

**FROM THE DIRECTOR**

Dear Friends,

Welcome to our Winter 2011 Amyloid Treatment and Research Program Newsletter! In this issue, we will update you on our clinical and laboratory research activities. These programs are critical in the battle against amyloidosis. Through research, we will develop a better understanding of the causes of amyloidosis, and this will lead to improvements in diagnosis and in treatment.

In this Newsletter, we will introduce you to some of the dedicated researchers in the Alan and Sandra Gerry Amyloid Research Laboratory, directed by Dr. Lawreen Connors. We are pleased to tell you that the laboratory, which for many years has served as a referral laboratory for patients and doctors from all over the country and all over the world, has recently been designated as a CLIA-approved laboratory, under the Boston Medical Center Department of Pathology and Laboratory Medicine. This gives our test results an official status as part of a medical record, like any other medical tests. Some of the available tests include Congo red staining; staining for TTR, AA, and light chain amyloid types; isoelectric focusing screening for TTR mutations; and genetic sequence analysis for mutations causing TTR and other inherited amyloidoses.

In addition to carrying out diagnostic testing, the Gerry Laboratory coordinates collaborative laboratory research programs involving investigators from many departments. These include:

- Dr. Connors' own studies of chaperones as accessory proteins and biomarkers in amyloidosis, and on the age-related form of TTR amyloidosis
- Dr. Flora Sam's research on mechanisms of remodeling in the heart of patients with amyloidosis
- Dr. Vickery Trinkaus-Randall's work on the role of sugar-based molecules called "glycosaminoglycans" in formation of amyloidosis
- Dr. Catherine Costello's work on proteomics and mass spectrometric analysis of amyloid proteins
- Dr. David Sherr's work on immunotherapy approaches for treatment of amyloidosis
- Drs. Frederick Ruberg, Stephan Anderson, and James Hamilton, investigating MRI imaging for amyloidosis
- Our work on development of animal models and siRNA therapies for AL amyloidosis

This work is made possible through the dedication of many colleagues and trainees, and has been facilitated by new equipment purchased through a U.S. Health Research Services Administration grant, obtained with the help of our advocates in Washington, Terry and Ann Peel; and also by grants from the Wildflower and Gruss Foundations.

A research highlight this past year was the XII<sup>th</sup> International Symposium on Amyloidosis that was held in Rome in April. Our investigators and trainees made a total of 22 oral and poster presentations there, which was made possible by travel awards from foundations and donors. This was a wonderful opportunity to get together with amyloidosis researchers from all over the world. We are grateful to Dr. Giampaolo Merlini and colleagues for organizing it.

In Rome, Dr. Martha Skinner was installed as the third President of the International Society for Amyloidosis. This is a great tribute to her remarkable contributions to the field in many areas of research and patient care, and she will provide wonderful leadership to the ISA. Also at the International Symposium, our former director Dr. Alan Cohen stepped down after many years of service as the Editor-in-Chief of the Amyloid Journal. We are grateful to Dr. Cohen for his leadership at the Journal over the years, and look forward to the future growth and impact of the Journal.

One of the most rewarding missions of our Laboratory is helping to train researchers and clinicians from other institutions. Dr. Maria Cibeira, a visiting hematologist from Dr. Bladé's program in Barcelona, Spain, was honored in Rome with a young investigator award for her outstanding work on long-term outcomes in our patients. For the next 6 months, we will be hosting a visiting nephrologist from Italy, Dr. Laura Econimo. We also would like to introduce you to our new Amyloid Internist, Dr. Sujata Ramamurthy. Dr. Ramamurthy recently completed a residency in Internal Medicine at Boston Medical Center, and next year will be a cardiology fellow at Washington University in St. Louis.

We hope you will enjoy learning more about our investigators and our laboratory research programs. Your support allows us to continue our work and our commitment to improving the diagnosis and treatment of amyloidosis.

With best wishes,



David C. Seldin

**Inside this issue:**

FROM THE DIRECTOR	1
MEET THE GERRY LABORATORY	2-3
PEOPLE, PHOTOS	4



**David Seldin, M.D., Ph.D.**



**Lawreen Connors, Ph.D.**



**Laura Econimo, M.D.**



**Sujata Ramamurthy, M.D.**

## Lawreen Connors, Ph.D., Director



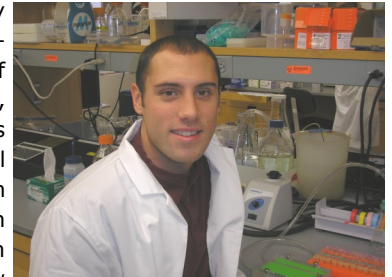
I have had the privilege and very good fortune to be a member of the Amyloid Program at Boston University/Boston Medical Center for most of my professional career. As a graduate student mentored by Dr. Martha Skinner, I studied transthyretin amyloid for my thesis research project and was awarded a Ph.D. in Biochemistry. My research currently focuses on the cardiac amyloid diseases, including the senile systemic form, and involves exciting collaborative studies with several clinician scientists in the Cardiology section at BMC. In addition to research, I oversee all activities performed in the Alan and Sandra Gerry Amyloid Research Laboratory. The Gerry Laboratory serves as an international reference center for diagnostic testing of amyloidosis and determination of amyloid disease type; an area for biochemical, biophysical, and cell-based research of amyloid proteins; and a repository for thousands of clinical specimens and de-identified patient data. This year, we took a big step forward and became a U.S. government approved, CLIA certified laboratory. With respect to research, our laboratory team is actively involved in several separate, but integrated, amyloid studies that are supported by federal, foundation, and institutional funding sources. Research projects include collaborations with colleagues in other laboratories at Boston University, as well as at other institutions. In this newsletter, I am delighted to introduce you to the dynamic group of people who are working so hard in the Gerry Laboratory to meet our goals of improving diagnosis, determining the cause(s) of amyloid, and developing improved treatments.

"... dynamic group of people who are working so hard in the Gerry Laboratory ..."

Lawreen Connors

## Michael Greene, Ph.D. Graduate Student

I am a fourth year graduate student in the Department of Pathology and Laboratory Medicine researching TTR amyloidosis with an emphasis on cardiac related forms of the disease. A particular interest of mine is understanding the role of a molecular chaperone protein, Clusterin, in amyloidosis and how it interacts with amyloid in tissues and in circulation. Using biochemical, immunological, and histological techniques I am investigating the molecular interactions of Clusterin with amyloid precursor proteins in serum and mature amyloid fibrils in cardiac tissues. These interactions can affect amyloid fibril formation and stability thereby leading us to a better understanding of how these diseases progress. In the Gerry Lab, I have the opportunity to do research in a multidisciplinary environment where I can learn scientific techniques from diverse fields and collaborate with researchers skilled on many levels. This has provided me with a positive graduate school environment where resources and knowledge are freely shared between investigators.

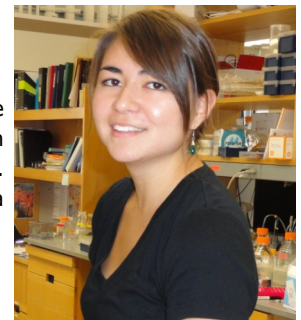


"In the Gerry Lab, I have the opportunity to do research in a multidisciplinary environment..."

Michael Greene

## Clarissa Koch, Ph.D. Graduate Student

I'm a second year PhD student in Pathology working on characterizing the effects of TTR and Clusterin on cardiac cells. In 2009 I received my BSc in Biomedical Sciences from the University of Amsterdam in the Netherlands. What I like most about working in the Gerry Lab is the fact that it is part of a great program where treatment and research go hand in hand.

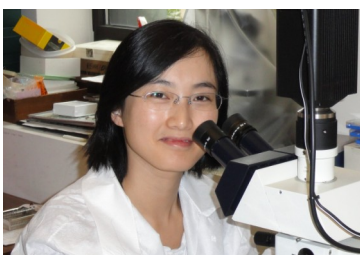


"What I like most about working in the Gerry Lab is the fact that it is part of a great program where treatment and research go hand in hand."

Clarissa Koch

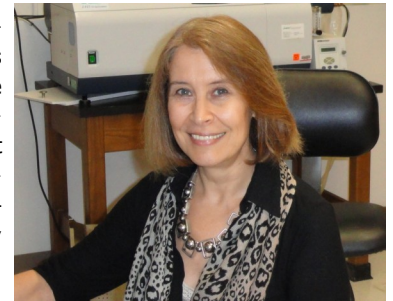
## Yanyan Lu, Ph.D. Graduate Student

I am a second year graduate student in Biochemistry. I am working on a project, which applies mass spectrometry to study the modifications and depositions of amyloid proteins. In this way, we hope to find more information about the mechanism of the formation of amyloid diseases. As a student of chemistry in the past, working on a biological and patient-oriented project makes me feel more motivated. Here in Gerry Lab, I can do research with practical samples and what I can do may help in some way; this idea makes me really excited.



**Tatiana Prokaeva, M.D., Ph.D., Research Scientist**

I received my medical training and doctorate degrees in Rheumatology from Moscow Rheumatology Institute in Russia, where I was involved in a research projects on secondary amyloidosis in patients with rheumatoid arthritis. In 1999, I was invited by Dr. Martha Skinner to join the amyloid research team. My current research is focused on genetic aspects of primary and familial types of amyloidosis. In particular, I am interested in studying the role of amyloidogenic light chain gene sequences on the variability of clinical presentation and survival of patients with primary amyloidosis. I am delighted to be a part of the Amyloid Research team, a group of enthusiastic and knowledgeable scientists working toward improving diagnosis, treatment, and quality of care in patients with amyloidosis.



**Elena Klimtchuk, Ph.D., Research Scientist**

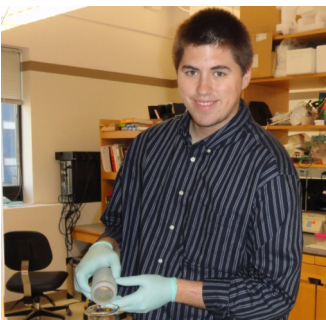


I have been a research scientist in the Gerry laboratory since 2005. I have a degree in Physical Chemistry and am trained in biophysics and protein structure. Since all amyloid proteins are tightly folded structures, changes in the folding structure caused by mutations or other events allow the protein to be unstable and deposit as amyloid in tissues. I measure changes in structure and stability of amyloid proteins using a precision instrument called circular dichroism spectrometer that can analyze tiny but important changes. I work on the light chain proteins and the transthyretin proteins to determine factors that lead to protein instability in amyloid disease.

"I measure changes in structure and stability of amyloid proteins using a precision instrument called circular dichroism spectrometer that can analyze tiny but important changes."

Dr. Elena Klimtchuk

**Darrell Cameron, Masters Graduate Student**

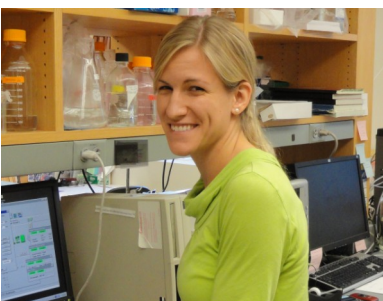


I am a second year graduate student in the Graduate Medical Sciences program working on quantifying two proteins associated with amyloid disease to see if their ratios in serum correlate with the formation of amyloid. I am also working on generating a mutant of one of those proteins so that we can study it in a different state. This will help us get a better understanding of how the protein acts in smaller units. I enjoy working in the Gerry Lab because all of the people are helpful and friendly, and I feel like my research is important and will help us understand amyloid disease better.

"...research is important and will help us understand amyloid disease better."

Darrell Cameron

**Kate Bailey, Research Technician**



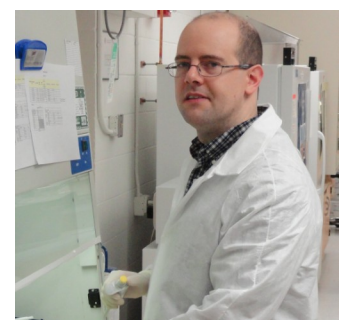
I have worked at the Gerry Laboratory since 2006, after studying biology at Gordon College. I am currently pursuing my Masters degree in public health from Boston University. I receive and process patient samples and purify them to study the amyloidogenic proteins. Over the last four years, I have been fortunate to learn to use several sophisticated instruments for the purification and analysis of the light chain proteins and the transthyretin proteins. I have enjoyed the investigative aspects of research and learning to think scientifically, both of which are encouraged by the collaborative environment of the Gerry Laboratory.

"I enjoy the chance to work on projects that have a positive impact for patients with systemic amyloidosis."

Brian Spencer

**Brian Spencer, Laboratory Manager**

I started out at the University of Rhode Island and earned a B.S. in Zoology. I went on to work at MIT's Genome Center during the race to decode the Human Genome. After years spent as a Team Coordinator, I moved from a production oriented, bioinformatics centered role, to my current role at the Amyloid Program. Here I have my hands full with a number of research projects and I support the lab through the laboratory training of students and by taking charge of sundry of organizational tasks. I enjoy the chance to work on projects that have a positive impact for patients with systemic amyloidosis.



## PEOPLE, PHOTOS

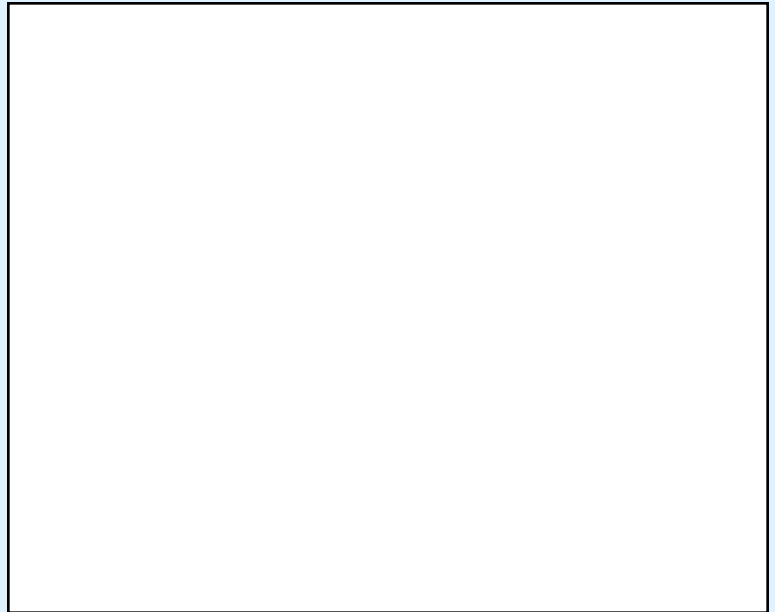
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The Amyloid Treatment and Research Program gratefully accepts financial support for our research and clinical programs from patients, family and friends.

For information on bequests and other planned giving instruments contact us at the address listed above or by phone.

Donations can be made through our website or by mail.

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**Dr. Carl O'Hara, pathologist**



**Pam Soo Hoo, Sophia and  
Dr. Martha Skinner**



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