

# Gateway to a Cure

Education • Awareness • Support • Research

Volume 8, Issue 1 • Spring 2015

[www.amyloidosis.org](http://www.amyloidosis.org)



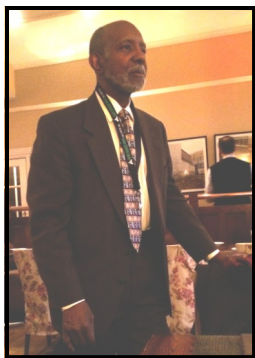
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## PATIENT ADVOCATES LOBBY CAPITOL HILL

### THE AMYLOIDOSIS FOUNDATION ATTENDS RARE DISEASE WEEK IN WASHINGTON, DC TO SEEK CHANGE



The Amyloidosis Foundation sponsored five patient advocates to attend Rare Disease Week on Capitol Hill from February 23-27, 2015. This eventful week in



*Len Strickland*

Washington, DC was organized by the Rare Disease Legislative Advocates (RDLA) and brought together the force of over 200 rare disease

patient advocates from across the nation. The NIH reports that there are nearly 7,000 rare diseases, affecting nearly 30 million Americans. By coming together, the ambassadors for these diseases used their



*Isabelle Lousada*

collective strength to make an impact on rare disease legislation.

Our Action Advocates were: Isabelle Lousada, Dena Heath, Len Strickland, Kim Ottone Tank, and Mary Ellen Thomas. Each one of these advocates has been touched by amyloidosis in a profound and personal way. They attended the conference and multiple panel discussions while learning about the current legislation on rare disease policies in the Federal Government. They each had appointments to meet with many congressional representatives (both Senate and House members) to address issues of concern and lobby for increased awareness of amyloidosis.



*Dena Heath*



*Mary Ellen Thomas*

In addition, our advocates focused on the FDA's implementation of rare disease provisions and clinical trial access for amyloidosis patients.

The week's events concluded at the National Institutes of Health (NIH) in Bethesda, MD on February 27<sup>th</sup>, which they designated as their "Rare Disease Day."

The Amyloidosis Foundation presented a research poster on amyloidosis diseases that was accepted by the NIH to be included



*Kim Ottone Tank*

in the poster session on this important day. This poster highlights the recent survey study results from the amyloidosis patient community. These results contain significant data on how the lack of diagnosis and the limited access to appropriate treatment directly impacts the health of amyloidosis patients.

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# 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.

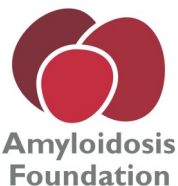
- Live Teleconferences with Experts (CD's available, and recording posted on website)
- Accurate Informational Pamphlets
- Website with information patients, caregivers and physicians
- Toll Free Number 1-877-AMYLOID
- Listing of experienced physicians that specialize in amyloidosis, it's diagnosis and treatment
- Email us anytime with questions: [info@amyloidosis.org](mailto:info@amyloidosis.org)

## Follow Us



Stay connected for all the latest information on Amyloidosis:

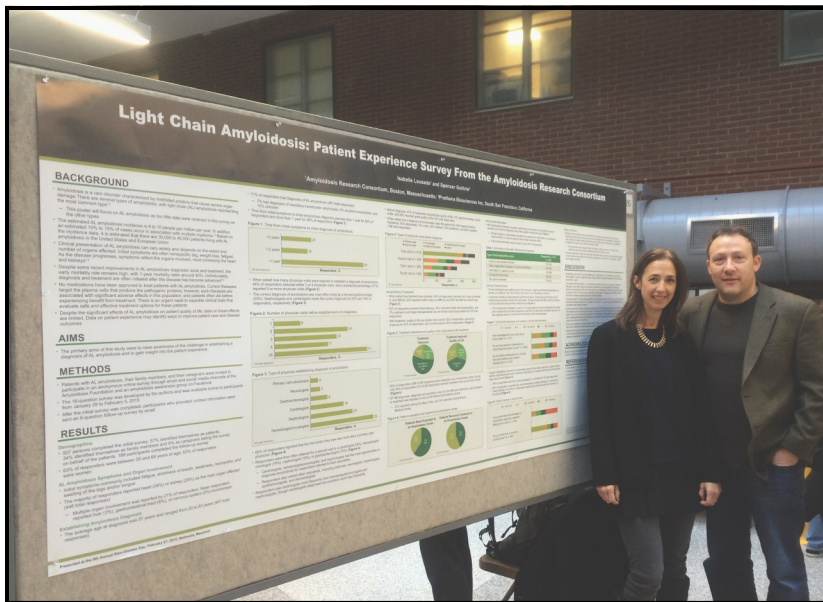
Web: [www.amyloidosis.org](http://www.amyloidosis.org)  
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## Rare Disease Week—Washington, DC

*(continued from page 1)*

The week was powerful and productive, putting amyloidosis front and center with the nation's legislators and the rare disease community. The Amyloidosis Foundation is proud to have been represented by such dedicated advocates during this week of events. **AF**



*Isabelle Lousada (AF) and Spencer Guthrie (Prothena Corporation) at the poster presented at the NIH.*

## Patient Webinar—Amyloidosis 101

The Amyloidosis Foundation is sponsoring our first patient webinar entitled “Amyloidosis 101”, presented by Dr. Morie Gertz, on Monday, March 16 at 4pm EST. He will review details on symptoms, diagnosis and treatment options. To register, click the link on the top of our homepage, [www.amyloidosis.org](http://www.amyloidosis.org). There is no charge for this webinar.



Dr. Gertz is the Roland Seidler Jr. Professor of the Art of Medicine and Chair of the Department of Internal Medicine, at the Mayo Clinic in Rochester, Minnesota. He is a Master of the American College of Physicians. Undergraduate degree was awarded with highest distinction from Northwestern University graduating Phi Beta Kappa. Dr. Gertz has served as a member of the Scientific Advisory Board of the Amyloidosis Foundation since 2006.

## President's Corner

Thank you to everyone who supported Rare Disease Day 2015. It's refreshing to see people all over the world come together to highlight diseases such as amyloidosis.

You are not alone in your amyloidosis journey and we are here to support you. This issue is filled with patient profiles, unique fundraising ideas and examples of patients, families and physicians advocating for a cure. Please join us in 2015 to bring awareness of amyloidosis in your community. Contact us at [info@amyloidosis.org](mailto:info@amyloidosis.org) or 1-877-AMYLOID and let us know how we can help you.

It is with sincere regret that I announce the passing of our longtime Treasurer, Rick Cwiek. He was a friend and valued asset of the foundation for many years. We will miss his integrity and passion for finding a cure for amyloidosis. Rest in peace, Rick.

Thank you to our Development Director, Boris Sellers, for seven years of service at the foundation. Your dedication and humor will be missed and we wish you well in your retirement in Nevada.

Enjoy the Spring flowers everyone and share time with your friends and family in this warmer weather. We all deserve it!

Sincerely,

Mary E. O'Donnell

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## Amyloidosis Physician Speaks at Michigan Rare Disease Day

On Thursday, February 27, over 60 residents in Michigan attended the Rare Disease Day event at the State Capitol in Lansing, sponsored by the National Organization of Rare Disorders (NORD). Governor Rick Snyder issued a signed proclamation for the event and many legislators attended and spoke as well.

Dr. Jeffrey A. Zonder, Associate Professor in the Departments of Oncology and Medicine at the Karmanos Cancer Institute and Wayne State University, was first to address the audience. His passion for rare diseases, especially amyloidosis, was evident throughout his speech. We applaud his efforts and dedication to his patients.

Thank you Dr. Zonder for participating and spreading awareness with us at this important day. We look forward to Rare Disease Day 2016!

*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](mailto:info@amyloidosis.org), 1-877-AMYLOID or 7151 North Main Street, Ste. 2, Clarkston, MI 48346

## Amyloidosis Education at the Ohio State University College of Medicine

by Kathy Koontz

I participated in a two-hour lecture to 120 medical students at the Ohio State University College of Medicine on

Wednesday, February 11 with my oncologist, Yvonne Efebera, MD.

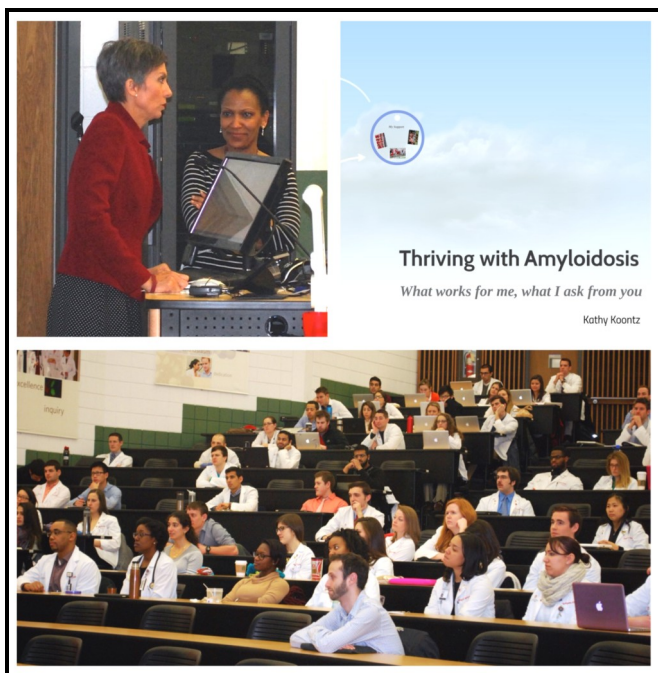
The students were very engaged. The lecture was divided into two parts, the medical overview and the patient profile. Students are required to attend the patient profile section, which I think is awesome because it's probably much more memorable.

The audience was really focused on my medical history, how challenging it was for me to be correctly diagnosed and the details of my

road to recovery. The publications and pens provided by the AF were well received.

When the lecture was done, Dr. Efebera invited students down after the lecture to speak with me and to feel my liver. Six students came down and felt my abdomen while we spoke. I get frustrated sometimes that my liver span isn't reducing but I'm happy being a bit of a medical sideshow if it will help current and future physicians think of amyloidosis more often during their diagnostic process.

I am so thankful Dr. Efebera gave me the opportunity to participate and that Ohio State devotes this time to amyloidosis education. I know those students will enter their medical practice better equipped to recognize amyloidosis. **AF**



## Ice Fishing Derby Fundraiser for Amyloidosis!

In January this year, Marla Walsh had a last minute idea to raise funds for the Amyloidosis Foundation and to honor her husband Tim, who passed away from the disease in 2009. She lives in Wisconsin and twice a year (January & July) they offer "fishing for free", so you don't need to purchase a fishing license to fish.

Marla decided to have an "Ice Fishing Derby Fundraiser". The entry fee was \$10 and they offered complimentary chili and coffee throughout the day on Lake Como, WI.

Her group of volunteers sold 50/50 raffle tickets with the winner to receive half of the raffle and the other half donated to the Amyloidosis Foundation.

They had a First, Second and Third Place challenge for the largest Bass and the largest Northern.



Steve Trimborn, lucky fisherman!



Kathy Kronwall, Ashley Klein, Marla Walsh (Organizer and wife of Tim) and Lauren Hatchett.

Once those winnings were paid out and after they split the 50/50 raffle, they were able to donate \$656 to the Amyloidosis Foundation.

Another person wrote a check to AF for \$25 for a grand total of \$681! Thank you Marla and your hearty group in Wisconsin for your generous donation! We appreciate your support & good luck next year! **AF**

## TTR-FAP: A Life Changing Diagnosis for Me and My Family

by Robert Hauge (Originally published in the *Neuropathy Association E-News* Oct. 2014, reprinted with permission by the author)

I was 65-years-old when I first started experiencing numbness in the soles of my feet, weakness in my leg muscles, and heart problems. My symptoms were progressive: my weakness was increasing and my heart function was declining. Two years later, after several mis-diagnoses and attempts to address the constellation of symptoms, my primary care doctor referred me to a neurologist.

But it was my second neurologist who diagnosed me with transthyretin-mediated familial amyloid polyneuropathy (or TTR-FAP). My biopsy results showed the fibril deposits - the hallmark of amyloidosis. Next, we proceeded to determine the underlying cause, i.e., was there a genetic component to it? The analysis confirmed I had familial amyloidosis.

I learned that TTR-FAP is a rare, progressive, and fatal form of neuropathy affecting approximately 10,000 patients worldwide. There are over 100 reported mutations for TTR, and the particular TTR mutation and the site of amyloid deposition determine the clinical symptoms of TTR-FAP, which include two clinical syndromes: - familial amyloidotic polyneuropathy (FAP); and familial amyloidotic cardiomyopathy (FAC). I have the Irish mutation - which involves replacement of the amino acid threonine by alanine at position 60 along the sequence of amino acids.

I was on a mission to learn about TTR-FAP—and fast. TTR-FAP is inherited in an autosomal dominant manner, meaning each child of a gene carrier has a 50% chance of inheriting the variant gene.

This explains the urgency to get informed and to share this news with my family.

Much of my information has come from a visit with the world's expert on familial amyloidosis at the University of Indiana School of Medicine in Indianapolis, Indiana. Then, I wrote a letter to my two brothers and two sisters detailing my journey: my diagnosis, the symptoms, the tests involved, and the general consensus that there is no known cure for TTR-FAP. But most important, I wanted to let them know that TTR-FAP is an inherited condition and that it would be a good idea to get tested for our specific family genetic defect.



*From left to right, the Hauge siblings: Tom, Mike, and Robert (back row); Betty and Mary.*

We found out that four out of five of us siblings had the genetic defect. In fact, my brother Mike (age 69) and my sister Betty (age 77) had the symptoms, but were never diagnosed with TTR-FAP. My youngest brother Tom (age 61) has not yet developed any symptoms, although he has the genetic defect.

The realization that TTR-FAP is a possibility for the extended family, cousins etc. caused us to share our new-found information with every-one who might be affected.

It was becoming clear that we had inherited TTR-FAP from our mother once we began to look at my mother's side of the family. TTR-FAP was evident in my mother's father's side of the family and among my cousins. It is highly likely that our mother's father died of TTR-FAP without it being diagnosed.

I have to wonder if my mother and her brother John had the symptoms, but never were diagnosed. Both my mother and her brother were also battling cancers that had much more immediate health consequences. The TTR-FAP diagnosis was a big surprise, but it made me better appreciate my friends and family - and especially my wife Djehane - for all the help and encouragement they offered. Unfortunately for me, Djehane died on Dec. 3, 2011, after her own battle with cancer. However, the gift she did leave me with was her example of courage and graciousness as she lived her life in her last years. It has essentially left me unafraid of what lies ahead for me. However, next time I see mother I do have a bone to pick with her. She may already have had the same discussion with her dad (my grandfather) who has probably also spoken to one of his parents depending on who is the guilty party. One wonders how far back this goes...

I have learned that the only approved treatment involves a liver transplant, since that is where the TTR is made; and, for some patients, a heart transplant as well if the heart has been damaged.

*(continued on page 8)*

## 2015 U.S. Figure Skating Scholastic Honors Team Member Chooses Amyloidosis Foundation as his Charity of Choice

Ten U.S. Figure Skating athletes were named to the 2015 U.S. Figure Skating Scholastic Honors Team on January 25. The team was honored last week during a ceremony at the 2015 Prudential U.S. Figure Skating Championships in Greensboro, North Carolina.

The program, which started in 1996, was created by U.S. Figure Skating to recognize high school athletes who excel in academic pursuits, community involvement and the sport of figure skating. More than 1,100 skaters have applied for the scholarship since its inception, with 210 team members selected.

In addition to receiving a scholarship, each member is awarded an additional \$1,000 to donate to their charity of choice. For 2015 Scholastic Honors Team member Eric Stinehart, that program is the Amyloidosis Foundation. Eric's father, James, has AL amyloidosis and he wanted to honor his dad with this charitable donation.

Yvette says her husband James (Jim) is preparing for a stem cell transplant this year. She said Jim has tried to be positive and optimistic throughout this whole ordeal, sometimes still going to work on good days. They are extremely proud of Eric, plus his older brother James—who attends Princeton.

The award is available to U.S. Figure Skating members of all disciplines who are juniors or seniors at an accredited high school. They must be fulltime students with a grade point average of 3.4 or higher and have competed at the novice, junior or senior level during the past two years. Eric is one of only two juniors to be accepted this year.

Eric will graduate from New Trier High School in Winnetka, Illinois, in 2016 where he has a GPA of 3.9. An honor roll student during each quarter of his high school career, he is co-head for This is Our Music club, the New Trier Figure Skating Club and a member of the New Trier Symphony Orchestra.



2015 U.S. Figure Skating Championship in Greensboro, NC. Eric Stinehart, from Winnetka, Illinois is third from the left. Below, Eric and his father James in 2013 at the U.S.F.S.A. Nationals competition in Omaha, NE.



A member of the Skokie Valley Skating Club, he has volunteered at club exhibitions and competitions, and is a skating instructor. Champion of the 2013 Midwestern Sectionals, he medaled at the 2013 Prudential U.S. Figure Skating Championships as an intermediate-level skater.

*Congratulations, Eric!! AF*

### 2015 Conferences

#### March 14-16

American Cardiology Conference  
San Diego, CA

#### September 26-29

Heart Failure Society of America  
National Harbor, MD

#### November 3-8

The American Society of Nephrology  
San Diego, CA

#### December 5-8

American Society of Hematology  
Orlando, FL

## Special Event in NJ Raising Funds for Research for Amyloidosis

by Patty Claus

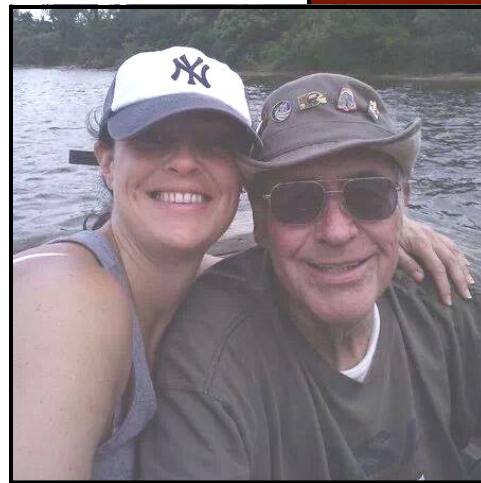
My father, Hugh Claus, was diagnosed in 2009 with Primary AL Amyloidosis and he passed away in December 2013. I joined the Chip Miller Charitable Foundation three years ago after his diagnosis. I've been fighting to spread awareness ever since. Chip Miller passed in March 2004, three months after he was diagnosed. The Millers then started the Chip Miller Charitable Foundation. For more info on this charity please go to [www.chipmiller.org](http://www.chipmiller.org).

The CMCF group has a long history with the Amyloidosis Foundation. The AF is grateful for their donations over the years that enable them to provide grants each year to fund much-needed research to find a cure for this tragic disease.

Join us on March 21st in Little Egg Harbor, NJ for a special night of fun and laughter, all in the name of research and awareness for amyloidosis. **Get your tickets today, contact Patty for more information: [patty4tino@yahoo.com](mailto:patty4tino@yahoo.com).** Please help raise money for research and awareness. **AF**



Patty and her father Hugh



**Chip Miller**  
CHARITABLE FOUNDATION  
[chipmiller.org](http://chipmiller.org)

PRESENTS:  
**A Night Of Laughter!**  
To raise money for Amyloidosis research and awareness.

Saturday March 21, 2015 5:00 pm  
\$40.00 PP advance sales by March 7, 2015.  
\$45.00 after March 7, 2015 and \$50.00 at the door  
18 years of age and older / Cash bar  
Cuisine on the Green 261 Country Club Blvd. Little Egg Harbor, NJ 08087

There will be raffles, food and fun!  
Not to mention the Comedic Stylings of:



Tickets can be purchased <https://www.eventbrite.com/event/chipmillerpresentsanightoflaughter-tickets-45567495>  
out to the Chip Miller Charitable Foundation.  
For more information contact Patty at [patty4tino@yahoo.com](mailto:patty4tino@yahoo.com)  
Remember people with the knowledge and understanding of  
the disease are the best people to help raise money for those afflicted with the disease and to help  
by the IRS as a not-for-profit 501 C (3) corporation.

## Family Journey of Amyloidosis

By David Jennings

The first time I heard the word amyloidosis was in January of 2010 when my mother, who was 68 at the time, called to discuss the results of her kidney biopsy. She had been seeing her nephrologist and internist frequently for a year or two, trying to get a diagnosis for something going on with her kidneys. I had not been following her medical situation very closely up until then, but that would soon change.

The diagnosis on the biopsy report was amyloidosis, but the pathologist could not definitively state that it was AL amyloidosis. A little bit of online research convinced me I needed to attend Mom's upcoming appointments and hear things directly from the doctors to put

myself in a better position to talk to her afterwards and also to help me relay information to my two sisters. So I prepared my list of questions and started going to doctor appointments with Mom, while at the same time seeking advice from both online and local support groups.

Her local hematologist realized he was not dealing with an AL amyloidosis patient after the bone marrow biopsy came back negative for amyloidosis, so Mom and I decided she needed to be evaluated at one of the amyloidosis centers. We found ourselves at the Amyloidosis Center at Boston University a few months later, talking to Dr. Martha Skinner after two days of testing and consultations.

They had ruled out AL and AA amyloidosis for Mom and suspected one of the rare familial types, but additional genetic testing would be required to determine which type. About a month later we were informed Mom has the mutation for fibrinogen amyloidosis.

Fibrinogen amyloidosis is a slowly progressing type of familial amyloidosis that impacts kidney function, eventually leading to kidney failure. Other than some drugs currently in the clinical trial phase, the only treatment for fibrinogen amyloidosis is organ transplantation.

I'm sure everyone reacts differently when they find out there is a hereditary disease in their family, but I made it my mission to learn all I could about this one and communicate to other potentially affected family members.

*(continued on page 8)*

## “A Life Changing Diagnosis for Me and My Family “

(continued from page 5)

With no more mutant TTR being produced by the liver, the thinking has been that amyloid deposition will cease.

However, I was told by my doctor that a liver transplant may not always help because normal TTR (referred to as wild type TTR) can continue to deposit on the existing amyloids. Given my age, we came to the conclusion that a liver transplant might not be a treatment option for me. After further research on TTR-FAP online, I found that there were several clinical studies underway on small molecules that enhance the stability of TTR as the desired four-fold tetramer form that circulates throughout the body.

My decision to participate in a clinical research study was a natural one for me since I have been involved in scientific research all of my adult life. I currently work in the nanoscience and technology field at Rice University. Another factor was that there are no FDA-approved medications for TTR-FAP. And, the medications in development – accessible via clinical studies -- were my only hope.

Several family members have also enrolled in the TTR-FAP clinical studies. We are excited to be part of the research efforts to either slow down the progress of FAP or perhaps to completely stop its progress. While Djehane and I did not have children, my siblings do...so our decision to participate is also driven by our children (my nephews and nieces) – so they, hopefully, will have more answers and treatment options for TTR-FAP. And, of course, we'd be helping bring forth the first wave of TTR-FAP treatments to the broader community.

I have realized I'm not alone in this fight. My family and friends have been very supportive.



*"My decision to participate in a clinical research study was a natural one for me since I have been involved in scientific research all of my adult life."*

This journey has also made me realize that there is a lot of love in this world: love from those who have provided all of the information on TTR-FAP; love from those who are doing the work to combat it; love from my friends who take time off from work and their own lives to accompany me to my appointments with the doctors; love from my family and especially my sister Mary (who does not have the TTR-FAP gene, but could not be more supportive). So I take it one day at a time – learning as much as I can about TTR-FAP and dealing with its effect on my daily life. But I do so with the conviction that the ongoing clinical studies will soon make TTR-FAP a treatable condition. **AF**

(continued from page 7) “Journey”

I saw no reason for any close relatives to go undiagnosed for years simply because of a lack of information. After informing Mom's closest relatives (her two brothers and my two sisters), I sent a letter to her surviving aunts and cousins that we knew how to contact. Since there was no family history of kidney disease, we did not know which of her parents my mother inherited the mutation from until a few years later when we learned of a cousin on her father's side who was diagnosed with it.

As for myself, I did test positive for the mutation but I am currently asymptomatic at the age of 52. Given the low penetrance of fibrinogen amyloidosis, I may never develop symptoms. Fortunately there are some very clear biomarkers that always appear as the first symptoms, so I have a good chance of early detection which should give me more treatment options.

As a result of being in touch with a few (very few) other people with fibrinogen amyloidosis, I began a blog in 2012 to not only tell about our family's experiences but also to provide a resource for others searching for information on this particular disease. I not only make available all of the relevant articles from the medical journals, but I try to explain the content of the articles in layman's terms to help those who are overwhelmed trying to figure out what they should do next.

Mom did not let her diagnosis slow her down by any means. She continued to travel extensively (including internationally) until she started hemodialysis in October of 2012. Her travels did slow down at that point, but she has still managed to go on a Caribbean cruise while on dialysis. As of early 2015 she is waiting on a kidney transplant yet remains reasonably active, maintaining a household and driving herself to dialysis three times each week. **AF**