ELLA JEWELL FOUNDATION

SUPPORTING NEUTROPENIA RESEARCH

Summer 2011

What is Neutropenia?

Neutropenia is a very rare blood condition that causes a reduced number or complete lack of neutrophils. Neutrophils are a type of blood cell that defends the body against bacterial infections. Sometimes the term Severe Chronic Neutropenia (SNC) is used to describe a group of conditions in which Neutropenia is the primary problem. Types of Neutropenia include Congenital, Idiopathic, Cyclic and Autoimmune.

For more information visit the National Neutropenia Network @ www.nationalneutropenianetwork.com

Ella Jewell Foundation Dinner Gala (Left to Right) Dr. David Dale, Ella Jewell McGuinness, Kristin McGuinness, Dr. Lawrence Boxer and Robert McGuinness.

Ella Jewell Foundation
Wine Tasting Event
Friday, October 21st
7:30 pm
Shenorock Shore Club
Rye, NY

Join us for an evening of Wine Tasting, Music and a Fabulous Food Selection.
$100 Dollars per Ticket
Space is limited; please reserve your tickets now @ www.ellajewellfoundation.com. Please go to the google check-out on the donations page to buy your tickets and be sure to e-mail us @ admin@ellajewellfoundation.com letting us know the full names of each guest attending. You can also send a check made payable to:

Ella Jewell Foundation
P.O Box 396
Bethel, CT 06801

Invitation to Follow

ELLA JEWELL DINNER GALA 2010

Our daughter, Ella Jewell McGuinness, was born on September 2, 2009 and shortly after diagnosed with Severe Congenital Neutropenia. Ella's diagnosis was devastating to our family but we decided to stay positive. We are very fortunate and Ella responds well to daily injections of Neupogen. Since this is currently the only effective medication available, we hope that she continues on this path. When we were first learning about Neutropenia, we quickly realized that many consider neutropenia an "orphan" disorder, unknown by many and unfortunately under researched.

In early 2010 we formed a Board and Committee and started The Ella Jewell Foundation. The Foundation's mission is to raise awareness and funds for research. We believe in our hearts that a cure will one day be found and that together we can make a difference.

Although the Ella Jewell Foundation is named after our daughter, it has been established to benefit all families affected by Neutropenia.

On November 5, 2010 The Ella Jewell Foundation held a Dinner Gala at the Westchester Marriott in Tarrytown, NY. Over 250 guests enjoyed cocktails, dinner, dancing, a silent auction and raffle prizes. We were honored to have neutropenia specialists Dr. Lawrence Boxer and Dr. David Dale attend the event. Hematologists from New York University also attended along with many doctors from the New York area. Dr. Lawrence Boxer spoke to the guests, educating them about Neutropenia. I, Kristin McGuinness, explained the emotional toll that comes along with having a child diagnosed with Neutropenia.

With a lot of hard work our team pulled off an amazing evening. As the parents of Ella Jewell, this was and always

Gala continued on page 2

Gala from page 1

will be one of the best evenings of our lives.

On that night we raised $30,000 and subsequently donated it towards neutropenia research being conducted at The University of Washington, led by Dr. David Dale.

We want to encourage others to help raise funds for research. Many of you out there also have great ideas on how to raise awareness and funds. Let's collectively share our thoughts and our experiences and work together to raise funds that are desperately needed for research. We would be happy to talk to anyone interested in pursuing Neutropenia Fundraising.

We would like to thank everyone that helps and supports The Ella Jewell Foundation and we ask that you please continue to do so.

Ella Jewell Foundation Goals

- Raise Awareness
- Raise Funds for Research
- Encourage other families affected by Neutropenia to raise funds for research.

"Together, we can make a difference"

Research Report

Neutropenia research being conducted at The University of Washington, Seattle WA.

Reported by Dr. David C. Dale

Research report:

We want to learn how mutations in one gene, the gene called neutrophil elastase, cause neutropenia, severe infections and predispose to leukemia.

With support from the Ella Jewell Foundation, we have found more than 60 different mutations in this gene. We are currently analyzing the relationships of the mutations to the disease severity.

We have laboratory evidence suggesting a new and alternative therapy for neutropenia due to elastse mutations. We want to find an alternative to the only current treatment G-CSF or Neupogen.

With support of the Ella Jewell Foundation, we have extended these studies and have submitted new proposals to the NIH and industry to proceed with development and clinical trials.

In other work, we have discovered a potential therapy for a rare for of severe neutropenia called WHIM syndrome and are proceeding with clinical development. We just presented this work and have submitted the results for publication.

We are working cooperatively with several groups to analyze and understand neutropenia in other diseases including, Shwachman-Diamond syndrome, Barth syndrome and diseases affecting glucose metabolism.

We developed and submitted a two major grants to the NIH for support for research on severe neutropenia. The grant brings together laboratories at the University of Washington and the Fred Hutchinson Cancer Research Center in Seattle and Washington University in St Louis. The focus is on understanding the basis causes for neutropenia and the routes to better diagnosis and treatment. This is a difficult time for new grants, but we are very hopeful and optimistic.

McGuinness family attends National Neutropenia Conference 2011 :

This past July, we attended a three day conference held by the National Neutropenia Network in Ann Arbor Michigan. During the conference we learned more about neutropenia, learned of new advances in the treatment of neutropenia and met with families that are also affected by the disease. We were again reminded how serious neutropenia is and how there are too many that struggle with the disease. We met with families that are preparing for Bone Marrow Transplants due to their child's low response to the medication or because the child has developed leukemia. A Bone Marrow Transplant is currently the only cure for neutropenia and, at this time, carries many risks. Doctors discussed the hope that one day this procedure will not carry such great risk. We were hopeful to learn that a new medication may be available in the future. Currently the only medication that treats neutropenia, Neupogen, does not work for all patients and also has side effects. We took both positives and negatives away from the conference. We feel more motivated than ever to contribute towards raising funds for research. We pray that Ella continues to do well and that one day she and all children affected by neutropenia are cured. We feel blessed that we had the opportunity to meet with great doctors and families. We ask that you please continue to support our cause. You all give us a great amount of hope and together we can make a difference.

Baby John's Story

by: John Merchlinsky III

This story is about our son John J Merchlinsky IV, John was born on August 31 2009 from a normal pregnancy and full term. Things were pretty much the standard for a newborn baby. Around six months of age John started developing repeat ear infections. John's pediatrician would put him on antibiotics and we would run the course of the antibiotics and the ear infection would go away, but shortly after he would get the ear infection again.

Around the same time John started getting lesions on his skin which the doctors said were staph infections, they would come and go and be on various places on his skin, including his face, back and groin areas. John spent his first Thanksgiving in the hospital being treated for an anal fistula which required him to have the first surgery of his life. John recovered from the surgery but was still getting the lesions and ear infections.

By this time, Angie (mom) and Me (dad) were getting frustrated with the doctors because of their inaction in not wanting to run any tests despite the fact that we thought that there was something wrong. So we decided to try a new route with a new doctor, we called Geisinger Medical Center.

Our first trip to Knapper Clinic was an uneventful one, Baby John had blood tests run and we went home. A few hours later we received a phone call from his doctor, we needed to bring John back to the hospital he was being admitted because his blood tests were way off. To start his counts looked like malnutrition, and his absolute neutrophil count was zero. And so began this odyssey, we were admitted for several weeks while they ran all kinds of tests and blood work.

The doctors had many ideas but no concrete definition. They had drawn blood for the ELA gene test but told us that it would take a few weeks, so I guess it was just a waiting game. We talked to the doctors about having a second opinion at The Children's Hospital of Philadelphia (CHOP) . We got to Spend Christmas at home, but shortly after we were back in the hospital. We traveled to CHOP and met with the doctors and immediately it was mentioned that it was Severe Congenital Neutropenia. Our suspicions were confirmed when the ELA gene test came back positive for the mutation. The doctors started John on medication, Neupogen it was not working for baby John and was the only medication available to treat him. Although very dangerous, a Bone Marrow Transplant was our only option to save our child's life. We finally had the answer, we were scared. There were follow ups and travel to Philadelphia. We had his three year old brother Cameron tested and it turned out that he was a perfect match. Hope and relief were the emotions we were feeling then.

Things moved rather quickly from there and we had our transplant on April 27th 2011, and here we are several weeks removed and his counts are improving. He developed Graft vs. Host Disease (GVHD) and we are working through that but we are getting better. The thing that we cannot express the most is that without the Bone Marrow transplant things might not have gotten better, we might still be looking for answers, we have a long road ahead of us and we are not out of the woods yet. We constantly thank God, the doctors, the foundations, and everyone we have met and who have helped us along the way.

ts mentioned above.

A very special Thank You to Erik Bodelsen for Designing and setting up our new Website!

Check it out at www.ellajewellfoundation.com

Ella Jewell Foundation
P.O. Box 396
Bethel, CT 06801

Build Rate
US Postage
Paid
Permit No.
00000

Mailing Address
Street Number and Name
City, State 98765-4321

SAVE THE DATE!!!

Ella Jewell Foundation

Wine Tasting Event

Friday, October 21st
7:30 pm

Shenorock Shore Club
Rye, NY