KAPOW!

A KIF1A.ORG UPDATE
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APRIL 2018

REFLECTIONS

This letter is written exactly one year after a group of *KIF1A* families met for the first time in New York at Columbia University Medical Center. Most of us had only recently received a *KIF1A* diagnosis for our child. We were scared and anxious, faced with the cruel uncertainty inflicted upon those affected by rare disease. Not only were we a "rare" group, with fewer than 70 *KIF1A* patients *in the world* known at the time, we had quickly realized that the resources, knowledge and expertise we needed to help us move forward in our *KIF1A* journey were scarce or nonexistent.

But that meeting was a turning point for our small but mighty community. We then realized we had to become the drivers of resources, knowledge and expertise we so desperately sought. As the saying goes, "If not now, when? If not you, who?"

We may be rare, but we are not alone. We have our growing number of families (including over 140 known patients today).

We have a growing nonprofit organization, which we are proud to have been described as "grassroots," "scrappy" and "blue collar" by our mentors and supporters.

We also have a growing number of experts and advocates who are committed to helping KIF1A.ORG advance our mission. Starting with our self-proclaimed sherpa, Dr. Wendy Chung, we now have other renowned scientists, organizations like The Jackson Laboratory and partners like Global Genes and NORD who are in our corner.

Even a year later, our *KIF1A* journey is just getting started. But we are wholeheartedly optimistic in the the progress that's being achieved today and the accomplishments that are ahead for our *KIF1A* kids.

With Hope,

KATHRYN ATCHLEY, PRESIDENT
LUKE ROSEN, FOUNDER

IN THIS ISSUE

3 WAYS YOU CAN GET MORE INVOLVED IN KIF1A.ORG | PAGE 10

Fundraising Update

STARTING STRONG

In just over a year, our organization has raised $169,000 in donations to advance KIF1A research led by Chung Laboratory at Columbia University Medical Center. Online crowdfunding has been a powerful fundraising tool for families and supporters. Check out page 10 to learn more about our crowdfunding platform, **GiveGab**, and how you can get involved. We have also seen families host a variety of fundraising events, including silent auctions, restaurant and retail fundraisers, and t-shirt sales. These family-led efforts have been critical in the early stages of our organization. In addition to directly funding KIF1A research, our efforts have caught the attention of local and national media, some of the largest rare disease organizations in the world and talented scientists who have been inspired to join our mission to advance KIF1A research.

Our 2018 fundraising goal is bold: $1.1 million. Now that our organization has been operating for more than one year and has submitted our Form 990, we will also start aggressively pursuing grant opportunities from corporations and foundations. While we aim to secure significant grant funding opportunities, keeping up our our strong, grassroots fundraising culture is important. If you have questions or ideas about fundraising, please reach out to Kathryn at kathryn@KIF1A.org.

TEAM JAYBRIEL RAISED OVER $2,000 FOR KIF1A.ORG AT ONE OF THEIR LATEST FUNDRAISERS.

GO TEAM!

$169,000
RAISED TO DATE

2018 TARGET
$1.1 MILLION

Fundraising Update

RARE CAROUSEL

KIF1A.ORG was selected to participate in a collaborative crowdfunding campaign powered by the **Festival of Children Foundation** (FOCF) and **Global Genes: The RARE Carousel of Possible Dreams**. We participated with 30+ other rare disease organizations, with our dream of raising $25,000 to accelerate KIF1A research.

$30,486
RAISED

In less than two months, KIF1A families, friends and supporters raised over $30,000 to realize our dream!

We truly appreciate each and every hard-earned dollar that was contributed to fund our dream. Because of your passion for our mission to cure KAND, KIF1A.ORG had the third-highest fundraising total out of the 30+ participating organizations. Our Rare Carousel campaign ended strong on **World Rare Disease Day, February 28, 2018**, and we couldn't think of a better way to celebrate our KIF1A kids on this special day.

KIF1A.ORG will receive the donations raised through the online RARE Carousel platform (minus credit card fees of approximately 3.3%) by May 15, 2018. We will submit an Outcomes Report to FOCF and Global Genes by August 2018 to highlight the research progress that has been made as a result of the fundraising. We look forward to sharing our report with the KIF1A community!

LEADERBOARD

TEAM SUSANNAH
$12,573

TEAM RYLAN
$5,825

TEAM JAYBRIEL
$2,938

TEAM SUTTON
$2,800

TEAM PARKER
$1,665

TEAM COLLIER
$1,115

TEAM JIM
$1,025

TEAM BRAYDEN
$810

TEAM EMILY
$605

TEAM TURNER
$165

TEAM TRISTAN
$105

TEAM KIF1A
$860.68

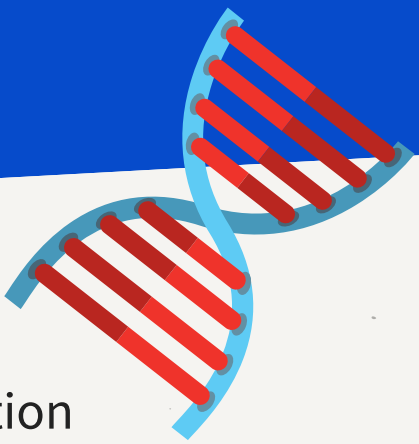
THANK YOU
PARTNERS & SUPPORTERS!

OUR PARTNERS

The Carousel of Possible dreams
Festival of Children Foundation

Global Genes®
Allies in Rare Disease

From the Lab



PRIORITIES

- Disease Characterization
- Disease Modeling
- Testing Novel Treatments for KIF1A

THE LATEST

The Chung Lab at Columbia University is actively working to understand the clinical characterization of disorders associated with mutations in *KIF1A*, as well as developing strategies for potential future therapies. Funds raised by KIF1A.ORG help support this vital research. In this section you'll find a description of each of our current priorities, as well as an update on the progress we are making in the lab.

DISEASE CHARACTERIZATION

Dr. Chung's team continues to expand an extensive natural history study of the clinical course of KIF1A Associated Neurological Disorder (KAND) and presented a poster with data collected at the American Society of Human Genetics. To read about this significant progress, click HERE. As our community grows, so does this study.

What is a Natural History Study?

A natural history study is a systematic collection and analysis of comprehensive information from people with KAND. The study is intended to advance understanding of the disease and how it is expressed and how it changes over time. This is vital in understanding KAND and developing strategies for future treatment.

Developing treatment is like building a house. "Both require a solid, sound foundation. For any rare disease treatment development program, that foundation consists of having a thorough understanding of the natural history of a disease" (fda.gov).

The natural history study also generates a robust registry of patients and families and plays a major role in our thriving, informed community. **Currently we know of 141 individuals with KAND in six countries.** This registry is a key component in understanding disease-causing mutations in *KIF1A*. The following is some helpful information to understand what exactly a disease registry is.

What is a Registry?

A registry is an organized online program for collection, storage, retrieval, and dissemination of a clearly defined set of data collected on identifiable individuals for a specific and specified purpose (nih.gov).

From the Lab

What types of data can be collected in a Registry?

All types of data can be collected, including data related to the following:

o Data that captures longitudinal information about the disease
o Diagnosis and treatment
o Management of care
o Quality of life
o Clinical testing samples
o Clinician reporting

Not only is it important to collect the right type of data that is meaningful to researchers, it is critically important that we collect and maintain data with the highest regard for its quality and integrity. In our conversations with other rare disease organizations, we have heard of natural history studies being scraped because these concerns were not adequately prioritized in the beginning stages of their research. This is why it is so vital for KIF1A.ORG to engage an experienced organization like Chung Lab for our study.

What are the uses of having a Registry?

Data from an effective patient registry can help to:
o Unite the patient and research community
o Educate patients, caregivers, and researchers about a disease or condition
o Improve quality of care
o Develop treatments
o Develop research hypotheses

Lia Boyle with Chung Lab is leading our natural history study and registry. Don't worry if you haven't heard from Lia yet; our rapidly growing registry of families has her working nonstop to understand how KAND affects every patient and family. **To join our registry, send your contact information to registry@KIF1A.org.**

DISEASE MODELING & WHY #WENEEDAMOUSE

The Jackson Laboratory is continuing efforts to design multiple mouse models in collaboration with Dr. Chung and Lia Boyle's extensive research and in vitro disease modeling. To understand why mouse models are vital in the discovery process, click HERE.

The primary goal of disease modeling is to replicate the physical characteristics, or phenotypes, associated with KAND. This is not an easy or inexpensive task, so we remain thankful to our collaborating research teams for their dedication to families affected by KAND. Currently we have one successful mouse model and the team is working to design more models of *KIF1A* mutations to observe, collect data, and test potential therapeutics.

Human cell lines, or IPSCs, are also in production to model the characteristics of *KIF1A* mutations and help researchers understand the disease. To read about how IPSCs work and why they are an important research tool, click HERE. In the coming months, patients in our registry might be contacted to discuss involvement in this work.

The disease models are useful to screen for medications or other treatments for KAND that improve symptoms and that are safe and do not have concerning side effects. It is critical to use these models to explore possible treatment and filter the list of possibilities down before trying any treatment in people. We are starting to test for treatments for KAND in these models now. We have a long road of discovery ahead of us before treatment becomes a reality, but we're making progress every day.

From the Lab

COLLABORATIONS

The Jackson Lab isn't the only team collaborating on *KIF1A*. Dr. Chung's work includes collaborations with Dr. Richard Vallee's Lab at Columbia University and Dr. Elliott Sherr's Lab at University of California, San Francisco.

What makes Dr. Chung's team so innovative is their commitment to the KAND community. Every family on our registry plays a role in discovery. KIF1A.ORG and families diagnosed with KAND are key collaborators in our mission to understand this disease and discover treatment for our loved ones.

COMMUNICATION

Our urgent need for research and support relies, in part, on strong, consistent and clear communication between our patient community and the research community. To create an efficient line of communication, questions about *KIF1A* research and how you can play an active role in discovery can be directed to KIF1A.ORG. Email Luke (luke@KIF1A.org) or Shannon (shannon@KIF1A.org) and we can provide information on research and facilitate communication with Lia Boyle at Chung Lab.

Stay tuned for information about webcasts and group chats with periodic updates about events, fundraising and research.

ONE MORE NOTE

All this innovative work brings the scientific community closer to developing therapeutic strategies for families affected by KAND. Stay updated on KAND initiatives by regularly checking our website and visiting our Facebook page.

Keep up the great work and remember:

TOGETHER WE ARE STRONG

Q&A with Lia Boyle

Meet Lia Boyle, a member of Dr. Wendy Chung's Laboratory at Columbia University Irving Medical Center. An MD and PhD student at Columbia University Vagelos College of Physicians and Surgeons with a particular interest in pediatric genetics, Lia has been working on *KIF1A* research since October 2016.

If you could have any super power, what would it be and why?

Telepathy. It would be interesting to know what other people are thinking and I've always been curious what goes through my cat's mind.

When did you know you wanted to be a scientist? What made you drawn to this field?

Ever since I was a kid, I've always been interested in understanding how things work – my "why" phase lasted long past my toddler years and through college. I didn't so much grow out of it, but now instead of asking other people why, I focus on searching for the answers myself. It wasn't until after college that I realized the best way for me to do this as a career was as a physician scientist. What I love about genetics as a scientific field is that because there is so much unexplored territory and so many things we are only just developing the tools to look at, there's a lot of space for discovery.

What does a typical day or week look like for you at the Lab?

There really isn't a typical day or even a typical week – part of what I love about what I do is how much variety there is. If Dr. Chung has patients coming in with KAND or any of the other conditions I study, I will participate in those appointments and might spend the whole day with families. Similarly, there are some days when I spend almost the entire time on the phone or Skype, talking with families all over the world. If it's a "mouse day," I might spend most of my day in the mouse room checking in on their health, studying their behavior, and genotyping (the mice need genetic tests just like patients!). Some of the mouse testing needs to be done every other day, so I'm often in lab at least one if not both days over the weekend. If blood or other samples arrive in the mail, I'll put everything else on hold to make sure I am able to process them in time so the samples don't go bad. Part of what makes Columbia a great place to do this sort of work is the wealth of resources: between the faculty here and visiting lecturers, I'll typically spend a few hours each week going to talks and learning from the experts in the field, in many cases hearing from people who developed the techniques we use. Finally, I try to set aside some time each week to read papers and catch up on the newest discoveries. Most weeks include some mix of all of the above.

Q&A with Lia Boyle

What do you love most about your job?

That's a tough question – I love so much of it! The highlight by far is getting to work directly with patients and families: I learn so much from you all. I also really enjoy the process of thinking through and planning which experiments will best answer specific questions. It feels like a logic puzzle.

What is the most challenging aspect of your job?

One of the challenging things about working with a neurodegenerative condition like KAND is knowing how much every day counts. This is especially difficult when I face the gaps in our current understanding of the condition: it's frustrating how many questions we just don't know the answer to yet. Even as we try to answer these questions, research is a process and many experiments take time. When working with mice, for example, it takes a few months to get from one generation to the next, and even then, there's no guarantee how many mouse pups will be in any litter.

What development are you most excited about in terms of KIF1A research?

Right now, I'm most excited about all the different disease models we have in the pipeline to look at multiple different *KIF1A* mutations. These models will enable us to start to answer some of our many questions about what exactly is going on and why KAND can look so different depending on what mutation someone has. Even better, these models are what let us explore possible treatments moving forward.

I couldn't get through a day at work without:

Caffeine! I can't leave my apartment in the morning without at least one cup of coffee, and most days, I'll also have a mug of (caffeinated) tea in the afternoon.

I am motivated by:

Every single family I have the privilege of getting to know and work with. My lab bench is surrounded with the photos you all have sent me!

I think mice are:

Really cute, but kind of smelly.

Community News

GROWING BY THE DAY

A year ago, there were 66 known patients with KAND. Now, we know of over 140 patients. With the increasing availability of genetic testing, more individuals are being properly diagnosed with *KIF1A* mutations. As more families join our community, our support circle grows stronger and we are able to engage additional resources and advocates to advance our cause.

GETTING SOCIAL

Like many rare disease organizations, social media platforms have been invaluable to the connectivity and growth of our community. Our public and private KIF1A.ORG Facebook pages have seen steady growth over the last year, while we are also cultivating a presence on Twitter, Instagram and LinkedIn. These platforms enable KIF1A.ORG to connect with other families, rare disease advocates and the scientific community. Encourage your family and friends to connect with us on our public platforms to stay updated on the latest developments at KIF1A.ORG.

CASTING A WIDER NET

While we have more than doubled our KAND population in the last year, we know there are so many more individuals out there who are either undiagnosed or misdiagnosed. One of our goals as an organization in 2018 is to begin targeted outreach to similar rare disease organizations in an effort to raise awareness of KAND and identify new patients. As many *KIF1A* families know, the odyssey endured before a proper diagnosis is found can be a long, frustrating and distressing process. By proactively reaching out to rare disease and undiagnosed communities, we hope to ease some of this struggle and connect newly found *KIF1A* families to the support and resources made available through KIF1A.ORG. Stay tuned for more information and updates as we develop this initiative in 2018.

LET'S CONNECT!

474 likes
PUBLIC PAGE

242 members
PRIVATE SUPPORT GROUP

148 followers

132 followers

16 followers

Get Involved

WE NEED YOU

KIF1A.ORG is a parent-led, 100% volunteer-powered organization. We have ambitious goals for our organization, from fundraising $1.1 million dollars in the next year to growing *KIF1A* awareness to creating new, valuable resources for our *KIF1A* families. There is no small contribution to our organization. **If are interested in any of the highlighted opportunities or have other ideas about how you or others in the *KIF1A* community can get involved, reach out to Kathryn at kathryn@KIF1A.org.**

SUPERHERO OF THE WEEK COORDINATOR

Gather and prepare profiles of *KIF1A* superheroes to feature on KIF1A.ORG. Our goal is to feature one superhero a week to highlight the strength and spirit of *KIF1A* kids. You'll be the go-to contact for parents who want to see their superhero featured on our site and help them complete their superhero's profile.

THE MIGHTY CONTRIBUTORS

KIF1A.ORG recently became a partner with The Mighty, a digital health community created to empower and connect people facing health challenges and disabilities. Share YOUR story and become a voice for the *KIF1A* community.

#WEDNESDAYWISDOM CONTRIBUTORS

Where ever you are in your *KIF1A* journey, you have something valuable to share with the rare disease community. Send us your bits of wisdom--in 125 words or less--to feature in our #WednesdayWisdom series on social media.

GIVEGAB

Did you know you can form your own online crowdfunding webpage to help KIF1A.ORG raise funding to advance *KIF1A* research? **Join our growing list of families, friends, and supporters on our fundraising platform, GiveGab!**

You can easily create a profile, set your personal fundraising goal and share your webpage with your network. GiveGab is an exclusive platform for registered nonprofit organizations and automatically generates donation receipts, so donors can be confident in their giving and KIF1A.ORG can efficiently manage donations.

GiveGab®
Nonprofit Giving Platform

START FUNDRAISING

Get Involved

EVENTS

Here are some upcoming events for the rare disease community, made possible by our partners at Global Genes and NORD. KIF1A.ORG representatives are scheduled to attend the RARE Patient Advocacy events in May and October. If you can represent KIF1A.ORG at any of these events or if you know of any other events that should be on our radar (including any outside the United States), get in touch with Kathryn (kathryn@kif1a.org).

RARE PATIENT ADVOCACY SYMPOSIUM

Philadelphia, PA | May 19

RARE ON THE ROAD: RARE DISEASE LEADERSHIP TOUR

Houston, TX | June 9

Salt Lake City, UT | June 30

Nashville, TN | July 21

GLOBAL GENES RARE PATIENT ADVOCACY SUMMIT

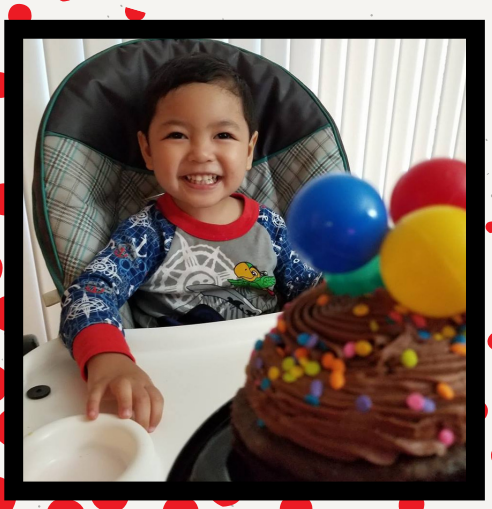
Irvine, CA | October 3-5

RARE DISEASES & ORPHAN PRODUCTS BREAKTHROUGH SUMMIT

Washington, D.C. | October 15-16

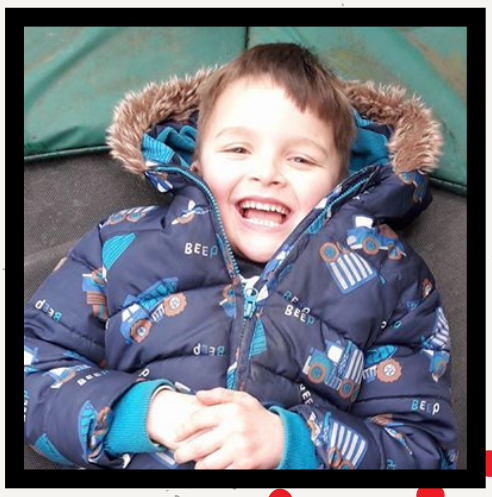
Spotted in Action

KIFIA SUPERHEROES



Spotted in Action

Spotted in Action



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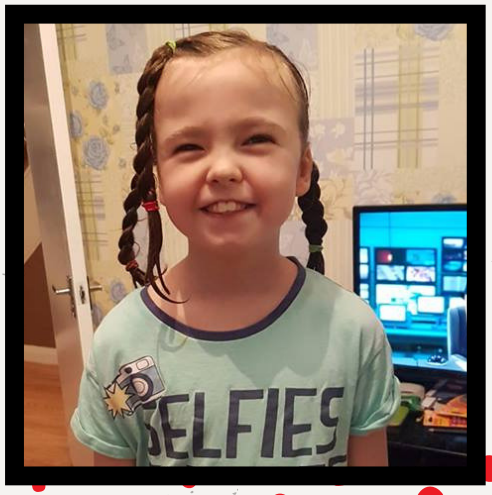

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KIF1A.ORG

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(Tax ID: 82-0714729)