THE ELLA JEWELL FOUNDATION

"TOGETHER, WE CAN MAKE A DIFFERENCE"

SUMMER 2013

BY: KRISTIN MCGUINNESS

Ella Jewell McGuinness was born on September 2, 2009 and diagnosed with Severe Congenital Neutropenia shortly thereafter. Ella's diagnosis was devastating to our family but we decided to stay positive. We are very fortunate that Ella responds well to daily injections of G-CSF. When we were first learning about Neutropenia, we quickly realized that many consider Neutropenia an "orphan" disease, unknown by many and, unfortunately, under researched.

When Ella was diagnosed we searched for an organization that was raising funds for research, in hope that Ella would one day be cured. We learned from Neutropenia experts, Dr. David C. Dale and Dr. Lawrence Boxer that there was no known organization/foundation in America raising funds specifically for Neutropenia related research.

We felt in our hearts that something needed to be done, not only for Ella but for all families affected by this disease. We have made it our life's mission to do all that we can to make a difference. As rare disease research moves forward, Neutropenia cannot be forgotten. Although we are extremely thankful for G-CSF, we want alternative therapies and hopefully, one day, a cure.

In early 2010 we formed a Board and Committee and started The Ella Jewell Foundation. Although the foundation is named in honor of our daughter, it is established to benefit all families affected by every type of Neutropenia including Congenital, Idiopathic, Cyclic and Autoimmune.

From the very start, it has been our goal to encourage other families to join forces with us. We feel that we all need to work together because, " Together, we can make a difference." We have never felt so passionate about anything. The foundation gives us a great amount of hope and we would like to spread that hope to all families affected by Neutropenia. We are very excited and honored that other families have recognized the strength in our mission. You will read further about their efforts and how they joined forces with us in this newsletter.

The Ella Jewell Foundation has held a number of great events in effort to raise awareness and funds for research. Our first event, held in November 2010, was a very successful Dinner Gala at the Westchester Marriott in Tarrytown, NY. Close to 300 guests enjoyed cocktails, dinner, dancing, a silent auction, and raffle prizes. Both Dr. Lawrence Boxer and Dr. David C. Dale attended the event. It was an amazing night and a kick off to something that has grown exponentially since. The Ella Jewell Foundation raised over $30,000 that evening. As the parents of Ella Jewell, it was one of the best nights of our lives. The outpouring of love and support allowed us to see that there is hope. Hope for every mother, father, sister, brother, grandparent, and patient affected by Neutropenia.

We have held various events in the last three years. Including an extremely successful wine tasting event. We also formed "Team Ella Jewell". Which brings me to our greatest honor of all, when the Long Family joined forces with us in late 2011. The Long's came up with a fabulous idea, running races! Many other families are now forming teams nationwide and running towards the ultimate goal, a cure.

Now, the Ella Jewell Foundation is excited to introduce a new, easy, fun, and exciting fundraising idea; Bowling 4 Neutrophils. This is something that every family can do and we are looking forward to everyone across America joining us in the fun! Read more on page 5 or visit www.bowling4neutrophils.org.

In conjunction with Bowling 4 Neutrophils we formed an alliance with the National Neutropenia Network (the NNN). Lee Reeves established the NNN, and it is a huge support system for all families affected by Neutropenia. The NNN brings together families and Neutropenia experts at an annual world-class conference. Although the Ella Jewell Foundation and the NNN are separate organizations, we are working as a team to strengthen our community. As the Ella Jewell Foundation raises funds through Bowling 4 Neutrophils, we will donate 25% of our net profit to the NNN to help fund peer-support programs.

With help from the families that have joined in our efforts, the Ella Jewell Foundation has donated over $100,000 as of December 2012 towards the research being conducted by Dr. David. C. Dale's research team at The University of Washington in Seattle. We strongly believe in the work that Dr. David C. Dale and his team are doing, and we are grateful for their dedication to the field of Neutropenia. We want to do all that we can to assist them in furthering their research. We strongly encourage you to either form a team of your own or host an event dedicated to the Neutropenic in your life. We would be happy to assist you in anyway that we can. Because, together, we can make a difference! We have high hopes for the Ella Jewell Foundation, lets do this together!

Message from Dr. Dale:

By: Dr. David C. Dale

Neutropenia Update: Twenty-five Years of G-CSF for Severe Chronic Neutropenia

We have been very busy for the last year in our studies on neutropenia. We continue to enroll new patients in the Registry (known as the Severe Chronic

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Dr. David C. Dale , M.D.

Neutropenia International Registry or SCNIR). Through our offices in Seattle and Hannover Germany, we maintain records and contacts with about 2,000 patients scattered over more than 60 countries, but chiefly in North America and Western Europe. It is a lot of work keeping up with medical records, blood counts and events in the life of so many people, but we have been doing this for 25 years! It was in the summer of 1987 that we began the studies which led to the SCNIR!

In our recent work we completed and submitted for publication a study of the diversity of mutations in the gene for neutrophil elastase, called the ELANE gene, which causes most patients to have the inherited conditions called "severe congenital neutropenia" and "cyclic neutropenia." We've identified now more than 100 different mutations in more than 300 patients, creating the largest data base for correlations of the genetic mutations and patient outcomes. We have found that there are some mutations cause mild disease and some mutations are more likely associated threat of leukemia or death from infections because of a poor response to granulocyte colony stimulating factor or G-CSF. In ongoing research we are trying to understand why this happens and the specific cellular and molecular basis for the different types of neutropenia.

Within the last few months we have also discovered a new genetic mutation marking the pathway for patients to evolve to develop leukemia; our colleague Dr. Karl Welte in Germany led these studies. This information should be very helpful in identifying patients at risk and correctly identifying patients in urgent need of stem cell transplantation. Because the risk of complications with transplantation still remains relatively high, we do not recommend transplantation to all patients. Understanding risk factors and mutations in the pathway for developing leukemia is a way that we are trying to assist physicians around the world in caring for patients with severe neutropenia.

We are very engaged also in trying to understand the cause for neutropenia in patients who do not have any known cause, but where we have a strong suspicion that the disease has occurred because of a specific mutation. Recently we believe we have found an additional rare causes for inherited neutropenia. We expect in the next few months this work will all be published and become part of our panel of studies to define the cause of neutropenia.

A few weeks ago, we helped produce a new documentary on neutropenia for the PBS series, "Health Mind, Healthy Body." This is the second time we have been invited to contribute to this series; the first time was apparently a big success and the program was shown in many stations across the country. This is part of our efforts through public education to improve diagnosis, understanding and treatment of neutropenia. The McGuinness family, as well as other families and patients, contributed to this effort and the Ella Jewell Foundation will be mentioned in the program. We hope that you will watch for this program on your PBS station.

In summarizing our research work, it is important to note that we are working cooperatively with a number of groups around the world to expand studies on the causes and treatment of neutropenia. Our research team at the University of Washington includes laboratory researchers- Vahagn Makaryan, Merideth Kelley and Elin Rodger. As well as a clinic group collecting and maintaining records on patients around the world: Audrey Anna Bolyard, Tracy Marrero, Lan Phan, Barbara Otto, and Amanda Jones. Our work in Europe is led by Drs. Karl Welte and Connie Zeidler and all of the work is overseen by a dedicated advisory board. We are currently supported by funds from the National Institutes of Health/National Institute of Allergy and Infectious Diseases, the Ella Jewell Foundation, and the Amgen Foundation. We are very appreciative of this support. We are dedicated to this work and the patients and families we serve.

David C. Dale, M.D.
Professor of Medicine
University of Washington

Message from Dr. Boxer:

By: Dr. Lawrence Boxer M.D.

As an investigator and a clinician with the Severe Chronic International Registry (SCNIR), the Ella Jewell Foundation represents hope for patients that I see as well as for many patients around the country with whom I consult. By supporting SCNIR, the Ella Jewell Foundation provides me with the ability to encourage patients that sometime in the future alternative treatments besides Neupogen (G-CSF) might become available to treat their child or in some cases their own neutropenia. As it turns out, about 5% of patients with severe congenital neutropenia do not respond to conventional doses of Neupogen and must undergo a bone marrow transplant. The basic research at SCNIR, headed by Dr. Dale, is examining alternative treatments including an oral treatment that might replace the use of subcutaneous injections of Neupogen. Many patients whom I consult or see as a new patient where the diagnosis and the cause of the neutropenia remains unclear, I have been able to identify new genetic mutations that are associated with their neutropenia with the resources provided by SCNIR. The funds provided by the Ella Jewell Foundation have allowed SCNIR to identify new causes of neutropenia and have provided a foundation for the research team at SCNIR to apply for further research funding from the NIH.

My particular role in support of the Ella Jewell Foundation has primarily centered on following the largest number of neutropenia patients in the United States. I have been privileged to identify patients with new causes of neutropenia as well as to identify patients with novel mutations for congenital neutropenia. I have received many emails and calls again from patients from all over the country for my advice regarding their diagnosis and treatment. In the later situation I try to work closely with the patient's treating physician to arrive at a solution for management of their neutropenia. Recently I had an opportunity to see a toddler from Connecticut. The family had contacted the McGuinness' to ask them for advice regarding whom they should take their patients to see. Following their advice, they sought advice from me and they came to Ann Arbor. Fortunately we were able to aid in the diagnosis of the cause of the neutropenia.

(l to r) Robert McGuinness and Dr. Lawrence Boxer

My hope for the future is that the foundation will continue to grow and expand beyond the region of the Northeast. It would be wonderful if many of the families that I follow in Ann Arbor could become acquainted with the activities of the foundation. There is so much need to provide hope for all patients with chronic neutropenia.

The long race 4 Tate:

By: Gretchen Long

In August 2011, when our son Tate Long was 17 months old, we found out he had Neutropenia. In that moment, the explanation for his chronic illnesses felt like we had crossed the finish line.

We quickly realized we had only finished our training for a much longer race. We were at the starting line and the finish line, a cure, was nowhere in sight. While thankful for the G-CSF injections, we knew the medication wasn't a cure. We refused to stand by waiting for answers, awaiting a cure for our son. We had to become part of the solution.

LONG RACE 4TATE

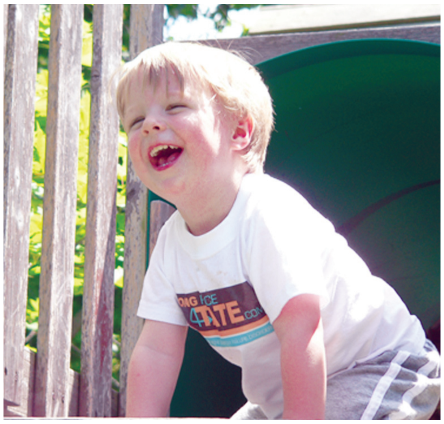
In order to breathe easier each day and rest easy each night, we had to be proactively involved in finding a cure.

We are not doctors or nurses or scientists. We are not independently wealthy. But we can raise money for research – starting with asking our friends and family. We were thrilled to quickly find the McGuinness family. Knowing there was another family out there who immediately decided to take action, and had already started a nonprofit dedicated to research gave us something very positive to focus on, even as we were grappling with understanding Tate's condition. All we needed to do was figure out how to help the cause.

There's a reason things happen – even if you don't know it right away. Tate's dad, Thad, started distance running soon after Tate was born. We decided that any race he runs will be dedicated to Tate in order to raise money for the Ella Jewell Foundation mission. Now all we had to do was ask our friends and family to "sponsor" Thad in his upcoming races by donating to the Ella Jewell Foundation. Longrace4tate.com was born!

We are still amazed as we reflect back on 2012. Not only did our friends and family donate, they told their friends and they dedicated their races to Tate. Our personal network of donors grew exponentially within weeks. The momentum has extended to other families. The McGuinness family and friends raced for Team Ella Jewell. The Lewis family and friends teamed up for Team Henry.

Altogether, in 2012, there were 26 members of Team Tate who competed in 17 races and raised more than $22,000. Team Ella Jewell and Team Henry brought in more races and altogether we raised more than $48,000!


Tate Long

The momentum continues. 2013 kicked off with Paxton's family and friends starting a team. The Long Race 4 Tate is not a single event; it's not a pledge per mile. It's not even really "for Tate." Not a single dollar goes towards Tate's medical expenses. Every penny we raise goes to the Ella Jewell Foundation, which donates all proceeds towards research for a cure.

As the race continues, all we need is you! Whether you walk, run, cycle or bike, it's easy to join this effort.

Interested? Contact Gretchen Long (longrace4tate@gmail.com or 330.242.9458).

Team Henry:

Raises $6, 920 Dollars for Research and Awareness:

By: Mariel Lewis

On November 3, 2012 Nate and Mariel Lewis joined with family and friends and ran their first 5K race in honor of their son Henry to raise funds for Severe Chronic Neutropenia. Their journey with neutropenia began when Henry was 9 months old. He has since been diagnosed on the spectrum between Cyclic and Congenital Neutropenia. Approaching three years old, Henry receives daily G-CSF and does very well.


Henry Lewis

Mariel Lewis serves on the board of directors for the National Neutropenia Network and was thrilled to join forces with the Ella Jewell Foundation and Long Race 4 Tate. All proceeds raised through team Henry were divided equally between the Ella Jewell Foundation (EJF) and National Neutropenia Network (NNN). In order to help mobilize our very small but mighty neutropenia community, EJF invites anyone to participate in the Long Race 4 Tate or create a fundraiser of your own and designate a portion of the proceeds to NNN.

Christian Lakis event:

Lakis Family Raises over $10,000 dollars for Research!

By: Kristin McGuinness

On March 14, 2013 the Ella Jewell Foundation and the Lakis family hosted a fabulous event in honor of Christian Lakis at Morton's Steak House in Great Neck, NY. Christian's parents, Jennifer and Greg, worked very hard to put together an amazing evening for their friends and family to honor their son in a very special way. The Lakis family raised over $10,000 dollars for the Ella Jewell Foundation. Guests enjoyed

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Christian Lakis event:

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an evening of fabulous food, cocktails, silent auction items and a heart-felt speech made by Jennifer Lakis.

Christian Lakis was born on October 8, 2009 and diagnosed with Severe Congenital Neutropenia. Jennifer Lakis understands the struggles of living with this disease, because she too is diagnosed with Severe Congenital Neutropenia. Jennifer's journey with Neutropenia began in the days before the life-saving medicine, G-CSF, was available for Neutropenic patients. Despite their diagnosis, Jennifer and Christian are doing well and living a healthy, beautiful life.

In October 2011, Jennifer and Greg attended an Ella Jewell Foundation event. They immediately became interested in joining forces with the Ella Jewell Foundation. They wanted to do all that they could to raise awareness and funds for research. They began talking with their family and friends and sharing their goal of hosting an event with the Ella Jewell Foundation in honor of their son. Their entire family was eager to get involved, especially their 5 year old daughter, Bella. The evening was amazing. The Ella Jewell Foundation congratulates the Lakis family on their great efforts and wonderful success.

The Lakis family is already thinking ahead and working on organizing a 2014 event. We are excited and honored to have them as a part of our team.


Christian Lakis

Team Paxton:

Raises $1,500 Dollars for Research

By: Kristy Adams

December 8, 2011 was the day our lives were changed forever. Our daughter, Paxton's, blood work returned saying she had zero neutrophils. Paxton was 18 months old at the time. After a few months of blood tests and constant fear, she was diagnosed with Autoimmune Neutropenia. Since then we have been on an emotional roller coaster. I never could have imagined our beautiful daughter would be diagnosed with a chronic illness. Even with a diagnosis, we have so many unanswered questions and unknown factors. As parents, we want to protect our children and keep them safe. If I could take this from her, I would. Since that isn't possible, our family is driven to do more to help families battling Neutropenia. It is important to us to help raise funds for Neutropenia research because we need a better treatment, a cure and answers. We were so excited to learn about the Ella Jewel Foundation and the Long Race 4 Tate. This provided us with an easy way to join the cause! So now, we are racing to raise awareness. We are racing to find new ways to raise funds for Neutropenia research. We are racing for Paxton and everyone else who is affected by Neutropenia.


Paxton Adams


Kara and Kate Loader

Loader Family:

By: Tammy Loader

The Loader family pitches in to raise money to find a cure for Neutropenia.

After attending the 2011 National Neutropenia Network (NNN) Family Conference, Katie, age 10, and Kara, age 7, set a goal to raise funds to find a cure. At the closing session of the conference, Katie heard Kristin McGuinness share information about the Ella Jewell Foundation. Katie, who has watched her sister live with Neutropenia, sprang into action. For the entire five hour trip home from Michigan, she came up with idea after idea of ways to raise money and awareness for the cause. Katie's hope is to help raise enough money to find a cure and keep raising money to ensure that even kids in "Third World Countries" can have access to the cure.

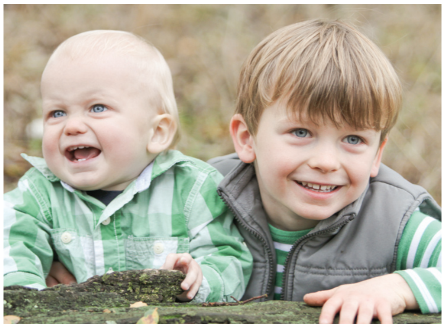
With the support of their parents, Bill and Tammy, the girls shared their plans with their small church, Christ Evangelical Lutheran Church in Cambridge Ohio. The church chose to take up collections during Sunday School to help the girls. The ladies of the congregation held a purse party and a collection was taken at the annual Easter Breakfast. As a result of their efforts and the support of their community, they raised over $1,600 for the Ella Jewell Foundation. In October the Loaders held their first annual, "Harvest A Cure" sale for Neutropenia. Despite drought conditions, pumpkins, gourds, and sunflowers were sold and $1,800 was raised for the Ella Jewell Foundation.

"DASH"ing Toward a Cure:

By: Victoria Taylor

Dash was diagnosed with Severe Congenital Neutropenia when he was 6 months old and soon after he started the only recommended treatment, G-CSF injections. His parents were devastated to find out that his body was not responding. Without neutrophils, Dash was at very high risk for infection and more serious complications. After meeting with numerous Bone Marrow Transplant teams across the US, Dash's parents made the difficult decision for Dash to undergo a bone marrow transplant in January 2013, 6 months after diagnosis, just after his first birthday. Dash received bone marrow and cord blood from his older brother, Declan and is doing well today. The road to recovery after Bone Marrow Transplant is a long and difficult one. Dash's family is dedicated to support other families dealing with Neutropenia by raising funds for the Ella Jewell Foundation so that research can move forward to provide other treatment options and someday, a cure.

Westarity is a not-for-profit organization established by Westward Management, a Chicago based residential property management company. On Saturday, May 18th, Westward supported several employees in their participation in the Chicago Tough Mudder Competition. The race was a 10-12 mile obstacle course designed by British Special Forces to test strength and stamina. The team raised $500 at the Tough Mudder Event, and all funds were donated to the Ella Jewell Foundation in support of Neutropenia research. "Dash"ing toward a cure!



Dash and Declan Taylor

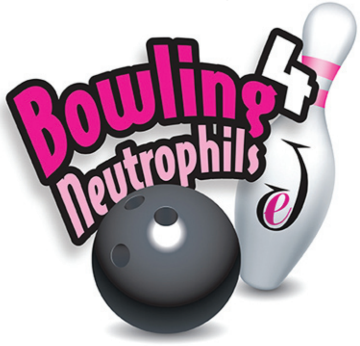
Bowling 4 Neutrophils:

Join us Across America for a Bowling Fundraiser Event. Proceeds benefit the Ella Jewell Foundation. This is an invitation to join an easy, fun fundraising event. You will be assisting in raising neutropenia awareness and donating towards neutropenia related research.

Have a great time bowling in honor of the Neutropenic in your life! Together, we can make a difference.

How to get involved (10 easy steps)

1) Pick a bowling alley near your home
2) Put together a team (or teams) of 4-6 players
3) Name your team(s) (example: " Team Ella Jewell")
4) Pick a captain to oversee your team(s)
5) Plan to play on any date between December 11, 2013 & December 15 2013.
6) Each player will pay a $10 registration fee to the Ella Jewell Foundation (the team captain will organize this collection).
7) Set a fundraising goal for your team: minimum $250 per team
8) Ask friends and family to go to www.bowling4neutrophils.org to sponsor you and your team by making a tax deductible donation.
9) Meet your team on the date chosen between December 11, 2013 and December 15, 2013 for bowling fun.
10) Team Captain, please contact Kristin McGuinness or Victoria Taylor (admin@ellajewellfoundation.com or (203) 664-1385) by NOVEMBER 1, 2013 to set up your team page and any additional questions. Be prepared to supply the following
    1) Team Name
    2) Fundraising Goal
    3) Location of Bowling, date and time
    4) Team Players Names
    5) A picture of each of your teammates.
    6) Shirt sizes of team members



www.Bowling4neutrophils.org

Prizes will be awarded to the:

1) Team that raises the most funds.

2) Best individual bowler (Male/ Female/Child)
3) Team with most creativity in raising awareness.

Prizes to be announced

Featured article of all winners in the Ella Jewell Foundation 2014 Newsletter

Upcoming events:

8/18/2013:
Team Tate:
Vermilion Harbor Triathlon

8/18/2013:
Team Ella Jewell:
5K Run at Yankee Stadium

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Welcome New Committee Members:

Jodi Barry
Grace-Marie Chapin
Jinny Garton
Victoria Taylor
Brittany Mothershead

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To All Those Affected by Neutropenia
Support Neutropenia Research!

The Ella Jewell Foundation is a Connecticut based Non-Profit recognized by section 501 (c) 3 of the Internal Revenue Code. All donations are tax-deductible.

Raising Funds & Awareness For Neutropenia Since 2010.

Ella Jewell and sister Alexa

We are always looking for stories for future Newsletters. If you hold a Fundraising Event, send us the information. If you have a story about an experience with Neutropenia, let us know. We are also looking for photos of people showing support for the Neutropenic in their lives. All submissions can be emailed to **ADMIN@ellajewellfoundation.com**