

FROM THE DIRECTOR

Dear Friends,

Best wishes from the Amyloid Treatment and Research Program at Boston University School of Medicine and Boston Medical Center for 2012. The new year is a period of change and rebirth, with spring just around the corner. I'd like to tell you about "change" in our program in three areas:

Amyloid Treatment and Research Program personnel. We welcome two new physicians to our program, Dr. Andrea Havasi and Dr. Lauren Stern. We were very sorry to see Dr. Adam Segal, and Dr. Laura Dember, our talented nephrology consultants, leave this year. Dr. Segal, ever busier with young children, went into private practice. Dr. Dember left us to take on a wonderful new opportunity as a Professor of Medicine at the University of Pennsylvania School of Medicine in Philadelphia. Dr. Dember will remain a close collaborator, and we are very pleased that she will be continuing to care for patients with amyloidosis and building clinical and research programs with colleagues at Penn. Dr. Havasi is a graduate of the Semmelweis University School of Medicine in Budapest, Hungary, and completed her training at Boston Medical Center. She is a recipient of awards from the National Kidney Foundation, American Heart Association, and NIH for studies of mechanisms of proteinuria and renal damage. Dr. Stern received her M.D. from the University of Medicine and Dentistry of New Jersey and trained in internal medicine and nephrology at Mount Sinai Hospital in New York. We are so pleased to have these outstanding young physicians join us as amyloid nephrologists.

XIIIth International Symposium on Amyloidosis. The XIIIth International Symposium will be held from May 6-10, 2012 in Groningen, the Netherlands. The Symposium is entitled "From Misfolded Proteins to Well-Designed Treatment", and will bring together researchers from all over the world who are dedicated to improving our understanding and treatment of the systemic amyloidoses. Investigators and trainees in our program have submitted 19 abstracts to the meeting, and we expect to have great participation there. We are actively seeking support to help defray the expense of travel to the meeting. If you are interested in helping fund one of our trainees or faculty, please contact us.

Improved Outcomes and Survival for Amyloidosis Patients. This newsletter focuses on a very important change, which is the improvement in outcomes and survival for our patients. The outlook for patients in 2012 is so different from just a few years ago. For patients with AL (immunoglobulin light chain) amyloidosis, the world changed for the better in 1994, when our brave patient, Lou Catania, underwent one of the first stem cell transplants in the world for a patient with AL amyloidosis. An expert team including Drs. Martha Skinner, Ray Comenzo, and Evan Vosburgh harvested Lou's bone marrow, stored it, administered high dose melphalan chemotherapy, reinfused his stem cells, and put his disease into remission, a remission that has lasted to this day. Also as you will read, Carolyn Gunn, the fourth patient we treated, enjoys an active and fulfilling life. Isabelle Lousada, 30 years old and newly wed, was also an early pioneer, coming to us from the U.K. Isabelle has gone on to adopt and raise three beautiful children, and now chairs the board of the Amyloidosis Foundation. Since those early days, more than 600 transplants for AL amyloidosis have been carried out at Boston Medical Center, and over the years, the safety and outcomes have steadily improved. Furthermore, through clinical research, we now have identified other drugs such as Revlimid and Velcade that are very effective for AL amyloidosis, so options for patients continue to improve. Our thanks to all, doctors, nurses, and most of all patients, who have helped change the world through their efforts and participation in clinical trials! The landscape has also changed for patients with AA (secondary) and AF (familial) amyloidosis, as new drugs are being developed and tested at BMC and elsewhere. In addition, for patients like Carol Pantazakos, liver transplantation has been successful in preventing progression of familial TTR amyloidosis. We work closely with Dr. David Lewis at the Lahey Clinic in Boston to find organ donors for our patients, replacing their liver, the source of the mutant transthyretin protein that causes disease, with a normal liver. Because the TTR patient's liver is otherwise healthy, it can often be used as a lifesaving "domino" transplant donor liver for patients whose liver is damaged from a hepatitis virus or cirrhosis. Our congratulations to these patients, and to all of you, for your efforts to make amyloidosis a treatable condition!

With Best Wishes,



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Director



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Amyloid Nephrologist



Andrea Havasi, MD
Amyloid Nephrologist

Lou Catania was the first patient treated with high-dose chemotherapy and autologous stem cell transplantation at Boston Medical Center in July, 1994. His wife Linda tells the story...

Lou was 43 years old when he got sick. We had 3 kids, and his illness had gone undiagnosed for nearly 9 months, and I was convinced that he would die. How long could I keep secret from Lou, and his family that all of the illnesses the doctors were looking for were fatal? With prayer circles formed in several states, we plunged through time with nothing more than a fist full of hope. Time went by without answers and I clearly remember thinking that the doctors would figure out what was making Lou sick. We just had to keep him alive long enough for the doctors to realize that they did have the answer, and they could fix him.

It was an answered prayer when Texan Jim Lang spoke the words "Boston University is doing research on amyloidosis, you need to go there". Our only problem was that we did not have a positive diagnosis. But Jim had also mentioned that Boston had "Congo Red Dye" and the equipment to get the diagnosis. We didn't want the diagnosis to be amyloidosis, but that would answer the questions about what was making Lou sick. So, I was beyond elated when the renowned Dr. Martha Skinner actually returned my phone call, and we soon headed to Boston. Lou now believed that the doctors in Boston knew what he had, and they could fix him.

Long story short, we were given the red carpet treatment and a team of the world's best doctors, all of whom believed that they could provide hope with the use of a stem cell transplant after high dose chemotherapy. Dr. Comenzo told us it was "experimental", but if Lou were his brother he would want him to try it. Hope grew as teams of doctors paraded through Lou's reverse isolation room followed by groups of students privileged to actually have an amyloid patient to poke at. What a thrill it was to see smiling doctors huddled in circles sharing good news that their patient was showing signs of improvement! Milestones were met; success recorded, studied, and restudied and at last news came that the patient could go home!

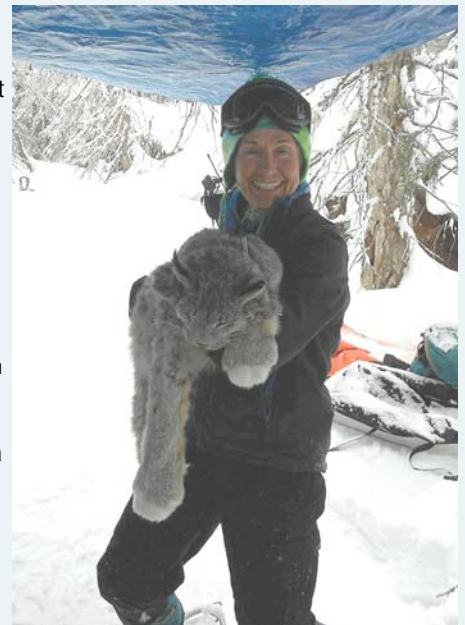
We left our new friends in Boston wondering how long it would last. What would the next step be? Would Lou need a bone marrow transplant? (We opted out of that one!) Everyone watched as Lou continued to improve for the next 17 years. We have celebrated birthdays (Lou just turned 60!), anniversaries (30 years), graduations (our middle child just received her doctorate as a Chiropractor), and all the ups and downs of family life. Lou continues in great health, and we are still easily brought to tears with thoughts of our experience in Boston. How proud we are to go back and visit and share our story, and see so many smiles. Thank-you is just not enough, and never will be.

"What a thrill it was to see smiling doctors huddled in circles sharing good news that their patient was showing signs of improvement!"

Dr. Carolyn Gunn was only 44 when AL amyloidosis was diagnosed.

In November, 1994, I received chemotherapy and an autologous stem cell transplant at the Boston University Medical Center for treatment of primary amyloidosis. The compassionate care I received during the procedure and the positive outlook I was given over the next several months was most encouraging. Prior to my diagnosis, I had been an adventurous and active person, and my wish was to return to that lifestyle. I also hoped to practice some of the life lessons that accompanied such a brush with mortality.

Recovery to full strength took a few years, but now, nearly 18 years later, I feel as strong as I did before I became sick. I am so fortunate to lead a healthy and active life, supported by family and friends. Since recovering, I have returned to a career in veterinary medicine and I currently work for the Colorado Division of Wildlife which presents me with many wonderful projects. One project is the lynx recovery project – capturing lynx, placing telemetry collars on them, giving them a microchip, doing a physical exam, drawing blood, aging, weighing and measuring. It is a most exciting project that gets me out snowshoeing to remote, beautiful spots. I also enjoy hiking, fishing, ornithological research, and restoring an 1898 home in southwest Colorado. I thank Dr. Martha Skinner and her team for giving me the chance to experience each precious day.



Isabelle Lousada writes...

The year I turned 30 I completed my architectural training, and was busy planning my wedding. It should have been a wonderful time, but the truth was that for over a decade I had been unwell. I had a lowered immune system, muscle aches and extreme fatigue, all attributed to Chronic Fatigue Syndrome. Gradually and almost imperceptibly my health was declining, and each day was becoming a physical struggle. By the time I tied the knot I was truly very ill. At our wedding my brother in law, a cardiologist, realized this and we ended the celebration with a visit to the Emergency Room. From that point it was several months of regular doctor and inpatient visits before finally a liver biopsy confirmed a diagnosis of AL Amyloidosis. The symptoms I had largely been overlooking included, abdominal pains, extreme weakness and fatigue, puffy eyes, fainting, tingling fingers, swollen ankles, nosebleeds and shortness of breath. Tests at the National Amyloidosis Center in London, where I lived, revealed my liver, spleen, kidneys and heart were involved and I had a massive load of amyloid deposits.

The painfully slow process of getting a diagnosis was in stark contrast to the speed in which I was admitted to hospital and started treatment. I had several weeks on a renal ward while doctors tried to get my fluid gain under control before I started what should have been six courses of chemotherapy. That didn't go well at all, while I was dealing with almost unbearable side effects of the first round of chemo, and fast deteriorating health, my family learned about the Amyloid Program at Boston Medical Center. Thanks to a cooperative airline, and my brother in law's assistance I was allowed to fly over to Boston, where I immediately started undergoing a stem cell transplant. There was grave concern over my condition, but thankfully, with some hesitation, it was agreed I could go ahead with the treatment. I had numerous complications and had to remain in the hospital for months. My husband and mother had to share an apartment in town. Not an easy start to a marriage for any of us!

It took several years for me to regain my health. Bit by bit my body recovered and my organ functions returned to normal. Now 16 years on, my life is amazing. There are many days when the word amyloidosis doesn't even pop into my head. I take a deep breath when I say this, but I have perfect health. Really, the best thing I can say is I now have the kind of health I don't think about. I am extremely active; busy with family and work, invariably I stay up way too late. We live outside of Boston now, with our 3 children, adopted from Vietnam and Ethiopia. The eldest just hit his teens, and the youngest is in first grade. I have a beautiful horse that I ride most days. My husband jokes that we have a Benjamin Button marriage; we started near death and are now almost back to dating! I feel absolutely blessed and I love my life.

**Carol Pantazakos** writes...

I was found that I had inherited a familial amyloidosis gene in January of 1985. At that time I was married, 34 years old and had two young children; 6 months and 2 years old. My first thought was that I would not be around to see my children grow up and envisioned the disabilities that this disease would cause me. I had grown up with a father and an uncle who had developed and ultimately passed away from complications of Familial TTR Amyloidosis.

At the moment of diagnosis my life changed forever. Having seen the devastating effects of this disease growing up I could only imagine my future health and the hardship it would cause my family. At first I went into a deep depression shutting my husband and family out but ultimately my husband made me realize that I was still healthy and should live my life appreciating each day.

Fortunately for me my mother came across an article about Dr. Alan Cohen's/Dr. Skinner's group in Boston and their research on amyloid. I contacted them and went to Boston to be evaluated. Following this I made at least two trips per year to be monitored for onset of amyloid disease and to donate blood for research purposes.

During the years that I was regularly visiting Dr. Skinner I was informed that studies had shown that liver transplantation would arrest the progression of the disease. Finally there was hope. However, the idea of a liver transplant was a fearful and daunting thought. After much consideration, prayer and weighing my options of developing the disease versus the transplant I made the difficult choice to put myself on the liver transplant waiting list as soon as I developed the first symptom and my biopsy was positive.

In 1995 after waiting for 2½ years I got the phone call that a liver had been found for me. By that time I was very excited to receive that call and get the transplant behind me. On May 23rd I underwent the surgery. My recovery went well and I was able to return home to New Jersey 17 days later.

My husband and I are now semi retired and have moved to North Carolina. I do take medication on a daily basis but it has become just a part of my daily routine. Since my surgery I have led an active life playing competitive tennis, skiing, golf and backpacking across the Grand Canyon, but most importantly I am now looking forward to attending both of my children's weddings this year!

Life has been good.

PEOPLE, PHOTOS...

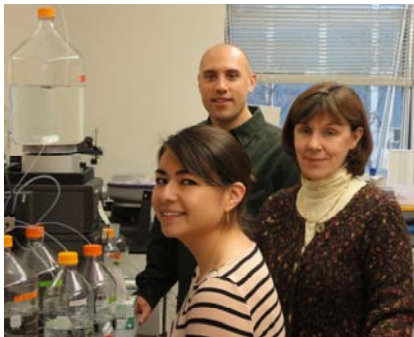
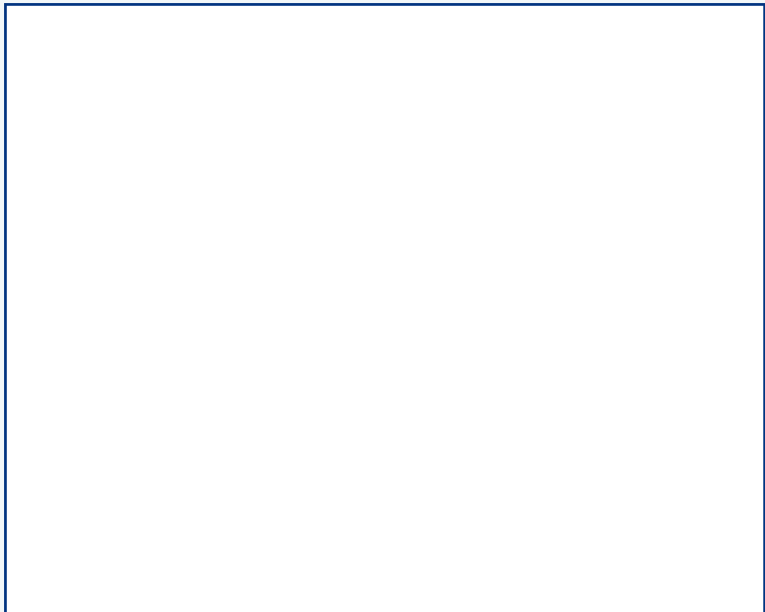
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The Amyloid Treatment and Research Program gratefully accepts financial support for our research and clinical programs from patients, family and friends.

For information on bequests and other planned giving options contact us at the address listed above or by phone.

Donations can be made through our website or by mail.

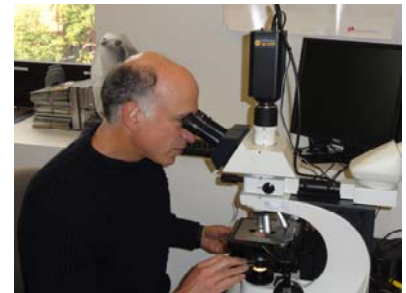
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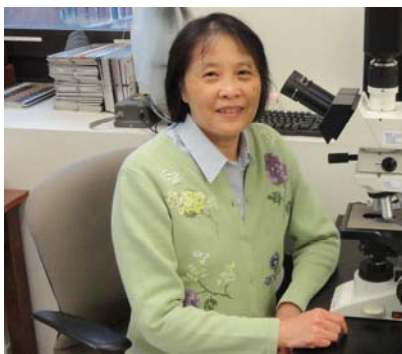
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**The Nassau Wings with Drs. Cowan, Connors
& Seldin**



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