



News and Stories - Fall 2019

Spreading Awareness

One of the tasks that the foundation has taken on is spreading awareness of the amyloidosis diseases among the medical community. We accomplish this in a variety of ways. With the help of dedicated board members and volunteers, we exhibit at a number of medical conferences throughout the year.

They include The American College of Cardiologists, American Association of Nurse Practitioners, Heart Failure Society of America, Kidney Week and the American Society of Hematology. By distributing informational materials and having one



on one discussions with the doctors and nurses that attend these meetings, we work to raise awareness which hopefully will lead to earlier diagnosis for patients. By no means are we trying to make amyloidosis

specialists out of the doctors that attend these meetings, but if they are familiar with the signs and symptoms of the disease, they have a better chance of thinking of

the disease as a potential diagnosis.

The foundation started this effort in 2005, and has attended/exhibited at nearly 50 medical meetings

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Did you miss our webinar events?

The Amyloidosis Foundation sponsored 2 webinars with live Q & A in June! The first one was titled 'PROGRESS IN AMYLOIDOSIS RESEARCH: WHERE ARE WE NOW?'

This was presented by Jeffrey Zonder, MD – Professor from the Department of Oncology at the Barbara Ann Karmanos Cancer Institute and Wayne State University School of

Medicine in Detroit, MI.

The second webinar was titled 'LIGHT CHAIN (AL) AMYLOIDOSIS: WHAT IS AL ANYWAY?'

This webinar was presented by Heather Landau, MD – Associate Attending Physician, Adult Bone Marrow Transplant Service

Memorial Sloan Kettering, New York, NY.

The webinars were recorded and are available on our website:

<http://amyloidosis.org/resources/#webinars>



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Global Genes: Rare Patient Advocacy Summit

One of our staff members attended the 8th Annual Global Genes Summit, the world's largest gathering of rare disease patients and advocates.

The new norm in the rare world is about patient engagement, which is critical

in driving progress in our community. The role of patients and caregivers in the future of amyloidosis treatments, and hopefully cures, continue to expand.

Patients are playing a key role in driving drug development. At this conference,



we have gained insight, made connections, and were able to bring home actionable strategies and tools to help accelerate change.

Here is how the days mapped out.

The first session was on Innovations In Rare Disease. It focused on improving clinical care, treatments and therapies by making patients a partner in the process. Data collection and sharing of that data was a key topic. With surveys, focus groups, and a quality of life (burden) study, we would be able to track severity of symptoms. This may lead to better treatments and targeted timing of treatments.

Session 2 was on Genome Sequencing. Nearly 80% of rare diseases have a genetic cause. For many of these patients, whole genome sequencing can provide a diagnosis, which

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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 with questions: info@amyloidosis.org
- Treatment Centers (US / International)
- Support Groups
- Newsletters
- Webinars
- Caregiver/Patient Binder
- Fundraising Toolkits

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President's Corner

Mary E. O'Donnell



What is Quality Adjusted Life Year (QALY)?

Because Rare Disease patients have the highest spending, policymakers and industry stakeholders are considering policies that will have a significant impact on what treatments patients and persons with disabilities can access. Approval does not guarantee access.

The use of flawed, discriminatory value assessments could threaten access to care for patients with chronic illnesses and people with disabilities. These assessments place a value on the life of a human based on their health status and assume every patient will respond the same way to treatments.

Methods for assessing value should meaningfully acknowledge diversity amongst all patients, avoiding the use of obscure methods that embody a "one-size-fits-all" mentality for determining value.

Mary

#GivingTuesday 2019



Celebrated on the Tuesday following Thanksgiving (in the U.S.) and the widely recognized shopping events Black Friday and Cyber Monday, #GivingTuesday kicks off the charitable season, when many focus on their holiday and end-of-year giving. **AF**

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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 anytime at: **info@amyloidosis.org**, **1-877-AMYLOID** (877-269-5643) 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



Living Rare, Living Stronger Patient and Family Forum

On June 21-23, 2019, one of our staff members attended a special event that gathers the rare community and presents tracks filled with tools, information and support needed to share with our amyloidosis community.

While there, Kathi attended many different classes on drug development, clinical trials and investigational therapies, the role of therapy and psychiatry in rare disease, and stress management for caregivers.

There were also networking sessions and a luncheon where there were medical

professionals and students who were eager to learn about amyloidosis. Things discussed included diagnosis and misdiagnosis, a doctor's need to have a good supportive relationship, and tips for future medical professionals to think outside the box!

During the session on Clinical Trials and Investigational Therapies, it was said that it takes approximately 9 years to bring a drug to market. Most patients were shocked to hear this news. In the rare community, the cost per approved

orphan drug that is a NME (new molecular entity) is \$242 million.



The next session was named 'Depression: Evaluation, Diagnosis and the Role of Therapy and Psychiatry'.

Living with a rare disease

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Shop Today! Together We Can Make A Difference!

Show your awareness and wear your Amyloidosis Foundation gear proudly! Be an ambassador for us and spread the word about amyloidosis.



You can place your order online at:
<http://www.amyloidosis.org/products>

You can also order by mail (form is on our website), or call **1-877-AMYLOID** (1-877-269-5643).

T-shirts— \$25

Wristband— \$5

Lapel Pin— \$8

Notecards (set of 12)— \$15



These items make great gifts and people purchase them to create awareness for amyloidosis at their fundraising events.

Show your spirit and shop today!

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Living Rare, Living Stronger (Continued from page 4)

can have a detrimental effect on mental well-being. While this could be said of any disease or chronic condition, the mental impact can be amplified in the case of a rare disease like amyloidosis. Anxiety, stress, low mood, emotional exhaustion, and suicidal thoughts, have all been identified in the rare disease population.

There are several factors that lead to increased stress in patients with rare diseases, one of the most significant being delays in receiving a correct diagnosis. According to Barbara Schildkrout, MD, assistant professor of psychiatry, at Harvard Medical School in Boston, Massachusetts, "A diagnosis is very powerful because it essentially sums up a huge amount of information. A diagnosis conveys an etiology, clinical course, and treatment. Often, it can predict that you will get better, and it provides information on how long it will take to get better. This cannot necessarily be said about a rare disease diagnosis because very little might be known about a particular disease."

If you or a loved one is exhibiting signs of anxiety or depression, contact SAMHSA at 1-800-662-HELP.

The final session was geared toward the caregivers. This is an area that the Amyloidosis Foundation has been focused on in recent months.

The name of the session was 'Stress Management: Self-Care and Emotional Health for the Caregiver'.

It was mentioned several times that caregivers need help from others. It is not something to take on all on your own. A great suggestion was for caregivers to make a "wish list" of ways that people can help when they offer. Often times, we are so wrapped up with getting things done, that we can't think of ways for others to help (and you can only have so many casseroles in the freezer!). Making a list can help you delegate some of the duties so that you can limit your stress level.

Self-care was talked about. Taking care of yourself refuels you instead of taking from you. It is necessary for you to take care of yourself so that you can take care of someone else. Seek out a support group so that

you don't feel isolated.

Often times, caregivers stress about the "What-ifs?". "What if something happens to me before the one I care for?", "What if we aren't making memories because we're too busy trying to care for/cure our loved one with amyloidosis?", "What if I can't care for my loved one?", "What if I do something wrong?".

All of these are valid questions, and proper



planning can make all the difference. Focus on READY.SET.GO! Get ready by doing your research and develop a medical support team. Get set by keeping medical history updated and preparing for hospital trips. AND GO! Get away from it in your head. Train yourself to be in the moment because you're prepared. For more support information, go to www.amyloidosis.org. **AF**



Spreading Awareness

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since.

We distribute hundreds of information packages at each meeting and have one on one discussions with dozens of doctors during each exhibit.

What we have noticed over the years is that although



many of the doctors still don't know a lot about amyloidosis, in the recent years at least many of them have at least heard of the disease.

Many years ago at one of the cardiologist meetings, an elderly doctor came to our booth and made the statement "Amyloidosis, it is just so sad that there is nothing we can do for these patients". So very frustrating to hear this because at that time there

were some treatments in clinical trials that were showing promise for these patients.

And now, we have three FDA approved drugs for ATTR amyloidosis, and many promising drugs in trials for AL amyloidosis. We will continue our efforts in raising awareness of amyloidosis and the progress that is made in the development of treatments.

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Global Genes (Continued from page 2)

can prevent unnecessary testing, lead to stronger medical management, and provide a link to the amyloidosis community for support.

What is druggable? New therapeutic target in rare disease was the title of the third session. Only a fraction of rare diseases have a FDA approved therapy. Advances in DNA sequencing have led to the cause of many rare diseases, which can lead to better therapies. This session talked about one of those clinical trials that had great success.

Contact us if you'd like more information at:
info@amyloidosis.org

Day 2 started with a session on Parents and Caregivers: Shaping Clinical Drug Development. There is a wide array of complex and closely monitored processes along the way to developing a protocol.

After all, starting a clinical trial is no simple task. From developing an agenda, to patient recruitment, to FDA submission, there are many steps, even within these steps. Patient and community involvement help ensure study questions are relevant and that the trial is achievable.

We were able to get the regulatory and industry perspective on what it takes to design a trial.

Session 2 was on Leveraging Digital Tools for Reach and Impact. This session focused on technology as it has empowered individuals to reach larger audiences, raise awareness, and ultimately drive change.

The last session was titled The Value of Rare Disease Therapies: Patient Perspectives Needed. There is a lot of public debate surrounding the 'Value' of rare disease therapies. We need perspectives from patients, families, and advocates to further these discussions and assessments.

This event focused on the future, new pathways to diagnosis, treatment and delivering care. **AF**



What is ATTRv?

Hereditary ATTR amyloidosis has been in the news a lot because of it's FDA drug approvals within the last year, but do you really know about ATTRv?

Even though you are born with a gene mutation, normally the harmful deposits don't occur until adulthood. Although all the variants of the hereditary amyloidoses can cause serious complications, there are some carriers of this genetic mutation that may not show symptoms of the disease at all. Others may have a few, more minor, health issues. A patient is tested to determine if they have amyloid proteins in their body. If amyloidosis is confirmed but the type is not clearly found in these tests, it will be important to do more tests to

find the exact type and also to determine the variation of ATTR.

The main diagnostic testing for any amyloidosis disease includes blood tests, urine tests and biopsies. Some tests are only done once to confirm a diagnosis, while others may be repeated to monitor the disease and response to therapy. For a diagnosis, blood and urine tests will be done to help your doctor determine the diagnosis of amyloidosis.

These tests can also help to show which organs are involved and how much damage they may have. A tissue biopsy will be performed. With ATTR, after amyloidosis is confirmed and it is determined that there is transthyretin

Today's treatment plans are two-fold:

Supportive treatment – treating your symptoms and organ damage. Supportive treatment varies depending on symptoms, including peripheral neuropathy, autonomic neuropathy, and cardiac and kidney problems. Some medications can help with pain relief and nerve damage.

Source treatment – slowing down, or stopping, the overproduction of amyloid at the source of the disease. It is possible that ATTR can cause serious health complications, however, do not assume that disability or severe health issues are stamped on your future. There are treatments available and research continues.

What is ATTR?

Hereditary amyloidosis is one type of the systemic amyloidosis diseases that are caused by inheriting a gene mutation. That genetic mutation then produces an amyloid protein that forms into an abnormal shape. These abnormal "misfolded" amyloid proteins can be deposited and cluster in the body's nerves and other organs and once they build up, this may affect and harm tissue and/or organ function.

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What organs can it affect?

- Protein in the urine
- Renal Failure
- Dizziness
- Urinary tract infections
- Sexual dysfunction
- Sweating abnormalities
- Carpal Tunnel Syndrome
- Glaucoma
- Dark floaters
- Abnormal blood vessels in the eye
- Eyelid swelling/ inflammation
- Irregular heartbeat
- Heart failure
- Nausea/ Vomiting
- Loss of appetite
- Diarrhea/ Constipation
- Wasting of muscles/ tissues
- Nerve death beginning in the feet and moving up the body

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amyloid protein (via biopsy and Congo red staining in the lab), the protein needs to be identified by protein sequence analysis and DNA sequencing must be performed.

Within the last year, 3 drug therapies have been approved by the FDA. Patisiran (Onpattro) and Inotersen (Tegsedi) treat polyneuropathy through suppression of TTR synthesis (anti-sense), and Tafamidis (Vyndaqel) treats ATTR related cardiomyopathy by stabilizing the TTR (stabilizer). To learn more about these treatments, please visit:

- www.onpattro.com,
- www.tegsedi.com
- www.vyndalink.com

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