KAT6 NEWS

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KAT6 Foundation

The KAT6A Foundation was founded in 2017 by a handful of parents of children identified with mutations on their KAT6A gene. At the time, there were less than 50 known KAT6A cases. In the span of 5 years, our numbers have expanded to over 370 known KAT6A cases worldwide.

In 2020, the KAT6A Foundation began fostering connections with the KAT6B community and in 2022, we formally became the KAT6 Foundation, an organization devoted to understanding mutations in both KAT6A and KAT6B genes. Today, there are more than 500 known cases of KAT6 syndromes worldwide, and we expect our numbers to continue to grow.

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LETTER FROM BOARD CHAIR

Dear KAT6 Community,

It has been another great year for the KAT6 Foundation thanks to the engagement and participation of our family community and the dedication of our volunteers.

Thank you to all of the organizers, participants and donors of our annual KATwalk awareness and fundraising event in raising over $175,000. Thank you as well to all the families, researchers, and clinicians, particularly the Kennedy Krieger Institute and Dr. Angie Serrano of Boston University, who attended our annual KAT6 Conference in April in Boston! We look forward to even greater participation at the 2024 conference on June 8th in Baltimore.

This year our research community, supported through your donations and fundraising, has been publishing their progress in understanding the fundamental aspects of the KAT6A and KAT6B disorders - you can find the new publications on the foundation's website.

To provide direct support to KAT6 families, the Foundation made 21 individual Empowered Grants to families to support assistive devices and therapies for their loved ones and we look forward to continuing this program in 2024.

The Foundation also found creative new pathways for spreading awareness of the KAT6 Disorder. First, a documentary movie about KAT6 called "Genetic Defekt" by filmmaker Niko Mylonas premiered in January and received the Austrian Child Welfare Award. Second, on the evening of October 7th, Niagara Falls were illuminated in the KAT6 colors. Please continue to spread the word about our KAT6 community and refer potential supporters to our website on ways they can donate or volunteer.

Lastly, I have two requests for you. First, we are always looking for volunteers interested in assisting the Foundation's activities - to include serving on the foundation's Board of Directors. If you are interested, please reach out to support@kat6a.org. Second, we are in the midst of our year-end Annual Appeal fundraising campaign. Please reach out to your network for folks who may not have been able to give earlier in the year or participate in the KATwalk.

Best wishes for a happy and healthy 2024!

Sincerely,
Jordan Muller
KAT6 Foundation, Board Chair

EVENTS RECAP

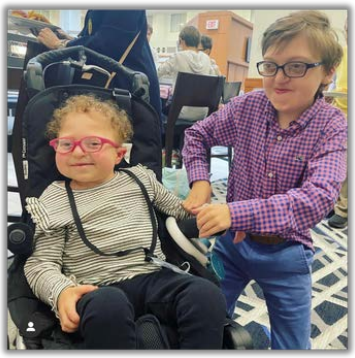
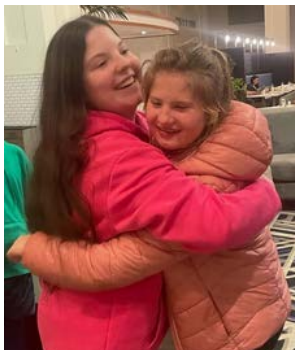
4th International KAT6A & KAT6B Conference

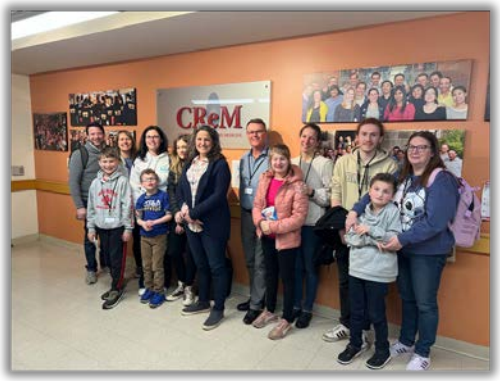
The 4th International KAT6 Conference took place on April 1, 2023. It was a patient-centered, collaborative event, organized and sponsored by the KAT6 Foundation, designed to solidify KAT6A and KAT6B research internationally and enable open dialogue between families, clinicians, and researchers.

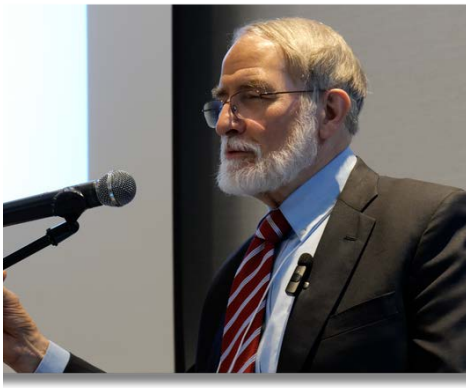
The event had a total of 200 registrants, including 60 families and 21 scientists from around the world. Taking place in Boston, Massachusetts, USA, the conference spanned a single day and covered a wide range of topics, such as personalized medicine, the role of iPSC cell lines, neuropsychological assessments, epilepsy, autism and its relation to KAT6A and KAT6B, special education, the KAT6A and KAT6B patient registry, and initiatives led by the KAT6 Foundation, such as the KATwalk.

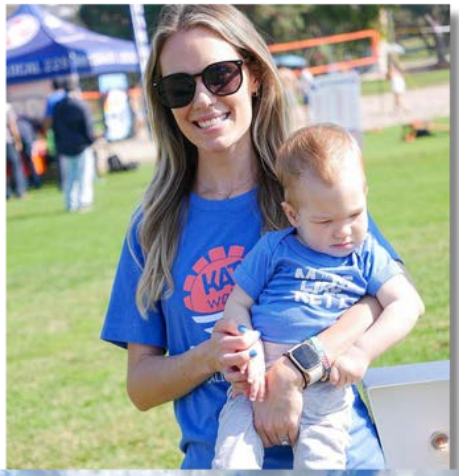
During the conference, assistance was provided to three research groups by creating a space for families to participate in research data collection. Additionally, the foundation organized consultation visits for families to meet experts in the field of KAT6A and KAT6B-related syndromes.

Many videos from the conference are available on the KAT6 Foundation YouTube page, including the Serrano Lab Tour, How the KAT6 Foundation Helps Newly Diagnosed Families, The KAT6A and KAT6B Registry with Dr Bhawika Sharma and Supporting Patient-Focused Research by Saylor Williams.



KATwalk 2023

KATwalk 2023 was our most successful walk to date!

Thanks to the incredible efforts of our KATwalk Chair, Katie Bator, our ambitious KATwalk hosts and their wonderful friends, families and sponsors, we raised **$188,000**!

We are blown away by such an incredible show of support from friends and family, allowing us to continue raising awareness and supporting families in our community.

In-person walks took place in 10 US states and our virtual teams spanned Canada, Germany and Ireland, with a total of 32 teams participating across the globe!

We want to extend a huge thank you to everyone who volunteered, donated time, money and resources, spread the word, and supported their loved ones in person or from afar.

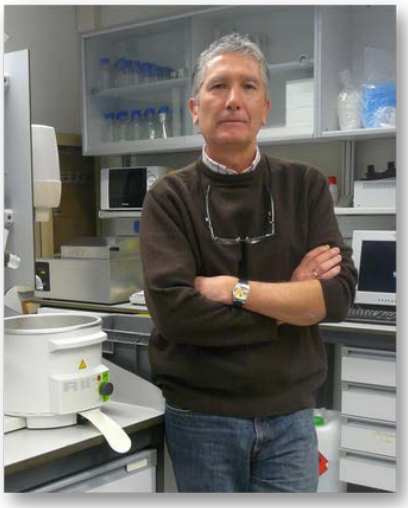
Top 10 Teams

Team Benjamin $26,663
More Like Kett $21,758
Forza Franco $17,932
Walk for Jack $14,830
Charlotte's Stampede $14,368
Gigi's Guardian Angels $13,551
Chloe's KATwalkers $11,279
Jack's KAT Pack $9,794
Will's Warriors $9,305
Miles for Moira $7,986

WALKING FOR A BRIGHTER TOMORROW!

Published Research

A KAT6 Foundation Funded Research Project Gets Published!



We are proud to report that research led by Dr. José A. Sánchez-Alcázar and his team was published on November 15, 2022 in a journal article titled, **Pantothenate and L-carnitine Supplementation Corrects Pathological Alterations in Cellular Models of KAT6A Syndrome**. The article is available on the Preprints Platform as it is waiting peer review. This is an important milestone for our Foundation as it is the first research project that we directly funded to reach publication, and is an important step forward on the path to finding a treatment for KAT6 individuals.

Three individuals with KAT6A gene variation participated in the study conducted at Universidad Pablo de Olavide in Spain. An initial series of experiments generated evidence supporting the use of patient-derived fibroblast to study KAT6A gene variation.

The team identified four critical pathophysiological processes altered by KAT6A gene variation: 1) Coenzyme A (CoA) metabolism, 2) Iron metabolism, 3) Enzymatic antioxidant system and 4) Mitochondrial function. Two compounds were identified to have a positive impact on the altered physiological pathways. These compounds are: 1) Pantothenate and 2) L-carnitine. Pantothenate is a CoA metabolism activator and L-carnitine is a mitochondrial boosting agent. Supplementation with pantothenate and L-carnitine supported the survival of the KAT6A fibroblast in a stress inducing medium. The concentration of pantothenate and L-carnitine varied in all three KAT6A cell lines suggesting that different type of mutations respond differently to these positive compounds. The KAT6A gene plays a significant role in histone acetylation which is a key process involved in cell progression and differentiation. Supplementation with pantothenate and L-carnitine resulted in significant increase in histone acetylation, recovery of gene expression patterns and expression levels of proteins affected due to the KAT6A gene variation.

We want to extend our sincere thanks to Dr. José A. Sánchez-Alcázar and his entire team for their professionalism and commitment to rare disease research and the KAT6 community. We look forward to building upon this partnership in the future.

KAT6A and KAT6B Research Published in 2023

Epigenetics of Cognition and Behavior: Insights from Mednelian Disorders of Epigenetic Machinery

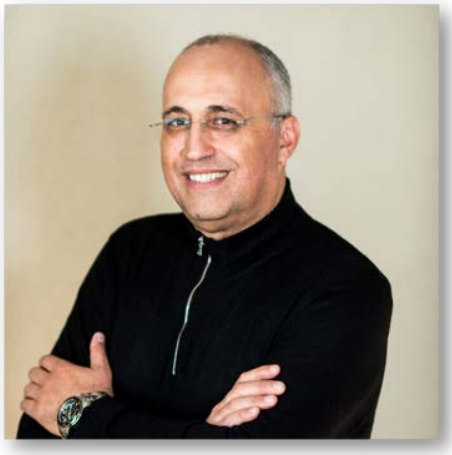
KAT6A Mutations Drive Transcriptional Dysregulation of Cell Cycle and Autism Risk in an Arboleda-Tham Syndrom Cerebral Organoid Model

Clinical Features and Underlying Mechanisms of KAT6B Disease in Chinese Boy

The Omics Era: A Nexus of Untapped Potential for Mendelian Chromatinopathies

Current Research Projects Funded by KAT6

Assaraf Research Lab at Technion Institute



Dr. Yehuda Assaraf of Technion Institute in Israel has been studying KAT6A gene mutations since 2017. In June, 2022 the KAT6 Foundation began funding new research at the Assaraf Research Lab. The current research aims to characterize the molecular loss of function of the KAT6A and KAT6B genes in dermal fibroblasts obtained from the mutation of the KAT6A and KAT6B gene when compared to healthy counterparts, using state-of-the-art metabolomics analysis, including cutting edge seahorse technology.

Quantitative and qualitative alterations in metabolites from mutant KAT6A and KAT6B fibroblasts could uncover the metabolic pathways that may be dysregulated or impaired in KAT6A and KAT6B patients. Upon cross-verification with the complementary omics, analyses which the team previously performed with a single KAT6A patient, paired with relevant literature, this information could be used for the selection of the most appropriate nutrients for personal supplementation of KAT6A and KAT6B individuals, as well as for possible therapeutic interventions.

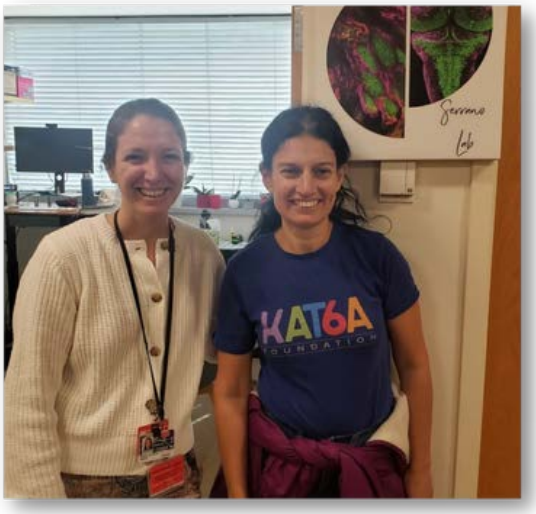
iPSC Bank at Boston University

The KAT6 Foundation and collaborators established the first patient-derived induced Pluripotent Stem Cells (iPSC) bank for KAT6A and KAT6B variants.

The foundation's Science Advisor, Dr. Angie Serrano, stores and maintains biospecimens, such as skin and blood samples of KAT6A and KAT6B patients at Boston University. This bank broadly shares iPSC to the whole research community and industry to spark collaboration and advance research in KAT6 syndromes.

Furthermore, the biospecimens can be used by multiple researchers for multipurpose research studies. Our foundation is responsible for the cost of the reprogramming services for the generation of 5 patient-specific iPSC lines and will continue to fund more cell lines in 2024 if funding is available.

Email support@kat6a.org to provide samples for the KAT6A and KAT6B (iPSC) Bank.



Dr. Serrano and KAT6 Foundation co-founder, Natacha Esber at Serrano Lab.

Webinars, Committees and Involvement with NORD

Webinars and Virtual Conferences

In 2023, the KAT6 Foundation hosted two virtual webinars and six virtual conferences with topics ranging from Early Treatment Pathways for Children with Neurodevelopmental Disorders to Swimming and Beyond with KAT6A.

These webinars and conferences are available on the KAT6 Foundation's YouTube page!

Involvement with NORD

Our scientific committee assisted NORD (National Organization for Rare Disease) in publishing the first NORD report on KAT6B-related disorders. Read it here!

NORD® | RARE. STRONG. TOGETHER. 40 YEARS

Poster Presented at CZI Meeting - October 2023

2023 offered our KAT6 families several opportunities to participate in studies and screenings and give their valuable input as a caregiver to a child with a RARE disease.

Sign up for the Guardian Study - Newborn Screening

New Study Seeks Input from KAT6A Caregivers Regarding Speech and Language Therapy

Seeking Participants for New Research on KAT6B with Dr. Rowena Ng, PhD.

In 2023, the KAT6 Foundation established a committee to study mortality within our community. While we are all here to surround and support those burdened with the loss of a loved one, the ultimate objectives for this committee are to guide parents in understanding how best to adjust to KAT6 disorders and to prevent suffering among our most vulnerable members. We ask that all families register with the KAT6 Patient Registry to support meaningful research.

NORD® 2022 MEMBER PLATINUM

Updates to the Patient Registry

- The KAT6A Patient Registry was renamed as KAT6A/KAT6B patient registry. This change acknowledges the presence of KAT6B families in the patient registry and reflects the Foundation's aim to support all KAT6 families.
- The format changed from 5 long surveys to 12 short, digestible, easier to fill out surveys

Join the Registry

KAT6 at Niagara Falls

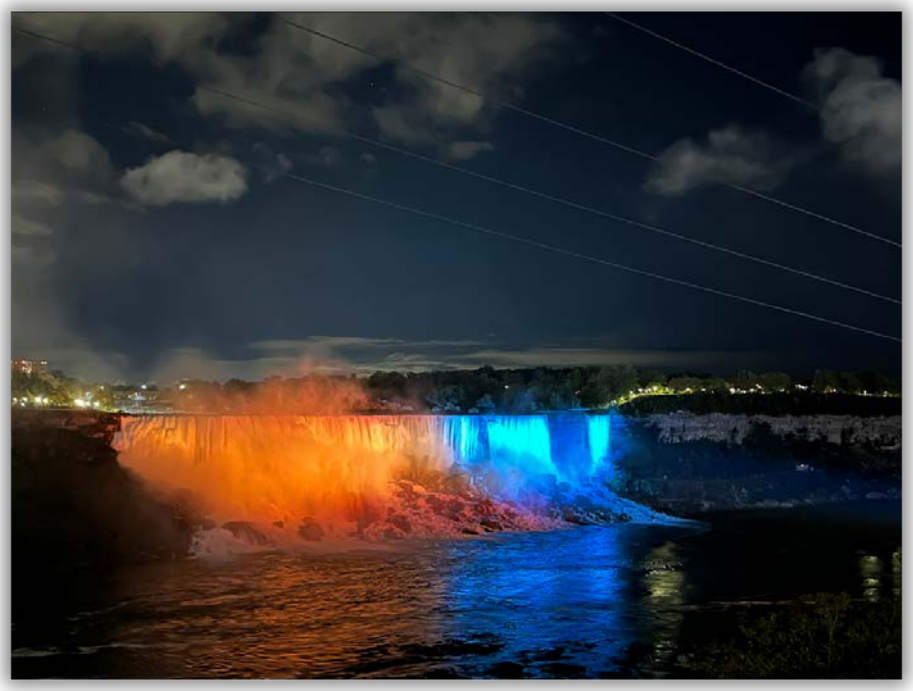
A Note from the KAT6 Foundation's Social Media Coordinator Megan Stetts

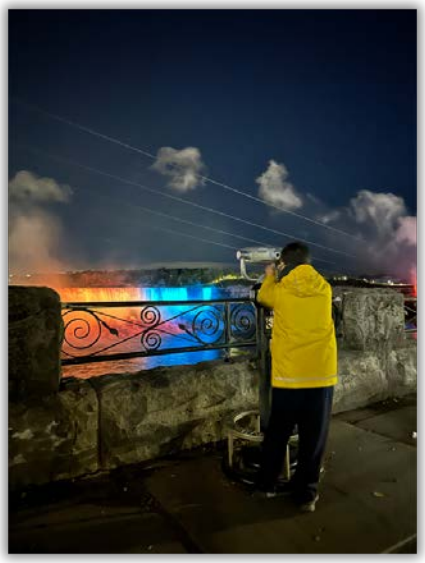
In the summer of 2023, I became aware of an incredible opportunity through the Niagara Falls Illumination Board that allows organizations to submit an application to have the falls lit in their colors to raise awareness. I immediately submitted an application on behalf of the KAT6 Foundation and was thrilled to hear we'd been approved!

I shared the good news far and wide, on all our social media platforms and hoped KAT6 families on both sides of the falls would gather to witness this wonder in person. Luckily, for those unable to make the trek, the illumination was live-streamed for all to enjoy from the comfort of their homes. As a global community, this was huge. We wanted as many families and friends to join us in this moment, even if virtually.

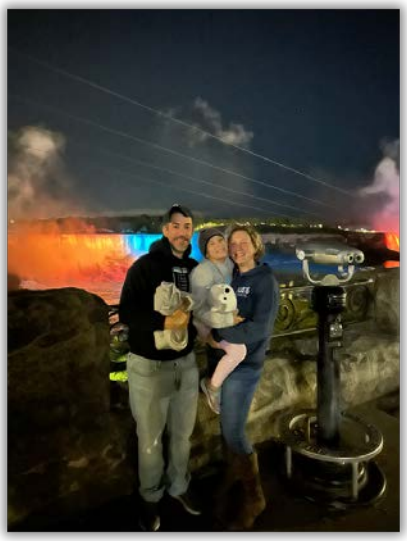
We packed our bags and headed East to Niagara Falls from Spring Lake, Michigan to see the colors firsthand. We had the pleasure of meeting the Lawrence family, watching the fireworks and seeing the falls change colors with them.

The Niagara Falls Illumination was an incredible experience and I am eternally grateful for the opportunity to witness it firsthand.

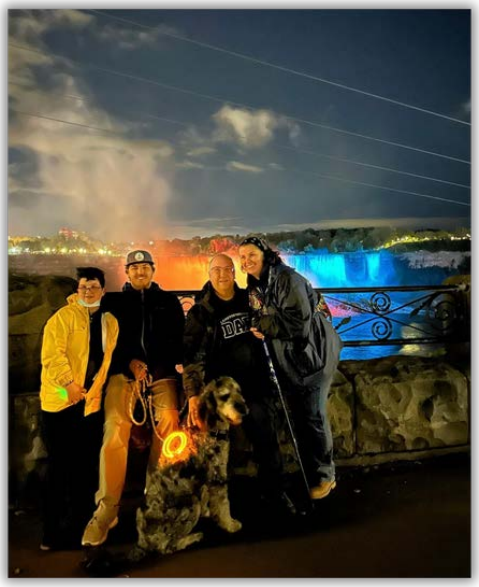




KAT6 kiddo Toby taking in the view.



My family at the falls.



The Lawrence family.

KAT6 on Screen: Genetic Defekt

Working through ZebraKinder, our KAT6 counterpart in Austria, filmmaker Niko Mylonas released the KAT6 documentary, "Genetic Defekt" earlier this year.

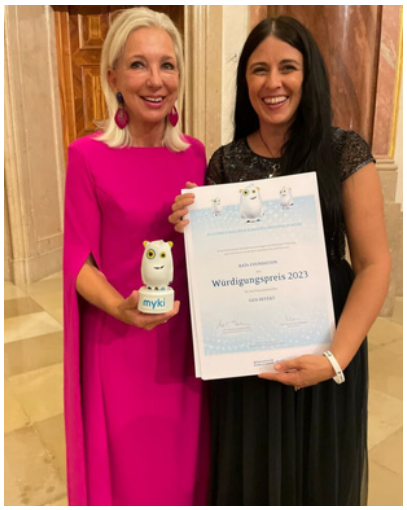
While the film touches on the technical aspects of KAT6, it is an opportunity to get acquainted with families around the world who live with KAT6 syndromes.

In the film, we see the full spectrum, from subtle, almost unnoticeable effect to severe impairment, depending on the type of gene anomaly — truncation, missense, deletion, and other variants.

We meet Ella in Innsbruck and her advocate-aunt, Monika Rammal. We visit Gianna in Michigan, Samantha in Germany, Will in New York, Warren, Bay, Max, Hadley, and many more.

We hear from some of the scientists and parent-advocates including Dr. Jacqueline Harris, Dr. Angie Serrano, Aimee and Jeff Reitzen, Susan and George Hartung. We visit with the Najm family, who, on behalf of their son Peter, had the inspiration to organize parents in starting the KAT6 Foundation.

In addition to a glimpse into the everyday trials and sweet triumphs of those who live with KAT6, the film lets us spend a poignant few minutes with the parents of Helin, a girl in Germany who fell ill and, although brought to a hospital, did not survive. Her parents share the message learned from such tragedy.



Once you've seen it, show it to others, speak of it, send it, share it widely. Make it the centerpiece of a gathering or fundraiser. At just 48 minutes in length, this award winning film is a tool we have long needed to promote awareness of the adversity that has brought us together.

ASSISTING FAMILIES

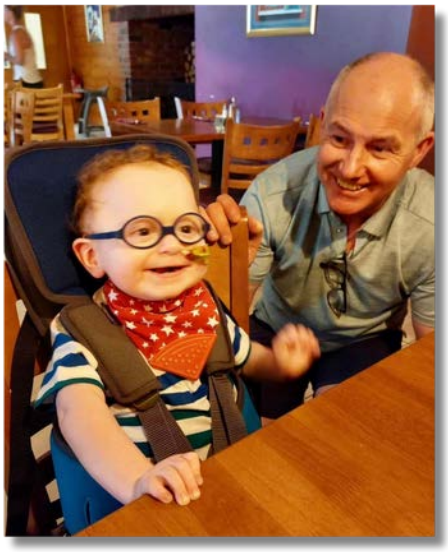
Empowered Grant Program

Thus far in 2023, we've awarded 21 Empowered Grants to KAT6 individuals living in 12 different countries!

Caregivers have received awards to help pay for a wide range of therapies and assistive equipment including hippotherapy, hydrotherapy, physical therapy, tablets with communication software, eye glasses, chairs, adapted tricycles and a portable wheelchair ramp.

In 2024, we hope to support even more families. Join us by making a donation today!

See https://kat6a.org/empowered-grant/ for a full set of guidelines. Applications will be reviewed in order of receipt and granted based on full completion.



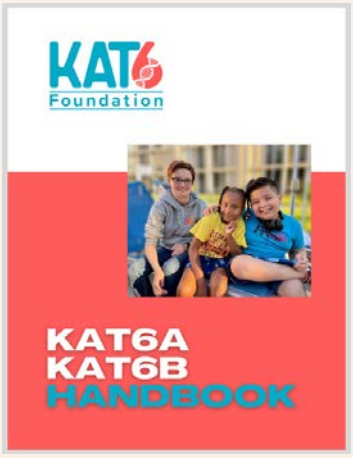
"With the Empowered Grant, we were able to purchase a Firefly GoTo Seat for Leo. This seat has made his life so much easier! He can sit at the table with everyone, go on the swings and his trike!"

KAT6 Advocacy

This year, the KAT6 Advocacy Group, has been very active, offering online presentations and workshops, and continues to work side by side with families on issues like guardianship and school advocacy.

Watch their Best Practices for Effective Education Plans presentation here!

Our team also compiled a comprehensive list of disability resources - available here.



The Foundation's KAT6 Handbook is now available in seven languages, including Arabic, French, Italian and Portuguese.

Our handbook continues to be a resource for parents, teachers, doctors and caregivers alike. Download your copy today!

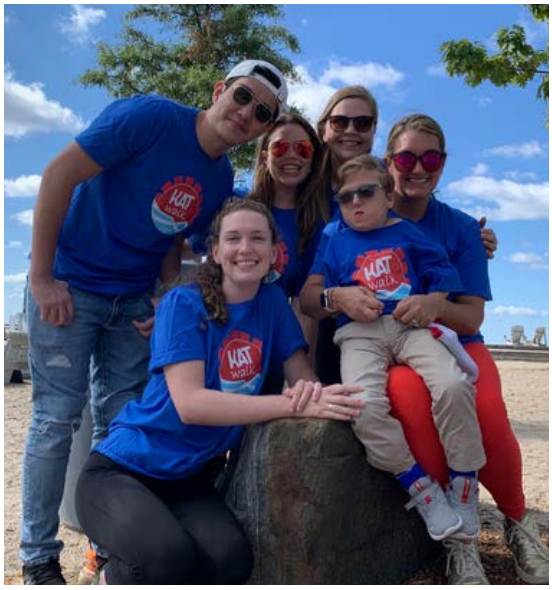
SUPPORT KAT6

We are incredibly grateful for all of the support we received throughout 2023.
As we look forward to 2024, we are eager to continue raising awareness, supporting families and driving research to better inform our patient community.

With the funding ending from the grant we received from CZI's Rare as One Network, it is critical that we increase our fundraising capacity to continue progressing as a Foundation.

How You Can Support Our Foundation

- **Donate to our *Annual Appeal* and YearEnd Giving campaign**
- **Support us on Giving Tuesday - the largest day of giving!**
- **Purchase KAT6 awareness merchandise from our *shop*.**
- **Ask your employer if they would consider supporting the KAT6 Foundation or enquire about a matching program**
- **Spread awareness on social media by sharing our posts.**
- **Support our *"I Care for Rare"* fundraising campaign by starting your own fundraiser!**
- **Join the KATwalk in 2024!**



Facebook Giving

Thank you to all of the individuals who showed their support via Facebook fundraisers this year.
2023 Facebook Donations Totaled **$6,793.70.**

Facebook fundraisers are a great opportunity to raise awareness among friends and family and folks who might not otherwise be familiar with our foundation. Creating a birthday fundraiser is a great way to raise money and awareness - every dollar counts!

Our Team

2023 Board of Directors

Jordan Muller, Board Chair

Emile Najm, CEO

Karen Ginsburg, Chair of Fundraising

Typhaine Lejeune

Myria Normann

Andrew Rankin

Maureen Martini

David A. Woodbury, Board Secretary

Kevin Young, Chair of Finance

Director of Science and Research
Dr. Natacha Esber

Director of Fundraising
Marjorie Weintraub

Registry Coordinator
Dr. Bhawika Lamicchane

Research Coordinator
Dr. Tanya Tripathi

Science Advisor
Dr. Angie Serrano

Website Coordinator
Aimee Reitzen

Registry Director
Amy Young

Advocacy Team Members
Susan Hartung, Director
Sue Carpenter
Beth Woodbury
Amy Young

KATwalk Chair
Katie Bator

Empowered Grant Coordinator
Jessica Wiemann

Social Media Coordinator & Fundraising Administrative Assistant
Megan Stetts

Dear KAT6 community:

We need YOU!

Did you know that our foundation is run by a very small group of committed parents? We have only part-time professional support for scientific research and fundraising.

The Board of the KAT6 Foundation is actively seeking new board members from the USA and internationally. They could be from within our support community or from without. We are especially seeking folks with experience with:

Pharmaceuticals, Scientific Research, Medicine, Marketing, Accounting, Fundraising, Media and PR, Graphic/Web Design, Communications.

The Board meets every other month. If you are unable to participate on the board, there are a number of committees within the foundation, available to join.

Please contact us at support@kat6a.org to join our team.

KAT6 Foundation

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