



Volume 11 | December 2025 | KAT6 Foundation, Inc. | [www.kat6.org](http://www.kat6.org)

**Every milestone the KAT6 Foundation has achieved this year is thanks to our incredible community of families, supporters, and donors.**

When we began in 2017, fewer than 75 individuals were known to have KAT6 syndromes. Today, that number has grown to more than 700. This remarkable progress reflects greater awareness, improved diagnosis, and the strength of our connected global community.

Together, we're building a brighter future for everyone affected by KAT6A and KAT6B. Inside this newsletter, you'll discover the impact of our research initiatives and the meaningful support we're providing to families worldwide.

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## LETTER FROM BOARD CHAIR



Dear KAT6 Community,

It has been another meaningful and productive year for the KAT6 Foundation, thanks to the continued engagement of our families, the dedication of our volunteers, and the steady progress of our research partners. Each of you plays a vital role in moving this work forward, and I am grateful for the strength and spirit of this community.

This summer, we hosted our largest International KAT6 Conference to date. Fifty-nine families and twenty scientists joined us from around the world to share experiences, learn from one another, and deepen our collective understanding of KAT6A and KAT6B disorders. The continued growth of this event reflects how far we have come as a community and underscores the importance of these connections.

Our support for research also expanded this year. For the first time, the Foundation released a public Request for Proposals (RFP) and received many compelling proposals from the research community. The Foundation selected eight new studies to fund, which aim at improving our understanding of the biology of KAT6 syndromes and identifying pathways that may lead to future treatments. These investments are possible because of your fundraising and generosity, and they are essential to accelerating progress. Additionally, the first official KAT6 Clinic is now underway in Boston and is scheduling appointments to see patients.

We also maintained our commitment to providing direct support to families. In 2025, the Foundation awarded twenty Empowered Grants to help with therapies, equipment, and other critical needs. These grants reflect our belief that advancing science and supporting families must go hand in hand.

Looking ahead to 2026, there is much to be excited about. Early in the year, we will release a new KAT6 Caregiver Handbook, designed to provide families with clear, accessible information and practical guidance. We are also planning a Family Connection Weekend to bring families together in a smaller, more informal setting to strengthen relationships and share experiences. In addition, we are working with the National Organization for Rare Disease (NORD) to update and enhance the patient registry, supporting researchers and ensuring that future studies are built on stronger, more comprehensive data.

As we close the year, I encourage you to stay connected, share the Foundation's work with your networks, and consider contributing to the year-end Annual Appeal. Every effort you make, whether attending an event, volunteering, or supporting research, brings us closer to a future where every family affected by KAT6 syndromes has the knowledge, care, and hope they need.

Thank you for being part of this incredible community. Together, we are building a brighter, stronger future for every individual and family touched by KAT6. Here's to a 2026 filled with progress, connection, and hope.

Sincerely,  
*Jordan Muller*

Chairperson of the Board of Directors  
Interim Executive Director

## Leadership Update

### *Important Leadership Update for KAT6 Community*

After eight years of incredible service to our community, Natacha Esber and Emile Najm, the founders of the KAT6 Foundation, have made the decision to step down from their positions as Chair of the Science Committee and CEO, respectively. We extend tremendous gratitude to them as the creators of our foundation, and as tireless advocates for our mission to advance scientific research and to support our families. There is no part of this organization that has not been touched by their incredible passion, vision and drive. Natacha has advanced scientific research and provided countless hours of medical advice to numerous families. Emile has run the legal and financial aspects of our foundation, as well as overseeing all of our committees. They will always be part of our community, and we hope you will join us in thanking them for their service.



The Foundation remains strong, stable, and mission focused. The Board of Directors is fully engaged, and Jordan Muller will serve as Interim Executive Director. Our Science Committee continues its work without interruption, and all research projects and partnerships remain active.

As we continue to grow, several new roles will be posted soon. We welcome parents, caregivers, professionals, and supporters who would like to join our mission. The future of the Foundation is bright and we're grateful to walk into this next chapter together.

[Read Full Announcement](#)

### *New Conference Committee*

We're excited to share that a new Conference Committee has been formed, and planning for our next annual gathering is already underway. The team is working to create an event that supports families, strengthens connections, and brings our community together in meaningful ways.

Our goal is to announce the official date for the 2026 conference in January. We know how important this event is for families, and we can't wait to bring everyone together again. Stay tuned for updates in the new year.

# WELCOMING NEW FAMILIES

## Welcome Committee

In 2025, KAT6 Foundation formally developed a Welcome Committee led by our Advocacy Team. When a caregiver receives a KAT6 diagnosis for their child, the team provides them with helpful resources and emotional support.

## Join the KAT6 Community



470 Families have joined our KAT6 Family Map.

In order to help share important research information, community events, and support services, we ask you to fill out the KAT6 Foundation's Community Survey. Having this basic information is vital to advancing our mission.

We will not share this information with anyone outside our community.

View our [privacy policy](#).

[TAKE SURVEY](#)

## Introducing Our KAT6 Welcome Video

We're excited to share a special new resource for families!

The KAT6 Foundation has created a **Welcome Video** for parents and caregivers of children newly diagnosed with KAT6 Syndrome.

This 6-minute video, beautifully produced by KAT6 father and filmmaker Jeff Worden, offers a heartfelt introduction to the KAT6 community and the many ways our Foundation supports families around the world.

 [WATCH IT HERE!](#)



### Newly Funded Projects in 2025

We are proud to fund **eight new studies** advancing our understanding of KAT6 syndrome. These projects address disease mechanisms, model development, potential therapies, and biomarkers, laying critical groundwork for clinical trials. Visit <https://kat6a.org/funded-projects/>

#### 1 **Engineering novel genetic tools to unravel the complex KAT6-disease phenotype**

**Chief Investigator: Effie Apostolou, PhD**

**Institution: Weill Cornell Medicine**

Dr. Apostolou's team is working to create a stem cell genetic model which will allow them to control the amount of KAT6A and KAT6B proteins available in a cell. This will allow them to see how different amounts of these proteins affect cell development and highlight future potential therapeutic targets. The group is committed to making their model available to the broader research community. *(This study is fully funded by an anonymous donor.)*

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#### 2 **Patient-Specific Neurodevelopmental Models for KAT6B Mutations**

**Principal Investigator: Valerie Arboleda, MD, PhD**

**Institution: David Geffen School of Medicine, University of California, Los Angeles**

This study leverages patient-derived induced pluripotent stem cell (iPSC) lines to investigate how specific KAT6B variants contribute to the phenotypic divergence between Genitopatellar Syndrome (GPS) and Say-Barber-Biesecker-Young-Simpson syndrome (SBBYSS). Brain-like cells grown from patient samples will help researchers understand condition-specific development and open the door to personalized therapies.

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#### 3 **Neurobehavioral Differences in Early- and Late-Truncating KAT6A Mouse Models**

**Principal Investigator: Valerie Arboleda, MD, PhD**

**Institution: David Geffen School of Medicine, University of California, Los Angeles**

Dr. Arboleda's team is creating new, KAT6A variant-specific mouse models to study how different types of KAT6A variants affect brain development and behavior. Based on data from their lab, they will test therapies to see if it can help improve symptoms in mice with severe KAT6A mutations. This study brings us closer to testing treatments that target the specific effects of different mutations in people.

## Funded Projects cont'd

**4 A Multidisciplinary Clinical Program and Identification of a Metabolomic Profile in KAT6A/KAT6B Conditions to Inform Clinical Trial Readiness**

**Co-Investigators: Olaf Bodamer, MD, PhD and William Brucker, MD, PhD**

**Institution: Boston Children's Hospital & Harvard Medical School**

Dr. Bodamer is launching a new clinical program for patients with KAT6A and KAT6B syndromes at Boston Children's Hospital. The team will collect detailed health data and samples from patients to better understand the natural course of these conditions. This project combines high-quality patient care with research aimed at preparing for future clinical trials.

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**5 Biomarker Discovery in KAT6A for Translation into Clinical Trials**

**Chief Investigator: Sarah Donoghue, MBBS, FRACP**

**Institution: Murdoch Children's Research Institute (MCRI), University of Melbourne**

This project seeks biomarkers in blood and brain tissues to further understand how cognitive function develops in KAT6A in hopes of using this information to measure disease progression and treatment success. We hope to understand the impact of KAT6A on brain function in mice and whether carnitine treatment improves this, paving the way for future human trials.

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**6 CA3 Neuronal Development in KAT6A and KAT6B Patient-Derived iPSCs**

**Principal Investigators: Thomas Durcan, PhD, Faiza Benaliouad, PhD and Gilles Maussion, PhD**

**Institution: Neuro: Montreal Neurological Institute-Hospital & McGill University**

Focusing on the CA3 hippocampal region linked to memory, this study uses patient-derived stem cells to uncover how brain cell development is altered in KAT6 syndromes. The goal is to find points for therapeutic intervention.

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**7 Epigenetic Landscapes and Gene Regulation in KAT6 Disorders**

**Co-Investigators: Maria A. Serrano, PhD and Gustavo Mostoslavsky, MD, PhD**

**Institution: CReM & Boston University Chobanian & Avedisian School of Medicine**

This research examines how KAT6 mutations affect gene regulation in brain, gut, and blood cells. The team will use an advanced method to see how these cells' "epigenetic landscapes" (chemical markers that control gene activity) are different from healthy cells.

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**8 When Proteins Go Wrong: Unravelling the Impact of KAT6 Variants on Protein Structure and Function**

**Principal Investigator: Shabih Shakeel, PhD**

**Institution: Walter and Eliza Hall Institute of Medical Research**

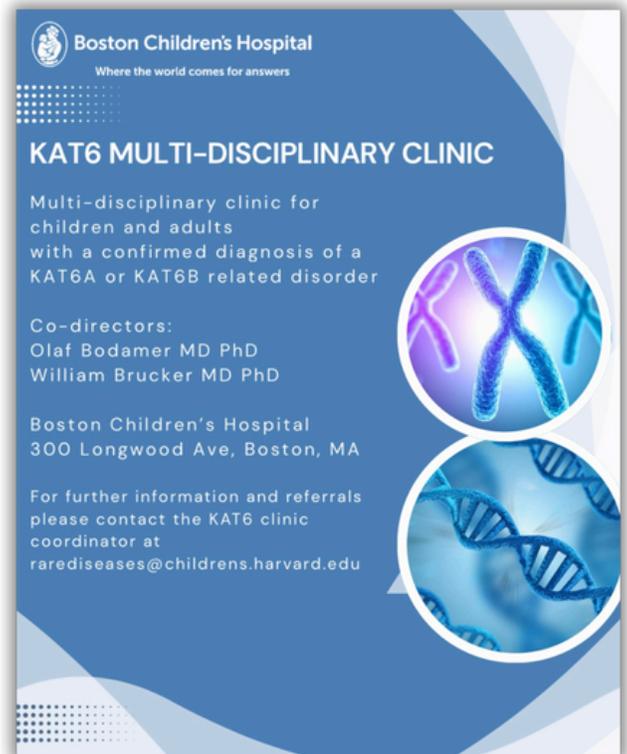
This project is focused on characterizing the effects of different KAT6A and KAT6B mutations on protein structure and important protein functions such as binding with protein partners and acetylation. They will do this first characterization by isolating the proteins and studying them in test tubes. Dr. Shakeel's team will then see how these changes in function lead to downstream changes to actual cells. *(This study is fully funded by an anonymous donor.)*

# KAT6 Clinic opens at Boston Children's Hospital

A new milestone for the KAT6 community has arrived. Led by Dr. Olaf Bodamer and Dr. William Brucker, Boston Children's Hospital has launched a dedicated multidisciplinary clinical program designed to serve as a true medical home for individuals with KAT6 syndrome. Families now have access to coordinated care across specialties, along with clinicians who understand the unique needs associated with KAT6A and KAT6B.

This clinic was made possible in part through funding from the KAT6 Foundation, supported by the generosity of our donors. Their commitment continues to open doors for improved clinical care and meaningful research progress.

In addition to providing care, Drs. Bodamer and Brucker will collect essential natural history data and biospecimens for biobanking. They also plan to collaborate with established clinical and scientific partners, including the Serrano Lab at Boston University, to advance research that will deepen our understanding of KAT6 and guide future therapies.



The flyer features the Boston Children's Hospital logo at the top left, with the tagline "Where the world comes for answers". The main title is "KAT6 MULTI-DISCIPLINARY CLINIC". Below this, it states "Multi-disciplinary clinic for children and adults with a confirmed diagnosis of a KAT6A or KAT6B related disorder". The co-directors listed are Olaf Bodamer MD PhD and William Brucker MD PhD. The address is "Boston Children's Hospital, 300 Longwood Ave, Boston, MA". At the bottom, it provides contact information: "For further information and referrals please contact the KAT6 clinic coordinator at [rarediseases@childrens.harvard.edu](mailto:rarediseases@childrens.harvard.edu)". The flyer is decorated with two circular images: one showing a blue X-shaped chromosome and another showing a blue DNA double helix.



[Olaf Bodamer, MD, PhD, MA](#)

To schedule an appointment or for further information, contact the KAT6 clinic coordinator at [rarediseases@childrens.harvard.edu](mailto:rarediseases@childrens.harvard.edu).

## Update on KAT6 iPSC Bank



In 2022, the KAT6 Foundation, in collaboration with Center for Regenerative Medicine (CReM) and The Serrano Lab at Boston University, established the first patient-derived induced pluripotent stem cell (iPSC) bank for KAT6A and KAT6B. These cell lines are invaluable tools that allow researchers to study KAT6 syndrome using cells that reflect each donor's unique genetic background. They can be differentiated into various cell types, including neurons and heart cells, to better understand how KAT6 variants influence development. Families may choose to contribute samples from their child to the iPSC bank, offering a meaningful way to support research and future treatments.

### Participate

By making these resources freely available, CReM and the Serrano Lab foster collaboration, reduce duplication of effort, and accelerate the translation of basic research into clinical applications. This approach helps ensure that patients, researchers, and the global community benefit from scientific progress as quickly and equitably as possible.

Currently, the Biobank has 42 samples available for research. Most donors are children and adolescents, and the collection includes families from diverse backgrounds. We remain committed to ensuring that research benefits all communities affected by KAT6 syndrome. Through our partnership with Boston Children's Hospital, we hope to continue expanding this important resource.

Biorepository Summary Statistics

Category	Count	% of Total
Total PBMC Samples	42	100%
iPSC Lines Established	8	19%
KAT6A Samples	28	66.7%
KAT6B Samples	14	33.3%

For inquiries concerning available iPSC cells please contact Serrano Lab at [mfjames@bu.edu](mailto:mfjames@bu.edu).



## KAT6A/KAT6B Patient Registry

Launched in 2019 through the National Organization for Rare Disorders (NORD), the KAT6 Patient Registry is the first long-term study of KAT6 syndrome. By collecting information from caregivers, the registry helps researchers better understand the full range of KAT6 characteristics and identify areas for new studies. **Today, 532 patients are registered.**

The KAT6 Foundation owns the data, which allows us to guide research and share findings responsibly. We also analyze the information and provide insights to families, helping parents and caregivers better understand and support their loved ones. In 2025, we began updating the registry to make it more user-friendly. The new registry will simplify updates and improve the experience for families — look for it in 2026.

[Register Now](#)

## 2025 Published Research Articles

### KAT6A

[Comprehensive review and outline of genotypes and phenotypes of Arboleda-Tham syndrome spectrum: insights from novel variants](#)

[KAT6B overexpression rescues embryonic lethality in homozygous null KAT6A mice restoring vitality and normal lifespan](#)

[Research themes in KAT6A syndrome: A scoping review](#)

### KAT6B

[KAT6B overexpression in mice causes aggression, anxiety, and epilepsy](#)

[Loss of KAT6B causes premature ossification and promotes osteoblast differentiation during development](#)

For an archive of KAT6 publications, visit [www.kat6.org](http://www.kat6.org)

# ASSISTING FAMILIES

## Empowered Grants

Donations made throughout the year support our Empowered Grant Program, which provides funding for the purchase of assistive equipment, technology, and therapy treatments for individuals diagnosed with KAT6.

In 2025, we awarded **19** Empowered Grants to support a variety of needs - including aquatic, physical, and speech therapy - as well as reimbursement for essential items such as safe beds, therapy swings, and communication devices. Your generosity makes a meaningful difference in the daily lives of those affected by KAT6!

[Learn more about the Empowered Grant Program and its impact.](#)



*Thanks to regular work with the speech therapist, he has made noticeable, even if still small, but very meaningful progress. Yegor does not speak like a typical child yet, but now he can say a few words that we are able to recognize — something that was almost impossible before. . . For us, this is a significant achievement and an important step forward.*

*We are sincerely grateful to the KAT6 Foundation for providing such essential support for Yegor.*



-Marina

## Family Resources

In 2025, we added many new resources to our website to better support families, including links to webinars, videos, and fundraising/awareness materials. Coming soon: the NEW KAT6 Caregiver Handbook!

### [Family Resources](#)



## EVENTS RECAP

### 6th International KAT6 Conference

On May 30–31, more than 200 members of the KAT6 community, including 58 families and 27 scientists and therapists, came together in Baltimore, MD for the 6th International KAT6 Conference! It was an unforgettable weekend filled with emotional connections, groundbreaking research, and joyful moments.

The event kicked off on Friday with family social gatherings, where participants connected over bowling and shared laughter and stories at the Guinness Brewery, setting a warm and welcoming tone for the weekend.

Saturday's one-day scientific conference focused on strengthening international collaboration and advancing research on KAT6A and KAT6B. Sessions explored a wide range of topics, including personalized medicine, iPSC cell lines, neuropsychological assessments, animal models, advocacy, and updates on the KAT6 Patient Registry and fundraising initiatives.

The Foundation supported three research groups by facilitating family participation in data collection. In addition, three family workshops on speech and communication, advocacy, and a roundtable with Dr. Richard I. Kelley were highlights of the weekend, offering opportunities for learning, sharing, and connection.

A special moment of the conference was honoring Dr. Richard I. Kelley for his unwavering dedication to KAT6 patients and his lasting impact on the community.



-  [Summary of 6<sup>th</sup> Annual KAT6 Conference](#)
-  [Conference Highlight Video by Jeff Worden](#)
-  [Conference Videos and Presentations](#)



# KATwalk 2025

*Walking for a brighter tomorrow!*



Thanks to the incredible leadership of our KATwalk Co-Chairs, Katie Bator and Aimee Reitzen, the dedication of our ambitious Team Captains, and the generosity of our community, KATwalk 2025 raised more than \$324,000 — our most successful KATwalk yet! We received over 1,600 individual donations. We extend our heartfelt gratitude to everyone who volunteered, donated time, money, or resources, spread the word, and supported their loved ones both in person and from afar. Your continued commitment makes our mission possible, and for that we thank you.

This year, 15 in-person walks took place across 10 U.S. states, with teams walking from coast to coast. We also had 17 KATwalk ANYWHERE teams who chose to fundraise virtually, giving their supporters the flexibility to participate throughout September.



- KATwalk San Diego - \$117,681**
- Team Luke - \$28,670**
- Team Benjamin - \$28,025**
- Walk for Jack - \$16,650**
- Bloom with Lily - \$16,070**
- Mattie & Everson - \$13,345**
- Will's Warriors - \$12,702**
- Roar for Robby! - \$11,635**
- Charlotte's Stampede - \$7,911**
- Go YaYa! Go YaYa! - \$7,851**



We also launched a new KATwalk t-shirt design and logo that highlights the strength and support of our community.



## Raising KAT6 awareness on a global level!

This year, members of our KAT6 community took part in major rare disease events, helping raise awareness on a global scale.

Susan Hartung, our Director of Advocacy, attended the **NORD Breakthrough Summit** in Washington, DC, which took place October 19–21. In the UK, Board member Dr. Andrew Rankin and KAT6 parent Vaila Morrison represented our community at **Rare Summit 2025**, hosted by the Cambridge Rare Disease Network on November 6.

Their involvement ensures that the needs and voices of KAT6 families are included in important conversations shaping the future of rare disease research and advocacy.



Dr. Lindsey Murphy and KAT6 mom Kristin O'Brien brought a dream to life by writing the first book about KAT6 — **KAT6 and Me**. This beautifully written book teaches, comforts, and celebrates children living with KAT6 disorders, while also helping others understand and include them. Read an in-depth interview [here](#).



[ORDER YOUR COPY!](#)

In March 2025, our KAT6 Spain families visited Dr. José Antonio Sanchez Alcazar and his team at Pablo Olavide University in Seville. There, families were given a tour and a chance to see advances being made in the KAT6 world.



# Raising KAT6 awareness on a local level!

We appreciate everyone who raises awareness within their own communities. This year, KAT6 families hosted a wide range of fundraisers to support our foundation, including a golf outing, coffee sale, Halloween banquet, bracelet fundraiser, 5K, and more. Special thanks to Bird Rock Coffee Roasters, Waconia Brewing Company, Merrick Dads Cup, the American Legion Auxiliary of Grand Haven, MI, and More Love Collective for their generosity and support.



Fundraise year-round with our  
**I Care for Rare** fundraising platform!  
[Create your page here](#)

## Rare Disease Day 2025



Each year, the KAT6 Foundation participates in international Rare Disease Day, which takes place on the last day of February. The 2025 theme was "More than you can imagine." Board Member Rachel Worden designed Ultra Rare tees for our community, and we encouraged individuals to donate \$6 on that day for KAT6. This social-media-driven campaign raised more than \$3,000 in a single day.

**Mark your calendars for February 28, 2026,** and stay tuned for our new T-shirt design.



### Help us Fundraise for the Annual Appeal

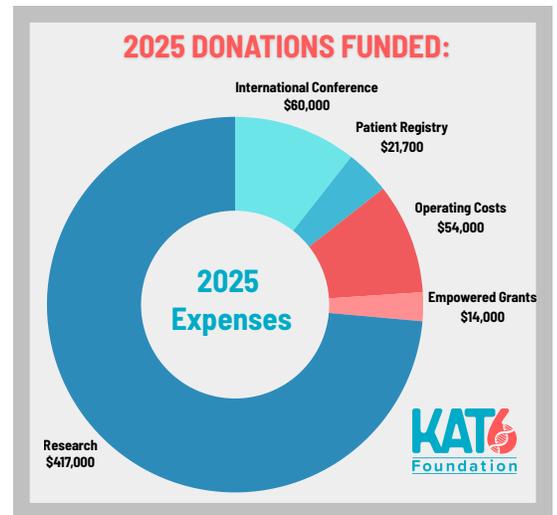
Every December, the KAT6 Foundation launches our Annual Appeal, chaired by Board Members Karen Ginsburg and Rachel Worden. **Reaching our goal of \$350,000 will allow us to continue our important work.**

Donations made during the Annual Appeal fund essential initiatives that support our community, including the new KAT6 Clinic at Boston Children's Hospital, groundbreaking research, updates to the KAT6A/KAT6B Patient Registry, the iPSC BioBank, and the Empowered Grant Program. These funds also help us provide valuable educational resources for families and caregivers, such as informational webinars, fundraising materials, KAT6 brochures, and a new handbook coming in January 2026.

Help us finish strong in 2025 — we have big dreams for 2026.



[Download Fundraising Toolkit](#)



### Three More Ways to Support our Annual Appeal:



**Ask your employer about their charitable donation matching program.**



**Share our posts on social media to spread awareness.**



**Email your friends and family.**

**DONATE**

## Five ways to help individuals with KAT6 today!

At the KAT6 Foundation, the season of gratitude is in full swing and we wanted to take this opportunity to thank you for all you do to help move our mission forward.

Interested in continuing your impact?

### Explore these five ways you can support us this year:

#### Make a one-time gift:



Every dollar donated supports the important work we do at the KAT6 Foundation. Your one time gift supports our Empowered Grant program, helps drive research and makes a real-time impact in the day-to-day lives of KAT6 families.

#### Start a recurring giving plan:



Recurring gifts help sustain our foundation year-round. Donate today and choose a recurring donation plan that fits your giving capabilities - monthly, quarterly, semi-annually or annually!

#### Leave a gift in your will:



Include the KAT6 Foundation in your estate planning, planting seeds for a brighter future for KAT6 families for years to come.

#### Make a gift in honor of a loved one:



Give a meaningful gift this holiday season! Donate to the KAT6 Foundation in honor or memory of a loved one and we'll email a card to let them (or their family) know. Just don't forget to check the box that says "Dedicate my donation".

#### A holiday two-for-one!



Celebrate the holiday season AND support the KAT6 Foundation this month by visiting the KAT6 store for branded hoodies, onesies, t-shirts, computer bags, coffee mugs and more! There's something for everyone on your list! [Shop now!](#)

**Questions?** Email board members  
Karen Ginsburg at  
kginsburglcs@gmail.com  
or Rachel Worden at  
rachelnworden@gmail.com



Give to KAT6

#### Send a personal check:

KAT6 Foundation  
8 Leland Court  
Chevy Chase, MD 20815  
Nonprofit EIN # 82-3118535

# OUR TEAM

2025

## Board of Directors

**Jordan Muller, Board Chair**

**Karen Ginsburg**

**Katie LaRow Brown**

**Maureen Martini**

**Myria Normann**

**Andrew Rankin**

**David Woodbury**

**Rachel Worden**

**Kevin Young**

The KAT6 Foundation is run by passionate KAT6 parents, with part-time professional support in research and marketing. As the Foundation evolves, we will post several new roles in the coming weeks. If you or someone you know would like to join our mission, we would love to hear from you.

Our team is mostly made up of parents and caregivers, but not entirely, and we welcome anyone with a passion for helping this community thrive. Please keep an eye out for position announcements soon.

For those interested in a larger role, there are also opportunities to serve on the KAT6 Foundation Board of Directors. The Board meets every other month.

### **Executive Director**

Jordan Muller, *Interim*

### **Marketing and Communications**

Aimee Reitzen, *Director*

Megan Stetts, *Social Media*

Thayer Glass, *Technology Advisor*

Jeff Worden, *Video Production*

### **Fundraising**

Karen Ginsburg, *Chair*

Katie Bator, *KATwalk Co-chair*

Aimee Reitzen, *KATwalk Co-chair*

Rachel Worden, *Annual Appeal*

Megan Stetts, *Fundraising Assistant*

### **Advocacy Committee**

Susan Hartung, *Director*

Susan Carpenter

Hyeryun Choi, *Liaison to Korea*

Jane Ellul, *Liaison to Australia and NZ*

Jennifer Sander

Beth Woodbury

Amy Young

### **Science and Research**

Katie LaRow Brown

Typhaine Lejeune

Andrew Rankin

Dr. Tanya Tripathi

### **Empowered Grants**

Jessica Wiemann, *Coordinator*

### **Grants and Sponsors**

Lindsey Blanch, *Co-chair*

Rachel Worden, *Co-chair*

### **Conference Committee**

Lindsey Blanch

Melanie Maas

Jordan Muller

Andrew Rankin

David & Beth Woodbury

### **NORD Patient Registry**

Susan Hartung

Rosa Strino

# KAT6 Foundation



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[www.kat6.org](http://www.kat6.org)  
EIN # 82-3118535



*In honor of those we lost this year,  
we extend our deepest condolences to the parents and  
families who are grieving. We mourn with you.*