amyloidosis foundation

News and Stories - Summer 2022

Amyloidosis and the Heart

By Melissa Lyle, MD

Amyloidosis is an umbrella term that incorporates a heterogenous group of disorders characterized by misfolded proteins. The misfolded precursor proteins bind together to create amyloid fibrils which can deposit into different organs and tissues. More than thirty different proteins can misfold and cause amyloidosis, but the two most common causes of amyloid heart disease are immunoglobulin light chain (AL) amyloidosis and transthyretin (ATTR) amyloidosis.

Amyloid fibril deposition in the heart can result in thickening of the heart muscle and stiffening of the heart, a condition sometimes called amyloid cardiomyopathy.

As a cardiologist specializing in the treatment of cardiac amyloidosis, I am often asked how amyloid affects the heart and what are the best management strategies. Laboratory testing and imaging studies, such as an echocardiogram or ultrasound of the heart, a cardiac MRI, or a specific nuclear scan in the case of ATTR amyloidosis can aid in the diagnosis of amyloid cardiomyopathy. A heart biopsy may be needed to confirm the diagnosis. The amyloid build-up in the heart results in thickening and stiffening of the heart, making it more difficult for the (Continued on page 5)

As We Look Toward The Future

By Mandy Gallaway Lacey

In May of 2012, my father received a diagnosis that no one saw coming: Heavy Chain Amyloidosis. He had been sick for years, decades even. He had atherosclerotic plaques throughout his heart and carotid arteries and suffered GI symptoms that would cause severe bleeding and nausea. I remember my father being sick for most of my life, but despite his failing systems, he fought every minute to work hard to provide for his family and to spend more time with those he loved.

Before he started chemotherapy, my dad had his kidney biopsy sample sent to Mayo Clinic. Our lives changed forever in July 2012 when we found out that my dad had an ultra-rare form of hereditary Amyloidosis called Fibrinogen A Alpha Chain Amyloidosis. Fifty-percent chance of passing the gene to his three daughters. Chemotherapy was not an option for treatment, and Dad was told he was not a candidate for an organ transplant. Instead, he would go through the years of his (Continued on page 4)

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There are many ways to support the Amyloidosis Foundation while you shop!

Every little bit adds up:

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iGive.com-
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ShopWithScrip (RaiseRight) App- order online or call our office at **248.922.9610** for the access code.

Kroger Community Rewards-
Organization Number: HS565

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
- Listing of experienced physicians that specialize in amyloidosis. Email us anytime with questions: **info@amyloidosis.org**

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

- Treatment Centers (US / International)
- Support Groups
- Newsletters
- Webinars
- Caregiver/Patient Binder
- Fundraising Toolkits

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President's Corner

Mary E. O'Donnell

We are excited to announce our new board member, Stacey Goodman, MD. Stacey Goodman, MD completed her hematology training at the NIH and Vanderbilt University before joining the faculty in 1993. She cultivated her career initially in bone marrow stem cell transplants (SCT) and was a leader within the Vanderbilt/VA SCT program for over 30 years.

In case you missed it, last month was a pivotal time for hATTR amyloidosis sufferers. Vutrisiran, to be marketed under the brand name Amvuttra, has won an FDA approval to treat hereditary transthyretin amyloidosis (hATTR) polyneuropathy. It is the first and only FDA-approved treatment demonstrating reversal in neuropathy impairment with subcutaneous administration once every three months. The drug, which belongs to an emerging class of RNA interference therapeutics works by ways of small interfering RNA that target the transthyretin mRNA to block the protein from forming into toxic clumps.

As we look to the future, we are hopeful for even more approved treatments for amyloidosis. We are thankful for those whose research has made great strides in the last few years.

We hope everyone enjoys the beautiful summer weather with your loved ones.

Mary

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, (248) 922-9610**
or **7151 N. Main Street, Ste. 2, Clarkston, MI 48346**

If you wish to receive an electronic version, please send us an email:

info@amyloidosis.org

As We Look...

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his diagnosis doing supportive therapies. Peritoneal dialysis. Albumin infusions. Arthrectomies. Stents. Cardiac catheterizations. Hemodialysis. We balanced our lives with caregiving, considering our own future health, and trying to live normal lives–something my dad wanted for my mom, my sisters, and myself. We continued this cadence until my father passed away on November 26, 2018, of a heart attack. He was a huge presence in our family, and he continues to be dearly missed by so many.

My dad always denied that his disease was hereditary. The thought of passing down a rare genetic mutation was devastating for my dad, so he somehow managed to interpret his results in a way that made them "inconclusive." We all dared not to argue with this, as we knew that believing the truth would devastate him to a point where he would not want to live. He ended up enjoying time with family and friends between dialysis and infusions. He and my mother would form close relationships with their treatment teams. They became family, and I credit them all for giving our dad six more years of life after being diagnosed with a disease that no one understood.

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I would wait until 2021 to begin the process of genetic testing for the FGA gene, the gene that causes Fibrinogen A Alpha Chain Amyloidosis (AFib). July 2021, eleven years after my father found out he had the gene, I found out I tested positive for the mutation as well. Almost simultaneously, my aunt (my dad's sister) had a transient ischemic attack and started going into kidney failure. She, too, tested positive for the FGA gene. I have since been evaluated at Boston Medical Center's Amyloidosis Center, and I am healthy for now with no sign of the disease.

As I look to the future for myself and my son, I see so many possibilities for improved treatment and cures for genetic diseases. I am encouraged by the research and drugs being produced for ATTR and gene therapy. My family and I are actively working on a fundraiser for the Amyloidosis Foundation for November 26, 2022, in memory of an incredible husband, father, brother, cousin, and friend: Glenn Dale Gallaway. We will continue to advocate, spread awareness, and support those who feel like they are not understood because of the rarity of their disease. Just because we are rare, does not mean that we are alone.
Please learn more about our family story on Instagram, Facebook, or Twitter @FibrinogenAF

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Amyloidosis and the Heart

(Continued from page 1)

heart to function effectively, resulting in heart failure.

What are the symptoms of heart failure? Patients may present with fatigue, shortness of breath, leg swelling, weight gain related to fluid retention, cough, and inability to lie flat at night because of shortness of breath. Although therapy should be tailored to the underlying cause of the amyloid cardiomyopathy, the treatment of heart failure is similar. The goal is to decrease shortness of breath and remove fluid with the use of diuretics or water pills.

Conservative measures that are often beneficial include compression stockings to help mitigate fluid accumulation in the legs, and patients can also follow a sodium and fluid restriction. Medications that are commonly used in other types of heart failure may not be well tolerated in patients with amyloid cardiomyopathy. Specific types of calcium channel blockers, diltiazem and verapamil, are avoided because of increased binding to amyloid fibrils increasing the drug effect, leading to slower heart rates and other rhythm abnormalities. Abnormal heart rhythms are common in cardiac amyloidosis, particularly atrial fibrillation, which can increase the risk of stroke. Unless there is a contraindication related to bleeding concerns, patients with atrial fibrillation and cardiac amyloidosis will be treated with blood thinners to lower the risk of stroke. Amyloid experts recommend a transesophageal echocardiogram prior to cardioversion despite anticoagulation because of higher risk of clot formation in the setting of amyloidosis.

Cardiac amyloidosis can result in progressive heart failure, and, in select patients, heart transplantation may be warranted, which requires evaluation and management by multiple different subspecialties. I see patients in the context of our multidisciplinary amyloid clinic at Mayo Clinic Florida, and patients are initially evaluated by hematologists, nephrologists, cardiologists, and neurologists, all specializing in amyloidosis. Given potential multiorgan involvement with amyloidosis, a multidisciplinary approach is always the best management strategy for patients with amyloidosis, particularly those with advanced heart failure, who will need close follow-up and monitoring.

Melissa Lyle, M.D., FACC is a board certified cardiologist and echocardiographer with subspecialty training and board certification in advanced heart failure and transplantation at Mayo Clinic.

Her clinical focus includes expertise in the diagnosis and management of cardiac amyloidosis. In addition to her clinical activities, Dr. Lyle is active in research and education, providing mentorship to both residents and fellows.

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THERE'S STILL TIME to register for the AF VIRTUAL Walk/Run/Roll/Bike!

REGISTER TODAY for the 2022 AF Virtual Run!!!

You can do your run at your leisure anytime between now and July 31, 2022.

Please tag us in your pics as we would LOVE to highlight YOU and your amyloidosis awareness!

REGISTER HERE https://secure.qgiv.com/for/amyfound/event/815306/

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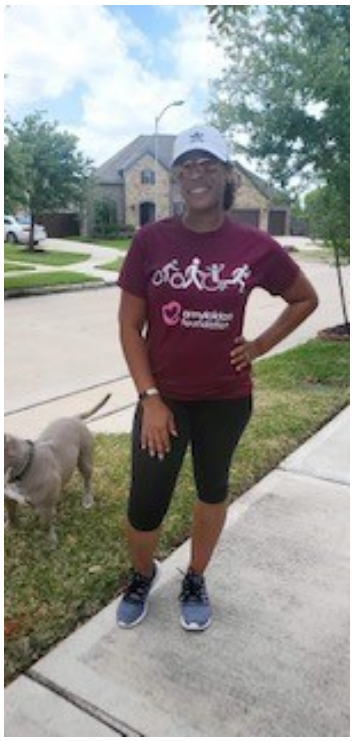

Carpal Tunnel Syndrome

Carpal tunnel syndrome can happen when amyloid deposits in the wrist area squeeze and irritate the nerve, causing tingling and numbness in the fingers and thumb. According to Mayo Clinic, you should treat carpal tunnel syndrome as early as possible after symptoms start. In the early stages, simple things that you can do for yourself may make the problem go away.

For example:

• Take more-frequent breaks to rest the hands.

• Avoid activities that make symptoms worse.

• Apply cold packs to reduce swelling.

Other treatment options include wrist splinting, medications and surgery. Splinting and other conservative treatments are more likely to help if you've had only mild to moderate symptoms that come and go for less than 10 months. If you have numbness in your hands, you need to see a health care provider. **AF**

Welcome to Stacey A. Goodman, MD

The Amyloidosis Foundation is pleased to welcome Dr. Stacey Goodman to our Board of Directors. Her SCT background and expertise in plasma cell disorders helped her to initiate and lead the Vanderbilt Amyloidosis Multidisciplinary Program (VAMP) from its inception in 2011 thru 2021 when she retired. We are fortunate to have her advance our mission for years to come!

Progress in Amyloidosis: 2022

View our latest webinar by Jeffrey Zonder, MD, who is a Professor in the Departments of Oncology at the Barbara Ann Karmanos Cancer Institute (KCI) and Wayne State University School of Medicine. To view, go to:

https://amyloidosis.org/resources/#webinars

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