KAT NEWS

Volume 7 | December 2021 | KAT6A Foundation, Inc. | www.kat6a.org

Empowered Grant Update

In 2022, we are proud to expand the Empowered Grant program to include reimbursement of expenses for therapy treatment. This includes private speech therapy, occupational therapy, physical therapy, ABA therapy, feeding therapy, vision therapy, hippotherapy, aquatic therapy, music, art therapy and more.

Inside This Issue

PG. 2-3 Events Recap

PG. 4-8 Research

PG. 9-10 Assisting Families

PG. 11 Who We Are

PG. 12 Fundraising & Support

THE KAT6A FOUNDATION MISSION

Our mission is to support people and their families living with KAT6 disorders. It is imperative to raise funds to further research and identify possible treatments that could lead to a better quality of life. We aim to inform, raise awareness, and identify more individuals with KAT6A and KAT6B gene mutations. This enables researchers and clinicians the opportunity to discover more about these rare disorders and give our community greater consideration.

EVENTS RECAP

Rare Disease Awareness Day

On February 28th, "Rare Disease Awareness Day," we asked everyone to "show their stripes" and sold Rare Disease Day Awareness t-shirts.

The three different designs had a total sales of US$1,413.82

Webinar Series: Advocating for Answers

The virtual panel on September 23rd was moderated by Kristen Angell, Director of Advocacy for NORD, and discussed the challenges of rare disease care, treatment and advocacy, with a focus on KAT6A/B.

Topics included:

- Living with rare disease from the perspective of KAT6A families
- The successes and goals of the KAT6A Foundation as an emerging nonprofit focused on research for effective treatments
- The status of current research highlighting progress to date including projects funded by the Chan Zuckerberg Initiative grant. **WATCH HERE**

KATwalk 2021

KATwalk 2021 was our most profitable fundraiser to date. In total, we profited over **US$151,000** for our foundation. The funds will go towards expanding research on KAT6A/B gene mutations and will provide Empowered Grants for assistive equipment and technology to families and individuals with KAT6A/B. We want to extend a massive thank you to everyone who donated, walked, spread the word, and supported your loved ones with KAT6A/B in person or from a distance. Walks were held across the U.S. and virtually around the globe.

READ MORE AT KAT6A.ORG

HIGHLIGHTS

"Walk for Jack" raised the most funds (over $35,000) with the most donors (over 200!).

Team Hadley" held their walk in West Greenwich, Rhode Island, and had the largest turnout with over 120 walkers.

Virtual walks were held across the globe including in Australia, Canada, Israel and the UK. Team "Brynna the Brave" had boots on the ground in 16 US states.

WALK IN THE PRESS

Katwalk Raises Awareness for Rare Disease Affecting 2 Long Islanders
Long Island News 12

Local Boy Fights Rare Disorder
LIHERALD.COM | Bellmore

Nyack Family on Mission to Help Children with Rare 'KAT6A/KAT6B' Disease
Nyack News 12

RESEARCH

KAT6 Patient Registry

Provided by Bhawika Sharma

The KAT6A Patient Registry serves as an official database to track, record and understand the KAT6A/KAT6B mutations through the patient perspective.

For rare diseases like KAT6A and KAT6B, information about patient experiences is critical because it helps researchers and clinicians understand the diseases, ask better questions and seek better treatment strategies.

Our patient registry has **221** patients registered as of November 2021.

UPDATES

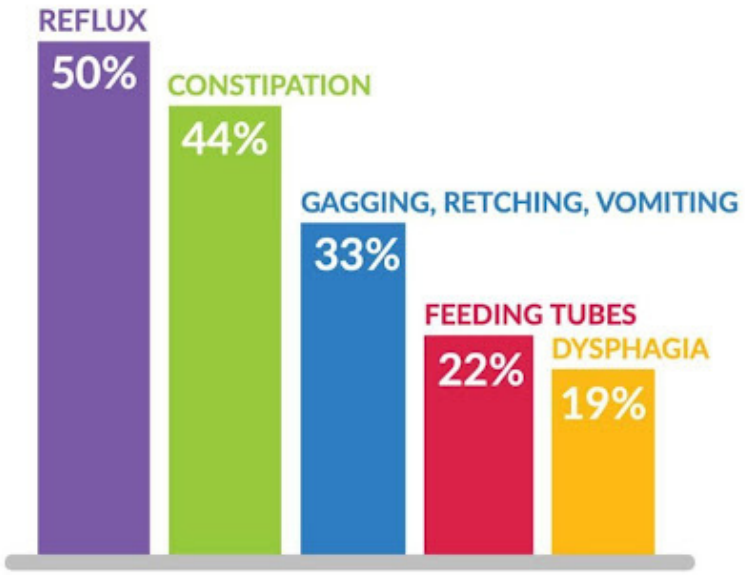
- The KAT6A Patient Registry is in the process of being renamed as KAT6A/KAT6B patient registry. This change acknowledges the presence of KAT6B families in the patient registry and reflects the Foundation's aim to be increasingly more inclusive to the KAT6B families.

- The KAT6A/KAT6B Patient Registry format is changing. We are going from 5 longer surveys to 12 short, digestible, easier to fill out surveys. We have also made edits on the questions to be more inclusive of KAT6B families and for patients from all backgrounds and ailments.

- The KAT6A Foundation has started analyzing patient registry data and sharing it with our patient families on the Facebook Support Group. **For the year 2022, we will work on the first publication using de-identified patient data coming out of the registry.** This is an exciting next step and a publication from the Foundation will be monumental in expanding our reach amongst researchers, patient families and fundraising entities.

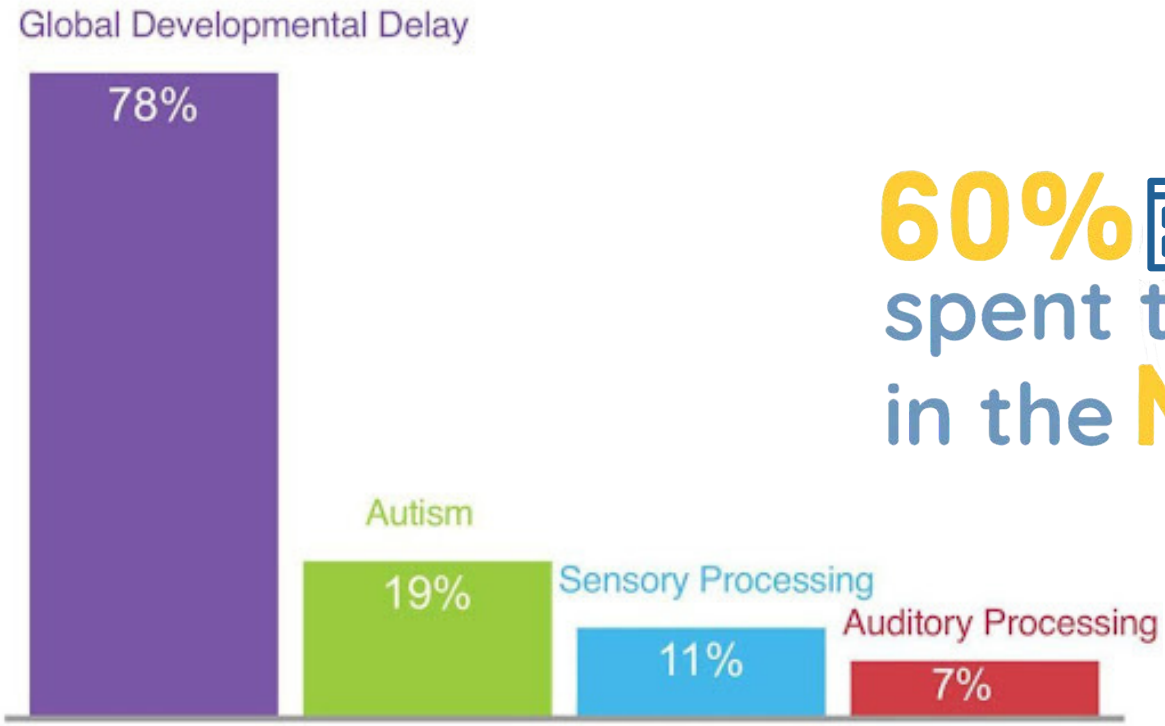
Stats Based On Registry Data, 2021

Most Common GI Disorders



64 %
Have high pain tolerance

Common Neurodevelopmental Disorders



60% spent time in the **NICU**

KAT6A/B Symposium

The 2021 KAT6A/B symposium was the first collaborative research event organized by the KAT6A Foundation. **It was designed to solidify the current KAT6A and KAT6B research network amongst clinicians and researchers through identification of research gaps, opportunities and collaborations.**

Over 45 participants attended the virtual meeting which included 16 speakers invited to present their research related to KAT6A or KAT6B genes.
The symposium had a duration of 2.5 hours and it was organized in three sessions:

- The first session was led by members of the KAT6A Foundation where topics such as **Collaborative Network Approach**, KAT6A/B registry and initiatives led by the foundation to raise awareness and strengthen research were discussed
- The second session had a series of five minute "flash talks" for 10 speakers to present their research on KAT6A/B related disorders
- The third session focused on speech and language in children with KAT6A related conditions a phenotype identified as a significant health challenge by the KAT6A/B community.

Patient-Led Research Collaborative Network Approach

Zuccato, Shilling & Fajgenbaum Emerging Topics in Life Sciences (2019)

2021 Published Research

KAT6A Research

First Case of Pan-suture Craniosynostosis Due to De Novo Mosaic KAT6A Mutation

Impaired Regulation of Histone Methylation and Acetylation Underlies Specific Neurodevelopmental Disorders

Sleep, Behavior, and Adaptive Function in KAT6A Syndrome

KAT6B Research

Impaired Regulation of Histone Methylation and Acetylation Underlies Specific Neurodevelopmental Disorders

Novel Variants in KAT6B Spectrum of Disorders Expand Our Knowledge of Clinical Manifestations and Molecular Mechanisms

Participants Needed for New Research Study

Dr. Rowena Ng, Ph.D is looking for participants for a new research study.
Dr. Ng's research team is completing a retrospective study that involves reviewing participants' past neuropsychological or psychological evaluations to determine the cognitive developmental pattern associated with KAT6A Syndrome. Currently, this area is not well published, and therefore determining appropriate early treatments can be challenging. These results will allow us to better understand the specific cognitive strengths/weaknesses seen among this syndrome and the differences found with the different gene variants, which in turn can inform more targeted interventions in early childhood.

If you are interested, please email Dr. Ng and she will follow-up with you with further instructions. This will largely involve you sending any of your child's previously completed neuropsychological or psychological evaluation reports so they can compile cognitive data across those affected by KAT6A mutation. Dr. Ng is happy to answer any questions you may have.

Rowena Ng, Ph.D. (#06263)
Pediatric Neuropsychologist
Department of Neuropsychology
Kennedy Krieger Institute

Assistant Professor
Department of Psychiatry and Behavioral Sciences
Johns Hopkins University School of Medicine
Email: ngr@kennedykrieger.org

Ongoing Research

Research Study at Universidad Pablo de Olavide Sevilla, Spain by Professor Jose Alcazar

Professor Alcazar is looking to firstly gain a fundamental understanding of the activity and potential role of mitochondria in fibroblast cells derived from the skin of 5 KAT6A patients with different mutations. Professor Alcazar and his team are studying fibroblast cell growth and the mechanics and workings of the mitochondria which is the engine room of every cell. Once an understanding of the basic cellular activity in KAT6A patients has been gained, and perhaps an indication of differences between individual patients and mutations, Prof Alcazar will look to evaluate individual biological elements that are active at the mitochondrial level in these fibroblasts. This will help identify the most effective components of mitochondrial cocktails currently in use for individual patients and potentially new therapeutic targets and approaches; leading us closer to scientifically proven and personalized medicines for KAT6A.

This open call is intended to specifically address disease mechanisms of neuro-developmental disorders in KAT6A and KAT6B syndromes. Year 1 funding from the KAT6A Foundation is US$52,650 (45,000 EUROS) and will provide additional years of funding if preliminary results show promise.

The KAT6A Foundation is in the Process of Submitting Two Proposals to RDMM in Canada

The Rare Diseases: Models & Mechanisms Network (RDMM) connects people discovering new genes in patients with rare diseases, and basic scientists who can analyze equivalent genes and pathways in model organisms. RDMM provides grants to fund projects that will allow rapid confirmation of potentially disease-causing genes, and fuel pilot studies to improve understanding of how specific gene mutations cause disease.

The KAT6A Foundation is working on submitting one proposal for the KAT6A gene mutation and one proposal for the KAT6B gene mutation. The focus of these 2 proposals is the development and validation of KAT6A and KAT6B research organisms suitable for neurodevelopmental analysis as well as the Identification of signaling pathways that might provide insights into therapeutic strategies.

ASSISTING FAMILIES

Empowered Grants

Gianna with her cooling vest which her family purchased with their Empowered Grant.

In 2021, we awarded **22** grants to KAT6 individuals living in **10** different countries. Caregivers received reimbursement for a wide range of therapeutic equipment including: tablets with communication software, adaptive bikes, feeding tools, physical therapy equipment, specialized strollers, standers, sensory swings and more.

In 2022, the KAT6A Foundation looks to support even more families with the expansion of this program to include reimbursement of therapies that parents pay for personally such as hippotherapy, music therapy, aquatic therapy and more.

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.

Francisco enjoys playing on his iPad purchased with his Empowered Grant. He uses different apps for work and play, and this technology helps him learn along the way.

KAT6A Medical Providers

Austria

Dr. Sara Baumgartner, A.Univ.-Prof. Dr. Daniela Karall, IBCLC Medical University of Innsbruck, Clinic for Pediatrics/Inherited Metabolic Disorders, Innsbruck, Austria.

France

Alain Verloes, MD, PhD, Chief of the Department of Medical Genetics, "CRMR Anomalies Développement & Syndromes Malformatifs et Déficiences Intellectuelles de causes rares", Robert Debré Hospital, Paris, France.

Iceland

Hans Tomas Bjornsson, MD, PhD, Clinical Director, Clinical Genetics, Landspitali University Hospital, Reykjavík, Iceland.

International

Richard Kelley, MD, PhD, Professor of Pediatrics at Johns Hopkins University, Baltimore, Md, Visiting Scientist at Boston's Children's Hospital, Boston, Mass.

United States of America

Valerie A. Arboleda MD, PhD, Founder of the Arboleda Lab at UCLA, Department of Pathology and Laboratory Medicine, David Geffen School of Medicine, UCLA, Los Angeles, Calif. Dr. Arboleda will arrange referrals to clinical genetic specialists at UCLA.

Jill Fahrner, MD, PhD, Assistant Residency Program Director at Johns Hopkins Genetic Medicine Residency Program and Assistant Professor of Pediatrics at Johns Hopkins Hospital, Baltimore, Md.

Jacqueline Harris, MD, MS, Assistant Professor of Neurology and Pediatrics Director, Center for Tuberous Sclerosis and Related Disorders, Kennedy Krieger Institute, Johns Hopkins Medical Institution, Baltimore, Md.

Dr. Anne O'Donnell, MD PhD, Epichroma Clinic Boston Children's Hospital, Boston, Mass.

Kenneth N. Rosenbaum, MD, Founder of the Division of Genetics and Metabolism, Rare Disease Institute, at the Children's National Medical Center, Washington, DC.

The KAT6A Foundation

Board of Directors

Karen Ginsburg

Typhaine Lejeune

Jordan Muller

Emile Najm

Andrew Rankin

Shelby Rau

Aimee Reitzen

Meg Salisbury

David A. Woodbury

Kevin Young

Technical Advisors

Natacha Esber, MD

Andrew Rankin, PhD

Kenneth N. Rosenbaum, MD

Angie Serrano, PhD

Chairperson
Jordan Muller

Chief Executive Officer
Emile Najm

Secretary
David A. Woodbury

Treasurer
Matt Salisbury

Follow Us On:

SUPPORT THE FOUNDATION

The KAT6A Foundation is incredibly grateful for all of the support throughout 2021. As we move forward into 2022, we're invigorated to continue assisting families, driving research, and inform, raise awareness, and identify more individuals with KAT6A and KAT6B gene mutations. **Wondering how you can help us continue our work?**

1. **Donate to our End-of-Year Annual Appeal via our website HERE**
2. **Select the KAT6A Foundation as your charity to support when you shop on Amazon via smile.amazon.com**
3. **Help us raise awareness by wearing your stripes for Rare Disease Day on Monday, February 28, 2022**

Other Ways To Support Fundraising

- **Purchase KAT6A and KAT6B awareness clothing and merchandise on our shop**
- **Start planning for KATwalk 2022**
- **Ask your company if they would consider donating to the KAT6A Foundation or enquire about a matching program**
- **Spread awareness on social media by sharing our stories and fundraising posts**
- **Join the KAT6A Foundation: KAT6A and KAT6B Research and Raising Awareness Group on Facebook.**
- **Hold a local fundraiser at a craft fair or community event**

Facebook Giving

Thank you to all of the individuals who showed their support via Facebook fundraisers this year.

A very special thank you to Courtney Teicher, who raised US$4300 with her Facebook initiative.

2021 Facebook Donations Total: US$20,885.30