amyloidosis foundation

News and Stories - Winter 2018

Fundraising Events Soar at the End Of 2017!

The Amyloidosis Foundation is lucky to have patients, friends, family and volunteers who organized events and fundraisers to fund research for a cure for amyloidosis.

Our 2nd Annual Pittsburgh Research Benefit was in October, raising over $50,000! AF Board of Director Darcy Tannehill and her team made sure the night was extra special. We enjoyed live music, a silent auction & delicious food.

The AF hosted our first 5k/10k run in Michigan on a sunny November morning with over 100 participants! Runners came from every corner of the state to show their support. We raised over $5800 and look forward to next year.

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What is Wild Type TTR Cardiac Amyloidosis?

by Mathew S. Maurer, MD, Arnold and Arlene Goldstein Professor of Cardiology, Columbia University Medical Center, New York Presbyterian Hospital

This form of cardiac amyloidosis is due to the deposition of normal or "wild type" transthyretin (TTR). This means that the transthyretin gene does not harbor any mutations. In other words, just the way that TTR exists in the "wild".

This form of transthyretin cardiac amyloidosis is different from the type seen in people who have a mutation in their transthyretin gene. That type is called familial amyloid cardiomyopathy. This difference is important. Since there are no known genetic causes for wild type TTR cardiac amyloidosis it means that family members are not at added risk for this condition.

What are other names for Wild Type TTR cardiac amyloidosis? Wild type transthyretin amyloidosis (which is abbreviated ATTRwt) has been called senile cardiac amyloidosis, senile systemic amyloidosis and age-related cardiac amyloidosis. These former names indicate that this condition predominantly affects older adults and that the principle organ affected is the heart.

How common is Wild Type TTR cardiac amyloidosis?

Among newly seen patients at most amyloid centers, ATTRwt cardiac amyloidosis is the most common type of amyloidosis. The increase in the number of patients being diagnosed with ATTRwt cardiac amyloidosis has been attributed to several factors including;

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Meet our new Board of Directors

Sheryl Kernodle, RN has called Vanderbilt Medical Center home for the last seven years, where she has worked as a Cardiac nurse in the Cardio-Oncology, Advanced Heart Failure/ Amyloidosis clinic at the Vanderbilt Heart and Vascular Institute in Nashville, TN. She has over 24 years of experience, with the last 17 focusing on Cardiac subspecialties.

In 2014, Sheryl began working with Daniel Lenihan, MD who was one of the Cardiologists at VHVI that specialized in the care of patients with amyloidosis in their Vanderbilt Amyloidosis Multidisciplinary Program (VAMP). at Vanderbilt as his clinic nurse.

She attended the American Association of Nurse Practitioners Conference (AANP) in June 2017 as a volunteer for the Amyloidosis Foundation and looks forward to attending more conferences in the future.

Sheryl joined the Board of Directors of the Amyloidosis Foundation in 2017. She is married and enjoys cooking, gardening and her favorite exercise is walking.

Mark Sutherland is a Financial Advisor/CFP, CRPC with LPL Financial in Southfield, Michigan. He attended Michigan State University, where he graduated with a B.A. in Business Administration in 1979.

Mark has supported the AF for many years, attending our annual golf outing events. He began working with AF President Mary O'Donnell and Treasurer Dante Burchi regarding investment recommendations for the foundation since 2015. He became a member of the Board of Directors in 2017.

Great to have you both on our team! ***AF***

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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.

- Webinar recordings posted on our website
- Updated informational pamphlets
- Toll Free Number **1-877-AMYLOID**
- Listing of experienced physicians that specialize in amyloidosis. Email us anytime

Our comprehensive website has information for patients, caregivers and physicians featuring:

- Treatment Centers (US / International)
- Support Groups (CA, TN, WA)
- Newsletters
- Webinars
- Fundraising Toolkits

Follow Us

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Stay connected for all the latest information on Amyloidosis:

Web: **www.amyloidosis.org**
Twitter: **@Amyloidosisfdn**
facebook: **@amyloidosisfdn**

President's Corner

I hope you all enjoyed your holidays with friends and family.

As we begin 2018, I am proud to welcome two new members to our Board of Directors, Sheryl Kernodle, RN and Mark Sutherland. Both share our passion to find a cure for amyloidosis. We've included their biographies and photos in this issue.

Thanks to all of our donors who gave in 2017, our total contributions are up over 29%! You are the reason we are able to carry out the mission of the Amyloidosis Foundation: supporting patients and families while promoting research, education and awareness. This year we will be celebrating our 15th anniversary! More details on how you can help us celebrate will be coming soon.

Have a happy and healthy new year,
Mary E. O'Donnell

2018 Amyloidosis Foundation Webinar:

"ATTRwt Cardiac Amyloid: Often Overlooked, Not Uncommon and Manageable"

Mathew S. Maurer, MD

Wednesday, February 7
3 p.m. (EST)

Register on our website: www.amyloidosis.org

Mathew S. Maurer, MD, is the Arnold and Arlene Goldstein Professor of Cardiology and Professor of Medicine at Columbia University Medical Center. A general internist and geriatric cardiologist with advanced training in heart failure and cardiac transplantation, he has received grants from the National Institute on Aging to study cardiovascular changes in older patients and specializes in the care of patients with heart failure and a preserved ejection fraction, including patients with amyloidosis.

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

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

If you no longer wish to receive this newsletter, please send us an email: **info@amyloidosis.org**

2018 Amyloidosis Foundation Research Grant Recipients

The Amyloidosis Foundation is proud to announce our 2018 research grant awardees, whose research targets the challenges in the field of amyloidosis.

We look forward to their success and hope for a cure in the near future.

Celia Torres Arancivia - PhD
Age-Related Cardiac Amyloid Disease (ATTRwt)
Amyloidosis Foundation David Seldin, MD, PhD
Memorial Research Grant, 2018
Boston University Amyloidosis Center

Matteo de Rosa - PhD
A New Route to the Development of Therapeutics
Amyloidosis Foundation Research Grant, 2018
National Research Council, Italy, Medicine Division: X-Ray Crystallography & Structure-Based Drug Design

Luke Berchowitz - PhD
A New Platform to Discover the Genes Involved in Amyloid Formation
Amyloidosis Foundation Donald C. Brockman Memorial Research Grant, 2018
Columbia University Medical Center, New York

Mario Nuvolone - MD, PhD
Investigating New Therapies to Treat AL Amyloidosis
Amyloidosis Foundation Research Grant, 2018
University of Pavia, Italy, Medicine Division: Amyloidosis Research and Treatment Center

Surbhi Sidana - MBBS
Understanding the Composition and Mechanism of Amyloidogeneis in Light Chain Amyloidosis
Amyloidosis Foundation Research Grant, 2018
Mayo Clinic, Medicine Division: Hematology

Alexandra Silva - B.S, Chemistry, PhD
Assessing Efficiencies in Machado-Joseph Disease (MJD) Therapies
Amyloidosis Foundation Research Grant, 2018
Instituto de Biologia Celular e Molecular, Portugal, Medicine Division: Biomolecular Structure and Function Group

Advocating for amyloidosis

On September 13, 2017, our Special Projects Director, Kathi Luis, represented the Amyloidosis Foundation at the 2017 Global Genes Summit in California.

The goal was to learn about how pharmaceutical/ biotech companies collaborate with nonprofit foundations and patient communities to ease the burden of patients participating in clinical trials. Says Kathi, "I was inspired by every patient, advocate, and fellow industry representative - it was truly a great conference that inspired me to act".

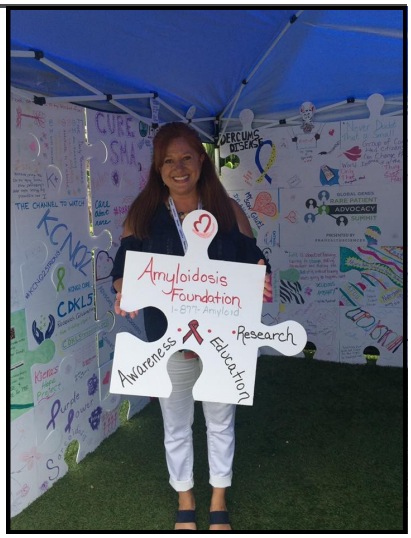
While at the conference, she rallied for medical research with the National Institute of Health (NIH) by emailing her senators and state representatives, giving rare disease patients a voice in Congress.

The AF is also a member of the Rare Disease Legislative Associates (RDLA) and Kathi is part of the Rare Disease Regulatory Science Working Group in the Community Congress.

The annual meeting was held last November in Washington D.C. and she participated in the Rare Disease Congressional Caucus Briefing to advocate with the RDLA on the challenges rare disease patients face. At this caucus, they urged members of Congress to support the OPEN ACT (Orphan Product Extensions Now, Accelerating Cures and Treatment), which would potentially double the number of treatments approved by the Food and Drug Administration (FDA) for rare diseases.

The News

While in Washington D.C., Kathi also attended the RareVoice Awards to recognize and celebrate advocates in the rare disease community who have made an impact at the state or federal level. On her last day, she went to the Community Congress Annual In-Person



Meeting, where the Regulatory Science group focused on payers and access. They discussed the gap between the FDA approval of a new therapy and the ability of patients to access it. During this meeting, Kathi learned of current legislative initiatives and activities plus urgent policy issues standing between rare disease patients and treatments.

The Amyloidosis Foundation is committed to partnering with these nonprofit patient organizations in the future to spread amyloidosis awareness and make sure patients are involved in decisions that affect their future. ***AF***

To make a donation to the Amyloidosis Foundation, go to **www.amyloidosis.org** or **scan the UR code** *(right)* **if you have a UR reader on your smart phone.**

Thank you for your support.

Fundraising Update

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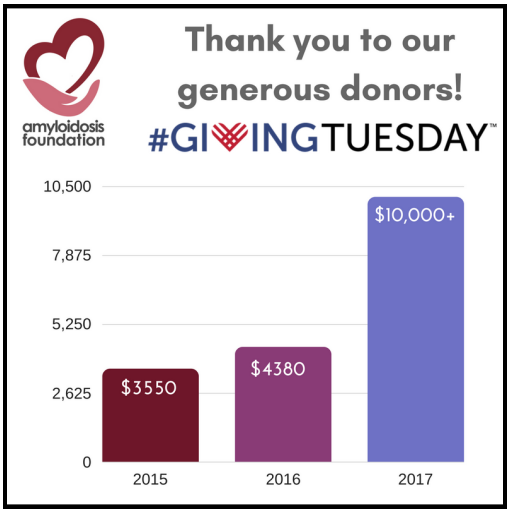
On November 4, Shawn Forman ran the 36th Annual Mountain Masochist 50 Mile Ultra Marathon with his best friend Joe to raise money and awareness for the AF. They ran in memory of Shawn's mom Kathy who passed away in 2011. They finished in under 10 hours and raised over $1650!

Pennsylvania to Georgia. We are so thankful for the support of everyone who participated in this annual day of giving. ***AF***

This year the AF raised over $10,000 on **#GivingTuesday**, November 28. Donations came from near and far - Poland to California,



What is Wild Type TTR Cardiac Amyloidosis?

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* the increasing age of the population (as people live longer, there is a larger cohort at risk for developing the condition),

* the use of non-invasive imaging techniques, especially nuclear scintigraphy, to diagnose the condition without the need for a heart biopsy and

* the appreciation that 10-20% of patients with heart failure, particularly in the setting of a preserved ejection fraction (which is a measure of the pumping function of the heart) can have ATTRwt

In fact, due to the aging of the worldwide population, it is anticipated that ATTRwt will become overall the most common form of systemic amyloidosis.

How is the diagnosis made?

In the past, diagnosis of ATTRwt cardiac amyloidosis required a cardiac biopsy to confirm that not only was amyloid present but that the protein causing the amyloid deposits was transthyretin.

Also required were special stains or use of a microscopic laser to dissect the amyloid fibrils,

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Wild Type TTR Cardiac Amyloidosis

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chop up the protein and to determine its origin (called laser dissection mass spectroscopy).

More recently, a relatively simple imaging test that employs an isotope called technetium pyrophosphate (Tc^{99}-PYP) (which was used for many decades to image bone) can diagnose TTR amyloidosis without a biopsy. The PYP scan needs to be coupled with blood tests to make certain that it is not a case of AL or light chain amyloid.

Finally, to confirm that the transthyretin protein does not have any mutations (e.g. is wild type) a genetic testing of the TTR protein needs to be performed.

How does wild type TTR cardiac amyloid manifest? Symptoms can be explained by their underlying heart condition. Such symptoms include fatigue, inability to exert much effort, shortness of breath, palpitation, loss of consciousness, swelling (edema), abdominal bloating or an inability to lie flat because of shortness of breath.

There is a pre-symptomatic period in which patients may be diagnosed with this condition because of amyloid detected by a biopsy during a different examination (carpal tunnel release/surgery or lumbar spine surgery or in the gastrointestinal tract). Such patients may have evidence of cardiac amyloidosis based on cardiac testing including an electrocardiogram or echocardiogram.

How is wild type TTR cardiac amyloidosis managed? The key to managing this condition is to keep the fluid retention under control, maintaining a normal amount of fluid in the body, which is called euvolemia. This is accomplished by monitoring one's weight on a daily basis.

For changes in weight that are significant (usually 2-3 lbs depending on one's body size) and occur quickly (over a few days), patients are encouraged to contact their providers. In these situations, medication adjustments, usually diuretics, can remove the excess fluid in the body relatively quickly and improve one's symptoms. It is important to eat a low sodium diet in the setting of cardiac amyloid, because the more salt is consumed the more fluid is retained.

Additionally, some medications, especially non-steroidal anti-inflammatory drugs, which are used to treat pain, can exacerbate fluid retention in patients with cardiac amyloidoses.

Are there treatments specifically for ATTRwt cardiac amyloidosis? At present, there are no specific treatments for this condition that have been absolutely shown to make one feel better or live longer. However, medical management including careful use of diuretics and other cardiovascular medications can be beneficial. For patients with atrial fibrillation (an abnormal heart rhythm) it is important to take an anticoagulant to prevent a stroke. Additionally, in some patients, restoration of sinus rhythm for patients in atrial fibrillation is quite beneficial. It is also important to avoid certain medications including the use of some calcium channel blockers (such as verapamil and diltiazem).

Of note, there are a host of emerging therapies for this condition that are either in phase 3 trials (e.g. if they work they may be approved by the FDA) or are entering into clinical trials.

All affected patients are encouraged to consider enrolling in a clinical trial. You can find out about these trials by looking at **www.clinicaltrials.gov**.

These emerging new treatments can be described as belonging to one of three categories: 1)those that attempt to **stabilize** the protein, and thereby would prevent the disease from progressing; 2) those that **silence** the production of the protein in the liver, which would also keep the disease from progressing, if they work; and eventually, 3) it is hoped that an agent will be developed that can be shown to **remove** the protein build-ups in the heart, reversing the condition.

Remember that none of these potential treatments have yet been proven. ***AF***

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