The 2009 Neutropenia Family Conference will lead us all back to Ann Arbor, Mich., July 24-26, at the lovely Eagle Crest Conference Resort, located at 1275 S. Huron Street, Ypsilanti, Michigan.

Eagle Crest is 15 miles west of the Detroit Metropolitan Airport and seven miles from downtown Ann Arbor. This location rests on 135 acres of recreational area which includes an on site 18-hole championship golf course and jogging paths.

Confirmed speakers to date for the 2009 conference include: Dr. Laurence A. Boxer, Dr. Peter E. Newburger, Dr. Mary Ann Bonilla, Dr. David C. Dale, Dr. Daniel C. Link, Dr. John Levine, and Dr. Linda DiMeglio. They will discuss a wide variety of topics related to SCN.

As in years past, SCNIR’s top researchers will be on hand to discuss the latest research findings and what they mean for the future of SCN. Breakout sessions per diagnosis/type of neutropenia also will continue this year.

Perhaps most important, the one-on-one consultations will again take place. Please visit www.neutropenianet.org for a sign up form for this.

Other important topics that will be covered will span from the latest bone marrow transplant protocols to osteoporosis/osteopenia and strategies for coping with long term illness.

An exciting addition that we hope to add to this year’s conference is that of a teen program, which would be held in conjunction with, but separate from, the children’s program (the teen program will be contingent upon adequate registration).

The following events are already slated for the program(s):

- Small groups which will rotate to a few stations including: making (and eating) bone marrow snacks; education about blood, neutropenia and how it effects someone
- A drum circle with a music therapist from CS Mott Children’s Hospital
- Teen stories, activities, advocacy and mentoring
- Outside large group/team building activity
- Games with treats for the winners

These events will be facilitated by Jessica Doletzky, BS, CTRS, a recreational therapist; and Genny Dillard, BS, CCLS, a child life specialist; both from the pediatric hematology/oncology program at the UM Comprehensive Cancer Center, Pediatric Infusion, Child Life Program. We regret there is no childcare available for children under age 4.

Conference registration costs are as follows:

- Early Bird (Before March 30, 2009)
  - $120 per adult, $50 per child
- Regular Registration
  - $135 per adult, $65 per child

A limited number of hardship grants will be available. Please contact Lee Reeves for more information and to apply.

The hotel rooms will fill quickly, so make your reservation as soon as possible. Make sure to mention the NNN to receive the discounted room rate of $99/night. Refrigerators will be made available for storing Neupogen.

Stay tuned to www.neutropenianet.org for more details as we get closer to the date.

For more information on the 2009 Neutropenia Family Conference

Call • (517) 294-0736 or Email • nnnconference@live.com

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The National Neutropenia Network newsletter is published three times a year.

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Comments, contributions, and article suggestions are always welcome and encouraged. Please email us or write to:
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http://neutropenianet.org

Linking Families Program

The NNN Linking Families Program links individuals with similar diagnosis and life situations. If you are interested in contacting individuals and families with the same type of neutropenia that you or a loved one is facing, please email Lucy at: lucyl3@aol.com, or sign up online at http://neutropenianet.org/patient-family-support/find-someone-me/.

The Prevention Zone: Staying Healthy at the Dr.’s Office

Neutropenia is an impartial force. It does not spare the wealthy or the poor, not does it favor one ethnic group or gender. Back when it was first identified, physicians thought its victims were primarily children, but now we know it can pay a visit at any stage of life.

Although Neutropenia can affect individuals from across the socioeconomic and cultural spectrum, its impact can vary dramatically. For one family a sickly child diagnosed with autoimmune neutropenia that resolves after a few years can spark a new outlook that never again takes health for granted. For others it can mark the beginning of a life governed by daily injections, bone marrow aspirations, mysterious illnesses and the discovery of untapped strengths and wisdom. For the healthy-bodied young adult or active older adult who is suddenly beset with mysterious illnesses that propel them into the complex maze we call health care, there will be grief over the loss of their prior-self before they can begin to find the inner gifts neutropenia might reveal.

This might seem like a strange thing to say about a disease—that it brings gifts. Yet my experience loving a child with neutropenia awakened me to a new vision of my world. Before Leta was born I had an image as me the wise teacher who would share all my knowledge with my new baby, who would absorb it like a sponge. As most parents do, I learned that it works the other way.

Leta was an extraordinary teacher; even as an infant she radiated strength and understanding, and never passed up a chance to laugh. The truth in my daughter’s eyes told me that the greatest dis-service I could do for her was to see her as weak and powerless, not up to the challenges before her. I learned to see beyond the IVs, and bruises and powerless, not up to the challenges before her.

These thoughts were prompted when I read the article about Mary and Carl who lost two boys and found a way to transform their tragedy by helping others get through grief and loss. I had the opportunity to visit with Mary and her son John at the Family Conference in Seattle this year. Both mother and son are an inspiration: they smile easily, they are open about the grief of the past, and yet they seem to live in the present moment, enjoying life with all its simple miracles.

My experience in recent years with the NNN has put me in touch with people from all countries and backgrounds. It’s become clear to me that the demographics that separate us are superficial compared to that which unites us: a health-condition that demands we look within for hidden strengths and wisdom.

When we are doing well and neutropenia responds to treatments, we don’t need a support group, we may not even want to talk about our condition. When my daughter made neutrophils for the first time in her life after taking G-CSF, our family relegated neutropenia to the background. That was the right thing to do at the time; it gave us a chance to have a more “normal” life. But when things started to fall apart, we needed to talk to others who had been through the same things. My hope is that the NNN will be available to anyone who needs it until that wonderful day when we are truly obsolete.

Letter’s From Lee

“Adversity has the effect of eliciting talents, which in prosperous circumstances would have lain dormant.” — Horace

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Leta was an extraordinary teacher; even as an infant she radiated strength and understanding, and never passed up a chance to laugh. The truth in my daughter’s eyes told me that the greatest dis-service I could do for her was to see her as weak and powerless, not up to the challenges before her. I learned to see beyond the IVs, and bruises and medical labels to the strong and powerful spirit that inhabited her body. I don’t want to suggest that I was always the picture of composure through those difficult years, or that I never embraced anger or fear. But I did learn that if I truly desired, I could replace destructive emotions with gratitude, trust, acceptance, even joy.

These thoughts were prompted when I read the article about Mary and Carl who lost two boys and found a way to transform their tragedy by helping others get through grief and loss. I had the opportunity to visit with Mary and her son John at the Family Conference in Seattle this year. Both mother and son are an inspiration: they smile easily, they are open about the grief of the past, and yet they seem to live in the present moment, enjoying life with all its simple miracles.

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Comments, contributions, and article suggestions are always welcome and encouraged. Please email us or write to:

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National Neutropenia Network
2009 FAMILY CONFERENCE REGISTRATION FORM

July 24-26, 2009
Marriott Ypsilanti at Eagle Crest
Ann Arbor, Mich.

Name (please print): _______________________________________________________
Address: _________________________________________________________________
City: ____________________________ State: ________________ Zip: ______________
Day/evening/cell phone: ____________________________________________________
E-mail address: ___________________________________________________________

Names of each adult and name & age of each child
___________________________________     ___________________________________
___________________________________     ___________________________________
___________________________________     ___________________________________
___________________________________     ___________________________________
___________________________________     ___________________________________
___________________________________     ___________________________________

(A children and teen program will be available for ages 4 and up.
The cost is included in registration fee.)

Person(s) with SCN: _______________________________________________________
Type of SCN (if known): ___________________________________________________

Please check here if OK to include SCN type on name badges: ______________________

Any food allergies or other special needs? ______________________________________
________________________________________________________________________
________________________________________________________________________

Do you prefer vegetarian meals? ___ yes ___ no • If yes, how many people per meal? ___

For questions concerning the conference or the SCN CAMP program,
please e-mail Mara at nnnconference@live.com.

_____ Check here if you are willing to volunteer to help with the conference.

_____ Please check here if you are signing up for a one-on-one consultation with the expert physicians. Please make sure
to submit the separate form for one-on-one consultations. The form is available on the conference registration page of the
Web site. Contact Lee Reeves at leereeves99@gmail.com or (517) 294-0736 with any questions.


REGISTRATION COSTS

Earlybird
(Before March 30, 2009)
___ $120 each adult
___ $50 each child

Regular Registration
(After March 30, 2009)
___ $135 each adult
___ $65 each child

_____ Total # of adults
_____ Total # of children

TOTAL enclosed:

$__________

Registration costs include all presentations, handouts, children and teen programs, Friday reception, Saturday breakfast, lunch and dinner and Sunday breakfast.

PLEASE MAKE CHECKS PAYABLE TO THE NATIONAL NEUTROPENIA NETWORK (NNN)
AND SEND TO NNN AT P.O. BOX 1693 BRIGHTON, MI, 48116

Payment is also available on our Web site via Google Checkout.
testing, what to expect with the disease, and Neupogen dosing.

Friday night brought about networking, coping tips, and a lesson in history. Dr. Laurence A. Boxer, one of the pioneers of SCN research, shared with attendees the history encircling the study of the neutrophil, his personal experiences in SCN research and treatment experiences, and how G-CSF (Neupogen) came to be the only effective treatment for this patient group to date.

Saturday was packed full of educational sessions. Dr. Peter Newburger started the day with an overview of the neutrophil—how it is formed in the body, what it is charged to do, and how the breakdown occurs in SCN patients.

Dr. David C. Dale followed with a presentation about the Severe Chronic Neutropenia International Registry (SCNIR) and what the Registry has learned over its 13 years of existence. Dr. Dale and the other experts shared with the attendees all the species currently known to be affected by neutropenia, which include mice, killer whales, grey collies, cattle and even Persian cats.

Dr. Akiko Shimamura then addressed the issues and criteria to take into account when considering a bone marrow transplant in a SCN patient. She shared the indications for transplant candidates, the intricacies of donor selection, and the risks and possible health improvements that may result from transplant.

The afternoon featured an expert panel where attendees were able to submit questions and receive expert advice and answers on many facets concerning SCN. This panel was followed by the Ask the Doctor breakout sessions where attendees were split up by specific disease categories and each were assigned an expert physician to both explain disease-specific topics and to answer individual questions. This year, the adult-onset idiopathic patient group heard from Dr. Sioban B. Keel, who delivered an in-depth view of the disease; how it is diagnosed, treatment options and the statistics available through SCNIR.

New this year was a panel discussion, led by a licensed social worker, which featured three people who shared their unique perspectives and experiences of living with neutropenia—one with lifelong neutropenia, one diagnosed with neutropenia as an adult, and the parent of a child born with neutropenia.

Other topics that were covered during the Saturday educational sessions included bone health—an overview of bone architecture and osteoporosis/osteopenia in SCN patients, presented by Dr. Gad Kletter; and a full debriefing of a recently-completed pilot study on SCN patients’ oral health issues, presented by Dr. Frank Roberts. In addition, the day’s educational program was coupled by “SCN Camp” for the children who attended. The program was a hit by those who attended and was led by child life workers from Children’s Hospital & Regional Medical Center in Seattle.

Friday night all of the attendees and many of the day’s experts attended a wonderful, pirate-themed dinner. Everyone was having such a great time talking and sharing stories that the hotel’s personnel had to kick everyone out of the room because it was getting so late! Many people then convened in the hotel lobby/bar area to continue the camaraderie.

Sunday morning the attendees met for a relaxing breakfast and open discussions. Talks included the mission of the NNN—recent accomplishments and hopeful projections for the following year.

In addition, Chris Blanz, Web designer and owner of cабedge.com, the company that designed the new NNN Web site located at www.neutropenianet.org, gave a brief overview of the Web site’s impressive statistics, it’s mission, how to get the most out of a visit to the site, and he sought feedback from the audience for future improvements.

“The networking with other patients with this same, rare disorder is priceless for me,” said Corina, an attendee. “At home, I have been completely alone with my SCN, without support from either friends or doctors. Having access to experts and other patients, and excellent frontline information to take home to my doctors, has helped me and my medical team in Vancouver, Canada, truly begin to treat my condition. I’ve learned a great deal toward understanding my own specific diagnosis here.”

Aaron said he too learned a lot about neutropenia over the weekend. “I learned how much neutropenia has affected the world. It affects more than just humans and is an even more complex disease than I thought.”

He also resonated a thought that proliferated many of the weekend’s discussions—that of the life-changing results that Neupogen has had on this patient group. “Many lives, not just my own, have been changed by the addition of Neupogen. My quality of life has improved 1 million percent!”
A LEGACY OF LOVE AND COURAGE

Carl and Mary are veterans at dealing with neutropenia, now having spent nearly a half century waging war against it. A war unfortunately not always won. The two began building their family in the early '60s, a mere 30 years following the discovery of the first antibiotic. In seven years, they created five new lives—five lives that they loved dearly; and three of which they soon found themselves fighting hard to keep. Their second-born, Louis; fourth-born, John; and their baby, Matthew, were each born with cyclic neutropenia.

They cycled every three-and-a-half weeks or so—each on their own schedule—and each cycle bringing its own challenge, especially in those early years. What became normalcy for Mary and Carl was hundreds of sore throats, countless cases of cellulitis, endless ear infections, a tonsillectomy, several bouts of pneumonia and group, double-hospitalizations, and even septicemia at the delicate age of two weeks.

“I remember at one time talking with the school counselor and she asked me how many times a night I get up. I said six times a night. I often had two sick at a time. I said ‘Oh, that’s just how it is.’ And she said, ‘No! That’s not how it is!’” Mary chuckled at the memory, but still recalls just how taxing it all was at that time. Carl said they just somehow “normalized a high degree of anxiety.”

The St. Paul, Minnesota family of seven took a lot of vacations together. They traveled and they went camping, fishing and skiing often. “On almost every vacation we took, somebody was sick,” Mary said. She recalled one trip to Seattle when she was hot packing Matthew’s leg for an infection. They often would drive into the next town and find a doctor, she said. “They always asked me, ‘What does your doctor do for this?’ and I always told them which antibiotic to give us and how much, and they did it. They would always do what we told them to do. There was no Neupogen back then, they were always only treated with antibiotics. Back then the bacteria was still pretty responsive,” she pointed out.

And Mary would know these things. Not just because she had to in order to care for her sons, but because she was a nurse. And sometimes knowing the severity of their illness amplified the worry. “I realized the danger of just how serious it could be. The undertow of worry was always there.”

The two recall vividly just how challenging the years had been. Carl said he remembers the hopeless, anxious feelings of having no cure for his sons. “There just seemed to be no end to it. We couldn’t seem to really get that expert advice. Our lives were totally turned upside down.”

Louie, as the couple call their second-born son, died in 1977 at age 16. His bowel perforated and he developed a series of severe infections. The doctors tried for over a month to save him. They tried injecting antibiotics straight into his abdomen and they performed several surgeries, but the infection was just too much for his neutrophil-depleted body.

Devastated, Carl and Mary found a child loss grief support group at the children’s hospital that their sons so often visited. Mary said they spent the first year getting out of it what they needed to help themselves heal. They then spent the next six years as parent facilitators of the group. “That’s what saved our lives,” Mary shared.

But that seventh year was anything but lucky. While away in Europe, Carl and Mary’s youngest son, Matthew, died in his sleep. He too suffered a perforated bowel. He was 17. “They both died from the same thing,” Mary said. “One was dying for a month, the other in one night. If Neupogen had been available back then, Louie and Matthew would still be alive. I have no doubt about that one.”

Mary said they had to quit their beloved work with the grief group. “After Matthew died, we became too threatening to the group,” she explained. Eventually though Mary did go back and she facilitated for another three years.

Those years were filled with worry though, worry for the health and well being of their third neutropenic son, John. “We were on pins and needles for a number of years after Matthew died,” they said.

Prior to Matthew’s death, they had acquired a “brilliant hematologist” for the boys. “We asked him, ‘Could you look all over the nation for someone who knows about this disease?’ And he did. He found Dr. Dale for us.”

“This is how we found help for our son, John,” Carl added. “It was like a big search light went off.” Dr. David C. Dale is an expert on cyclic neutropenia, and upon the advent of Neupogen, he began the treatment on John. “On Neupogen, life was totally different from what it had been,” Mary said.

John went on to marry and had two daughters of his own. Olivia, who is now 13, also has cyclic neutropenia. “She was diagnosed right away and has been on Neupogen from the beginning,” Mary said.

Carl said John doesn’t take a moment of life for granted—especially when it comes to his girls. “He treasures every second of every day with those girls,” he said proudly of his son. “Things happened so quickly with his brothers, he knows.”

To this day, the couple still has Louis’ ficus tree and Matthew’s aquarium. And Mary said she and Carl have been able to become “very, very happy again.”

“It wasn’t all blood and guts,” she chuckled. “We had some good times.” Carl agreed, “While they were sick often, they still had a lot of fun.” They were boys, for sure. Carl recalled one time when the boys were all ice skating in the back and John’s head got injured. To escape getting in trouble while Mary and Carl were on their way out the door, one of the older brothers held John’s head down on the ice. After their parents were gone, they brought John in and cleaned him up. “And it didn’t even get infected either!” Mary laughed.

“The illness has a leverage all its own,” Carl noted. “They had good lives. Unfortunately, they were painful and short.”

Mary agreed, and added, “We packed an awful lot of love into those years.”
The National Neutropenia Network wishes to recognize Dr. David C. Dale for his 40 years of dedication to researching neutropenia.

Dr. Dale’s lengthy career began when he received a medical degree from Harvard University. He went on to serve his residency training at Massachusetts General Hospital and at the University of Washington. He then served as a senior investigator with the National Institute of Allergy and Infectious Diseases’ Laboratory of Clinical Investigation at the National Institutes of Health (NIH).

Dr. Dale has worked as professor of medicine in the department of medicine at the University of Washington, Seattle, for more than 25 years. Dr. Dale recently served in the prestigious role of president of the American College of Physicians.

His research in neutropenia spans from Border Collies to all ages of humans. His “first love” began with cyclic neutropenia, which the Border Collie shares with humans, and his passion to seek answers has led him all around the World. His interests today include neutrophil physiology, neutropenia, and hematopoietic growth factors.

Dr. Dale also is active on the National Board of Sponsors for Physicians for Social Responsibility and currently serves as codirector of SCNIR.

Thank you, Dr. Dale, for all you have done for all of us affected by neutropenia.

Patients Write Letters in Support of NIH Grant for SCNIR

Over 60 families responded to the NNN’s call for letters of support for the National Institutes of Health grant that SCNIR director, Dr. David C. Dale, recently submitted. The response was wonderful. The letters were personal, unique and heartfelt. Some were from children, some from grandparents, and many were from patients.

“It was wonderful to receive so many letters of appreciation for the work of the Registry and Amgen’s provision of G-CSF for SCN patients in the United States,” wrote Dr. Dale. “We received responses from 67 patients and their families. We sent a representative sampling of the letters to the National Institutes of Health (NIH) with our request for renewal of funding for the Registry for another five years. We also sent a copy of all the letters to Amgen so that they were aware of how meaningful it is to patients and their families to have their support.”

Dr. Dale continues, “Just as the downturn in the economy has affected many individuals, these are very tough times for getting federal grant support for medical research, as well. It is very meaningful to the officials at the NIH and the experts who review grants to know that patients and families are truly helped through the NIH grants program. The letter from all the patients and families gave our grant application a much better chance for success. Thanks very much.”

Contacts:

National Neutropenia Network (NNN)
PO Box 1693 Brighton, MI 48116
(517) 294-0736 • www.neutropenianet.org

Severe Chronic Neutropenia International Registry (SCNIR)
Plaza 600 Building 600 Stewart Street, Suite 1503 Seattle, WA 98101
(800) 726-4463 • http://depts.washington.edu/registry/

Determine Your ANC

SCNIR offers a tip sheet on how to read a complete blood count (CBC) report and how to determine your absolute neutrophil count (ANC). Visit: http://depts.washington.edu/registry/Calculate.ANC.pdf

World Experts Meet to Discuss Neutropenia

The Third Neutropenia Conference was held in Heraklion, Greece Sept. 26-27, 2008. The hosts for the European conference were Dr. Helen Papadaki of the University of Crete School of Medicine, Heraklion, Greece; and Dr. Jan Palmblad of the Karolinska Institute, Stockholm, Sweden.

More than 100 physicians attended, including Dr. David C. Dale of the USA’s SCNIR. Discussions were held on cyclic, idiopathic, and autoimmune neutropenia, as well as the many types of congenital neutropenia.

The experts also discussed the neutropenia associated with primary immune deficiency disorders. Dr. Dale closed the conference by talking about G-CSF treatment for neutropenia, based on the work of the SCNIR over the last 14 years.

Did You Know?

The consumer genetics testing company, 23andMe cut their prices a full 50 percent. The company, which specializes in DNA analysis and recently formed a partnership with Ancestry.com, maintains that the price cut is intended to make their services more attainable to the average consumer. For more information, visit www.23andMe.com.

Canadian Support Group Plans Conference

The Neutropenia Support Association Inc. in Canada is planning its 2009 Marrow Failure and Myelodysplasia Conference March 28, 2009 in Toronto, ON, Canada. For more information visit their Web site at http://www.neutropenia.ca/
De-myth-tifying Autoimmune Neutropenia

By Peter E. Newburger, MD

Autoimmune mechanisms are responsible for many cases of acquired neutropenia in children and adults, and some cases of neutropenia in the setting of congenital disorders of the immune system.

Although most cases in infants and young children resolve by age 4 or 5, autoimmune neutropenia in older children or adults may be chronic or lifelong.

The limited availability of information on the diagnosis and natural history of this disorder has led to a number of prevalent myths, which I will try to bust in this article.

1. Anti-neutrophil antibody tests are readily available and reliable

The diagnostic test for autoimmune neutropenia is a measurement of anti-neutrophil antibodies, performed as either an “indirect” test (looking for antibodies in the patient’s serum that react with normal neutrophils) or a “direct” test (looking for neutrophils from the patient with antibodies attached). Most clinical laboratories do not perform these tests.

As neutrophils do not travel well, send-out testing is usually performed by the less sensitive indirect test on serum that can give a false negative result. An excellent reference laboratory for this test is provided by the American Red Cross: http://www.redcross-lab.org/neutrophil/

2. A positive anti-neutrophil antibody test means you definitely have autoimmune neutropenia

Not necessarily … False positive results are quite common, particularly with the direct test, but can also occur with the indirect assay. The results need to be interpreted in conjunction with other tests and the clinical features of each case. Dr. Laurence Boxer recently reported several severe congenital neutropenia patients initially thought to have autoimmune neutropenia due to positive anti-neutrophil antibody assays.

Similarly, a negative test does not rule out autoimmune neutropenia, which I often diagnose based on other features, such as a bone marrow examination or response to very low dose G-CSF (filgrastim, Neupogen®). A bone marrow exam is indicated in patients with recurrent bacterial infections and a history of mouth ulcers, to determine if there is a cause for neutropenia other than autoimmune.

3. A low ANC always means high risk of infection

In autoimmune neutropenia, production of neutrophils by the bone marrow is normal or increased, then they are cleared rapidly from the circulation by the autoantibody. However, during their rapid transit through the bloodstream, the neutrophils function well and can be delivered to sites of infection.

Thus, there is often a much higher number of neutrophils in the tissues (e.g. skin, lung), where they perform most of their work fighting infection, than in the veins where blood is drawn for laboratory counts.

Many patients with autoimmune neutropenia need no treatment, even with very low absolute neutrophil counts. If they do need treatment, the neutrophil count usually rises very rapidly in response to extremely low doses of filgrastim; in fact, “normal” doses (calibrated for chemotherapy patients) can lead to severe bone pain.

4. Autoimmune and idiopathic neutropenia are separate entities

There is considerable overlap between these disorders with different names but similar clinical features, laboratory values, and bone marrow findings.

In fact, many “idiopathic” (i.e. unknown cause) neutropenia patients probably have autoimmune neutropenia with a false negative anti-neutrophil antibody test—or the test was never performed.

Peter E. Newburger, MD, professor of pediatric hematology/oncology and vice chair of pediatrics at the University of Massachusetts Medical School, now serves as the NNN’s Board Medical Advisor.

~ TIPS: Traveling With Neupogen ~

Sharon White, who has idiopathic neutropenia and is a NNN Board member, shared with the attendees of the 2008 Neutropenia Family Conference some tips for traveling with Neupogen and safely storing and dispensing injection supplies.

First, the following is a list of items Sharon buys to travel with her Neupogen:

1. Icy Cools: www.containerstore.com, item #10004715, $4.99/45
2. Everyday Diabetes Organizer: www.medportllc.com, #10007, click on the link to find a retailer
3. Dry Sack: www.seatosummit.com, small-4 ltr, 9”x15”, click on link to find a retailer

“I put my box of vials in the MEDport Organizer, then surround it with the Icy Cools,” she shared. “Sometimes if I only need a few vials, I’ll use something smaller than the box like an empty Rx bottle. I put the whole pack into the Dry Sack. This Sack is in case something happens, like a delayed flight, and I need to use ice. The bag will not drip and make a mess. Also, the Dry Sack comes in bright colors, mine is red, so I’m always on the alert as to where my Neupogen is. I often hook the Dry Sack to the hotel door knob or to my suitcase to remind me to take my meds when I leave.”

Do you have a tip to share? Send it to us!
Email us at: nnnnewsletter@yahoo.com
Or snail mail it to: PO Box 1693 Brighton, MI 48116

Spring 2009
A newly discovered gene mutation has revealed that a rare form of SCN occurs as part of a complex syndrome involving several organ systems.

The mutation involves the glucose-6-phosphatase, catalytic subunit 3 (G6PC3), SCNIR researchers concluded in their recently published article in this year’s first issue of the New England Journal of Medicine. “The mutations of G6PC3 result in a loss of phosphatase activity. G6PC3 catalyzes the hydrolysis of glucose-6-phosphate to glucose and phosphate, the terminal step of the gluconeogenic and glycogenolytic pathways.” That is, the mutation prevents neutrophil precursors from being able to complete the metabolic steps necessary for the formation and storage of glucose, the primary sugar used by neutrophils.

Homozygous mutations of G6PC3 were identified in five children with the disorder from two parents who each carry the gene, but are not symptomatic of neutropenia. Since the mutation was discovered, European SCNIR researchers have identified an additional seven unrelated patients with the same mutation.

The children with G6PC3 mutations frequently exhibit cardiac abnormalities, thrombocytopenia, and urogenital abnormalities, the article notes.

In an accompanying editorial written by U.S. SCNIR researchers, Dr. David C. Dale and Dr. Daniel C. Link, the genetic basis of severe congenital neutropenia still has yet to be discovered in “a substantial proportion of children with the disorder.” They go on to note that in a study involving North American patients, nearly 40 percent did not fit into the already known mutations: ELA2, HAX1, WAS, or GFI1 (G6PC3 was not sequenced).

Dale and Link wrote that the effect of the genotype on the clinical course of the disorder remains unclear, but with continued discoveries such as this one, there is hope that “effective targeted therapies can be developed.”

### Genetic Variants of Severe Congenital Neutropenia

<table>
<thead>
<tr>
<th>Genes</th>
<th>Gene Function</th>
<th>Incidence of Variant</th>
<th>Inheritance</th>
<th>Associated Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>ELA2</td>
<td>Serine protease</td>
<td>50-60%</td>
<td>Autosomal dominant, sporadic</td>
<td>Isolated neutropenia</td>
</tr>
<tr>
<td>HAX1</td>
<td>Mitochondrial function</td>
<td>Unknown</td>
<td>Autosomal recessive</td>
<td>Neurologic and neuropsychological abnormalities in some cases</td>
</tr>
<tr>
<td>GFI1</td>
<td>Transcription factor</td>
<td>Rare</td>
<td>Autosomal dominant</td>
<td>Monocytosis and defects in lymphocyte number and function</td>
</tr>
<tr>
<td>WAS</td>
<td>Cytoskeleton function</td>
<td>Rare</td>
<td>X-linked recessive</td>
<td>Monocytopenia and T-lymphocyte activation</td>
</tr>
<tr>
<td>CSF3R</td>
<td>G-CSF receptor</td>
<td>Rare</td>
<td>Autosomal dominant</td>
<td>Severe myeloid hypoplasia in the bone marrow; resistant to G-CSF treatment</td>
</tr>
<tr>
<td>G6PC3</td>
<td>Glucose metabolism</td>
<td>Unknown</td>
<td>Autosomal recessive</td>
<td>Cardiac defects, thrombocytopenia, and urogenital abnormalities</td>
</tr>
</tbody>
</table>