2023 ANNUAL REPORT

KDM5C related genetic disorders

PRESENTED BY

KARES BOARD OF DIRECTORS

info@kares.foundation

kares.foundation

KARES FOUNDATION

BY THE NUMBERS

59
Individuals enrolled in RARE-X

905
Contacts in our database

$30K
First seed grant given for KDM5C research

647
Facebook Support Group members

9
Board members

6
Scientific Advisory Board members

$98K+
Revenue raised in 2023

1426
Social media followers

3.5K
Website visitors in 2023

416
5K participants

THE BEGINNINGS

Amy Robl, Board Chair and co-founder of the KARES Foundation, received a KDM5C diagnosis for her daughter, Gabby, in 2017. Gabby was 10 years old at the time, and the family had been searching for answers for years.

Not finding a community or a source of support, Amy created the KDM5C Support Group on Facebook to connect with the few families that had also received this ultra-rare diagnosis. Since 2018, this group has grown to over 600 members, representing an estimated 250 patients worldwide.

In 2020, a group of 12 KDM5C families met for the first time with researchers at Albert Einstein College of Medicine. This Rare Disease Day gathering was the spark that ignited collaboration amongst families that eventually became the KARES Foundation.

In late 2021, four founding moms (Amy, Lara Erekson, Amanda Lowry, and Melissa McNeilly) came together to begin writing the Articles of Incorporation for the KARES Foundation. We became officially incorporated in April of 2022.

As a relatively new nonprofit foundation, we are immensely proud of all we have accomplished in just two short years. This report will outline our successes in the year 2023.

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.

ABOUT KDM5C

CHARACTERISTICS

KARES FOUNDATION

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

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

“Disability parents are some of the strongest people I know. They never asked to be in this category of parenting, but every day they continue to show up.”

Taylor Leal, KDM5C parent

KARES FOUNDATION

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.

OUR VISION

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

OUR CORE VALUES

KARES FOUNDATION

HOPE

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

COMMUNITY

We collaborate with KDM5C families, friends, 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 our community.

INNOVATION

We strive to tap 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.

OUR PILLARS OF WORK

KARES FOUNDATION

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

We 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 embrace the ever-changing needs of our community.

FUNDING PRIORITIES

OF ALL FUNDING RECEIVED IN 2023, OUR TOP SPENDING PRIORITIES WERE...

1. **KARES FAMILY & SCIENCE CONFERENCE**
   OUR FIRST OFFICIAL CONVEVNING OF FAMILIES AND RESEARCHERS IN JUNE 2024
2. **KDM5C RESEARCH**
   INCLUDING THE FIRST CALL FOR PROPOSALS FOR THE KARES RESEARCH GRANT
3. **FAMILY SUPPORT**
   TOOLS & RESOURCES FOR FAMILIES INCLUDING THE FAMILY EDUCATION SERIES
4. **ADMINISTRATIVE COSTS**
   SUCH AS OUR WEBSITE AND ORGANIZATIONAL MEMBERSHIPS
5. **EDUCATIONAL OPPORTUNITIES**
   FOR OUR VOLUNTEERS TO LEARN ABOUT RARE DISEASE ADVOCACY
6. **RESERVES**
   SAVINGS FOR FUTURE PROGRAMS & PROJECTS

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 3 kids, Julian, Javi, and Emi. Julian was diagnosed with KDM5C in 2020 at age 4. Melissa works in nonprofit digital content management and is a strong advocate at the intersection of diability rights and human rights. Melissa manages KARES' website, social media, email marketing, and student intern program.

KARES FOUNDATION

BOARD OF DIRECTORS

JONI CHU

At-Large Board Member

Joni lives in Texas with his wife April and their five KDM5C affected kids Eva, Ellie, Eric, Eli and Eugene. The first of their KDM5C diagnoses came in 2021 with the others quickly following thereafter. Having always been an advocate for those with special needs, upon getting the kids' diagnoses Joni has been involved in the KDM5C community as much as possible including meeting several families from the US and abroad in person to share their experiences.

HEATHER COFER

At-Large Board Member

Heather lives in New York with her husband, Dave, and their 2 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.

SHAWN FILIPPI

At-Large Board Member

Shawn lives in Portland, Oregon with her husband David, her daughter Olivia, son Max and fur-baby Emma. She has been an attorney since 1999 in the areas of corporate, securities and finance law, with certifications in Business Law and Corporate Governance. She currently serves as Vice President, Chief Compliance Officer and Corporate Secretary for NW Natural Holdings. Shawn's daughter Olivia is an intern for KARES. With a life-long passion for disability rights and justice, Shawn is particularly excited about pursuing near-term therapeutics to alleviate symptoms of KDM5C.

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 KDM5C in 2021 at age 3. Since, Corey has been active on Facebook group and passionate about spreading awareness of this rare disorder.

KARES FOUNDATION

SCIENTIFIC ADVISORY BOARD

DR. JULIE SECOMBE

SAB Chairperson

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.

DR. 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.

DR. CATHERINE KEEGAN

Member

Professor of Pediatrics and Human Genetics at the University of Michigan in Ann Arbor. Director of the Division of Genetics, Metabolism, and Genomic Medicine in the Department of Pediatrics and Co-Director of the Michigan Medicine NORD Center of Excellence.

Dr. Keegan completed her undergraduate degree at University of Michigan, followed by her M.D. and Ph.D. at the same institution as part of the Medical Scientist Training Program. She completed her Pediatric Residency at Children's Hospital of Boston and returned to U-M for Medical Genetics fellowship training. She started her faculty position in the Department of Pediatrics at U-M in 2002.

DR. CONNIE KRAWCZYK

Member

Associate Professor in the Department of Metabolism and Nutritional Programming at the Van Andel Institute in Grand Rapids Michigan.

Dr. Krawczyk obtained her B.Sc. in Guelph, ON, Canada and her Ph.D. at the University of Toronto. She performed her postdoctoral studies at the University of Pennsylvania, then worked in drug discovery at Merck Frosst before starting her independent faculty position at McGill University in 2011. She moved to the Van Andel institute in 2018, to help establish the new Department of Metabolism and Nutritional Programming. Her lab studies metabolic and epigenetic programming of immune cells and has found that KDM5C functions at the intersection of these processes.

SCIENTIFIC ADVISORY BOARD

DR. 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.

DR. NHUNG TRAN

Member

Dr. Tran obtained her MD at the University of Texas Medical School (San Antonio) and is board-certified in Developmental and Behavioral Pediatrics practicing in Texas.

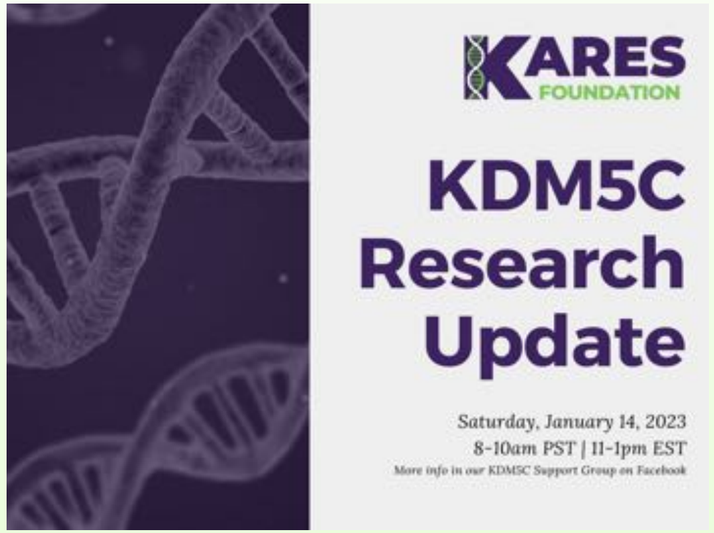
Dr. Tran is a developmental and behavioral pediatrician assisting families in the prevention, diagnosis and management of developmental difficulties and behaviors that can compromise development. She also works to foster understanding and promote the optimal environment for children and families through clinical care, education and advocacy efforts.

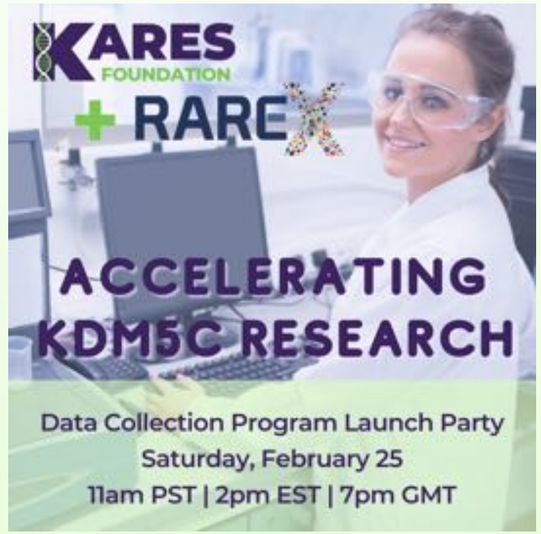
MARIAM REBOLLAR

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.

2023 EVENTS & ACCOMPLISHMENTS





JANUARY 14, 2023

VIRTUAL KDM5C RESEARCH SYMPOSIUM WITH 7 PRESENTERS & 42 ATTENDEES

JANUARY 27-29, 2023

BOARD STRATEGIC PLANNING MEETING WITH OUR 7 INITIAL BOARD MEMBERS

FEBRUARY 13, 2023

FAMILY EDUCATION SESSION ON COMMUNICATION: SUPPORTING AAC USERS

FEBRUARY 25, 2023

LAUNCHED THE KDM5C RARE-X DATA COLLECTION PLATFORM

2023 EVENTS & ACCOMPLISHMENTS

MARCH 1, 2023

OPENED THE FIRST CALL FOR APPLICATIONS FOR THE KARES RESEARCH GRANT PROGRAM

APRIL 29, 2023

REGIONAL MEET-UP OF KDM5C FAMILIES AT CANDLELIGHT RANCH, TEXAS

JUNE 1, 2023

FIRST KARES RESEARCH GRANT GIVEN TO DR. WEKSBERG'S LAB AT SICKKIDS, TORONTO

JUNE 3, 2023

REGIONAL MEET-UP OF FAMILIES & INTERNS FOR KDM5C AWARENESS DAY IN NORTH CAROLINA

2023 EVENTS & ACCOMPLISHMENTS

JUNE 5, 2023

FIRST OFFICIAL KDM5C AWARENESS DAY WITH 45 ATTENDEES AT VIRTUAL CELEBRATION

SEPTEMBER 19-21

KARES BOARD MEMBERS ATTEND THE GLOBAL GENES RARE ADVOCACY SUMMIT IN SAN DIEGO, CA

SEPTEMBER 30

MORE THAN 400 PARTICIPANTS RAISED OVER $75,000 IN THE 2ND ANNUAL KARES 5K

LOOKING AHEAD TO 2024

JUNE 5, 2024

2ND ANNUAL KDM5C AWARENESS DAY

JUNE 7-9, 2024

FAMILY & SCIENCE CONFERENCE IN SAN ANTONIO, TX AND VIRTUALLY

SEPTEMBER 28, 2024

THE 3RD ANNUAL KARES “5K YOUR WAY” EVENT

FALL, 2024

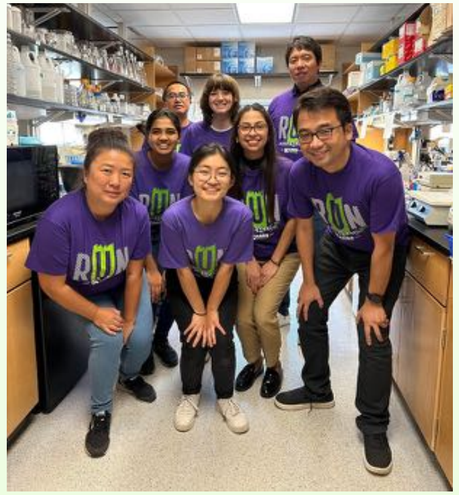
2ND CYCLE OF THE KARES RESEARCH GRANT OPENS

THE KARES 5K FOR KDM5C

Sept 29 - Oct 1, 2024

416 participants ◆ 24 teams ◆ 30+ sponsors ◆ raised $75,000+

THE KARES 5K FOR KDM5C



Top Teams

Jack's Pack $22K+

Jenna's Journey $12K+

Gift of Gab $6K+

Whitney's Warriors $6K+

Jog for Julian $3.8K

Team Jax $3.8K

Porter Proud $3.7K

Intern Impact $2.8K

KARES FOUNDATION

KDM5C ADVOCACY, RESEARCH, EDUCATION, & SUPPORT

In March 2023, we opened our first Call for Applications for research to advance understanding of the KDM5C gene and highlight therapeutic targets.

In June 2023, we funded a $30,000 research grant for...

Establishing a DNA Methylation Signature for Claes-Jensen Syndrome

Hospital for SickKids and University of Toronto

Dr. Rosanna Weksberg & Dr. Zain Awamleh

2023 FAMILY MEET-UPS

NC

TX

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or subscribe to our email newsletter.

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Instagram: instagram.com/karesfoundation
Twitter: twitter.com/KaresFoundation
LinkedIn: linkedin.com/company/kares-foundation
YouTube: @KARESFoundation

THANK YOU TO OUR STUDENT INTERNS

As a 100% volunteer-run organization, our student intern program provides us with much-needed help with administrative and communications tasks, while providing our student representatives with knowledge of genetic disorders and experience in rare disease patient advocacy.

BRIANA RAMOS

Senior, UNC Chapel Hill
Finance & Administrative Intern

GEETHIKA VENKATARAMAN
ASHA THAMODHARAN

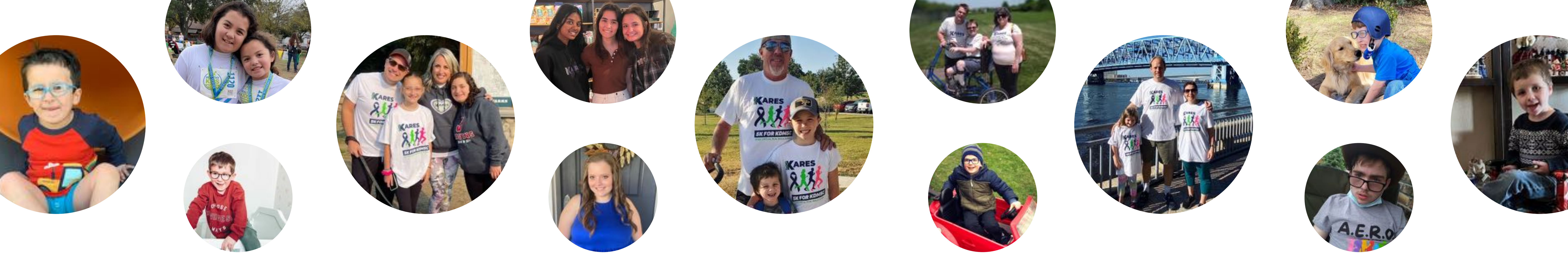
Seniors, Lake Norman Charter
Social Media Interns

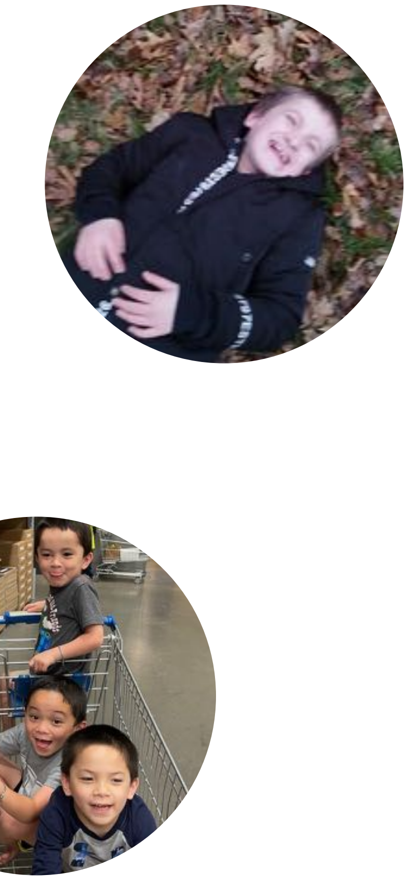
ADDISON LORD

Junior, Lake Norman Charter
Social Media Intern

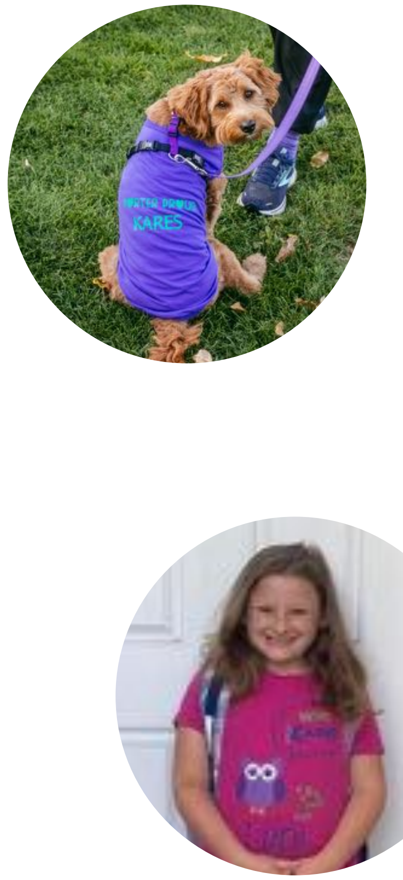
OLIVIA FILIPPI

Sophomore, University of San Diego
Administrative Intern





Thank You!



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

KARES FOUNDATION