

6TH INTERNATIONAL



CONFERENCE

PROGRAM AGENDA

May 31, 2025
Baltimore, MD, USA



Venue: Doubletree Baltimore by Hilton Baltimore, BWI Airport
890 Elkrige Landing Road Linthicum, Maryland, 21090, USA



MEET THE SPEAKERS



**Ms. Amy Bergeson
and Ms. Diane Callari
AAC Specialists**



**Ms. Leigh Broughan
Board Certified
Behavior Analyst**



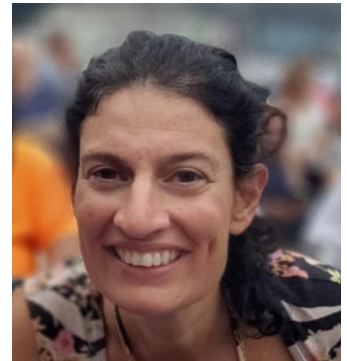
Ms. Kristen Carpenter



**Ms. Sue Carpenter
Advocacy Team Member
KAT6 Foundation**



**Dr. Sarah Donoghue
Metabolic Physician
and Pediatrician**



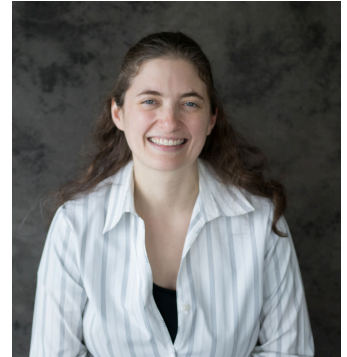
**Dr. Natacha Esber
Director of Science and
Research, KAT6 Foundation**



**Dr. Jill Fahrner
Molecular and Epigenetics
Researcher**



**Ms. Zoe Goldstone-Joubert
Genetic Counseling Student**



**Dr. Jacqueline Harris
Pediatric Neurologist**



**Ms. Susan Hartung
Advocacy Director
KAT6 Foundation**



**Dr. Richard Kelley
Pediatrician and
Biochemical Geneticist**



**Ms. Lusía Langi
Content Creator and
NCAA athlete**

MEET THE SPEAKERS



Dr. Paul Marcogliese
Biochemistry and Medical
Genetics Researcher



Dr. Jordan Muller
Chairperson
KAT6 Foundation



Mr. Emile Najm
CEO
KAT6 Foundation



Ms. Aileen Nava
Doctoral Student
Human Genetics



Dr. Rowena Ng
Pediatric
Neuropsychologist



Dr. Andrew Rankin
Board Member
KAT6 Foundation



Ms. Aimee Reitzen
Director of Marketing &
Communications, KAT6 Foundation



Dr. Angie Serrano
Assistant Professor, BU
Researcher



Ms. Paulina Varella Castillo
Doctoral Student
Experimental Medicine



Ms. Patricia Wilson
Speech Therapist
(PROMPT)



Ms. Beth Woodbury
Advocacy Team Member
KAT6 Foundation



Ms. Amy Young
Advocay Team Member
KAT6 Foundation

MORNING SESSION (7:30 AM - 12:00 PM)

All presentations to be followed by 5 minutes of Q&A

REGISTRATION AND BREAKFAST (07:30 - 08:00 AM)

Speakers	Title
Jordan Muller, KAT6 Foundation	Conference moderator
Natacha Esber KAT6 Foundation	Welcome note 8:00 - 8:05 AM
Rowena Ng, and Jacqueline Harris Kennedy Krieger Institute & Johns Hopkins Medicine, Baltimore, USA	TBA 8:05 - 8:35 AM
Paul Marcogliese and Zoe Goldstone- Joubert Rady College of Medicine, University of Manitoba, Canada	Understanding the spectrum of KAT6A/B variants using fruit flies 8:40 - 9:10 AM
Aileen Nava Arboleda lab, David Geffen School of Medicine, University of California, Los Angeles, USA	TBA 9:15 - 9:40 AM
Serrano Lab Center for Regenerative Medicine, Boston University, USA	TBA 9:45 - 10:15 AM

BREAK (10 minutes)

Sarah Donoghue Victorian Clinical Genetics Services & Murdoch Children's Research Institute, Melbourne Australia	Cellular mechanisms that cause intellectual disability and how this applies to KAT6A 10:25 - 10:50 AM
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MORNING SESSION (8:00 AM - 12:00 PM)

All presentations to be followed by 5 minutes of Q&A

Speakers	Title
Paulina Varela Castillo Rosalind and Morris Goodman Cancer Institute McGill University, Montreal, Canada	A highly efficient mutagenesis method to engineer KAT6 and other gene variants for functional analyses 10:55 - 11:20 AM
Jill Fahrner Kennedy Krieger Institute & Johns Hopkins Medicine, Baltimore, USA	Mendelian disorders of the epigenetic machinery 11:25 - 11:40 AM
Jordan Muller KAT6 Foundation	Charting new paths: Funded research initiatives for 2025 and beyond 11:45 - 11:55 AM
Aimee Reitzen, Jordan Muller KAT6 Foundation	Fundraising for KAT6 Foundation 11:55 AM - 12:05 PM
Emile Najm KAT6 Foundation	KAT6A/B Patient Registry update 12:05 -12:15 PM

GROUP PHOTO (12:15 PM - 12:30 PM)

LUNCH (12:30 PM - 1:30 PM) Provided by KAT6 Foundation

AFTERNOON SESSION (1:30 PM - 2:00 PM)

All presentations to be followed by 5 minutes Q&A

Shared Journeys: Stories from Families, Siblings, and Individuals with KAT6A/B Variations 1) Ms. Lusia Langi 2) Ms. Kristen Carpenter	1) Sibling Bonds (1:30 - 1:45 PM) 2) Resiliency in a life of Unknowns (1:45 - 2:00 PM)
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WORKSHOPS FOR FAMILIES (2:00 PM - 4:00 PM)

Workshop duration - 20 - 25 minutes

Workshop A - Speech therapy “Freedoms of Speech”	Ms. Patricia Wilson, PROMPT Therapist
Workshop B - KAT6 Foundation Advocacy Update	<p>Ms. Susan Hartung, Ms. Sue Carpenter, Ms. Beth Woodbury, and Ms. Amy Young KAT6 Foundation Advocacy committee</p> <p>Members of the Advocacy Committee will share a personal story on how they have effectively advocated at the school, medical and legislative level and share their tips and suggestions on how to be an effective advocate and open the floor to questions.</p>
Workshop C - AAC Strategies to Support Speech and Language Development	<p>Ms. Amy Bergeson, Speech language pathologist and KAT6 Foundation Member</p> <p>Ms. Diane Callari, AAC Specialist</p>
Workshop D - Applied Behavior Analysis (ABA)	Ms. Leigh Broughan, Board certified Behavior Analyst and KAT6 Foundation, Member

RESEARCH WORKSHOP (2:00 PM - 4:00 PM)

Closed Scientific Meeting	<p>Moderator: Dr. Andrew Rankin, Scientific committee, KAT6 Foundation</p> <p>This closed meeting brings together researchers working on KAT6A and KAT6B gene variations. The session will facilitate in-depth scientific discussions, and collaboration among experts in the field. Participation is limited to invited researchers actively engaged in KAT6A/KAT6B-related research.</p>
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RAFFLE PRIZES AND THANK YOU NOTE

Jordan Muller KAT6 Foundation	4:00 - 4:10 PM
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KAT6 Foundation: Driving Research for a Better Future

Participate in Blood Collection & Advance KAT6 Research

Exciting News from the KAT6 Foundation!

For the first time, the KAT6 Foundation is sponsoring blood sample collection to support KAT6A/B patient registry. Thank you for your generous donation!

What's being collected?

The blood samples will be analyzed for a range of key metabolic and biochemical markers, including:

- Complete Blood Count (CBC)
- Comprehensive Metabolic Panel
- Lipid Panel
- Total Homocysteine Levels
- Plasma Amino Acids
- Carnitine Levels
- Acylcarnitine



How will the data be used?

All collected data will be securely stored in the KAT6A/B Registry, contributing to a growing resource that empowers researchers to uncover new connections between KAT6 mutations and health outcomes.

What's in it for families?

Families will receive a copy of their child's report at no cost. This provides valuable clinical information and helps empower families with actionable insights, all while advancing scientific discovery.

To join the KAT6A/B patient registry, click [here](#).
To participate in blood sample collection click [here](#).

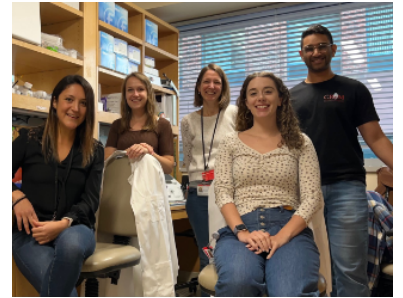
Note: Blood sample collection is only available for those who have joined the KAT6A/B patient registry.

KAT6 Foundation: Driving Research for a Better Future

Join the Effort: Research Labs Need Your Contribution to KAT6A/B Studies

Project 1 - The **Serrano Lab** at the **Center for Regenerative Medicine (CRoM)**, in collaboration with the KAT6 Foundation, is working to establish the first KAT6A and KAT6B patient-derived iPSC bank. This important initiative will create a repository of KAT6A and KAT6B human iPSC lines, which will be shared with both internal and external investigators for basic and translational research.

To sign up for iPSC biobank, click [here](#)



Project 2 - The **Arboleda lab** at **UCLA** are inviting individuals with rare genetic disorders and their families to participate in a research study funded by the NIH and California Institute for Regenerative Medicine. The study will involve collecting 3 vials of blood to identify disease-associated biomarkers for diagnosis and future therapies. Participants of all ages are welcome, and parents and siblings are encouraged to participate as control samples.

Your participation is crucial to advancing this research, including specialized testing of DNA epigenomes and blood cells to identify therapies based on their promising findings in a mouse model.

To contribute to Dr. Arboleda's research, click [here](#)



Important: You only need to provide a blood sample once, but you will need to sign up separately to consent to how the data will be stored and shared.

KAT6 Foundation: Driving Research for a Better Future

Join the Effort: Research Labs Need Your Contribution to KAT6A/B Studies

Project 3 - Background - The **Chromatin Disorders Research** team at the **Murdoch Children's Research Institute, Australia**, is currently using a mouse model, alongside human cortical neurons to study gene expression and metabolomics in KAT6A syndrome, in collaboration with **Professor Anne Voss** at the WEHI. This work is being led by PhD student **Dr Sarah Donoghue** and supervised by **Professor David Amor** and **Professor Paul Lockhart**. The goal of this project is to understand the differences in brain development that occur in KAT6A syndrome, and to identify biomarkers that may show response to treatment in clinical trials.

Rationale - This project aims to extend the work on blood biomarkers in Kat6a mice to children and adults with KAT6A syndrome. In this project, a range of molecular compounds will be measured in blood samples collected from human participants with KAT6A syndrome, using untargeted metabolomic and proteomic analyses. The plasma profiles of 50 participants with KAT6A syndrome will be compared to those of 20 participants without KAT6A syndrome.

Project aim - To identify biomarkers that are detectable in the plasma of participants with KAT6A syndrome, with the hope that these can be translated for use in clinical trials, as an objective measure of treatment efficacy as the community proceeds to clinical trials.

For participate in this research, click [here](#)

Important: You only need to provide a blood sample once, but you will need to sign up separately to consent to how the data will be stored and shared.

Consultation with Dr. Richard I. Kelley



Meet Dr. Richard I. Kelley—a brilliant pediatrician, clinical geneticist, and a longtime champion of the KAT6 Foundation!

Former director of the Kennedy Krieger Institute's Clinical Mass Spectrometry lab, Dr. Kelley is renowned for unraveling the biochemical mysteries behind genetic disorders. And guess what? He's back this year, bringing his expertise and warmth to families like yours!

Here is the best part: Dr. Kelley is dedicating an entire day to meet with families in small, personalized sessions. This is your chance to ask questions, get insights, and receive one-on-one guidance from a true expert in the field.

Spots are limited, so don't wait—secure your appointment now!

Book in advance to guarantee your family's time with Dr. Kelley. It's an opportunity you won't want to miss!

Scan the QR code to sign up or click [here](#)



Science with a Twist: Fun Activities at the KAT6 Conference!

Music Therapy

The KAT6 Foundation invites children to a special musical experience with a talented music therapist, **Ms. Amy Gardiner!**

This interactive session will encourage kids to engage in music through rhythm, melody, and song, creating a fun and joyful atmosphere. Don't miss this wonderful opportunity to explore the power of music and connection!

Date: May 31, 2025

Time: 10:00 AM - 11:00 AM



Spike Your Skills: Volleyball Tips & Tricks with a Pro

We are thrilled to welcome the incredible

Ms. Lusia Langi to our conference!

A standout volleyball player and former high school star, she is sure to inspire! With an impressive high school career at Hays High School in Kyle, Texas, including numerous awards and records, she is now making waves as a sports management and media studies major at Utah Tech. A three-year letter winner, team captain, and AVCA All-American selection, she holds multiple records at her high school, including the all-time career blocks record.

Join us for a fun and interactive session where kids can learn volleyball tips and tricks from a star player!



@lusialangi

Date: May 31, 2025

Time: 11:00 AM - 12:00 PM

Science with a Twist: Fun Activities at the KAT6 Conference!

Meet Our Therapy Dog - Pets On Wheels

The KAT6 Foundation invites you to enjoy "Fur Time"—a pawsome chance to relax, cuddle, and connect with our friendly therapy dog! This furry friend brings comfort, joy, and plenty of tail wags, making every moment a little brighter. Don't miss out on the cuddles!

Date: May 31, 2025
Time: 11:00 AM - 12:00 PM



Let's Science Together! Exploring Tiny Worlds with Foldscopes

Join Serrano Lab for a fun, hands-on science adventure!

We'll use Foldscopes (paper microscopes) to explore tiny, fascinating worlds hidden in everyday samples. Children, parents, and family members are all welcome to discover, observe, and learn together. Best of all, each family can take their Foldscope home, continuing their journey of scientific discovery!

Duration: Approximately 40 minutes

Who can participate? All ages and family members welcome (no previous science experience needed!)

To sign up: Click [here](#)

Date: May 31, 2025
Time: 4:20 PM - 5:00 PM



SOCIAL ACTIVITIES

TGIF - Bowling & Dinner

Join us for bowling and dinner!
It's a great way to meet other KAT6 families and spend a fun afternoon and evening together.

Date - May 30, 2025

Bowling @ 3:30 PM - 5:30 PM

Venue - Glen Burnie Bowl
6322 Ritchie Hwy, Glen Burnie, MD 21061
Phone no. - 443-312-8538
To sign up for bowling, click [here](#)

Dinner - 6:00 PM - 8:00 onwards

Venue - Guinness Open Gate Brewery
5001 Washington Blvd, Halethorpe
MD 21227
Phone no. - 443-575-6893
Outdoor seating—come prepared for the weather!
To sign up for dinner, click [here](#)

Note: You will be responsible for paying for your bowling and/or dinner.



SOCIAL ACTIVITIES

Saturday: Pizza by the Pool

Continue the fun by socializing at the pool.
Pizzas will be provided by the KAT6 Foundation.

Date - May 31, 2025

Time - 5:00 PM - 8:00 PM onwards

Location - Doubletree Baltimore BWI Airport



SunnySide Start Breakfast



Start your day with us before everyone heads back home.

Date - June 01, 2025

Time - 7:00 AM - 9:00 AM

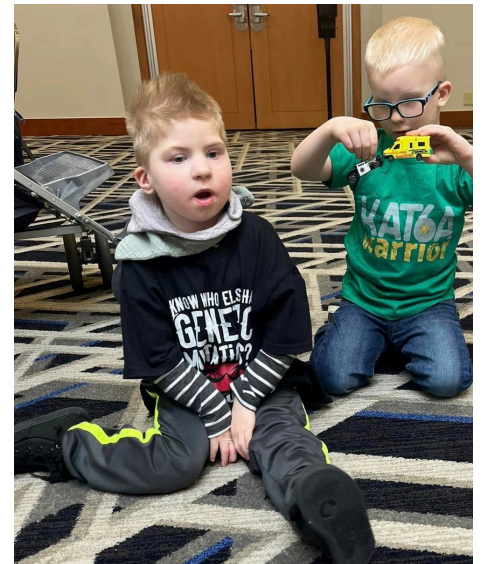
Location - Grand Ballroom, Doubletree Baltimore BWI Airport

Sponsored by: KAT6 Foundation

Childcare Services

Childcare assistance will be available from
9:00 AM - 1:00 PM on May 31st.

This service aims to support attendees by providing a safe and nurturing environment for children, allowing parents and caregivers to fully participate in the event without worry.



IMPORTANT SIGN UP INFORMATION

Activities	Sign up link	Date and Venue
Meeting Registration	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
Consultation with Dr. Richard Kelley	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
Join KAT6A and KAT6B Patient Registry	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
Participate in Blood Sample Collection and Advance KAT6 Research*	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
Join the iPSC Bank (Blood sample collection)*	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
Sign up for Dr. Arboleda's (UCLA) research (Blood sample collection)*	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
Sign up for Dr. Sarah Donoghue (MCRI) research (Blood sample collection)*	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
Let's Science Together! Exploring Tiny Worlds with Foldscopes	Click here	May 31, 2025 Doubletree Baltimore By Hilton Baltimore - BWI Airport
TGIF Bowling (03:30 - 05:30 PM)	Click here	May 30, 2025 Glen Burnie Bowl 6322 Ritchie Hwy, Glen Burnie, MD 21061
TGIF Dinner	Click here	May 30, 2025 Guinness Open Gate Brewery 5001 Washington Blvd, Halethorpe, MD 21227

*Participants will only need to provide **one** blood sample but must enroll in each study separately to provide consent for how their data will be stored and shared.

RAFFLES & MERCHANDISE

Handmade Quilts for Raffle winners!

Two lucky winners will take home beautifully handcrafted quilts (50x60 in), lovingly made by Susan Carpenter, a talented member of the KAT6 Foundation.

Plus, a variety of exclusive prizes will also be up for grabs!

Each attendee will receive one free raffle ticket, with additional tickets available for purchase at **\$5** each. Don't miss your chance to win!



KAT6 Merchandise for Sale

Conference Tshirts and car magnets will be available for purchase at the conference during check in (7:30 AM - 9:00 AM). Quantities limited.

Cash, check, venmo and credit card will be accepted.



\$5



\$20

ACKNOWLEDGEMENTS

We'd like to thank the following individuals, families and sponsors for making this conference possible.

- Families and volunteers of KAT6 Foundation
- Scientist members of KAT6 Foundation
- Ms. Susan Hartung for co- sponsoring Ms. Amy Gardiner, music therapist
- Bay Tower's family for providing babysitting services
- Melanie Maas for organizing networking and fun activities for families attending the conference
- Susan Carpenter for donating two handmade quilts
- Jeff Worden for video documenting the event
- Pets on Wheels for providing a fur friend!

**SCAN THE QR CODE TO
REGISTER FOR THE CONFERENCE**

Advance registration is mandatory.



This is a FREE event. A courtesy shuttle will be provided by Doubletree hotel to and from the airport.

Free Parking included.