KATNews

Volume 6 | December 2020 | KAT6A Foundation, Inc. | www.kat6a.org

Rare as One Grant Recipient

In January 2020, the KAT6A Foundation was chosen to become a part of the **Chan Zuckerberg Initiative's** Rare as One Network. Chan Zuckerberg Initiative (CZI) announced US$13.5 million in funding for their Rare as One Project, which is a group of 30 patient-led organizations that are accelerating research and driving progress in the fight against rare disease. Founded by Dr. Priscilla Chan and Mark Zuckerberg in 2015, CZI is "proud to support patient-led organizations as they pursue diagnoses, information, and treatment options in partnership with researchers and clinicians."

As part of the Rare as One Network, the KAT6A Foundation received a two-year grant totaling US$450,000. The grant's intention is to assist us in developing and launching collaborative **research networks** in partnership with clinicians and scientists. CZI provides us with the funding, tools, support and training needed to pursue our mission.

Inside This Issue

EVENTS RECAP

KATwalk 2020 Breaks a Fundraising Record

Our largest fundraising event of the year looked a bit different in 2020. In the past, we were able to hold large scale events in person in hopes that our KAT6A families from across the globe could attend. This year, we held a virtual KATwalk on October 3-4, and invited family and friends of individuals with KAT6A and KAT6B gene mutations to raise awareness. We encouraged our support system to register to walk under the team name of their loved one, donate online, purchase custom KATwalk2020 t-shirts, share our social media posts, and spread the word.

Families held individual walks in their communities across the globe. We saw an amazing turnout, raising an incredible amount of awareness for KAT6A/B.

KATwalk 2020 was our most profitable fundraiser to date. In total, we profited over US$45,000 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. During these uncertain times of COVID-19, we are humbled to have received such a large outpouring of love and generosity in support of our foundation.

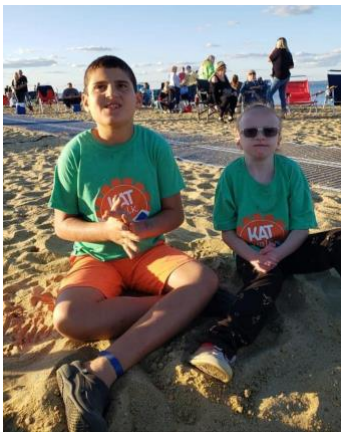
Check out a slideshow of KATwalk2020 highlights on our YouTube channel, linked **here!**

Walk for Jack was our top fundraising team for the third straight year!

Rüzgar walked with his family in Istanbul, Turkey!

Top Fundraising Teams

Walk for Jack	**$17,227.22**
Will's Warriors	**$4,777.00**
Ginger's Ninjas	**$2,620.00**
Team Benjamin	**$2,585.00**
Jackman's Crew	**$2,450.00**
Team Gianna	**$2,295.00**
The Tom Squad	**$2,155.00**
Freya's Cheer Crew	**$1,830.00**
KAT6A James	**$1,715.00**
Savannah strong	**$1,118.20**
Team Chase	**$1,085.00**



Peter and Savanah walked together with their families in New Jersey.

Ginger's Ninjas showed love all the way from Australia! Go Ninjas!

Team Benjamin (left) and Team Gianna (right) showed up strong in Michigan this year.

Three of our top fundraising teams walked in their hometowns in Minnesota. Below: Freya's Cheer Crew, Team Chase and KAT6A James.

Jackman's Crew showed up in a big way for their first walk in Paso Rablos, California.

In New York, The Tom Squad (left) and Will's Warriors (below) had a sunny day for their walks in Long Island.

Informational Webinars

We are proud of the partnerships we have formed with the medical doctors at the Kennedy Krieger Institute and are grateful for their continued support and look forward to coming together in-person again in the future. In place of an in-person 2020 KAT6A Clinic, several of our scheduled speakers presented webinars to members of our community via Zoom. Each webinar is available on the **KAT6A Foundation YouTube channel.**

September 12: **Genetic and Neurodevelopmental Features of the KAT6A syndrome by Dr. Harris, MD, Dr. Fahrner, MD, PhD and Dr. Smith, PhD**

September 16: **Importance of the Registry in Advancing Research by Suzanne Rossov, Senior Research Program Manager, NORD**

September 29: **Treatment of KAT6A – An Update presented by Richard Kelley, MD, PhD**

October 19: **When the School Bus Stops Coming by Susan Hartung, M.A. Ed in Special Education**

November 2: **Lysine acetyltransferases (KATs) in developmental disorders presented by Xiang-Jiao Yang, PhD**

December 3: **A Review on KAT6B Disorders by Gabrielle Lemire, MD, FRCPC**

December 8: **Special Financial Planning Considerations for Special Families by Stephen Ehrens, Wealth Advisor, CPA, CLTC, and Kevin Young, CFP.**

Stay tuned for more webinars in 2021. Check the Events page on www.kat6a.org often.

RESEARCH OPPORTUNITIES

KAT6A/B Patient Registry

The KAT6A Foundation and National Organization for Rare Disorders (NORD) Launched Natural History Study of KAT6A/B Syndrome

Research study is open to participants worldwide to advance understanding and treatments for the rare gene mutation causing KAT6A/B Syndrome.

One of the most important purposes of the KAT6A/B Patient Registry is to bring the KAT6A/B Syndrome community together and collect data which could be used to create therapeutics and improve the quality of life for patients. Some other goals of the KAT6A/B Patient Registry are to:

- Conduct a prospectively-planned natural history study that will result in the most comprehensive understanding of KAT6A/B Syndrome and its progression over time.
- Characterize and describe the KAT6A/B Syndrome population as a whole.
- Assist the KAT6A/B Syndrome community with the development of recommendations for standards of care.
- Assist researchers studying the pathophysiology of KAT6A/B Syndrome.
- Assist researchers studying interventional outcomes.
- Support the design of clinical trials for new treatments.

GET INVOLVED

What you want handy when completing the surveys:

- recent weight and height
- names of any publications the patient has been in
- genetic diagnosis (you have the option to upload a document or simply write out the diagnosis)
- medications & supplements (amount, usage, purpose)
- medical issues at birth
- any MRI or EEG results
- heart conditions
- digestive issues
- respiratory issues
- kidney and/or bladder problems
- hearing problems
- any allergies or immune system issues
- sleep disorders
-hospitalizations (when, length of stay, for what purpose)
- current therapy programs

Visit **https://kat6a.org/registry/** to begin today!
Currently we have 141 individuals registered.

For assistance email **kat6aregistry@gmail.com.**

2020 Published Research Articles

Deficient Histone H3 Propionylation by BRPF1-KAT6 Complexes in Neurodevelopmental Disorders and Cancer

Five New Cases of Syndromic Intellectual Disability Due to KAT6A Mutations: Widening the Molecular and Clinical Spectrum

The Key Roles of the Lysine Acetyltransferases KAT6A and KAT6B in Physiology and Pathology

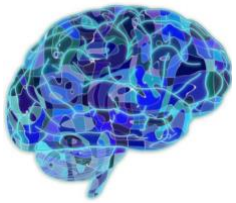
A Novel Pathogenic Frameshift Variant of KAT6B Identified by Clinical Exome Sequencing in a Newborn with the Say–Barber–Biesecker–Young–Simpson Syndrome

Further Delineation of the Clinical Spectrum of KAT6B Disorders and Allelic Series of Pathogenic Variants

KAT6B Disorders

KAT6B Genetic Variant Identified in a Short Stature Chinese Infant: A Report of Physical Growth in Clinical Spectrum of KAT6B-Related Disorders

Participants Needed for Investigation of Behavior and Sleep in KAT6A



This is a survey-based study. No travel or in-person visits at this time. Contact Clay Smith, MD, at **smithcl@kennedykrieger.org** or Jennifer Johnson at (443)923-2746, **JohnsonJen@kennedykrieger.org.**

KAT6A Foundation Funds New Research Study in Sevilla, Spain

Summary of a Research Proposal from Professor Alcazar and Universidad Pablo de Olavide, Sevilla, Spain for The KAT6A Foundation

by Andrew J Rankin, PhD.

The clinical features in KAT6A syndrome are highly variable across patients with basic characteristics of intellectual disability, speech delay, microcephaly, cardiac abnormalities and gastrointestinal complications amongst others. Furthermore, it is unknown whether specific mutations may lead to different degrees of impact on individual organ systems and clinical features. The long-term goal which could stem from this research proposal is to understand if a personalized therapy approach could ultimately be applied based upon the disruption of the normal working of the cell and mitochondrial activity. Identifying if different mutations or interindividual genetic variation can contribute significantly to how each patient expresses clinical features of KAT6A and ultimately how each patient could respond to individual components of mitochondrial active elements currently in therapeutic cocktails or even identify new targets for drug treatments. The goal of personalized medicine is then to maximize the likelihood of therapeutic efficacy and minimize the risk of drug toxicity for each individual patient.

Recent hypotheses (Dr. Richard I. Kelley, Kennedy Krieger Institute, Department of Pediatrics, Johns Hopkins Medical Institutions) suggests that mutations in the KAT6A gene secondarily affect mitochondrial function and that biologically active elements (such as carnitine and vitamin B5) that act at the mitochondrial level may potentially modulate the clinical features of the disease.

In this project, Prof Alcazar of Universidad Pablo de Olavide, Sevilla, Spain proposes to firstly gain a fundamental understanding of the activity and potential role of mitochondria in fibroblast cells derived skin fibroblasts donated from KAT6A patients with different mutations, 5 of our KAT6A patients in the first instance.

Prof Alcazar and his team will study fibroblast cell growth and the mechanics and workings of the mitochondria which is the engine room of every cell. For those interested in the science and details, he we will use sophisticated and proven scientific techniques, familiar expertise in his laboratory, such as the enzymatic activities of the mitochondrial respiratory chain, the levels of coenzyme Q10, the expression levels of mitochondrial proteins, the mitochondrial membrane potential, and the activation of mitophagy and / or apoptosis.

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 derived from KAT6A patients. 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.

Participate in a KAT6A Foundation Crowdsourcing Project

Parents and guardians of KAT6A/B individuals, we need your assistance. Help us gather information that will allow us to identify and prioritize high-impact research questions that will have the potential to improve KAT6A/B patient health and quality of life.

Click here and participate in a 3-question survey.

IN THE PRESS

Jack Nicklaus, Barbara Nicklaus, Lily Jaeger

Lilly, a Cover Girl

The Jaeger family was honored to be invited as special guests to Creighton Farms Invitational Golf Tournament in 2019. Barbara Nicklaus, the wife of golf great Jack Nicklaus spoke about the Genomics Program at Nicklaus Children's Health Care Foundation (NCHCF) in Miami. Through their collaboration with RADY San Diego, they were able to give the Jaeger family answers via whole exome sequencing, which is technology that changed their lives. Christy Jaeger says, "the relief of having a diagnosis of KAT6A, of seeing that list of characteristics that read like a checklist of Lilly's symptoms is not easily described."

This month, because of their involvement with NCHCF, Lilly was featured on the cover of their 2019/2020 issue of **Fore the Children**. Christy says that the family was honored to be chosen to represent the thousands of children that NCHCF helps.

Read more about Lilly's journey on the **KAT6A Foundation Blog.**

"Debating with Children" Radio Program in Spain

Each week, children are invited to speak about a topic or present personal stories on a radio program in Spain. Victoria and her mother had an interesting experience visiting the studio for a live program. They had the chance to discover a new atmosphere and were shown warmth from the radio staff. The first part of the program was centered around information about the KAT6A Foundation, and the second part focused on Victoria's rare disease and personal life. Victoria's mother spoke about the foundation and the great support and sense of belonging that she feels from other KAT6A families across the world, as well as Victoria's improvements since birth and how she surprises her family all the time.

Cincinnati Children's Hospital Committed to Rare

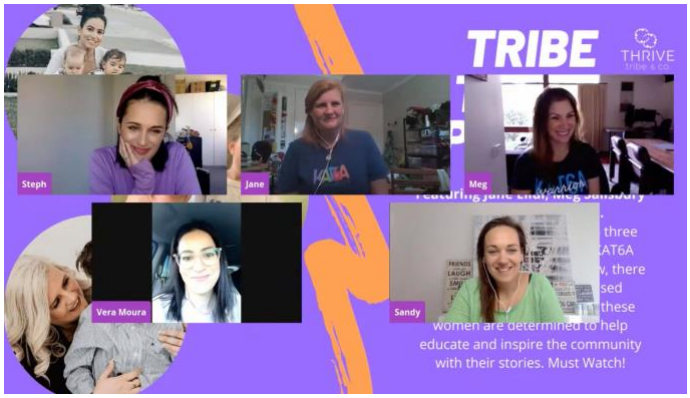
Elena, age 6, was diagnosed with KAT6A Syndrome in 2019 after being adopted from China. Recently, she was featured in an article titled, **"Diagnosis of Rare Disorder Leads to Even More Progress for Elena,"** which is available on the website for Cincinnati Children's Hospital Epigenetics Clinic.

Camden Featured in Local News

Camden was the 80th child diagnosed with KAT6A syndrome. His parents work hard to support the KAT6A Foundation's mission to raise awareness. Recently Camden's father, Stephen, decided to contact his local news outlets to share their unique journey in hopes of inspiring other families living with a rare disease. Check them out on **Wink News TV,** and read about their journey in the article, **Cape Coral Child Only the 80th in the World Diagnosed with Rare Disorder.**

Three Mums Team up for a Podcast in Australia

Jane Ellul, Vera Moura and Meg Salisbury met with Thrive Tribe & Co founders Steph Wicks and Sandy Golder as part of their Tribe Talks series. Jane, Meg and Vera did a great job of raising awareness for KAT6A syndrome and The KAT6A Foundation by discussing their personal journeys and educating on the various characteristics of KAT6A Syndrome. Check them out on **Tribe Talk Episode #6.**



From Global Delay to Literary Award

Diane Slonim, PhD, CCC-SLP was introduced to Peter Najm in 2016 when his family came to her for a thorough speech evaluation. Slomin's speech-language pathology work with Peter has improved his behavioral issues and has given his intellectual abilities an opportunity to shine. Slomin declares, "What makes for a rewarding career in speech-language pathology? An open heart and a curious mind."

Read their story published in the October journal of **The American Speech-Language-Hearing Association.**

View more media stories at **www.kat6a.org/press/**

FUNDRAISING

KAT6A Foundation Earnings

Total: US$93,198.46

Jan 1, 2020 – Dec 15, 2020

Ways to Support Fundraising:

- Hold a Birthday Fundraiser on Facebook
- Shop using **AmazonSmile**
- Purchase KAT6A and KAT6B awareness clothing and merchandise on our **Spreadshirt Shop**
- Start planning for **KATwalk 2021**
- 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

A special thanks to the following individuals for showing support in 2020 via **Facebook fundraisers**:

Hayley Albert, Andrea Goes, Terri Fallis, Anada Filipitsch, Oumou Fofana, Matt Goes, Kimberly Harvey-Chase, Brittney Hochhalter, Simone Kabelitz, Jessica Lafferty, Ema Langi-Colon, Carol Langley-Hensley, Amanda Marie, Danielle Mineo, Sonia Nations, Sandra Neville, Stacie Norris, Anna-Louise Peschel, Tina Schoenike-Schriber, Dale Serfass, Traci Starn, Ashley Trowbridge, Andre Voor and Yin Yoga Solidario/Pilar Jaen.

2020 Facebook Donations Total: US$19,377.32

(Note: Facebook pays out donations at the end of each month, therefore December donations will be reflected in the 2021 earnings report.)

Bonfire T-Shirt Fundraiser

This year we raised **US$1,026.86** in our annual t-shirt fundraiser for Rare Disease Day awareness.

Holiday Fundraiser

Thanks for your support of our inaugural holiday wreath/decor fundraiser from Gift-it-Forward. We sold 14 items and raised **US$112.** Every little bit helps us achieve our mission.

CALENDAR OF EVENTS

Empowered Grant

Application Period: Now - Jan 31, 2021

Details and requirements are available on https://kat6a.org/empowered-grant/

Email KAT6Aempowered@gmail.com with any questions or to request a translated application.

T-Shirt Fundraiser

January 2021

Stay tuned for a link to purchase a new design to wear on Rare Disease Day. This year we will be selling a KAT6B option to accompany our KAT6A design.

Rare Disease Day

February 28, 2021

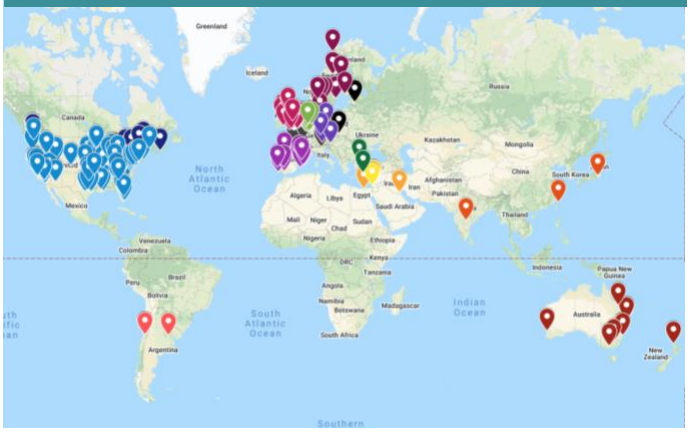
Wear your stripes and KAT6A Foundation clothing on this day to raise awareness for our very rare community. Last year we had several families advocate for RDD in their communities, schools and even in the press. Be sure to look for our specialized photo frame on Facebook and use it on your profile picture.

The KATwalk

October 2-3, 2021

Save the date for our annual fundraising and awareness event. More details to follow.

231 Families Join Our World Map



VOLUNTEERS NEEDED!

We are looking for individuals to assist in the following areas:

- Writing grants
- Fundraising
- Event planning
- Newsletter
- Translating informational resources
- Contributing to our Blog on KAT6A.org
- Raising awareness in the press

SUPPLIES NEEDED!

We are compiling a list of durable medical devices and gently used speech aids that your children have outgrown so that these items can be passed on to other KAT6A/B individuals. Please contact Lindsey Geiger via Facebook messenger if you can help support this cause.

Please like/follow/subscribe to us on social media.

FAMILY SPOTLIGHT

Korra's KAT6A Journey

Korra is a child who radiates positivity and joy. She is one of our Empowered grant recipients, which she used to a purchase a new iPad with communication software. We recently spoke with her mother, Miri, about Korra's inspiring journey with KAT6A. Through this Q& A session, we received some insight on her family's process of learning her diagnosis, and how they have adapted since. We believe the following Q&A will resonate with our support group.

Q: Please tell us a little bit about your family and Korra.
A: Korra is such a happy kid. She is three and a half years old, and was diagnosed with KAT6A in September of 2017, when she was five months old. She has a large group of adults in her life who love and care about her. She has two sets of parents and many grand and great-grandparents. She has an older sister who is her best friend. Korra loves to play doctor and dance. She loves baby dolls, and recently went roller skating for the first time, and didn't want to stop.

Q: How did you feel about the process of Korra being diagnosed, from beginning to end?
A: It was a long and stressful journey, and we spent the first five months of her life in and out of the hospital with no real answers. Having a diagnosis was a relief, regardless of the challenges that we now knew she would face.

Q: What were some things that your doctor told you upon the diagnosis?
A: They gave us links to the KAT6A Foundation website and Facebook group, and told us that they really didn't know a lot about the disorder.

Q: In what ways has navigating KAT6A syndrome changed your life?
A: Korra is just Korra, and she wouldn't be the same without her disorder. She is perfect and wonderful, and we wouldn't change anything about her if we could. One thing that has changed is our perspective. We have a greater appreciation for healthcare workers and therapists and celebrate accomplishments that we previously took for granted.

Q: What are some tasks that are difficult for Korra, and how does she overcome those difficulties in her everyday life?
A: Korra is still learning how to talk. With the help of therapists and her speech device, she has made great strides in speech and language.

Q: What is one accomplishment that Korra has achieved that stands out to you?
A: Despite the time she spent in the hospital early in life, Korra continues to develop and flourish. She works hard to accommodate for the things that are harder for her and has an amazing sense of humor.

Q: What are some resources that have helped you over the years with questions/concerns relating to KAT6A/B?
A: The KAT6A foundation Facebook page has been a great resource, and an encouraging group of people who understand the struggles we face as a family. The KAT6A clinic has been such a wonderful place to learn firsthand what other families with similar struggles look like. First Steps in our state helped Korra for the first three years of her life with in home therapies before she transferred to her school. Our local children's hospital has literally saved her life and been an unwavering support through our journey.

Q: What is one piece of advice you'd give someone whose family member was just diagnosed with KAT6A/B?
A: Don't be afraid to ask for support. Parenting a disabled child is hard, but so rewarding. Allow your child to create their own path, and don't set expectations for them. They will surprise you.

FOUNDATION UPDATES

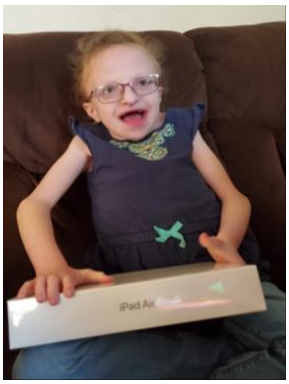
We Officially Support People and Their Families Living with KAT6B Related Disorders

We are happy to welcome KAT6B families to our support system! KAT6B and KAT6A syndromes have many similarities, and are considered a part of the same overarching family. We want to provide support to families coping with KAT6A and KAT6B disorders, as well as increased awareness of both across the globe. KAT6B families can find a list of KAT6B research articles on our website linked **here** and join **our KAT6A and KAT6B Research and Raising Awareness Group** on Facebook.

KAT6A Foundation Supports Families through the Empowered Grant

In response to the global pandemic, the KAT6A Foundation established the Empowered Grant to support individuals diagnosed with KAT6A syndrome and KAT6B related disorders. Empowered grants provide individuals with therapeutic equipment and technology so that parents can support their child's growth and development at home. Last June, the KAT6A Foundation funded 13 Empowered Grants totaling US$7000. Families from seven different countries were reimbursed up to US$600 for iPads with communication software, therapy swings, orthopedic shoes, therapy tables and more. We look forward to rewarding more Empowered grants in 2021.

Applications are open now. **The deadline to apply is January 31, 2021.**



New Website Made Possible through CZI Funding

We have a wonderful new logo, awareness ribbon and an upgraded website thanks to funds obtained through our CZI grant award! All information on **www.kat6a.org** has been updated including new faces in our photo gallery and donation software that accepts various currencies. New pages include: About KAT6B, KAT6B Published Research, Newsletters, Family Resources, Press, and Volunteer. At the KAT6A Foundation, we strive to make information meaningful and accessible for families, and valuable to medical doctors/therapists treating our children. Use Google Chrome to access our website in your native language.

KAT6A FOUNDATION

CZI Celebrates Immigrants in Science

Our founder, Emile Najm and his wife Dr. Natacha Esber were featured in a **video** by CZI highlighting immigrant scientists such as themselves. Esber says, "the global approach coming from a different country, helps a lot in research with a different point of view. It's like having a second opinion for science and research."

It is with great sadness that we share the news of the passing of **Dr. Susan Marie Hull**, an advocate and educator for the KAT6A community. Rest well, Dr. Hull, and thank you for the work you've done on our behalf.

WELCOME

Angie Serrano, PhD

Dr. Angie Serrano is the newest science advisor to the KAT6A Foundation. Dr. Serrano is a postdoctoral fellow in H. Joseph Yost Lab at the University of Utah. Presently, she is researching Kabuki Syndrome (KS) using a zebrafish genetic model and human iPSCs-derived brain organoids in order to understand the key biological processes that trigger cardiovascular and neurological defects in KS patients.

After spending more than a decade working in basic science, Dr. Serrano has learned that what truly drives scientific discovery is not only based on sound research and efficient use of state-of-the-art technologies but also the connection and understanding of the very people you represent and work with.

Dr. Serrano was recently awarded a 2020 Warren-Alpert Distinguished Scholar and is currently an advocate for bridging the gap between rare disease research, healthcare providers and patients.

Yan Campbell, PhD

Dr. Yan Campbell joined the KAT6A Foundation earlier this year as the new KAT6A Patient Registry coordinator. She is currently a Clinical Trial Risk Management Lead and Global Strategic Intelligence Manager in the pharmaceutical industry.

She has a great love for children, especially after becoming a mother. She says, “if I can make only one single wish, I would wish that there are no more sick kids in the world.” Campbell received her PhD degree in Biochemistry and Biophysics and master’s degree in Business Administration. She is knowledgeable in molecular biology, drug development, dietary supplements, as well as strong interests and experience in clinical studies. She says that she is always willing to push her boundaries a little further in order to help more patients out there.

WHO WE ARE

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

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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.