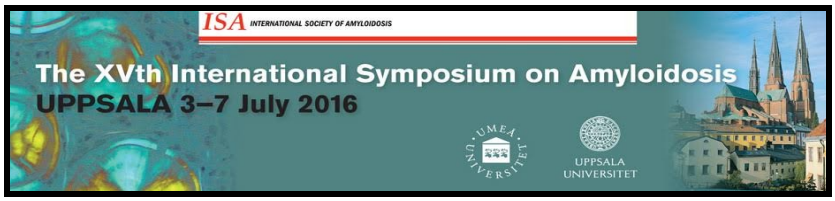
amyloidosis foundation

News and Stories - Fall 2016

Report from International Symposium on Amyloidosis

by Raymond Comenzo, MD, Director, John C. Davis Myeloma and Amyloid Program; Professor, Tufts University School of Medicine



The 2016 International Symposium on Amyloidosis was held in Uppsala, Sweden in early July. Over the years the meeting has grown in size and it was impressive to see over 400 scientists and experts come together to spend three days sharing knowledge and learning more about amyloidosis.

The Amyloidosis Foundation supported many young investigators from around the world, allowing them to go to the meeting and present their work. The Foundation's sister organization, the Amyloidosis Research Consortium, had a booth and presentations and several meetings that brought together all stakeholders to work on collaborative initiatives moving forward.

Among the meetings held by the ARC were a meeting of industry partners, patient representatives and experts to discuss collaborative research programs that will advance our knowledge and support the development of new treatments and also the kick-off meeting of the ARC's Collaborative Network.

The network stretches across the globe and is comprised of twenty-six amyloidosis centers,

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Wild-type Transthyretin Amyloidosis: From Suspicion to Treatment

by Cristina C. Quarta, MD, PhD, National Amyloidosis Centre, London, UK and Anna Laura Tinuper, MD, University of Bologna, S. Orsola-Malpighi Hospital, Bologna, Italy

Wild-type transthyretin (TTR) amyloidosis (ATTRwt) was previously known as "senile systemic amyloidosis" since it was mainly diagnosed in elderly people, in most cases above the age of 70. However, it is now clear that the disease can also manifest below the age of 65. The incidence and prevalence of the condition are unknown, but ATTRwt is definitely underdiagnosed.

Indeed, newer imaging techniques have shown that disease caused by ATTRwt deposits may be far more common than anyone thought.

TTR is a blood protein produced by the liver, which is present in everybody.

Normally, TTR circulates within the bloodstream and functions as a transporter of the thyroid hormone (thyroxin) and vitamin A (retinol), hence the name: 'transthy-retin'.

While genetic alterations (mutations) in the *TTR* gene lead to familial (i.e. inherited) TTR amyloidosis, in which patients have structurally abnormal, amyloid-forming 'variant' TTR in their blood, ATTRwt is NOT hereditary.

For reasons that remain unclear, normal (i.e. 'wild-type') TTR proteins clump together and form amyloid deposits. Although advancing age is undoubtedly a risk factor, the mechanisms that lead to amyloid formation in some people and not in others starting from 'wild-type' (i.e. normal) TTR, are still unknown.

Also, for unknown reasons, ATTRwt is far more common in men than in women.

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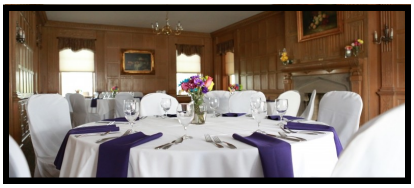
Pittsburgh Benefit for Amyloidosis Research

Join us on **Friday, October 28, 2016** for a special night as we raise funds for research and celebrate our amyloidosis community.

Tickets are $175 and we have tables of eight for $1400. Event sponsorships are also available. Please find a link to purchase tickets, become a sponsor or donate on our website at **www.amyloidosis.org**.

We have a block of rooms reserved for this event at the **Sheraton Pittsburgh Airport Hotel**. The room rate is $118 per night. They will provide complimentary transportation to and from the event as well.



Rooms at this rate will be available until September 30.

We look forward to seeing everyone in Pittsburgh this October!

For more information, please contact us at: **info@amyloidosis.org**.

MONTOUR HEIGHTS COUNTRY CLUB

amyloidosis foundation

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**

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

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

www.amyloidosis.org

Follow Us

Stay connected for all the latest information on Amyloidosis:

Web: www.amyloidosis.org
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President's Corner

The Amyloidosis Foundation has much to be thankful for this year. Many of you have organized, participated in or donated to one of the many fundraisers held this year. We could not function without the generous support you all have shown us. We are forever grateful.

Make sure to register for our patient webinar on **October 17**, **"The Changing Paradigm of Amyloidosis Treatment"**, with Jeffrey Zonder, MD. Details on page five and on our website.

We look forward to meeting some new faces (as well as seeing many familiar faces) at the **Pittsburgh Research Benefit on October 28**. Please visit our website for details, purchase tickets or donate, **www.amyloidosis.org**.

Enjoy this beautiful season of change. Continue to show gratitude for your friends and family as we do for all of you.

- Mary O'Donnell

Black Friday. Cyber Monday.

#GIVINGTUESDAY™

November 29, 2016

#GivingTuesday is a global day of giving, fueled by the power of social media and collaboration.

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.

We ask you to donate to the AF on **#GivingTuesday,** follow us on social media and share our posts with your friends and family to create amyloidosis awareness in your community. *Thank you for your support!*

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

Symposium on Amyloidosis

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each one having met rigorous criteria. The Collaborative Network is focused on expediting and optimizing clinical research through efficient clinical trial design and addressing the barriers that slow down the delivery of promising therapies.

The Amyloidosis Foundation with the ARC was the only nonprofit organization represented by a booth. AF and ARC board member Dena Heath staffed the booth for the duration of the conference. She spoke with doctors and scientists from around the world, sharing our mission of accelerating the development of advanced diagnostic tools and effective treatments for systemic amyloidosis through collaboration and innovation. Ms. Heath also presented the new Amyloidosis Clinical Resources App for smart phones and tablets and the two new laminated pocket cards for health care professionals – the Cardiac Alert pocket card and the Clinical Trials pocket card.

The meeting included presentations by AF and ARC board members, physicians Giampaolo Merlini and myself. Dr. Merlini discussed the many advances in diagnosis and understanding of the amyloid disease process and I sketched a futuristic vision of therapies for AL amyloidosis. Other highlights of the meeting were Dr. Morie Gertz's presentation of results with the anti-amyloid monoclonal antibody, NEOD001, and Dr. Mark Pepys presentation of the anti-amyloid anti-SAP antibody therapy. Both of these approaches are extremely promising for patients and are in clinical trials at this time.

The AF and the ARC look forward to participating in the 2018 ISA meeting in Kumamoto, Japan. ***AF***

Kelli's 50th #4Tres—A Spinning Success!

Kelli Heald celebrated her 50th birthday organizing a spin class on August 18 & raised over $11,000 for the AF, and most importantly – honored the memory of her husband, Tres. Friends and family joined in on the fun as well (*see below*).

Thank you to everyone who donated and joined her at Grit Cycle in Costa Mesa, CA. We also appreciate the support of these local businesses: Cafe Gratitude, Nekter, Happy Dough and Ink & Think. ***AF***

Amyloidosis Foundation - October Webinar

On **Monday, October 17, 2016**, Dr. Jeffrey Zonder will host the AF webinar titled "The Changing Paradigm of Amyloidosis Treatment" at 2:30pm (EST).

This webinar will focus on treatment options for amyloidosis patients.

Please visit our website to register: **amyloidosis.org**.

Dr. Zonder is a Professor in the Departments of Oncology at the Barbara Ann Karmanos Cancer Institute and Wayne State University School of Medicine.

He is the leader of the KCI Myeloma Multidisciplinary sub-team, Medical Director for Karmanos Cancer Center Therapeutic Apheresis Unit, and a member of the International Myeloma Working Group, the International Myeloma Society and the International Amyloidosis Society.

Register today and share the details of our webinar with your friends and family to spread amyloidosis awareness.

Thank you to Alnylam Pharmaceuticals for sponsoring this educational webinar. ***AF***

Wild-type Transthyretin Amyloidosis: From Suspicion to Treatment

(cont. from page 1)

ATTRwt mainly manifests with cardiac symptoms, although it often also affects the part of the wrists called carpal tunnel, causing carpal tunnel syndrome - tingling and pain in the wrists, pins and needles in the hands. Around 50% of patients affected by ATTRwt have carpal tunnel syndrome, which, interestingly, can precede the cardiac symptoms by several years.

Amyloid deposits in the heart lead to a progressive thickening of the cardiac walls and stiffening of the heart, which becomes progressively unable to pump the blood around the body efficiently. Therefore, patients affected by ATTRwt become more subject to build up fluid and develop heart failure symptoms, including breathlessness, swelling of the legs, palpitations (mainly due to atrial fibrillation and atrial flutter), bloating, dizziness and collapse (syncope), and fatigue. Amyloid can also compress the electrical system of the heart. As a consequence about 40% of patients end up needing a pacemaker at some point during the course of the disease.

The combination of electrocardiogram, echocardiogram and cardiac magnetic resonance (MRI) can prompt a strong suspicion of amyloid heart disease. However, since different proteins can lead to cardiac amyloid deposits, a definite diagnosis of ATTRwt would require the detection of TTR amyloid deposits on a heart biopsy in conjunction with genetic testing showing that there is no mutation in the *TTR* gene. However, cardiac biopsy is an invasive procedure and can be associated with risks. Furthermore, biopsies from other parts of the body, such as abdominal fat or the rectum, are negative in many patients with ATTRwt since the disease mainly affects the heart.

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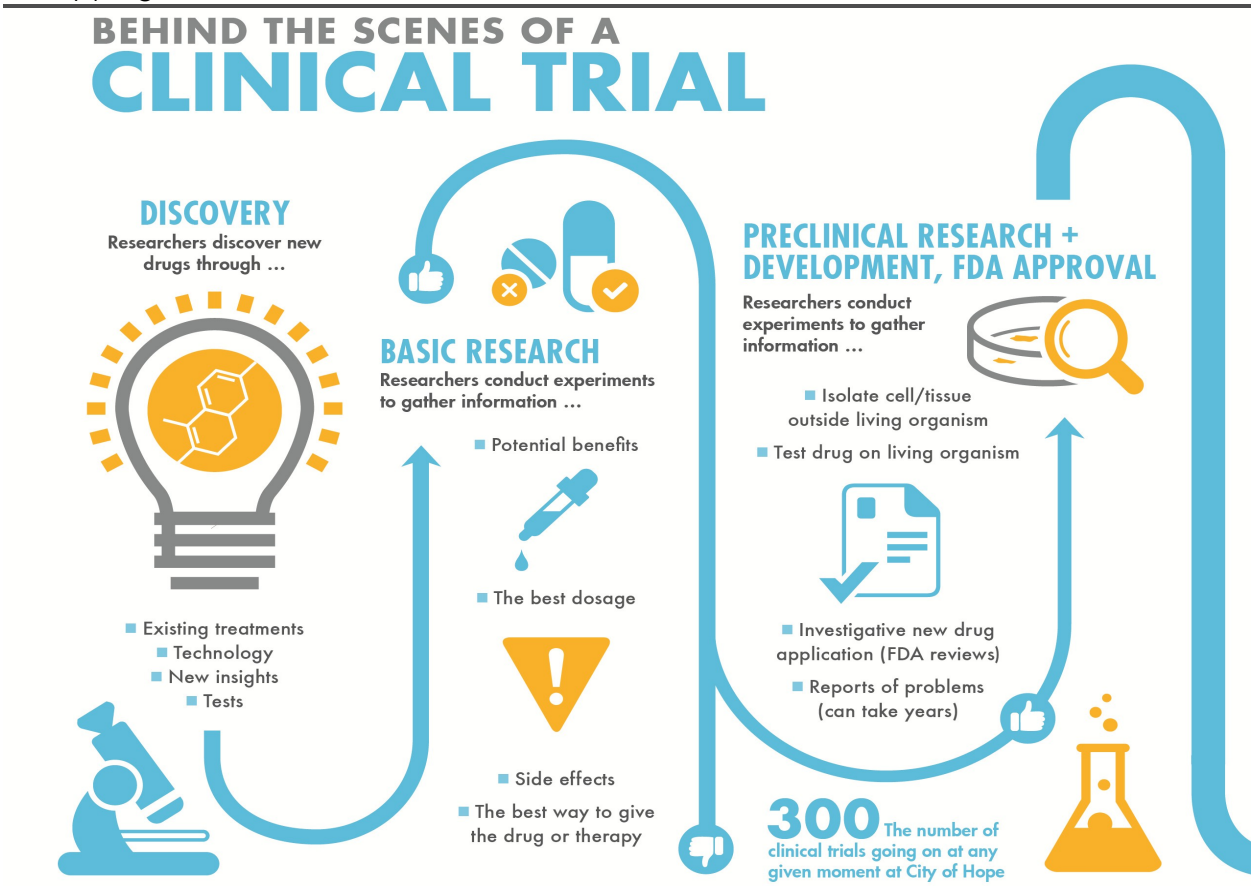
In recent years we have witnessed a paradigm shift in the diagnosis of this rare condition. Indeed, it is now possible to avoid a tissue (especially cardiac) biopsy in many cases with suspect ATTRwt thanks to a non-invasive and widely available diagnostic tool, called 'bone scintigraphy' (DPD in Europe, PYP in U.S.). This test in fact can identify the presence of TTR amyloid depositsin the heart with an extremely high level of accuracy and is playing an increasingly important role in the diagnosis of ATTRwt. It is not yet possible to know when the disease starts during life, although it is believed to be a slowly progressive condition and patients are estimated to survive for about 7-10 years after being diagnosed with ATTRwt.

Treatment of ATTRwt is symptomatic and mainly relies on diuretics ('water tablets), drugs that help patients to get rid of extra fluids, but restriction of fluid (maximum 1.5 liters per day) is extremely important for the patient to benefit from the positive effects of diuretic drugs and possibly delay the need of higher dosages. A number of new drugs for either familial or wild-type TTR amyloidosis are in various stages of development. Some of them, such as Tafamidis and Diflunisal, bind to TTR in the blood making the TTR protein less amyloidogenic. They are available in a number of countries worldwide. However, they have been mainly studied in patients with familial TTR amyloidosis and their role in ATTRwt is still under investigation.

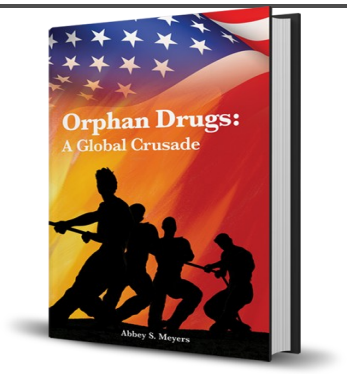
Other therapies, not yet available but currently in clinical trials, such as small interfering RNAs and antisense oligonucleotides, are able to 'switch off' the gene for TTR in liver cells, so that TTR is simply not produced.

This offers hope for the future and makes awareness and diagnosis of ATTRwt even more important. ***AF***



Orphan Drugs: A Global Crusade,

A New Book by Abbey Meyers



Abbey Meyers, PhD is the founder and past President of the National Organization for Rare Disorders (NORD), a coalition of national voluntary health agencies and a clearinghouse for information about little known illnesses.

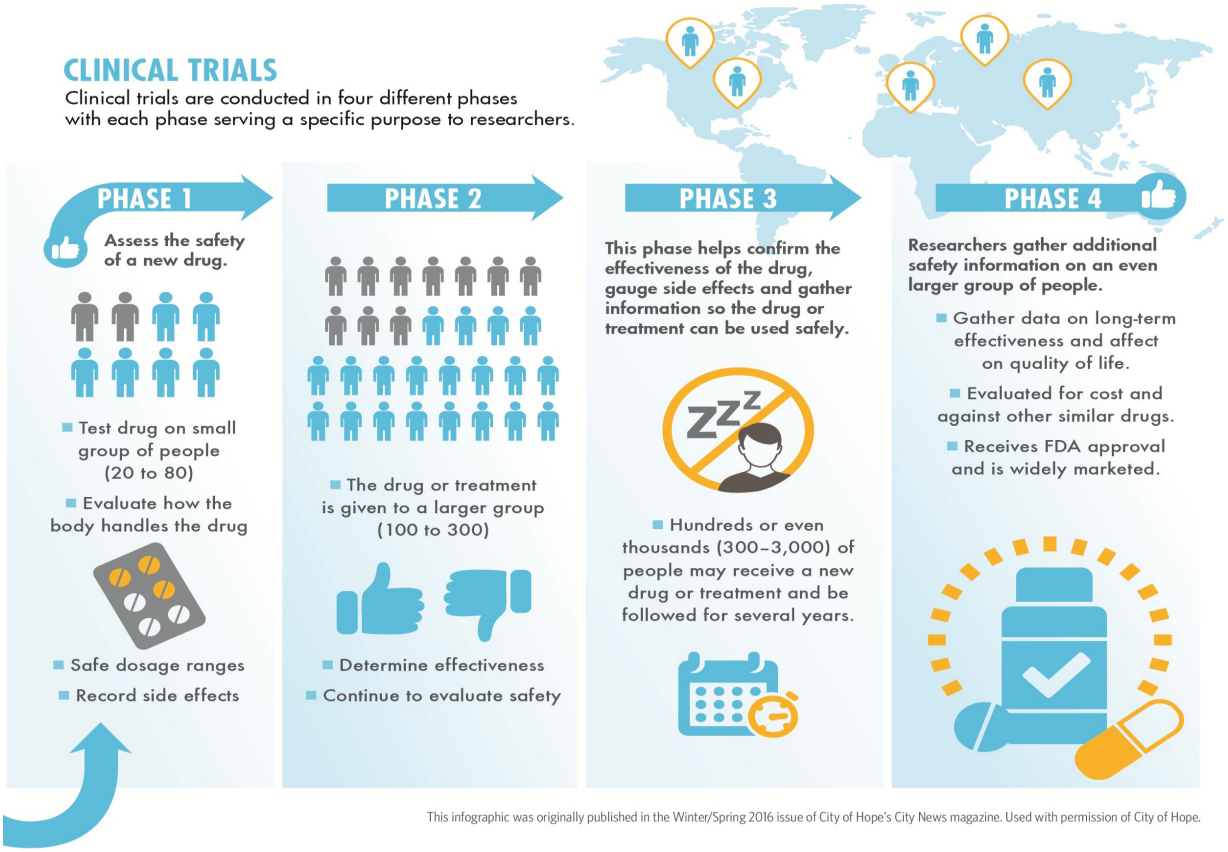
She became involved with the rare disease community when her oldest son, at two years old, had unexplained symptoms that no one could diagnose.

She fought for a diagnosis for him (now known as Tourette's syndrome) and pushed for the Orphan Drug Act to be signed by President Reagan in 1983.

If you are passionate about rare diseases and learning the history of this movement, please read this book.

Her memoir is now available online for free: **www.abbeysmeyers.com/**. ***AF***

Amyloidosis Foundation disclaimer*: We have no financial gain from sharing the details of her book other than to provide a resource you might find interesting.*



This infographic was originally published in the Winter/Spring 2016 issue of City of Hope's City News magazine. Used with permission of City of Hope.

The Prieber Open Was a Hole in One!

On August 13, 2016 more than 300 golfers took to the links in The Prieber Open, to honor the memory of Christopher Priebe and raise awareness for amyloidosis.

He was diagnosed in October 2015 and passed in February 2016. Chris loved cigars, golf and Grand Marnier. His wife Johna wanted to share his spirit for life and raise funds for others struggling with this rare disease.

The event raised thousands of dollars for the Amyloidosis Foundation Research Grant Program and we are so grateful for their support.

The two golf courses used were in Livonia, MI – Fox Creek and Whispering Willows. Players enjoyed lunch donated by Panera and a delicious steak dinner to end the day.

The weather was unpredictable but the sun finally came out with a beautiful rainbow *(see below)*.

Thank you to all the volunteers and generous family and friends for making this day so special. ***AF***

The Amyloidosis Foundation appreciates your continued support.

If you would like to become more involved in the foundation, interested in starting a fundraiser or becoming an amyloidosis ambassador—we would enjoy speaking with you and helping in anyway we can.

Please call our office today **1-877-AMYLOID** (877-269-5643) or send us an email at **info@amyloidosis.org**.

Thank you!