2022 Annual REPORT

KDM5C

KARES FOUNDATION

PRESENTED BY

KARES BOARD OF DIRECTORS

info@kares.foundation

kares.foundation

OUR STORY

In 2017, when their daughter, Gabby, was 10 years old, The Robl family finally received a diagnosis that explained her symptoms and challenges: a genetic variant that disrupted the function of the KDM5C gene.

Amy Robl searched online for other parents and families affected by this rare syndrome, but couldn't find much support. That's when Amy created the KDM5C Support Group on Facebook, which now has hundreds of members representing approximately 100 diagnosed individuals worldwide.

In 2020, a group of families and researchers met up for the first time at the Rare Disease Day conference at Albert Einstein College of Medicine. It was inspiring to see families, clinicians, and scientists come together in support of KDM5C research for the first time. There continues to be strong collaboration amongst these groups and a growing volume of research.

As the Facebook support group continued to grow, several families came together with the idea of creating a Foundation to improve the lives of individuals living with KDM5C-related disorders. The KARES Foundation was born in May of 2022, a nonprofit 501(C)(3) dedicated to KDM5C advocacy, research, education, and support.

ABOUT KDM5C

KDM5C is a protein that controls the expression of a number of genes that are involved in a range of cellular processes. It is expressed in most cells of the body but is particularly important in neurons, where it is needed for developing neural networks.

Genetic variants that disrupt the KDM5C gene cause Claes-Jensen syndrome, which is characterized by an array of developmental and cognitive changes. These genetic variants can range in severity, some leading a small change to the KDM5C protein, while others result in no KDM5C protein being produced.

DNA changes within the KDM5C gene can occur within the affected individual (de novo) or can be inherited via the X-chromosome.

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CHARACTERISTICS

KDM5C genetic changes are associated with a range of symptoms.
Some of the most commonly reported characteristics include:

- Altered muscle tone
- Attention Deficit Hyperactivity Disorder
- Autism
- Challenging behavior
- Developmental delay
- Epilepsy
- Gastrointestinal issues
- Intellectual disability
- Motor delays
- Ocular findings
- Short stature
- Speech impairment

There are vast differences in the ways individuals with KDM5C genetic changes are impacted. Symptoms affect both males and females to various degrees of severity.

A KDM5C variant is diagnosed through genetic sequencing or whole exome sequencing, which has only started to become widely accessible in recent years. This means there are likely to be many undiagnosed adults and children living with KDM5C-related conditions.

"Stay patient, let the joy and grief coexist, but always love and appreciate your child for the perfectly imperfect person they are. They shouldn't change to fit in the world; the world should change to accommodate them!"

JULIAN'S MOM

OUR MISSION

Our mission is to improve the lives of those affected by KDM5C genetic variants through advocacy, research, education and support for affected individuals and families.

KARES FOUNDATION

OUR VISION

We envision a world where patients and families affected by KDM5C genetic variants are able to achieve their highest attainable health in order to thrive in their communities.

CORE VALUES

HOPE

We believe in a positive outlook for the future. We maintain ambitious goals for improved quality of life for those affected by KDM5C variants. We recognize that our challenges and trauma can coexist with joy and hope.

INNOVATION

We believe in tapping into the best resources to cultivate new skills, opportunities, therapies, and more. We are committed to utilizing technology to improve our organizational structure and research practices, pushing the limits to bring positive impact to our KDM5C community.

COMMUNITY

We collaborate with KDM5C patients, families, friends, caregivers, clinicians, researchers and partner organizations to build relationships that maximize our impact. We strive for all people to feel a sense of inclusion and belonging in our community.

INTEGRITY

We conduct our work with honesty and transparency. We demonstrate financial stewardship of the resources entrusted to the KARES Foundation. We put relationships first, prioritizing trust and privacy in all interactions with our community.

KARES FOUNDATION

KARES

OUR PILLARS OF WORK

ADVOCACY

We advocate for inclusion within our communities. We encourage KARES leadership and families to use their voices to create positive change in rare disease policy and funding.

RESEARCH

The KARES Foundation strongly encourages interdisciplinary collaboration and the sharing of resources as a means of accelerating progress in KDM5C research. Research funded by the KARES Foundation is for public use in an effort to educate and promote further discovery.

EDUCATION

We aim to empower KDM5C families and caregivers with the tools and resources to navigate the challenges of living with a KDM5C-related disorder. We build the capacity of staff and professional communities (educators, doctors, therapists) through ongoing education and professional development opportunities.

SUPPORT

We provide a safe space for exchange and support amongst KDM5C families and caregivers. We partner with other organizations and professionals to support the ever-changing needs of our community.

"You’re the only expert on your kid. Be the best advocate you can be and that’s all you need. When times are tough, remember it's not your kid’s fault. Your fight is with KDM5C. That’s the enemy. Use other families' experiences with KDM5C [genetic variants] to help your kiddo.”

JOSH'S DAD

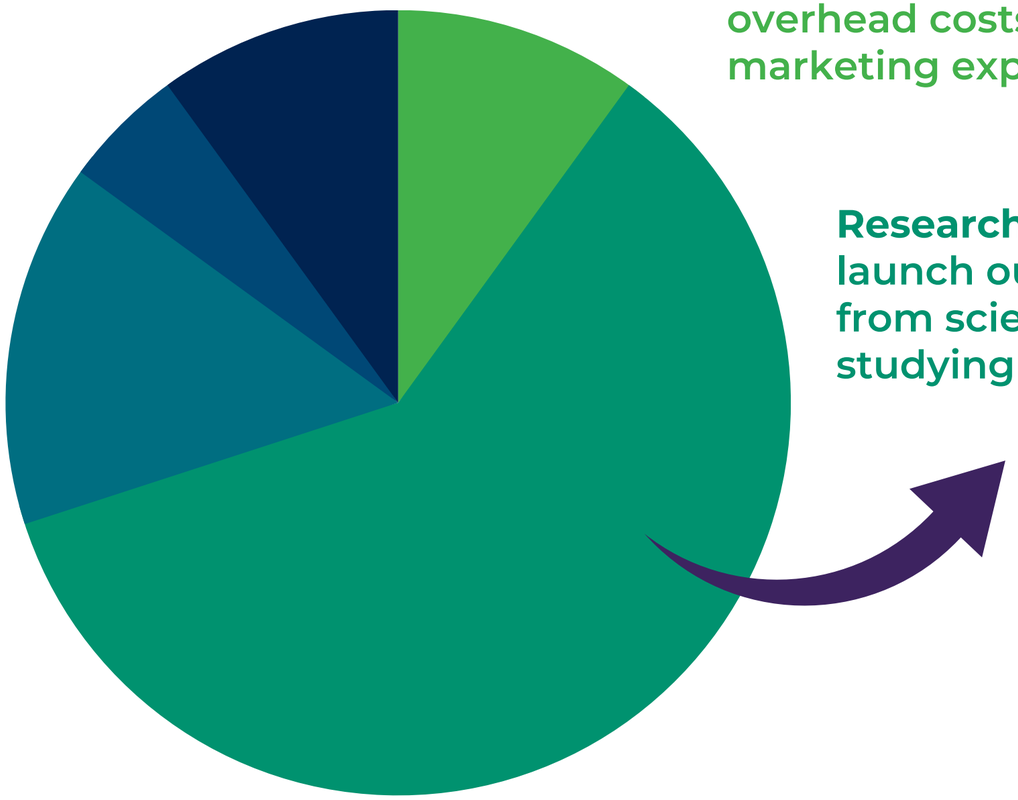
WHERE YOUR DONATION GOES

OF ALL DONATIONS RECEIVED IN 2022...

Savings: (10%) stays in our reserves for future programs and projects

Administrative Costs: (10%) goes to overhead costs such as website and marketing expenses for our events

Education: (5%) allows our volunteer staff to learn more about rare disease research & advocacy



Research: (60%) allows us to launch our first Call for Proposals from scientists interested in studying KDM5C

Family Support: (15%) provides tools & resources families need to better the lives of their loved ones, including our family education series

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*Percentages are approximate

BOARD OF DIRECTORS

AMY ROBL

Cofounder, Board Chair

Amy and her husband Chris own and operate a general contracting business in Washington state, where they live with their two daughters, Gabby and Ashley, and yellow lab, Newey. Gabby is affected by a KDM5C variant and was diagnosed at age 10. Amy created the Facebook Support Group to bring KDM5C families together in 2018, and she has been an integral part of the formation of the KARES Foundation in cooperation with families and researchers.

LARA EREKSON

Cofounder, Vice Chair

Lara lives in Idaho with her husband Jeff and daughter Whitney, and their miniature schnauzer, Milo. Whitney was diagnosed with a KDM5C variant in 2019 when she was 13. Lara's oldest daughter Savannah, is currently studying Music Therapy at Utah State University. Lara is passionate about advocacy, inclusion, and spreading KDM5C awareness. As the Fundraising Committee Chair, she helped implement KARES first successful Virtual 5K Run, Walk and Roll. She also serves on the Family and Community Engagement committee.

AMANDA LOWRY

Cofounder, Treasurer

Amanda lives in Illinois with her husband Jerry, and their children, Jack, Owen, and Ella. Jack was diagnosed with KDM5C disorder when he was 7. Amanda acts as the Treasurer for KARES and Chairs the Finance Committee. Amanda is a CPA and CHFP and works as the Vice President-Controller for OSF HealthCare System.

MELISSA MCNEILLY

Cofounder, Secretary

Melissa lives in North Carolina with her husband, Steven, and three kids, Julian, Javi, and Emi. Julian was diagnosed with a KDM5C change in 2020 at age 4. Melissa works in nonprofit Digital Marketing and is a strong advocate for diversity, equity, and inclusion. As Marketing Committee Chair, she manages KARES' website and social media, including the Family Spotlight.

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BOARD OF DIRECTORS

HEATHER COFER

At-Large Board Member

Heather lives in New York with her husband, Dave, and their two children, Natalie and Porter. Porter was diagnosed with KDM5C disorder in 2015 when he was 2. Heather has been a part of the Facebook Support Group since its establishment, and her family attended the in-person Rare Disease Day in 2020.

COREY MOSS

At-Large Board Member

Corey lives in Austin, Texas with her husband, two sons, Brian and Austin, and boxer and Great Dane pups. Corey works in technology as a Content Management specialist. She enjoys outdoor activities and is a green belt in Tang Soo Do. Austin was diagnosed with a KDM5C change in 2021 at age 3. Since, Corey has been active on the Facebook group and passionate about spreading awareness of this rare disorder.

JULIE SECOMBE

At-Large Board Member

Scientific Advisory Board Chair

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SCIENTIFIC ADVISORY BOARD

JULIE SECOMBE

SAB Chair

Professor of Genetics and Neuroscience at the Albert Einstein College of Medicine in New York, as well as a member of the Rose F. Kennedy Intellectual and Developmental Disabilities Research Center.

Obtained her B.Sc and PhD at the University of Adelaide in her native country of Australia. She then did postdoctoral research at the Fred Hutch Cancer Center in Seattle before beginning her faculty position at Einstein in 2009. Dr. Secombe's research focuses on understanding how the KDM5C protein regulates gene expression programs that are critical for neuronal development and function.

SHIGEKI IWASE

Member

Completed his B.Sc and PhD at the University of Tsukuba in Japan. He then completed his postdoctoral training at Harvard Medical School in Boston in 2012.

Dr. Iwase now serves as an Associate Professor in Human Genetics at the University of Michigan in Ann Arbor, where he also carries out his research. His work focuses on chromatin dynamics in health and disease in the brain. In particular, Dr. Iwase and colleagues discovered that the human KDM5C gene encodes an enzyme that removes methyl marks specifically from histone H3 lysine 4, and this enzymatic function is broken in human KDM5C disorders.

JULIAN MARTINEZ

Member

Dr. Martinez is a pediatric geneticist at UCLA. Dr. Martinez received his PhD in 1999 and MD in 2000 from Yale School of Medicine, then carried out his residency at UCLA and his fellowship at Cedars-Sinai Medical Center in 2005.

He became a Clinical Instructor and Chief Resident of the UCLA Intercampus Medical Genetics Program in 2005. Dr. Martinez has a clinical interest in genetic syndromes and his research interests include understanding the pathways that govern progenitor and stem cell maintenance.

MARIA MIANO

Member

Dr. Maria Giuseppina Miano is a Senior Researcher of National Research Council of Italy and Head of the Human Neurogenetics Laboratory at the Institute of Genetics and Biophysics "Adriano Buzzati-Traverso" in Naples (Italy). She obtained her BSc and PhD at the University Federico II Naples and Specialty Degree in Medical Genetics at the University La Sapienza in Rome. She completed the research training at Medical Research Council-Human Genetics Unit in Edinburgh (UK).

Her studies focus on the identification of convergent disease pathways damaged in brain diseases. Particularly, she established that KDM5C is an epigenetic regulator at the intersection of transcriptional axes involved in multiple neurodevelopmental disorders.

MARIAM REBOLLAR

Parent Liaison

Mariam lives in Los Angeles, California with her husband, Ivan, and son Nicholas. Mariam is a former Dentist from Cuba, now practicing as a Dental Hygienist. Mariam's son Nicholas is affected by a KDM5C variant and was diagnosed at the age of 2. She is a strong advocate for research and the importance of collaboration between families, researchers and health care providers. Mariam is the Parent Liaison to the Scientific Advisory Board and a member of the Finance Committee.

KARES FOUNDATION

OUR ACCOMPLISHMENTS TO DATE

The KARES Foundation was founded by a group of parents in 2022, and launched with the support of other parents, family members, and key members of the medical and scientific community. There has been significant progress planning and collaborating with key stakeholders to create a framework for success over the last year.

KEY ACCOMPLISHMENTS

RARE X

KDM5C - Data Collection Program

- **Formed in May 2022** and developed governance structure and organizational policies.
- Engaged ~500 members in a growing **Facebook Support Group.**
- Held a **KDM5C Research Update Symposium** in 2023 bringing together researchers, clinicians and patient families from all over the world.
- Partnered with RareX to build a **patient data collection program** for the KDM5C community.

KEY ACCOMPLISHMENTS CONT.

- Held a successful **virtual 5K event** with 250 participants in 17 US states raising $60,000.
- Built website and social media infrastructure with cohesive branding guidelines.
- Launched a **family education series** to provide information and support.
- Held a **Strategic Planning** meeting.
- Launched a **Research Grant Program** to support long-term therapeutic targets for KDM5C genetic variants.

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KEY ACCOMPLISHMENTS CONT.

- Launched a **volunteer internship program** that allows high school and college students interested in rare disease studies to lend their time and talents to KARES
- Established a **KDM5C Awareness Day** on June 5

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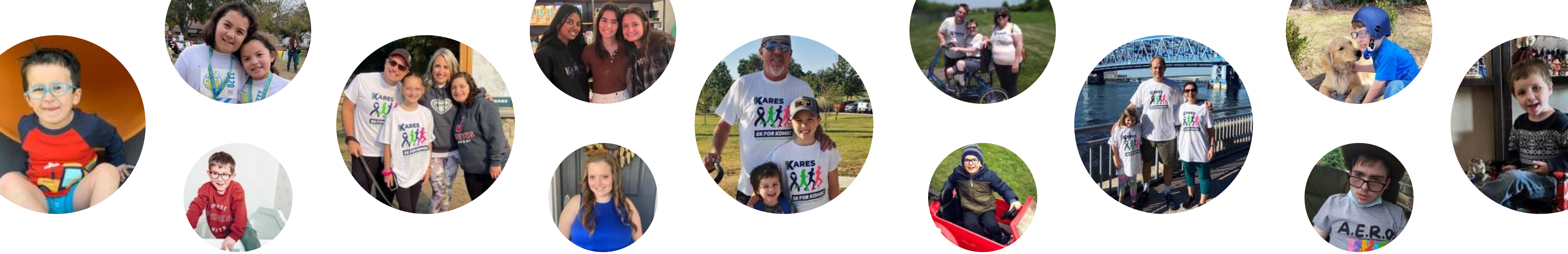
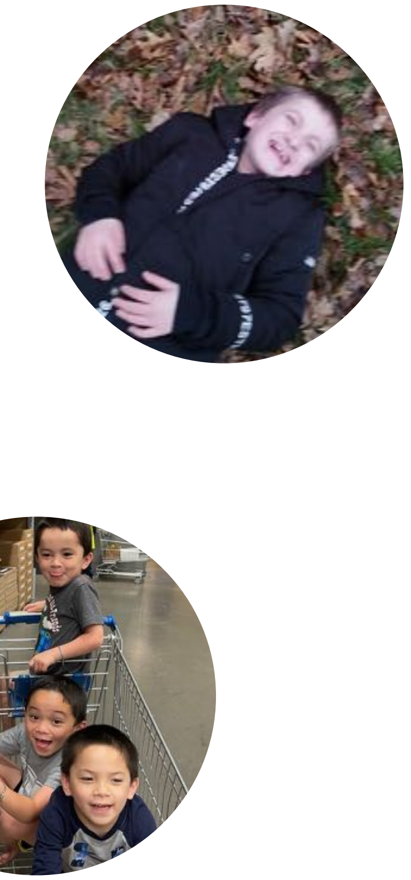
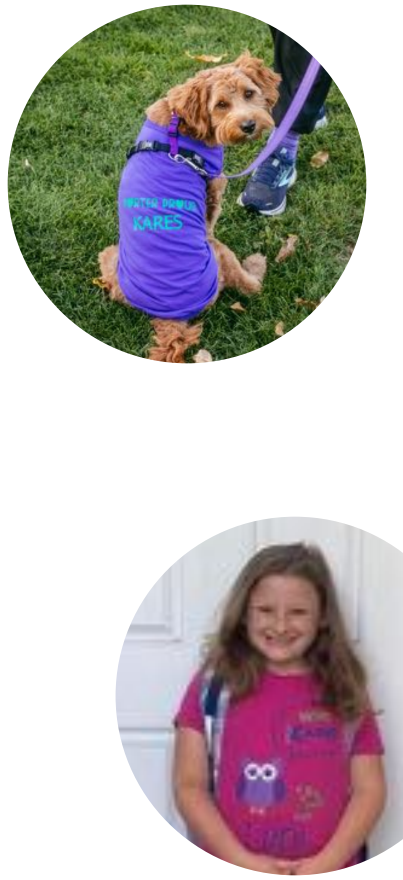




Thank You!

On behalf of our team at the KARES Foundation, we would like to thank our volunteers, leadership, partners, researchers, clinicians and interns for their help in advancing the work of our organization. We are so grateful to have created such a supportive and inclusive environment that all families can access when they receive a KDM5C-related diagnosis.



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