



May 31, 2025 | Baltimore, MD, USA

# 6th International KAT6A and KAT6B Conference

Venue: Doubletree Baltimore by Hilton Baltimore, BWI Airport

890 Elkridge Landing Road Linthicum, Maryland, 21090, USA

## Preliminary Program Agenda

### MEET THE SPEAKERS



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



**Dr. Jacqueline Harris**  
Pediatric Neurologist



**Dr. Rowena Ng**  
Pediatric  
Neuropsychologist



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



**Dr. Meghna Singh**  
Postdoctoral Scholar



**Dr. Paul Marcogliese**  
Biochemistry and Medical  
Genetics Researcher



# MEET THE SPEAKERS



**Dr. Xiang-Jiao Yang**  
Molecular and Epigenetics  
Researcher



**Serrano Lab| CReM| BU**



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



**Ms. Kristen Carpenter**  
Member, KAT6 Foundation



**Ms. Lusia Langi**  
Content creator  
(Volleyball) and Member,  
KAT6 Foundation



**Ms. Patricia Wilson**  
Speech Therapist  
(PROMPT)



**Ms. Leigh Broughan**  
Board certified Behavior Analyst  
and Member, KAT6 Foundation



**Ms. Beth Woodbury**  
Member, KAT6 Foundation



**Ms. Amy Young**  
Member, KAT6 Foundation

# MEET THE SPEAKERS



**Ms. Aimee Reitzen**  
Member, KAT6 Foundation



**Ms. Karen Ginsburg**  
Member, KAT6 Foundation



**Ms. Susan Hartung**  
Member, KAT6 Foundation



**Ms. Sue Carpenter**  
Member, KAT6 Foundation



**Mr. Emile Najm**  
CEO, KAT6 Foundation



**Dr. Andrew Rankin**  
Member, KAT6 Foundation



**Ms. Amy Bergeson and  
Ms. Diane Callari**  
AAC Specialists and  
Members, KAT6  
Foundation



**Jill Fahrner**  
Molecular and  
Epigenetics Researcher



## Morning Session (08:00 AM - 12:00 PM)

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

Speakers	Title
Jordan Muller, KAT6 Foundation	Conference moderator
Natacha Esber KAT6 Foundation	Welcome note 08:00 - 08:05 AM
Rowena Ng, and Jacqueline Harris Kennedy Krieger Institute & John Hopkins Medicine, Baltimore, USA	TBA 08:05 - 08:35 AM
Paul Marcogliese and Zoe Goldstone- Joubert Rady College of Medicine, University of Mannitoba, Canada	Understanding the spectrum of KAT6A/B variants using fruit flies 08:40 - 09:10 AM
Meghna Singh Arboleda lab. David Geffen School of Medicine, University of California, Los Angeles, USA	TBA 09:15 - 09:40 AM
Serrano lab Center for Regenerative Medicine, Boston University, USA	TBA 09:45 - 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 (08:00 AM - 12:00 PM)

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

Speakers	Title
Xiang-Jiao Yang McGill University, Montreal, Canada	TBA 10:55 - 11:20 AM
Jill Farhner Kennedy Krieger Institute & John Hopkins Medicine, Baltimore, USA	TBA 11:25 - 11:45 AM
Karen Ginsburg KAT6 Foundation	KAT6 Foundation Fundraising 11:45 - 11:55 AM
Aimee Reitzen KAT6 Foundation	KATwalk 2025 11:55 AM - 12:05 PM
Emile Najm KAT6 Foundation	KAT6A/B Registry update 12:05 -12:15 PM

## Group Photo (12:15 PM - 12:30 PM)

Lunch break (12:30 PM - 1:30 PM)

## Afternoon session (01:30 PM - 05:00 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) TBA 2) Resiliency in a life of Unknowns 01:30 - 2:00 PM
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# Workshops for Families (02:00 PM - 04:00 PM)

*Workshop duration - 20 - 25 minutes*

<b>Workshop A- Speech therapy “Freedoms of Speech”</b>	Ms. Patricia Wilson, PROMPT Therapist
<b>Workshop B - KAT6 Foundation Advocacy Update</b>	<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>
<b>Workshop C - AAC Strategies to Support Speech and Language Development</b>	<p>Ms. Amy Bergeson, Speech language pathologist and KAT6 Foundation Member</p> <p>Ms. Diane Callari, AAC Specialist</p>
<b>Workshop D - Applied Behaviour Analysis (ABA)</b>	Ms. Leigh Broughan, Board certified Behavior Analyst and KAT6 Foundation, Member



# Consultation with Dr. Richard Kelley



**Meet Dr. Richard I. Kelley**—a brilliant paediatrician, 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](#)





## Research Workshop (02:00 PM - 04:00 PM)

Closed scientific meeting

Moderator: Dr. Andrew Rankin, Scientific committee, KAT6 Foundation

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,

## Raffle Prizes and Thank You Note

Jordan Muller  
KAT6 Foundation

04:00 - 04:10 PM



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

## Music Therapist

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 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 AM - 12 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 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 - 05:00 PM





# 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 \$10 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. Quantities limited. Cash, check, venmo and credit card will be accepted.



\$5



\$20

# 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 more information about this research, please contact Sarah Donoghue at [sarah.donoghue@mcri.edu.au](mailto:sarah.donoghue@mcri.edu.au).**

**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.

## TGIF - Bowling & Dinner



Join us for bowling and dinner! It's a great way to meet other KAT6 families and spend a fun 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 - 08:00 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: Buy your own ticket and dine event*

## Saturday: Pizza by the pool



Dive into relaxation at the pool and savor some pizza fun!

**Date - May 31, 2025**

Time - 5:00 - 08:00 PM onwards

Location - Doubletree Baltimore BWI  
Airport

Sponsored by: KAT6 Foundation

## SunnySide Start Breakfast



Start your day with us before everyone heads back home.

**Date - June 01, 2025**

Time - 07:00 AM - 09:00 AM

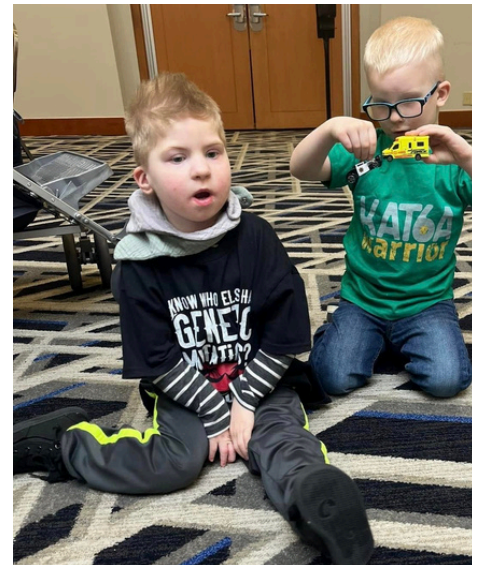
Location - Grand Ballroom, Doubletree Baltimore BWI Airport

Sponsored by: KAT6 Foundation



# Childcare Services

Childcare assistance will be available from **9:00 AM-01:00 PM** on the conference day. 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 ACTIVITIES AND SIGN UP INFORMATION

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

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# ACKNOWLEDGEMENTS

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

- Families of the KAT6 Foundation
- Scientist members of the KAT6 Foundation
- Ms. Susan Hartung for co- sponsoring Ms. Amy Gardiner, music therapist
- Bay Tower 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
- Pets on Wheels for providing a fur friend!

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

Advance registration is mandatory.

