



June 08, 2024 | Baltimore, MD, USA

5th 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



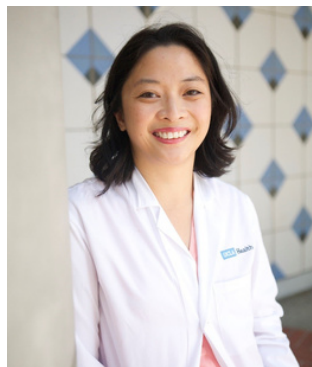
Dr. Jacqueline Harris
Pediatric Neurologist



Dr. Rowena Ng
Pediatric
Neuropsychologist



Dr. Jose A Sánchez-Alcázar
Molecular Biologist & Medical
Genetics Researcher



Dr. Valerie Arboleda
Molecular Biologist and
Medical Genetics Researcher



Dr. Paul Marcogliese
Biochemistry and Medical
Genetics Researcher



MEET THE SPEAKERS



Alejandra Laguillo-Diego
Postdoctoral Researcher,
Human Genetics



Dr. Angie Serrano
Molecular Biologist and
Medical Genetics Researcher



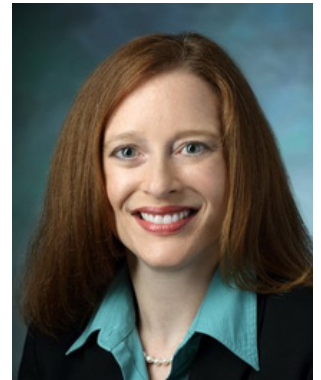
Dr. Bhawika Sharma L.
Postdoctoral Researcher
Neurology and
Neurological Sciences



Susan Hartung
KAT6 Foundation



Patricia Wilson
Speech Therapist
(PROMPT)



Jill Fahrner
Molecular and
Epigenetics Researcher



Katie Bator
KAT6 Foundation



Karen Ginsburg
KAT6 Foundation



Bethany and Ireland Harris
KAT6 Foundation

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, Jacqueline Harris and Jill Farhner Kennedy Krieger Institute & John Hopkins Medicine, Baltimore, USA	Overlapping Cognitive and Behavioral Features Across KAT6A and KAT6B Disorders 08:05 - 08:40 AM
Jose A Sánchez-Alcázar Sánchez-Alcázar's lab, Universidad Pablo de Olavide, de Sevilla, Spain	Precision Medicine for KAT6A and KAT6B Syndromes 08:40 - 09:10 AM
Paul Marcogliese Rady College of Medicine, University of Manitoba, Canada	Using Fruit Flies to Assess KAT6A/B Variants, Model Symptomology, and Screen Drugs. 09:10 - 09:40 AM
Angie Serrano Lab Center for Regenerative Medicine, Boston University, USA	TBA 09:40 - 10:15 AM

15 minutes break

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

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

Speakers	Title
Valerie Arboleda David Geffen School of Medicine, University of California, Los Angeles, USA	Unraveling the Molecular Mechanisms for KAT6A in Human Development and Disease 10:30 - 11:00 AM
Karen Ginsburg and Katie Bator KAT6 Foundation	KAT6 Foundation Fundraising 11:00 - 11:15 AM
Alejandra Laguillo-Diego Apostolou lab, Weill Cornell Medicine, New York, USA	Dissecting the Role of Mitotic Histone Acetylation and Acetyltransferases in Epigenetic Inheritance of Stem Cell Identity 11:15 - 11:45 AM
Richard Kelley Boston Children's Hospital, Boston, USA	The Metabolism of KAT6A and KAT6B: A Comparison 11:45 AM - 12:10 PM

Group Photo (12:10 - 12:20 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

Bhawika Sharma Lamicchane Neurology and Neurological Sciences Stanford University, California, USA	Lessons from the Patient Registry and Beyond 01:30 - 02:00 PM
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Afternoon session (01:30 PM - 05:00 PM)

Closed Scientific meeting (02:00 - 04:00 PM)

Workshops for Families (02:00 PM - 04:00 PM)

Workshop duration - 35 minutes

Workshop 1 - Expert consultation	Dr. Richard Kelley
Workshop 2 - Speech therapy "Freedoms of Speech"	Patricia Wilson, PROMPT Therapist See page 6 for a full description of this workshop.
Workshop 3 - Advocacy Update: "Important Legislation, Government Services, Long Term Planning, Siblings"	Susan Hartung, Amy Young, Sue Carpenter, and Beth Woodbury, KAT6 Foundation Join Susan and her team to learn about important Federal Legislation, the NORD State Report Card Tool for state health services, Self-Directed Services for families, medical and educational updates, advocacy success stories, and engage in a Q&A session with KAT6 families.
Bethany Harris and Ireland Harris	Horses, Homeschooling and the Human Genome: Meet Ireland and Beth Harris 04:00 - 04:10 PM
KAT6 Foundation (presentations and raffle time) **Raffle participants have the chance to win two handmade quilts: one adult-sized and one kid-sized. Crafted with care by Susan Carpenter, a member of the KAT6 Foundation. See page 8.	04:10 - 05:00 PM

WORKSHOP II

Title: Freedoms of Speech



Patricia Wilson
CCC-SLP, TSSLD
Otto Specht School

I. Communication

II. Acrobatics Analysis

- abilities/individual presentations
- potentials/therapeutic approaches (answers Why and How questions)

III. Juggling Programs

- brain plasticity
- coordination of systems for communication

IV. Lion Taming

- social emotional regulation
- activities to foster growth mindset (I am capable)

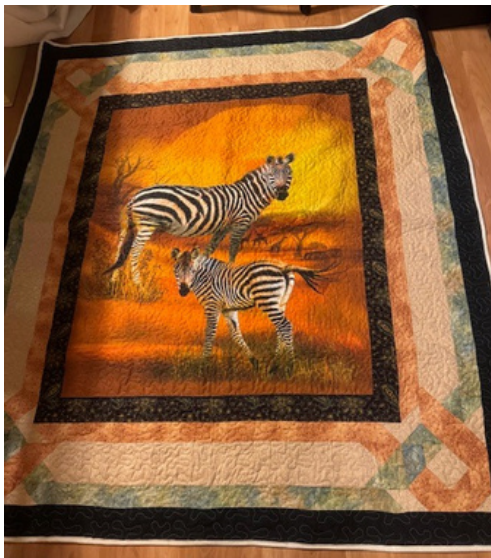
V. Magical Acts

- systems integration (sensory signs/communication developments that support brain changes)
- motor planning for speech (approach analysis: PROMPT, ASL and AAC)

Handmade Quilts for Raffle winners!

Two raffle winners will receive meticulously crafted quilts, one tailored for an adult and the other sized perfectly for a child, lovingly created by the skilled hands of Susan Carpenter, a member of the KAT6 Foundation. KAT6 Foundation merchandise will be included in the raffle.

Each attendee will receive one free raffle ticket with extra tickets available for purchase for \$10 per ticket.



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

Study 1: KAT6A and KAT6B Cognitive and Speech Profile for children **ages 5-18** with Drs. Harris and Ng.



Participants Needed: Study on KAT6 Disorders

What is this study about?

This research study aims to identify the unique cognitive profile of KAT6 disorders.

Why participate?

- Before clinical trials can be constructed to treat cognitive impairment in genetic diseases, scientists must first understand the cognitive phenotype of the disease in order to identify potential outcome measures, which has to be applicable across age and severity of cognitive impairment.
- Given our prior research findings, a new battery focused on nonverbal cognition will be tested to determine its sensitivity in measuring cognitive functioning in KAT6 disorders across different developmental periods and range of severity in intellectual impairment.

Who is eligible?

- Any individual between age 5 to 18 years who is attending the KAT6 Conference in Baltimore (June 8, 2024) or willing to be seen at the Kennedy Krieger Institute. A copy of the child's genetic test results is required to molecularly confirm the diagnosis.

What does it entail?

- Caregivers will be given an online research intake form and consent form. In addition, a copy of your child's genetic test results is needed to confirm the diagnosis.
- This study has three components:
 - 1.) In-person testing with your child: Your child will be given a battery of cognitive tests (e.g., paper and pencil or computerized puzzles and games) that will take about 90 minutes to complete. 2.) Your family will be contacted for a follow-up virtual Zoom session to complete a structured interview on your child's communication skills. 3.) Caregivers will be given a battery of questionnaires to complete remotely to index their child's day to day behavioral functioning.

Who do I contact to enroll in this study?

- If you would like to enroll in this study or have questions, please contact the principal investigator: Rowena Ng (ngr@kennedykrieger.org).
- *Please note, study enrollment and scheduling for this study at the conference are based on a first come first serve basis.*

PARTICIPATE IN RESEARCH

Study 2: KAT6A and KAT6B Cognitive and Speech Profile for children **ages 0-5** with Drs. Harris and Ng.



Baby and Toddler Participants Needed: Study on KAT6 Disorders

What is this study about?

This research study aims to examine early developmental profile and trajectory of KAT6 disorders.

Why participate?

- Before clinical trials can be constructed to treat cognitive impairment in genetic diseases, scientists must first understand the cognitive phenotype of the disease in order to identify potential outcome measures, which has to be applicable across age and severity of cognitive impairment.
- Currently, there are limited to no existing literature on early developmental profiles of individuals with KAT6 disorders, including developmental progress or milestones. As such, it is unclear if developmental domains (cognition, language, motor functioning) make gains at different pace, warranting focused domain-specific interventions in early childhood.

Who is eligible?

- Any individual under 5 years of age and is willing to be seen at the Kennedy Krieger Institute outside of the conference dates. We are happy to schedule interested families who are in town for the conference between June 3-6, June 10-14, or at a later date. A copy of the child's genetic test results is required to molecularly confirm the diagnosis.

What does it entail?

- Caregivers will be given an online research intake form and consent form. In addition, a copy of your child's genetic test results is needed to confirm the diagnosis.
- This study requires two in-person visits, one year apart from each other.
 - 1.) In-person testing with your child: Your child will be given a battery of tests to assess early cognition, language and motor skills that will take between 30-90 minutes to complete pending on their age. We will schedule the same testing a year later for your child so we can assess changes in these domains. Part of the in-person visits include a structured interview with parents which will take about another 30 minutes, totaling 60-120 minutes per visit. 2.) Caregivers will be given a battery of questionnaires to complete remotely to index their child's day to day behavioral functioning across both visits.

Who do I contact to enroll in this study?

- If you would like to enroll in this study or have questions, please contact the principal investigator: Rowena Ng (ngr@kennedykrieger.org).

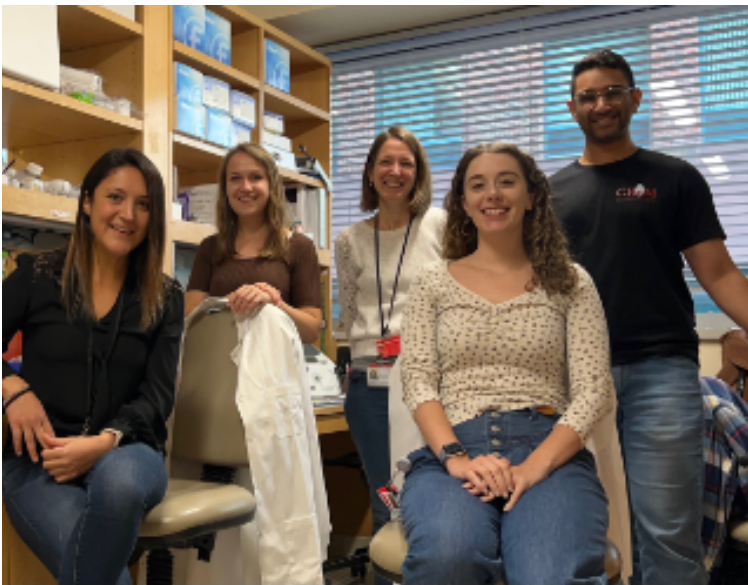
PARTICIPATE IN RESEARCH

Study 3: KAT6A and KAT6B Patient-Derived induced Pluripotent Stem Cell (iPSC) Bank: Capturing the Spectrum of KAT6 Pathogenic Variants

The Serrano Lab at the Center for Regenerative Medicine (CReM) is working together with the KAT6 Foundation to establish the first KAT6A and KAT6B patient-derived iPSC bank. The CReM bank serves as a repository for sharing KAT6A and KAT6B human iPSC lines with internal and external investigators to conduct both basic and translational research.

To participate in the iPSC bank, scan the QR code or click [here](#).

SATURDAY, JUNE 08, 2024



Study 4: Genomic Evaluation of Rare Diseases

Dr. Valerie Arboleda and UCLA researchers are seeking individuals with rare genetic disorders and their family members to participate in a research study funded by the National Institutes of Health and California Institute for Regenerative Medicine.

They will draw blood for laboratory studies of cells. Blood samples will be used to identify disease-associated biomarkers for diagnosis and for future assessment of any new therapies.

They will draw **three** vials of blood for this study, all ages are welcome and parents and siblings are encouraged to participate as control samples for this study.

To register for this blood draw, [click here](#).

Interested parents can opt for their child's involvement in the blood draws by reviewing the **consent form** [here](#).



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 - June 07, 2024

Bowling @ 4:00 PM - 6:00 PM

Venue - Glen Burnie Bowl

6322 Ritchie Hwy, Glen Burnie, MD 21061

Phone no. - 443-312-8538

Dinner - 6:00 PM onwards

Venue - Guinness Brewery

5001 Washington Blvd, Halethorpe

MD 21227

Phone no. - 443-575-6893

(Buy your own ticket and dine event)

To sign up for this event please click [here](#)

Saturday: Pizza by the pool



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

Date - June 08, 2024

Time - 5:00 - 08:00 PM onwards

Location - Doubletree Baltimore BWI Airport

Sponsored by: Medical Management and Rehabilitation Services

SunnySide Start Breakfast



Start your day with us before everyone heads back home.

Date - June 09, 2024

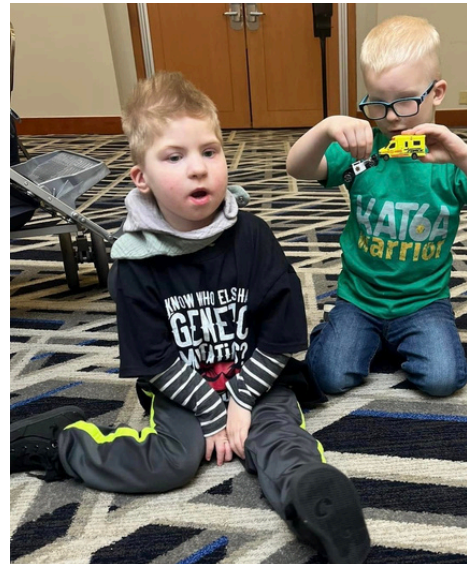
Time - 07:00 AM - 09:00 AM

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.



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
- Protomac Community Resources for sponsoring Amy Gardiner, music therapist
- Medical Management and Rehabilitation Services
- Bay Tower family for providing babysitting services
- Melanie Mass for organizing networking and fun activities for families attending the conference
- Susan Carpenter for donating two handmade quilts
- Pets on Wheels for providing a therapy dog

SCAN THE QR CODE TO REGISTER FOR THE CONFERENCE

Advance registration is mandatory.

