



national neutropenia network

awareness, education, research and support

A Letter from Lee

"I've had neutropenia for 20 years, but I've never met anyone with it. When I walked into the hotel lobby filled with people registering for the Conference, I suddenly felt nervous. I had to catch my breath as I realized I was surrounded with people who have neutropenia just like me." These are the words of a first time attendee at the Neutropenia Family Conference in July. An emotional experience for many, the Conference was also a time of sharing and learning in a safe and comfortable atmosphere.

In addition to participating in a weekend chock full of information, the NNN board members met with physicians to discuss issues of common concern. With the wildly diverse and often misguided advice given on social networking sites regarding hygiene and infection prevention, there was agreement that we should publish guidelines on these topics. Dr. Newburger will initiate an effort to update the Patient Handbook and include specific infection prevention guidelines. There was also discussion on the difficulty of finding physicians with experience in treating chronic neutropenia. This has been a problem for years. Patients often tell stories of frustrating experiences going from doctor to doctor before finding one who understands the challenges and complexities of treating chronic neutropenia. We plan to develop a list of physicians in the coming months to publish on our website.

Our next Conference is set for the weekend of July 11, 2014 in Ann Arbor. We will be skipping a year for the first time in nearly a decade, but the National Neutropenia Network will not be standing still. We are discussing several new programs as well as working to strengthen current offerings such as the Peer Support Volunteer initiative, outreach programs such as the newsletter, the website and a social networking presence. We are considering the possibility of creating named funds to honor families and individuals who wish to support the Network. We're exploring ways to work with the Ella Jewell Foundation.

Our board is growing in numbers and talent. With new leaders in place the NNN will benefit from fresh ideas and energy. We are pleased to welcome Rajan Lakhia DO who has served for ten years as a hospitalist with The Christ Hospital of Mt. Auburn Ohio. His also serves as the hospital's associate internal medicine residency director, ethics chair and palliative care medical director. He is currently working on his masters in bioethics and healthcare policy. His seven-year-old son Liam has congenital neutropenia. Raj and his wife Tanya, a nurse, want to be active participants in the NNN. They want to assure that Liam and all neutropenic individuals have the benefit of a strong and relevant support organization now

Cont. on page 13

WHAT'S INSIDE

2012 Neutropenia Family Conference Recap

Chronic Trauma and the brain	3
Coping Skills for Challenging Times.....	3-4
My Favorite Cell.....	4-5
How to Get the Most out of your Doctor Appointments	5
Extremism in Defense of Health in Neutropenia.....	6
The Severe Chronic Neutropenia International Registry	7
NNN Update	8
Four in Forty	9
Breakout Sessions	10-11
Kids Camp	12

A Letter from Lee Cont. 13

How Can We Help 14-15

The National Neutropenia Network newsletter is published several times a year. Lee Reeves, NNN Executive Director: lee@neutropenianet.org P.O. Box 1693 • Brighton, MI 48116 • 517.294.0736 • neutropenianet.org

Many thanks to Mariel Lewis, owner of Trapdoor Design, for updating the look of our newsletter. She also designed an attractive fundraising brochure which is now available to anyone wishing to raise funds for the NNN. (Contact lee@neutropenianet.org) We are most grateful to Mariel for making a difference.

2012 Neutropenia Family Conference Recap



The conference kicked-off on Friday night with a bittersweet testimonial from Steve R. His son Mitch was born with neutropenia. The young family struggled to keep him well and fought hard to keep Mitch from death's clasp.

Tearfully, Steve shared with those in attendance about his long nights of researching his son's bone marrow failure syndrome and how at that time the survival rate was dismal. Devastated, he held out hope that his son would be a survivor.

When Mitch turned two, Steve received the most helpful news he'd ever heard: a drug had been developed. The drug was successful in creating those most sought-after neutrophils and the human drug trials soon would begin. Steve immediately signed the papers for Mitch to partake in the studies

because even the unknown specifics of the drug paled in comparison to what they already had faced.

Soon thereafter, the family received a call from Dr. Laurence Boxer MD, professor of pediatrics and communicable diseases at the University of Michigan, who was one of the expert physicians studying neutropenia and helping to spearhead the drug trial. He told Steve that unfortunately Mitch had been selected for the control group and would have to wait to receive the promising new medicine. The family was devastated. All that hope and anticipation they had for their young son to become healthy and have a future had been shattered in that moment. With a few close calls they made it through the six months trial required for patients on the control group. Then they got the wonderful news—the medicine was proving effective and Mitch could begin to take it.

Thanks to regular injections of G-CSF Mitch has neutrophils and can fight infection. He has led a full and rewarding life. In fact, just this May, Mitch graduated law school from the University of Michigan, "The very institution that saved my son's life, kept him living, and 23 years later he chose it for law school," Steve choked through his tears. "If there is one piece of advice that I can offer, it is to have hope and remain optimistic. As difficult as all this is, you can do it and your child can make it."

Mark your Calendars now for the 2014 Family Conference the weekend of July 11-13. We plan on having a full day of programs on Friday, and the private consultations with doctors will occur Saturday afternoon. Once again we will be at the beautiful Marriott Eagle Crest in the Ann Arbor/Ypsilanti area. Our room rate is \$110 plus tax. The Conference fee is not yet determined. Watch our website for more information. Registration will be available in the fall of 2013.

Chronic Trauma and the brain

The first Friday session featured psychiatrist Shelley Uram, MD who discussed how chronic and severe health conditions can be traumatic and how it all combines to affect the brain. She said that the level of trauma a human will experience is dependent upon that individual's brain and how they perceive the event. The accuracy of that perception does not matter, she added. Interestingly, she shared that a person's reaction to similar situations is determined by that first experience. For example, a young child undergoes a medical procedure and becomes frightened because of the pain he felt, the lack of control he had over the situation and his lack of understanding that the procedure is to help him and not to harm him. Thereafter, the child unconsciously may react the same way later in life that he did in that first occurrence of the event.

"Your brain makes up the story," she explained. "When the implicit memory gets triggered, you are not remembering it you are becoming it. The 'there and then' is constantly hijacking the 'here and now,'" she added.

She advised that patients of severe, chronic illnesses and their caregivers could benefit from seeing a therapist and undergoing what is termed "bottom-up therapies." This type of therapy enables the stimulation of the innermost parts of the brain (starting with the brain stem) and works up from there to essentially retrain the brain to better cope with the perceived traumas it has experienced over the course of that person's lifetime. She suggested that instead of the typical cognitive behavioral treatment, that the therapy be based more on somatic experience (SE) and eye movement desensitization and reprocessing (EMDR).

"Parents, the more you can clean-up your own baggage, the better you will be able to care for your child. If a family has insurance that covers 10 sessions a year, I'd want to use eight of them on the parents," she concluded.

Coping Skills for Challenging Times

Shay Jones, MS, LPC, led a "Getting to Know You" session. Shay has congenital neutropenia, and it wasn't until he read a newsletter in 2005 that he'd first seen a photo of another person with congenital neutropenia. The photo was of Leta Reeves, daughter of NNN executive director, Lee Reeves.

Shay asked attendees to answer questions about how they each cope with neutropenia in their lives and asked them to share within their respective diagnosis groups (i.e. congenital, cyclic and idiopathic/autoimmune). They shared their experiences and challenges with each other.

"The scientists tell us we are one-in-a-million, but here we can be a dime-a-dozen. Feels good, doesn't it?!" said Lucy Lyman, who serves chairperson of the National Neutropenia Network Board of Directors. Lucy has cyclic neutropenia, as do two of her three daughters. Interestingly, her father also has the gene mutation for cyclic neutropenia but has been asymptomatic his whole life. "I want to thank the doctors who have dedicated their lives to this disease," she continued. "I can't even say Dr. Dale's name without getting emotional and choked-up."

"Neutropenia has been the greatest monster of my life. I was born with it and I will die with it. The first person I met with neutropenia was my daughter. It has been my greatest teacher, but I am scared. I often wonder what would happen to my husband and kids if something happened to me."



2012 Neutropenia Family Conference Recap



Below are several other exchanges:

“It is a relief to be amongst other parents who understand. I’ve become so worn down. I feel my character is building and I find myself tearful in certain settings and situations.” – Karl M.

“Every year I have learned more and more from attending this conference. I feel great and I feel blessed. I have a great doctor and a great support system. Neutropenia has always been a monkey on my back. I was kicked out of high school because of it. I began Neupogen injections when I was 17. Neupogen has brought me my second life.” – Aaron H.

“I knew when I had a baby that I could have a baby with neutropenia. Still, when Olivia was born, I was crushed when she was diagnosed.” – John S.

Attendees listened intently to each other, soaking up every word. Finally, they were in a setting where others really understood everything they as patients and as parents have been feeling. The relatable storylines that they each had experienced were met with understanding ears and open hearts.

“There were some very powerful exchanges,” Shay noted. “Neutropenia interweaves itself into every aspect of our lives.”

My Favorite Cell

Saturday’s sessions kicked off with a presentation titled, “My Favorite Cell,” by Peter Newburger, MD, professor and vice chair for research, Department of Pediatrics, University of Massachusetts Medical School.

Dr. Newburger showed the attendees how the circulating neutrophil in the bloodstream is just one part of the whole life of a neutrophil. He explained the varying points of maturation arrest in the different types of neutropenia, and he shared slides on where exactly those places of maturation arrest occur.

A bone marrow examination is the most telling study when a patient is suspected to have severe chronic neutropenia (SCN) as it likely will reveal if a maturation arrest occurs. Annual bone marrow studies are advised for severe congenital patients to screen for conversion to myelodysplasia (a pre-leukemic condition) or leukemia; however, annual bone marrow studies are not necessary for those with other forms of neutropenia (e.g. idiopathic, autoimmune, chronic benign).

Dr. Newburger explained the process of neutrophil development, including how the neutrophil flows through the blood vessels and then sticks to the vessel wall before it exits through the vessel wall. If bacterium is present, the neutrophil chases and consumes it. “Neutrophils are ready to give their all for their country,” he said, jokingly. Another interesting aspect he shared is that when the neutrophil generates reactive derivatives of oxygen, they combine with chloride (i.e. salt) to create the active chemical in Clorox bleach!

He discussed the application of G-CSF as a stimulant to create these most coveted cells and he shared how the various stages of bone marrow failure will depict how much of the drug is needed and how long it will take for a patient to respond to the drug.

“In fact, it’s almost a diagnostic tool to see how quickly the body responds to an injection of

G-CSF,” he said. This is because, for example, patients with congenital neutropenia will take much longer to produce circulating neutrophils. Patients with autoimmune neutropenia usually will respond immediately because every point of the neutrophil production cycle occurs in a healthy manner up to the neutrophil being released to the bloodstream (where it then is destroyed by an antibody).

As anti-neutrophil antibody tests are very difficult to perform reliably, and may vary considerably from one laboratory to the next, he recommended asking your doctor to send the test to a very experienced lab, such as the Blood Center of Wisconsin, in Milwaukee.

How to Get the Most out of your Doctor Appointments

Dr. Mary Ann Bonilla, MD, assistant professor of pediatrics and pediatric hematology/oncology specialist at St. Joseph’s Children’s Hospital, Peterson, NJ, was up next with how best to work with your physician. “It is a basic human need to understand and be understood,” she first explained.

Her tips include:

- It is up to the patient to fully inform the physician of all their symptoms.
- Communication is key.
- Ask other professionals including nurses, child life specialist, etc., because their experiences can help you make informed decisions by gaining a broader knowledge base.

“Getting a second opinion is your legal right. If a doctor refuses to allow a second opinion, consider if the physician is worth staying with,” Dr. Bonilla warned. “You have to do whatever you have to do to walk away at peace with your knowledge and with your treatment. The last thing you want to do is say, ‘I should have.’”

“Just as you find the camaraderie with other people, I also enjoy getting to meet you. I walk away thinking of your questions and your challenges, and I think of how to address them.” – Dr. Mary Ann Bonilla



John S. has attended the Neutropenia Family Conference four times. He said what keeps him coming back is the fact that there is always something new to learn. “I love seeing all the other people who are here, especially those with neutropenia. It’s kind of like a family, too, seeing some of the same faces over the years. I always sign-up for a one-on-one with the experts and it’s nice to connect with them; I’ve been part of the studies through the Registry since the 1990s.

“I also am here to show my support. We only have momentum as a group. I would hate for us to one day wake up and it’s over. You know things don’t last forever. We’re so lucky to be here. You know, there’s life before Neupogen and all this support, and there’s life after. I’d never want to go back to that life before.

John’s wife (Jane) and two daughters (Olivia and Sophie) attended with him, as they have in the past. Her parents often attend, too. Sophie, his youngest daughter, usually attends Kids Camp with her sister Olivia who like her dad has cyclic neutropenia. This year, Sophie also attended some of the adult sessions. She said they helped her to better understand what her sister and dad experience with their health. “It was sometimes hard to follow but their slides and pictures helped me to follow along,” she said.

Extremism in Defense of Health in Neutropenia

Laurence A. Boxer, MD, professor of pediatrics and communicable diseases at the University of Michigan, addressed many common concerns in dealing with neutropenia. The main point to his talk is to remain calm throughout your experience with neutropenia and not seclude yourself or allow “extremism in the pursuit of cleanliness” to overcome you and your family’s habits. In particular, he said that anyone achieving an adequate level of neutrophils on Neupogen therapy should be able to have an entirely normal lifestyle, without taking precautions.

He noted that even for those with low neutrophil counts, the two most common infections are skin infections and pneumonia. Humans have more bacteria in the body than they have human cells, so most infections come from our own “resident” bacteria and it doesn’t do any good to wipe down the entire house or playground with Lysol.

Dr. Boxer advised that all infants and young children absolutely should be regularly immunized. He said that the safety and efficacy of both the dead and live strain vaccines are safe for those with neutropenia because the response to vaccines (and virus infections) depends upon lymphocytes, a type of blood cell unaffected in virtually all forms of neutropenia. “I can’t emphasize enough the importance of receiving all scheduled immunizations,” he said.

An interesting aside he shared is the fact that one-third of African Americans run a normal lower absolute neutrophil count (ANC), and that they now have linked this instance to a red blood cell protein that was found to protect against malaria. “We are not yet sure how that relates to white blood cells; however, it is an interesting finding,” he added.



As mentioned above, his main piece of advice is to live as normal a lifestyle as possible. “Extremism is not healthy,” he warned, “and excessive hand washing can damage skin barriers.” He offered the following tips for those patients successfully treated with G-CSF:

Prevention:

- Reasonable good hand washing
- Good hand and nail care
- Treat cuts with washing and an antibiotic ointment such as Bactroban

Mouth Care:

- Cleanings 2-4 times a year
- Regular brushing /flossing
- No need for prophylactic antibiotics before dental cleanings

Gastrointestinal Care:

- Good food handling
- The usual food safety guidelines are sufficient

Respiratory Care:

- Avoid exposure to sick or coughing people
- Get the annual flu vaccine

He emphasized that these are all common-sense measures applicable to anyone, with or without neutropenia!

He also conveyed the importance of no rectal temperatures for neutropenic patients because it could endanger the integrity of the skin around the anus, leaving the area susceptible to infection from fecal matter.

The Severe Chronic Neutropenia International Registry

Wrapping up the morning’s round of expert physicians was an annual update on the Severe Chronic Neutropenia International Registry (SCNIR) by David C. Dale, MD, professor of medicine for the Department of Medicine at the University of Washington and founding director of the SCNIR.

He discussed the Registry’s many milestones, explaining the SCN categories and subcategories, and sharing some of SCNIR’s research findings. He also credited the Registry’s clinical manager, Audrey Anna Bolyard RN for her knowledge and understanding of neutropenia, and her attention to patients’ needs.

There is great interest, he noted, for the SCNIR researchers in the family history of SCN patients, as well as great need for bone marrow samples of patients over the course of many years. Dr. Dale said they are “desperately interested” and that he would be happy to discuss with any patient’s physician the possibilities of donating marrow when already undergoing an aspirate.

He spoke of this year’s change in pharmacy from CVS to Crossroads for delivery of the G-CSF

(Neupogen), and shared slides on dosing guidelines for neutropenic patients. “With proper dosing a plateau can be maintained for an extended period of time,” he said.

Treatment Pearls:

- Start low on Neupogen dosing and work your way up
- Stick with consistent G-CSF administration
- Get consistent blood counts and bone marrow studies
- Maintain good health records
- Stay active with the National Neutropenia Network – “not only are you helping yourself, but a lot of other people, too,” he said.

Currently, Dr. Dale’s team is working on a safety study on G-CSF and pregnancy; the risk factors of leukemia in severe congenital neutropenia patients; genetic analysis to discover causes of SCN; new, targeted treatments; sub-registries for rarer forms of congenital neutropenia; and a new clinical trial to discover new, molecularly-targeted treatments.

“Your questions at this Neutropenia Family Conference provides guidelines to our studies at SCNIR,” – Dr. David C. Dale

DID YOU KNOW?

The SCNIR Advisory Board is made up of 18 MD/PhD members; plus one layperson – Lee Reeves, executive director of the National Neutropenia Network. Each year, the Board meets at the annual American Society of Hematologists (ASH) Conference. “Her insights have been invaluable to us,” Dr. Dale said of Lee’s participation.

The SCNIR has helped 1,158 U.S. patients since its inception in 1994. Moreover, 2,000 additional patients have been served in 60 other countries.



NNN Update

Lee Reeves expressed appreciation to board members present: chairperson Lucy Lyman and directors Mariel Lewis and Kate Bottiger. She also noted those who could not attend: Taylor Carlton, Silke Deeley and Sharon White. She recognized Kim Blanz who recently retired from the board, and was instrumental in getting the website up and running.

Lee then shared an update of the many happenings throughout the National Neutropenia Network (NNN). They include:

- **The volunteer peer support group.** Training is available and volunteers still are needed. Family members and caregivers also are encouraged to volunteer. "When people seek us out, the peer support enables us to stabilize them emotionally and then very quickly we move them over to the SCNIR," said Shay Jones, who heads the group and the trainings.
- A wealth of information on the **NNN website**, located at neutropenianet.org, including a downloadable emergency room protocol card in English and Spanish, and The Patient Handbook.
- A **social networking taskforce** has been initiated.
- A **new educational fundraising brochure** has been developed. Patient's may insert their personal story and journey with neutropenia, and share them friends and family.
- The NNN hopes to develop over the next year a **list of physicians** who knowledgeable about

treating neutropenic patients. The NNN is collecting names and geographical information of such physicians. See more information on page 15. The NNN also hopes to begin work with the four expert physicians in the development of continuing education units on the topic of severe chronic neutropenia for physicians and other healthcare providers. In addition, exciting news was shared that Dr. Boxer will be a speaker at the annual ASH Conference and will host a private session for the hematologists in attendance who would like to learn more about SCN.

"Ultimately we'd like to support research," added Lee Reeves. "As a patient advocate charitable organization, we have an interest in seeing that the groundbreaking neutropenia research we've seen in the past continues into the future."

The NNN awarded 9 scholarships for families to attend this year's conference. "We haven't had to turn anybody away," said Mara Lim, NNN Conference Coordinator. "The cost of the conference does not even cover the cost of the food, per person," Mara added. "We are thrilled to be able to keep the cost low for families. The NNN appreciates the support of patients, family and friends. If you would like more information on how you can become involved with the National Neutropenia Network, please contact Lee Reeves at lee@neutropenianet.org or visit us online at neutropenianet.org.



Saturday afternoon kicked-off with Four in Forty.

HERE'S THE HIGHLIGHTS:

Dr. Boxer – Bone Health

Dr. Boxer addressed the topic of bone health and neutropenia crediting Linda Dimeglio MD, MPH and associate professor of Pediatric Endocrinology/Diabetology at Riley Hospital in Indianapolis for research on bone loss as it relates to neutropenia. It is not clear why individuals with neutropenia may be more susceptible to bone loss. Data does not clarify if G-CSF contributes to bone loss, but low bone loss and vertebral fractures are seen in individuals with SCN prior to starting G-CSF.

He emphasized the need for DXA scans on the same machine. They should be done every year if abnormal in adults. There currently is not enough data to make recommendations for children under five but every two years after diagnosis is a reasonable time frame for DXA scans in children. It is important they are conducted in a center used to measuring children.

Dr. Boxer stressed the importance of children to lead physical active lives and for all patients to get enough calcium and vitamin D in their diet. More details on this topic can be found at neutropenianet.org. Click on Conference Highlights and scroll down to find several slide presentations on this topic.

Dr. Dale – Dental Health

The Patient Handbook, available on the NNN website, has all dental guidelines listed:

- G-CSF helps with inflammation of gums/gingivitis
- Use a fluoride mouth rinse first and then brush
- Brush in an up and down motion on each tooth

- Use an electronic toothbrush
- A mouth rinse made up of half-strength peroxide is more comfortable than alcohol-based mouthwash for people with mouth ulcers
- Be doubly cautious with orthodontic appliances as they can harbor bacteria and have rough edges that can cause cuts in the mouth

A University of Washington study showed that G-CSF early in life has benefits in maintaining gingival health and reducing loss of teeth later in life.

Dr. Newburger – Pain Management

It is better to treat the cause than to treat the pain.

- Use the least potent analgesic that is effective
- Combine drugs from different classes (e.g. ibuprofen + codeine)
- Use complementary/alternative medicine alone or in combination with analgesic drugs
- Continuous treatment is better than intermittent ("prn") dosing
- Lower doses of G-CSF taken more often reduces bone pain

He then discussed the differences between addiction (psychological) vs. dependency (normal and physiological).

Dr. Bonilla – Integrated Therapy

Complimentary alternative therapies can help the mind, body and spirit.

- Essential oils have many uses and are used by inhalation or topically
- Meditation helps with self-regulation
- Yoga helps to open your body through use of your muscles and breathing
- Massage therapy, reflexology, acupuncture and acupressure all may be helpful in relaxation

Help us serve you better. In an effort to improve and update our website, we have recently placed a survey online. Please take a minute and complete it. Visit neutropenianet.org. Click on any tab and you will see a feedback button on the right. Click on it and complete the survey. Thank you—it will make a difference.

Breakout Sessions

Conference goers were able to spend time in a breakout session geared solely on their respective diagnosis. The attendees who are patients, or loved ones of patients, with congenital neutropenia attended an informational session with Peter Newburger, MD.

Dr. Newburger went over the specifics of the disease and the many gene mutations that have been found to cause congenital neutropenia. "It looks like the G6PC3 may become the second most common gene mutation," he said. The most common is still ELANE (formerly known as ELA2). He shared that a gene mutation analysis is helpful but not essential. If interested in having them done, he recommends using Dr. David Dale's lab at the University of Washington or a company such as GeneDX.

Prior to G-CSF (Neupogen) therapy, patients with congenital neutropenia had a 50 percent mortality rate in the first year. There are three case reports of patients with congenital neutropenia who developed acute myeloid leukemia (AML). (AML is seen in 20 percent of SCN patients in the SCNIR after 10 years on Neupogen.) Dr. Newburger explained that it is still unclear if or what the connection to Neupogen could be, but it could be hypothesized that the diseased cells develop more rapidly when bone marrow experiences the increased production that the drug initiates. Since there were cases of leukemia before the advent of Neupogen therapy, there is also a contribution from the basic biology of SCN.

Congenital neutropenics demonstrate a delayed response to G-CSF treatment and some patients do

not respond at all to G-CSF. Sepsis still develops in 8 percent of SCN patients. It is likely, but unproven, that it is more likely in those who do not respond to Neupogen or who stop taking the drug.

Among the congenital group at the conference were parents of children ranging in age from 9 months to 10 years; and Shay Jones, who was diagnosed at 6 months and is now 43 years old. Three of the children represented at the congenital session have been found to have the ELANE mutation.

Dr. Newburger went into great detail about bone marrow transplant, as this is the group most likely to have to face this type of decision sometime in the future. He explained the hierarchy of effectiveness and best-matched donors, which include a sibling match, another relative such as a parent or more distant relative (unlikely, however), an unrelated person and finally cord blood from a sibling or unrelated donor (which has shown a reduced rate of graft vs. host disease). Testing for a match is now DNA-based and not done by serology.

"Anyone who goes in for a transplant consultation and doesn't come out in tears, hasn't been given an honest consultation," Dr. Newburger told the group. He discussed all of the risks and concerns, as well as the entire process and the most common complications. He said the first three weeks is the most critical and that there is a 5-10 percent mortality rate in the first four to eight weeks. The patient will have to be completely re-immunized. They will miss school (or work) for at least six months and not be completely normalized for a full year. The caregiver and patient will need to remain near the transplant center for at least four to six months. Ten to 20 drugs will be given three to six times daily for several months.

Among the largest considerations to fully understand include the increased risk of death during transplant in SCN patients. Other possible complications

include chronic graft vs. host disease, and loss of the graft and return to pre-transplant cells. The endocrine system also may be affected.

Survival curves have improved by 25 percent between 1987 and 2007.

Who should consider a bone marrow transplant?

- If the patient has an absent or very poor response to G-CSF.
- If annual bone marrow studies show cytogenetic abnormalities predictive of myelodysplasia.
- Conversion to myelodysplasia or leukemia.

Shay asked in what ways he could watch for transformation to leukemic stages and Dr. Newburger suggested that regular blood tests will reveal abnormalities in other blood cells (unusually lower numbers) but annual bone marrow examinations are more sensitive and accurate.

Noah and Michelle, the parents of a 15-month-old son with congenital neutropenia who has been recommended for transplant attended the Conference in hopes of a second opinion from the expert physicians on hand. They were very pleased for the chance to meet with SCN experts who convened a special meeting to discuss their son's case. "The access to the experts has proven priceless for us. We had a one-on-one with Dr. Dale and managed to speak with every one of the other doctors throughout the weekend.

The young parents also commented on the benefit of camaraderie at the Conference, "It is comforting to us to meet other parents in similar situations," they shared.

The family shared that Dr. Newburger will keep in contact with their child's doctors in New York.



Kids Camp

The children who attended the 2012 Neutropenia Family Conference had several amazing treats in store for them. From pet therapy to body art to acting classes!

In fact, the children put on a stellar performance for all attendees during lunch on Saturday. They learned how the body will remain healthy when all the working parts of neutropenic care are in place and functioning. For example, Samuel (who had a birthday over the weekend!) was the person in the middle while Phoenix Children's Hospital's Child Life Specialist, Chantelle Johnson, demonstrated hand washing; while Abby gave Neupogen shots; Jessie did regular blood testing; and Lacie was eating healthy. They acted out the moving parts and demonstrated how everything "gets slumpy" when the parts aren't all working together.

They then acted out how the white blood cells are the soldiers of "Bodytopia" and when bacteria invade, the white blood cells capture them and carry them away.



The children all compared notes on where they take their shots and they came to the consensus that no one likes the shots in their legs. Abby, who is 10, said she gets hers on her arms and legs. When Abby's mom told her they were coming to the Neutropenia Family Conference she said, "Do you mean there will be other kids like me?"

"I liked it," Abby said of Kids Camp. "I liked meeting other kids with SCN." Her advice to the other children about taking shots was, "Just try to take it seriously and try to be brave."

Olivia S. will be a senior in high school this upcoming school year. She was born with cyclic neutropenia (as was her father and two of her uncles). She has come many times to the Neutropenia Family Conference and attended Kids Camp. This year, she helped with much of the Kids Camp activities. "I've never been sad for myself," she shared of her diagnosis. "I'm just sad for the children younger than me."



A Letter from Lee Cont.

and into the future. Not long ago Liam expressed an interest in telling his own story. Tanya found a website called Slide Rocket where she helped Liam to create his own presentation to share with his class. You can find Liam's presentation on neutropenianet.org under the Stories tab. Tanya is working with the NNN to facilitate the creation of more slide rocket presentations by other children to post on neutropenianet.org, and serve as a source of hope and inspiration to the neutropenic community.

We also welcome Mike McGuire PhD. Mike has spent thirty years working in the area of alcohol and substance abuse recovery and rehabilitation. This year he is retiring from his position as executive director of Sitka Counseling and Prevention Center in Alaska. As an infant Mike was diagnosed with cyclical neutropenia. His daughter and two grandchildren also have CN. Mike's childhood was difficult as there was little known about neutropenia and he spent three years in the hospital. His struggles with his disease were one factor that led him to seek a PhD in psychology from Columbia University and a career helping others to live healthy lives. Mike is glad for the chance to lend his energy and skills to the Network. Mike wants to work to assure the Network is available for his grandchildren, and that they have an active and fulfilling life despite their neutropenia.

As we welcome new board members, we send our appreciation to Kim Blanz for her service. Kim resigned from the board late this summer, due to new work responsibilities. Kim and her husband Chris were instrumental in creating our website in 2008. As owners of the web design company Cabedge, they put their resources to work and created a wonderful web presence for the Network. Kim has been a great supporter of



the Network since her son was first diagnosed with idiopathic neutropenia six years ago. We appreciate Kim's years of service and commitment to the Network.

Kim said it was a hard decision to resign but wanted to assure us that she will remain a loyal supporter of the NNN.

At this time of Thanksgiving, I wish to thank the many individuals and families who have contributed to the NNN this year. Your funds, resources, volunteer time, and excellent suggestions are essential to our ability to serve you and to help ease the difficulties of living with neutropenia. May the holidays be a time of peace and joy for all of our readers.

"The National Neutropenia Network is my personal place of peace. The people of the NNN taught me there were other parents who were dealing with the same things/same feelings I am. I met amazing people with neutropenia who live great lives, knowing them continues to give me a great amount of hope, it carried me forward and helped me deal with the impact on our family. The doctors are fantastic, so personal, and willing to share information. I was able to come home from conferences and set realistic baselines of what could (and should) happen in my child's life. The patients also gave me LOTS of good information about things I would never know...like where to give shots, growing up with neutropenia, dating with neutropenia, keeping a positive attitude, and so many other things I just can't write them down. I am friends with a number of NNN people on Facebook and will continue my friendship with them and I will continue to touch base with the NNN and hope to make it to the next conference. I will also continue to help in any way I possibly can." – Kim Blanz

How Can We Help?



THE DOCTORS' VIEW

We sat down with Mary Ann Bonilla, MD and Peter Newburger, MD to discuss how patients and their families can assist them in their work toward finding a cure.

"We appreciate the involvement in the SCN Registry, and the donation of marrow and blood for research," Dr. Bonilla immediately responded. During the conference, it was mentioned that there is a definite need for continuous donations of bone marrow as the researchers continue to learn the progression of the disease.

"I think we also appreciate being put in contact with other physicians so that we may create a good physician network," Bonilla continued. "Right now we especially are attuned to the very sensitive time when an adolescent transitions to adulthood. We need to identify internists or adult hematologists to refer to for that patient demographic. Of course, advocacy for both the National Neutropenia Network and SCNIR are both very important, as well."

Dr. Newburger pointed out that advocacy for the SCNIR could begin with the National Institutes of Health (NIH), as they are the government funding source for the Registry and much of the research they do. In the past, the NNN has collected letters from patients who have benefitted from Neupogen, and the research and help they have received through the SCNIR.

Familial studies are another area of interest for the researchers throughout the International Registry. The results and findings of gene mutations have taught them much about neutropenia, but using the DNA from the family members is very helpful in the studies, too. This is because it helps to identify how the relationship conveys between being a carrier of the gene and becoming somebody who is symptomatic.

Dr. Newburger explained that these studies are useful for identifying interactive genes. "What makes one member of the family with the gene have symptomatic disease and another member of the family with the same gene be relatively asymptomatic? There must be other genes. And

once you identify those, there's a potential to develop a drug to imitate or target the function of that gene."

He continued, "Have you heard of the Alzheimer's protection gene they discovered in Iceland? There's a gene, I think a polymorphism, that almost completely protects against Alzheimer's – even in people who have other genes that have an 80 percent chance of Alzheimer's. So, if you can find out what that gene does, than you might be able make a drug that does the same thing. The same could be true for neutropenia."

The four expert physicians (Bonilla, Boxer, Dale, and Newburger) have been advocates for neutropenic patients and very supportive of the National Neutropenia Network over the years. Dr. Bonilla has attended many Neutropenia Family Conferences. But what keeps them so engaged?

"I come back because I like to think I can contribute in some way to patient care," Dr. Bonilla explained. "I come from a small medical center, so to feel like I can contribute some helpful medical information is very rewarding to me. I also like to see and hear what is going on with patients that I have met before. Their problems and concerns are things that I think about and try to find answers to."

One common thread concerns her, however. "I am hearing very loud and clear something that has made me unhappy although I have to be honest that I have run into it personally. There is a lack of interest in contacting someone on the (SCNIR) advisory board or other colleagues, or not being exactly helpful in obtaining a second opinion and things like that. You should always be able to go beyond that," she said.

"That is such a widespread problem," Newburger agreed. But, he pointed out that people do have a voice and can be heard. "People can be advocates at their own institutions and make available information and resources for people with

nonmalignant blood diseases. That still leaves a lot of places uncovered, but it's a start." Dr. Newburger is working with the NNN to develop a database of pediatric hematologists around the country who interested in consulting with local doctors or directly taking care of children with neutropenia.

For more information on ways you can support the NNN, SCNIR, and the doctors and their research, email lee@neutropenianet.org.

DO YOU HAVE A PHYSICIAN WHO IS PARTICULARLY SENSITIVE TO THE NEEDS AND ISSUES OF NEUTROPENIA.

We'd like to develop a list of physicians throughout the country to refer patients to. Since SCN is such a rare disease the physicians on our list will not have a large number of referrals. Please write us with a short note with the following details: Name of physician, medical specialty, how long he/she has been treating you, are you on the Registry? What qualities do you think make your doctor exceptional?

(Please send to lee@neutropenianet.org.)

Dennis and Gwen F. attended for their first time this year. Their son is nearly three years old and has been diagnosed with idiopathic neutropenia. They shared that their physician back home is very interested in learning more and that they look forward to bringing back for him the plethora of information they've gathered.

"We've learned so much here this weekend," they shared. "So many things have completely changed," Gwen said. "All the information we've received about using a lower dose of Neupogen, the normalcy we can have in our lives that Dr. Boxer talked about, and the fact that we don't have to worry about our son going to the dentist now. I feel this conference has freed us this weekend. We felt so alone and secluded before."

Then, she shared the best news of all. "My son with neutropenia attended Sunday School this morning with all his friends for the first time in eight months. We called our older son at home and told him it was OK for him to go."

The National Neutropenia Network is grateful to the Severe Chronic Neutropenia International Registry for helping us get this newsletter into the hands of Registry members since 2005. Their help has been invaluable as we have worked to grow and develop the NNN. In 2013 the Network will take on the distribution of the newsletter, either electronically or through the postal service, depending on your preference.



IF YOU WISH TO CONTINUE TO RECEIVE THIS NEWSLETTER, IT IS IMPORTANT TO

LET US KNOW! You can sign up after December 1. Visit neutropenianet.org/newsletterregister.

You will be invited to complete a form which will assure that you will continue to receive this newsletter and updates of interest to neutropenic patients and their families.

For those who do not have access to the internet, please complete this form, cut it out and send it to NNN, PO Box 1693, Brighton, MI 48116.

I would like to be included on the National Neutropenia Network mailing list:

Name: _____

Address: _____ City: _____ State: _____ Zip Code: _____

Phone Number (optional): _____

Help us serve you better. What types of articles and stories would you like more of:
