The Amyloidosis Foundation Newsletter

Gateway to a Cure

Education • Awareness • Support • Research

Volume 7, Issue 2 • Winter 2014

www.amyloidosis.org Amyloidosis Foundation 1-877-AMYLOID

My Amyloidosis Journey

by Kathy Koontz

In October 2012, I was a healthy, very active 50 year old when I noticed my liver was enlarged and I started having some episodes of edema. I ate healthy, was trim and exercised regularly—riding 100 miles on my bike and attending exercise classes during which I kept up with folks 15 years younger than me. My primary care doctor ran some tests including an ultrasound and CT but came to the conclusion that my enlarged liver was of no concern. I knew I just didn't feel right and kept pushing for a diagnosis. Eventually, I was referred to a Gastroenterologist at the Ohio State University Medical Center who ordered many tests, including an upper endoscopy.

During that procedure, he biopsied a few spots in my stomach and the following week diagnosed me with AL Amyloidosis on February 25, 2013. What a scary time that was. Would I see my 15 year-old daughter graduate from high school, would I be there to help my 27 year-old daughter adjust to her new marriage, were my plans to grow old with my husband now shattered?

Three days after my diagnosis, I had my first appointment with my oncologist, Yvonne Efebera, MD, at the Ohio State University Comprehensive Cancer Center. I was diagnosed with kappa AL Amyloidosis effecting my GI tract, liver and kidneys, although all my organs were functioning well.

Kathy giving the keynote address in October at a professional conference in Nashville, TN.

Less than two weeks later, I completed one round of Velcade/Dexamethasone with a plan for high-dose chemo and an autologous stem cell transplant in April. The day after my last chemo infusion, I went on a weeklong ski trip with my family in Steamboat Springs, Colorado. Even though I was weak, in pain and had about 30 pounds of fluid in my abdomen, I skied with my family every morning.

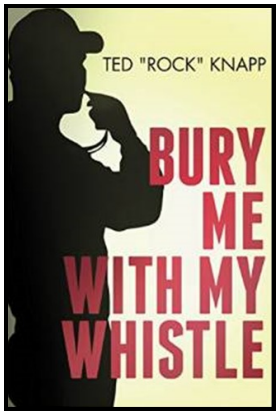
I didn't know if I'd ever have a chance to go on an active vacation—or maybe any vacation—with my family again. It was important for me to spend this time with them.

While in Colorado, I started my neupogen shots and four days after returning, my stem cells where harvested. The following week I was admitted into the hospital and began my stem cell transplant.

continue to page seven

The Courageous Story of Football, Family and Reaching Your Dreams

by Ted "Rock" Knapp



I was diagnosed with amyloidosis and multiple myeloma in the spring of 2008. I made it through that season, but it was a struggle. I did chemo three days a week and was taking over 40 pills a day. I finished my career at the end of that season...I was selected the South Georgia Coach of the Year.

As I entered retirement (full-time disability) I knew the challenges ahead would be many and overwhelming. But that was not my greatest concern. I'm a tough old linebacker and felt like I could handle whatever physical hardships awaited. My real concern was what I would do without football! I was 52 years-old and had either worn a helmet or a whistle for 48 of those years.

What could possibly fill that void?

Easter, 2009. I was sitting in the den watching all of our grandbabies playing on the floor. The thought occurred to me that if I passed in the next few years I'd only be a picture on the wall to these precious children. So, I decided to write a book for them; something that would connect us even after I was gone.

continue to page four

Amyloidosis Foundation Updates

Patient Resources

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

- Live Teleconferences with Experts (CD's available, and recording posted on website)
- Accurate Informational Pamphlets
- Website with information 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**

Follow Us

Stay connected for all the latest information on Amyloidosis:

Web: www.amyloidosis.org
Facebook: Amyloidosis Foundation
Twitter: Amyloidfnd
LinkedIn: Amyloidosis Foundation

Amyloidosis Foundation

2014 Research Grant Award Winners

We are pleased to introduce three researchers who were recently awarded grants by the Amyloidosis Foundation. Each year our Board of Directors chooses two junior researchers and one senior research scientist.

The primary purpose of the foundation in funding scientific research is to advance our mission and support research for all types of amyloidosis. With your generous donations this year, we were able to offer a monetary award of $50,000 provided to each of the junior awardees and $100,000 to our senior research grant winner. In their own words, here are their abstracts for each grant:

Guillermo A. Herrera, MD—Senior Research Grant

Kidney involvement and eventual kidney failure are common in amyloidosis. If the kidneys fail, the only options are dialysis and transplantation, both of which can cause significant complications. Today, amyloidosis is diagnosed earlier resulting in prolonged survival. The need for repairing damaged organs is obvious.

This grant investigates the use of stem cells administered intravenously to repair/heal damaged kidneys in an animal model. Stem cells are undifferentiated and can transform into specialized cell types. Results from this study will pave the way to use stem cells in human patients with amyloidosis to improve survival and quality of life. Dr. Herrera works at LSU in Shreveport, LA.

Jennifer Kollmer, MD —Junior Research Grant

A severe impairment of the peripheral nerves is one of the main manifestations in hereditary amyloidosis (TTR-FAP). Our grant will develop a new and highly sensitive diagnostic tool **1)** for the detection of very early nerve damage in gene-carriers without symptoms (family members of patients with already symptomatic disease), and **2)** for the monitoring of nerve lesions in patients with symptomatic amyloid polyneuropathy under treatment. Our study of participants will be done in different groups of disease severity. We will then perform a MRI of the lower limb peripheral nerves to find out which have the highest sensitivity in detecting peripheral nerve damage and determining a certain stage of polyneuropathy. This will hopefully lead to earlier treatment after initial diagnosis and successful therapies for patients with advanced disease. Dr. Kollmer works at the University of Heidelberg in Germany.

Rockland L. Wiseman, PhD—Junior Research Grant

The systemic amyloid diseases are a group of diseases caused by the build-up of unstable proteins that form toxic tangles in the blood. These tangles accumulate on organs such as the heart, gut, and kidney, leading to organ failure and ultimately death. Currently, no treatments besides invasive surgery exist to treat the majority of these diseases, making systemic amyloid diseases a large, unmet medical need. We are developing new strategies to reduce the lethal accumulation of unstable proteins associated with systemic amyloid diseases by enhancing the natural, protective pathways that regulate the levels of unstable proteins in the blood. Our establishment of this strategy will demonstrate that a single therapeutic approach can be used to treat many different systemic amyloid diseases, and therefore that it may be possible to use a single drug to broadly treat these devastating disorders. Dr. Wiseman works at The Scripps Research Institute in La Jolla, CA. AF

President's Corner

This is a special time for us here at the foundation. Awarding the research grants to the talented recipients and looking forward to their success that will surely follow is very exciting. These grants ensure your support is dedicated to projects that will provide the best outcomes for patients in the amyloidosis community.

We sincerely appreciate everyone who is committed to this important cause: our donors, patients, corporate sponsors, board members, physicians and community partners. You all work tirelessly all year to push for medical discoveries that will have a positive impact on the treatments and cure for amyloidosis.

Please join me in welcoming Dena Heath and Charlotte Haffner to our Board. Both have been active volunteers with the foundation for many years and we are thrilled to have their passion and drive on our BOD. We know 2015 will bring new endeavors to our foundation and we are proud to have these ladies on our team.

On behalf of our Board of Directors, Scientific Advisors and staff at the foundation, I wish all of you a wonderful holiday season and Happy New Year.

Sincerely,

Mary E. O'Donnell

Letters to the Foundation

I am both a volunteer as well as an Amyloidosis patient. If it were not for this wonderful organization spreading the word, educating all of us and helping young researchers in the medical community, I would most likely be dead by now. Since I was diagnosed over six years ago, I have seen so many advancements in medicine. Patients are surviving longer than the predicted 18 months we were originally told.

Many of us are fortunate enough to be living very productive lives and without financial support to this wonderful organization the wheels will begin to slow down. We can't let this happen as more and more people are diagnosed every year. Come join our choir, we can always use a new voice to help us stop this dreadful disease or at least make our lives easier to live.
—Donna U.

All of the amyloidosis diseases are confusing and complicated, there is little information out there that is straightforward and helpful. The Amyloidosis Foundation went above and beyond to help me and my family when my brother was diagnosed with AL amyloidosis. The website contains medical articles that are informative. The volunteers with the organization are very supportive. I am very impressed with their fundraising efforts and the annual grants that further awareness and research.

If it weren't for this foundation, I would have been unable to navigate the medical jargon. They know all the specialists in this disease so they can guide anyone, worldwide, to medical professionals who can help them. I am very grateful that they were there for my brother and my entire family. When I call them, they talk to me right away and always call back when needed.
—Anonymous

Gateway to a Cure 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 or **7151 North Main Street, Ste. 2, Clarkston, MI 48346**

Welcome to the Amyloidosis Foundation - Board of Directors

Dena Heath

Dena is happily retired from the corporate world, mostly spent at AT&T and consulting firms. She has a BA and MBA, both earned while working full time and as a single parent. She is an Amyloidosis widow who lost her husband to AL Amyloidosis (renal) in 2010, seven years after his stem cell transplant at Stanford and two years on dialysis. Dena has been volunteering at the Amyloidosis Foundation awareness booth at medical conferences for the last seven years and loves every minute of it.

Dena is also the Northern California Amyloidosis Support Group Facilitator (they just celebrated their tenth anniversary of quarterly meetings.) They are a well-informed and growing group of 40-50 participants that includes AL and hereditary patients, heart and stem cell transplants, kidney and liver transplants and all forms of the various chemo therapies as well as NEOD001 participants. Dena is passionate about Amyloidosis awareness and she is currently working on several exciting projects that will support both physicians and patients.

She believes retirement is a gift and a blessing. This has given her the opportunity to enjoy her work with the Amyloidosis Foundation. Dena also values time with her family and travel adventures that include white water rafting and trips to foreign countries and to pursue artistic interests.

Charlotte Haffner

Charlotte was diagnosed with primary AL amyloidosis in 2008. She was the first patient to receive both a heart transplant and a stem cell transplant at Vanderbilt University Medical Center.

Ms. Haffner has become a titan in the amyloidosis community. She volunteers on behalf of the Amyloidosis Foundation at their awareness booth at medical conferences, leads the amyloidosis support group at Vanderbilt University Medical Center (2nd Tuesday of each month) and has made numerous press appearances to generate awareness of amyloidosis. She is an excellent community builder, and has been involved in many fundraisers.

Charlotte was an equine midwife in Lexington, Ky., is an avid fly fisherman, and an accomplished horsewoman who still enjoys riding her horses on her farm in Franklin, TN. AF

Bury Me With My Whistle *(continued from page 1)*

So, I wrote, "The Great Adventures of Sparky the Angel." It has sold over 6,000 copies in the past 2 years! It continues to sell and has had real impact for many...including the grandkids!

The idea for, "Bury Me With My Whistle" came from years of friends and family prodding me to write a book about my career as a football coach and all the amazing stories and even miracles of those magical years of wearing a whistle around my neck. The release date was September 30, 2014 and already it has sold out 3 times on Amazon! It is about so much more than football. It is about relationships, it is about big dreams and reaching those big dreams. It is about love and it is about overcoming impossible odds.

It will make you cry and it will make you laugh. I made sure the entire manuscript was marinated in spiritual truth and written to provide hope for those facing major health issues.

Looking for a good read? Looking for one that will inspire you to celebrate each day and to live each of those days to the fullest of your potential? Well, today's your lucky day! And, may I wrap this up by encouraging you to keep fighting, to press on...taking hold of the unspeakable joy of the Lord and his peace that passes all understanding. AF

Biography: Ted "Rock" Knapp was diagnosed in 2008 with AL and Multiple Myeloma. He had several stem cell transplants, lost both his kidneys and decided against a kidney transplant (currently on dialysis 3x/week). Coach Rock is 58, lives outside Atlanta with his wife Shari and they enjoy their five kids and many grandchildren. He has written two books since he was diagnosed (self-published in 2012 "The Great Adventures of Sparky the Angel" and 2014 "Bury Me With My Whistle"). He will be on a 10 city book tour in 2015 that includes Houston, Chicago, San Diego and San Francisco.

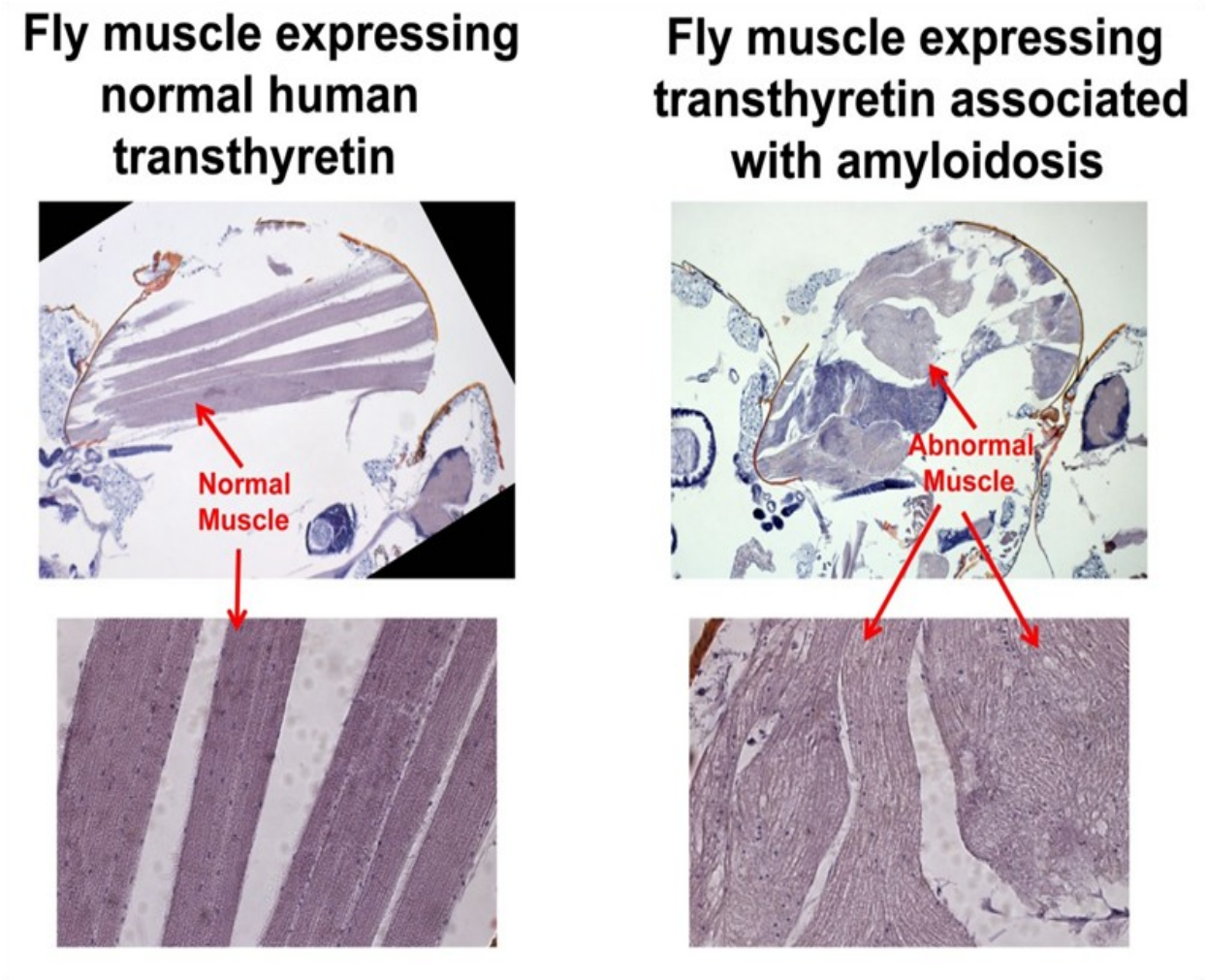
Coach Rock began his coaching career in 1980. As a head high school football coach, he amassed a 144-39-1 record, and won nine state championships. He was named Coach of the Year several times, including the prestigious 1998 Texas High School Private School Coach of the Year. AF

2013 Senior Research Grant Update

Matthew Wolf, MD, PhD—Duke University

Inherited changes in a protein called transthyretin (TTR) are associated with a condition known as cardiac amyloidosis or familial amyloid cardiomyopathy. I care for a family that has this condition and these interactions inspired my research project. My laboratory uses the fruit fly, *Drosophila melanogaster*, to model human cardiomyopathies and we are using this approach to develop new insights into cardiac amyloidosis. To accomplish this goal, we made transgenic flies that express either normal human transthyretin or a variant of transthyretin that occurs commonly in individuals who have cardiac amyloidosis.

The transgenic flies that have a variant of transthyretin develop changes in muscle consistent with changes observed in human amyloidosis and have problems with the way it functions. We also measured cardiac function in the transgenic flies using a method that is similar to echocardiography in humans and found abnormalities. These exciting results suggest that we can use fly genetics to identify new molecules that may prevent or slow the progression of cardiac amyloidosis. We have also made transgenic mice and hope to build tools that are necessary to translate research from the fruit fly models to mammals.



Human transthyretin (TTR) that is associated with amyloid causes abnormal muscles in fruit flies. The left panels show a fly that has normal human transthyretin and has normal appearing muscle. The panels on the right show that a fly that has human transthyretin associated with amyloidosis has abnormal appearing muscle. AF

A Life of Fighting Against FAP-ATTR Amyloidosis

by Fabio Figueiredo de Almeida, Brazil

After so many years watching my Father languish to the point where he couldn't get up from bed suffering from terrible pain, knowing that the end was inevitable - what could I do? Nothing, just wait. I was 23 years old when my father passed away in 1999. He was 47 years old and had no chance to win the fight against this terrible disease. We faced 11 years of atrocious suffering, which marked the childhood and adolescence of his 4 sons and his wife.

I was aware that I could be carrying the mutated gene. The years went by and we started living lighter because that nightmare had ended, but somehow I lived with the threat of the disease in my mind. The internet made things easier and when I thought of the disease I always researched about it to keep myself updated. Liver transplant for the FAP was already a successful treatment even in Brazil, though it didn't reduce our fear of having to face everything again. It is a complex surgery, with chances of death and post surgical complications, in addition to waiting in a long line for two to three years for the transplant. We didn't have time to wait.

In 2004, I received my Portuguese citizenship with the intention of one day, if needed, going to Europe to seek treatment for the FAP.

In 2006 I decided to do the DNA test, because I was already feeling some tingling in my feet which kept coming and going.

I had no escape and the result was as expected: the mutation MET 30 (codon Val30Met) in the TTR gene had been detected. This result is compatible with the diagnosis of Familial Amyloidosis Transthyretin. I lived each day at a time, praying the disease did not progress.

I was very happy for a few years until the end of 2011, when I was 36 years old and things changed. Every time I got home I felt leg fatigue and which was progressing to terrible pain: numbness, shocks, burning and needle feelings, superficial pain, muscle pain. After many medical consultations, I had few results: I was not eligible for the transplant list because the neurological tests had normal results. I prepared to go to Portugal to consult with specialists because I had read on the internet that the first drug to treat the FAP had been approved: Tafamidis.

I heard that Dr. Marcia Waddington Cruz, from the Federal University of Rio de Janeiro Hospital, specialized in FAP. She was participating in studies with this drug (Tafamidis, Pfizer's trade name now is Vyndagel). She and her wonderful team changed the course of my life.

My brother and I went to Rio de Janeiro because he, who is five years younger, was sick too. We did all the tests and amyloid deposits were detected. With such results, we could also finally qualify for the transplant list in São Paulo, but the liver transplant doctors were very skeptical about the simultaneous treatment with Vyndagel. We went ahead and chose the hard choice: to get Vyndagel! We filed a lawsuit against the Federal Government and, after just one month, the judge gave us a favorable decision: we were granted the legal injunction. Five months later, I could barely believe when I put my hands on the much awaited drug.

I write now after months of treatment with Vyndagel. I have gained weight and have less pain. I have been on the transplant list for more than a year but no call yet, I hope not to need the transplant.

I'd say never lose hope and always believe that everything is possible. We can't always agree with things, we must also act. AF

2012 Junior Research Grant Update

by Jennifer Ellis Ward, PhD—Boston University Medical Center

We are continuing the studies funded by the Amyloidosis Foundation, which we presented in two posters at the ISA meeting in April 2014. There has been much success with therapies that target the plasma cell source of the Light Chains that form amyloid. However, there are no approved therapies to remove the existing amyloid burden and that is what my work is interested in targeting. Currently we are working on two publications, one describing a method for quantifying the amount of amyloid in tissues and another describing the effects of different tetracycline antibiotics on Light Chain amyloid fibrils. We are using the experience we have gained and assays developed working with ex vivo fibrils to collaborate with a biotech company with a therapeutic targeting amyloid fibrils.

I am grateful for the foundations' support to develop innovative therapies for AL amyloidosis. AF

Fundraising & Crowdfunding = Success for Amyloidosis Foundation

The Amyloidosis Foundation participated in the RiseDetroit Charity Challenge earlier this year. Over 125 nonprofits in the metro Detroit area took the challenge to raise online donations via crowdfunding on a site called crowdrise.com.

Crowdfunding is the practice of funding a project or cause by raising monetary contributions from a large number of people, typically via the internet. Social media is used to promote donations and has become a very popular way to raise money with very little overhead. People worldwide have used various sites to raise awareness and increase fundraising for their charity and/or nonprofit.

The RiseDetroit challenge was six weeks, September 15—October 31st. We were lucky enough to have four teams raise funds along with the foundation. The teams were made up of friends and families affected by amyloidosis. They reached out to their own social networks and in total, we raised $4,855 with thanks to 60 donations.

We are very grateful to have such generous donors who fund our mission: to increase education and awareness of amyloidosis and support research towards a cure. Thank you to everyone who donated, we appreciate you all. AF

My Amyloidosis Journey (*continued from page 1*)

I had a lot of fluid issues in the hospital and gained 60 pounds of fluid, so much that I was barely able to move for many days. This resulted in me being able to walk only 40 feet upon discharge from the hospital after three weeks.

I spent one week and two days in a rehab hospital so I would be able to walk 500 feet unassisted and climb six stairs. My how my physical condition had declined! I returned home on May 10, 2013 and began the work of rebuilding my stamina and creating a new life of living with Amyloidosis. My husband pushed me every day to do my exercises and complete my physical therapy so I could return to my prior level of fitness. I began traveling to my daughter's lacrosse tournaments over that summer, requiring long car trips out of state for the weekend. In July, I returned part-time to my work as analytics executive and went back to full-time the following month.

That summer, I received wonderful news that the transplant was a success. I had a complete response and I was in REMISSION!! As time progressed, I regained more strength and energy, and life was returning to normal. I was able to return to some of my previous exercise classes and I also started cycling again. The following winter I took a few ski trips and was able to ski as hard and as long as I had before.

In August 2014, I rode my bike 180 miles over two days as part of a fundraising ride for cancer research (after riding more than 1,000 training miles.) I rode the first day with my husband and my oncologist's nurse practitioner, and the three of us crossed the finish line side by side. That was the line when I crossed over from being a patient, proving to myself and others that I had left the effects of the disease and treatment behind.

My organs have recovered well and continue to have normal function. My liver size has reduced and my kappa free light chains continue to be below the low end of normal. Aside from a slight elevation of my 24-hour urine protein, all my lab results are normal and I don't take any medication related to Amyloidosis.

This fall, I started competing in a new cycling discipline called Cyclocross. I am performing better than women much younger than me. I joke that I always win the Amyloidosis division.

In October 2014, I gave a keynote address at a professional conference to 3,000 colleagues from 60 different countries. I now have a new appreciation for life and loved ones, and try to live each day as fully as possible in the service of others. My faith in God, support of my loved ones and an incredible medical team brought me through this difficult time. From the beginning, I felt God telling me we would beat this, and we have so far. My life is normal and full and wonderful. Of course I occasionally think about relapse, but I know that new drugs and research will provide me with more effective treatment options than I have today. I don't think about it because I'm too busy living this life that God has given me.

As Amyloidosis patients are diagnosed earlier and new therapies are developed, our future will continue to grow brighter and brighter!

If you want to know more about my story, you can visit my blog at http://**katseyeview.blogspot.com/**or view a video about my recovery http://**vimeo.com/92131205**. AF

National Rare Disease Day

February 28, 2015

Need a unique holiday gift item?

$10 from each item purchased will be donated to the Amyloidosis Foundation.

https://www.bravelets.com/bravepage/amyloidosis-foundation

Upcoming 2015 Conferences

March

American Cardiology Conference
San Diego, CA

September

Heart Failure Society of America
National Harbor, MD

November

The American Society of Nephrology
San Diego, CA

December

American Society of Hematology

Amyloidosis Foundation

We Need Your Support!

We are truly thankful for the generosity of our donors. With this support we are able to provide all of our publications and services free of charge.

We have several volunteers who organize local fundraisers to support the foundation, golf outings, art exhibits, walk-a-thons and other activities. If you are interested in gathering your family and friends to organize a fundraiser please contact us (1-877-AMYLOID or info@amyloidosis.org) and we will provide support and assistance.

The foundation's success is totally dependent on donations and these fundraisers.

Thank you

26% of Each Online Purchase Helps Amyloidosis Foundation.

Start iGiving in 3 easy steps:

1. Join iGive.com for FREE
2. Shop online at any of the 800+ stores in the iGive network. You'll see all your favorites, including Amazon.com, Best Buy, Staples, eBay, and Pottery Barn, etc.
3. Amyloidosis Foundation receives a check for up to 26% of each purchase! It's free, it's easy, and every purchase you make generates a donation to your favorite cause.

iGive.com™
Change online shopping for good.