

Fighting for Fiona & Friends

ND-NKH News Today

Volume 2, Issue 1
December 2019

A Bi-Annual Newsletter for the Non-Ketotic Hyperglycinemia Rare Disease Community & Beyond

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UPCOMING EVENTS

- ◆ **FUNDRAISING HOW-TO WEBINARS**
January 16 & 17, 2020
- ◆ **ND RARE DISEASE DAY CONFERENCE**
February 28-29, 2020
- ◆ **NOTRE DAME DAY 2019 Matching Fundraiser**
April 2020—Dates TBD

Giving Thanks for 5...4...3...2...1...

By Kerry Molina & Sean Nohelty

This Thanksgiving marks a number of significant milestones in the fight against NKH and all of us at "Fighting for Fiona & Friends/ND-NKH News Today" wish to express our deepest gratitude to everyone who is in this fight together. Today we give thanks for:

- 5 - The "ND-NKH" fundraising initiative to support research at Notre Dame's Boler- Parseghian Center for Rare & Neglected Diseases launched 5 years ago by Pat and Lynda Sarb in December 2014.
- 4 - The "Fighting for Fiona & Friends" fund for NKH research at Notre Dame established 4 years ago by the Fitzpatrick & Nohelty families on Thanksgiving Day 2015.
- 3 - The 3rd issue of the bi-annual "Fighting for Fiona & Friends/ND-NKH News Today" newsletter published today, following the 1st (Fall 2018) and 2nd (Spring 2019) issues.
- 2 - Based on the resounding success of the inaugural event in September 2019, plans are underway for the 2nd Annual *Walk for NKH* at Notre Dame in Fall 2020.
- 1 - New *NKH Leadership Board* formed by Kristin Archibald of NKH Crusaders this November 2019, bringing together the active U.S.-based fundraising groups into 1 collaborative conversation about NKH research funding needs and priorities.

With all the success and progress over the past 5 years, there remains much more to be done. Alongside you, we have ever-increasing hope in our NKH community and beyond, especially with the launch of the new NKH Leadership Board, for we know that "Alone We Are Rare and Together We Are Strong!"

Walk For NKH Recap

By Kerry Molina & Sean Nohelty

On the cool, crisp morning of Saturday, September 7 in South Bend, Indiana, the inaugural **Fiona & Friends' Walk for NKH** was launched. Held to commemorate the 5-year anniversary of the founding of the "Fighting for Fiona & Friends" fund for NKH research at Notre Dame, the **Walk for NKH** covered just over just over 4 miles through Notre Dame's beautiful campus.

(see *Walk for NKH*, continued on page 8)

Latest in NKH Research at Notre Dame

By Dr. Kasturi Haldar, Ph.D.

Boler-Parseghian Center for Rare & Neglected Diseases

NKH is a neuro-metabolic disorder characterized by intractable seizures, failure to thrive, lack of developmental milestones, and premature death. Loss of function of glycine decarboxylase (*GLDC*; also known as P protein) is a leading cause of NKH. Mutations and deletions in P protein are responsible for greater than 80% of NKH cases. CRND executes a four pronged approach to their research, which includes:

MOUSE MODELS

We have overcome hurdles to developing mouse models that mimic NKH disease as seen in patients with genetic mutations. The mice developed show the full spectrum (severe to mild) of neuro-metabolic disease with measurable increase in glycine levels and defects in brain, liver and kidney function. Two mouse models have been developed and a third is planned for 2020.

GENE THERAPY

We have partnered with Dr. James Wilson and his team at the University of Pennsylvania Vector Core for developing gene therapy for NKH. ND has signed an agreement with U Penn. Vectors for Gene therapy have been designed and are in process of production at the Penn Vector Core. We expect their delivery to ND in December 2019.

DISEASE PREDICTION & MANAGEMENT

Computational, Cellular & Mouse models: We have completed the first large scale computational analyses of all patient mutations to predict and separate severe from attenuated NKH based on patient mutation. We want to expand working directly with patient families their medical records and tissue specimens to better help clinicians, patients and researchers improve understanding and managing major symptoms like seizures.

ZEBRAFISH

We have continued work with new zebrafish models to study NKH mutations that affect brain and kidney development and develop screens for new drugs.



Dr. Kasturi Haldar, Ph.D.,
James Parson and Carrie Quinn
Director, BP-CRND

“ND has signed an agreement with U Penn for vectors for gene therapy.”

- Dr. Kasturi Haldar



Boler-Parseghian Center for Rare & Neglected Diseases NKH Research Team

Dr. Kasturi Haldar, Ph.D. Barb Callhoun, Outreach Coordinator

Dr. Suhail Alam, Ph.D. Joe Farris, Graduate Student

Dr. Rebecca Wingert, Ph.D.

... and all the NKH Families & Friends like YOU!! ...

NKH Leadership Board Newly Formed

By Kristin Archibald

There has been so much happening in the NKH Community since the NKH Conference! Many know I have spent the last 5 months working on a grant to help grow the NKH Crusaders Foundation and better support NKH Research and families. This grant has opened important dialogue with NKH Families, the University of Notre Dame, University of Colorado and Boston Children’s Hospital. During this time, it became apparent that more transparency was needed within the NKH Community and between NKH Researchers and families actively raising funds for NKH research.

To assist in achieving that goal, NKH Crusaders has formed a NKH Leadership Board. The Board will include the active U.S. NKH Research fundraising groups. The Leadership Board will work as one to review research funding needs of ND and UC and collectively decide on funding models with both universities a year at a time. Notre Dame and the University of Colorado were asked to submit a letter of need for December 1, 2019 - December 1, 2020. The amount needed to fund both research programs is \$245,000 for the next year. It is important to remember that we need to fund both doctors and their work as we need more than one researcher and one plan in place to achieve the overall goal for better treatments and/or a cure.

It is equally important at this stage of research for the doctors to keep families informed on financial needs and how those funds will be used. The \$245,000 figures can be overwhelming when looked at from one organizations’ viewpoint. The fact is many NKH families are raising funds (and have for years) but often do not communicate with one another.

The Leadership Board will change that and be a vehicle for us to further support each other and the research efforts as a whole. The Board will hold quarterly meetings to update one another on current plans for fundraisers and ways we can support one another better. Each organization will continue to raise funds under their name.



The Board will also review the needs of both doctors’ research programs and, with consensus, establish quarterly payments to fund both programs by evaluating what and when each NKH organization raises funds and can support a payment. The doctors have committed to providing quarterly updates on research progress and this will include information that can be shared publicly. This is exciting and will increase transparency for all families!

“The Leadership Board will...be a vehicle for us to support each other and the research efforts as a whole.”

- Kristin Archibald, NKH Leadership Board

Joseph's Goal (based out of the UK) has provided financial support for many years to keep NKH research moving forward. Emma and Paul Kendrick (Joseph's Goal) have been wonderful and with their support has been vital to reach achievements made to date. However, at this time they have decided to focus their support on Dr. Nick Green (and not send funds to Dr. Van Hove). The absence of their financial support, creates a shortfall of funding expected in the coming year (approximately \$52,000).

Research needs to keep moving forward. Without new families willing to get actively involved, funding levels cannot be sustained. To offset and surpass this shortfall, we need all of you to step in and join our efforts. Fundraising may be new to you, or maybe you have not fundraised in a while. Fundraising can be a daunting task on many levels. Not to worry! Another role the Leadership Board will take on is hosting informational webinars (open to all) on topics including: how to get started with fundraising, fundraising ideas, and ways to collaborate and support between NKH organizations/families.

NKH Crusaders (on behalf of the Leadership Board) also plans to establish a process for families to send fundraising dollars to a dedicated NKH general fund (not directed to either university) where every dollar will be looked at and a Board decision made where the money is needed and will be best utilized.

We are a determined community and together nothing is impossible! We can do this!

(see Leadership Board, page 7)

Meet Nora Almany

By Amanda Almany

At home, Nora still was not waking up to eat, waking up to noises, or crying when she was hungry or needed a diaper change. Nora had her first visit with the pediatrician that Friday; however, it was not Nora's pediatrician but the only one available that day. She informed us that Nora lost 10% of her body weight and that was alarming.

When we told her that she wasn't waking up to eat, and she wasn't crying, she brushed it off. She sent us to the lab to have a heel poke done to test for jaundice. When Nora did not wake up from the heel poke, we knew something was wrong. We took Nora home and hoped that she was okay and that she would start waking up.

On Saturday, after not waking up to eat once again, we decided to head to Mercy Hospital in St. Louis. Andrew called ahead and when we arrived, they were waiting for us. They rushed Nora to a room in the ER and began doing every test imaginable. When they started crossing things off the list that could be wrong, and their questions to us were becoming more in depth, we began to realize that something was seriously wrong.

Once Nora was "critical, but stable," they moved us to a room in the Pediatric Intensive Care Unit (PICU). The doctor was convinced at this point that Nora had a metabolic condition. Her lab results were rushed to the Mayo Clinic in Minnesota. On Tuesday, the results arrived and Nora was diagnosed with a rare, genetic, metabolic disorder called Nonketotic Hyperglycinemia, or NKH.



Nora Almany



Nora and Big Sister Alex

When I first heard it, I remember thinking, oh, so she can't break down sugar? Boy, was I wrong. The attending doctor, neurologist, and metabolic specialist sat us down and explained the disorder.

Even though the information was grim and heartbreaking, we were happy to finally know what we were dealing with. Our next step was to transfer to St. Louis Children's Hospital and begin treatment. That afternoon, we were transferred by ambulance to SLCH. Nora spent six weeks at Children's Hospital. In those six weeks, we experienced things I could have never imagined experiencing. Nora went through more in the first two months of her life than most people go through in a lifetime. We were told multiple times that our baby probably wouldn't make it home from the hospital. The option was given to us more than once to take her home, make her comfortable, and watch her pass. That was unacceptable to us.

Nora is now three years old and Andrew and I are so incredibly lucky and grateful. Even though Nora doesn't do everything a "typical" toddler would do, we are proud of her strength and determination. This is our new normal, and she is our superhero.

Visit the Nora Jane Foundation for more information and for details about or upcoming fundraisers at:

NoraJaneFoundation

NKH Crusaders Convention Recap

By Kristin Archibald

The 7th annual NKH Conference was held October 2-4, 2019 at the Masonic Lodge in Weymouth, MA. Nineteen NKH families from across the globe came to learn more about research, network with other NKH families and fundraise for research. Speakers from Boston Children's Hospital shared information on genetics, future pregnancy options, ketogenic diet and immune health and NKH. Dr. Kasturi Haldar from the University of Notre Dame and Dr. Johan Van Hove from the University of Colorado updated the attendees on ongoing NKH research efforts at their universities. They are both making great progress!

Dr. Haldar shared news of their work on mouse models, Gene Therapy (and the exciting news surrounding the creation of a vector!). She also shared information on disease management and the use of zebrafish to study NKH mutations affecting brain and kidney development. This work assists in developing new screens for drugs.

Dr. Van Hove spoke about his mouse model studies, chaperone development and CSF and brain development studies. We are hopeful for what is to come in the next 12 months with all the great research being done.

The event ended with a beautiful dinner fundraising event held in the Tirrell Room in Quincy, MA. We raised over \$25,000 in support of NKH research. The amount of awareness about NKH continues to grow each year since NKH Crusaders began in 2010. The organization now supports both the University of Notre Dame and the University of Colorado research programs. Since 2010, over \$397,745 has been raised with \$377,745 given to the University of Colorado and \$20,000 to the University of Notre Dame.

Save the date for next year's conference: September 30 – October 2, 2020!



Learn more about NKH Crusaders at NKHCrusaders.com

Fundraising Events & Opportunities

By Kerry Molina, Pat Sarb & Kristin Archibald

There are several current fundraisers happening now. Share with your friends, colleagues, and social networks. The more we share, the greater our circle of influence and the more monies that can be raised to advance the research efforts of the amazing teams at the University of Notre Dame and University of Colorado.

Sarb Family Fundraiser with matching gift!

We are excited to announce that thanks to a matching gift challenge from a generous couple, they will match donations dollar for dollar up to \$5,000 made to Notre Dame's NKH Research Fund between Giving Tuesday and December 31, 2019. That's a total impact of \$10,000. One hundred percent of the money donated will go towards NKH research. Donations can be made at the following link: <http://supporting.nd.edu/NKHResearchFund>. Please include the note "Sarb matching gift" in the Additional Details box.

Yankee Candle Fundraiser – perfect for holiday gift giving!

Do you love Yankee Candles? We would love for you to consider ordering for our online fundraiser to support NKH research: <https://www.yankeecandlefundraising.com/store.htm>

Group Number: 999975598
Group Name: NKH CRUSADERS
Catalog/Group Sales Ends: 31-Dec-19
Online Sales Ends: 08-Jan-20

Lynch Creek Fundraiser

Find beautiful wreaths, centerpieces and more for yourself or gifting! NKH Crusaders earns 15% on every purchase. Sale Ends December 15 – Act now!

Lynchcreekfundraising.com

Fundraising How-To Webinars

Thursday, January 16 & Friday, January 17, 2020

Never fundraised before? No idea where or how to start?

NO PROBLEM! We're here to help! Mark your calendar and join us for our upcoming webinars on Thursday, January 16 and Friday, January 17, at 7pm EST. More information and how to log in and join instructions will be posted on NKH Crusaders website and Facebook pages soon.

NKHCrusaders.com

**Got an Upcoming
Event or Fundraiser?
Want to share your
story?**

**SUBMIT YOUR
INFORMATION, STORY, OR
EVENT DATES FOR OUR
SPRING 2020 ISSUE TO :**

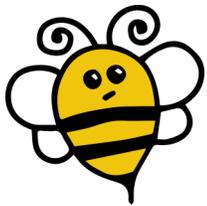
kerry@fourcloversconsulting.com

Leadership Board

(continued from page 3)

The Leadership Board members are NKH Crusaders, Nora Jane Almany Foundation, Carson Fund, Brodyn’s Friends, Swarm for NKH, Hope Link, Fighting for Fiona & Friends (Fitzpatrick & Nohelty families), and ND-NKH (Sarab family). More information will be shared in the coming weeks as the Board finalizes the information from ND and UC. We and the research teams recognize how important it is to keep the NKH community informed so you know where the money is going that you work so hard to raise. The Board is committed to keeping communication flowing and will utilize the existing “Fighting for Fiona & Friends ND-NKH News Today” newsletter to keep you informed.

It has been an amazing experience to put this plan together and see the connections being made. We look forward to so many more of you joining forces with us and all the amazing things yet to come!



“Research needs to keep moving forward. Without new families willing to get actively involved, funding levels cannot be sustained... We are a determined community and together nothing is impossible! We can do this!”

- Kristin Archibald, NKH Leadership Board



Patient Histories and their Importance in Research

By Barb Calhoun, MSN, RN, PNP



Barb Calhoun, MSN, RN, PNP

As the Outreach Coordinator for the Boler-Parseghian Center for Rare and Neglected Diseases at the University of Notre Dame, my overall goal is to support rare disease patients and their families. This is accomplished through a variety of educational, research and community focused programs.

Through courses (BIOS 40450/60565) co-taught by Kasturi Haldar, Ph.D. and myself, students become adept in the study of natural history of rare diseases. Patient families provide medical records that are reviewed for information to better understand the pertinent rare disease symptoms and their progression.

NKH Patient Histories are an important part of the work the Haldar

Lab conducts. In addition to medical records, researchers request skin biopsies and plasma from patients to evaluate the molecular and cellular factors contributing to disease. This data is then used to develop targeted therapies for the disease.

“The data is then used to develop targeted therapies for the disease.”

- Barb Calhoun, MSN, RN, PNP

If you are interested in participating in our NKH studies, please contact the Boler-Parseghian Center for Rare and Neglected Diseases at 574-631-3372 or bcalhoun@nd.edu.

Walk for NKH Recap

(continued from page 1)

A pre-walk program led by emcee Kerry Molina included inspirational comments from Mary Fitzpatrick and Pat Sarb, "America the Beautiful" and "Back Home Again in Indiana" sung by Aaron Reeder, warm-up exercises led by John Fitzpatrick, and a group prayer and blessing over Pat and Fr. Eliaona's Notre Dame Trail walking sticks.

The parade of 88 walkers strong - including NKH families, friends, advocates, researchers and students - made their way by many of Notre Dame's most storied landmarks including the Hesburgh Library, Golden Dome, Basilica of the Sacred Heart, St. Mary's Lake, Grotto and Log Cabin. Pausing momentarily at Badin Hall for a Mass concelebrated by Rev. Francis Murphy, C.S.C. and Rev. David Eliaona Lyimo, C.S.C., the walk concluded at Jordan Hall of Science with a luncheon buffet and research update from Dr. Kasturi Haldar and Dr. Rebecca Wingert.

The inaugural **Walk for NKH** was a huge success in both raising a total of \$16,622 for NKH research via a [GoFundMe](#) campaign (far exceeding the \$10,000 goal), but also in expanding awareness and support beyond the NKH community. Plans are already underway to make the **Walk for NKH** an even larger, annual event at Notre Dame with the organizational support of the University. We look forward to seeing you on campus to walk with us in **Fall 2020**. Be sure to bring your comfortable shoes!



Fitzpatrick Family leading the Walk



Pat Sarb & Fr. Eliaona



Hergott Family



Notre Dame BP-CRND Students



Walk for NKH Cookies

Learn More

More Ways to Help

Donate to the NKH research funds at Notre Dame supporting Dr. Haldar and her research team:

FIGHTING FOR FIONA & FRIENDS

giving.nd.edu/fionafund

- and -

ND-NKH

supporting.nd.edu/NKHResearchFund

100% of all donations to both funds

Other NKH Related Organizations

For more information on these organizations, click on their logos:



The Foundation for NONKETOTIC HYPERGLYCINEMIA

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**Alone We Are Rare,
Together We Are Strong!**

Share your story, ask a question, and/or submit ideas for future articles to:

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