Stage is Set for 2006 Family Conference

Sessions to feature top docs, discussions on condition-specific concerns, dental hygiene, infection prevention, and much more.

The National Neutropenia Network (NNN), in conjunction with the Severe Chronic Neutropenia International Registry (SCNIR), will be hosting the 2006 Neutropenia Family Conference in Seattle, Wash., August 11-13.

The conference will feature the latest medical updates concerning neutropenia presented by Dr. David Dale, co-chair of the SCNIR; Dr. Laurence A. Boxer, SCNIR board member and expert on neutropenia; and Audrey Anna Bolyard, RN, BS, SCNIR clinical manager. Other sessions will include a Q&A with Dr. Dale, a Neupogen dosing session, and a dental hygiene overview with periodontist, Dr. Frank Roberts.

Dr. Wayne Katon, a professor of psychiatry at the University of Washington Medical School, will address depression and chronic illness and the symptoms and special risks related to the chronically ill patient. This year's conference also brings with it breakout sessions for each type of neutropenia to facilitate individualized, informative talks. Special program activities also are being planned for the children.

Other scheduled events will include a dinner party on Saturday night, separate rooms for childcare, and meals with a view of Portage Bay. The conference will provide a family friendly, intimate experience which will be reflected in the agenda schedule, venue, and accommodations. The agenda will be formulated to allow time for attendees to talk with each other, share stories, information, and support.

To help offset a portion of the conference costs, we are requesting a conference attendance fee of $35 for adults and $10 per child. Please do not allow this donation request to deter your attendance. Special arrangements may be made with Lee Reeves for those unable to pay.

Rooms are reserved at the beautiful Talaris Conference Center. The rooms are being held under “Family 2006” for the rate of $120 per night, and include a refrigerator. Call (206) 268-7000 for reservations. Please also visit the Talaris Web site at www.talarisconferencecenter.com for more information and to view the rooms and property. Please reserve a room ASAP as there are only 31 rooms available and many events are occurring at the University that weekend. Area hotel are expected to fill rapidly.

The hotel spillover site will be the Silver Cloud Inn, University location. Call (800) 205-6940 for reservations and ask for the University hospital rate.

Arrangements also have been made at the Silver Cloud Inn to accommodate overflow requests for double beds. The rate is $139/night for the two beds, and includes breakfast, and shuttle bus transportation to the conference center. Additional events are also available for children.

RSVP Today!

Send an email to:

NNN_SCNIR_2006@hotmail.com

Serendipity: good luck in making unexpected and fortunate discoveries.

I believe in serendipity. I believe it's one of the magic ingredients behind any endeavor that makes the world a better place. I'd like to share with you how serendipity has left its imprint on the National Neutropenia Network in recent months.

In the winter edition of this newsletter we featured Erin Bogart. She is one of the most enthusiastic and talented individuals I've ever met; the kind of person who has great ideas and the ability to execute them. I asked her to help with the upcoming Family Conference.

Without hesitation she jumped in and took on the task of making the 2006 Family Conference the best ever. She set priorities: a new, more family friendly venue, more topics and breakout sessions focused on specific types of neutropenia.

Unfortunately personal changes in Erin's life cut into her free time making it impossible for her to continue as the coordinator of the 2006 conference. But Erin didn't drop the ball, she handled it off to Mara Lim, the sister of Matt Solomon who was among the first recipients of Neupogen therapy. I was concerned about making the change,...
If you are interested in attending this event, please respond with your name, number of adults and number of children who will be attending, and the ages of the children to: NNN_SCNIR_2006@hotmail.com.

For additional questions concerning the event or to volunteer in assisting with the final touches of the conference, please email Mara Lim, conference coordinator, at NNN_SCNIR_2006@hotmail.com or write to: 2006 Neutropenia Family Conference, 7728 18th Ave NW, Seattle, Wash. 98117.

For a tentative schedule of the sessions to be held on August 12, see page 5.

We hope to see you all in Seattle this summer!

For more information concerning the Family Conference call the SCNIR at (800) 726-4463

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...Letters from Lee continued from pg 1

but Erin assured me that Mara was perfectly capable and willing to help.

She was right. Mara didn’t miss a beat. With the help of her mother, Charlene Teel who also lives in the Seattle area, she contacted numerous venues and found one that fits our needs for a relaxed and comfortable atmosphere at a reasonable price. Mara has dedicated hundreds of hours to conference planning.

She brings grace to the process and skillfully handles the inevitable glitches that come with preparations for an event where families from all over the country will gather to learn about the rare disease that has changed their lives.

I can’t overstate the value of Mara’s high energy and can-do attitude. It’s just what we need to take the 2006 Family Conference to the next level.

In late May, Jennifer Schraag’s email landed in my inbox with the golden offer to help with this publication.

Jennifer is a healthcare journalist and mother of a one-year-old daughter with autoimmune neutropenia. As a staff writer for the healthcare trade publication Infection Control Today, she is familiar with the ins and outs of various infection prevention strategies. She will be sharing those strategies in a new standing column titled The Prevention Zone. You can read her first column, “Summer Safety” on page 7.

I am pleased to announce that Jennifer has volunteered to write and edit our newsletter. As a journalist by trade, she is wonderfully qualified. Jennifer is an unexpected and fortunate discovery for our organization.

Debbie Dicesare, featured on page 4 of this newsletter, came to us by way of Dr. Boxer. She expressed to him her frustration with the lack of information on neutropenia and her interest in helping to change that.

Debbie has idiopathic neutropenia, works full time and is the mother of two small children. Nonetheless, she has given generously of her time and talent. Soon we will benefit from the fruits of her labor.

Finally we have two families who I consider our organization’s guardian angels: the Serra family, who we featured in the winter 2005 newsletter, and the Jacobson family, who have two children with congenital neutropenia.

Inspired by our efforts to revive the National Neutropenia Network they reached out to family and friends requesting donations to support our work.

Thanks to their belief in our cause and their willingness to step out of their comfort zones, we can continue to move forward as an organization.

Maybe someday we can support research that leads to a brighter future for those afflicted with neutropenia. That would be the ultimate serendipity.

Lee Reeves is the president of the National Neutropenia Network. Her daughter Leta, who passed away in 1997, had congenital neutropenia.
Surviving and Thriving:

You could call Silke Deeley a pioneer in neutropenic advocacy. She’s been fighting the battle since the 1980s. Well, it is her daughter who has actually been on the front line of the battle, but Silke was there, advocating for her the whole way.

Alyssia, Silke’s daughter, started getting sick when she was six years old. The Deeleys spent over three years searching for some kind of diagnosis for her - let alone an effective treatment. Young Alyssia endured countless tests and doctors visits along the way.

“We had referrals to any number of physicians at that time,” Silke recalls. “And one would think that in a city as large as Chicago, someone would know something.” But they didn’t.

The hematologist treating Alyssia at that time had chalked it up to cancer; possibly some kind of leukemia. But after all her bone marrow biopsies and the barrage of other testing all came back fine, the diagnoses didn’t make sense. It was really only Alyssia’s low neutrophil count that was continuously flagged.

That hematologist eventually transferred, and the Deeleys received a new hematologist; and a new hope. Jong Kwon, MD, came from Memorial Sloan-Kettering Cancer Center in New York where he had worked under Dr. Mary Ann Bonilla, a long-time researcher of white blood cell disorders. With his gained knowledge, Kwon was familiar with neutropenia and was able to finally make a diagnosis: idiopathic neutropenia.

Kwon advised the Deeleys that there was little to be done, but he had heard about another physician, Dr. Laurence A. Boxer, an expert on neutropenia. Silke sought him out and found that he was conducting the research trials for Neupogen. Alyssia was soon enrolled in the trials for gaining the drug’s FDA approval.

“I don’t know if that was as scary as the idea that your child might die,” Silke says of enrolling her daughter in a trial of an unapproved drug. “When they tell you there isn’t really anything else that could be done ... she had continuous infections and running fevers, abscesses and fissures ... she was on steroids for a year prior to treatment with Neupogen, and that was awful. They had also given her gammaglobulin transfusions, repeat bone marrow aspirations, and she had two surgical procedures. I think that was probably scarier than having someone tell me that there is an experimental drug and ‘We’re having a lot of success with it; we would like to try it with Alyssia.’ Nothing else seemed to be working and she was miserable. We said, ‘Yes, great, let’s try it.’”

Silke says Alyssia didn’t have a good quality of life leading up to that point. “One year she was hospitalized probably three weeks out of every month for an entire year,” Silke asserts. “I don’t know how we did it when I think back about it. She was little girl and having to go through countless procedures, doctor visits, and hospitalizations, and nobody knew what to do to help her.

“She was constantly sick and not able to do the things the other kids were doing. Kids not understanding, schools not understanding; and then Alyssia always asking if she was going to die.

“So finding something that we thought might help, approved or not, was not nearly as scary as everything we had already dealt with. We still worry about the possibility of leukemia, although it hasn’t been linked to the idiopathic group. It is something that remains in the back of your mind.”

Today, Alyssia is 28 years old - and a new mother. Her son Matthew Edward was born on March 21, 2006. Alyssia has been able to live a much higher quality of life since she began taking Neupogen in 1988, and Silke says she took it throughout the pregnancy.

As for Matthew’s health, “So far so good,” Silke says cheerfully. The proud new grandmother says it never ceases to amaze her how they were able to get through all that they have faced over the years.

“I think both my husband and I, when we look back and we think about what our lives were like for all those years, we wonder how we managed. I believe there is a capacity for families to cope with a lot more than they think they might when faced with a child’s illness. So what was hardest? I don’t think any of it was easy. But somehow we found the strength and got through it.

“I went through that period of time when Neupogen wasn’t available. I think when you have a neutropenic child and you don’t have any means of keeping your child well, you can’t see a light at the end of the tunnel. Not knowing if your child is going to make it through the next year, or wondering what kind of infection they may get next; that’s the worst part.”

This is what spurred Silke’s pioneering days. During the Neupogen trials, Amgen pulled five families together and flew them out to California. They wanted a family-based support group and thus the National Neutropenia Network was born.

“We came from different states, and we were scattered all over. It was difficult to stay on task because we were never able to meet other than by phone,” she shares. But they pulled through and eventually became organized enough to become a 501(c)3 nonprofit group.

In 2000, before leaving Chicago to move to Georgia, Silke organized a conference where she brought in Dr. Boxer and Dr. David Dale, and approximately 65 families attended. She did a lot of work in terms of sending information to families and physicians, attending the American Society of Hematology (ASH) con-
Frustrating Search Leads One Woman to Take Action to Benefit All Neutropenic Patients

A physicist by trade and researcher at heart, Debbie Dicesare was crushed when her Internet search turned up sparse information on neutropenia.

"When I search things on the Web I want to see more than one thing," she says of her initial Internet search. "I felt like I was starved for information. I thought, ‘If I am this frustrated, there must be people out there that are really frustrated with this situation.’"

That frustration fueled plans to create a Web site for the National Neutropenia Network. The new site, www.neutropeniainet.org, is expected to go live by the end of June.

Her research about idiopathic neutropenia began in the fall of 2003 when she began feeling tired frequently and had little energy. A severely low white blood count led her internist to refer her to a hematologist.

Unfortunately, the hematologist did not have any experience treating idiopathic neutropenia. She was being treated "like a cancer patient," she recalls.

"I was the only patient (with idiopathic neutropenia) they ever had in their practice," she says. "They had no idea what the symptoms were."

When she received Neupogen, she would get very high doses of it - about 480 mcs. Debbie’s condition see-sawed with each dose, and in the spring of 2004 she was missing a lot of work because she was always so sick.

"It was really bad," she said. "I was in constant pain all the time. It was horrible. I would feel horrible for days afterward."

In desperation, she searched the Internet to learn why she was having so many problems. One of the resources she found was contact information for Lorna Stevens, president of the Neutropenia Support Association Inc., in Canada. She wrote to Stevens to see if she could get in touch with Dr. Laurence A. Boxer. For Debbie, the experience with Stevens’ recommendation to Boxer highlighted the need for strengthened network abilities.

"There needs to be more information out there," she asserts. "I thought, ‘If I ever get better here and I am back up to speed, I am going to make sure there is something available.’"

For about a year, Debbie worked to gather information for the Web site. She worked with her brother-in-law, a Web site designer, to create the site. The site will not only include contact information for the National Neutropenia Network, but she also hopes to have ways for members to contact one another.

Ideally, Debbie wants to include links to journal articles regarding neutropenia and other helpful Web sites. She wants to post details about events such as the Family Conference in Seattle in August. In the future she would like the site to host a neutropenia forum for those who have questions about the illness.

"Most of the people that write me or call me; they are starved for that kind of contact," she says.

She expects that she will probably be the site moderator at least for the time being.

"I anticipate it will grow and change over time depending on what people’s needs are," she says. "I am just trying to get something on the Web to start out with."

I extend my appreciation to my friend Cyndi Lieske for her critical role in writing and editing our first two newsletters. In this issue, she wrote the feature articles on Melissa Little and Debbie Dicesare. Her earlier stories have moved many of you to share your own stories and to let me know how isolation has been one of your greatest challenges in living with neutropenia.

~ Lee Reeves

National Neutropenia Network
No Life Challenge Too Big for Melissa Little

Melissa Little remembers the shock she felt when she learned she was going to have a baby.

She was 16 years old when doctors told her she was about seven weeks from giving birth. At that time, not much was known about the effects of severe congenital neutropenia (SCN or Kostmann's Syndrome) on pregnancy. Little was told her baby might not be born alive and that its chances for survival were slim.

"They didn't know if his lungs would be developed," she said, adding that her parents did not buy any items for the baby based on the dire predictions.

Imagine her surprise when her son, James, was born "perfectly healthy."

"He was a perfect 10," Little recalls. "My Dad called my sister and told her that we needed a crib, high chair ... all kinds of things. We didn't have anything for a baby."

James, now 11, plays hockey and is active as a boy scout. While James' good health has continued through the years, Melissa has not been so fortunate. Vasculitis, hypothyroidism, and severe bone pain are some of the health problems she frequently deals with.

"He doesn't know what it is like to not have a mom that is not sick," Melissa says.

Whether she is feeling sick or not, Melissa makes time to be with her son. One of the ways she likes to spend time with her son is to drive him to and from school each day. She enjoys their one-on-one time in the car. It gives her a chance to hear about his daily activities.

"When it comes to my son, I will go to the end of the world and back for him," she says. "It doesn't matter how I will feel. There are days when I don't feel good and I will still want to drive him to school - that's our time together."

Melissa has not been hospitalized recently, but she has had several problems with her mouth and gums.

"I still have a lot of mouth sores that I am dealing with," she explains. "I got full dentures when I was 21. The bones in my mouth had deteriorated and had caused my teeth to loosen and fall out. I had a lot of problems with sensitivity; anything I drank or ate had to be room temperature. My dentures have been the greatest thing in the world. I don't have any pain in my mouth anymore."

We would take our shots together. We would sit in the living room and say 'One, Two, Three, Go!'"
- Melissa Little

The time has come for her to get new dentures. She is considering either new dentures or dental implants. She has been doing research on the implants and hopes to make a decision soon.

Melissa is in the process of applying for disability benefits through social security. For a short time she was working as an insurance agent.

One day she left for work with a small sore the size of a mosquito bite on her leg. By lunchtime the pinprick-sized spot was the size of a tennis ball and was very painful. When she asked her supervisor if she could leave, she was told there was no one to cover for her and she would be out of a job if she left.

"I had to work the rest of the day," she said. "That night I had to be hospitalized and I was in the hospital for two weeks."

When she returned to her job, Little's supervisor informed her that the incident was Melissa's fault because she had not called in sick the day the sore showed up on her leg.

"I decided enough is enough," Melissa says.

As a teen, Melissa became friends with Leta Reeves, daughter of the National Neutropenia Network's president, Lee Reeves. The two girls became good friends. They sometimes would have sleepovers at the Reeves' house.

"She and I were really close," Melissa recalls. "We would take our (Neupogen) shots together. We would sit in the living room and say 'One, Two, Three, Go!'"

Melissa was happy to receive the first NNN newsletter and have a chance to reconnect with Lee. She hopes to meet others with SCN.

She and her husband, Dan, hope to attend the family meeting in Seattle this summer. In the past they have wanted to go but have not been able to work it out with her husband's work schedule.

"As far as the newsletters go, and the National Neutropenia Network goes, I think it is a great thing because it helps you get in touch with other people," she says. "It helps you know that you aren't the only one in that boat."

Jennifer Schraag: "Infection Prevention in the Healthcare Setting."
1:00-2:00 Dr. Wayne Katon: "Depression and Chronic Illness, Symptoms and Special Risks in the Chronically Ill Patient."
2:00-3:00 Audrey Anna Bolyard, RN, BS: "Neupogen: Dosing. Getting It Right."
3:00-4:00 "Ask the Doctor" Break out sessions: Idiopathic - Congenital - Cyclical
4:00-4:30 Final Thoughts and Evaluation: Audrey Anna Bolyard, Lee Reeves, Mara Lim.
6:00 Dinner: Luau Theme at South University Campus

Schedule subject to change without notice.
SCN Hits International Headlines
Sperm Donor Passes on Congenital Neutropenia to Five Children

Severe congenital neutropenia (SCN) proliferated international headlines in May when research from Laurence A. Boxer, MD, and colleagues from the University of Michigan and the Severe Chronic Neutropenia International Registry (SCNIR) were published in the May 2006 issue of the Journal of Pediatrics. The study investigates the cases of five children, from four different families, all of whom were conceived by in-vitro fertilization by donor insemination, and all have SCN.¹

Boxer and his colleagues conducted DNA testing to ascertain SCN inheritance. Because the donor sperm was not available, alternative methods were used to determine whether the sperm donor transmitted SCN. DNA isolated from leukocytes was used to sequence the ELA2 gene in the affected children and their mothers. ELA2 was amplified by polymerase chain reaction (PCR), and the product was sequenced. PCR was also performed with genomic DNA from the mothers and affected children using a set of 22 microsatellite PCR primers on chromosomes 14 and 19 to establish linkage to the paternal allele.

None of the mothers were found to have a mutation in ELA2, but all five affected children had the same mutation affecting the fourth exon at site S97L. Linkage mapping analysis confirmed that all affected children had the same paternal allele on chromosome 19, which contains ELA2.

“Our findings indicate that the father provided consistent haplotypes leading to the expression of SCN in all affected children, supporting an autosomal dominant inheritance in which ELA2 mutations occur,” the researchers wrote.

When the first cases of Kostmann Syndrome were reported in 1950 and 1956 by Dr. Rolf Kostmann, the cases were documented as being transmitted in an autosomal recessive pattern of inheritance.² Since, other cases of SCN have been described, and some show an autosomal dominant or sporadic pattern of inheritance.³

Dr. Boxer, in interviews with the press, said that while he had no information about the donor, he and his colleagues suspect that the man had an unusual condition called mosaicism in which the mutant gene occurred only in his sperm and not in the rest of his body. If in fact he did have mosaicism, he would have shown no physical symptoms, Boxer pointed out.

According to a correction in the May 20 issue of the New York Times, the sperm bank says it is unable to get in touch with the donor because he no longer knows his whereabouts.

Citations:
Summer Safety

Summer brings with it a whole new set of health concerns. But don't sweat it! With flu season behind us and by simply adhering to a few preventative measures, we can all have a happy, healthy, active and safe summer packed full of fun.

Infection rates generally decrease during the summer months because more outdoor activities are executed - thus limiting our exposure to respiratory secretions. The most pertinent infection prevention concerns related to summertime includes traveling, heat and sun exposure, waterborne pathogen exposure, and insect bites.

We'll begin with traveling concerns. We can avoid acquiring infections during traveling by taking some pretty simple precautions.

Hand hygiene. Hand washing is our No. 1 defense. Organisms are most commonly transferred by the hands from the surfaces we touch. We expose ourselves by touching high-touch areas found in airports and other high-traffic areas. Such high-touch areas include elevator buttons, escalator handrails, door handles, and even the pens we use at the check-in counters.

The very best thing you can do is to always carry an alcohol-based hand gel, and use it frequently. These hand gels are 99.9 percent effective in killing the organisms found on hands. Also, avoid touching your nose, mouth or eyes without first washing your hands or using the hand gel.

Other tips include always using a seat cover when using a public restroom, and washing your hands every time you visit a public restroom. Use a paper towel to open the door when leaving the restroom - you'd be shocked to know how many people do not wash their hands and transfer their germs to the door handle.

For those on Neupogen injections, make sure to pack your supply with ice packs for transfer. It is also a good idea to adhere to sharps safety standards while traveling by bringing along a small sharps container for disposal of your used needles. If one is not available, a small Clorox bottle or other heavy duty plastic container will work fine.

Gather important information of your destination. Talk with your doctor and ask if they recommend any particular hospital in that area should you fall ill while you are away. Make sure to pack insurance cards and your doctor's contact information (if you don't already know it by heart!).

Hotel rooms. Researchers have sampled hotel rooms and have found a plethora of organisms lurking. A good antibacterial spray down of the high touch areas around the room and bathroom, and all around the toilet area, could be of benefit. Also, many organisms have been found on hotel bedsprads. The simple cure for this is to take the bedsheets off and put it away during your stay. Furthermore, for those with small children, most hotels set their water temperatures very high. Always use care when bathing the little ones that they do not get burned.

This brings us to skin care. Our skin is our first barrier to infection. Anything that compromises that barrier can potentially facilitate a breeding ground for infection. Cuts and scrapes should be washed thoroughly and an antibiotic skin cream should be applied.

Sunburns are another such compromise. Apply sunscreen amply and often. One slather for the whole day is never enough. Sunscreen should have a SPF factor of at least 15, and should feature both UVA and UVB protection. Coverage should also include the lips and face. Use accessories to cover up as well. Wear a hat, wear sunglasses, and when swimming, it never hurts to wear a t-shirt for extra protection.

Swimming is the all-time favorite summer activity, and it too can present special considerations. According to the CDC, recreational water illnesses (RWIs) are spread by swallowing, breathing, or having contact with contaminated water from swimming pools, spas, lakes, rivers or oceans. The most commonly reported RWI is diarrhea.

Other RWIs can cause various symptoms which can include skin, ear, eye, respiratory, and neurologic infections. The transmission of the waterborne pathogens that cause these infections are more commonly found in lakes, rivers, and ponds. Generally, swimming pools are cleaned regularly and do not present as much of a risk. It is good to wear ear plugs when swimming as well.

Other suggestions on promoting healthy swimming practices are available at: www.cdc.gov/healthy-swimming/tools.htm.

When fishing, ensure proper hand hygiene protocols. Most marine bacteriums are not commonly transmittable to humans, but there have been some documented lately. Wash your hands frequently when fishing and always cook the fish well before eating it.

Insect bites are becoming increasingly more troublesome. Both mosquito and tick-borne diseases have increased in the United States over the last few years. It is very important to stay covered, wear long pants and long sleeves, especially in the dusk and dawn times, and to use insect repellent, as directed, to reduce the chance of a bite.

Last, but not least is food safety. Summer brings with it numerous cook outs and picnics. This is can promote exposure to infectious diseases that are transmitted through food.

Cook meat and poultry thoroughly and avoid cross-contaminating foods. For example, if preparing chicken with one set of utensils, the utensils need to be thoroughly cleaned and replaced before using the same utensils on any other foods. Refrigerate leftovers as quickly as possible, or simply throw them away.

Following these simple tips should be an everyday occurrence no matter the season, but by applying them during the summer months, maybe we can all go out and have a great, relaxing time - and stay healthy doing it!

Jennifer Schraag, mother of two, is a healthcare journalist residing in Chandler, Ariz. Her daughter Brooklyn (pictured above) has autoimmune neutropenia.
CIMFR Investigates Disease Progression, Diagnosis

Researchers at the Canadian Inherited Marrow Failure Registry (CIMFR) located in Toronto, Ontario, Canada released a study in May concerning disease progression and diagnosis characteristics of inherited bone marrow failure syndromes (IMFSs).

IMFSs are genetic disorders characterized by defective single-lineage or multi-lineage hematopoiesis. According to the researchers, IMFS patients are at risk for severe cytopenias, development of marrow cytogenetic abnormalities, myelodysplasia, and malignancy.

The rate of disease progression and proportion of patients at risk for these complications is currently unclear, they wrote, so they examined recently diagnosed IMFS patients to determine distribution of diagnoses, disease progression, and development of significant outcomes.

The results showed that 74 CIMFR patients were considered "recently diagnosed." The median age at diagnosis was 2.7 years of age (range of study participants was birth to 40.6 years of age). Annual follow-up data were available for 53 (72 percent) patients.

The five most prevalent diagnoses were Fanconi anemia, Shwachman-Diamond Syndrome, Diamond-Blackfan anemia, dyskeratosis congenita, and Kostmann's Syndrome. Eighteen (24 percent) patients were unclassifiable.

Twenty-eight (53 percent) follow-up patients had disease progression as indicated by new or worsening cytopenias, new marrow changes, or initiation of transfusion support and/or medical therapy. Fourteen (19 percent) fulfilled minimal diagnostic criteria for myelodysplasia. Eleven patients had hematopoietic stem cell transplantation by first follow-up. Five patients have died, but the researcher note that survival at 36 months is approximately 89.8 percent.

The researchers concluded that IMFS patients are often diagnosed at a young age. The relative distribution of diagnoses is similar to previous reviews of published cases; however, 25 percent of patients are currently unclassifiable.

Perhaps the most distressing finding is that disease progression has occurred in approximately 50 percent of the follow-up patients. Early mortality is noted among this patient group. Therefore, the researchers note that continued prospective observation of these patients is warranted.

The CIMFR is a prospective multi-center study established in 2001 to register all IMFS patients in Canada.

Citation: