Spring 2024 Newsletter

national neutropenia network

NNN Updates

Thank You Stephanie!!!

Stephanie Long joined the NNN as the Executive Director in August 2022 after her now 7-year-old daughter, Hannah, was diagnosed with SCN at the age of 5. Hannah was early on in her diagnosis and Stephanie was looking for a way to learn as much as she could about SCN by hitting the ground running, hoping to connect with the community, and making an impact. During her time with the NNN, she fostered multiple community partnerships, secured grants to support the NNN 2023 Family Retreat and helped to raise funds to support the NNN Mission. Stephanie gave a heartfelt address at the Family Retreat and really captured the essence of what it's like being a parent of a child with SCN looking for "that community" to help bear the load. **Watch it here!**

Stephanie has decided to step down from her position as Executive Director to focus on her daughter's journey with neutropenia. We look forward to supporting Stephanie and Hannah with the community and resources they will need. We thank you Stephanie for all that YOU have given to the community!!

Welcome Jeanne Siegel, Executive Director

Jeanne lives in Cincinnati, Ohio with her husband Chris and son, Will. Jeanne brings almost a decade of experience in nonprofit leadership and a relentless drive to advance awareness, support, and research initiatives within the neutropenia community. Her dedication stems from being a mom to their youngest son, Alex, who was diagnosed with Severe Congenital Neutropenia when he was 8 weeks old. Alex passed away in July 2022 at the age of 9 after complications from a bone marrow transplant. This profound loss has fueled her commitment to support families affected by neutropenia and continue efforts to find a cure so one day, no family will have to endure similar heartache.

When not working, Jeanne loves golfing, walking her dogs, and traveling with her family. Because of their experience, Jeanne and her family firmly believe in living every day to the fullest.

Jeanne is excited to be joining the NNN and states, "From teaching kids the game of golf, to working with organizations committed to finding cures for congenital diseases, I have been lucky to work with organizations that align with my passions, interests, and values. This opportunity to join NNN is no different. I can't think of a better way to devote my time or honor my son than to work for and support this community."

Jeanne can be reached at jeanne@neutropenianet.org

How Can I Volunteer and Get Involved with the Network?

We have a number of ways to get involved with us:

- Serve as a Board Member
- Become a Peer Support Volunteer
- Join the Ambassador Program (applications will be accepted until August 31st; terms run Sept 1-Aug 31)

There are also additional ways to volunteer:

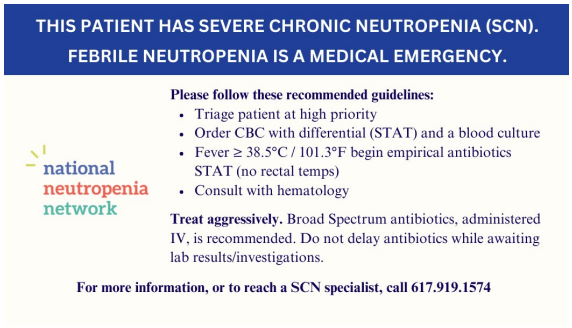
- Community Outreach/Advocacy Volunteer
- Become a Story Writer for our website/newsletter
- Virtual Fundraiser Coordinator
- Support Group Moderator
- Conference Committee Member (in-person or virtual)
- Share an idea! We would love to hear it!

The success of the Network has been largely due to volunteers and their commitment over the years. Thank you for your consideration in joining our efforts.

Click the button below to complete the interest form. A member of our team will be in contact with you for the next steps! (Please note some positions have a limited number of positions which will be filled with the most qualified candidates.)

Volunteer Today

New Emergency Room Cards Available



Click HERE for your updated ER card.

Community Spotlight

Rare Disease Day by Maria Diploudis

Rare Disease Day occurs annually on the last day of February. In 2024, it fell on the rarest day of the year - Leap Day. The first Rare Disease Day was in 2008 in Europe and Canada, and it is now observed in over 80 countries. Conditions that affect less than 1 in 2,000 people are considered rare, and there are over 6,000 identified rare diseases which afflict hundreds of millions of individuals worldwide. Rarediseaseday.org works in collaboration with 1,400 organizations in over 100 countries and territories to raise awareness, share resources, and unify the global rare disease community.

The majority of rare diseases are genetic and diagnosed in childhood - that was the case with Hannah, who was born with Severe Congenital Neutropenia. Although Hannah was born with this condition, she was diagnosed at 5 years old after frequent hospital visits due to fevers and infections. Hannah, now 7, helps raise awareness for her rare condition with the help of her mother, Stephanie Long. Stephanie is currently the Executive Director of the National Neutropenia Network (NNN), though she will be stepping down from that position to focus on Hannah's health. Hannah and Stephanie have organized several fundraising events over the past couple of years in their home state of West Virginia in collaboration with politicians, local news programs, restaurants, and Hannah's school. These events include an annual Neutropenia awareness 5K run/walk, a restaurant fundraiser in which 10% of an evening's earnings were donated to the NNN, and official recognition by West Virginia Governor Jim Justice proclaiming February 28 as Rare Disease Day.

Maison, 9 years old, was diagnosed in 2021 with Severe Chronic Neutropenia of unknown origin and Bone Marrow Failure (with probable Myelodysplastic Syndrome). He has undergone 6 bone marrow biopsies under the care and observation of specialists at the National Institute of Health (NIH) and the Severe Chronic Neutropenia International Registry (SCNIR) - further genetic testing and evaluation will hopefully provide a more complete diagnosis. Maison and his mother, Tracy Lemmons, actively work towards raising awareness about Neutropenia. Maison was recently featured in an online video clip highlighting the reality of living with neutropenia as a child. This video was created by Kosair for Kids, an organization that helps deliver financial and other support services to the families of children living with serious health conditions. For Rare Disease Day this year, Tracy and Maison created flyers to help raise awareness. The flyer included the mission statement of the National Organization for Rare Diseases (NORD): "Alone we are rare. Together we are strong." Maison also handed out gift bags to his classmates that included barcodes to more information about NORD and the NNN.

Living with Neutropenia as a child means often having to stay home from school due to being sick, frequent hospitalizations, missing out on sports or other social activities, and avoiding activities that could potentially expose the child to dangerous bacterial or fungal infections. All of that is on top of having to frequently communicate the realities of living with an invisible - but very serious - medical condition to schools, doctors, hospitals, and the caregiver's employers. Any event, organization, observance, or recognition that helps the general public understand the reality of living with rare health conditions helps make life just a little bit easier for those living with rare health conditions and their loved ones.

Proclamation
by Governor Jim Justice

Whereas, there are nearly 7,000 diseases and conditions considered rare each affecting fewer than 200,000 Americans in the United States, according to the National Institutes of Health (NIH); and

Whereas, many rare diseases are serious and debilitating conditions that have a significant impact on the lives of those affected; and

Whereas, while more than 450 drugs and biologics have been approved for the treatment of rare diseases according to the Food and Drug Administration (FDA), millions of Americans still have rare diseases for which there is no approved treatment; and

Whereas, individuals and families affected by rare diseases often experience problems such as diagnostic delay, difficulty finding a medical expert, and lack of access to treatments or ancillary services; and

Whereas, while the public is familiar with some rare diseases, many patients and families affected by less widely known rare diseases bear a large share of the burden of funding research and raising public awareness to support the search for treatments; and

Whereas, residents of West Virginia are among those affected by rare diseases because nearly 1 in 10 Americans have rare diseases.

Now, Therefore, Be it Resolved that I, Jim Justice, Governor of the Great State of West Virginia, do hereby proclaim February 28, 2023 as

Rare Disease Day

in the Mountain State and encourage all citizens to join me in this observance.

In Witness Whereof, I have hereunto set my hand and caused the Great Seal of the State of West Virginia to be affixed.

Done at the Capitol, City of Charleston, State of West Virginia, this the First day of February, in the year of our Lord, Two Thousand Twenty-Three, and in the One Hundred Ninetieth year of the State.

Jim Justice
Governor

By the Governor:

Mac Warner
Secretary of State

SOURCES:
https://rarediseases.org/rare-disease-day
https://www.rarediseaseday.org
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9632971
https://en.wikipedia.org/wiki/Rare_Disease_Day
https://www.kosair.org

Physician Corner

SCNIR Update

The Severe Chronic Neutropenia International Registry (SCNIR) is now established in its new home in Boston. We are reconnecting those who have already signed up and welcome new registrations. The updated patient and family handbook is now available on both the SCNIR and NNN websites. Below you will find the SCNIR's contact information:

SCNIR-dl@childrens.harvard.edu
www.scnir.org
617-919-1574

CLICK HERE FOR THE UPDATED PATIENT HANDBOOK.

News from Seattle, March 2024

Over many months we worked steadily to move the US office for the SCNIR from Seattle to Boston. It was more work than any of us expected. We had to satisfy the review boards at the University of Washington and Boston Children's Hospital that the transfer was secure, intact, and complete. As you can imagine, we had a lot of records from more than 30 years of working with you in defining the causes, consequences, and treatments for neutropenia.

At our meeting in July in Cincinnati, we spoke enthusiastically about our work with Emendo, based in Tel Aviv, Israel, on selective gene editing as a potential therapy for neutropenia caused by mutations in the ELANE gene. Really, much of our enthusiasm rested on the promise of this technology as a way to treat many genetic diseases causing neutropenia and other conditions. In December, it was devastating for us to learn that Emendo had nearly collapsed, dismissed many of its employees and closed our project, at least for the time being. The science was very promising, but the problem was collapse of the parent company in Japan. We remain hopeful, and we are continuing to collect DNA samples that may be used in the future for this work. Thank you to all of you who have helped us with this promising work.

We are now in the late stages of planning a study of home monitoring of blood counts, neutrophil counts, to diagnose cyclic and congenital neutropenia. As you know, for years we have recommended getting blood counts at least 3 times a week for 6 weeks to determine if someone has cyclic neutropenia. The pattern of symptoms (recurrent fevers, mouth sores, bacterial infections etc.) and genetic sequencing helps with the diagnosis, but serial counts showing regular neutrophil count oscillations really makes the diagnosis. Lacking true cycling with very low counts at the low point leaves doubt.

We have worked for several years with a California company, Athelas, on developing a small, FDA approved device that is easy to use for home monitoring of blood counts. We have a plan to test the acceptability doing daily counts yourself at home with this simple device. For this study, we will focus examining its utility to distinguish between cyclic and congenital neutropenia.

We also have exciting work dissecting the mechanisms and potential treatments for novel causes for severe chronic neutropenia discovered through family studies. I

cannot emphasize too much how valuable it is to know family history in trying to understand neutropenia.

Audrey Anna and I remain very committed to helping you, and continuing the work of the SCNIR just as long as we can. It has been “a good ride,” and we appreciate so much your support and help to us over these many years.

Sincerely,
David

David C. Dale MD
Professor of Medicine, University of Washington

Community Partner Connections

X4 Pharmaceuticals

Click here for information on X4 Pharmaceutical's chronic neutropenia study. Phase 3 study updates will be released soon!

Exciting Partnership Announcement: Free Genetic Testing!

In an effort to increase accessibility to important resources for neutropenia patients, NNN is partnering with Probably Genetic to ensure our community is aware of their free genetic testing program for Primary Immunodeficiencies.

This collaboration aims to eliminate the barriers that often prevent patients from accessing crucial genetic insights. Through comprehensive analysis, patients and their families can gain a deeper understanding of their condition, enabling personalized treatment plans and better management strategies.

Key benefits of the Probably Genetic’s program:
- No Cost: Eligible participants will receive genetic testing at absolutely no charge!
- Comprehensive: Eligible participants will receive whole exome sequencing and will have the opportunity to discuss their results with a board-certified genetic counselor.
- Clinical grade & HIPAA compliant: The entire testing process is CLIA and CAP certified, and Probably Genetic adheres to the most stringent data security and privacy guidelines.

How to Participate:
Click here to report your symptoms and apply for free genetic testing. It takes <10 minutes to complete!

If you have any questions about the program, please contact the Probably Genetic team at hello@probablygenetic.com.

From Our Community Partners in Toronto, Canada:

8th National Marrow Failure and Myelodysplasia Patient and Family Virtual Conference

Saturday April 13th, 2024 @ 1:00-6:00 PM EST

We are pleased to invite you to this free interactive meeting. It is an ideal opportunity to learn, network with other families and meet experts in the field. Families/patients outside of Canada are welcome to join.

This meeting is for patients and families with aplastic anemia, bone marrow failure disorders and myelodysplastic syndrome (MDS), physicians, other health-care workers, fellows, residents, and all others who are interested in the field. Children and adult patients are invited.

Virtual attendance will be provided through webcasting by Microsoft Teams.

The Conference Program includes:

New treatments and clinical trials, adult care, cancer risk, fertility, new transplant strategies (e.g. haploidentical), long-term effects of transplant, Canadian pediatric and adult centers which specialized in caring for patients with bone marrow failure/MDS/aplastic anemia.

Access our website and registration via https://cvent.me/WW8W3G.

The National Neutropenia Network depends on support from the friends and families of those whose lives have been forever changed and challenged by this rare disease. Thank you for your support.

Donate Today

Our mailing address is:
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