is in lower case letters for rodents while in capital letters for humans and non-human primates.

E3 Ubiquitan Protein-Ligase Mouse Protein *(Ube3a)* Rodent
Ube3a is the rodent model protein coded by the *Ube3a* gene. The Ube3a protein has many of the same functions as the human UBE3A protein. The conservation of function between humans and animals allows researchers to use the AS animal model and test therapeutic efficacy.

E3 Ubiquitin Protein-Ligase (UBE3A)
UBE3A is the human protein coded by the *UBE3A* gene. Absence or loss of function of UBE3A causes AS. UBE3A is a protein with many functions in the human body including targeting other proteins for removal. Note that the human UBE3A protein is capitalized but not italicized. The rodent Ube3a protein is in lower case and not italicized.

E3 Ubiquitin Protein-Ligase Antisense Transcript (UBE3A-ATS)
The long, noncoding piece of RNA that blocks paternal *UBE3A* gene expression in humans.

E3 Ubiquitin Protein-Ligase Antisense Transcript *(Ube3a-ATS)* Mouse
The long noncoding piece of RNA that blocks paternal *Ube3a* expression in rodent models.

E3 Ubiquitin Protein-Ligase Gene *(UBE3A)*
The gene that codes for the UBE3A protein in humans. *UBE3A* is on chromosome 15, in the 15q11.2-13.1 region and is generally expressed from both the maternal and paternal copies throughout the body. In neurons (cells of brain), only the maternal copy of the *UBE3A* gene is expressed. This is called imprinting. The *UBE3A-ATS* is silencing the paternal *UBE3A* gene from being expressed. Please note that the human *UBE3A* gene or RNA is capitalized and italicized. The rodent *Ube3a* gene or RNA is in lower case and italicized. The protein for both mouse and human are not italicized.

Endpoints
Quantitative and/or qualitative measures that can be assessed in a clinical trial based on the symptoms of a disorder like communication, sleep, behaviors, motor function, etc.

Enzyme Replacement Therapy (ERT)
A therapeutic approach replacing the missing or nonfunctional UBE3A protein in the brain.

Food & Drug Administration (FDA)
A government agency in the USA that is responsible for protecting the public's health by ensuring the safety, efficacy, and security of human and veterinary drugs, biological products (like cell and gene therapies), medical devices, food supply, cosmetics, etc.

Gamma-Aminobutyric Acid (GABA)
An inhibitory neurotransmitter in the brain that plays a crucial role in regulating brain activity. GABA's primary function is to inhibit or reduce the excessive firing of neurons, which can lead to overstimulation and conditions like anxiety, stress, and even seizures.

Gene
A segment of DNA that provides the instruction, or code, for a specific protein. Typically, each individual has two copies of each gene, one from the mother and one from the father. Missing genes or differences within the gene that change the code can then result in absent protein, not enough protein, excessive toxic protein, or a protein that is nonfunctional.

Gene/RNA Editing
A technology that enables changing or editing parts of the genome, either the DNA or the RNA. This can be used to create tools that mimic genetic disorders (for example, to make an animal or cell model) or potentially to correct certain genetic disorders. Gene editing results in either the removal of existing DNA or RNA or the insertion or replacement of DNA or RNA.

Gene Therapy (GT)
A therapeutic approach that involves introducing, modifying or replacing specific genes within target cells with the goal of correcting or alleviating a condition caused by missing or nonfunctional genes.

Genome
A complete set of an organism's genetic material, including all its genes. This serves as a genetic blueprint or instruction manual for the growth, development, functioning, and reproduction of that organism. The genome is encoded in the DNA (deoxyribonucleic acid) of an organism's cells.

Genotype
The specific genetic makeup or combination of genes that an individual organism possesses. AS has 5 genotypes: Deletion, Mutation, UPD, ICD, and Mosaic.

Global Angelman Syndrome Registry (GASR)
A registry that assists families, researchers, clinicians, and pharmaceutical companies in understanding the scope of AS based on the data contributed by those who know the individuals living with AS best: caregivers.

Hematopoietic Stem Cell – Gene Therapy (HSC-GT)
A therapeutic approach where an individual's own bone marrow stem cells are removed from their body, modified ex vivo, or outside the body, and returned to the body with a replaced copy of the missing or nonfunctional gene (*UBE3A* in this case). Once the bone marrow cells are injected back into the individual, they go to the bone marrow to grow and repopulate. The goal is for those cells to continuously supply the body with a healthy version of the gene. These cells can cross from the blood to the brain, which is called crossing the blood brain barrier (BBB). Once they cross the blood brain barrier they become a cell type called microglia and secrete the UBE3A protein throughout the brain for neurons to take up the protein and use it.

Imprinting
The process by which only one copy of a gene in an individual is turned on, based on whether the gene was inherited from the mother or the father. The copy from the other parent is turned off or "silenced."

Imprinting Center Defect (ICD)
A genotype of AS that is caused by an error in imprinting, either as a result of a random event at the imprinting center or because of an imprinting center deletion. This results in silencing of both the maternal and the paternal copies of *UBE3A* and therefore lack of expression of the *UBE3A* gene or protein.

Induced Pluripotent Stem Cells (iPSCs)
iPSCs are derived from mature cells in the body, like skin or blood, and can be reprogrammed back into very young cells enabling them to grow into determined cell lines, like neurons. This allows them to be used as tools for testing and research purposes.

International Angelman Syndrome Research Council (INSYNC)
A council that brings together world experts in and outside of the Angelman syndrome ecosystem to help support advancing AS drug development, ensuring all research avenues are identified, de-risking novel therapeutic platforms, and encouraging collaborative efforts within the field of neurogenetic disorders.

Investigational New Drug-Enabling (IND-enabling)
Research experiments performed in animals and cells to study safety, toxicology, pharmacology, and drug metabolism of a potential therapy. These studies are required by the regulatory organizations, like the FDA, to help define the properties, define the dose, and reduce risks of potential therapies before it is provided to humans in a clinical trial.

Maternal
Genetic traits, variants or chromosomes inherited from the mother. UBE3A is only expressed from the maternal copy of the 15th chromosome in neurons and the paternal copy is silenced due to the *UBE3A-ATS*.

Methylation
Methylation is a chemical modification of DNA that can affect gene expression. Methylation testing is a common type of diagnostic testing in AS used to determine if *UBE3A* is abnormally methylated. Methylation testing can detect deletion, UPD, and ICD genotypes. The mutation genotype is not identified on methylation testing.

Mouse Model (AS Mouse)
The mouse is the foremost mammalian model for studying human disease. Several mouse models of AS exist, which have been able to recapitulate many of the symptoms like balance disorders, anxiety, learning and memory challenges, motor dysfunction, increased seizure susceptibility, and an abnormal EEG.

Natural History Study (NHS)
An observational clinical study that aims to conduct a prospective, longitudinal evaluation of children and adults living with Angelman syndrome using investigator-observed and parent-reported outcome measures, or endpoints, to obtain data that will be useful for future clinical trials. Understanding how individuals perform on these measures without a potential therapy can help to guide if that trajectory of development can change after a therapy is given.

Neurodevelopmental Disorders (NDD)
A group of conditions that are associated with differences in brain development. This can impact language, emotions, behavior, learning, memory, motor function, and more.

Novel
Something that is new and unique to the research field, for example, a delivery method or therapy.

Observational Clinical Study
A type of clinical study in which participants are identified, observed, and assessed for biomedical or health outcomes. Usually there is no drug or intervention in this type of study (e.g. Natural History Study).

Observer-Reported Communication Ability Measure (ORCA)
A caregiver reported outcome measure that separates communication into three main concepts: expressive, receptive and pragmatic communication. This measure was developed by FAST in collaboration with Duke University for AS specifically and is now being advanced for 14 other rare NDDs. The ORCA is now being assessed in all active clinical trials for AS and is the first validated endpoint specially developed for the AS population.

Organoids
Three-dimensional tissue cultures that are derived from stem cells. Organoids are self-organized cultures that can be crafted to replicate much of the complexity of an organ to characterize and test various therapeutic modalities. For AS, brain cortical organoids have been developed for every genotype.

Outcome Measures
A measure, or test, to determine if a potential treatment or intervention has an effect. Typically, this assessment is collected before a treatment/intervention for baseline results, then re-administered after the treatment/intervention to measure for changes due to intervention.

Paternal
Genetic traits, variants or chromosomes inherited from the father. *UBE3A* is only expressed from the maternal copy in neurons and the paternal copy is silenced due to the *UBE3A-ATS*.

Phenotype
An individual's observable characteristics. In AS this can be their ability to walk, talk, sleep, have seizures, etc.

Pillar 1
Pillar of FAST's strategic roadmap which focuses on replacing the missing or non-functional maternal copy of the *UBE3A* gene or protein in neurons of the brain. This includes therapeutic platforms like AAV-GT, HSC-GT, ERT, etc.

Pillar 2
Pillar of FAST's strategic roadmap which focuses on activating the silent copy of the paternal *UBE3A* gene in the brain. This includes therapeutic approaches like ASOs, CRISPR, ATF-ZF, miRNA, etc.

Pillar 3
Pillar of FAST's strategic roadmap which focuses on different molecular pathways and proteins impacted by the missing UBE3A protein. These drugs generally aim to improve the communication of neurons at the synapse (junction between the two neurons) and are often referred to as downstream targets.

Pillar 4
Pillar of FAST's strategic roadmap which focuses on work supporting necessary research tools, clinical developments, and community efforts to prepare for AS clinical trials and drug approvals. This includes the development of clinical trial training centers, newborn screening efforts, advancing endpoints and biomarkers (A-BOM), and driving policy and visibility globally.

Pre-Clinical
Refers to any research investigating a potential therapeutic approach prior to clinical assessment in humans.

Rescue
Refers to a technique or experimental approach aimed at restoring or improving a specific biological function or phenotype that is disrupted in a genetic or disease model.

Ribonucleic Acid (RNA)
The molecule that translates genetic information from DNA into proteins. Unlike DNA, RNA is usually single-stranded and helps carry instructions from the genes to the cell's machinery that makes proteins. RNA is composed of four chemicals, abbreviated A, G, U, and C.

RNA interference (RNAi)
A natural cellular process that helps control gene expression. RNAi occurs when small RNA molecules inhibit or reduce the expression of a particular gene(s).

RNP (Ribonucleoprotein)
A complex, often associated with CRISPR, that consists of a nuclease protein and a guideRNA (gRNA) to taget DNA or RNA for gene or RNA editing. This is most commonly used for
non-viral genome editing.

Scientific Advisory Board (SAB)
A group of FAST volunteers made up of scientists and clinicians who review grants, advise on new scientific ideas, and support ongoing programs in academia and industry.

Small Molecule
A low molecular weight compound that is small enough to easily get into tissues, enter cells, and interact with specific biological targets. In AS, small molecules could be developed that function as drugs by modulating biochemical pathways, inhibiting or activating specific proteins, or altering cellular processes shown to be altered in AS neurons. These targets of small molecules can be proteins, DNA, RNA or the ATS.

Synapse
A neuronal junction, which is the site of electric nerve impulse communication between two neurons or between a neuron and a muscle cell. This junction is impacted in AS.

Translational Research
The process of turning observations in the laboratory, clinic, and community into interventions that improve the health of individuals.

Upregulate
The process of increasing the expression or activity of a gene or protein.

Vector
A delivery system or carrier used to transport therapeutic agents, such as gene therapies, vaccines, or other medical treatments, to their intended target within a patient's body. Terms you might hear are Adeno-associated Virus Vector or Lentiviral Vector.

Wildtype (WT)
When animal models are designed, the WT genotype refers to an animal without any mutated genes. The phenotype of a WT mouse is considered to be "typical" functioning and can be used as a comparison group for animals with the mutated gene, or the AS model.

HOPE IN ACTION

PROGRESS THROUGH ADVOCACY

angelman SYNDROME FOUNDATION fast

Calling all AS Families!

The Foundation for Angelman Syndrome Therapeutics (FAST) and the Angelman Syndrome Foundation (ASF) have formed a strategic partnership to drive a coordinated, multi-tiered advocacy strategy to improve the lives of those affected by Angelman syndrome.

Both organizations understand the need to ensure key decision-makers understand Angelman syndrome to influence and inform policy, legislation, and funding related to AS. Without YOU—we cannot accomplish our goals.

We need your voice to to ensure those who make decisions on behalf of you and your loved ones – make informed decisions.

Upcoming Events

Jan 29, 2025

Externally-led Patient-Focused Drug Development (El-PFDD) Meeting with FDA

Be sure to complete the survey and register to attend virtually at the link below.

8 am - 1 pm ET

Virtual

Mar 4-5, 2025

2nd Annual Angelman Syndrome Congressional Advocacy Day:

Training (March 4, Marriot Capitol Hill) and Day on the Hill (March 5)

Washington, DC

Visit **AngelmanAdvocates.org** to learn more.

Going beyond every day.™

Ultragenyx is proud to sponsor the 2024 FAST Global Science Summit & Gala and support FAST's ongoing work with the Angelman community.

Together, we will continue to Dream Big and transform the lives of people living with Angelman syndrome.

To learn more visit **Ultragenyx.com**

ultragenyx

GPRC-GTX102-00020 September 2024

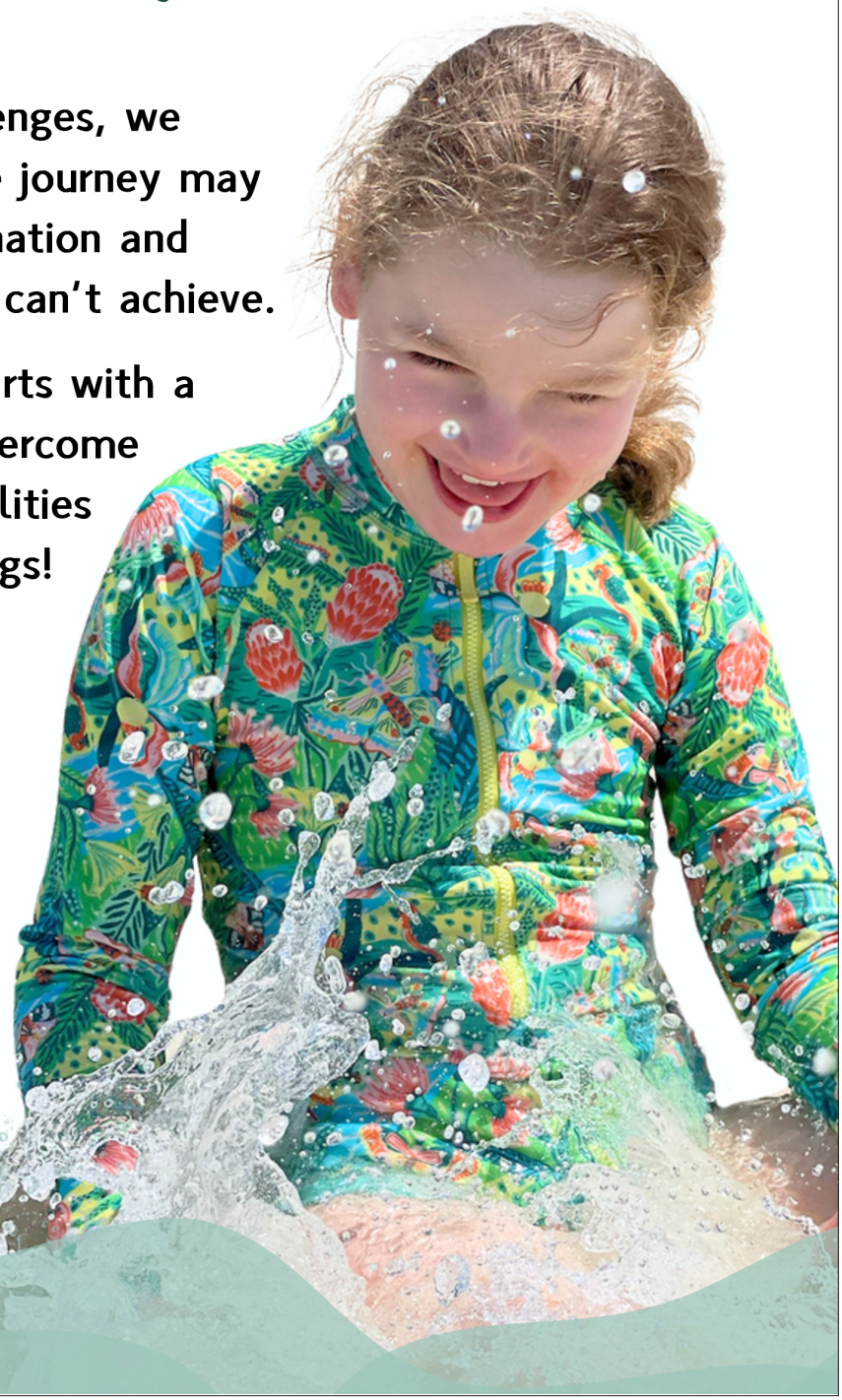
DREAM BIG

hope together

In a world filled with challenges, we believe in dreaming big. The journey may be tough, but with determination and support, there's nothing we can't achieve.

Every great achievement starts with a dream- together, we can overcome obstacles and create possibilities beyond our wildest imaginings!

Join us in embracing hope, supporting FAST, and *dreaming big!*



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#CUREANGELMAN

DREAM BIG

MAGAZINE

Quincy and Elijah : Love at First Sight - Page 3

Quincy's Quest Foundation
a mission to cure Angelman Syndrome

"they have so much TO SAY"

- Allyson Berent, Quincy's mom

CAN'T STOP WON'T STOP

Vivi and Sydney: Who Wore it Best?

NOVEMBER 2024

Notes

Notes

A Special Thank You

To all of our partners, thank you for your generosity and commitment to advancing therapeutics for our loved ones living with Angelman syndrome.

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