



News and Stories - Fall 2015

2015 Nashville Gala—October 16

Join the Amyloidosis Foundation for our Nashville Gala in the beautiful Country Music Hall of Fame, which honors the history and the stars of country music. Actor Michael York will speak about his experiences with amyloidosis.

Enjoy dinner, live music from The Kentucky Lineman, silent auction and more! Please show your support and make plans now to attend. Proceeds will be donated to the Amyloidosis Foundation and Amyloid Research Fund at Vanderbilt University Medical Center.



Tickets are \$175/per person and \$125/Amyloidosis patients. Please go to our website www.amyloidosis.org for more information and a link to purchase tickets. We look forward to seeing you in Nashville!

A New Life and A Life-Threatening Challenge

By Darcy Tannehill

My AL (light chain) amyloidosis journey began around 2005. Gastrointestinal issues for which there appeared to be no cause and a colonoscopy showed nothing. In 2007, I noticed differences in my hair and nails and began to have symptoms of carpal tunnel. Routine blood work showed only a low level of Vitamin D. I was told to take vitamins.



A nerve conductor test showed carpal tunnel in both hands but I put off the surgery due to a lack of time. In December of 2008, I became dizzy and fainted. I was going to get that checked, however two weeks later my husband had a heart attack and died. I did eventually see my doctor who ordered an MRI, which showed nothing. Over the next two years, I became increasingly tired and everyone said that I was working too hard and long. They were right and I've always done that, but this was a different kind of tired. More blood work again, which showed nothing. I was convinced that something was wrong and I thought it might be hormonal or found something that matched my symptoms—amyloidosis.

(continued on page 7)

Education • Awareness • Support • Research

www.amyloidosis.org



Foundation Updates

2015 Patient's Day - October 17, Nashville, TN

Amyloidosis: Raising Awareness, Improving Care, Enhancing New Treatments

A team of highly-regarded medical specialists will join patients and their families for a day of amyloidosis information and discussion. The agenda will include medical presentations in the morning, and an open panel discussion in the afternoon. Special focus will be on these disciplines: Gastroenterology, Pulmonary, Nephrology, Cardiology, Neurology and Pathology.

Registration is complimentary and includes breakfast and lunch. Onsite event registration will begin at 7:00 am, presentations will start at 8 am - 4:00 pm at the Nashville Hilton Downtown, 21 Fourth Avenue South, Nashville, TN.

Please visit our website www.amyloidosis.org for more information, a link to register for this event plus a link to the Nashville Hilton Downtown (please mention the AF for our group rate). You can also register by sending us an email at info@amyloidosis.org with your name and how many people will be attending with you. Remember, this event is open to patients, family members, caregivers and friends. Please share with anyone you think would benefit in attending our event, your participation will make this event a success.

TTR Patient Webinar—Presented by Giampaolo Merlini, MD

We will offer this recently recorded webinar on our website later in September. Please visit our Patient Resources page on www.amyloidosis.org to view this and other webinars. Thank you to Dr. Merlini and Alnylam Pharmaceuticals for sponsoring this educational webinar.

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Patient Resources

The foundation has several programs that benefit patients and their families. All of these are provided free of charge upon request.

- Webinar recordings posted on our website
- Accurate Informational Pamphlets
- Comprehensive website with information for patients, caregivers and physicians
- Toll Free Number **1-877-AMYLOID**
- Listing of experienced physicians that specialize in amyloidosis, it's diagnosis and treatment. Email us anytime with questions: info@amyloidosis.org



President's Corner

As you can see, we have been busy here at the foundation—we have an updated “look” and logo, a brand-new website filled with updated information that is easy to understand plus exciting events happening in Nashville this Fall.

We are currently reviewing all of the research grant applications that were recently submitted and will be announcing the awardees in December.

There has never been a better time to make a donation of support. Because the Amyloidosis Foundation is a nonprofit organization, your support provides resources that end up in the hands of promising scientists working to find a cure for amyloidosis.

Please stand with us by making a generous donation today. We appreciate all that you do.

We hope to see many of you at our Nashville Gala and Patient Day in October!

- Mary O'Donnell

David Seldin Memorial

The passing of David C. Seldin, MD, PhD, director of the Amyloidosis Center at Boston University School of Medicine, on June 27, has left a large hole in the amyloidosis community.

A longtime friend of the Amyloidosis Foundation, he served on our scientific advisory board for many years. David was generous in lending his support, recently volunteering much of his time to read and advise on the amyloidosis disease information and content for our new website.

We will miss him greatly.



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Our newsletter is published quarterly (Spring, Summer, Fall and Winter) by the **Amyloidosis Foundation**. We welcome letters, articles and suggestions.

Please contact us at: **info@amyloidosis.org**, **1-877-AMYLOID** (877-269-5643) or **7151 North Main Street, Ste. 2, Clarkston, MI 48346**



Stop and Smell The Roses

By: Kathy Whitney

As published in *The Reporter*, Vanderbilt University Medical Center's Weekly Newspaper

Charlotte Haffner is the first Vanderbilt patient to receive both a heart transplant and a stem cell transplant, she has certainly learned that life is all about the journey.

In 2008 Charlotte was a healthy, active 55-year-old who loved the outdoors, particularly taking care of her horses. Then, one day she started to feel different. She was fatigued, retaining fluid, experiencing shortness of breath.

Routine diagnostic testing by her physicians did not reveal the cause of her heart failure. As the months dragged on, her health deteriorated. She sat on a hillside on her farm one evening and looked up at the star-filled sky and prayed for guidance. She sensed she was dying.

The next day she received a letter stating her doctor was relocating and she would have to find a new one. A friend encouraged her to call Vanderbilt Heart & Vascular Institute and gave her the name of cardiologist Joseph Fredi, M.D. Fredi had a good idea what was wrong with Haffner after her first visit in July 2008.

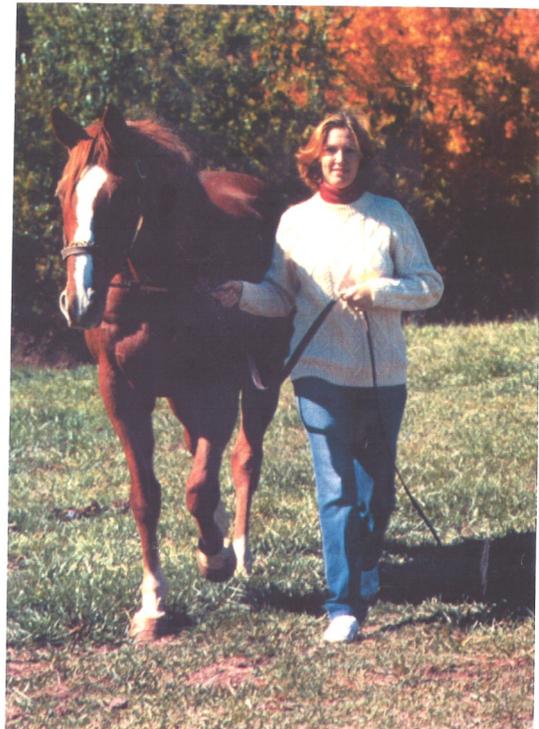
Several tests and a biopsy later, he delivered the bad news to her as she lay in the recovery room. Charlotte has primary AL amyloidosis, a plasma cell disorder that originates in the bone marrow. The disease results when amyloid protein builds up in one or more organs, causing them to malfunction. Amyloidosis is typically treated the same way as cancer: chemotherapy and a stem cell transplant. Because the disease had taken up residence in her heart, she was in heart failure.

"I asked Dr. Fredi if it was fatal, and he held back for a minute, and he said, 'I'm afraid it could be,'" she recalled. "I said to him, 'Well, it won't be this time.'"

Before Charlotte could undergo treatment, she needed a heart transplant. She met with a team of Vanderbilt cardiologists, surgeons and hematologists who jointly agreed to take her case despite the high risks associated with it.

Charlotte had a heart transplant on Nov. 29, 2008. She cleared the first hurdle in her journey back to good health. It would pale in comparison to the second.

After recovering at home, she re-entered Vanderbilt University Hospital in February and had her stem cells harvested, which means that all of the blood was taken out of her body, the stem cells removed, and the blood replaced. **(continued on page 6)**





Jonathan Wall, PhD—Inspired by Patients

As published in the Greater Knoxville Business Journal

Every year, thousands of Americans go to their doctors with an inexplicable pain that worsens with time and eventually kills them. Much like cancer, their ailment eats at their body, causing organs to shut down one after another. Unlike cancer, there aren't ways to see the disease in the body and know where it is and what it is doing. And few ways exist to specifically treat it. The disease is amyloidosis and Dr. Jonathan Wall has dedicated his entire adult life to helping doctors diagnose it quicker and treat it better.



As a professor of medicine and director of the Preclinical and Diagnostic Molecular Imaging Laboratory at the University of Tennessee Graduate School of Medicine, Wall has made great strides in developing treatments for amyloidosis as well as ways to image it. Until recently, the only way to see amyloids in patients was with an autopsy.

"Amyloidosis is a rare, complicated, and devastating disease," he says. "There are probably only 3,500 new patients diagnosed every year in the U.S. Until recently, these people got very little attention." Amyloidosis is caused by an amyloid, which is formed when proteins that people make normally stop working correctly. The amyloids propagate and stick together into hard clumps that collect in tissues and organs, disrupting their functions. When amyloid builds up in the brain, for example, it can kill brain cells and contribute to Alzheimer's disease. When it clumps elsewhere in the body, it causes amyloidosis.

Wall came to Knoxville in 1995 from the United Kingdom to work with the Dr. Alan Solomon, whom he calls "a god in the field of amyloid research" who is on staff at the UT Graduate School of Medicine at UT Medical Center. (Dr. Solomon was honored as a Business Journal Health Care Hero in 2011.) A couple years later, the two made a groundbreaking discovery — an antibody with therapeutic and diagnostic potential for amyloids. When injected into mice, the antibody bound to amyloid and caused it to be dissolved by cells in the immune system — a first. The researchers decided to see what happened if they made the antibody radioactive. They discovered it imaged the amyloid perfectly in mice.

Ten years later, Wall and his team became the first in the world to image people with amyloids using antibodies at UT Medical Center. As the first of 40 patients were undergoing their tests, Wall was undergoing his own medical test down the hall. "When patient three was getting imaged, I was running on a treadmill getting a stress test," he recalls. "It was so nerve wracking. Here I thought I was dying because I was worried about the patients down the hall being injected with something we invented." Wall's concern for the patients exemplifies the type of scientist he is since he was a doctoral student.

"Dr. Wall is a brilliant, but introspective individual. Most think of him as personally outgoing, however, he considers other people greatly in all interactions," said Dr. Stephen Kennel, who has worked with Wall for 15 years. Wall met with all the patients in his study, and says their spirit was inspiring. "These patients came from all over the U.S. and they knew that what they were about to enter into would not benefit them," he says. "But every one of them said the same thing — that they want to help so that the next generation of patients with this disease can benefit from what they are doing. They wanted to leave a legacy."

Wall is also keenly interested in seeing his research applied in treatment. "In addition to being a leading researcher in his field, Dr. Wall is passionate about bringing the results of his research into the clinic to treat patients fighting this disease," says Michael Paulus, director of technology transfer at Oak Ridge National Laboratory and former executive at Siemens Molecular Imaging, who has collaborated with Wall. Wall and his team are working on developing new therapeutics. **(continued on page 7)**



An INVITATION to all Patients, Caregivers and Families, Join Us on Nov. 16: Amyloidosis Patient Forum with the FDA

The Amyloidosis Research Consortium (ARC) is hosting an extremely important meeting with the FDA in Bethesda, Maryland on November 16th. This is an unprecedented opportunity for amyloidosis patients to inform and educate the FDA about the need for new treatments.

Amyloidosis experts will present the pressing need for new and effective treatments to be safely and quickly evaluated. This is our chance to address the urgent patient need for additional treatments while educating the FDA about our community. This effort will help to avoid delays in the evaluation and approval of these drugs.

This is a call for action. The FDA has requested that we hold this meeting. We need patients and their advocates to demonstrate their support by attending - and there will also be an opportunity for patients to speak. The ARC is working closely with the Amyloidosis Foundation, Amyloidosis Support Groups and the core amyloidosis centers to drive this initiative forward. Admission to this meeting is free. To find out more and reserve your place, please contact us at admin@arci.org.

(continued from page 4)

"All of my doctors exude confidence. I never had a worry. This is going to work. I know it is," she said. "I could not have gotten through this without Drs. Fredi, Sawyer, Mark Wigger, Tom Di Salvo and Adetola Kassim. Not only are they brilliant physicians, they are also compassionate men of great character."

Since her diagnosis seven years ago, Charlotte has dedicated her life to promoting amyloidosis awareness and support to those affected by this disease. She volunteers on behalf of the Amyloidosis Foundation at their awareness booth at medical conferences, leads the foundation support group at Vanderbilt University Medical Center and has made numerous press appearances to generate awareness of amyloidosis. She is an excellent community builder, and has been involved in many fundraisers, including Co-Chairman of the 2015 Nashville Gala. Charlotte has served as a member of the Board of Directors of the Amyloidosis Foundation since 2014. Her support group meets monthly in The Vanderbilt Clinic.

"I decided to start the amyloidosis support group because I had no one to turn to when I went through my heart and stem cell transplants. Having a rare disease is difficult, but having no one to talk to about what to expect makes it even harder," Charlotte said. "The support group is very important to me. It gives me a chance to give back, to share my experiences and maybe help smooth the way for someone who is confused and afraid."

Vanderbilt formally established Vanderbilt Amyloid Multidisciplinary Program (VAMP) in 2011 to advance the understanding, treatment and research of amyloidosis, joining Boston University and the Mayo Clinic as one of the few places to offer a multidisciplinary approach to the often fatal disease.

Aside from streamlining clinical management of these patients, VAMP is dedicated to improving diagnosis, disease monitoring and treatment through research, including studies of potential biomarkers, noninvasive imaging and drug trials.

Charlotte now says, "I am not in such a hurry anymore. Things aren't as urgent as they used to be," she said. "You've got to stop and smell the roses." **AF**



(continued from page 5)

His team is also preparing for a clinical trial to test a new imaging agent. Since not all patients' amyloids lit up with the radioactive amyloid, Wall found and patented a peptide that has shown specificity for binding to and lighting up amyloids. It has also shown promise for helping people with Alzheimer's, diabetes and certain forms of cancer. Clinical trials in patients with amyloidosis are slated to begin next year.

"If our imaging agent works, which we think it will, it will help patients understand their disease and get a diagnosis quickly," he says. "That is so important because most of the treatments for amyloidosis, such as chemotherapy, are toxic and are not tolerated well if you are extremely ill. So catching the disease early is key." Imaging the disease will also help pharmaceutical companies test drugs under development.

"What I would like is for us to develop the U.S.'s first imaging agent for this disease here in Knoxville and then see it fan out across the country and benefit these patients," he said. Wall credits many co-workers for their contributions to this work. They share his belief that amyloidosis patients are "worth working for" — even if it may force them to take a stress test every now and again. **AF**

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I knew about amyloidosis. I live near Pittsburgh, PA and both the Mayor of Pittsburgh and the Governor of Pennsylvania died from amyloidosis, in 1988 and 2000, respectively. I hoped that I was wrong.

A referral to a rheumatologist came next. She found no autoimmune disease, however MGUS was diagnosed as a result of my blood and urine tests. By this time, I was experiencing swelling in my legs and feet and shortness of breath, but it was not often and not terrible and it would always go away. My heart would sometimes skip beats as well but always went back to a normal rhythm.

In January of 2012, I decided to have carpal tunnel surgery. The EKG for the surgery showed something odd. A stress test was next and during the recovery phase I went into atrial fibrillation. A cardiologist thought that I had blockages, valve issues, or both. However, the cardiac CT showed nothing. The cardiologist said to take an aspirin each day and cleared me for surgery, which went without incident. That was the end of my appointments with a cardiologist at that time. I now know that this is often the path.

The rheumatologist told me that she wanted me to see an oncologist since MGUS could be a sign of multiple myeloma. I saw the oncologist in April of 2012. She ordered a bone marrow biopsy and other tests. The results showed that I had 15% plasma cells in my bone marrow. The oncologist wanted two more things: a second opinion from a multiple myeloma specialist; and an abdominal biopsy to rule out amyloidosis. She had listened to everything I described happening over the past and she said she just wanted to rule it out. But, my blood ran cold.

Four days later, on May 29, 2012, the results came back as positive for amyloidosis. My first question was, "Will my granddaughter ever remember me?" My daughter was due to have a baby on July 5th - my first grandchild.

(continued on page 8)

(continued from page 7)

I saw the specialist on June 1, 2012, who ordered tests for staging within two days and the results were that I was Stage II (tests prior to my stem cell transplant showed Stage I). Beginning on June 19, 2012, I did 20 treatments of Velcade, cell retrieval the Monday after Thanksgiving, and entered the hospital on December 10th. I had two days of high dose Melphalan and my stem cells were received on December 12th. I went through all of the issues, including C-diff infection, but I made it through them all. I hit clinical remission in the summer of 2013.

My light chains almost immediately began a gradual increase and it was decided that I should go back on Velcade in May of 2015. I completed my 16th treatment on July 24, 2015. I am going to take a break now, get more bloodwork in a few weeks and I will see my hematologist/oncologist on August 25th to discuss my next steps. I may need to do monthly blood monitoring again, until the light chains are high enough to demand more treatment.

I am very lucky and fortunate to have a doctor who partners with me in this journey-- he is the best and I am forever grateful for him. All of the pre- and post- tests show no organ damage. I want to be here and to benefit when they find the cure. I want to help to spread the word and increase the chances for a quicker diagnosis and patient survival. The support of my daughter and son-in-law have been incredible. I am truly not alone.

My granddaughter was born three weeks early and on the day I was to begin my first chemotherapy. A sign? I believe so. I skipped chemo, was in the delivery room to see her enter this world, and started chemo the next day. She turned three years old in June of 2015, she is my best friend and I plan to be here for a long time to watch her grow. **AF**

The Amyloidosis Foundation appreciates your continued support.

If you would like to become more involved in the foundation, interested in starting a fundraiser or becoming an amyloidosis ambassador—we would enjoy speaking with you and helping in anyway we can.

Please call our office today **1-877-AMYLOID** (877-269-5643) or send us an email at **info@amyloidosis.org**.

Thank you!

